Psychological Consequences of Rare Diseases

MATURA PAPER

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Preface

'Rare diseases' is a topic I can empathize with and associate to, since I am affected by one myself. It poses not only a challenge, but also a unique opportunity to explore the "world of rare diseases" and interact with other people who share the same or similar problems, from mild to severe impacts. Furthermore, it was my desire to be familiar with other experiences of rare diseases, particularly in respect to psychological consequences and treatments. Ultimately, this study has facilitated the process of handling my personal rare disease, *Takayasu's arteritis*, and enabled a productive link between my disease and school.

"There's no rose without a thorn"

Dedicated to all people who suffer from a rare disease and their families. May this help you in bad times as it has helped me…

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1. Abstract

My personal experience with Takayasu's arteritis, a rare disease, encouraged me to this study on the psychological consequences of rare diseases in Switzerland. The current situation in Switzerland and the survey published in this paper show that only few people are really aware of the set of problems with rare diseases and its consequences. The lack of information and the generally inappropriate education of physicians and psychologists can be devastating for concerned patients. This is also one of the main differences to 'general' diseases. First, I present my personal experience of concernment based on my own rare disease. Second, I follow with a short overview of the initial position and current situation in Europe and Switzerland, which is also reflected by two important associations (Eurordis and ProRaris) and the information portal Orphanet.

The main question of this paper is if the psychological consequences of directly and indirectly affected people (i.e. patients and their families) are negative, positive or both. For that matter, I have proposed six hypotheses, which I tested on 19 people affected by a rare disease and on four experts as a qualitative method to systematically check the hypotheses. At the time of interrogation, all the interviewees were in a mentally stable condition. Every interview has been listed singly in a chronological order. In order to get a quantitative measure, a survey with 250 participants was evaluated, of which 29 were directly affected by a rare disease. Furthermore, many different aspects of patients with rare diseases and affected families as well as valuable insight from "experts" complement this paper, all based on the experience and information of the interviews.

Even though a generalization for all victims cannot be made and there is a large panel of different psychological consequences, I was able to filter the important points to be considered when dealing with people affected by a rare disease, like changes in attitude, personal values, lifestyle, or perceived quality of life. The core result is that the psychological consequences of rare diseases are not only negative, but can even be positive.

2. Introduction

2.1 My story

The realization of a new phase in my life started with horrible repeating nightmares. Every time I had this nightmare, I saw a black shape at the end of my room. I could not say whether this shape was human or not. However, in every dream, I noticed how this dark shape approached my bed, on which I lay. And every single time when the mysterious shape approached, I was paralyzed. During this experience, I could neither move nor scream. All I wanted was to shout for help – or scream out of fear – because I knew I was going to die, or at least I thought I would while I was dreaming…
Half a year after the first nightmare had occurred, my physical and mental state had evidently suffered. I had severe back pains, which worsened from day to day. In addition, I caught angina with high fever and a sore throat. But on 23 July 2010, everything changed.

On that morning, the pain had become unbearable. My mother was distressed to see me in such a condition. Therefore, she packed my bags and dragged me to the hospital, ignoring my reluctance. From my point of view, going to the hospital was completely exaggerated. But, as we all should know, mama always knows best!
As soon as I arrived at the hospital, they started asking questions I didn't really know how to answer. After all, I wasn't in the mood and didn't have the strength to think. Then, they started to do various examinations. An X-ray seemed to show nothing, but the blood tests indicated a very high CRP (C-reactive protein) and many white blood cells. A doctor then started checking my neck and looking into my throat. My lymph nodes were extremely swollen. Maybe a hyper- or hypothyroidism? An ultrasonic examination somewhat solved the case. However, I was not told immediately. "You have got something, but we are not sure what. We do not deal with such cases very often. That is why we are going to send you to Berne, now!", they answered. I was shocked. And slowly but surely, I felt the fear growing inside me.
After a while, I was sent to Berne in an ambulance. I felt horrible during the ride to the hospital spending so much time without any idea of what was going on whatsoever. This lack of knowledge was a psychological burden to me. "What is wrong with me?" was the first question I asked myself. And then I was told the problem – I suffered from a rare disease called *'Takayasu's arteritis'*. Funnily enough, this news did not really upset me. I was surprised of course. But I accepted it, or at least I thought I did…

In the beginning, I had no difficulties at all. Everything was fine, even perfect. I felt perfectly well, my pain was gone, and I felt like I had endless energy. My thoughts were always positive, and my ambition stronger than ever. I had never felt like this before. During almost half a year, my motivation was very high. But suddenly, something changed. While my physical state remained stable, my psychological state started to get worse and worse. I could not explain why at first, but I started to feel miserable. Questions bothered me like: "Why me? Why can't I be healthy? Why do I need to take this medicine?!". It was frustrating! It felt like an endless spiral going downwards. About mid-

winter, I made a decision. I was fed up with the pills I had to take. Surely, it was not that much of an amount. But this amount of cortisone I had to take was enough to drive me mad. Even though cortisone is not a medication that leads to addiction, in my mind, I felt this enormous pressure. I was angry and at the same time afraid because I was dependent on medicine, especially on cortisone. And when I look back, I notice now what an effect this medication had on me – on my personality. I had become a different person, happier than I ever had been in my life, and full of enthusiasm. What frightened me was that I had become like this only due to the cortisone. That was the moment I stopped taking it. And shortly after, I fell into a deep depression. It was one of the worst depressions in my young life. I was only able to think negatively. There was no end in sight.

Eventually, I came out of this black whole. Only with the help of my family and friends, I was able to get some strength back. And some time after, I fully recovered. In summer 2011, one year after the incident, I went through heaven and hell, just to finally get back to earth. Only this time, I have changed to a new person. My way of seeing things has changed as well as the ability to live consciously. Now, I have learned to appreciate the smallest and most unimportant things and to value life and health, all thanks to my *'Takayasu'*. I am thankful to the doctors who detected the disease immediately, and also to my family and especially my mother, who has probably saved my life on that 23 of July 2010.

2.2 What is it about?

In this matura paper I want to show the various psychological consequences that people who are directly or indirectly affected by rare diseases are faced with. Originally, I proposed five hypotheses to cover a large spectrum of different effects rare diseases could cause. After the first few interviews, I added a sixth hypothesis to include additional consequences of rare diseases. Finally, I decided to take a sixth into consideration.
I intended to answer following questions in subsequent chapters:

- *What is the emotional well-being of someone who is affected by a rare disease?*
- *How do people with rare diseases handle daily life?*
- *How do other people see and approach affected people?*
- *What further influences has the disease got on the concerned person?*

To test the hypotheses, I interviewed 19 people affected by a rare disease and four experts in this field. The volunteers who suffered from a rare disease were people in a good psychological condition and were able to talk about their experiences themselves. Exceptions were two families with Huntington's disease, and partly also the Poincilit family with NP-C and the Rietzschel family with hemophilia type A, where most of the information about personal experiences was provided indirectly through another family member. The results I achieved concerning the hypotheses are demonstrated in chapter 7.1. The interviews were evaluated qualitatively.

Important to note: Volunteers who wanted to stay anonymous are marked with a *. Their real name has been replaced by a false name.

Before I started with my matura paper, I wanted to know whether there are other sufferers who also experienced a hard time due to their disease. Subsequently, I wanted to know more about the effects of a rare illness and what it might provoke in people's life. This was the first aim of this study.
The second aim was to raise "awareness" about rare diseases. People who have never heard of rare diseases should get the chance to learn more about this topic. That is why I listed every single interview with a sufferer. It contains their personal story.
My last aim was to establish a reference for people who suffer from a rare disease themselves. I hope that this matura paper will help people to gain ground again – people who were and are in the same situation as I was recently.

In this paper, I specifically concentrated on the psychological effects of a rare disease. Chapter 3 points out the initial position, as for the definition of an orphan disease, the set of problems, organizations and current situation.
To find out if a study or research about the psychological consequences of rare diseases had already been carried out, I researched the Internet and several libraries. However, I could not find anything in this field, except for some single papers, which are based solely on an individual rare disease, and not on orphan diseases in general.

3. Background, initial position and actual situation

3.1 Orphan diseases

3.1.1 What is an orphan disease?

A rare disease – also known as orphan disease – is defined by its number of affected people: In Europe, the prevalence of an orphan disease has to be less than 1 per 2,000 habitants in a population to count as such. Prevalence means the theoretical probability of occurrence of a rare disease (the total number of cases in a population, divided by the number of individuals in the population in a statistical population).[1] Worldwide, about 30,000 known diseases exist, of which 20-25% are rare. Hence, there are 6,000-7,500 orphan diseases – 80% of those are genetic disorders. It is estimated that 6-8% of the population suffer from an orphan disease. Therefore, the number of people who suffer from a rare disease in Europe is about more than 30 million.[2]

The number of inhabitants in Switzerland is currently 8 million. More than 6% of the Swiss population is affected by a rare disease. As a result, 500,000 people in Switzerland are estimated to suffer from an orphan disease.[2]

A lot of orphan diseases have a chronic and irreversible progression. Some lead to invalidity and some are life-threatening. Also, due to low prevalence, complex and highly specific treatment is required. Yet, the tendency of finding out new diseases is increasing. Diseases, which originally belonged to one clinical picture, suddenly are being classified into many single diseases due to new knowledge of their genetic cause.[3]

3.1.2 Characteristic problems of orphan diseases

Although many different rare diseases exist, many of the patients' problems are very similar. Mostly common are isolation, administrative problems, no treatment possibilities, missing hope for cure, insufficient support and absorption of costs. These characteristic problems raise ethical, social, economic, scientific and clinical questions. Here, two specific and connected problems are being pointed out:

Diagnosis and therapy (problem of recognition and treatment)

The problem of diagnosis and therapy is very common for an orphan disease. Low number of cases, missing experience and lack of information, unsystematic capture and barely existent perception make it difficult for a punctual recognition or treatment. Diagnoses can be wrong or come too late. In some cases, the space of time between the first symptoms and the final diagnosis are 5 to 30 years.[3] And sometimes after a

[1] http://en.wikipedia.org/wiki/Prevalence
[2] Printzen, Gert, 2011. Von der Häufigkeit seltener Erkrankungen... . *SÄZ (Schweizerische Ärztezeitung)*
[3] Lazor, Romain and D'Amato Sizonenko, Loredana, 2011. Seltene Krankheiten und Orphan-Medikamente: eine Herausforderung für das Gesundheitswesen. *SÄZ (Schweizerische Ärztezeitung)*

diagnosis is finally achieved, there is no treatment available at all. Furthermore, many patients stay without a diagnosis.
Nowadays, the diagnostic capabilities are improving, safer and more precise, thanks to rapid developments in molecular diagnostics and mass spectrometry. Therefore, a suspicion can be affirmed surer and maybe even cheap screenings can be developed. An advantage of the molecular genetic analysis is the case-oriented and specific classification of a disease. In this way, a variability of different groups of orphan diseases and new, more targeted therapies can be found for individual treatments.[2]
However, diagnosis and therapy of a rare disease are very cost-intensive (see Production and Financing).

Production and Financing

Another important problem is the one of drug production and financing. Pharmaceutical industries show only little interest in supporting research for orphan diseases and development of orphan drugs (medicine for a small number of patients). Sometimes treatments for rare diseases do exist, but the low prevalence (see chapter 3.1.1) leads to a reduced sales market and thus, lack of potential profitability.
To solve this problem rudimentarily, several acts in many countries have been introduced with the aim to motivate pharmaceutical companies to develop treatments for orphan diseases, like financial incentives and marketing exclusivity for seven to ten years. This tactic has proven to be very helpful: Within 25 years, the U.S. has brought out 370 orphan drugs and the EU within ten years about 60 medicaments.[3]
Unfortunately, abuses of the regulations could not be avoided. For example, it is known that some pharmaceutical companies have declared already known and tested drugs as 'orphan drugs'. Subsequently, they were able to extremely increase the price of a new 'orphan drug'. As a result, health insurances refused to pay for the medicine, stating that the cost-benefit ratio was inappropriate.[3]

Nowadays, treatments for many rare diseases are still very expensive. Even when new orphan drugs are developed for a certain disease, the costs are still unaffordable for a sufferer. Health insurances often search for an excuse to outrun the assumption of costs. As an example, the Swiss federal court made a decision for a single case in November 2010 that the health insurance of a victim with Pompe disease only had to cover 100,000 CHF per saved year of life, even though the actual treatment with the medicine *Myozyme* originally costs 600,000 CHF annually. *Myozyme* is permitted by Swissmedic from a scientific point of view as orphan drug, but the economic efficiency was still not recognized from the BAG (Bundesamt für Gesundheit). As a consequence, health insurances started to draw back their assumption of costs for other patients, even though this federal court decision was for a single case.[4]

Another common problem is when an affected person uses 'off-label use' medicine. 'Off-label use' means that a drug is being used that is not on the list with approved medicine

[4] Roadshow for Swissmedic, 1.11.2011, ProRaris

for the disease one suffers.[5] In these cases, the health insurance company is at times not willing to pay for the treatment, because in their opinion, it would be as if they were paying for a test drug.

In conclusion, if orphan drugs are too unprofitable, they are not developed or produced. But on the other hand, if they are too expensive, there is the possibility that the costs are not taken over. In both cases, the access of treatments is being hindered for people who suffer from a rare disease.

This economical approach may be a mistake, since experience gained by orphan diseases may lead to scientific improvements, which will have positive scientific and economical consequences on common diseases.

[5] http://flexikon.doccheck.com/Off-Label-Use

3.2 Organizations

About 100 patient organizations exist in Switzerland for people who suffer from a rare disease. Because these patient organizations are able to work closely together with their members, they are optimally aware of their problems and acquire a lot of valuable information. They also are an important source of information for physicians, if they want to know if a patient suffers from this rare disease and how to treat it.

Patient associations are very important for rare diseases, because the concerned people, who mostly live far apart, are brought together. Furthermore, the knowledge exchange about diagnostic and treatment options, convenient medical accommodations and research projects is supported. In addition, they can offer psychological and social support and break through the isolation.
These patient organizations represent their members and act for their interests facing political decisions, social insurances and private establishments.

3.2.1 EURORDIS

Eurordis is a big pan-European network for rare diseases. The nonprofit organization was founded 1997 in France and represents more than 479 patient organizations for rare diseases in more than 45 different countries and more than 4,000 rare diseases. Patient associations and individual sufferers lead this alliance. They are being financially supported by the members, the French association against muscular dystrophy (AFM), the European commission, corporate donations and from the industry for Medical Devices. Eurordis aims to fight the consequences of rare diseases in the life of affected people and to improve the quality of life for all people in Europe who suffer from a rare disease (through social, caring and pedagogic services). It gives a voice to 30 million patients in Europe with a rare disease.

With the representation of interests on the European scale, the support of research and drug development, the linking-up of patient associations, educational advertising and other activities, Eurordis does everything to support sufferers and to fight against the consequences of rare diseases.
One of Eurordis' main tasks is to raise awareness of the public, national and international institutions regarding rare diseases and its consequences. In addition, they want to improve the access to information, treatments, care and support for affected people as well as support the scientific and clinical research of rare diseases, so that the development of treatments and adequate medication can be assured.
Eurordis is a Europlan associated partner (see 3.3.1).[6]

[6] http://www.eurordis.org/

3.2.2 ProRaris

ProRaris (Allianz Seltener Krankheiten – Schweiz) is a nonprofit organization that was officially founded on June 26, 2010. The representatives of 42 patient organizations and isolated victims were involved in the foundation. The majority of the board members are directly affected by a rare disease, including family members.

ProRaris wants patient organizations and isolated sufferers to work closely together in order to ascertain more information about rare diseases. In that way, these can gain approval by the public authorities and public. Representatives of medical professions and research, social insurances, health insurances, social services and public authorities are involved in the 'rare disease problem'. By forming an alliance, all the patient organizations become an acknowledged dialog partner. Also, ProRaris was founded with the aim to support patient organizations in the accomplishment of their missions.

ProRaris is economically and politically independent and is a member of Eurordis (see chapter3.2.1). ProRaris gives all affected people in Switzerland a voice. Their goal is the recognition and acceptance of the social significance of rare diseases as well as awareness-raising of their consequences in the population and government agencies. Their duty is to make the difficulties and needs for patients clear and to make sure that patients get an equal access to diagnosis, treatment and care provision. Victims shall get the chance to take part in political decisions and be treated as equal partners in the health care system. Another important point for ProRaris is the development of a clear regulation regarding the reimbursement of drugs, therapy and health care, if possible at federal level. Also, the further education about rare diseases for patients and experts (physicians, psychologists etc.) should be guaranteed. At last, ProRaris wants to improve the general conditions for scientific and medical research.

ProRaris' priorities: establishment and enhancement of the dialog with stakeholders, preparation of means of communication, increase of ProRaris' visibility in the media and politics with regard to the development of a national plan, organization of the 'International Rare Disease Day' in Switzerland, awareness-raising of the necessity of centers of expertise.

At the moment, ProRaris is in contact with the BAG, many stakeholders, the media, has connections to politicians and is founding member of the *Rare Disease Community of Interest* (IG Seltene Krankheiten).

ProRaris works closely together with patients, families, doctors and medical experts, representatives for research, public authorities and politics and everyone who is willing to join the fight against rare diseases.

The following diagram shows the interactions between ProRaris, its members and the public agencies:

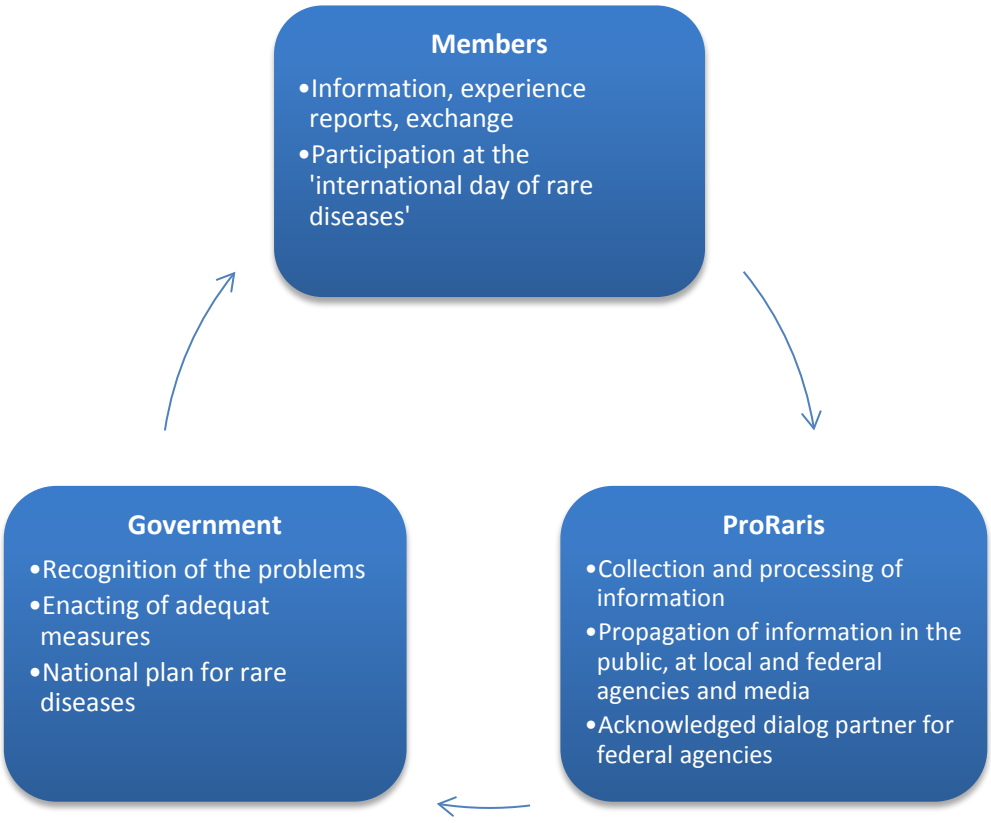


Fig. 1: Diagram showing interactions between ProPraris, its members and the public agencies

On 19 February 2011, the first 'International Rare Disease Day' took place in Berne ('Internationaler Tag der seltenen Krankheiten'), whereat about 450 people participated. The participants had the possibility to interact and get to know each other. 25 member organizations had their own booth. Speakers where among others the national councilor and member of the health commission of the national council (Ruth Humbel, AG, CVP) and the secretary general of Eurordis (Christel Nourissier). A total of 19 articles (online and print) were published, eight radio performances and six television performances (TSR 31.8.11). The approval and success of this event was very triumphant. Finally, they caught the attention of the public and politicians.[7]

"Together we are stronger"

[7] http://www.proraris.ch/

3.2.3 Orphanet

Orphanet is the European web portal for information on rare diseases and orphan drugs. It is meant for the general public and for all stakeholders. The aim is to improve the access to diagnosis, care and treatment of patients with rare diseases.

Orphanet provides information all around the topic of orphan drugs and diseases and several services in this area. It offers an inventory of rare diseases and orphan drugs, an encyclopedia, a directory of services (specialized clinics, laboratories, ongoing research, registries, clinical trials and patient organizations), a newsletter and thematic studies and reports amongst others.

There are currently 40 European countries that work together in partnership for Orphanet. France takes the leading position as a coordinating team. Dr. Loredana D'Amato Sizonenko is the coordinator for 'Orphanet Schweiz'. The national teams are responsible for the data collection of existing specialized services in their countries, for example current research activity, clinical studies and self-help groups. The coordinating team from France takes the responsibility for the infrastructure of Orphanet, as well as the actualization and classification of the diseases and creation of the encyclopedia. Several committees are responsible for Orphanet – both on a European and national level. Independently, they are in charge of this project, so that coherence, progress and the availability of the data bank can be assured.

Orphanet's infrastructure and coordinating working areas are being financially supported by INSERM (the French National Institute of Health and Medical Research), the French Directorate General for Health and the European Commission. National activities are being financed through national institutions and/or separate contracts. [8]

"The diseases are rare, the patients are numerous!"

[8] http://www.orpha.net/consor/cgi-bin/Education_AboutOrphanet

3.3 Current situation

3.3.1 Situation in Europe

In the last few years, the European Commission has taken several steps in order to solve the problem of rare diseases. In the USA, action has been taken several years before. A first big step was the foundation of Eurordis and the development of Orphanet in 1997. Then, on December 16, 1999, the 'Orphan Drug Directive' (REGULATION (EC) No 141/2000) has been introduced by the European parliament (Verordnung über "Arzneimittel für seltene Leiden"). The aim of this regulation is to offer certain incentives for pharmaceutical industries, like marketing exclusivity, for the development of drugs for the prevention, diagnosis or treatment of rare diseases (=orphan drugs).[9]
Further important steps were the '6th and 7th Framework Programme' (FP6 & FP7) within the EU. The 'Framework Programme' is a support program of the European Commission. The FP6 has successfully taken place from July 3, 2002 until 2006. Presently, the FP7 is running for research, technological developments and demonstration. Officially, it started on January 1, 2007. The next 'Framework Programme' is predicted to be in 2014 with the name "Horizon 2020", which will integrate the program for competitive capability and innovation.[10]

In April 2008, a three-year project started with the name 'Europlan' (European Project for Rare Diseases National Plans Development). It is a guide for the European countries and member states of the EU, in order to provide National Health Authorities recommendations how to create a national plan for rare diseases and to motivate them to create national strategies for rare diseases.[11] A national plan is a method and process of defining, developing and outlining various possible actions to meet the existing or future needs for a country or a group of people who share the same problems.[12]
The first national plan was in France from 2005-2008 with a budget of 108 million Euros.

3.3.2 Situation in Switzerland

The Simplified Orphan Drug Authorisation is governed by the Swiss Medicines and Medical Devices Law (Art. 14.1.f of LPTh, the Law on Therapeutic Products) and by the Ordinance of the Swiss Therapeutic Products Institute relating to the Simplified Medicines Authorisation.[13] Only a few single initiatives have been launched in the parliament in 2010/11 by Cassis, Gutzwiller and Humbel.[14] Furthermore, the foundation of a 'Rare Disease Community of Interest' (IG Seltene Krankheiten) has been founded on

[9] http://europa.eu/legislation_summaries/internal_market/single_market_for_goods/pharmaceutical_and_cosmetic_products/l21167_de.htm
[10] http://cordis.europa.eu/fp6/dc/index.cfm?fuseaction=UserSite.FP6HomePage
[11] http://www.europlanproject.eu/Home.aspx
[12] http://www.eionet.europa.eu/gemet/concept?ns=1&cp=5486
[13] http://www.swissmedic.ch/daten/00081/index.html?lang=de
[14] Roadshow for Swissmedic, 1.11.2011, ProRaris

24 August 2011.[15] An Orphanet-portal for Switzerland (Orphanet Schweiz) merely exists since 2001, based at the University Hospitals of Geneva. The public authorities and the public were not aware of the set of problems about rare diseases and its consequences until recently. When it comes to taking over the costs of a treatment or medical product, it mostly depends on the medical adviser of a health insurance for a final decision. Often, there is a danger of different interpretations of the scope of the basic insurance benefits when it comes to an assumption of costs. Also, because of the low prevalence, the experience with orphan diseases is smaller.
Furthermore, the phenomenon of the low interest of pharmaceutical companies to produce orphan drugs is well known in Switzerland, too. There are many rare disease patients who are dependent on "off-label use" medicinal products. As a result, health insurances will not cover the costs of these essential drugs, on the grounds that they are not responsible for paying for a "test drug". As an example: Niemann-Pick patients are dependent on the medicament Zavesca®, which has originally been developed for Gaucher's disease. For Niemann-Pick patients, Zavesca® is therefore an "off-label use" medicine. Consequently, health insurances are not willing to pay for the costs.
This conflict between health insurance, patient and the pharmaceutical industry (health insurance → patient ← pharmaceutical industry) is an urgent problem that needs to be solved by the politicians. In comparison to other EU countries and the USA, Switzerland is behind the times.

To change this situation, ProRaris was founded. The set of problems was unknown. About 100 organizations existed, but they were alone and with that small number of members, they had very low influence. By pressuring the public authorities and more presence in the media, the BAG is finally ready to develop a national strategy concerning rare diseases in Switzerland.

[15]http://newsroom.interpharma.ch/sites/default/files/download/pdf/positionspapier_ig_seltene_krankheiten_d_def.pdf

4. Hypotheses

To find out specific psychological effects of patients who suffer from rare diseases, I have proposed six hypotheses. These were built-in to the list of questions for the interviews. Initially, I created five hypotheses. However, after the first few interviews, I noticed that I had to add one more (B), for the reason that I had received several common statements ("Health is not self-evident" or "Health has become much more important to me").

⇒ **Hypothesis 1 (A):** People who suffer from a rare disease value life more than before the diagnosis or illness.
⇒ **Hypothesis 2 (B):** People who suffer from a rare disease value health more than before the diagnosis or illness.
⇒ **Hypothesis 3 (C):** People who suffer from a rare disease live more in the present and enjoy the moment.
⇒ **Hypothesis 4 (D):** People who suffer from a rare disease are more prone to depressions than before the diagnosis or illness.
⇒ **Hypothesis 5 (E):** People who suffer from a rare disease are being excluded by society (e.g. state, health insurance, environment).
⇒ **Hypothesis 6 (F):** People who suffer from a rare disease see life in a more realistic way than before the diagnosis or illness.

It is clear that the formulation of these hypotheses is very delicate. Then again, it was very difficult to formulate more precise hypotheses due to the fact that there were no existing assignments to go on. Hence, I had to think of the simplest possible effects of a rare disease.

Hypothesis A was devised because the affected people have survived the disease, some close to death, and consequently look differently at the value of life. Hypothesis B was created out of A. Many interviewees often stated that health had become much more important since their diagnosis. Therefore, I had to make a difference between valuing life and valuing health. The idea of hypothesis C was that sick people try to enjoy life (by living in the present), since they are aware of the fact that life can end very fast. It contains living consciously and to take every day as it comes. Hypothesis D arose from the idea that affected people could feel lonely and helpless because it is quite unknown and not many people are affected by the illness. Moreover, they could fall into depression just due to the knowledge of being sick. Another idea, which is shown in E, was that people who suffer from a rare disease could get excluded because of the rareness of their disease. This could be the case when non-affected people show a lack of understanding towards affected people, especially when the disease is visible. At last, I created F out of the idea that concerned people could have a new perception of things, because the disease shows them that even when it is rare, everyone can become affected.

5. Material and Methods

To analyze and test the hypotheses, the following methods have been used:

- Structured interviews with sufferers
- Structured interviews with experts
- A survey for the general public (no target subjects)
- A visit in the '*camp for children and adolescents with phenylketonuria'*
- An afternoon with the self-help group '*Junge mit Rheuma'*

Furthermore, to find out if a study already exists depending on similar hypotheses or an analysis of psychological consequences of concerned people, I researched in the *'Zentralbibliothek Zürich', 'ETH-Bibliothek', 'Medizinbibliothek Careum'* and on the Internet. The key words I used were: psychological studies, research, effects, and consequences of rare diseases. For further research, I checked articles in PubMed, Eurordis an orphanet. **Unfortunately, I could not find anything that spoke for rare disease patients in general.** The nearest results I came to were:

> *"Stress and well-being among parents of children with rare diseases: a prospective intervention study"* – Dellve L., Samuelsson L. & Co., 2006 Feb.[16]
>
> *„Psychological Effects of Carcinoid Disease: A study for carcinoid Patients and their Caregivers"* – Soliday E. and Smith S., 2002 Apr. [17]
>
> *„Psychosocial aspects of patients with Niemann-Pick disease, type B"* – Henderson SL. , Packman W. & Co., 2009 Nov. [18]

As already mentioned, these studies are only based on individual diseases. That is why I have not gone into further analysis of these works.

5.1 Structured interviews

As a basis in my paper and the main part of my work, fifteen people who suffer from a rare disease were interviewed personally (face to face) and in German. The meeting point was usually in a café or in the house of the concerned person or family, so that a comfortable atmosphere could be created. In addition, four concerned people were interviewed per telephone, for the reason that they live so far away. That is why three of the interviews were held in French, and only one in German. A personal interview took about 1-4 hours, a telephone interview about 30-60 minutes.

[16] http://www.ncbi.nlm.nih.gov/pubmed/16448482
[17] http://www.carcinoid.org/content/psychological-effects-carcinoid-disease-study-carcinoid-patients-and-their-caregivers
[18] http://www.ncbi.nlm.nih.gov/pubmed/19877061

The concerned people were contacted through self-help groups, ProRaris, self-made brochures situated at the Inselspital Berne and University Hospital of Zurich, and a publication in the *Glückspost (No. 33/18.08.11)*.

The interviews were 'open interviews'. This means that the interviewee was also allowed to ask questions and that not every question on the questionnaire had to be answered strictly. It should be more of a conversation. Yet, I tried to have all the questions regarding the hypotheses answered.
Before the interview, I prepared a questionnaire that was adjusted to every interviewee, depending on his or her rare disease and questions that were prepared to verify the hypotheses. To have a proof, I used a voice recorder and a mobile phone to record the whole interview. Simultaneously, I took notes during the interview. Furthermore, every interviewee had to sign a letter of agreement, in which he or she had to give his or her name and date of birth. In addition, he or she had to mark with a cross if he or she wanted to stay anonymous or be mentioned. Those whom I contacted per telephone had to send me the signed letter of agreement per mail.

As experts in the field of rare diseases, I interviewed altogether 4 people:

- Esther Neiditsch, the president of ProRaris
- Dr. Pierre Krayenbühl, doctor of the USZ who is in charge for two groups with rare diseases
- Dr. Michael Fischer, psychiatrist, who has experience with some cases of rare diseases
- Alessia Perifano, psychologist who attends to patients with lysosomal storage diseases, exchange per e-mail, since she lives in France

The material and methods here are analog to the interviews with the affected persons. All the obtained results were qualitatively evaluated.

5.2 Survey

The structured survey is an additional method to the interviews. In this way, the paper gets a further – this time quantitative – data collection. The questions were adapted to the hypotheses and were originally written in German, since the participants mostly came from the German speaking part of Switzerland. A first aim was to obtain knowledge about rare diseases and a general opinion of the public. Therefore, there were no specific target subjects. No extra knowledge was required to complete the survey. A single criterion was that the participant had to live in Switzerland. A second aim was to have a comparison between the affected people and the non-affected people in reference to the hypotheses.
The participants were able to fill out the survey online on the platform *"www.sondageonline.com"*. This method is very practical, for the reason that a lot of participants can be obtained in a relative short time span.

The survey link was sent per e-mail to friends, acquaintances, teachers, physicians and sufferers. To achieve a high response rate, the additional demand was made to remit it to further people. In addition, the survey link was published on the social network 'Facebook'.
250 people participated in the survey, in which 29 were affected by a rare disease themselves.

5.3 Events

Furthermore, I visited two events as extra: the *'camp for children and adolescents with phenylketonuria'* and the self-help group *'Junge mit Rheuma'*. During these events I wanted to collect further impressions and experience with affected people in whole groups (i.e. associations and camps).

6. The interviews

6.1 Personal interviews

6.1.1 The Poincilit family – Niemann-Pick type C

Niemann-Pick is an inherited metabolic disorder. Three different types of the Niemann-Pick disorder exist: Type A and B are caused by a lack of an enzyme. For type C the cause is a scarce transport of self-made cholesterol in the cells, following a toxic accumulation of lipids on neurons. This leads to structural and functional damage in the cells and tissue. Symptoms of NP-C are gaze paresis (disturbance of eye movement, gaze straightforward), learning and concentration problems, progressing intellectual decontrol, sudden deficit of tonicity, epileptic seizures, depressions and hearing loss. The prevalence of this autosomal-recessive disorder is 1:130,000 individuals. Thus, less than 20 individuals are estimated to suffer from NP-C in Switzerland.[19]

Mathias (31), Hanny and Christoph's eldest child, grew up as a normal boy at first. Christoph says, looking back, indications for Mathias' illness did exist. His symptoms were very weak at the beginning, so that nobody really noticed anything. It started with insomnia (problems sleeping). Then, he was brought in a hospital for the first time when he was five, due to a big spleen. Various tests were performed, with the ultimate result that at least it was not cancer. Mathias was sent back home, which was a big mistake. Once, he crashed into a pole with his bike. "That can happen", Christoph thought. Still, he had the feeling that these things were happening too often. When the boy was nine, further restrictions started to emerge. He had more falls, psychological problems, difficulties swallowing and epilepsy. Mathias went through many tests, but without any success. At the age of 20, his parents frustrated gave up searching for a reason for Mathias becoming more and more handicapped.

At the age of nine, Alix (29), their second child, started suffering from hearing loss. At school, her IQ started to decrease, without any particular reason. She went to secondary school first then to *'Kantonsschule Zug'*, which she had to give up. At last, Alix made an apprenticeship as businesswoman and landed a job in her field. Nonetheless, her first psychological problems started when she already was an adult. She had difficulties handling her daily life and suffered from permanent headaches. Her parents told her that she needed professional help, which she refused for a long time. Yet, her psychological condition worsened, and Alix was diagnosed with depression. She was treated with medication and after no improvement, sent to psychiatry. But even with treatment, her condition still got worse. Nevertheless, the psychiatry released her, something which the Poincilits find irresponsible till today. The physicians seemed to have worsened her condition. After Alix had been mentally ill for two years, her parents wanted a precise neurological examination. Subsequently, they brought her to the University Hospital of Zurich. But the doctors could not find anything wrong, and so they were ready to release

[19] http://www.npsuisse.ch/Niemann_Pick.htm

her, again. But this time, the Poincilits insisted that Alix should stay, and pressured them that the physicians should keep searching. In April 2010, an eye specialist noticed a gaze paresis and had the idea that it could be NP-C. At last, they made a bone marrow puncture (analysis of samples of bone marrow) to see if this assumption was right, and indeed: it came out to be Niemann-Pick type C. Back then, Alix was 27.

After Alix' diagnosis was clear, Hanny and Christoph were sure that Mathias also suffered from NP-C. He was finally diagnosed at the age of 30.
Then, another problem appeared: Was it possible that their youngest daughter Zita (23) was also affected by this rare disorder? With 18, she already suffered from hearing loss. Looking back, the parents remember her also having concentration and linking problems, a short-term memory problem and a gaze paresis. A few months later, Zita decided to do the test, which proved to be positive – Zita also suffers from NP-C. Nevertheless, she seems to have fewer symptoms than her elder brother and sister. At first, Zita was relieved to know from what she suffers. Afterwards however, she started to feel miserable and desperate. By seeing her brother grow up, she knew what the course of disease was. Thus, she got angry with her parents and the world. Later anyway, she contacted other families with NP-C. Being able to talk to other victims helped her a great deal.

Before the diagnosis, Zita was often bored and not really happy. She never really thought about the future. She had to give up her studies on economics and apprenticeship due to progression of the disease. Now, Zita is working in a laundry, which she finds interesting and diversified. She is doing well and tries to not let herself get into too much stress. Zita comments that some old friends have grown apart since they know about the illness, out of the reason that she cannot do everything anymore. Yet, her good friends have grown very close. She is thankful for their and her parents' support. Due to her disease, Zita has grown strong and lives her life consciously. Because she cannot plan her future in advance, she makes sure to enjoy every day and to take it as it comes. It does not matter how long she will still live. In her eyes, every new day is a gift. Zita values small things now and does not get annoyed that easily anymore. Time has become very precious to her. Therefore, she schedules it very carefully. More than ever, she says to appreciate life now.

Two years ago, a new medicine called Zavesca® came to market. Zavesca® does not cure NP-C, but slows down or stops the course of the disease. Side-effects are lactose and fructose intolerance. Both Alix and Zita are under this medication. Zita states to be able to read better now thanks to the medication.
Unfortunately, Zavesca® is only on the list for patients with Gaucher's disease. Therefore, the Poincilit children need to use it as "off-label use" medicine. As a consequence, the health insurance does not want to pay for it. That is why Actelion is paying the medication at the moment. For how long, the Poincilits do not know… Hanny and Christoph are still fighting with the health insurance to take over the costs of the medication. In their opinion, everything would be much easier then.

Nobody of the family had ever heard of the Niemann-Pick disease before. Hanny is more than glad that her children are finally diagnosed. However, at first, she was shocked. After the diagnosis, Hanny and Christoph started researching day and night. Never would they have thought that their children suffered from such a disease. At the beginning, they had trust in the physicians. According to a physician from the USZ, they were just in the wrong place at the wrong time. However, they do not believe that, stating that medicine has progressed enough. Christoph believes that a great problem was that physicians were not enough informed about this rare illness. Therefore, it did not come in question as a possible diagnosis.
Nowadays, Christoph affirms that he will try everything for a better future for his kids and prospective sufferers. They also want to help other people and inform parents in time. Determined to raise awareness, Christoph has founded an organization for Niemann-Pick patients, called NPSuisse, which also is a member of ProRaris. In addition, the Poincilits have shot a documentary about their story, “Living with NP-C”. This was also a way of processing this happening.

Sometimes, Hanny wishes to have never heard about NP-C. Nevertheless, she is happy to know what it is and consequently, what can be done. It is something to start with and makes it easier. In her eyes, "it is as it is". Thus, you need to make the best out of the situation.
Nowadays, Mathias is in an institution for severely disabled people, and Alix needs additional support. Luckily, Zita remains independent. Christoph cut back on work, in order to concentrate on the fight against the disease. The Poincilits try to enjoy life as much as possible and to live more in the present. That is why several months after diagnosis, they spontaneously decided to take a vacation on a faraway island. They believe that together, they are stronger and can manage.

6.1.2 Niklaus Hirsig – Neurofibromatosis

Neurofibromatosis type 1 has many different forms of appearance, which makes diagnosis difficult at times. The prevalence of this autosomal dominant disorder is 1:3'500, whereby men and women are affected equally. Typical symptoms can be café-au-lait spots, neurofibromas and axillary freckling. Neurofibromatosis can affect the bones, nervous system, soft tissue and skin. Certain abnormalities can occur during the development of almost half of the concerned children, such as slowed motor, linguistic and emotional improvement.[20]

Niklaus Hirsig (19) has suffered from neurofibromatosis since birth. Nonetheless, to notice this, you would have to ask him first. Niklaus' symptoms are a few café-au-lait spots and a lump on the visual nerve. When he reads for a long time, Niklaus gets exhausted, although his view is not restricted. Once a year, he goes to the eye specialist for a checkup. The diagnosis was very easy. His café-au-lait spots were checked and after that, it was clear. But for Niklaus, this is not a big deal at all. He once in a while searches on the Internet about this rare disease, and sometimes goes to the meetings for people with neurofibromatosis. The reason for this sparse interest is a very tactical one: Niklaus is afraid that, if he discovers too much information about his illness, his attitude towards life will suddenly change in a negative manner. That is why he does not bother so much about it. He doesn't need any medicine or therapy. The only situation when he needed medication was during his younger years as his bone structure started to change due to the neurofibromatosis. He subsequently suffered from scoliosis and required occupational therapy until the age of 5. Anyhow, since Niklaus has an absolute normal daily routine and is barely restricted, he does not believe to be different compared to other adolescents. Because he grew up with this illness and does not know how it would be otherwise, there has never been a significant change in his life. Even if the doctors told him now for the first time that he had such a disease, Niklaus believes that he or his personality wouldn't change because of this revelation.

Because his mother is a board member of the SNFV (*"Schweizerische Neurofibromatose-Vereinigung"),* he visits the annual meetings. He says that although his mother is much more engaged in this topic than he is, they both do not talk much about it. He thinks that the reason for his mother dealing so much with neurofibromatosis and the association is that he himself is affected by this disease. But Niklaus did meet people, which were much more affected by neurofibromatosis and showed noticeable symptoms. People who see such a concerned person mostly get out of his or her way. Niklaus only knows such people from the SNFV meetings and says that these people clearly have more troubles than he does. This is another reason why he counts himself as lucky, because he is healthier and doesn't have to worry about criticism so much. He also notices that parents of children who are more affected than he is get much more involved with this illness. As an example, he knows a child that he babysits regularly with a tumor in the head due to

[20] http://www.neurofibromatose.ch/wasistnf.htm

the neurofibromatosis. The child's parents do everything to protect and increase its quality of life.
Niklaus' family supports him very much, and concerning his friends, only the most important ones know about his disease and accept it. "If they didn't accept it, they wouldn't be good friends", Niklaus states. Till the present, there have never been problems due to Niklaus' illness. He was never excluded by the people who knew from what he suffers. Also, it never came to his mind that this could become a burden to him. Niklaus is very satisfied with the situation and of course, happy and thankful to be quite "healthy". Though his momentary situation could always change, his prognosis looks good. He himself is very optimistic about the future, as he maintains a positive attitude. Because he does not look or feel ill, the disease has always been in the background of his life, never really bothering him.

6.1.3 The Rietzschel family – Pulmonary hypertension and Hemophilia type A

Pulmonary hypertension, or PH, is a very rare pulmonary disease, marked through a high blood pressure in the arteries between the heart and the lungs. It is a chronic and incurable disease with a prevalence of about 1:100,000. The heart and lungs are affected: Structural changes in the vascular system of the lungs and the right chamber of the heart can become visible. This leads to restricted physical capability and even to a shortened life span.[21]

Hemophilia is a bleeding disorder. Men are predominantly affected. It is a hereditary illness, whereat the affected person lacks clotting factor 8 (type A) or 9 (type B). Therefore, the blood clotting is defect and people who are affected tend to bleed for a longer time. Consequences could be difficulties during operations and inner bleeding that are hard to control. Its prevalence at birth is 1:10,000.[22]

Heinz (68) and Melitta (69) have been together for many years. Even after all that has happened in their family, they still stay together. Their son Glenn (*17.2.69; †34) suffered from hemophilia type A and passed away 9 years ago. The way it happened was an unforgettable scandal, and they will never forget the story that tore them apart before his death. Glenn's disease, which normally is heritable, was unusually activated by a spontaneous mutation. It was an enormous shock for the parents when they were told the reason for all the bruises their one-year-old son had. Glenn was kept at the children's hospital in Zurich under constant supervision for one year. From then on, his parents fully attended to his illness and researched all about it.

Nevertheless, besides the intensive care at home, as Glenn grew up, his parents always tried to give him as much freedom as he needed. Together with Glenn and his sister, they didn't miss the summer vacation at the beach or skiing holidays in the winter. And skiing, not to forget, can be extremely dangerous, especially for a boy with hemophilia. Once, it even happened that the boy, curious as children are, took his father's skis without permission and went skiing on his own. It was a great fright for Heinz and Melitta when they realized that their son had gone skiing alone. Fortunately, nothing happened and the boy came home safely.
This was the difficult decision that Glenn's parents had to make very early: Should they lock him in a "glass cage" or let him be as free as possible? "The most difficult part is to decide for your own child," they remark. "We gave him the freedom he needed. Yet, you need to be aware of the risks and do everything with a lot of care and patience".

For Glenn, it wasn't always easy either. He gained a lot of information about his illness. When he was younger, children used to mock him because of his bruises. Even adults used to claim unknowingly that his parents probably beat him. Heinz and Melitta state that puberty was hardest for him. Sometimes, he used to say: "Why must I have this disease?" Anyhow, Glenn had many friends and was never excluded.

[21] http://www.lungenhochdruck.ch/php/index.php
[22] http://www.wfh.org/index.asp?lang=EN

Later, during his studies at university, he met a woman who he eventually married. His parents were sad to see how poorly his wife cared for him and how distanced they have become because of her. Yet, the parents' greatest fear was that Glenn's wife could leave him one day because of his illness.
Several years passed without any contact, when Heinz and Melita found out that their son was lying in intensive care. That is where they saw him for the last time before he died. The cause of his death was a contaminated infusion with hepatitis C – a disgrace in that time. Glenn's parents assert that it was what Glenn had always been afraid of, already since he was a small boy: to catch an unhygienic infusion.

Melitta and Heinz went to see a psychologist for five years because of all that had happened. In the present, Melitta says that it is never easy to process a disease and the loss of a child. That is why she has decided to write a book about it, so that she can learn how to deal with it. Heinz' comments: "What can you do? You have to draw a line, or you will break down because of it."

The next slap in the face was when they were told that Heinz suffers from a rare disease, too: pulmonary hypertension with a pulmonary emphysema.
It started with a severe pneumonia in October 2010. Heinz was hospitalized, and after a stay of several days, he was said to be healthy again. Heinz was released with good oxygen content, but with a wrong diagnosis. The doctor said that he had to use the oxygen concentrator round the clock now, because his alveoli had a bit of purulence and were inflamed, so the oxygen would help against the inflammation.
Then, Heinz caught pneumonia for a second time. He landed in hospital again, and this time, the doctors were motivated to find out what was the cause. "It was a great psychological burden to not know the cause for this bad state of health", Heinz and Melita affirmed. After going back and forth from one hospital to the next, a diagnosis was finally able to be made by the head physician of the pneumology in February 2011 thanks to an ultrasonic examination. They were extremely relieved. From then on, Heinz tried to accept his "destiny" and has ultimately learned how to deal with this new situation. "It depends on age, the experience and how you perceive something", he comments. The only thing he cannot accept up till now is that he cannot help his wife with the daily work in the household or garden. "This is something I still need to learn!", he claims.
Because of the disease, Heinz had to give up many things he used to love doing. It was a large blow for Heinz when he was told that he had to give up sports in general. What he especially misses is skiing and swimming. Sometimes, he goes to the public swimming pool to swim two or three laps. Then, he always needs a short break to breathe in some oxygen with his concentrator, and then he can go on. "This tube can be really annoying sometimes", he comments teasingly. Another matter the Rietzschels are restricted to is travelling. Before, they used to travel a lot to different countries, visiting foreign cultures. But because of the oxygen concentrator, this has become rather difficult. The portable concentrator constantly needs to be filled up at the "main oxygen tank" that has its place

at home. At first, Heinz had to get accustomed to use the “oxygen station”. But now, he has learned how to deal with it.

A prognosis for pulmonary hypertension is not easy to make. Some doctors think that the life expectancy after diagnosis is about two to three years. Others believe that affected people can still live up to 10 years with the disease. However, Heinz is optimistic: “It is my body, so I decide”. Both Heinz and Melitta are not afraid of death. Because they know that life could end any day, they have talked about it a lot. Heinz refuses to have a lung transplantation. He has the opinion that when the time comes to leave, he will be ready. “It is what it is”, Heinz believes. Until then, they like to spend time together or sit in a café and chat with their friends, who also encourage him morally.
Heinz is an example for someone without any psychological problems who can handle his disease nearly on his own. Yet, he is very thankful for his wife’s support – mentally and physically. He wouldn’t know what to do without her. Melitta is always with him, giving him a lot of strength. Even though the Rietzschels have lost some confidence in the doctors owing to the many experiences they went through, Melitta’s attitude stays very positive: “I will never give up hope!”. After all these incidences, Melitta and Heinz still stick together. They are happy again and thankful to have each other!

6.1.4 Maria K. *– Takayasu's arteritis

Takayasu's arteritis, also called the "pulseless disease" owing to not detectable pulse in arms and legs in some cases, is an autoimmune disease, belonging to the group of the vasculitides (inflammatory vascular disease) and is a chronic illness. The vascular walls of the aorta and its larger descending branches are inflamed due to the immune system attacking the own body. Consequently, this leads to aneurysms, stenosis, and thromboses. It is best known in young Asian women. The prevalence is less than 1:1,000,000. Symptoms are blood pressure difference between the right and left arm, fever, night sweat, fatigue and weight loss. If not treated, this disease can lead to death.[23]

In June 2009, Maria (33 and Swiss) was diagnosed with Takayasu's arteritis and subsequently began with the corresponding therapy. But symptoms had already existed much earlier at the ski camp in January 2009, where she fell awfully sick. She had high fever, fatigue and a cough. And even though it seemed to get better, she never fully recovered to her previous healthy state. Since the coughing was the most severe of the symptoms, the doctor took an X-ray of her lungs and prescribed her antibiotics after not finding any obvious problem, but measuring a high CRP. Nevertheless, it appeared not to help substantially, as her physical condition fluctuated up and down. When no improvement occurred, Maria went to the University Hospital of Basel, where diverse blood tests and another X-ray were taken. During interrogation, she confirmed having problems with weight loss and night sweat. A CT (computed tomography) made the structural change of her carotid arteries visible. To achieve a definite diagnosis, Maria had to do a PET (Positron emission tomography). The results were clear: Maria suffered from Takayasu's arteritis.

Since then, Maria has moved to Eastern Switzerland and goes for regular whole body checkups and blood tests to the University Hospital of Zurich every three to six months. She continues her therapy on Imurek® and cortisone and takes diverse medicine against its side effects. This medical cocktail should suppress her immune system, in order to prevent further inflammation of her arteries. She says that the medicine did help her to get better, even if the side effects were annoying. For example, she became nervous and hyperactive through the cortisone and lost hair caused by the Imurek®. However, Maria takes her new life situation with a reasonable approach. Finally, it was important to know what was behind all the symptoms that she experienced in early 2009. She was not very surprised to hear the diagnosis, as she had researched about the symptoms before in the Internet, trying to find out what was possibly behind it, and soon came across the term "Takayasu's arteritis" for the first time. Hence, she received the news relatively well prepared.

"I don't have any pain and I am barely restricted in my daily life activities, so I can't complain", Maria says. And because they found out about it in early stages of the illness, the consequences are not that severe. Yet, it was not easy for her to accept it at first. After

[23] http://www.deutsche-gefaessliga.de/takayasu.html

more than two years of medication and no end in sight, she had to learn that her disease will remain forever. Also, side effects due to the suppressed immune system, for example dermatophyte, have been very annoying. Because it is a very rare disease, Maria feels a bit lonely sometimes, since she does not know anyone with the same disease whom she could share her experiences with. But her family is very supportive and researched right from the beginning on. They took her seriously. Her husband was initially very affected by it, but handles the situation very well by now. Her colleagues and other people who were introduced to her disease were curious at first and wanted to know more about it. But soon after, the unusual situation returned to normal. She was never excluded nor did she exclude herself.
At the beginning, Maria thought that she had to "live her life more" than she did before the diagnosis, since she survived a disease which can be perilous without therapy. But afterwards, she didn't feel like changing much in her life perceptions. She just wanted to live a normal life, as she did before. That is exactly what she does now. "The disease is just here, a constant companion", Maria says. "Yet, I try to not let it become center stage of my life". To her, it is important that her sickness doesn't play a too big role in her life. In her eyes, her attitude towards life hasn't changed since the diagnosis, except that she has found a fondness for sports. Even though no cure exists for Takayasu's arteritis, Maria is optimistic towards the future. She is happy to become a mother in March 2012! In an e-mail, Maria wrote me:

"The psychological consequences of it are great! When you're pregnant, you can't think of anything else. It means that my disease is actually for the first time really in the background and I barely have to think about it."

6.1.5 Bruno Bosshard – Pulmonary hypertension

The first lung transplantation in Switzerland was carried out in 1992 at the University Hospital of Zurich. At that time, it was an experimental procedure. Nowadays, this intervention has become a therapeutic alternative for patients with a severe lung- or heart-lung-deficiency. Nevertheless, a lung transplantation still hides many problems, for example the risk of rejection and infections. On average, 50% survive after a time span of 5 years.[24]

Bruno (54) received his diagnosis in 1998 at the age of 42. Prior to this key moment, he did not experience any particular health problems. It took him three months to finally figure out the causes for his worsening health. Finally, a simple coincidence revealed the deeper physical problems behind his bad health.
His first symptoms started with constant syncopes during hard labor. A heart specialist at the University Hospital of Zurich ran some cardiac tests, without conclusive results. Subsequently, Bruno's condition got worse. At a medical congress, the heart specialist presented this bizarre case to other physicians and specialists. Applying the exclusion principle, they finally formulated a likely cause: Bruno could suffer from pulmonary hypertension. The only way to test this assumption was by measuring the pressure between the arteries of the lungs and the heart. Therefore, a right-heart catheter was used to measure Bruno's pressure in the heart, resulting in unusual high values. Because of this discovery, the physicians were finally able to make a diagnosis. From then on, he was no longer allowed to do any kinds of sports or other activities leading to high physical stress. Unfortunately, neither the physicians from the hospital nor the health insurance could help him. Consequently, this new situation made him feeling lonely and helpless. Nobody in the medical field in Switzerland really knew enough about this disease to improve his condition. At that time, there was no official and tested treatment to cure, or at least dampen the effects of his disease. Hence, Bruno received a medication that was still in the testing phase, called Ilomedin. According to the manufacturer, it should dilate the vessels. It helped him a little, though not enough. This probably was based on the lack of information on the right dose, since it was an experimental medication. In addition, the health insurance refused to pay for it, arguing that its effects were not known well enough, a common problem for rare diseases.

Meanwhile, Bruno's condition became worse; especially since his heart was affected. He fell into depression and had thoughts of life resignations. The only chance to survive was a lung transplantation. But he didn't feel comfortable with that option, as this surgery appeared to be too risky. For example, his body could reject a newly implanted pulmonary organ, a frequent problem in transplantation methods. Furthermore, it is not the last step in the process of a recovery, but only the first one. For Bruno, it would mean a new fight without any guarantees of a positive outcome. But with the great support of his family, he finally gave in and agreed upon a transplantation. Nonetheless, it was a huge burden for his family, although Bruno was ready for any, even fatal, outcome of the surgery. In his mind, he had already lived a fulfilling life, and if he had died after the

[24] http://www.novaria.ch/php/index.php

surgery, he would have accepted it. He was tired of the pain and daily health struggles to wish nothing more than to get over with it, the quicker the better. But his physician wouldn't let that happen. Shortly before his resignation, his physician made it clear to him that it is the loved ones that are left behind with sorrow and grief after a patient's death. Bruno started to realize that he needed to survive for his family. His children would not be able to live a pleasant childhood. And with his death, he indeed would abandon more his family than himself. It was a tough decision to accept a risky surgery procedure at that time. But in the year 2000, Bruno finally had successful bilateral lung transplantation.

To Bruno, it was never an option to exclude himself from the society. He actually wanted quite the contrary. Therefore, he founded an association several years ago – the SPHV *(Schweizer PH-Verein)*. Death is an important topic discussed among the members of SPHV. Because chances of a long life are very reduced when you suffer from pulmonary hypertension. And since there are little references of lung transplantation, Bruno has already discussed the relevant issues surrounding his potential early death with his family, so it is not a burden or surprise anymore. In fact, personally to him, he would be ready for it. "If the moment comes, it will be the right one". Today, death is always present in Bruno's daily life. To cope with this, you need a positive environment.
He receives a lot of support from his family and has an open ear whenever he wants to talk about his illness. His family has learned to deal with it. Yet, now and then, the fear something bad could happen seeps through. They will never be able to live as relaxed anymore as they have in the past.
Some people had misconceptions at the beginning. Some were even shocked as they heard about Bruno's disease, and consequently wanted to skip those issues or Bruno entirely. "Good" friends suddenly distanced themselves from Bruno, stating the reason that they were short on time. They never visited him or asked from what he suffers. In contrast, other relationships grew more profound. Those people sacrificed much of their time and really wanted to know more about this rare illness.

Bruno believes that after all what he experienced since the first diagnosis, his basic attitude has changed fundamentally. Before, he had clear defined aims, where deviations and distractions were not tolerated. Now, he has become much more open, and he does not get angry that easily anymore. "It's of no use at all to be annoyed all week because of little matters". Also, he has become much more flexible and tolerant, and thus developed a different handling with certain issues. Since his lung transplantation, he sees things more consciously and in a more realistic way. Generally, he is a happy and content person today. After the lung transplantation, he had new perceptions about the quality of life. Now, material goods are less important than health, consciously living and social interactions with family and friends. Self-pity has no more space in his life.
Bruno doesn't want to diminish the quality of his life because of the transplantation. That is why he still is travelling yet being aware of the potential risks. "You need to be clear about the consequences from that disease", he states. In Switzerland, lung transplantation has only existed for about 20 years. That is why there is no long-term

experience. But Bruno feels good and thinks that it will turn all right for him, though he will remain with a reasonable skepticism. To stay mentally fine, his solution is to not always bother whether he is going to live longer or not, but instead to live more in the present day. Of course, his daily life has drastically changed since the diagnosis and he does have some restrictions. “But you have to accept it to reach the best quality of life possible under those circumstances”, he comments. He even perceives an improvement in his life, since he retains more time for himself nowadays. Because the issue of time has become much more important, you have to use it meaningfully according to Bruno.

6.1.6 Josef Keusch - Hidradenitis suppurativa

Hidradenitis suppurativa (HS) – also referred as 'Acne inversa' – is a chronic and inflammatory disease of the sebaceous glands. It is marked through persistent and sometimes painful abscesses and knots (lumps) with a purulent content, mostly situated under the armpits and in the groins. Common are also cicatrizations and fistula tracts. The course of disease varies: Some barely suffer from complications, whereas with others, the disease constantly progresses, which leads to enormous pain and restricted mobility. Very little is known of this disease, so that the affected patients normally have a long life of suffering until a final diagnosis can be made. Unfortunately, the illness is mostly already in a severe state by then.[25]

It was 15 years ago when Josef Keusch (60) noticed first symptoms. He became an eczema sufferer, which got worse and worse. Two years later, he went to see a doctor for the first time. He had abscesses under the arms and at the left groin. The doctor recommended having it cut out, which he then had. It helped for a while, but then, the same problems started again. Today, Josef assures that it was the early stages and (psychological) cause of the disease which had yet to be diagnosed.
However, in October 2008, there was a rapid progression of the disease. He went to see his general practitioner, who could not say what it was either. Instead, he prescribed him antibiotics. Nonetheless, this did not help, so he went to see a dermatologist. He received Roaccutan® against the symptoms. But since it did not help either, he stopped taking it. Josef decided to find out by himself what the cause of all this stress was, and consequently began to research by typing in his symptoms in the search machine. Finally, Josef found out from what he suffered, and thus told the doctor. He was in extremely severe pain by then and started rejecting his own body mentally. It was a feeling as if he would rot from the inside out.

In January 2009, he got operated under the armpits once again, where 300 g of abscess was cut out on each side. As a wound treatment, he got a vacuum device under his arms for the suction of the wound fluid. The Spitex had to visit him every two days to change it, so that further infections and the sticking of the vacuum-foam-wound dressings could be prevented. But then, the Spitex stopped coming. The responsibility was too big. Josef tried to find someone else who would help him to take the device away, but he remained unsuccessful. So at the third day, one day more than it should be left by the wound, he went to the hospital so that they could change it. According to Josef, he was never in such an agony than at that time. After the hospital changed the vacuum-foamed material, he still couldn't find anybody who would help him for further changing. Yet, at the third day, he finally found a wound expert at the cantonal hospital of Baden who agreed to help him. Again, it was agony all over. The wound expert decided that it was time to try a new method for the wound treatment. Instead of the vacuum device, she used a bandage with gel, which had to be changed every two days. Henceforward, he constantly used this

[25] http://www.akne-inversa.de/

method, which turned out to be the best one. On March 2011, Josef could finally take off the bandage, forever.

However, in 2009, there were still abscesses in both his groins that also made problems, at first. Back then, the dermatologist who operated under his arms said that this time, he could not operate again. The intervention was too risky. Instead, he told Josef to look for someone else to operate, on his own. Nevertheless, Josef did not want any more surgery. He went to see a psychotherapist, with whom he could learn to find his own way of dealing with the disease. Together, they came to the conclusion that an inflammation means basically war in your body, and that you can link this with psychological aggressions. Josef was never an aggressive person. But now he knew that unconsciously, he was being aggressive. He noticed because of how he always tried to win and achieve what he wanted. So for Josef, it was clear that to stop the disease in progressing, he had to let go of these aggressions. To him, it was a really hard task to find these aggressions. Yet, on August 2010, the growth of the abscesses in his groin stabilized, and three to four months later, the extremely terrible pain that was caused by this had largely disappeared.

Josef is glad that he did not get operated on again and that he found another possibility to cure his illness. Now, he barely has any sufferings. He does not take any medication and does not go to any medical examinations. From his point of view, after all the rejection and incomprehension from the doctors' side, he has no more interest in visiting a doctor, and he has also lost trust in them. "I have never lost the faith that one can heal the illness oneself", he comments. For Josef, it is clear now that a disease is like a direction sign that wants to show us the right way to go. According to his opinion, his disease has broken out because of a character trait of his – his inner aggressions. The illness just wanted to point this out to him. Josef believes that to better your condition, you have to try to change your attitude, instead of walking away or putting it aside. You need to be attentive. "With 50 years, I started to become a person I like and can be proud of", Josef states. In his eyes, every destiny is related with a mission. For example, Josef's mission was to find his inner aggressions and confront them. Therefore, nobody really carries a punishment. In the end, it is what you do with it.

6.1.7 Walter and Itala Bucher – Huntington's disease

The Huntington's disease, also known as Chorea Huntington, is a neurological disorder, caused by a defect gene. This dominant gene is responsible that particular brain cells die off. People who inherit the defect gene are in every case affected. The risk to inherit the Huntington's disease from an affected parent is 50%. The Huntington gene was detected in 1993. Since then a genetic test is possible.
The sufferers are affected physically and psychologically. The disease is well known for the jerky reflexes that seem to appear like dancing moves. Other symptoms are agitation, difficulties with speaking and swallowing, personality changes that are associated with increased irritability, depressions, decreasing physical strength and social retreat. The disease usually breaks out between the ages of 35 to 50.[26]

The first symptoms were already noticeable 18 years ago. It started with memory problems, then with movement disorders, and much later after diagnosis, difficulties in swallowing. Itala's mother was also affected by this disease and committed suicide in Italy, where she jumped in front of a train. The suicide rate of Huntington patients is higher than normal because of the psychological ballast and guilt feelings when you have got children.

In the year 1995, Itala (63) decided to do a genetic testing. She went to see a recommended doctor, who then told her that he will not let her do the test unless she arranged a good social environment. It was important for her, so that in case the test turned out to be positive, Itala would have supportive friends around here. Since she only had colleagues from work, she contacted an old school friend and a former neighbor. Slowly but surely, the relationship was rebuilt. They informed themselves about the disease and from then on, became very close friends again. After she built up her social environment, Itala did the test. The result came after two months and, as expected, was positive. Since the result was clear, everything got easier, because they knew with what they were dealing with. Itala and Walter (66), her husband, wrote information letters to everyone they knew: friends, neighbors, colleagues from the working place and so on. They were not intending to hide and wanted to be able to speak openly about the situation. "The openness is very important", Walter states. Yet, the beginning after the diagnosis was not easy for the family. Walter barely knew about the Huntington's disease. Therefore, he did not know how to handle his wife's illness at first. He only knew that her mother had suffered from it, and that it is incurable. However, his wife stayed strong and did not let herself down because of it. "Now, we will make it", Itala said determined. And so they did.

Itala and Walter immediately started to do things they always wanted to do, like traveling through the USA. Itala also finished the New York marathon twice, the second time together with her son Reto, even though she already had the first symptoms of the disease.

[26] http://shv.ch/de/huntington-krankheit

Sometimes, Itala got angry out of nowhere. Until she realized that it was unnecessary, and that everything is okay. Nevertheless, both took it as it came and learned not to plan too much in advance. “We take it step by step and take every day as it is”, Walter comments.

Itala’s disease has progressed a lot by now. Walter and Itala have made a lot of new friendships since they joined the SHV *(Schweizerische Huntington Vereinigung)* about 16 years ago. He says that without the illness, they would have never met so many new people. In his eyes, it has helped them a lot. “You know what you’re talking about”, he says. When they joined first, the association gave them a lot of information about the disease and therefore helped them to become prepared. They now have an active collaboration with the SHV.
Doing something together as a group, for example going out for dinner, is great for Itala because she can be around people who are like her. They can laugh together and don’t have to be embarrassed when their forks fall to the ground. For the affected members, it is naturally disagreeable being stared at by unknown people.

Itala is lucky that she does not need to take any medicine unlike other Huntington patients. Except for Rudolac, which helps with the action of the bowels. Many therapies for Huntington affected exist, for example a regular visit in the “Memory clinic”, breathing therapy, occupational therapy, music therapy, hippotherapy and physiotherapy. Itala has been to the “Memory clinic” for a very long period of time, which has helped her a lot. Unfortunately, she had to stop because the clinic believed that the therapy didn’t help her anymore. Now, she goes to breathing therapy, since suffocation is a great factor in HD deaths. She also visits occupational therapy and goes to a daycare center three times a week, where she does additional physiotherapy. Walter says that it is good for him when Itala can be at the daycare for some time, because he also needs some free time every now and then. Unfortunately, the communication between Walter and Itala has become difficult and limited lately. Walter comments that occasionally, you notice how she really wants to tell something, but just can’t anymore. What is left are yes or no answers. Nonetheless, Walter affirms that after 40 years of marriage, you get to know what your partner wants to say. You notice with the help of her reactions and facial expression.

Itala was fortunate to have a very comprehensive boss. She has worked at ‘Tourasia’ and was allowed to stay there for a long time, even after the symptoms got severe. She tried to be as helpful as possible and was given all tasks she could manage. Also, her sons, who fortunately are not affected by the Huntington’s disease, are very supportive. From the rest of the family, they don’t hear very often. To Walter, it is clear that he will stay and look after his wife, at least for the next few years. For many, it is self-evident to stay with their partner after symptoms get worse. Still, he does know couples where this is not the case.

“At the moment, everything is fine”, Walter says. From his point of view, an illness has not only got negative sides. After the diagnosis, their personal values have changed. For example, the smallest things – like little gestures – already makes them very happy. All in all, they have become more modest. Objects or matters that have bothered before suddenly become irrelevant. And small worries are of no major importance anymore. You start living more consciously. Altogether, both are very happy: “You can’t determine your destiny. Therefore, it is important what you do out of it”.

6.1.8 David and Heidy Moser – Huntington's disease

Heidy (49) and her father both sit at the garden table, preoccupied with themselves. Both suffer from Huntington's disease. It is a sad picture for a first impression. But fortunately, I get the chance to speak to Heidy. However, the communication is limited and has gotten harder, even for her husband David (50), since she is practically only able to answer with yes or no. Complex questions are too hard for her to answer, but she tries hard to express her thoughts. Sometimes, she suddenly tells a story that has nothing to do with what she was saying before. She is trying to share her remaining memories with us, and so tells us what she thought at that moment. She can only concentrate on herself, and not even with her husband or anyone else. The disease has progressed too much to allow that. When she doesn't do that, she listens to audio books, music or watches TV. After the interview, she shows me her drawings, which are beautiful abstract pictures and even gives me one to take home. It is a picture with lines of red, orange and yellow flowed into each other by the help of the brush and water. She tells me that she likes to paint. Unfortunately, nobody knows how much time will pass until this hobby gets impossible…

After the genetic testing Heidy did in the year 1996 was positive, it was very hard for Heidy and David. Both were informed about this rare illness, though at that time, knowledge about it was not too big. They got married in the year 1989, and David, who learned about the disease shortly afterward, knew that there was a possibility that she could have inherited it. She has inherited it from her father. Still, their attitude changed and they enjoyed life more. Both loved to travel. There was no real change in daily life at first, until the first symptoms started about ten years ago. Certain activities like writing or stirring an egg in a pan became difficult and eventually impossible. Later, walking got more and more difficult, and Heidy's psychological capacity seemed to decrease. For example, if something went wrong during a registration, Heidy got angry and wanted to cancel it immediately. Back then, she had depressions and also had to take antidepressants. What has helped her a lot was joining the association. Heidy even became president of it. She made a lot of new friendships there and got the feeling to be understood. Also, the environment is generally comprehensive. Now, they go to the meetings four times a year. Nevertheless, she has never been excluded. Heidy herself believes that she is handling the situation well. She is being treated by a neurologist and goes to the rehabilitation facility in Zurzach for logopedics, massage, music and painting therapy, and to dance salsa. In addition, she needs to take sleeping pills and medicine to lessen the jerky movements.

Heidy's family supports her and handles the situation well, especially her mother. Heidy and David moved back to Oberhasli into their house about 10 years ago. Now, they live door to door, so that Heidy's mother is always within reach. In that way, she can care for both of them. Indeed, she is so supportive and helpful that David says that he wouldn't know what to do without her. And that is what sometimes frightens David. What will happen if Heidy's mother will not be around anymore? He works full time and wouldn't have the time or the nerves to look after his wife. For David, the disease is not such an

easy task to process and it is tiring. Yet, Heidy says that David is very supportive, but he believes that he could do better. He even feels guilty sometimes because of it. He was president of the association for ten years, but will stop it now because he's always gone and has no time left. In David's eyes, life is too short and that's why he tries to enjoy it a lot. He tries not to think what could be in ten years. He says that there are other priorities now. Yet, the only time when he is really happy is when he flies.
Heidy and David try to do best out of every day and appreciate life. On the whole, they are happy and content.

6.1.9 Hannelore Wälchli – Takayasu's arteritis

She already noticed it long ago as an adolescent when she went jogging. She was often dizzy and had dizzy spells. But it was 29 years ago when it really started to get serious. Hanni (64) says that she had shreds in front of her eyes and did not see so clearly. She went to see an eye specialist who then measured her blood pressure. And it was a big surprise when the blood pressure was extremely high: 270/190 mmHg (around 115/70 mmHg is normal). Very strange that three to four months earlier, it was very low. Normally, she was a person with rather low blood pressure. The eye specialist sent her to the intensive care unit in Aarau, where she stayed a while and then was sent back home with medication for very high blood pressure. The doctors did notice a strange sound at her carotid arteries. Yet, they could not tell what it was. But just a little time later, the head physician called her up and demanded that she should go to the Inselspital in Bern. One week later, in March 1983, after examinations of the vessels and X-rays, she received the diagnosis Takayasu's arteritis. Her doctor told her that at first, he didn't know at all what it could be, so he looked up on books every evening to find it out.

After a discussion, the doctors refused to give her cortisone, an all day medication nowadays. They were skeptical about the side-effects, which were already very well-known at that time. Instead, Hanni got in total 14 different tablets (five different types of tablets) that she had to take in regularly: six drugs against the high blood pressure, one beta-blocker, two drainage aids, three blood dilutors and two potassium tablets. Hanni didn't know what this disease was, she has never heard of it before. After the diagnosis, Hanni's doctor told her that this disease is so rare that practically nobody has it. That is why she felt very lonely and abandoned at first, and her nerves were on edge. She could not talk to anyone about it, even the doctors did not know much about it. The first years were hard for her. At first, she was a little depressive and asked herself why she had to take so many medicaments. She was glad of course that she was alive, as consequently they found out that in a few days she would have been be dead. However she could not stop thinking: "I don't want this disease!" Later then, she was able to accept it and thought: "I have no other choice, what else can I do?". Nevertheless, if someone had asked her at that time if she was confident, she would not have known how to answer.
Eight years after the diagnosis, doctors wanted to dilate the vessels, which were still very narrow. So they decided to do an angioplasty. It means, with supervision of the medical ultrasonography, you enter the vessels with a balloon catheter, which start to dilate where the vessels are narrowed. After this intervention, Hanni believes that everything started to become better.

Even with her illness, Hannelore never stopped working as a commercial employee. Her colleagues and her boss knew about her illness, and fortunately were very understanding. Her family was shocked after the diagnosis, especially her mother, who needed medical treatment at first. She had and still has a lot of support from her family and has never lost any friends because of it. Everything remained the same. In her environment everyone treated her same as before, telling her to 'Cheer up!'. And after a

while, they forgot about it anyway. She has never been excluded from society and mostly felt very normal.

Even though Hanni still cannot feel her own pulse on her wrist and has vascular narrowing in the arms, the legs and the aortic arch, she does not have any more symptoms or troubles of the disease. Headaches, which she often had before, are gone. And if it were not for the few drugs she has to take, luckily many less than before, she would forget that she is sick. She thinks that the disease has in the meantime become inactive, since there have been no further changes. Every year she needs to run blood test and all three years she goes for an ultrasonic examination. Now, she lives a perfectly normal life with almost no restrictions, except swimming and strength-weight training difficulties. Her attitude has not changed much, except for the fact that she is glad that her life has been saved. And after her diagnosis, she appreciates her life more than ever. She believes to see things more realistic now and in an easier way. She doesn't think a lot about the sickness. "Everything has turned to good account", she states happily. She takes every day as it comes, and is confident about the future.

6.1.10 Agatha Meier – Scleroderma

Scleroderma is a chronic autoimmune disease. Consequences of this incurable illness are swelling and hardening of the connective tissue, mucosa, skin, arteries and inner organs.[27]

In May 2008, cardiac arrhythmia was noticed in Agatha Meier (57), and the pressure between the lungs and the heart was too high. She had difficulties walking and her fingers were nearly white, so they inspected them more closely under the microscope. The diagnosis was found out quickly, she had scleroderma. After further examinations, they could see that her arteries, heart, kidneys and her digestive tract were affected, they were hard and that she had water on the lungs. Agatha receives intensive drug therapy for this chronic condition which also requires painkillers. In addition, she tries out medicine that still is in its testing phase. She goes to therapy for the whole body and for her fingers, which helps a lot. She has always very painful episodes again and again, with muscle pain. She says that if you survive, after six years the illness normally stands still. It was a huge change in her life and she had to give up a lot. Agatha says that she has always cold, even in the summer. And in winter, it gets really bad, does not matter how much clothes she puts on. She often massages her hands when they get stiff. It is very time-consuming, but helps a lot. She is always thirsty, but she cannot drink so often because it hurts. She was also restricted in eating certain foods. And she cannot do any physical effort, so she had to stop working on the field as a farmer.

Agatha had never heard of this sickness before. Four to five times a year, she goes to the University Hospital in Zurich for examinations. The doctors presume that she has already had the disease for a long time. The first year after the diagnosis was a catastrophe for Agatha. “This can’t be happening. Why me? What did I do wrong? Why did I get this?”, she constantly repeated and gave herself the blame. She could not believe that she really had this, and that was very hard. She felt helpless and blocked it out. “It takes time until you really believe it, until you accept it, what I finally did now”, she says. She says that she is lucky that her doctors always inform her well and give her good advice. Unfortunately, her brothers and sisters stay away from her since then, but the rest of her family is being very supportive. They handle the situation very well and always tell her: “Tell us if you are tired!”. She appreciates their regard a lot. They are very understanding and she can even talk with them about it. In the beginning, she was partially excluded from other people, but not anymore. She has lost some friends, but has gained a lot of new ones in the association she has joined now. There, you talk differently about the disease, because everyone knows it. You can talk openly about the disease. Sometimes, she even calls someone from the group up, just to talk.

Agatha appreciates her life more now: “It is not self-evident at all to be healthy”. She is able to handle her situation much better than before, by always telling herself to calm down. “Letting the head down doesn’t help. I live with it now and make the best out of it!”, she says. Agatha takes every day as it comes. If you have a bad day once, then you

[27] http://www.sclerodermie.ch/

just have it. She takes time for herself and doesn't feel so helpless like in the beginning. She also sees herself as lucky because her lungs are not affected. In her eyes, it would probably be much worse then. Agatha believes that she has changed as person. She enjoys life more than before and tries to make best of her situation. Little things that Agatha didn't seem important suddenly become positive. And as for the future, she is confident: "Hope dies last!"

6.1.11 Anna V. * – Systemic lupus erythematosus

Systemic lupus erythematosus (SLE) is an inflammatory autoimmune disease that attacks the connective tissue of diverse organs. Depending on which organs are affected, the disease can have lower or higher severity level. Mostly, the sufferer has to go through several episodes. Women are more affected than men. The course of disease is very individual. General symptoms are weight gain or loss, fever, fatigue, pain and weakness. Unfortunately, the illness is mostly diagnosed very late. [28]

It all started with a trip to India between January and February 2010. After the first days, Anna (29) suddenly caught a fever and breathing problems. She decided to see a doctor for tourists, who then told her that she had no problem and that everything was all right. But after lying in her bed in the hotel for five days, with fever, sweat and nightmares, her boyfriend called up REGA. They in turn told him that she has to go to a hospital there immediately. But as she stayed there for a while, the hospital in turn informed REGA that everything was okay. Staying in India for such a long time was a big mistake. After the flight from Delhi to London, Anna was barely able to walk. Back home, she went to see her general practitioner right away. And he told her that she has taken too many medicaments there and that it will get better if she returned home to sleep. But after a few days, her doctor called her up, telling her that something was not good. Anna decided to go to the father of a friend of hers, who is an oncologist. He noticed that all her lymph nodes were swollen and told her that she instantly needed to do a CT. It showed that she had water in the lungs, but nothing more was clear. Water from the lungs was taken to see if it was any infection, but it was not. They could not tell her what it exactly was, only that it was something inflammatory. Because Biel found no proper diagnosis after ten days, Anna was sent back home. Nevertheless, she still did not feel good and suffered from fever and pain. She had to miss her courses at university for more than a month and in conclusion change the test dates for the next semester. It was tedious.

In April 2010, she went back to her courses at university, still feeling bad. And in June, she had to go to hospital in Winthertur, due to strong pains, and stay there. After three weeks, she was dismissed and afterwards, she attended a rehabilitation center in Bulgaria where her condition improved. They told her that it seems that she has some problem with her heart, but it could also be nothing. During her visit to the Black Sea, she started to have strong pain again and took strong painkillers. When she came back – it was September 2010 – it still did not get better. She claimed to be in a strange condition, being in constant weariness. In the middle of September, her legs and eyes got swollen. She started doing urine samples by herself, noticing that the protein in the urine was very high. She constantly told herself that the next day, it will get better. But after a while, since that did not happen, she wrote an e-mail to the head physician, describing the symptoms. He answered back that she had to go to hospital right away. After hesitating, she went and with a urine sample, they found out that she had 23 gram of protein in the urine, normal would be 0 gram. She got cortisone and Endoxan® at once. Nevertheless,

[28] http://www.slev.ch/index.php?id=0:130:public/lupus

Anna still didn't get really better. In one week she gained 10 kilos due to the cortisone. She started to have enough of hospitals, though they were looking after her. Two days later she had blood in the urine and a kidney specialist came. Everything was under control, and Anna had to stay in hospital for ten days.

Since August 2011, Anna feels physically and psychologically better, what partly could be thanks to the medicine. She is thankful to be in good and trustworthy hands. Twice a month, she goes to hospital for blood tests, once a month for a general checkup and just recently started with the medicine MabThera®, which seems to work out well. At first, she couldn't believe or accept that she had Lupus. It was very difficult in the first place, since it took so long to finally be sure if it was Lupus. Now, at least she knows that the organs are affected. This fight against the disease was like a vicious circle: Because of the illness, she felt bad mentally. And that again affected her physically and so on. The whole time, she has just felt weary. It was a hard time. There were phases where it seemed to get better, and then suddenly, she was in pain again. She did not see an end… After Anna was told to have SLE, she started researching. But in her opinion, too much research is bad for the psyche. "You get crazy", she says. "But the psyche is very important, and you have to try to stay positive. This is of course very difficult…"
A few years before, her GP found many antibodies. In the year 2005, she was already tested for Lupus. But at that time, it was very unclear. She has a general bad perfusion and sometimes white fingers (Raynaud syndrome), the reason why she was tested for lupus. At first, the doctors had thought that Anna suffered from MCTD (mixed connective tissue disease). This wrong diagnosis was a reason why she did not trust doctors at first, and why she tried to avoid hospital whenever she could.
Anna doesn't know anyone with this illness. But in her opinion, the support of the family is much more important. She has got the full support from her husband and her mother, even though her mother lives in Bulgaria. The rest of the family doesn't know the whole truth. She believes that support is very important when you are sick. Her friends always give her tips. She does not want to start working until she fully recovers. And she wishes to have children later, but she can't have at the moment because of the medication. She is not restricted and is still able to play the violin at concerts. As a musician, she has to be careful with whom she talks about it, because if the wrong person gets to know it, it could be that she would be excluded due to the illness.

The disease did change Anna. She tries to stay normal and observant, and very important, to not stress herself! Before, she often felt stressed and put herself under pressure. Now, she takes things more easily and doesn't put herself under pressure. And stress has a strong influence in life. Due to this change of thinking, Anna has become more realistic. She has to rethink about her future, what to do, and she thinks a lot about it. She knows that something could always happen, but she still sees the future in a positive way. Because she was able to accept her disease, she feels much better now. Furthermore, she is very thankful for the support of her husband and family, relieved that the Inselspital is near and appreciates health very much. "If you want, you can do everything!, she believes.

6.1.12 Monica W. * – Phenylketonuria

Phenylketonuria (PKU) is a metabolic disorder, caused by the gene defect of an enzyme. The affected gene is inherited recessive. Subsequently, the amino acid phenylalanine (Phe) cannot be metabolized into tyrosine. Therefore, if the disease is not recognized in the first few weeks of life, the concentration of Phe in the blood rises to a dangerously high level. This can lead to irreversible mental and physical damage and after a while even to death. To prevent this, a protein-free or protein-low diet needs to be followed strictly. If the diet is maintained, the concerned person can live a practically normal life.[29]

Monica (28) is like any other person. She does all things any other normal person would do. But if you were invited for dinner at her place, you would notice one difference: the food. Monica has been on a special diet since she was born. Because she cannot eat any proteins, she never tasted meat, fish, cheese, milk or other products containing too many proteins. Except once when she was younger, her mother told her that it was okay if she tasted a bit of pizza. But after she did it, she felt psychologically bad. She felt guilty, as if what she did was intolerable. Probably, Monica felt like that because she knew the effects it would have if she took too much of this pizza containing cheese. But Monica was never scared because of her illness. She claims to have no restrictions whatsoever apart from the strict diet. Of course, every now and then, she wished to be also normal, like everyone else. But then again, she doesn't know anything else, since she grew up with this sickness. "The disease just belongs to me", she says.

In her eyes, Monica was very lucky. Her parents enabled her a lot. They took great care of her diet, always trying to get the best out of what was allowed for her. When Monica had a new teacher, the first thing her parents did was talk to him or her and make their daughter's situation clear. And every year, it was a pleasure for them to send their daughter to the camp for children and adolescents with phenylketonuria. Her mother has been a member of the CHIP ("*Schweizerische Interessengemeinschaft, Phenylketonurie und andere mit Eiweisseinschränkungen behandelten Stoffwechselstörungen"*) since 1988, which organizes the "cooking house", a youth club and the camp. Now, Monica is in the executive committee herself. She finds it great to be a part of it. Monica has met many new people of "her kind" through the camp, people who she even eventually met privately. It is a camp for children and teenagers, where they can meet once in a year to have fun and be together with the "same" (see chapter 6.4.1). The camp wants to take away the fear from parents and their affected children. There, you have got the feeling that you are not alone. Especially thanks to the camp, she has never been excluded. There, you rather indirectly learn how to deal with your disease, also when it comes to get an intuition of how much of a certain product you are allowed to eat and start measuring it by feeling.

Till today, Monica never had a serious problem. "It all depends from the support and education you get at home!", she says. There are parents who distance their affected

[29] http://www.chip-pku.ch/pages/pku---ein-ueberblick.php

children from the “outside world”. They do not send them to any camps, even though it would help both. Sometimes, they fear that they could eat something forbidden, or even out of laziness to prepare food for a whole week. Because of that unfortunately, some children cannot deal with their disease. That is why Monica appreciates her parents’ support and openness so much. Also, she is very thankful for her boyfriend, who never really had any problems with her sickness. He has always been very thoughtful, especially when it came to planning vacation, since they need to know where she could eat. She also gets great support from her office. She only told someone when she thought it was important for that person to know. When it came to telling her colleagues or other people, there never seemed to be a problem. At first, the people she tells are curious or show sympathy, then they offer their help, and after a while, the people even start to forget that she has phenylketonuria, since it is not obvious. For Monica, this is really positive!

Phenylketonuria affected people need to take in a phenyl-free powder. It is a protein alternative, which they need to take from 2x till 5x a day, depending on how much you need. Monica affirms that it is something very normal for phenylketonuria affected. To her, it has become routine and daily life, it just belongs to it. Once in a year, she does a blood examination and a checkup, and afterwards talks with the doctor if everything is all right. Monica admits that once in a while, she cheats on her diet. And when she does, she notices it mentally by being forgetful and impatient, lacking concentration and suffering from high blood pressure. That is why she does not like to do it.

Monica handles her disease very well. Of course she had phases where she was irritated, but that is self-evident. A decision you already need to do as child is: Do I make my life difficult or do I accept it and enjoy life? In her opinion, you just need to organize and plan ahead to live easily with this sickness. For example, Monica wishes to have children one day. To be safe during pregnancy, you need to maintain your diet very strictly. If you do not, there is a risk that the child becomes handicapped. You need to plan ahead!
In her life, food is the center point. There are times when she is in a grocery and sees all this different variations of food. At that moment, she would like to try it. However, she stays optimistic. There are times where she is happy and thankful that she “only” has that. Monica counts herself lucky and is very confident of the future, as she has many plans she wants to fulfill, like traveling or having children. She is very content with her life and lets nothing stop her.

6.1.13 Hans-Rudolf Wenger – Thymoma

When a person suffers from a thymoma, malignant cells form on the outside surface of the thymus. The thymus is a small organ that lies in the upper chest under the breastbone and is part of the lymph system. It is responsible for the production of white blood cells (lymphocytes), which should protect the body against infections. The types of tumors of the thymus can vary; thymoma is one of the very rare cases. Symptoms of a thymoma can be constant coughing, chest pain and trouble with breathing. [30]

When Hans-Rudolf (61) looks back, it seems to him as if he already had symptoms ten years ago. But his first verifiable symptoms started in 2006. He had a skin rash, bubbles on the mucosa, consequently diarrhea and costiveness, and a visual impairment. After a while, he went to his general practitioner to clarify what this all was. Unfortunately, the GP did not know either and so sent him to a dermatologist. But the dermatologist couldn't tell either, and Hans went back to his GP. Finally, he was sent to the Inselspital in Berne, where they detected the problem: He had a malignant tumor by the thymus – a thymoma.

In the year 2009, Hans was operated. The tumor and infected tissue around it was taken out. But after the operation, the symptoms did not stop. When he returned to the Inselspital, the doctors told him that his changed immune system was attacking his mucosae. He got cortisone against the inflammation of the mucosae, which caused as a side effect bone atrophy. So, to suppress his immune system, the doctors gave him Imurek®. However, afterwards, his immune system became even weaker. As a consequence, Hans had to deal with three pneumonias, herpes zoster, mycosis on the lungs and in the esophagus and his nails started to dissipate. Hans could not comprehend why his immune system suddenly became so weak. He remembers always having a strong immune system, enough strong to not catch any cold after swimming in the lake in the middle of winter. Now, it is a trial and error with the medication: How much can he bear to not have all this side effects and at the same time to obtain an effect?
Every month, Hans has one appointment for a general checkup and two to three other appointments with specialists of oncology, pneumology and gastrology. There are big conferences and little experience of the doctors, and so a constant experiment. It is a constant checkup of his state of health, giving him more, less or other tablets. However, Hans does not really care about it.

Hans remembers his first reaction when he got his diagnosis: "I thought that you have to take it as it comes, since you can't do anything about it". He had never heard of this disease before, and his doctors partly neither. He remembers how once 14 doctors were standing around his bed. He went to look up on the Internet himself. He didn't care that it was so rare and that he didn't know anyone else with the same problem, and he didn't feel alone. Luckily, he always became good support from his family and friends, even though his wife sometimes worries. Hans asserts that his attitude, which has always been

[30] http://my.clevelandclinic.org/disorders/thymoma/hic_thymoma_and_thymic_carcinoma.aspx

very calm, has still stayed the same and that he doesn't think too much about the future. "My momentary situation is what it is, what can I do?", is Hans response to the question what he thinks of his situation now.

Because of all his appointments and problems, Hans cannot work 100% as a business data processing specialist anymore. He had to reassign his work position and therefore, his job-related environment has changed. Because he only works 50% now, he is not informed about everything anymore and thereby is being excluded a little. This was one big change for Hans. At first, his main goal was to be successful and get big projects. Now, he just wants to enjoy life and look that everything is fine NOW. Thus he has become more spontaneous. He recounted how he recently went to an artist's workshop: "Before, I would have never gone to something like that because it would have been 'lost time' ". Now, he does. Hans is generally content. "Though it would be nice to be 20 years younger", he says laughing. The symptoms he had before the operation have all vanished, but the side effects of the medicine are bad. He is thinking about a retirement or an IV-rent (invalidity insurance). There is no prognosis for his illness, but he wants to see what the future brings him. "What will be, will be", is Hans' opinion. His only wish is that his health stays as it is now and doesn't get worse.

6.1.14 Laura M. * – A rare rheumatic disease

In February 2005, Laura (30) went to India. She was in the midst of her studies at university. One and a half weeks later after she came back from her trip, she felt strange. She felt hot and tired. In a first visit at the general practitioner‘s, she was diagnosed with a simple cold. Laura was relieved and regenerated. But six months later, the horror really started. Laura felt bad again, but this time she had sort of a high fever in addition, which lasted for four days. The first thing that came into Laura’s mind was that she could have been infected with malaria. Even though blood tests proved her wrong, her CRP and folic acid values were extremely high, and she had increased liver values. However, the doctor did not think that it was a symptomatology for an illness, and therefore did not investigate further.

Laura stayed tired throughout the whole fall and even started asking herself if she was not just lazy. But why was it so itchy?
Before Christmas, it started to get worse. She had a severe headache and felt very bad, as if she had the flu. And then suddenly, on 30 of December, as she was arranging her documents, she got a sort of terrible fever, which got worse and worse. The next day, she was so weak that her friend brought her to an emergency doctor. But after an examination, he said that it was just a severe cold. Yet, on the next day, she started vomiting and felt awful, and additionally lived through episodes of high spiking fever again. And then, her face appeared jaundice. She thought that she was going crazy. So Laura was brought to the emergency of a cantonal hospital where she lived back then. The people at the hospital emergency started laughing at her because they were wondering why she came to the emergency for simple symptoms of a flu. Subsequently, the doctor there only scanned her abdomen and did not do blood examinations. Laura’s friend brought her to another emergency doctor in a smaller hospital then. There, she stayed for two weeks with constant episodes of very high fever around 41°C during eighteen hours every day. Yet, in her condition, she should have been sent to a bigger hospital. Unfortunately, they did not do anything at all! Finally, she was hospitalized at a University Hospital. After two and a half days, they brought her into intensive care. Her state was critical, 20 doctors were studying this strange case, and after seven days without finding anything, Laura was put under quarantine. Finally, one doctor came to the idea that Laura could suffer from a rare rheumatic disease which is known to produce high spiking fevers in a chronic condition. And after several examinations, this idea proved to be right. Laura was at the end of her strength: Not only was she vomiting and in an extremely bad shape – she was also not able to move her joints anymore, for the reason that it hurt so much.

After a while of recuperation, Laura was sent home. But she noticed that she could not live alone anymore. The option was to move to her parents, what she finally did. For the next five years she lived in pain and constant fever episodes. Her daily plan was to endure the fever. She was at the hospital most of the time, and it seemed as if she were living there. It was a back and forth from her home to the hospital. In addition, it seemed

as if she was resistant against classical antirheumatic medication. Her life was chaotic. In the hospital, she was not able to move at all, she could not even ring the bell. Breathing was painful, she could barely walk, and everything hurt.

It took one year to get the new medicine. However, this time, it really helped! She was able to move again without having constant pain. Nevertheless, the fever did not go away fully, it just got a little better. Then the high fever episodes restarted in their hellish way again: Once overnight she got a 41° C fever. She could not ask her parents to help her, since they were not on friendly terms with her. She carried herself to the phone and called a taxi that brought her to emergency, where she barely could walk inside. The next morning, the doctors told her that for two hours, they were not sure if she would make it. Her life went on with more or less fever episodes, which she bravely endured. “It was a miracle that I survived this and that I was released”, Laura comments.

One day, Laura looked at her agenda, which was full of appointments with doctors. She decided to cancel all appointments and do something for herself. She invested her last energy to go clubbing. The problem was that she did not know anyone in the bigger town where she lived now. She was completely isolated. Still, she went out regularly. And even if she often had to go back to emergency, Laura felt that it had been worth it. After a while, she started to feel better. Her parents though were no support at all. They did not help her during that time. One night, they fought very much, and the parents decided to throw her out of the house. As a consequence, Laura had to look for a new home. She then found a temporary stay. Laura could not actually work: On one side because of her fragile health condition, on the other side because of her disability pension. Theoretically, she was not even allowed to study because of the invalidity degree she had gotten from the disability insurance (IV). Consequently, Laura started working off the books in order to pay her studies and all the medical extras, which come along when one is not really healthy. In that way, she started to rebuild her life again and was able to survive. It was hard and exhausting due to her illness. But Laura needed to go through this, all alone. She was working in her field or working for studies permanently. Nobody knew of her disease. She practically did not have help from anyone. “I pulled myself out of this on my own”, is Laura’s opinion. She was not able to hold friendships because she lived so far away from her old home. Rarely did anyone visit her or did she receive phone calls or text messages during hospital stays. And rarely somebody offered her help or money.
After a while, the effect of that first drug she had tried decreased. She changed to another one, which helped miraculously. The fever stopped, and Laura started to feel extremely better and healthier.

Laura’s attitude has totally changed by now. She has become a fully new person due to the disease. In all that time where she was able to think, she never lost hope. She believes to value things others would never do, like paying the bills on her own. Laura is fine and happy now. She feels healthy, capable and full of potential. Nonetheless, she has got a new problem at the moment: the integration back into society. She has lost so much time

and friendships. But she is confident that everything will turn out fine. Laura is so thankful to be healthy again. She believes that now, after having survived those terrible years, she still has got her whole life before her and so much that she has to catch up, like having a family, marrying, staying healthy and so on.
She has been at the lowest point in her life when the doctor told her to not give up. So she forced herself to fight. The pain back then was unimaginable: Like a hammer that hit on her joints over and over again. It was hell to put a finer point on it. However, she came to the point where she thought: “Now I need to make a decision if I want to fight or give up.” She decided to live. With this attitude, she was able to survive.
“Those were five dark years. I can’t really remember that time and I guess I am traumatized by it”, she says. Only thanks to her job off the books and the contact to at least some people there at work, Laura was able to survive. “If I had had people at that time who would have helped me rebuild my life, things would have gone faster”. But she did not really have anyone and was isolated. In addition, she has no contact to her family anymore. Laura finished university and got a degree, even though it took so much longer due to her illness and her work.

6.1.15 Seraina Wäspi – Cushing's syndrome

Cushing's syndrome is a hormone disorder caused by too much cortisol in the blood. Cortisol is a corticosteroid hormone that is produced by the adrenal gland. There are several reasons for a high cortisol level in the blood: First, it could be a consequence of glucocorticoid drugs (e.g. prednisone). Yet, the Cushing's disease itself is described by a tumor (adenoma) on the pituitary or adrenal gland. The symptoms can vary, for example moon face, weight gain, muscle weakening, high blood pressure and many more.[31]

When Seraina (15) was told she had a benign tumor (good tumor), which caused a cushing's syndrome, she was quite surprised. Never had she dreamed to get something like this (then it was naturally for her to be healthy). She had not noticed anything, except that she had gained a bit of weight and bloated a bit, especially during the bathing season in the summer 2009 (half moon face). Also, she got worse in sports lessons, and she in her opinion, was already bad. What she did notice was that she had become truly better in school, was able to concentrate more during lessons and her grades started to improve. But never would she have related it with too much cortisol in the blood. However, the person who did notice was Seraina's mother. Seraina, who normally was a thin girl, suddenly bloated up for no particular reason. Puberty, you could say. But her mother could not let go and knew that something was wrong, and so she brought her to the pediatrician. After a urine and saliva examination, the doctor said that she had too much cortisol, and told her that she had to go to the children's hospital in Zurich. Another urine test was taken with the same conclusion. Finally, they were able to make a diagnosis, shortly before Christmas 2009. They found out the reason why she had too much cortisol, which was the tumor in her head on the pituitary gland. The doctors told her that she had the cushing's syndrome, a rare illness which is even rarer too see by children. Seraina was shocked at first: "If this disease is so rare, why me?", she asked herself. But since she did not feel bad at all, she started to block it out quickly. Instead, she was sad when the doctor told her that she would not be able to go to the ski camp this winter, due to the operation. Especially her family was shocked and worried. Yet, their support was great. Also, friends and teachers visited or called her in hospital, or sent her flowers and cards. But Seraina stayed calm, since everything was fine and she was feeling well. Furthermore, she was never excluded back then. The only annoying issue was the load of sympathy.
Since Seraina did not know anything about this sickness, she started to research a little. But her mother didn't want her to look up 'tumor', out of the reason that what you would find then are horrible things. She was afraid that it would upset her daughter.

The only way to help Seraina was an operation. During the sports holidays in February 2010, she was operated. After the operation, Seraina felt strange. Not only was she thirsty like never before. Also her high motivation had disappeared. Instead, she felt a little down: The normal effects after an abrupt change from a lot of cortisone to none. Since then, the high cortisol-level effects have vanished forever. She has gotten back her

[31] http://csrf.net/

normal figure, and her motivation for school was gone, and she is normal again in school. Her daily routine has become normal again.

She had to take cortisone tablets after the operation, but since the adrenal gland started producing it again, she does not stand under medication anymore. Because of her illness, the physicians found out that she had a growth disturbance. She was very small at that time. Since then, she needs an injection every evening. Without the disease, they never would have found that out, and she would have stayed small forever. To her, this is at least something positive.

Looking back, Seraina would have never thought of the possibility that she could have died at that time, which could have been the case if they hadn't found out in time. She felt perfectly fine back then. And she does now, too. But in her opinion, she would feel a lot worse now if she had a throwback. She often looks back and thinks: "Seraina, you were lucky!". After the diagnosis, one important attitude towards life changed: "It is not self-evident to be healthy!". And she remembers the time where she had to take a lot of medicine, realizing that she was not healthy then. Now, she appreciates health a lot more than before and is happy that everything has turned out well.

6.2 Telephone interviews

6.2.1 Corentin Joye, 19 years, Kleine-Levin syndrome (KLS)

Corentin has been suffering from KLS since fall 2008. KLS (also Sleeping Beauty Syndrome) is a neurological disorder, characterized through phases of increased need for sleep (hypersomnia) and cognitive and behavioral disturbances (prevalence: 1-2/million individuals).[32]
Up to now, Corentin has experienced 7 episodes of hypersomnia, usually lasting 1-2 weeks, sleeping 20 hours per day. Since this disease is little known about, doctors don't know what to do. Nevertheless, according to Corentin, they assure that the illness should be gone around 30. To repress the illness a bit, he takes in medicine every day.

For Corentin, it was difficult to accept his situation at first. Yet, he has never been excluded. On the contrary, his family supported him more than ever, and his friends were rather interested to know what he had.
Except from that, there has not changed much since he was first diagnosed with KLS. As his disease is not lethal, his attitude has remained the same as before. He enjoys every day and appreciated life also before his illness. What did change was his point of view towards health. He estimates health much more now, especially in his normal phases. Knowing now that everybody could become affected from a disease, Corentin thinks he looks at things in a more realistic way. Now, he gives more attention to his needs and has become more cautious.

6.2.2 Chantal Kneuss, 36 years, Crohn's disease

13 years ago, Chantal was diagnosed with Crohn's disease, a chronic and inflammatory bowel disease (IBD), caused by a defective immune system (autoimmune disease). The whole gastrointestinal tract can be affected, reaching from the mouth down to the anus, including the esophagus, stomach and intestine. The consequences are diarrhea, vomiting, weight loss and abdominal pain.[33]

For Chantal, the pain was one of the worst symptoms. Luckily, her family always supported her, even though the situation was very difficult at the beginning. Still, she has lost some friends and not only that, but she has also lost her job due to her illness. According to her boss, she had too many absences – by reason of several hospitalizations and days of sickness.
Now, Chantal lives from the IV. But instead of being angry or sad, she is very happy. She has got much time now and can do things at her own pace. The psychological pressure owing to her old job is finally gone. Because of her disease and a close-to-death experience in account of a lung embolism, Chantal thinks she has learned a lot. Her way

[32] http://www.orpha.net/consor/cgi-bin/Disease_Search.php?lng=EN&data_id=10326&Disease_Disease_Search_diseaseGroup=Kleine-Levin-syndrome
[33] http://www.morbuscrohn.ch/mehr_verstehen/

of looking at life has totally changed; she has learned to appreciate life and especially health (her healthy moments) more, which she says is most important. She believes that, without this disease, she would have never learned to appreciate the little things; for example walking through the forest, enjoying nature and taking the time to breathe. Chantal has learned to listen and react to her body's needs. She lives consciously in the present and enjoys every second, because she never knows what could be (state of health) the following day.
In Chantal's eyes, health is not self-evident at all and can change very quickly, like luck. Also, she has become more realistic concerning reality, and how it really works when you are sick. "What doesn't kill us makes us stronger", Chantal believes. It makes us stronger to love life more, the present and yourself. Her mentality has changed much, and she does not live in the negative anymore.

6.2.3 Martin H. *, 39 years, Myalgic encephalomyelitis (ME)

Since July 2007, Martin is suffering from ME, also known as the chronic fatigue syndrome (CFS). ME is a neurological disorder characterized through a long-lasting fatigue, memory and concentration problems, disorientation, hypersensitivity towards noises and so on. Often, the sufferers are seen as malingerer, even though the disease is real and approved by the WHO (world health organization). Although the disease seems to be not rare, it frequently remains unrecognized. The illness drastically diminishes the quality of life.[34]
Martin is usually awake 4-6 hours a day. If he sleeps less, he feels insufficiently rested. During these periods, he eats, reads or watches TV. But he can only follow an activity 30-45 minutes; then, his concentration disappears and he needs a break. He had to give up his hobbies, driving and unfortunately, his job.

Martin doesn't value his life or health now at all, since he cannot live like he did before his disease. He often looks back and regrets not having his old life back. During one year, Martin was forced to take antidepressants, so that the chances for a depression were smaller. His friends avoided him more and more, leaving him alone. Only his parents stayed by his side, helping him especially with food (cooking and buying), the washing and household in general.
Martin looks at reality differently than before his disease. Especially at the difficulties some people have when someone is sick. His attitude towards the world has totally changed. Even after he has lost his job, the IV was not ready to help him, because the disease is not approved in Switzerland. He is surprised about this abandonment of the state insurance companies. He thinks it is very unfair. Now, he lives from the social service.

[34] Tion, Michaël Sam, 2010. *Le syndrome de fatigue chronique* (Brochure)

6.2.4 Kathrin H. *, 32 years, Galactosemia

Galactosemia is an inheritable metabolic disorder. Due to a missing enzyme (galactose-1-phosphate uridyl transferase), galactose can't be transformed into glucose. After birth, there are various examinations to test this disorder amongst others. If not immediately treated, the accumulation of galactose and galactose-1-phosphate can lead to mental and physical damage, and after a while even to death[35].

As Kathrin has suffered from galactosemia since birth and grew up with it, she never knew what it was like to be healthy. She is not allowed to eat any milk products that contain milk sugar (lactose). So she has to be on diet permanently and attentive to what she eats. As a child, she suffered from dyscalculia on account of the disease, a learning disability involving mathematical problems. Now, she has been happily married since August and works in a retirement home, instead of receiving from the IV, which she could. Kathrin confesses that she sometimes reaches her limits while working, physically or psychologically. She needs to take calcium and antidepressants. She thinks to have an addiction to depressions, but it is unsure if it is due to her illness or genetic disposition.

Kathrin believes to value her life and health a lot, especially since she was drawn in the conflict if she wants to live or not. She enjoys every day and is glad that she is fine. And because she guides a self-help group, Kathrin knows that not everyone has the luck she has. Also, her family, friends and now husband have supported her as far as she can remember. In her opinion, to be healthy (or unhealthy and happy) is not self-evident at all.

[35] http://www.galactosaemie.ch/index.php?site=galactosaemie

6.3 Experts

6.3.1 Esther Neiditsch, president of ProRaris

Esther Neiditsch has been president since ProRaris was founded in 2010. Thus, she has a lot of experience and knowledge when it comes to the field of rare diseases.
Furthermore, she has lost her eldest daughter who had a brain tumor (astrocytoma), another family member suffers from acromegaly.

First of all, Neiditsch wants to make clear that just as the rare diseases vary, the victims do to. The affected people are throughout different when it comes to the disease, severity level and their social situation. However, from Neiditsch's point of view, the rarity secures that everyone sticks together. It often happens that the sufferers feel alone because their disease is so rare. That is why patient organizations are crucial. They are very helpful for the reason that you can talk there about the disease with others who have got the same problems.
You can compare rare diseases with the example of the 'black swan' and 'white swan'. The 'black swan' is rare, absurd and tries to hide and can therefore be compared to people with a rare disease. The 'white swan' is the contrary and stands for the common diseases. Neiditsch wants to improve this situation with the help of ProRaris, by trying to turn the 'black swan' into a 'grey swan'.
Neiditsch comments that it is difficult to state if people who are affected by a rare disease are being more excluded from society. It mostly depends on how visible the disease is. Therefore, the reactions as well as the sympathy from other people vary a lot.
Depressions also can be caused by many factors. For example, it could be caused by the gravity of the disease or the disease itself, more precisely the clinical picture of the disease. Some other factors are the support you get when you are sick and the pressure you are under – like the boss or keeping the job.

Neiditsch has the opinion that attitudes of sufferers vary a lot. The new situation is hard and difficult to manage, especially when prognosis is death in a short time. Many think then: "What does it help to know about it? I can't change it anyway…". As you can see in rare cancers, the individual forms of the diseases often need to be treated specifically.

When you are told your diagnosis or when you are not diagnosed at all, it is "as if you are on an island", Neiditsch asserts. However, the first reactions vary. It could happen that some immediately start to deny their state of health and others who are shocked. Yet, many are extremely relieved about the news, because they finally know from what they suffer. In Neiditsch's eyes, the problem is that people who are not yet diagnosed are often looked as a malingerer.
Anyhow, the long-term response can differ from cases where the affected person starts to live more in the present to others who constantly moan about their situation. In conclusion, it is a whole personal process the concerned people need to go through.

In addition, life gets extremely difficult for the parents or family who need to care for an affected child of family member. The burden of caring for a person around the clock can quickly lead to isolation from society. Neiditsch believes that family members of a sick person often experience a similar fate to the one of the patient himself. Therefore, they are also concerned.

6.3.2 Dr. med. Pierre Krayenbühl, internal specialist at the USZ

Dr. Pierre Krayenbühl has been working in the ‚Klinik und Poliklinik für Innere Medizin' at the University Hospital of Zurich for 15 years now. He has been dealing with the Fabry disease and Hereditary Hemochromatosis (HHC) for a while now, the first a lysosomal disorder and the other a genetic disorder, which leads to an accumulation of iron in the body. Dr. Krayenbühl and three assistant doctors are in charge of two groups, one with 60 Fabry patients and the other with 180 HHC patients. Fabry patients from the whole German speaking part of Switzerland are treated by Dr. Krayenbühl. In that way, he wants to assure a stable relationship. In his eyes, it is the best opportunity to gain their trust. The idea is to become a centre point for people with these diseases. Dr. Krayenbühl has international contacts and regularly visits congresses and further education, so that he always is up to date.

Based on his experience with Fabry and HHC patients, people with a rare disease are often insecure and have difficulties understanding when they are first diagnosed. They are afraid of the uncertainty what this disease exactly is. That is why they need to be informed properly, at which a center of expertise can help. A question the sufferers often ask first is: "Why do I have to be affected by a *rare* disease?". Thus, they are mostly frustrated at first. Nevertheless, they learn to accept it after a while.
Dr. Krayenbühl believes that his patients do not live more consciously since they have been diagnosed, except for patients with a severe form of Fabry disease.

Dr. Krayenbühl's patients partially went to many doctors before diagnosis. Some of these physicians even claimed that they had psychological problems. When these people are then diagnosed, it is a huge relief for them. However, they afterwards feel that their trust has been abused by the treating physicians. Krayenbühl admits that with rare diseases, finding a diagnosis is most difficult. Many times, it is a long life of suffering for the concerned people. On the other side, doctors are human as well and thus make mistakes. Because rare diseases are *rare*, it is mostly not the first idea that comes up as a possible cause for a problem. In addition, Dr. Krayenbühl affirms that there do exist many patients who overdo it or even simulate. Nevertheless, he believes that rare diseases and the following set of problems should be made more "public", so that really affected people can be diagnosed more quickly. In his opinion, 'rare diseases' are the future.

6.3.3 Dr. med. Michael Fischer, psychiatrist

Dr. Fischer has been working as a psychiatrist for five years and in the psychiatric clinic of Königsfelden for one year. Patients with a brain disease and a psychological disease simultaneously have always been interesting to him. At the moment, he is treating several patients with Niemann-Pick type C (NP-C) and the Frontotemporal dementia (FTD), the first a lysosomal storage and the second a degenerative neurological disorder. He is not specialized in rare diseases. The patients are rather young. He tries to help them medicinally by changing or increasing the medication and with the organization. Furthermore, he tries to lead them into a targeted conversation as a psychotherapeutic help. Nevertheless, the therapy is not meant for talking about deeper thoughts or feelings. In his cases, the problem is that the psychological effects mostly belong to the clinical picture.

During his education, it is possible that he may have heard of these rare diseases a few times. Yet, he thinks that the problem for a late diagnosis, like it is in NP-C, is that most physicians do not think about such a case, for example because they do not remember or never had such a case before. Also, patients without a diagnosis are often been sent to a psychiatrist or psychologist. When a patient is being referred to a psychiatrist or psychologist, he usually assumes that the doctor was right. However, he admits that the physicians are not perfect. It is the experience, also “case-load”, which makes them more aware of such cases. He advises to be very open-minded and yet critical. Of course, this is not a safe method. “It is not possible to do a screening for rare diseases for every patient”. Nevertheless, Dr. Fischer is aware of the negligence of patients with rare diseases. For example, they need to fight hard to get a medicine paid. The support of the health care system is sometimes rather poor. This is very wearing for the concerned people, since they know that something exists, but they cannot pay for it and the health insurance refuses to take over the costs. This helplessness can be a risk factor for depressions.
Another important point to Dr. Fischer is the support of the sufferer’s environment. To him, it is clear that the support is very important for such patients.

A risk factor for a psychological disease is when you can see the effects of it. People start looking at them and those people subsequently retreat.
It is a big change for the affected people. Depending on the symptoms, they succeed in integrating back or not. For some, it is much more difficult to get a job. Following, they need to live from the IV.

Dr. Fischer has got the opinion that diagnosis is always a big shock for the affected. The stroke of fate is something rare disease patients have in common, though every person can react differently. However, he believes that after the hole, they start living more consciously and thankfully and in the present. “You can learn a lot of such patients”.

6.3.4 Alessia Perifano, psychologist

Alessia Perifano is psychologist in France and has been helping people suffering from a rare disease, exclusively lysosomal storage disorders, for two years and a half now. She gives them psychological support and helps them build up their life project, taking their disease into account. Moreover, she accompanies them towards medical professionals who know about their disease, as well as other families affected by a similar rare disease.

Alessia affirms that a typical and frequent problem for people who suffer from a rare disease is that most of them have to wait for a long time until they know the diagnosis. Even so, it is usually hard for them to find medical information about their disease. Most of the time, they are relatively lonely to manage the disease in their everyday life. Based on her experience, until achieving a final diagnosis, it can take from a few days up to several years, depending on the disease. In addition, Alessia thinks that concerned people often feel alone because the disease affects few people and because it is not very known.

In her opinion, the reactions after diagnosis are usually twofold. On the one hand, the patients might be relieved because they know what they suffer from at last. On the other hand, it might be a traumatising experience: in other words, she believes that there is a "before" and an "after" diagnosis.
Reactions of family members and friends can be varied: Some families can turn out to be of great support. Some others may have difficulties to realize the importance of the disease in such a way that they are unable to support their relative. As far as friends are concerned, it also depends. Some of them give precious support, whereas others do not know how to behave. Therefore, in some cases former friends alienate themselves from the sufferer and the patient might lose them as friends.
Nevertheless, the family's support is crucial. And this is not only concerning everyday life care. More important, they are able to help the sufferer psychologically.

From what Alessia has experienced, affected people are often neglected. In France for example, an ever-increasing importance is given to "technical acts" whereas less and less importance is given to the quality of relationship between medical staff and the sufferer. Moreover, there exist very few medical structures which are able to take care of people affected by rare diseases. As a consequence, parents often have to quit their job to take care of their relative.

As far as social life is concerned, some people commit themselves in associations, where they can meet new people facing the same kind of situation and thus who are able to understand them. Concerning their working life, a lot of sufferers have to work less given the tiredness and pain, among others. Some others are simply not able to work anymore. Fact is that everything in life gets more complicated for affected people.

Except for the fact that depression is possibly part of the clinical picture of some rare

diseases, physical pain can also trigger a depression. In addition, the evolution of the disease leading to the emergence of new symptoms, like the inability to walk, can cause mental distress.

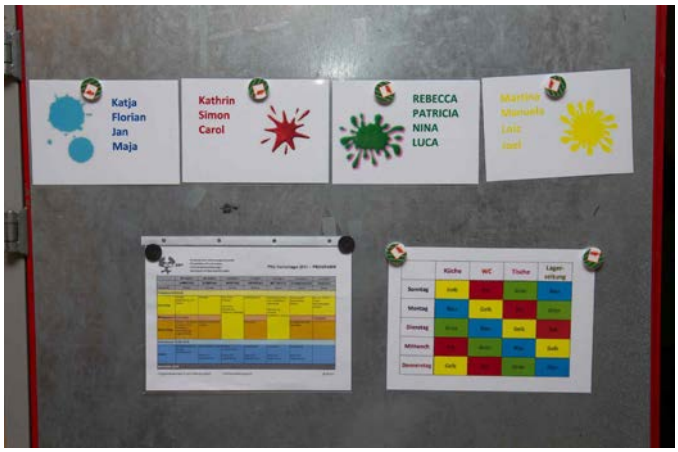
From Alessia's point of view, the people who suffer from a rare disease often go through a sort of "metamorphosis" after diagnosis: life priorities, relationships as well as everyday life change. However, if the affected can live a normal life depends on the disease as well as on the gravity of the disease and level of disability. It is hard for them to make future projects, since it is often associated with the evolution of the disease. Nevertheless, to Alessia, it seems as if they enjoy life and simple pleasures in a greater extent, such as dining with friends or spending time with their family.

6.4 Events

6.4.1 The camp for children and adolescents with phenylketonuria (PKU)

9.10.11, Pfadihuus Lachen

The visit at the camp was truly a valuable experience. I have learned a lot about team spirit and supportive care. During this pleasant evening, I was accompanied by Christoph Hofmann, who kept hold of the moments by taking pictures.



ill.1: All four groups of the camp

At first, we could not find the location where the kids were situated. For a few times, we drove back and forth. Our navigation system did not really help us. Finally, we found the place and arrived with a little delay. We were friendly received by the camp leaders and organizers. Some of the kids were in the main room, curiously observing us. But they quickly found back to their preceding action. We were showed around. We were told that the children and adolescents were split into four groups: the yellow, green, red and blue group. Each group was given "Ämtli" and has got a responsible person, who has to be at least 16. You could say a godmother resp. godfather. This person has to care for the group and is available for questions or when someone has got any worries and wants to talk about it. It is a person of trust.

Every day, different activities were offered (a program), like a guided tour through the 'Bergwerk Käpfnach' or a visit at the 'Alpamare'. At the day of our visit, the children had organized a "sports day". The prizes were given out before the dinner. Everyone got a prize. Afterwards, we were shown the kitchen. The delicious smell reached our noses within a few seconds. Cäcilia Smith and Martina Seiler, the cooks, were about to finish dinner. Cäcilia, a graduated nutritionist, tried to describe to me how they had to cook for the children. Since PKU causes that the amino acid phenylalanine (Phe) cannot be metabolized, the meals have to be cooked practically protein-free. Therefore, she showed me an index with every food product and its protein- and Phe-content listed. This was their fundament to cook the meals appropriately. Soon after, the dinner was served. Prepared was mussaka with a greek salad and tzatziki – for PKU and "normal"

ill. 2: A girl of the camp with a prize she has won

member. I must admit that the PKU meal was much better than I thought it would be – a real pleasure! Every kid had to know its allowed maximum Phe-intake per day by heart. Especially during infancy and adolescence, these limits may not be surpassed but have to be approached as closely as possible. The values were quite individual, namely from 200-1,000 mg of Phe. The cooks made sure that the contained protein and Phe in the meals were indicated as good as possible. Before every meal, they wrote it down on a big placard. Then, the children had to weigh their plate in grams and calculate the exact content of Phe and protein in their meal. In that way, they could check if they had enough or had to eat more. Like that ("how much am I allowed to eat?"), the children learn to become independent in the camp. Mostly, they ate 'Darvida' crackers to reach their Phe-value if it was not enough. The "godmothers" were there to help if it was too difficult.

ill. 3: Children of the camp at dinner with protein-free meals

ill. 4: An elder member as an idol function, calculating how much protein and Phe is in her meal

After dinner, the PKU members have to take in their "medicine". It is a powder, which is available with different flavors. Everyone has got their own individual powder that is adjusted to what they need. Because the powder smells really bad (like fish!) and in addition tastes bad, every affected person has found his or her own way of taking it. Commonly, they drink it with water, coke or orange juice. I tried the powder myself, just to have an idea of how bad it really is. And to be honest, you would not want to drink this voluntarily!

After the responsible group had done the wash-up, it was time for me to riddle them with my questions. Therefore, we sat in a circle. My first question was if someone could explain me what PKU is. The younger ones had a bit of difficulties and answered with: "We've got 20 of them, and 1 doesn't work". But at least they

ill. 5: Different boxes of powder as a protein-alternative


ill. 6: A circle with the PKU kids

knew what it is. The older ones were already able to explain it more precisely. After that, I asked how many people know about their illness. It came out that in most cases, their nearest friends, class comrades, teachers and family members were aware of the situation; mostly people that have to know about it. The parents mostly mention it at the first parent-teacher conference, so that everyone is informed. For the concerned kids, it is sometimes annoying that many know about their disease. “Everybody stares at me, and I need to explain it to everyone. Sometimes, I tell my best friend to explain it for me”. And occasionally, when others notice how and what they eat, they come to ask what that is and why they have to eat it. Apart from that, it is not really significant.

Moreover, I wanted to know which meaning the camp has for the participants. For everyone, the camp is very important. It is the place where you see each other and where everyone knows what the other has. So you do not need to constantly explain, like at school. “Before, I’ve always thought that I am alone”. Instead, the camp members are like a big family, where the older ones look after the younger ones and need to be a role model.
The children and adolescents there do not constantly talk about the illness. They are there to have fun and a good time together. But when they do, they are happy that they can talk about things, which others do not understand.
In addition, the kids do not need to cook themselves or bring their own food, as they need to do for school. Thus, they do not need to worry about food once a year.
The kids are having a great time during camp. The different activities and the program are always varied: visits and tours at special places, games, sport days, movie and disco nights, bathing and so on. Not to forget the “camp journal” at the end of the week.
Before we left, we played “Werwölfle”.

The camp takes place once a year during fall vacation. The children and adolescents with PKU are always excited when the time has come. It is a week where they can be with their own kind, even if they have not got a lot of contact during the year. However, the solidarity I have witnessed and the caring way of how they treat each other is truly amazing. Without a doubt, I have enjoyed the visit very much and I am very happy to have met the people there.


ill. 7: The kids at playing “Werwölfle“

ill. 8: All the PKU kids and me posing for a group photo

ill. 9: The camp leaders and cooks of the PKU camp: Kathrin Stalder, Diana Nussbaumer, Stefan Mützenberg, Cäcilia Smith, Martina Seiler

6.4.2 The self-help group 'Junge mit Rheuma'

5.11.11, Zurich

The self-help group ‚Junge mit Rheuma' has already been active for 16 years. The members reach between the ages of 20 to 50. At the moment, moment the group is headed by Thea Reich, Claudia Casanova and Lucie Hofmann (who also is in charge of the Swiss scleroderma association). They meet four times a year. Mostly, they have a program, like visiting an occasion, a presentation, or a workshop. One meeting is organized as an excursion with the whole family. When I came to visit the group, they had planned lunch at a Moroccan restaurant and afterwards, a Yoga-nidra workshop. During and after this experience, I learned some interesting things.

The group's aim is that members get a chance to relax and exchange themselves, especially about medicine and alternative treatment possibilities. They also talk about experiences, their condition, the family etc. However, even though many members have problems in finding a suitable medication, it is not their intention to constantly talk about their illnesses. Rather, they prefer to spend time together and have a good time. It is a nice change to their daily routine. Thea believes that the difference between other self-help groups and this group is that the "normal" ones tend to have rather older persons, especially when it comes to rheumatic illnesses. In such groups, members seem to be a little discouraged and are not at all interested in excursions or other activities. "This group is for young people and young at heart", Thea assures.

My personal opinion is that even though only a few members were affected by a rare disease, this group is a good example for an ideal self-help group. There, people can be together with others who share similar problems and have fun together.

ill. 10: Self-help group 'Junge mit Rheuma', posing for a group photo after the Yoga-nidra workshop

7. Results

7.1 Results of the interviews

The interviews have proven that the hypotheses cannot be completely confirmed or denied. Table 2 and 3 show the individual responses of the people who were interviewed concerning the theses. The green, red and yellow fields represent the individual attitudes (confirmed, denied, unclear[yellow]) of the interviewees. 'Unclear' can mean that the interviewed person is not sure him-/ herself if the hypothesis is applicable or that the statement has not changed after diagnosis. At the right end of tables 2 and 3, a total number of confirmed, denied and unclear answers have been listed for each hypothesis. The interviewed people were in a stable psychological condition and able to answer questions by themselves. Exceptions were both families concerning Huntington's disease (Bucher and Moser), and partly the Poincilit and Rietzschel family, where the information mostly came from a third party. The results concern the directly affected person or, in the case of whole interviewed families, all closer family members, which are also indirectly affected. The interviews were evaluated qualitatively.

⇒ **Hypothesis 1 (A):** People who suffer from a rare disease value life more than before the diagnosis or illness.

⇒ **Hypothesis 2 (B):** People who suffer from a rare disease value health more than before the diagnosis or illness.

⇒ **Hypothesis 3 (C):** People who suffer from a rare disease live more in the present and enjoy the moment.

⇒ **Hypothesis 4 (D):** People who suffer from a rare disease are more prone to depressions than before the diagnosis or illness.

⇒ **Hypothesis 5 (E):** People who suffer from a rare disease are being excluded by society (e.g. state, health insurance, environment).

⇒ **Hypothesis 6 (F):** People who suffer from a rare disease see life in a more realistic way than before the diagnosis or illness.

Table 1: List of interviewees

1. Poincilit family	**11. Anna V.***
2. Niklaus Hirsig	**12. Monica W.***
3. Rietzschel family	**13. Hans-Rudolf Wenger**
4. Maria K.*	**14. Laura M.***
5. Bruno Bosshard	**15. Seraina Wäspi**
6. Josef Keusch	**16. Joye Corentin**
7. Walter and Itala Bucher	**17. Chantal Kneuss**
8. David and Heidy Moser	**18. Martin H.***
9. Hannelore Wälchli	**19. Kathrin H.***
10. Agatha Meier	

Table 2: Personal interviews

	1. Poincilit	2. Hirsig	3. Rietzschel	4. Maria K.*	5. Bosshard	6. Keusch	7. Bucher	8. Moser	9. Wälchli	10. Meier	11. Anna V.*	12. Monica W.*	13. Wenger	14. Laura M.*	15. Wäspi	**Confirmed**	**Denied**	**Unclear**
$\Rightarrow A$																11	1	3
$\Rightarrow B$																12	0	3
$\Rightarrow C$																11	4	0
$\Rightarrow D$																3	12	0
$\Rightarrow E$																5	9	1
$\Rightarrow F$																12	1	2

Table 3: Telephone interviews

	16. Joye	17. Kneuss	18. Martin H.*	19. Kathrin H.*	**Confirmed**	**Denied**	**Unclear**
$\Rightarrow A$					2	2	0
$\Rightarrow B$					3	1	0
$\Rightarrow C$					3	0	1
$\Rightarrow D$					0	2	2
$\Rightarrow E$					2	2	0
$\Rightarrow F$					4	0	0

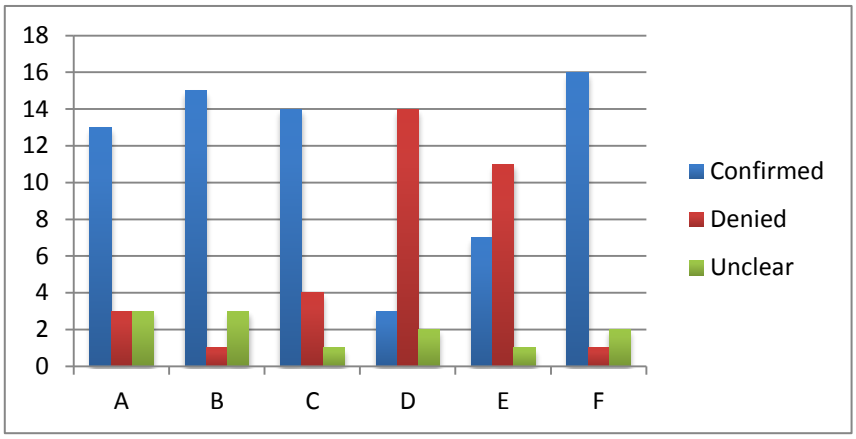


Fig. 2: The total number of results is shown in absolute values in this diagram

7.2 Results of the survey

The results of the survey are demonstrated here. It has been divided into five parts: Personal questions, Rare diseases in the personal environment, Organizations that deal with rare diseases, Rare diseases in the society (situation in Switzerland) and Personal opinion. The diagrams show how many participants have answered a response possibility in percentage. The diagram on the left stands for all the participants (n=250), while the diagram on the right stands for the participants who are affected by a rare disease themselves (n=29). It is clear to me that 29 people are not enough for a scientifically significant result.

Personal questions:

1. What do you think of your current life situation?

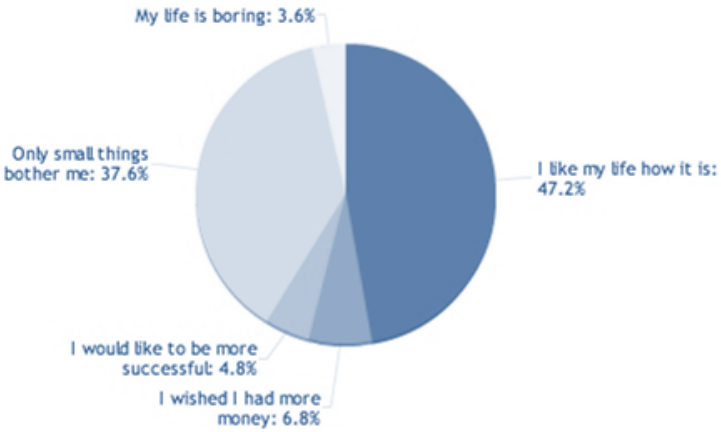


Fig. 3: All participants

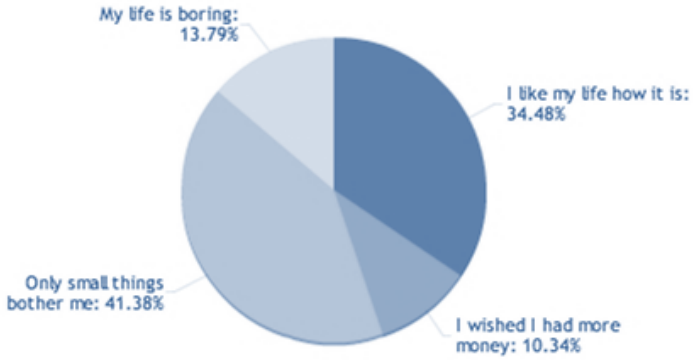


Fig. 4: Participants who suffer from a rare disease

2. What is most important to you?

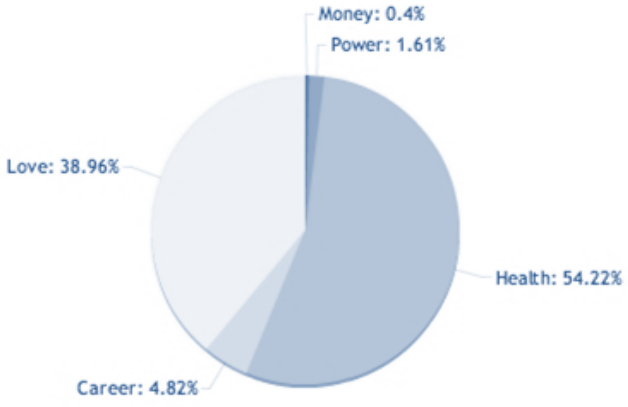


Fig. 5: All participants

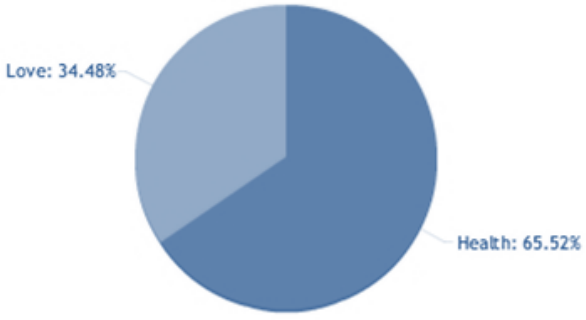


Fig. 6: Participants who suffer from a rare disease

3. How is your general attitude?

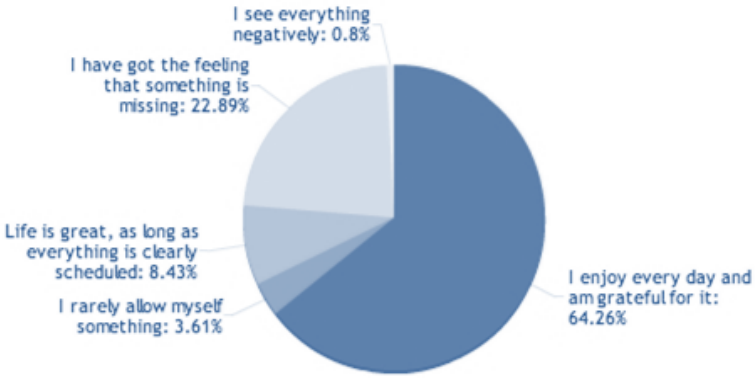


Fig. 7: All participants

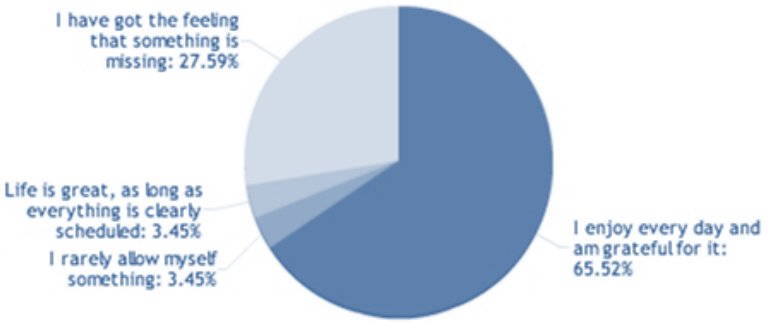


Fig. 8: Participants who suffer from a rare disease

4. I am a realistic person.

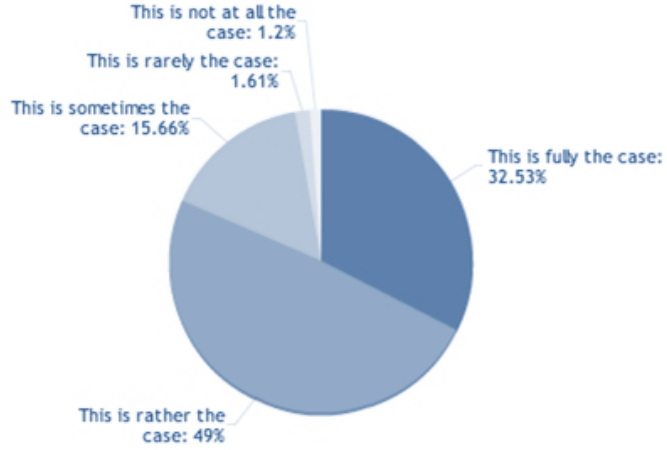


Fig. 9: All participants

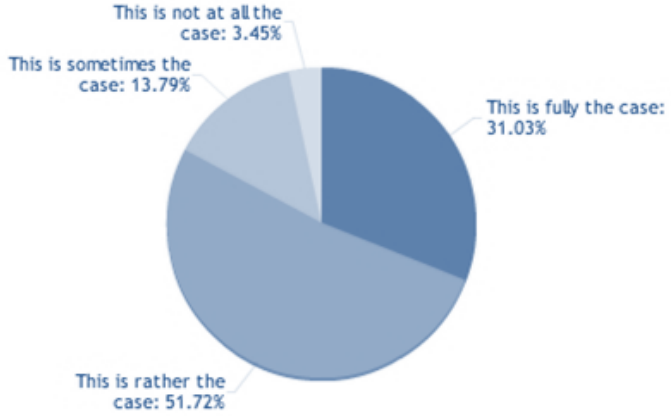


Fig. 10: Participants who suffer from a rare disease

5. I believe that to be healthy is self-evident.

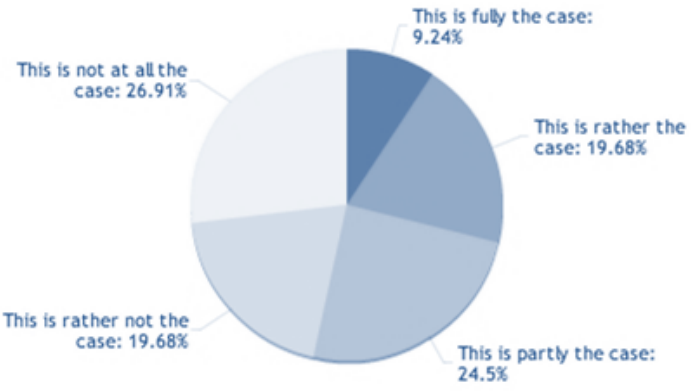


Fig. 11: All participants

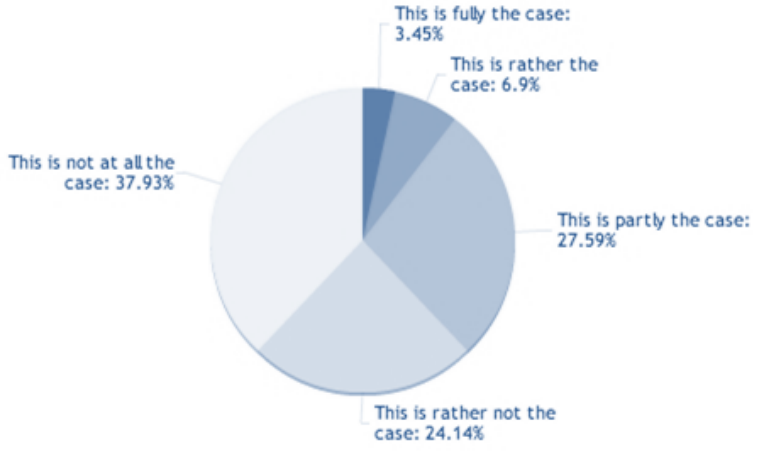


Fig. 12: Participants who suffer from a rare disease

Rare diseases in the personal environment:

6. Did you ever here about the term 'Rare diseases' resp. 'Orphan diseases'?

Fig. 13: All participants

7. If yes, in which context?

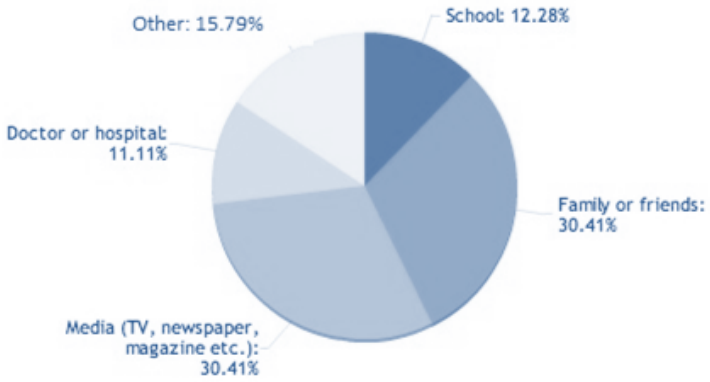


Fig. 14: All participants – Only people who answered 'Yes' in question 6 (n=130).

8. Do you know anyone who is affected by a rare disease?

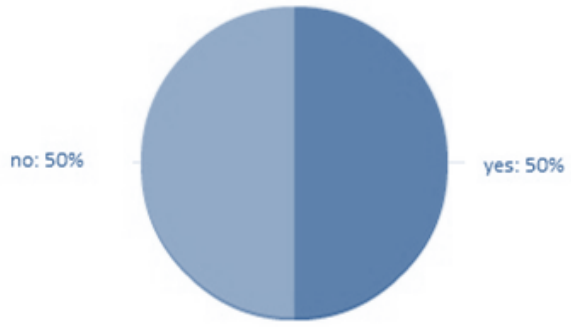


Fig. 15: All participants

9. **In which way are you related to this person?**

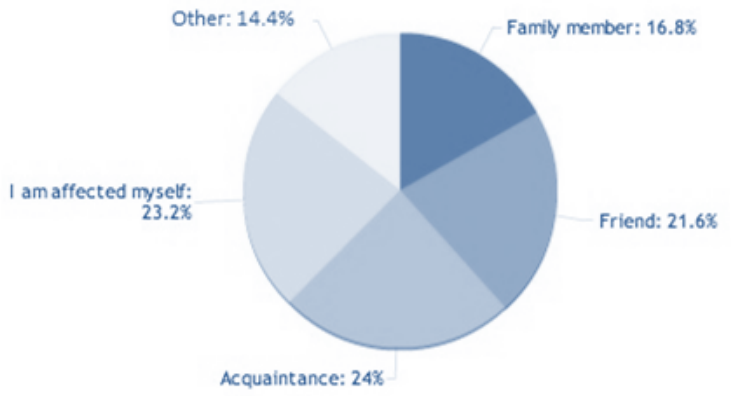


Fig. 16: All participants – Only people who answered ‚Yes' in question 8 (n=125).

Organizations that deal with rare diseases:

10. Have you heard about the European organization Eurordis?

Fig. 17: All participants

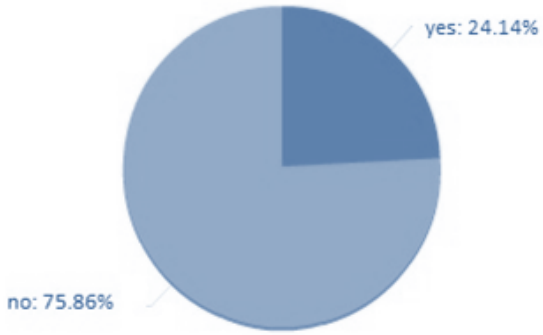


Fig. 18: Participants who suffer from a rare disease

11. Are you familiar with the organization ProRaris (Allianz Seltener Krankheiten – Schweiz)?

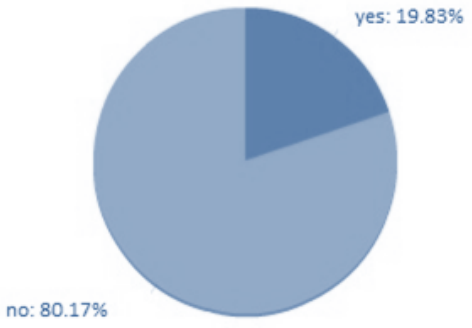


Fig. 19: All participants

Fig. 20: Participants who suffer from a rare disease

12. Have you heard about the website http://www.orphanet.ch resp. http://www.orpha.net ?

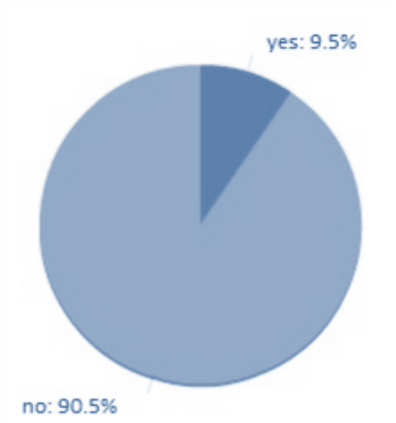


Fig. 21: All participants

Fig. 22: Participants who suffer from a rare disease

Rare diseases in the society (situation in Switzerland):

13. In the Swiss health care system, people with a rare disease often have disadvantages or are discriminated.

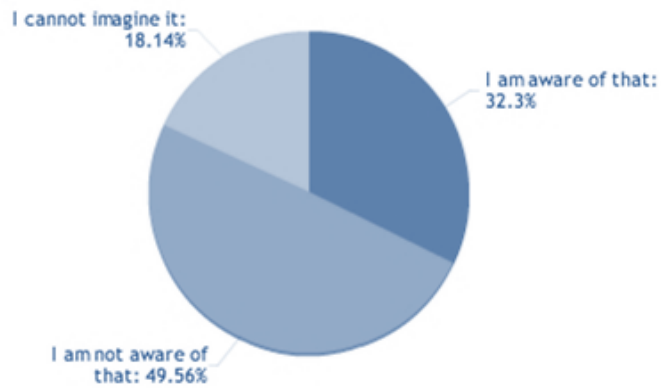


Fig. 23: All participants

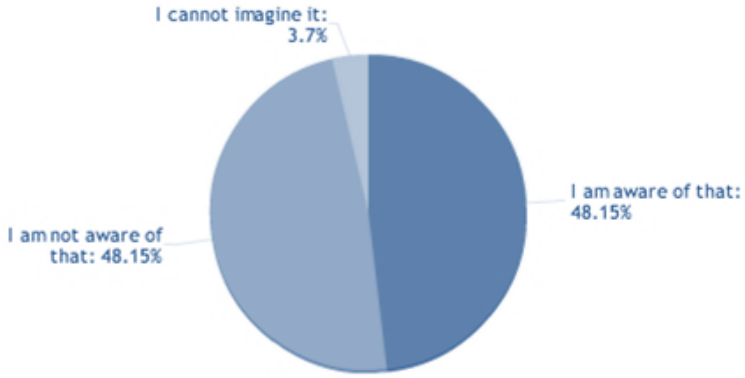


Fig. 24: Participants who suffer from a rare disease

14. While the U.S. and many European countries have already developed a national plan for the typical problems of 'rare diseases', Switzerland is still in the midst of the design phase of a national strategy.

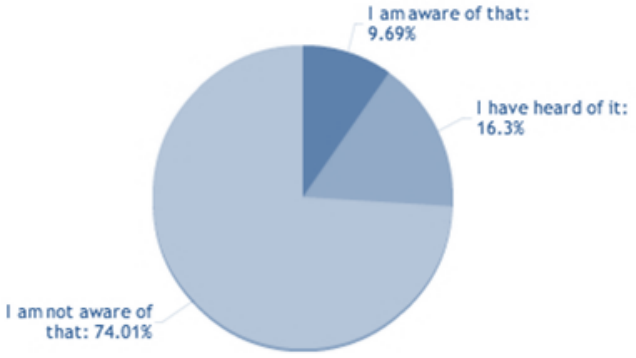


Fig. 25: All participants

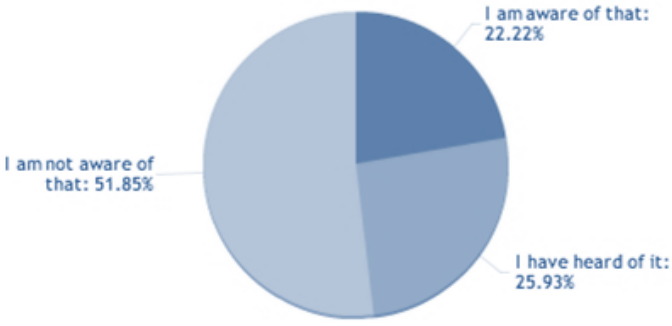


Fig. 26: Participants who suffer from a rare disease

15. In Switzerland, research projects concerning 'rare diseases' often lack of financial support from public and private institutions.

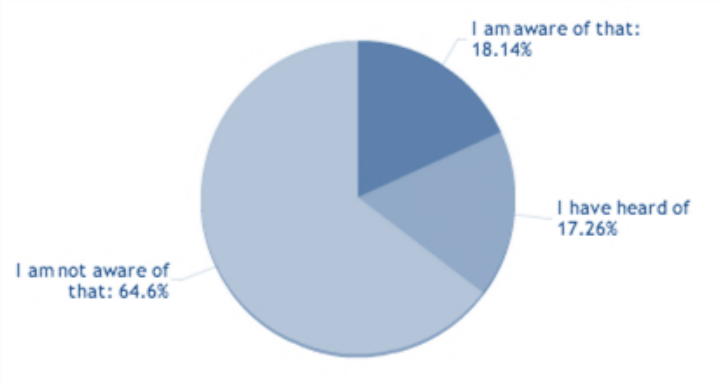


Fig. 27: All participants

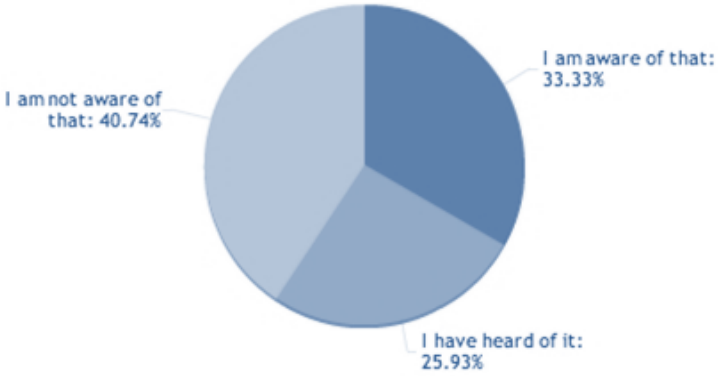


Fig. 28: Participants who suffer from a rare disease

16. Discoveries made concerning rare diseases can often be employed for treatments of common diseases. Do you think this is true?

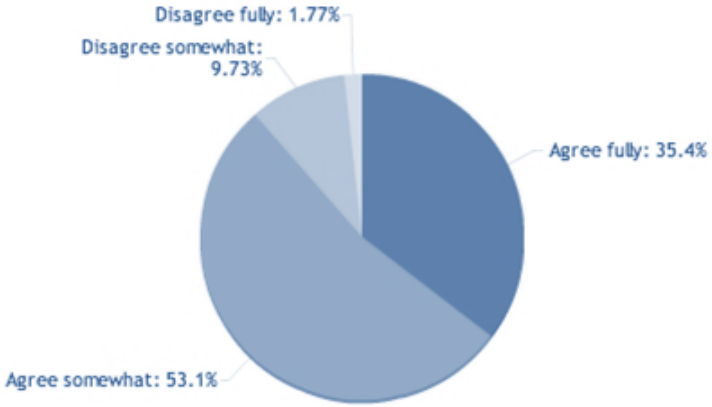


Fig. 29: All participants

Fig. 30: Participants who suffer from a rare disease

17. There exist over 8,000 rare diseases. A disease is considered rare when it affects no more than 1 in 2,000 people (often many less!). 6-8% of the population (1 out of 20 people) suffer under a rare disease. In Switzerland, this corresponds to about 500,000 people.

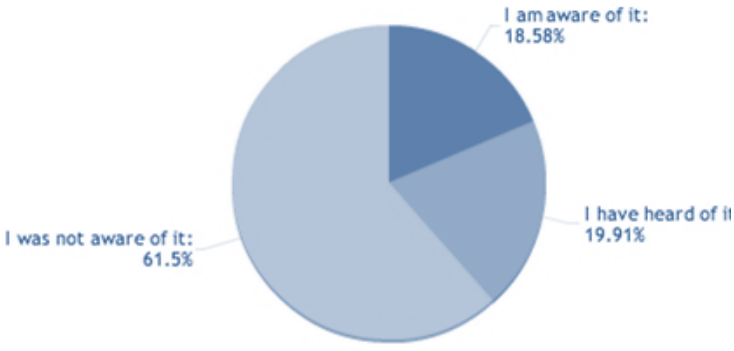


Fig. 31: All participants

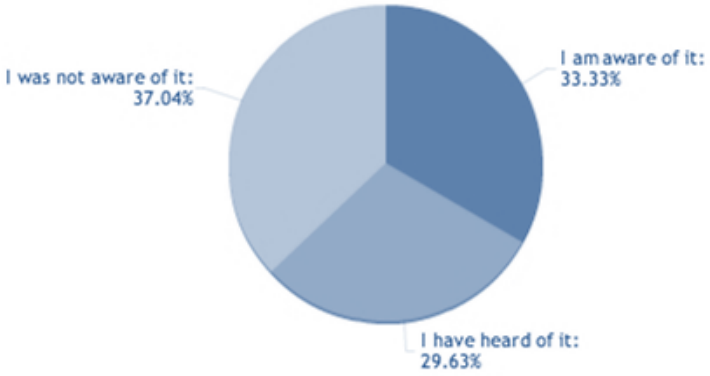


Fig. 32: Participants who suffer from a rare disease

Personal opinion:

18. What is your opinion on the topic 'rare diseases'?

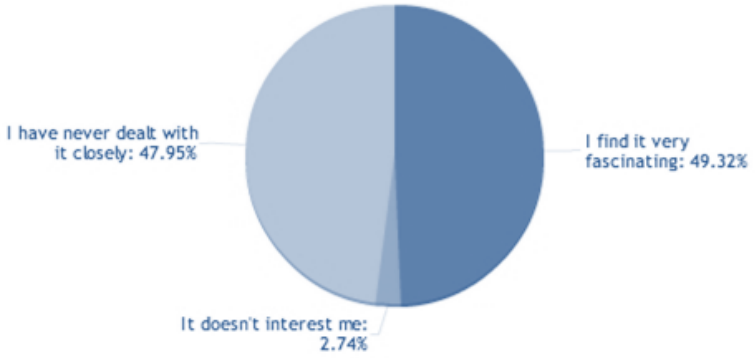


Fig. 33: All participants

8. Discussion

8.1 Analysis of the interviews

Here, different aspects of a rare disease and the consequences it can cause are listed. Important to note is that this analysis is generated through my own experiences, experiences of others and, most importantly, based on the interviews. At this point, I want to point out again that the interviewed people who suffer from a rare disease were people in a steady psychological condition and able to talk about their experiences themselves. Anyway, I wanted to find out if the consequences are only negative, or if they can also be positive. This is what I have found out:

Different degrees of severity

There are many different types of rare diseases with different degrees of severity. Also, within only one disease, there can be different degrees of severity, as you could read in the story of the Poincilit family or Niklaus, which affects the sufferer differently depending on how badly he or she is affected. As in the case of Seraina, there are diseases where you barely feel or notice anything. Then, there are diseases which make you mentally ill and so partly change your personality, and others which make you also physically handicapped or even both together. And last, there are illnesses where you get depressed. Depressions also vary from disease to disease. Not to forget, there are also depressions caused by the clinical picture of the disease. From Alessia Perifano's point of view, pain is a factor that can trigger depression. In addition, the course of disease leads to the emergence of new symptoms, which can also cause mental distress.
Besides the degree of severity of a disease, there is the resulting phenotype. There are some cases – severe or not – where you might not notice any difference to a non-affected person. But unfortunately, sometimes you can clearly see what a disease can cause. In the SNFV meetings (*Schweizerische Neurofibromatose Vereinigung*), Niklaus observed other people who suffer from the same disease. However, in those cases, the illness was clearly visible. These people rather tend to be excluded by society. Therefore, there is a chance that their psychological state can become extremely bad. Martin also went through this experience, even though his illness is not directly noticeable. It expresses itself that he cannot go out very often, since he needs more than 15 hours sleep per day. Yet, because of the incomprehension of his environment, he is isolated. If an illness is not noticeable, there is no reason to behave differently towards that person.

Important differences between affected people

Another fact is that, besides the differences of personality, a difference has to be made between inherited disorders (mostly disorders which you carry since birth or break out in early childhood) and diseases like autoimmune diseases, which you rather get later in life. Mostly, the affected people who grow up with an inborn disorder are better able to accept this and live with it, than people who get it later in life. Monica would be an example for an inherited disorder, phenylketonuria. She has much less of a problem to

handle her illness than other interviewees. The adaption to this "new life" or "different life" can be very difficult in some cases, of course depending on the severity level and psychological state. It involves a lot of effort, patience and time. The victims need to first learn how to deal with this new situation, which is a whole process.
There is also a significant difference between adolescents and adults. For young adolescents, it can be very hard in one way because this heavy load is an addition to the regular load a teenager has to carry: the stress of becoming an adult, the hormones that drive them crazy, the fear of responsibility and the future, rebelling against parents and school, psychological problems etc. Many teenagers, like Glenn for example, deal with at least one of these mentioned problems during this period of life. When they are sick, the disease makes daily life even more difficult. Of course, this does not apply to all juveniles, since Niklaus did not have any problems whatsoever.
Another difference has to be made with the different amount of restrictions. Sometimes, you are barely restricted at all, as in the cases of Niklaus or Maria. In those cases, there is little reason to become upset or even get depressed. Occasionally, you even learn to appreciate some things more than before the diagnosis, things you know now that you can do them. And then again, as seen in Bruno's or Martin's case, if you are so restricted that you cannot do anything you did before (hobbies amongst others), this grave impairment of quality of life can lead to depressions. Like for Agatha, it is a huge change when you have to give up nearly everything.

Who is right after all?

I have learned that there are doctors who have got no understanding for patients who suffer from a rare disease. If no diagnosis is found, they are postmarked as "lunatics" and malingerers, and are often sent to see a psychologist or psychiatrist. Or like in Josef's case, they are just abandoned in the worst case, even with diagnosis. That is why some sufferers start to mistrust doctors. They feel misunderstood and rejected, like Laura did. On the other hand, doctors cannot always be given the fault for such happenings, as Dr. Krayenbühl stated. Not many doctors think at first of a rare disease when they cannot find the cause of the patient's suffering. Dr. Fischer affirms that it partly has to do with the education and experience of a doctor.
Some patients, like Maria or Josef, find out themselves from what they suffer, by searching in the Internet. That is why it is important that articles about rare diseases are existent. The doctors are then still able to test if that is the case or not.

"Before" and "after" diagnosis

As maybe noticed before, a big problem is to find a diagnosis. Some patients need to wait for several years until they get a clear diagnosis. Until then, the disease keeps progressing. Therefore, their life of suffering is very long. The problem then is, are you happy to finally have found out, or upset to know what it is? The opinions differ here. For some, it is a huge psychological burden to not know what the cause is for their suffering, as many interviewees have confirmed. Some are really relieved to find out what they

have, as for Maria or Heinz. At least you can then try to adjust to this new situation and learn how to handle it. In that way, you can specifically fight against the illness, with medication or psychologically, and do not have to float around in illusion, asking yourself if you are really sick or not, is it chronic or just acute, is it curable or not?
However, some affected people are overstrained and shocked to hear such news. For them, it is a traumatizing experience. Many times, these cases appear together. This is what Alessia calls a "before" and "after" diagnosis.

Searching for information

Some do not want to find out more about their disease. They are afraid that if they do, their attitude will change. For example, Niklaus did not want to look up too much information on the Internet. Also, Seraina's mother did not want her to look up too much information. The fact is that sometimes, looking for information can lead you to fearful thinking. Or you can go crazy when you see too much information. Especially when there is no necessity to worry and the state of health of an affected person is okay, this person should not be researching too much. If you then check up the dark sides of an illness, this can psychologically affect you, even if there is no reason for it.

Involvement

Depending on how badly you are affected by a disease, you get more or less involved with this topic. Someone who has never heard of "orphan diseases" obviously does not attend this topic as much as a direct involved person. These affected persons do not only have to be people with a rare disease themselves, but could also be the parents of a child respectively several children who suffer from a rare disorder. They are equally affected and commit themselves to endless support and care. This phenomenon is very well known and there even exist several web pages for affected parents. Mostly, parents of an affected child are known to be more active, as we see with the Poincilit family, Niklaus' mother or Monica's parents. They join patient organizations or attend to self-help groups, or they search in the Internet and seek medical or professional advice to be perfectly informed and prepared about their child's disease. Often, it occurs that parents give up their social life, and their full time job is then to look after their child round the clock. The results are exhausted parents. Therefore, they need to endure big psychological weight. Sometimes, it even goes that far that some parents get psychological problems because of this.[36]
Especially the family members of Huntington's disease patients have got it very hard. They need to see how their loved ones slowly but surely lose the capacity to communicate, move and probably think. Mostly, it is more a psychological burden to them than to the direct affected.

[36] *Trop cher: tu meurs!* 36,9° broadcast, TSR. 2011 August 31

Loneliness

The fact that not many people share the same disease is a typical problem for rare disease patients. As in Hanni's or Bruno's case amongst others, some start feeling lonely and abandoned because of this issue, since they do not know anybody they could talk about the disease with. Sometimes, the disease is so rare that they cannot even talk to a doctor, so they start feeling helpless. They get no help from the hospital or physicians because they do not know much themselves. This can also be psychological weight. When there is no help and the condition of a victim gets worse and worse, it is clear that he or she is then more prone to depression. Laura is a very good example for a person who became completely isolated and excluded from society. Also, when medicine is not being paid by the health insurance, the affected people start becoming helpless, according to Dr. Fischer. The exclusion and negligence of these people is in some cases evident. This is a very important difference to more 'known' diseases. On the other hand, inexperienced physicians are badly informed about such cases. Psychiatrists often only want to fight the symptoms and search too little for the actual cause of a psychological problem.
Therefore, Dr. Krayenbühl believes that national centers of expertise are very important. If there is a place where all affected people of one rare disease can go to, it is much easier to gather information and thus treat the disease specifically.

The importance of associations and camps

The need of patient organizations proves to be very important, especially for people who suffer from rare diseases. People who are the same can meet and talk openly about the illness without feeling ashamed. Or in some cases, not talk about it all, but rather spend a good time together, like the self-help group *'Junge mit Rheuma'* does. It gives a good start anyway and helps them a lot to deal with it, like it is shown with the Buchers, the Mosers or Agatha. Also, it gives a feeling of being understood.
Also, camps can be very helpful, like I have experienced at the '*Camp for children and adolescents with phenylketonuria*'. During camps, the affected children learn how to deal with the illness and are there to support each other. The team spirit is being assisted. Especially the older and therefore more experienced participants can help the younger ones with specific problems. It is also the time were the children can relax and have fun and be with other of their own kind. A change of daily life, and a place where you are understood and do not need to think about your illness constantly. Therefore, they do not need to talk about it all the time. It is especially important to show children who suffer from a rare disease that they are not alone.

Comprehension and confrontation

For some people, it helps to see a disease as a direction sign that guides you in the right direction. This has helped Josef a lot to process the happening. The funny thing is, once you are affected, the situation cannot be changed, since most rare diseases are incurable. So the sufferers need to confront the situation and if necessary, change their attitude. For example, seeing the disease as punishment does not help to process it. Hence, some start

to see it as a lesson for life. Most of the patients I have interviewed told me to have learned something. Certain are more stamped by this experience, some less. In the end, it is as worse as you believe it is. That is why you always need to try to get the best out of it. In addition, death becomes more present to some sufferers. What they need then is a good environment. "If there's no cure, you are more prone to depression", Monica commented. Many also start asking themselves: "If this disease is so rare, why me?". The most common problem is when you comprehend that the disease cannot be cured and hence, realize that you will carry your disease forever. Some need to deal with a more severe disease, others with a less severe one. But the psychological weight remains. Many are then confronted with the question: "Do I accept this new situation? Or do I spend my life in constant despair?" In most cases, the sufferer accepts it.

Acceptance

For nearly everyone, it takes time to accept an illness. It is a process. The time until you accept it varies a lot, from disease to disease and from person to person. At first, you start questioning things like: "This is not fair! Why me? What did I do wrong?". Most of the interviewed people had thoughts like these or similar ones at the beginning.
At the Poincilit's, I was told a very good piece of advice: "Accept the disease as a companion and treat it well. In that way, both will do better."

After affected people have overcome this first phase, they start thinking differently: "The disease is here now, so I will make the best out of this situation". This is what most of the interviewees stated. After this insight, they start feeling much better. As soon as a sufferer is able to accept his or her illness fully, everything gets easier. Naturally, there are not many people who afterwards live with no doubts at all. Instead of depressions, many interviewees claimed to have suffered of phases of not being well and unpleasantness due to their illness. However, after acceptance, many are more open and feel freed. To finally accept an illness can be equalized with a huge weight falling off of your shoulders.

Alessia Perifano and Dr. Fischer have confirmed that the family's and friends' support is essential when a person suffers from a rare disease. Without their support, the sufferers are more prone to depression. If they do get support, it is very much easier to accept the situation. Sometimes, family is the only matter that gives you a reason to live for, as Bruno experienced. Unfortunately, there are persons like Laura who need to fight on their own, because they are isolated. This shows that it is manageable alone. Yet, with support, it is very much easier. That is why it is so very important.

Important changes in life

The attitude of many affected changes after diagnosis, as for Bruno or David amongst others. For example, they become more open, flexible, spontaneous and tolerant or do not let themselves get annoyed so easily, because of small and unnecessary things. They develop a different handling or approach of things. Or they start seeing things more

realistically and consciously, like Bruno. For example, they have a new conception of quality of life and therefore start living more consciously and in the present day. “We take step by step and every day as it is”, Walter said.
Material things are suddenly not so important anymore, and health becomes a priority. For some, the life (or daily life) totally changes; everything is put upside down. Also, there is a new definition of time. It becomes very important, and you start to use it meaningfully. For example, Chantal had the opinion that you start investing more time for yourself and become more attentive for your body’s needs.

This shows that a disease has not only got negative sides. Personal values change, the smallest things make you happy. You become more modest in some cases. You do not let little things bother or worry you anymore. Also, you start to appreciate your life or life in general more as well as your health**.** “You can’t determine your destiny. Therefore, it is important what you do out of it”, Walter said. Alessia confirmed that sufferers seem to be aware of the fact that life is not fair. In addition, she thinks that they seem to be aware that being in a good health is not self-evident at all. Amazingly, nearly all of the interviewees have said to be much more aware of this now.
Yet, results show that not all necessarily appreciate or live their life more than before the illness became a part of their lives, as it is in Maria’s case. It depends on the attitude, which helps a lot!
Many may believe that the future is “controllable” and clearly scheduled. Some affected do not know how tomorrow will look like, that is why they live from day to day and more spontaneous. Holidays they planned to do in the future are taken now, things they always wanted to do are done now.

It is very important to not forget that there are sufferers who have not got the choice how to live and how to see things. As an example, Mathias Poincilit is not able to look after himself and needs to be looked after. Even though he is happy and lives in the present, the disease forces him to think in that way. It is a course and a blessing at the same time. The same applies to Huntington’s disease patients, who are also not able to think of the future or past after the disease has progressed.

8.2 Analysis of the hypotheses

This is a short analysis gained on the information gathered basing on the hypotheses. Here you can see the results. It is clear that the hypotheses are difficult to answer. Aim was to confirm or foreclose hypotheses using the interviews of the affected persons:

⇒ **Hypothesis 1 (A):** *People who suffer from a rare disease value life more than before the diagnosis or illness.*

13 confirmed, 3 denied, 3 unclear:

The majority of the interviewees have clearly confirmed hypothesis A. Mostly, the reason was that they are glad to have survived their illnesses. For some, it is a second opportunity, especially when they have near-death experience.

⇒ **Hypothesis 2 (B):** *People who suffer from a rare disease value health more than before the diagnosis or illness.*

15 confirmed, 1 denied, 3 unclear:

Hypothesis B shows even clearer the agreement of the people who have been interviewed with reference to the health question. As mentioned in chapter 4, this hypothesis developed after the first few interviews, when I noticed that the affected people make a difference in appreciating life and health.

⇒ **Hypothesis 3 (C):** *People who suffer from a rare disease live more in the present and enjoy the moment.*

14 confirmed, 4 denied, 1 unclear:

This hypothesis also proved to be rather confirmed than denied. In a lot of cases, the interviewed person agreed to live more consciously and also value little things. Also, a lot answered not getting bothered so quickly by unimportant things anymore. Many said to take the day as it comes and not think too much about what could happen the next day. That is why many have become more spontaneous and started to do things they never did before, for example, going to a journey they always wanted to go. They decide to live in the present.
Yet, there are some cases where the affected person has got no other alternative than to live in the present, as for patients with progressed Huntington or NP-C. Nevertheless, this belongs to the clinical picture of a disease. Therefore, I mostly tried to answer this question when the concerned person still had the possibility to decide.

⇒ **Hypothesis 4 (D):** *People who suffer from a rare disease are more prone to depressions than before the diagnosis or illness.*

3 confirmed, 14 denied, 2 unclear:

Most of the interviewees denied having undergone a depression. Those who have confirmed had a depression because of the physical effects (≠ clinical picture) of the disease. I have thought that because most of the rare diseases are incurable and chronic,

many would be more prone to depressions. However, many have affirmed to have experienced several phases of despair, uneasiness as well as misgiving and weariness due to their illness. People who have assured this were still categorized under 'denied'. Also, some of the people commented to have felt lonely and helpless at some times because of the disease, one of the reasons why I established this hypothesis. Yet, this did not always immediately lead to a depression.

Those who answered with 'unclear' never suffered from a depression, but they take antidepressant, so that the chance for a depression is smaller or does not occur (and because they are prone to depression).

⇒ **Hypothesis 5 (E):** *People who suffer from a rare disease are being excluded by society (e.g. state, health insurance, environment).*

7 confirmed, 11 denied, 1 unclear:

Here again, many have denied to have been excluded in any way. Nevertheless, more than one fourth of the people who have been interviewed confirmed to have been in that situation. Incomprehension and lack of interest of friends or family members was one of the reasons. Another one is the lack of understanding with regards to the boss of an affected person. The disease can lead to his or her dismissal. In addition, problems with the health insurance happened to be mentioned quite a few times, for the reason that they were not willing to pay for the medication.
The person who answered with 'unclear' believes to become excluded, especially from the job, when somebody knew of her disease.

⇒ **Hypothesis 6 (F):** *People who suffer from a rare disease see life in a more realistic way than before the diagnosis or illness.*

16 confirmed, 1 denied, 2 unclear:

Hypothesis F has proven to be confirmed by the majority. Nonetheless, this hypothesis was most difficult to answer because it depends on the definition. Mostly, they answered positively because they know now that health is not self-evident. Furthermore, some thought themselves to be more realistic because now they know what happens and how others react when you become sick, e.g. friends, job, health insurance etc.
One of the interviewees states to be more realistic with regard to experiences she made with the illness (support etc.) but not in the social or job-related field, since the disease did not permit this.

One important remark I have to mention is that victims who have difficulties in handling their disease, victims where the disease is visible and victims who are undergoing a depression would rather not volunteer to a study. I believe this is the case because most of the interviewees did not undergo a depression or were not excluded. This is why the other hypotheses were mostly confirmed.

8.3 Analysis of the survey

The analysis of the survey is just a small addition to the rest of the paper. The intentions were to obtain the knowledge about rare diseases and a general opinion of the public and to have a comparison between the affected people and the non-affected people, especially in reference to the hypotheses:

Personal questions:

1. **What do you think of your current life situation?**

Most participants either answered to like their life how it is (47.2%) or that only small things bothered them (37.6%). From the people who themselves suffer from a rare disease, 41.38% answered that only a few things bother them and 34.48% answered to be happy about their life how it is. I assume that the former was answered that frequently due to the disease. However, the latter option has still been chosen very often, what indicates that many of them do not complain about their illness.

2. **What is most important to you?**

Here, the options most frequently chosen were Health (54.22%) and Love (38.96%). It is clear that health is important to all, since it is a fact. However, the affected have to learn it the hard way and therefore, they are really aware of it. This could be a reason why Health is 65.52% for the people who suffer from a rare disease.

3. **How is your general attitude?**

At this question, there are practically no differences between the answers of all the participants and only of those who suffer from a rare disease. The majority says to enjoy every day and to be grateful (around 65%). The second option answered most frequently was to have a feeling that something is missing (average of all participants and only affected ones about 25%). I can imagine that these could be smaller issues, like a partner or enough money.

4. **I am a realistic person.**

Again, the answers are distributed very similarly. The majority thinks to be a rather realistic person (around 50%), and a smaller majority to fully be a realistic person (about 30%). About 15% said to sometimes be a realistic person. It probably depends on the context and definition.

5. **I believe that to be healthy is self-evident.**

The different options are very balanced here. However, when it comes to the total answers of only affected persons compared to the whole, nearly 10% more sufferers do not believe at all that health is self-evident, and a smaller percentage believes that this is fully (3.45%) or rather (6.90%) the case compared to all participants (9.24% and 19.68%). On one hand, this shows that not all of the 29 people who suffer from a rare disease believe that health is not self-evident. On the other hand, it seems that sufferers are more aware of this fact than people who do not suffer from a rare disease.

Rare diseases in the personal environment:

6. **Did you ever here about the term 'Rare diseases' resp. 'Orphan diseases'?**

Another very astonishing result is that a bit more than half of the participants (53.66%) have already heard of rare diseases. Since about a fourth is affected by one, this makes a fourth who has heard about it.

7. **If yes, in which context?**

This question shows that most of them know through the media or family members and friends (30.41% each). The field 'Others' (15.79%) contains physicians, job and patient organizations.

8. **Do you know anyone who is affected by a rare disease?**

The next surprising results is that exactly 50% says to know somebody with a rare disease.

9. **In which way are you related to this person?**

Here, the answers are again balanced. 23.2% (n=29) are affected themselves. 'Others' are again mostly doctors, teachers or know the person through their job.

Organizations that deal with rare diseases:

10. **Have you heard about the European organization Eurordis?**

The result is very clear here. The majority has never heard of Eurordis (93.39%). Of the total number of participants who have heard about it (n=16), 7 are affected themselves.

11. Are you familiar with the organization ProRaris (Allianz Seltener Krankheiten – Schweiz)?

Again, a high percentage does not know about the Swiss organization ProRaris (80.17%). Of the total number of participants who have heard about it (n=48), 13 people are affected themselves.

12. Have you heard about the website http://www.orphanet.ch resp. http://www.orpha.net ?

The same here: 90.5% has never heard about 'Orphanet'. Of the total number of participants who have heard about it (n=23), 11 people are affected themselves.

Rare diseases in the society (situation in Switzerland):

13. In the Swiss health care system, people with a rare disease often have disadvantages or are discriminated.

This question clearly shows that the majority is not aware that people with a rare disease often have got disadvantages (49.56%). 18.14% cannot imagine this situation. Interesting is that half of the participants who suffer from a rare disease themselves experienced it themselves, whereas the other half did not (and 1 out of 27 cannot imagine it).

14. While the U.S. and many European countries have already developed a national plan for the typical problems of 'rare diseases', Switzerland is still in the midst of the design phase of a national strategy.

Only 9.69% is aware of this fact, a total of 22 people, where from 6 people are affected. A very high percentage (74.01%) is not aware of it, and 16.3% have heard about it. This shows that very few people know about this.

15. In Switzerland, research projects concerning 'rare diseases' often lack of financial support from public and private institutions.

Again, most of the participants are not aware of this (64.6%). 9 of 41 who say to be aware of it are affected themselves.

16. Discoveries made concerning rare diseases can often be employed for treatments of common diseases. Do you think this is true?

53.1% agree somewhat that results concerning rare diseases can be employed for common diseases, 35.4% agree fully. It is very interesting that the majority could imagine this.

17. There exist over 8,000 rare diseases. A disease is considered rare when it affects no more than 1 in 2,000 people (often many less!). 6-8% of the population (1 out of 20 people) suffer under a rare disease. In Switzerland, this corresponds to about 500,000 people.

Like in the other questions about rare diseases in the society, most of the participants are not aware of this (61.5%).

Personal opinion:

18. What is your opinion on the topic 'rare diseases'?

This question shows that almost half of the participants find the topic about 'rare diseases' very interesting (49.32%). 47.95% have never dealt with it closely, which could also mean that if they were more informed about it, they would start to get interested. 2.74% (n=6) is not interested in this topic, a rather small number compared to the rest.

9. Summary

9.1 Conclusion

The principal conclusion of this matura paper is that the psychological consequences of a rare disease do not necessarily lead to drastic negative psychological consequences. There are even patients who do not suffer any psychological problems whatsoever, whereas others experience a terrible time. Not to forget that some victims suffer from a degenerative illness, that changes the personality as in the Huntington's disease. In this case, the psychological consequences of the disease exert an involuntarily positive side-effect, as the affected person progressively loses the perception of his/her illness. However, it is unreasonable to discuss the effects of rare diseases on a common ground, because they are just too different among each other. Furthermore, as variable the diseases are, so are the individuals suffering from them. So even within one type of rare disease, the experience of the affected persons varies as well. Nonetheless, a disease marks you, no matter if negatively or positively, and a change is mostly present or evident.
Surprisingly, the results came out rather positive, because the interviewed people were in a good psychological condition with only a few exceptions. Many assure to have "learned a lesson" through their disease. Either they have a new perception of life or death, or they appreciate things they never did before. What appears to be the opinion of all though is that they look at the relevance of health differently now. Before, it seemed to be self-evident to be healthy. Now, those who survived and continue to live a more or less "normal" life again gained a much greater appreciation of every life period, in which health is not worse than before.

Even though my illness was a personal motivation to initiate such a matura paper, it was not the only reason why I chose to emphasize on *rare* diseases in this work, instead of on diseases in general. I found it interesting to know what determines rare diseases and how they differ from common diseases. It became soon evident that a low prevalence is not the only difference between rare and common diseases. Further issues linked to rare diseases concern the problem of production and financing of orphan drugs, lack of knowledge about an orphan disease, social exclusion of people with rare diseases and thus loneliness and helplessness they experience. This was the first thought that came into my mind after hearing the term 'rare diseases'. This ignited my interest to know more about the causes and effects of rare diseases. The proposed hypotheses, interviews, events and the survey have helped me to gain a clearer picture on them.

In comparison to other European countries, Switzerland is lagging when it comes to perception and treatment of rare diseases. I was able to point this out with the survey published in this paper: Many participants were not acquainted with this subject and the problems hidden behind it. Nevertheless, thanks to ProRaris and the *'International Rare Disease Day 2011 & 2012'*, i0ntentions for a national strategy in Switzerland are being set in motion.

9.2 Final word

This matura paper has helped me a lot to learn how to deal with my disease 'Takayasu's arteritis'. Seeing and speaking to other people who also suffer from a rare disease offered me different perspectives of how to perceive an illness. It gave me a deep insight into the "rare disease world". I got to know which consequences a rare disease can cause, and that these do not necessarily need to be detaining in one's life.

First of all, I want to thank all the people who volunteered for the interviews, especially those who were directly or indirectly affected by a rare disease themselves. Thank you for allowing me to spend time with you and sit next to you and just listen. I also want to thank those in charge of the self-help groups who helped me find volunteers. Special thanks to Esther Neiditsch and Christoph Poincilit, who motivated me from the start and helped me throughout writing this paper. Another thank-you to Prof. Dr. Peter M. Villiger and the whole medical staff of the department Rheumatology at the Inselspital Berne. Thank you for supporting, enduring and keeping me alive. Also a thanks to my supervisor Yolanda Bysäth, who has been as enthusiastic and curious as I have been from the beginning. Finally and most importantly, I want to thank my family and closest friends, for everything they have done for me, for all the love and sympathy they shared with me. Whenever I needed to be comforted or motivated to never give up, you were there for me.

Iliana Mebert *04.03.12, Zug (CH)*

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Illustrations

11. List of abbreviations and glossary

Aneurysm	Dilation in the blood vessels
Assumption of costs	Taking-over of costs
BAG	Bundesamt für Gesundheit – Federal Office of Public Health
Blood vessels	Part of the organ system that transport blood throughout the body
Board member	A member of the management of an enterprise or organization
Cardiac arrhythmia	Condition of irregular heartbeat sequences
Carotid artery	Artery that supplies head and neck with oxygenated blood
Clinical picture	Theoretical course of disease
Clotting	Process of solidification of the blood
CRP	C-reactive protein – The levels of this protein in the blood rise in response to inflammation
CT	Computed tomography – A medical imaging in radiology in order to see functional processes in the body
Degree of severity	The state of disease
Diagnosis (medical)	Precise attribution of symptoms to define a syndrome, arising with the cause and development of disease
Dose	The amount of a substance or radiance that is applied to an organism and has a biological impact on it
Drug	Medicine
Emphysema	Long-term progressive disease of the lungs that principally causes shortness of breath
Enzyme	Proteins that increase the rates of chemical reactions
General practitioner (GP)	Medical practitioner (physician) who is the first place to go for patients with a medical problem
Hyper- / hypothyroidism	Overactive tissue within the thyroid gland that causes an overproduction of thyroid hormones / condition where there is not enough production of thyroid hormones
IV	Invalidenversicherung – Disability (or Invalidity) insurance

Life of suffering	Time span during one endures suffering
Mass spectrometry	Analytical technique that measures the mass-to-charge ratio of atoms or molecules
Molecular diagnostics	Molecular genetic analysis
Off-label use	Usage of medicine which is not on the list of approved medicaments for the disease one suffers
Oxygen concentrator	Device that provides oxygen
Perilous	Dangerous to life
PET	Positron emission tomography – Nuclear medicine imaging technique that produces a picture of functional processes in the body
Physician	Doctor
Prevalence	Theoretical probability of occurrence of a disease within a population at a given time
Prognosis	Prediction of the likely outcome of an illness
Right heart catheterization	Procedure to measure pressure in the heart and large blood vessels
Sales market	Market in which the manufactured products are being offered to potential and actual purchaser
Sebaceous gland	Very small glands in the skin that secret an oily matter
Spitex	Hospital extern help and health care (at home)
Stenosis	Narrowing in the blood vessels
Symptom	Not habitual function or feeling of a person, indicating the presence of disease or abnormality
Syncope	Fainting
Thrombosis	Formation of blood clot inside a blood vessel
Vascular	Referring to blood vessels
Vasculitis (pl. *vasculitides*)	Inflammatory vascular disease
WHO	World Health Organization

12. Attachment

Interview Leitfaden

- Was für eine Krankheit haben Sie?
- Wie lange haben Sie schon diese Krankheit?
- Wie haben Sie bemerkt, dass etwas nicht stimmt?
- Wann traten die ersten Symptome auf? Was für Symptome waren es?
- In welchen und in wie vielen verschiedenen Spitälern waren Sie, was für Spezialisten mussten Sie zur Rate ziehen?
- Wann und mit welchen Mitteln bzw. durch welche Symptome kam man zur Diagnose?
- Wie lange brauchte man für eine Diagnosestellung?
- Wie haben Sie sich gefühlt, als Sie die Diagnose bekamen? Waren Sie erleichtert zu wissen, was es ist?
- Kannten Sie diese Krankheit schon vor der Diagnose, oder war es etwas, das Sie zum ersten Mal hörten?
- Wie sah es nach der Diagnose aus: Haben sie recherchiert? Wie? Wo? Mit welchen Leuten nahmen Sie Kontakt auf?
- Welche Medikamente mussten Sie schon einnehmen? (Auch diese vor der klaren Diagnosestellung.)
- Waren darunter auch einige, die Ihnen gar nicht halfen oder sogar Ihren Zustand verschlechterten?
- Welche müssen Sie immer noch einnehmen?
- Wie sieht Ihre Therapie aus? Ist sie aufwendig? Ist es eine individualisierte Therapie?
- Wie sehen Ihre Symptome jetzt aus? Kann man durch die Behandlung eine Besserung erkennen?
- Wie lange wird man diese Behandlung fortsetzen müssen?

- Es gibt ja leider Fälle, wo die Krankenkasse die Kosten der Therapie aus ökonomischen Gründen nicht übernehmen will. Wer übernimmt die Behandlung in Ihrem Fall? Wie teuer sind die Kosten pro Monat?

- *Weshalb will die KK die Kosten nicht übernehmen? Was ist der Konflikt?*

- *Fühlen Sie sich vom Staat, der Krankenkasse, etc. im Stich gelassen/zu wenig unterstützt?*

- **Wie sieht es mit Freunden und der Familie aus? Wie gross ist die Unterstützung (mental, finanziell, …) und das Verständnis bzw. die Toleranz ihrerseits?**

- Was haltet Ihre Familie von der Situation? Wie gehen sie damit um?

- Haben Sie einen Partner bzw. eine Partnerin?

- Wie gross und wie wichtig ist die Unterstützung ihrer- bzw. seinerseits? Welche Rolle spielt sie bzw. er jetzt in Ihrem Leben?

- **Hatte jemand der Familie wegen der Erkrankung eine Depression erleiden müssen? Mussten Sie eine Depression erleiden?**

- *Wünschen Sie sich Kinder, eine eigene Familie? Denken Sie, das funktioniert?*

- Haben Sie durch die Erkrankung Freunde verloren oder neue Freunde dazu gewonnen?

- Wie gehen andere Leute damit um, wenn sie erfahren, an was Sie leiden?

- Wie gehen Sie mit der Situation um? Wie fühlen Sie sich jetzt?

- **Haben Sie sich von der Familie, von Freunden, allgemein von der Gesellschaft, abgeschottet? Wurden Sie ausgeschlossen?**

- Sind Sie in einer Gruppe, einem Verein oder einer Selbsthilfeorganisation?

- Wie viele andere Betroffene derselben Krankheit haben Sie neu kennengelernt?

- Wie viele kannten Sie schon vorher? Ist jemand der Familie auch ein Betroffener?

- *Haben Sie Kontakt zu den anderen Betroffenen, die Sie kennen?*

- Fühlen Sie sich alleine, weil so wenige diese Krankheit haben und wenige Leute diese kennen?

- Wie sieht Ihre Prognose aus? (lang- und kurzfristig, aus ärztlicher Sicht)

- Gab es eine grosse Umstellung in Ihrem Leben?
- In welcher Weise sind Sie eingeschränkt, falls überhaupt?
- Wie sieht heutzutage Ihr Alltag aus, können sie ein normales Leben führen?
- Wie fühlen Sie sich gegenüber der Zukunft? Denken Sie, Sie werden es schaffen, ein schönes Leben zu führen, indem sie ihre Träume und Ziele erfüllen können?
- **Inwiefern hat sich Ihre Einstellung gegenüber anderen Menschen und dem Leben geändert, falls überhaupt? Sehen Sie jetzt alles auf einer *realistischeren* Ebene?**
- ***Leben Sie jetzt mehr in der Gegenwart (bsp. jeden Tag geniessen)?***
- **Wie sehr schätzen Sie Ihr Leben und Ihre Gesundheit seit der Diagnose? (Vergleich vorher/nachher)**
- Denken Sie, Sie sind jetzt ein anderer Mensch im Vergleich zu vor der Diagnose? (Veränderungen, neue Sichtweisen, mentale Lebensumstellung)
- **Sind Sie glücklich und zufrieden mit Ihrem Leben (trotz der Krankheit)?**

Interview with Alessia Perifano

How long are you attending people who suffer from a rare disease?
I have been helping people suffering from rare diseases for two years and a half.

In which way do you help them? Do your procedures help?
I help them build their life project, taking into account their disease. I give them psychological support. Moreover, I accompany them towards medical professionals who know their disease, as well as other families affected by a similar rare disease.

Which are frequent and typical problems for people with a rare disease?
Most of them have to wait for a long time to know the diagnosis; then, it is usually hard for them to find medical information about their disease. Most of the time, they are relatively lonely to manage the disease in their every day life.

According to your experience, how long does it take to achieve a final diagnosis?
It depends on the disease: from a few days up to several years.

What is a frequent reaction of a person who has just been diagnosed with a rare illness?
The consequences are usually twofold. On the one hand, they might be relieved because they know what they suffer from at last. On the other hand, it might be a traumatising experience: in other words, there is a "before" and an "after" diagnosis.

What is a frequent reaction of family members and friends?
Reactions can be miscellaneous: Some families can turn out to be of great support (brothers and sisters, grandparents, etc.). Some others may have difficulties to realise the importance of the disease in such a way that they are unable to support their relative. As far as friends are concerned, it also depends. Some of them give precious support; some others do not know how to behave. In some cases, friends alienate themselves from the sufferer.

How important is the family's support? Which new role do they play in the new life of the affected person?
Their support is crucial. First, concerning everyday life care (they often become "helpers"). Secondly, because they possibly help psychologically the sufferer.

Do you think affected people are being neglected? (by health insurance, state, social environment)
Yes, I think so. In France for example, an ever-increasing importance is given to "technical acts" whereas less and less importance is given to the quality of the relationship between medical staff and the sufferer. Moreover, they are very few medical structures able to take care of people affected by rare diseases. As a consequence, parents often have to quit their job to take care of their relative.

Do patients loose or gain new friends due to their sickness?
The answer is twofold. On the one hand, they might lose some friends. On the other hand, they can meet new people facing the same kind of situation and thus able to understand their own.

Do concerned people feel alone because the disease affects few people and because few people know about it?
Yes.

According to your opinion, do many patients fall into depression or get psychoses due to their sickness? Under which circumstances is it more usual?
First, psychiatric disorders (e.g. depression) are possibly part of the clinical picture of some rare diseases. Secondly, physical pain can trigger depression. Finally, the evolution of the disease leading to the emergence of new symptoms (e.g. the unability to walk) can cause mental distress.

Is it a big change in their lives? To what extent?
It is a metamorphosis: life priorities, relationships as well as everyday life change.

Which role do affected people play after the diagnosis in society (still a chance to find a job)? Are they being expulsed ore accepted?
As far as social life is concerned, some people commit themselves in associations. Concerning their working life, a lot of sufferers have to work less given the tiredness and pain, among others. Some others are simply not able to work. They are neither accepted nor rejected, the fact is that anything in life gets more complicated.

How does their daily life look like: Can they live a normal life?
1It depends on the disease. It also depends on the gravity of the disease. Finally, it depends on the level of disability. Most of the time, health care takes time.

Does the attitude of an affected person change in reference to life and health (comparison before/after diagnosis)? Do they become more realistic?
See 12.

Do they live more in the present than before the diagnosis (e.g. enjoy the moment, not always planning ahead)?
Rather than enjoying the present, it is hard for them to make future projects, since it is often associated with the evolution of the disease.

How much do they estimate their life and health after the diagnosis (comparison before/after diagnosis)?
It seems they enjoy in a greater extent simple pleasures such as dining with friends or spending time with their family.

Are your patients happy and content with their lives, even with the disease?
It depends on the person

Umfrage zum Thema ‚Seltene Krankheiten'

Ihre Antworten werden nur im Rahmen meiner Maturaarbeit *„Psychische Auswirkungen von seltenen Krankheiten"* in Form einer Auswertung bzw. Statistik verwendet und streng vertraulich behandelt. Ihre Angaben bleiben auf jeden Fall anonym.

Der Fragebogen ist in vier Kategorien aufgeteilt. Es geht unter anderem darum, herauszufinden, wie sehr das Thema ‚seltene Krankheiten' bekannt ist und einen Vergleich von Betroffenen zu Nicht-Betroffenen zu machen. Der Fragebogen sollte darum so wahrheitsgetreu wie möglich ausgefüllt werden. Kreuzen Sie das an, was für Sie am nächsten der Wahrheit entspricht.

Zeitaufwand ca. 10 Minuten. Bitte pro Frage jeweils nur **eine mögliche Antwort** ankreuzen!

Persönliche Fragen:

1. Was halten Sie momentan von Ihrem Leben?
   a. Mir gefällt mein Leben, wie es jetzt ist ☐
   b. Ich hätte gerne mehr Geld ☐
   c. Ich wäre gerne erfolgreicher ☐
   d. Mich stören nur gewisse Kleinigkeiten ☐
   e. Mein Leben ist langweilig ☐
2. Was ist für Sie das Wichtigste im Leben?
   a. Geld ☐
   b. Macht ☐
   c. Gesundheit ☐
   d. Karriere ☐
   e. Liebe ☐
3. Wie ist ihre Einstellung?
   a. Ich geniesse jeden Tag und bin dankbar dafür ☐
   b. Ich gönne mir selten etwas ☐
   c. Das Leben ist schön, solange alles klar geplant ist ☐
   d. Ich habe das Gefühl, dass mir etwas fehlt ☐
   e. Ich sehe alles negativ ☐
4. Ich bin ein realistischer Mensch.
   a. Trifft vollkommen zu ☐
   b. Trifft eher zu ☐
   c. Trifft manchmal zu ☐
   d. Trifft selten zu ☐
   e. Trifft gar nicht zu ☐
5. Gesund zu sein finde ich selbstverständlich.
   a. Trifft vollkommen zu ☐
   b. Trifft eher zu ☐
   c. Trifft teilweise zu ☐
   d. Trifft eher nicht zu ☐
   e. Trifft gar nicht zu ☐

Seltene Krankheiten im persönlichen Umfeld:

6. Haben Sie schon einmal etwas vom Begriff *Seltene Krankheiten* bzw. *Orphan diseases* gehört?
   a. Ja ☐
   b. Nein ☐

7. Wenn ja, in welchem Kontext ? (hier können Sie mehrere Antworten ankreuzen)
   a. Schule ☐
   b. Familie oder Freunde ☐
   c. Medien (TV, Zeitung, Zeitschrift etc.) ☐
   d. Beim Arzt oder in einem Spital ☐
   e. Andere, bitte präzisieren Sie: ☐

8. Kennen Sie jemanden, der von einer seltenen Krankheit betroffen ist?
   a. Ja ☐
      i. Welche Krankheit: _____
   b. Nein ☐

9. In welcher Verbindung stehen Sie zu dieser Person?
   a. Familienangehöriger ☐
   b. Freund ☐
   c. Bekannter ☐
   d. Ich bin selbst betroffen ☐
   e. Andere, bitte präzisieren Sie: ☐

Organisationen, die sich mit seltenen Krankheiten befassen:

10. Ist Ihnen die europäische Organisation *Eurordis* bekannt?
    a. Ja ☐
       i. Woher: _____
    b. Nein ☐

11. Haben Sie jemals von der Organisation *ProRaris* (Allianz Seltener Krankheiten – Schweiz) gehört?
    a. Ja ☐
       i. Woher: _____
    b. Nein ☐

12. Kennen Sie die Webseite www.orphanet.ch bzw. www.orpha.net ?
    a. Ja ☐
       i. Woher: _____
    b. Nein ☐

Seltene Krankheiten in der Gesellschaft (Situation in der Schweiz):

13. Im schweizerischen Gesundheitswesen werden Personen mit seltenen Krankheiten oft benachteiligt oder diskriminiert.
    a. Ist mir bekannt ☐
    b. Ist mir nicht bekannt ☐
    c. Das kann ich mir nicht vorstellen ☐

14. Während die USA und viele EU-Länder schon einen nationalen Plan zur Problematik ‚seltene Krankheiten' entwickelt haben, ist die Schweiz noch mitten in der Entwurfsphase einer nationalen Strategie.
    a. Ist mir bekannt ☐
        i. Woher: _____
    b. Ich habe davon gehört ☐
    c. Ist mir nicht bekannt ☐

15. Für Forschungsprojekte bezüglich ‚seltener Krankheiten' mangelt es in der Schweiz an finanzieller Unterstützung von öffentlichen und privaten Institutionen.
    a. Ist mir bekannt ☐
        i. Woher: _____
    b. Ich habe davon gehört ☐
    c. Ist mir nicht bekannt ☐

16. Die Ergebnisse von Forschungen über seltene Krankheiten haben oft auch einen Nutzen für die Behandlung allgemeiner bzw. häufiger Krankheiten. Denken Sie, das trifft zu?
    a. Ja ☐
    b. Eher ja ☐
    c. Eher nein ☐
    d. Nein ☐

17. Es existieren über 8'000 seltene Krankheiten. Eine Krankheit gilt als selten, wenn sie durchschnittlich nicht mehr als 1 von 2'000 Einwohner betrifft (meistens sind es aber viel weniger!). Es leiden 6-8% der Bevölkerung (1 von 20 Personen) an einer seltenen Krankheit. In der Schweiz entspricht dies einer Zahl von rund 500'000 Betroffenen.
    a. Das ist mir bewusst ☐
    b. Ich habe davon gehört ☐
    c. Das war mir nicht bewusst ☐

18. Was ist Ihre Meinung zum Thema ‚seltene Krankheiten'?
    a. Finde ich sehr spannend ☐
    b. Interessiert mich nicht ☐
    c. Ich habe mich noch nie genauer damit befasst ☐

19. Weitere Bemerkungen und/oder Anregungen?