

Within time one finds a normal life within the borders of changing directions

On how MFS patients experience living with MFS

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Summary

This thesis will answer the question ‘How do MFS patients understand their condition?’. To find an answer on this question semi-structured interviews were conducted with 19 adults with MFS. These interviews were qualitatively analyzed with Atlas.ti. around three themes: longer-term effects, effects on the social life of MFS patients, stigma experiences and identification. The theoretical framework that was the point of departure for the analysis consisted of theories on meaning, stigmatization, outsiders, identity establishment and embodiment.

From the analysis it turned out that the respondents regarded MFS as a source of limitations which made them live in a social and spatial smaller world. Furthermore, MFS was a source of guilt towards the people closest to them and towards their (future) children. Third, MFS was experienced as a source of insecurity. On the one hand because the respondents felt they could not always show the right line of behaviour. On the other hand because the respondents felt judged on their appearance. In addition, almost all respondents showed one or more characteristics Goffman ascribes to the stigmatized person. Not only the consequences of MFS but also MFS itself is regarded as (potentially)stigmatizing. The diagnosis with MFS was by some respondents regarded as legitimating who they were, while others experienced the diagnosis as a fateful moment.

The respondents turn out to use different labels for their situation. Although not all respondents regarded MFS as an illness, most of them regarded themselves as a patient because MFS was more to them than a biological condition. Furthermore, for some respondents MFS was a source of identification and new contacts, while others distanced themselves from this identification because this felt as loss of control over their image. Finally, some factors turned out play a role in these differences. The most compelling factors were the distinction between internal and external consequences of MFS, age of- and factors leading to diagnosis and presence of MFS in the family.

Although MFS was regarded as a source of limitations, guilt, insecurity, stigma, disturbance of what is regarded as normal and, for some, a source of identification, this was not how most respondents experienced living with MFS in their daily life. This was due to the fact that most respondents embodied one or more of their characteristics of MFS. As a result they became unconscious about living with MFS. In sum one can conclude that MFS has an effect on different aspects of the lives of these respondents, but most respondents have embodied living with MFS and as a consequence they do not feel living with MFS is a tough life. Instead, they adapted themselves to their situation and fatigue, pain and a ‘deviant’ body had become part of their typification of a normal life.

Introduction

In the tradition of medical sociology a lot of different patient groups are researched. This is done both quantitatively, with surveys for example, and qualitatively with interviews or observations. The focus of this research is diverse: some point at the experiences of the patients, others focus at the relation between doctors and patients and yet others are interested in the relations between patients and their relatives. These are only three examples of many possibilities.

In this thesis research is reported on the experiences of people with Marfan Syndrome (MFS). MFS is a rare¹, genetic disorder that affects connective tissue. People have connective tissue throughout their whole body, which has the result that MFS can manifest itself in different body parts. “Given that all tissues contain connective tissue, the clinical manifestations of MFS involve multiple foci, especially in the bones and ligaments (the skeletal system), the eyes (the ocular system), the heart and blood vessels (the cardiovascular system), the lungs (the pulmonary system), and the fibrous membranes covering the brain and spinal cord (the nervous system)” (Fusar-Poli, P. e.a., 2008:243).

MFS is caused by a construction error in one’s DNA. It is mainly caused “by mutations in the fibrillin-1 gene (FBN1) on chromosome 15” (Hilhorst-Hofstee, Y., e.a., 2011: 247). There are a lot of different mutations possible² and as a consequence MFS manifests itself in many different ways. The word syndrome already indicates that it is about a collection of symptoms. The symptoms appear in a specific combination and they are all explained by the construction error of one’s DNA. This one cause makes that it’s labeled MFS, which causes disorders in the cardiovascular system, the ocular system, the skeletal system, the pulmonary system and the nervous system³. As turns out from this list, people with MFS can have both internal and external characteristics.

This diversity in patients is one of the points that makes MFS-patients an interesting group to study from a sociological perspective. Furthermore, there is no cure for MFS. Although a lot of MFS-patients are operated on and take medication, these measures are not able to cure someone from MFS but merely mediate the impact of symptoms. In addition, MFS is ‘negatively progressive’ but one never knows when an attribute starts to get worse. For example, a MFS-patient can have no cardiac problems for years and suddenly the aorta starts to dilate. This unpredictability makes MFS-patients an interesting group to study, because it might result in feelings of insecurity and uncertainty. Finally, MFS is unknown by most people outside the

¹ Estimations range from 1 to 3 per 10,000 (Fusar-Poli, P., e.a., 2008), (Hilhorst-Hofstee, Y., e.a., 2011). In the Netherlands the amount of MFS patients is estimated on 1500 people (www.marfansyndroom.nl).

² In the Universal Marfan Database (www.umd.be) over 600 mutations have been published (Hilhorst-Hofstee, Y., e.a., 2011).

³ For more medical details and a diagnose-criteria of MFS, see attachment nr. 1.

medical field. As a consequence, an encounter with someone with MFS with visible characteristics, might raise a lot of questions and thus impact on their social relations.

This thesis will research the effects of MFS on the lives of MFS patients and what this means to them. The motivation for this research is twofold. To start with, the researcher has MFS herself and when thinking about this subject, she discovered that MFS had more influence on her life than expected. Although she was unconscious about this before, she realized that MFS had both direct, and indirect effects on her life.

Related to this is the second motivation for this research. When looking for social research on this subject, it turned out that most research done on this subject was medical research. The few social research studies that have been done, are quantitative in nature. These research studies used cross-sectional survey data and often focused on quality of life experiences. According to *Determinants of Quality of Life in Marfan Syndrome*, written by Fusar-Poli et al. (2008), people with MFS reported decreased quality of life compared to healthy people. This result was confirmed by research done by Rand-Hendriksen e.a. (2010), which focused on quality of life related to health and which compared people with MFS with the general population. In their article they concluded that persons with MFS have reduced scores on health-related quality of life measures. Furthermore, according to Peters et al. (2005) 32% of the survey respondents “reported feeling discriminated against or socially devalued because of having Marfan syndrome” (Peters, K.F., e.a., 2005:6).

These few social research studies form the point of departure for this thesis. They show interesting results, but there is not much room for the subjective experiences of respondents to answer a survey question with a certain score. In this thesis research will be done to ‘the story behind the survey scores’. It is about the experiences of people with MFS that makes them score lower on these quality of life measures and the meaning they give to these experiences. The main research question that will be answered is ‘How do MFS patients understand their condition?’

To answer this question, it will be researched how MFS affects their social life and what the long-term effects are. Furthermore, it will be analyzed whether they feel stigmatized because of MFS and what this means to them. In addition, it will be researched whether the respondents identify themselves with MFS and whether this is related to the patient association and/or the presence of MFS in the family. Finally, while answering all these questions it will be analyzed what is regarded as normal by the respondents and which meaning they give to all these experiences.

Theory

As made apparent in the introduction, this thesis is about meaning, stigma, identification and perceptions of what is normal. In this chapter different sociological theories will be outlined which will be the point of departure for the analysis of the data.

Intersubjectivity

To explain the concepts of meaning, stigma and identity and the applicability of different theoretical frameworks around these concepts in this thesis, it is important to outline first the concept of *intersubjectivity*. Each person on earth has a personal history and personal experiences – a socially rooted biography. By this is meant that there are no two people on earth who have met the same persons at the same times at the same locations. Everyone has their own experiences with other people, so every person has its own life-world (a concept from Schutz, which will be explained below). Although every person has a unique life-world, people are still able to live with each other and to communicate. This is because these life-worlds of different people also have a lot in common. This commonality between the life-worlds of different people is called intersubjectivity. This intersubjective life-world consists of norms, values and rules of conduct and interaction. In what follows, the concept of intersubjectivity will be explained more extensively and it will be described how this leads to meaning establishment, according to Schutz (1973), potential stigma (following Goffman, 1963) and to social inclusion and exclusion (following Becker, 1963).

Meaning

The founding father of the concepts of intersubjectivity and life-world was Edmund Husserl, who inspired Alfred Schutz for his theory on meaning establishment. The concept of life-world refers to the world of every day social life. “This is an intersubjective world in which people both create social reality and are constrained by the pre-existing social and cultural structures created by their predecessors” (Ritzer 2004:64). Schutz (1973) does not regard the life-world as an objective world. Although lots of actors share a big part of the life-world, every actor has his or her own private aspects of this world. Schutz is most interested in the intersubjectivity of the life-worlds. As said in the introduction, with intersubjectivity we mean that part of the life-world that, despite every one's personal history and experiences, is common to many people.

Schutz (1973) wondered how people were aware of each other's meanings and how mutual understanding and communication is possible. According to Schutz the essence of intersubjectivity is the simultaneity of sharing time and space with others. This simultaneity

means that one can grasp the subjectivity of someone else at the same time as this person lives in their own stream of consciousness (Schutz 1973). The challenge Schutz faced himself was to develop an analytical framework using objective social-scientific constructs, of how it is possible that people subjectively give meaning to the world.

Schutz solves this paradox with his concepts of first-order and second-order constructs. In short, first-order constructs are ideal types we use in everyday life, while second-order constructs are the constructs scientists develop on the basis of these first-order constructs. Here most attention will be paid to first-order constructs while these are important to explain Schutz's theory on the establishment of meaning. One of the terms Schutz uses when he talks about first-order constructs is that of *typifications*. "In any given situation in the world of everyday life an action is determined "by means of a type constituted in earlier experiences" (Schutz and Luckman, 1973:229). Typifications ignore individual, unique features and focus on only generic and homogeneous characteristics" (Ritzer 2004:415).

Typifications are important in Schutz's theory on meaning establishment. According to Schutz, every time people enter a situation, one starts to 'analyze' this situation on the basis of typifications. To interpret a situation or a conversation, one starts, unconsciously, to compare this situation to situations of the same type one has experienced in the past. If this situation or conversation is comparable to one of the many typifications, one recalls the meaning and values attached to the typification of this past situation and applies these to the new situation.

This applying of typifications and at the same time giving meaning to situations and conversation happens mostly in a less than conscious way. "Schutz believed that in the everyday world, as things are running smoothly in accord with recipes, reflective consciousness is relatively unimportant, and actors pay little attention to what is going on their minds or in the minds of others"(Ritzer 2004:424). Crucial here is that people really start to think about a situation and conversation and give real meaning to it, when none of the typifications one 'knows' is applicable to this situation. "Only when it is abundantly clear that our recipes won't work do we abandon them and seek to create, to work out mentally, new ways of dealing with situations" (Ritzer 2004:415).

This theory of Schutz on meaning establishment and the use of typifications is useful in this thesis, because some of the attributes of MFS might have the result that typifications are not applicable anymore. Because MFS is negatively progressive it is possible that people are not able anymore at a certain moment to act in accord with the typifications they always used to give meaning to the situation. In these situations MFS patients might get conscious about their situation and the meaning it has for them. Furthermore, it is possible that some characteristics of

people with MFS are incongruous with the general typifications used by others. This might result in stigmatization of people with MFS.

Stigma

An important aspect of intersubjectivity in general and communication in particular is that one person 'knows' what another person 'knows': that one is aware of the norms and values of other people. This is important in the theory of Erving Goffman (1963) on stigma. When Goffman talks about stigma, he means a characteristic someone has that is discrediting for this person. This characteristic is "incongruous with our stereotype of what a given type of individual should be" (Goffman 1963:3). This characteristic makes other people judge negatively about the beholder, because this beholder differs from the expectations (norm). These expectations are part of the intersubjective life-world. Goffman distinguishes two kinds of attributes: discredited and discreditable attributes. In the first case "does the stigmatized individual assume his differentness is known about already or is evident on the spot" (Goffman 1963:4), while in the second case "does he assume it is neither known about by those present nor immediate perceivable by them" (Goffman 1963:4). So, in the case of a discreditable characteristic, this attribute is hidden. This attribute is only stigmatizing when it is revealed.

Goffman makes this distinction between discredited and discreditable attributes to illustrate that stigma is not about the attribute itself, but about the social relations in a specific situation. These social relations define what is normal and what is not. An attribute is not stigmatizing in itself, but it depends on norms and values of a society whether an attribute is stigmatizing or not. These norms and values of a society, which again are part of the intersubjective life-world, determine which rules apply in a specific situation and these rules determine whether an attribute is stigmatizing.

Whether this attribute is stigmatizing can vary in time, place and the persons present in the situation. For example, someone with a dark skin tone (discredited) might be stigmatized in one country where most people have a light skin tone, while in another country the person with the dark skin tone is regarded normal and the person with the light skin tone is stigmatized. Mental illness (discreditable) can be stigmatizing in particular settings, at one's work for example, while when one is in company of other people with mental illness, it is not stigmatizing. These examples show the most important point about stigma: a stigma is a social relation between an attribute and the social reactions towards this attribute (Goffman 1963).

According to Goffman the stigmatized is mostly aware of the rules that are applied in a situation. Therefore the stigmatized is aware that he is different than 'normals' in this situation.

He knows he cannot get in contact with the ‘normals’ like they are equals. As a result the stigmatized often is ashamed and this shame is often amplified by the reaction he gets on his stigmatizing attribute. Another result of these reactions is that the stigmatized often takes over, or internalises, the negative judgments about himself. Goffman calls this self-stigma. This is more likely to happen with discredited attributes, because all ‘normals’ can see this attribute and can judge negatively about this attribution. As a result a lot of people with a discredited attribute feel ‘like the whole world is against them’. This happens a lot less with people with a discreditable attribute, because they can try to hide their attribute (Goffman 1963).

Both Goffman and Schutz state that actors are aware of the rules, norms and values of other actors and this awareness affects the way people behave in a situation or conversation. In addition, both point at the role of norms. These core themes and connections make them both useful for this thesis. Because MFS is a rare condition, people with MFS can be regarded as differing from the norm. The skeletal problems that might be a result of MFS are in some cases visible on the spot: they can be regarded as discredited attributes. Other consequences of MFS, cardiac problems for example, can be hidden by people. These might be regarded as discreditable characteristics. Furthermore, to research how MFS patients understand their condition, it is important to know whether they feel treated differently because of MFS. Do they consciously feel themselves to differ from the norm and do they feel that others act toward them like they are different?

Because some of the consequences of MFS can be regarded as discreditable attributes, persons can have different ways of dealing with these attributes. Nancy Herman (1993) describes different tactics of handling one’s discreditable attribute. According to her, four strategies of information management can be discerned. The first is that of selective concealment, which is “the selective or withholding or disclosure of information about the self perceived as discreditable in cases where secrecy is the major stratagem for handling information about an attribute”(Herman 1993:307). According to her, persons with a discreditable attribute make a difference between safe others and risky others. The risky others are expected to react in a negative way when the discreditable attribute is revealed, while the safe other are expected not to stigmatize the person because of his attribute.

The second strategy described by Herman is that of therapeutic disclosures. Which is “the selective disclosure of a discreditable attribute to certain ‘trusted’, ‘empathetic’ supportive others in an effort to renegotiate personal perceptions of the stigma of ‘failing’ ” (Herman 1993:311).

Furthermore, she discerned the tactic of preventive disclosure. This is the selective disclosure to ‘normals’ with the goal to influence the perception of that ‘normals’ towards people with such an attribute (Herman 1993). The one with the attribute wants to show that he is normal and is able to act normal despite of his attribute. Another goal is to show that it is not one’s own fault that he has this attribute: he is not to be blamed. Finally, the strategy of political activism is a way of handling one’s attribute. Where the above mentioned three strategies are individual strategies, this last one is a collective strategy. This is the collective variant, where people with the same attribute gather together to meet the same ends.

Outsiders

These tactics might be applied by some MFS patients in handling their discreditable attributes. They might do this because these attributes make them different from normal and as a consequence they risk being labelled as an outsider. When people do not act in accordance with the rules or typifications, they risk being labelled as an outsider. This is another concept which is well known by a lot of people, but needs further theoretical exploration. According to Howard Becker (1963) an outsider is someone who breaks the social rules of a group. It depends on the rules in a situation who is an outsider and who is not. A person can be an insider in one situation while this same person can be an outsider in another situation. Who is an outsider and who is not is thus socially defined. Deviance is created by society: “... social groups create deviance by making the rules whose infraction constitutes deviance, and by applying those rules to particular people and labeling them as outsiders. (...) deviance is not a quality of the act the person commits, but rather a consequence of the applications by others of rules and sanctions to an ‘offender’. (...) deviant behavior is behavior that people so label” (Becker 1963:9).

Like Goffman and Schutz, Becker points at the situational: the way people communicate, act and are perceived depends on the situation and thus varies per situation. In relation to this thesis, it is interesting to analyze in what kind of situations people with MFS feel themselves to be an outsider and in what kind of situations they feel themselves to be an insider. In particular it is interesting to research what the role of the patient association is in relation to feelings of being an outsider or an insider. Furthermore, it can be expected that those respondents who have family members with MFS feel themselves less an outsider than those who are the only one in their family. When people have relatives with MFS, one can expect that having MFS is normalised in this family. The respondent is not deviating from the norm within his family.

Identity

Identification

When the concept of life-world was explained above, it was suggested that every person has their own life-world, because of personal history and personal experiences. This historicity is also an important aspect of Traas's theory on identity. According to Marinus Traas (1990), personal history is the most important element of one's identity. Personal history is the most important reason why people feel themselves connected to the group someone is part of. Personal history and the corresponding identity is seen as part of this larger group (for example ones' family).

In the last part of this theory chapter attention will be paid to identity and identity establishment. This is of interest in this thesis, because one can wonder whether MFS patients identify themselves with MFS and whether they feel they are part of the group of MFS patients. As suggested above, one can expect that some MFS patients, those with the discredited attributes, do not feel themselves to be part of the group of 'normals'. If this expectation is true, it is unlikely that these MFS patients identify themselves as a normal. Stigma might thus be regarded as a threat to identity: although people might want to identify themselves as normal, when other people stigmatize them, they are not identified as 'a normal', but as a 'deviating person'. Here it will be researched whether this is the case and if so, whether they identify themselves with MFS in general and/or with the group of MFS patients as a whole.

As said, for Traas an important aspect of identity is history. Furthermore, Traas (1990) has a relational concept of identity. "Identity implicates a relation of the person towards himself and a relation towards the world. That world includes all issues in the world of the other people, but that relation towards the world also gives colour to the relation of the person towards himself" (Traas 1990:25, orig. in Dutch). Finally, Traas has an eye for the dynamical aspect of identity development. According to Traas (1990) identity is about the ability of people to choose their own way and make their own decisions in a situation in which their surroundings are changing.

Embodiment

Where Traas's theory is about identity establishment in general, and Goffman describes stigma as a threat to identity, Maurice Merleau-Ponty (1962) focuses on how stigmatizing attributes can become part of one's identity. In his theory on embodiment he relates Goffman's concept of stigma to that of identity. In short, embodiment means "taking one's body image into account unconsciously" (Iwakuma in: Corker & Shakespeare 2006:81). This needs some further

explanation to show why this concept is interesting in relation to Goffman, Becker and theories on identity.

According to Goffman the stigmatized person is aware of his differentness and sometimes he takes over the negative judgment about himself. In Becker's theory on outsiders he puts it slightly different. Becker agrees that people are aware of their differentness, but they do not always feel themselves outsiders. Sometimes the one labelled as different feels that the people who regard him as outsider, are themselves outsiders and he himself is an insider. Although this is a difference between Goffman and Becker, they agree on the point that the one labelled as different, knows this.

According to Merleau-Ponty (1962) people with a physical differentness can lose this awareness in a process which he calls embodiment. For example a person who becomes blind, will in the first period bump into every obstacle, because he was used to see things and manoeuvre between objects. After a while, one learns to depend more on other senses than his visual sense and one starts to adjust his environment to his visual impairment. When it becomes more easy for the blind person to move, he will realize less and less that he isn't able to see. Not seeing becomes usual for this person and he will less and less consciously feel impaired. The process of embodiment is completed when the disabled person is unconscious of his impairment and of his own bodily image. When an attribute is fully embodied, it has become part of the identity of the person who has this attribute.

This theory of Merleau-Ponty is mentioned in this thesis, because people with MFS can possess both discredited and discreditable attributes. Because MFS is already present from the moment one is born, it is interesting to research whether people with MFS stay conscious of their attributes throughout their life, or that they embody their attributes. On the other hand, MFS is negatively progressive which means that attributes only get worse throughout life. Some people with MFS develop attributes during their life, for example a dilated aorta. In this case a process of disembodiment can take place. One might be unconscious about having MFS until a certain characteristic develops at a particular moment in time.

Methods

This research is about meaning, typifications and experiences of respondents. The goal is to map these experiences and to reveal regularities. The best research strategy to accomplish this goal is that of qualitative research. One of the characteristics of qualitative research is that its aim is to see through the eyes of the people being studied (Bryman 2004). In this chapter it will be explained which research steps are taken and why certain choices are made.

Research group

As the research question ‘How do MFS patients understand their condition?’ already suggests, the research group consists of people with MFS. In general, this group is chosen because there has not been much qualitative research on this group yet, both in the Netherlands and internationally. By estimation this group consists in the Netherlands about 1500 people (www.marfansyndroom.nl). Of these estimated 1500 people, only adults were of interest for this research. For several reasons it was decided that all respondents should be 18 years or older. To start with, if one wants to interview people under aged, parental consent is an issue. Secondly, adults are more settled than adolescents and as a consequence are better able to consider in what way MFS affects their life. This is not to say that under aged people are not able to do this, but adolescents do not know yet what consequences it has for example on employment. Furthermore, it is expected that the lives of adults are less tumultuous than the lives of adolescents, which makes it more easy for adults to discern what the consequences of MFS are for their lives on the longer term. Finally, by leaving out the under aged, one narrows down the focus. If one includes them, the research will probably become too broad and it would be harder to discover regularities (Bryman 2004).

Method

Of this research group, the experiences regarding different aspects related to MFS were of interest for this research. To collect all these experiences, semi-structured interviews were conducted. A semi-structured interview is a conversation between the researcher and the respondent in which the researcher makes use of an interview guide, but in which the respondent “has a great deal of leeway in how to reply” (Bryman, 2004:321). The researcher has a topic- and question list which is an instrument for the researcher to guide the interview. At the same time, this research guide is not determining the course of the interview. The respondent is free to answer the questions as he or she wants and these answers guide the sequence of the questions.

Thus, the respondent has a great amount of freedom in telling his experiences, but the researcher steers the respondent on basis of his research guide (Bryman 2004).

The strength of semi-structured interviews is that the respondent has a lot of freedom to speak. He can expatiate whenever he wants. As a consequence, it is possible that subjects are touched upon which might not have been expected by the researcher but that are useful for the research. Furthermore, because the researcher makes sure that the subjects he is interested in are talked about, it is possible to compare the different interviews with each other (Bryman 2004). This makes it possible to discover regularities, which is the purpose of this research.

These interviews were recorded on a voice-recorder. This is done because it has several benefits, in comparison with writing down the answers. To start with, if one writes down the answers it is not possible to write down every word that is spoken. If one records the interview, no or almost no information will be lost. Furthermore, a voice-recorder does not suffer from memory loss, so everything that is said, is stored. Another advantage is that every interview can be literally transcribed, what makes a more thorough analysis possible. Finally, the research in total gets more transparent, because it is more easy for other people, if they get the data, to follow the research steps that are taken (Bryman 2004).

Respondent recruitment

To find these respondents, the researcher got in contact with Contactgroep Marfan Nederland (CMN). This is the Dutch patients association for people with MFS. After the patients association agreed on helping to find respondents, the researcher wrote a letter to appeal to the members of CMN to participate in this research⁴. In this letter the goal of this research was explained. Furthermore, it was explained why this research was done and why it would be useful if people responded. To put possible respondents more at ease, it was told that the researcher has MFS herself. It was expected that for some people this would make it more 'easy' to react. Furthermore, it was told that people could be quoted anonymously if they wanted. This too was done to make it easier for people to react on the appeal.

This letter has been published in a digital newsletter, in the paper magazine which is issued every three months and on the website of CMN. Via CMN 21 respondents were recruited, of which 19 were interviewed. Two respondents cancelled the interview appointment⁵ and when new dates were suggested, they did not react anymore. Because of a lack of time, it was chosen to leave these two out of the research.

⁴ See attachment 2. The original letter was in Dutch.

⁵ One respondent heard one day before the interview that she had to go abroad because of her work, while the other respondent had a lack of energy that whole week.

In advance, one of the goals of this thesis was to make a comparison between the experiences of members of CMN and the experiences of people with MFS who were not a member of CMN. To reach these people, a cardiologist of the Academic Medical Centre in Amsterdam was contacted. When he agreed on helping to find respondents, an almost identical letter was sent to him to give to his patients. Unfortunately, this did not result in respondents⁶.

The interviews

The nineteen interviews were held in a period of seven weeks. Thirteen interviews were held at the home of the respondent, three were held in an eatery, two in the office of the respondent and one in a canteen. Most respondents were alone in the room. In one case, the partner of the respondent sat beside him. She did not interfere, only when he asked her something or when the researcher asked her something. The interviews lasted from between 40 minutes to 115 minutes. All respondents agreed on being recorded on the voice-recorder.

Before the interviews started, the researcher explained again to the respondent what the interview would be about, that one was free to answer as desired and that one could refuse to answer a question. Furthermore, it was emphasized that it was about their experiences and there were no good or false answers.

The researcher made use of an interview guide, which contented a topic list and a question list⁷. To put the respondents at ease, each interview started with some general questions about age, hobbies and daily activities. The order of the rest of the topics and related questions was dependent on the answers on the first few questions. The only regularity was that the researcher started with the general questions to put the respondents at ease.

One of the topics that the researcher wanted to address, was that of stigma. It is chosen not to use the word stigma itself. This was done because stigma has a negative connotation and the researcher wanted to avoid that the respondents were 'scared' by these questions. Instead of using the word stigma, respondents were asked whether they felt themselves to be different or to be treated differently.

Analysis

Each interview was transcribed verbatim. This was done as soon as possible after the interview was held. About a week after the last respondent was interviewed, all interviews were transcribed. After transcribing the interviews, they were analyzed with Atlas.ti. This is a computer program in which digital texts can be coded and analyzed.

⁶ A few months after the period of interviewing it turned out that unfortunately he forgot to give the letter to them.

⁷ See attachment 3

The codes that were used in the analysis, were both deductive and inductive. Some of them were used on the basis of the theoretical framework, while others did arise from the interview itself. For those codes that were used a lot, new codes were made which made different categories (or sub-codes) within that one code.

Remarks

CMN has about 300 members, of which 24 members reacted on the letter. Only nineteen of them were interviewed. It is important to be aware of the fact that there might be differences between those who did react and wanted to be interviewed and those who did not react. One must be aware of these differences, although one does not exact know what these differences are⁸.

The respondents were asked if they tell other people that they have MFS and what their considerations are regarding disclosure. Because these respondents wanted to be interviewed, they have a certain amount of openness about their situation. It might be that those who did not react lack this amount of openness. This means that because certain people do react and others do not, there is a bias regarding the subject of disclosure. Thus, it must be emphasized again that the results of this research are not generalizable to the whole group of MFS-patients. On the other hand, when the respondents were asked about their motivation to participate it turned out that some respondents wanted to show that they live a good life despite MFS, while other respondents wanted to show to ‘outsiders’ how tough life could be with MFS. As a result this self selecting bias was balanced out by those who wanted to ‘complain’.

Another important remark is on the fact that the researcher has MFS herself. On the one hand this leads to a bias in the topic list and the question list, because the cause to start this research was partly inspired by personal experiences and by personal observations in waiting rooms in the hospital. On the other hand, for some respondents the fact that the researcher told in the letter that she has MFS herself was an important factor to participate in this research. They thought that this researcher could better understand what they meant than a student who was not familiar with MFS. This was only true for some of the respondents. Others said it would have not made any difference to them. The researcher is aware of the fact that her familiarity with MFS will lead to biases, but at the same time had benefits for establishing rapport and the accessibility of the group of MFS-patients as a whole. In the analysis of the data personal experiences did not play a role. Maybe it did unconsciously, but this is true for all researchers. If the subject was another patient group, the analysis would be done in the same way.

⁸ Although the respondents were asked about their motivation to participate, the motivation of those who did not participate is unknown.

Finally, one should realize that other research methods could have been used as well, for example participant observation on a meeting of CMN. As a consequence of only using semi-structured interviews, one only researches what respondents tell about how they act and about their behaviour, but one does not research whether this corresponds with actual conduct. But because the goal of this research is to map out feelings and experiences, this should not be a big problem.

Data and Analysis

For this research nineteen respondents were interviewed. In the methodological chapter the format of these semi-structured interviews was described and various strengths and weaknesses of this approach were discussed. In the next five chapters the results of these interviews are presented and analyzed. In this first data chapter an overview of the respondents will be presented. This entails that some characteristics of the respondents will be described. In the following four chapters, the content of the interviews will be analyzed around four key themes which emerged within the analysis: long-term decisions, every day interactions, stigma and outsiders, and finally identity. These four themes are related to, and follow from, the various conceptual frameworks described in the theory chapter.

Data Overview and Characteristics of the Sample

In this first part, some main characteristics of the respondents will be described, to give an illustration of the group of respondents. To start with, among the nineteen respondents were ten women and nine men. The youngest respondent was 21 years old and the oldest respondent was 65 years old. The mean age was 40 years, but relatively the most respondents were aged 30 till 39 (see table 1). The median age was 37.

Besides a variation in the age of the respondents, there was also a variation in the age on which the respondents were diagnosed. Three respondents were diagnosed in their early childhood, by this it is meant that they were diagnosed between the ages of zero and five. Four respondents were diagnosed between the ages of six and nine. Four respondents were diagnosed when they were teenagers (aged from ten till twenty). Furthermore four respondents got the diagnosis of MFS in their twenties, while two of them were diagnosed in their thirties. Finally, two respondents received their diagnosis when they were forty.

Age	Frequency
20-29	3
30-39	8
40-49	3
50-59	4
60-69	1

Table 1: Age of respondents (frequency)

Age of diagnosis	Frequency
0-5	3
6-9	4
10-19	4
20-29	4
30-39	2
40-49	2

Table 2: Age of diagnosis (frequency)

Furthermore, respondents were asked about their employment status. Six of them were officially considered unable to work. Two of these four did voluntary work, a third one was busy setting up his own business and finally two of these respondent worked a maximum eighteen hours per week, but continued to receive welfare benefits. Three of the respondents already had their own business. Six respondents worked 30 hours or more per week. One respondent was unemployed and looking for a job. Another one was unemployed but not looking for a new job, because this respondent was unable to work, although she did not mention to be officially considered unable to work. The oldest respondent was retired. Finally, one respondent studied and worked less than 20 hours per week, without welfare benefits.

Occupation	Frequency
Considered Unable to work	6
Unemployed, looking for a job	1
Unemployed, not looking for a job	1
Working at least 30 hours per week	6
Having one's own business	3
Studying and working	1
Retired	1

Table 3: Occupation of respondents (frequency)

On basis of the theory set out in an earlier chapter, it was expected that the respondents who had relatives with MFS had different experiences than respondents who had a spontaneous mutation of their genes and consequently were the only ones in their family with MFS. Nine respondents had a parent with MFS and six of them also had one or more brothers and sisters with MFS. Three of these nine respondents also had children with MFS. Seven respondents had a spontaneous mutation and were the only one in their family with FMS. One respondent had a spontaneous mutation, but got a son who turned out to have MFS as well. Both this son and this respondent itself were diagnosed with MFS after this son was born. This respondent also had a daughter and it was probable that she had MFS as well, but she was never tested. One respondent doubted whether he had a spontaneous mutation or that his mother had MFS and he inherited it from her. Because his mother died shortly after he was born, it was not possible anymore to test this. Finally, one respondent was very recently diagnosed. Although this respondent had a lot of characteristics, he was not yet diagnosed officially based on his DNA. Because his own DNA-sample is still in the process of being researched, his relatives are not yet tested on MFS.

Besides having relatives with MFS or not, another factor that was expected to make a difference between respondents was whether they had 'inward' characteristics and/or 'outward' characteristics. Especially the persons with more severe outward problems were expected to

differ in their experiences from the ones who had little or no outward problems⁹. First, the inward characteristics. Except for one respondent, all had cardiac problems. The most common were dilatation of the aorta and problems with valves. Another inward problem most respondents suffered were trouble with the eyes: severe short-sightedness, lensluxation or retinal detachment. The outward characteristics mentioned by most respondents were length of body above average, long limbs and abnormalities of one's skeleton (e.g. scoliosis and deviant sternum). All respondents had both inward and outward characteristics, but differed in severity. Whether and how this different severity explains differences in experiences, will be analysed in the next chapters.

Except for two respondents, all mentioned having been operated on at least once. Twelve respondents were operated for their cardiac problems and one respondent will be operated for these problems within a few months. Furthermore four respondents were operated on their eyes, while three were operated on their sternum. Finally, seven respondents had an operation to correct deviations of their skeleton, for example a correction of scoliosis¹⁰, a correction of one's sternum or to strengthen their wrists.

Furthermore the respondents were asked whether they take medication related to MFS. All respondents, except for one, took medication. The medication most respondents mentioned to take were beta-blockers, antihypertensive and anticoagulant medication. All these medications were taken in relation to heart problems referred to above. Whether the respondents attached meaning to this and what kind of meaning, will be explored in the chapter on identity.

One last characteristic that was expected to make a difference in the experiences of the respondents, was whether they visited the so called 'Day of Contact' organized by the patients association CMN. Nine respondents were visiting this Day of Contact almost every year, while five of them used to visit this day in the past, but have not visited it in the last few years. Two respondents visit this Day of Contact rarely, but more than once, and one respondent visited it only once. Furthermore, one respondent was going to visit the Day of Contact for the first time a few days after the interview took place. Finally, one respondent never visited this day and he would only visit it when a seminar would take place on a subject he was really interested in.

⁹ With severe outward problems are meant those characteristic that differ a lot from normal. These adjectives, 'a lot' and 'severe', are still subjective. To illustrate what is meant by them, think about body length. The average length of Dutch men in 2009 was 181 cm, while the average length of women in that same year was 167,4 cm (www.cbs.nl). A length of body above the average is one of the characteristics of MFS. If a male person with MFS has a body length of 190 cm for example, this is only 9 cm above the average (5%) and is not regarded as a severe abnormality. If a male person with MFS has a body length of 205 cm, this is regarded as a severe abnormality.

¹⁰ A scoliosis is a curve in one's spine. On an X-ray, this spine looks more like a 'S' or a 'C' instead of a straight line. This may be corrected by planting in metal instruments like pins.

As with this last respondent, most respondents who visit this Day of Contact only occasionally, were motivated by the information they could get on this day. Most, but not all, respondents who visit this day every year were motivated by the contact they could get with other people who have MFS. Gathering information was also important to them, but was of less interest: it was not their main motivation. Probably this is because a lot of the information offered, flyers for example, stays the same over years, so these respondents gathered this information in the past. On the other hand respondents referred to a variation in the content of the workshops and the seminars.

The respondents who used to visit this day in the past, were by that time motivated by getting information and to get in contact with other persons who have MFS. If they would go now, this would be for information. For example when a seminar held is of interest to them:

“Yeah, I thought it was good to go to this day and my parents needed a lot of information off course. But the contact with other parents was also important for them... Also the contact with boys of my age who had Marfan as well. (...)Now I will only cancel football for it and visit this day if the subject of my interest, like I said Marfan and getting children, is addressed in one of the seminars. That is important enough to me.” (R6)

Whether and how these different ‘groups’ of respondents differ in their experiences and the meaning they give to these experiences, will be explored in the next chapters.

Before moving to these chapters which explore the data thematically, first a methodological issue is addressed. It is important to state that the researcher is aware of the fact that these nineteen respondents cannot be generalized to the whole population of MFS patients. It is hard to say in what way the people who read the appeal and did not reply differ from those who did reply and wanted to be interviewed (self-selection bias). Because of this, one cannot generalize the results of this research to all MFS patients.

One qualification to this is that although generalisability is not attempted here, what is imperative within qualitative research is the inclusion of a broad range of experiences and perspectives and that these should be explored in depth. The sample characteristics outlined in this short chapter suggest a broad range of experiences and MFS-related characteristics are included in this research. The following chapters will develop an in-depth analysis of the experiences and perceptions of the respondents.

Changing Directions of Life: Analysis of Longer-term Effects of MFS upon life plans and everyday experiences

In this data chapter, the focus will be on the long term consequences of living with MFS on the lives of the respondents. It will be researched in which way MFS affects the ‘directions’ of the lives of the respondents. According to Bury (1982) a chronic illness can be regarded as a biographical disruption. He suggests that chronic illness is “that kind of experience where the structures of everyday life and the forms of knowledge which underpin them are disrupted” (Bury 1982:169). This is similar to Schutz’s statement that we become aware of the typifications we use when they are no longer applicable to a situation.

According to Bury the unfolding of chronic illness is biographically disrupting because it has consequences for different aspects of life. First of all, people are confronted with issues like pain, suffering and sometimes even death. When one is diagnosed with a chronic illness, one is often forced to think about these issues and finds himself confronted with a process of recognition of these features. Secondly, chronic illness forces a person to take another look at his or her social relations. The illness can disrupt the normal rules of reciprocity and mutual support, which can affect the character of the relationship of patients with their friends, families and wider social networks. Finally, the unfolding chronic illness may have an effect on the plans one holds for the future (Bury 1982). In other words: a chronic illness has an influence on the direction the life of a chronically ill person takes.

All three aspects are related to experienced limitations and the way people have to adapt to ‘fit’ the problems faced in their daily life. In this chapter the data analysis will explore whether MFS patients regard living with MFS as a biographical disruption, which limitations they encounter, how they try to adapt to these limitations and how their working- and family life look like as a consequence of, adaptation to, the diagnosis MFS.

Effect diagnosis

To start with, the analysis will focus on the effect of the official diagnosis of MFS on the respondents. In the interview the respondents were directly and indirectly asked whether the diagnosis had an effect on them and whether this diagnosis had an effect on their self-image. Furthermore they were asked about the factors leading up to this diagnosis. Finally, some of the

respondents¹¹ were asked whether there was a moment or a period in their life when they started to realize what it meant to have MFS.

It turns out that age of diagnosis and factors leading up to diagnosis were important factors in the experiences of getting the diagnoses. Five of the eight respondents that were ten years or younger when they were diagnosed clearly stated that the official diagnosis had no effect on them, because they were too young to realise what it meant. At that age it was just a word for them without any content or meaning. When they got older they started to realise what it meant to live with MFS. For some of them this moment came when they themselves or a relative needed to be operated on.

“Look, the first time I was diagnosed, I was six. That did not really hit me, no. (...) In one year and a half my aorta had grown 4 cm and this was crucial. Within no time I was in the hospital. This was a wake-up call for me. I thought: stop! This is Marfan! But what is Marfan? What the hell do I have? Only then I dived in to information resources.” (R5, diagnosed at the age of six, operated at the age of 22)

“Well in that age, I think I was nine or ten years old, I knew I ‘had’ something, but it did not really bother me. In puberty I had a period of not wanting to be sick, but over time you find some resignation towards it. At the moment I heard I had to be operated on my heart, then it hit me. I thought okay, something is really going on.” (R11, diagnosed at ten, operated at the age of 25)

These experiences of respondents who were diagnosed at a young age differed from the ones who were older. Three of them described exactly what Bury called a biographical disruption. These respondents expected that some aspects of their life would change now they were diagnosed. Although these respondents do experience consequences of having MFS, according to themselves it did not have the effect they expected initially.

“Yeah, this broke me, although I think I already knew it... But it was definitive now. Because now I am not normal or healthy anymore and I have to pay attention to it the rest of my life. It really meant something to me, it hit me. Later on, I thought I am not myself anymore. I had a negative image of that, but it turned out to be not that bad. I stayed the same person. I expected I would feel different in my social life, but it turned out to be not true. In the end these thoughts kind of disappeared, but it still is somewhere in your head.” (R13, officially diagnosed at age 28)

“I expected that I would possibly start to feel more sick, although I did not like this. But still... There is always something. Although, not yet at that age. I know that at that time I thought: this is strange I’m working here with that little bomb in myself. Eventually I got over this thought. You know, you are the way you are and you feel the way you feel. A diagnosis does not change this suddenly... Well, not really. It’s is more that you know it now, but you are still the same person.” (R18, diagnosed at the age of 30)

For yet another group of respondents who were diagnosed on an age older than ten, this diagnosis felt like legitimating their behaviour or like an explanation for their behaviour and their looks. Some of them even felt they were proud of themselves because they now knew they were

¹¹ This question was initially not part of the interview guide, but during the interviews it turned out that a lot of respondents experienced a specific period in life, or a specific situation, in which they became conscious of the effects of MFS. It depended on the course of the interview whether this question was asked or not.

special. For this group having MFS can be regarded as biographical disruption, but the diagnosis made it legitimate¹².

“Suddenly it has a name. First you have a strange body and then it has a cause, you know where it comes from.” (R1, diagnosed at the age of 17)

“And then a lot of things in my life fell into their place. All the burdens you experience since you were a baby, without knowing it. (...) It did not change my self-image but I thought: it is alright, it is allowed. It is not affectation. I gave all I had and I could not have given more.” (R15, diagnosed at the age of 40)

“Now I think, I have got Marfan, it is not my fault. It has got a name. It’s just a mistake in my DNA. And I am not weird, I’m not allowed to say that anymore, I’m different. Yes I am different. But it’s medically objectified, it’s a fact. (...) Yeah, this diagnosis changed everything for me. I’m looking at myself with admiration.” (R16, diagnosed at the age of 30)

Finally, one could see a difference between those who were diagnosed acute because of cardiac problems and those who were diagnosed on basis of milder characteristics. This last group was mostly diagnosed at an earlier age than those who got an acute diagnosis. Those who were diagnosed acute, were more often aware of the consequences for the rest of their life. These respondents realized that their life would take, as a consequence of MFS, another direction than they expected for their own life. This direction of my had in its turn an effect on the identity of the respondents, which will be addressed in the last analysis chapter.

“ Well when I heard it, we went with the two of us to the hospital and at first I intended to go to work after this hospital visit. But I went to my work and I said: I won’t be here for the rest of the day. I was totally upset. (...) It had a huge impact. Partly because of my sister, partly because I already had two children. Maybe also because of life-expectancy and ambiguities about what is going to happen and what is it? Yes, the insecurity. And also towards each other, what do you do to each other, what do you do to the children? Will I be dead the next day or what? What’s the effect on our family-life?” (R10, diagnosed at the age of 40)

“Well I was hospitalized acute because I did not get any oxygen anymore. I was intubated for three weeks, without the doctors knowing what was wrong with me. Then I got an ECHO and they said your heart is not right. And I thought: Oh no, I can never start my own business then!” (R3, diagnosed at the age of 19)

Overall the data suggests that the older the age of diagnosis, the more living with MFS is experienced as a biographical disruption. Related to this are the factors leading up to the diagnosis: the respondents diagnosed at an early age are mostly diagnosed on the basis of milder characteristics, while the ones diagnosed at an older age, were often diagnosed as a consequence of cardiac problems. These problems have significant impact on someone’s life and are a biographical disruption in themselves¹³.

¹² This touches the subject of sense-making, which will be further explored in the chapter on the analysis of identification.

¹³ Cardiac problems were also a reason for those who knew since their early childhood that they had MFS to consider on a later age the consequences of MFS.

These different effects of the official diagnosis might be explained by different notions of risk. Because MFS has several consequences and these consequences might develop themselves over time, the diagnosis of MFS is accompanied by certain risks. According to Alaszewski (2006) diagnosis of a chronic illness can be regarded as a fateful moment. This is a concept of Giddens (1991) which refers to a key personal event that irreversibly changes the ways in which people perceive themselves and the world in which they live (Alaszewski 2006:3). A fateful moment is comparable to a biographical disruption as described by Bury (1982), but Alaszewski refers to a specific moment which has a major impact, while Bury refers more to the long-term consequences. According to Alaszewski (2006) the diagnosis of a chronic illnesses can be regarded as a fateful moment, because certain risks are related and the awareness of these risks changes the way people experience themselves and the world around them. Normal activities that used to be taken for granted, may be experienced now as risky activities. Due to the risks that are connected to a diagnosis, it may feel like one is carrying a time-bomb. The same is true for living with MFS. The greatest risk connected to MFS is that one gets a dilatation of the aorta. For those respondents who expected that MFS would change them and their life (see the quotes above from R13 and R18) and those who were diagnosed on basis of their cardiac problems (see the quotes above from R3 and R10), this diagnosis was a fateful moment. The effect of the diagnosis on them might be regarded as a result of their reaction on the risks that are related to MFS.

On the other hand, there are some respondents who saw the diagnosis as legitimating their looks and behaviour. This reaction is in line with the notions of risk of Douglas (1966). According to Douglas labelling something as a risk always has a function. When people are unsure about the effects and the characteristics that a certain phenomenon contains, it is helpful for these people to label it as a risk. By categorizing it as a risk, they can keep it at a distance and make it more comprehensible for themselves at the same time. When one gets diagnosed with MFS, the consequences of MFS are labelled as risks. The respondents who saw the diagnosis with MFS as legitimating their looks and behaviour applied this as a sense-making tool, where uncertainty about their looks and behaviour had previously existed. Now that it is called MFS, it has been categorized and it is comprehensible and explainable for themselves.

Limitations and adaptations

Here the limitations or difficulties that the respondents experienced will be analyzed. This will reveal what the respondents regarded as normal: when they experienced something as a limitation, this was not regarded as normal. According to Bury (1962) this 'normal life' is

disrupted by a chronic illness. Did the respondents experience MFS as a disruption of what they regarded as a normal life and how did they adapt to this?

To start with, twelve of the nineteen respondents mentioned the experiences of pain. This difficulty is in line with the second aspect mentioned by Bury: people with chronic illness will experience confrontations with pain, suffering and sometimes even death (see above). Eight of these respondents recounted that they felt pain more or less incessantly.

“That pains stays. That pain does not get less. We can try what we want, but somehow it does not work. That is really tiresome.” (R17)

Two of these eight respondents recounted that they did not really feel the pain when they are doing what they like, while they know that it is there. According to Johansson et al (1999) the experiences of pain are related to the social contexts of people, the narratives available to them and whether they feel their experiences of pain are legitimized and recognized (Brown and Vickerstaff 2011). This means that it depends on the situation how pain is experienced. Here the social context of doing what one likes, means that these respondents did not feel the pain in that context although it were still the same bodily consequences of MFS.

“I know that at the moment I am home, I feel all those little pains. But at the moment I am playing basketball, I just scored 30 points, we won and I was topscorer, well I did not feel anything, I can tell you. Really. While everything hurts now, but just now it did not.” (R2)

The other four respondents who mentioned pain, experienced pain only when they carried out specific activities like sports or when they ‘crossed their border’. In other words, when they were more active, or did too much, than their bodies could handle, they felt pain. This crossing of borders was also mentioned a lot in relation to fatigue. Fatigue is another difficulty or limitation, which was mentioned by fourteen respondents.

“I notice that the older I become, the more tired I am. After a day of work, I was knocked out. After dinner I needed to sleep for an hour, otherwise I wouldn’t make it through the evening.” (R5)

“I get tired quickly. I need a lot of sleep.” (R7)

“Fatigue, that is one of the chronic factors of Marfan that always hits you.” (R19)

Related to both the experiences of pain and of fatigue is that most (all except for two) respondents felt limited by MFS because they were not able to do everything they wanted. Mostly this was prohibited by the pain or the fatigue they felt. Strikingly, four of the nine women who run their own household mentioned housekeeping as an activity they were not able to do the way they wanted it. It turned out that these women tend to split up every task to be able to finish it.

“Look, in fact, in my opinion I do everything. It is just that...for example vacuuming the house, I am not able to do that in one turn. I vacuum in different periods. The same is true for cleaning the dishes, I do it in different stages. First I rinse the dirt off and then I let it soak. In this way it is okay for me, I am happy. If I would do it different, thus in one turn, that is not good for me.” (R9)

In addition, gardening was mentioned by both some men and women as an activity they could not perform the way they wanted to. The same was true for playing sports and spending the holidays.

“I would like to play sports more. I would like to play a team sport, but that is infeasible for me. Now I do sport twice a week for half an hour and actually that is already too much for me. (...)It is not really sport, it is more just making movements. I call it sport myself and that sounds good, and I attend it in my sports outfit, but I do not sweat. In that way it is very limited again, it is just moving and that is kind of frustrating.” (R17)

Finally and strongly related to all the other limitations experienced, is that eleven respondents mentioned ‘not being able to keep up with other people’ as a limitation. This points also to the second reason why Bury regards chronic illness as a biographical disruption: it forces one to take another look at his or her social relations. Some respondents felt that they were not such a good friends, because they could not always join activities.

“That Marfan bullshit! Not being able to keep up with someone else, not being able to keep up with your friends, not being able to go out for two nights. That stuff.” (R2)

“Listen, I am 34 and if you look to others of 34, rushing with their children and their work...I am just not able to do that” (R4)

As a result of all these limitations experienced (pain, fatigue, not being able to do what they want and not being able to keep up with others), ten respondents experienced life in a smaller world than when they would not have MFS. This was felt as a burden. This world was smaller in a social way (see the quote above of R2) or in a spatial way or sometimes both. In a spatial way it meant that some of them could not visit the places they wanted to visit. This last burden was often due to having poor vision.

“I do have an electrical scooter and I am not able to go wherever I want, because this electrical scooter has only a limited range. That is all a burden, so I do feel enormously limited. (...) If I am going to some place new and I don’t know what the entrance looks like, then that is a burden for me. I will not go to that place that fast, because I might stumble for example.” (R8)

As pointed out before, the limitations experienced are closely related: not being able to perform the way one wants is often a consequence of pain, fatigue and bad vision, while the smaller world is a result of not being able to do what one wants.

According to Schutz, when typifications are no longer applicable, people will look for new typifications to give sense to the situation. Bury too pointed out that when people experience a biographical disruption, they try to adapt to the situation. “There is the response to disruption involving the mobilization of resources, in facing an altered situation” (Bury 1982:170). In analyzing the interviews with these nineteen respondents, it turned out that they knew how to adapt to this new situation as well. For example by knowing how to deal with limited energy, by making use of certain resources, by turning ones hobby into another form or by making adjustments to the furniture¹⁴.

Working life

Now the limitations faced by the respondents are mapped, this section will analyze how living with MFS affects the working life of the respondents. According to Bury a biographical disruption affects different aspects of one’s life. For people in general work is an important, long-term, aspect of life. This section explores how these respondents felt their working life was affected by MFS and what they thought their working life would look like if they did not live with MFS.

As pointed out several times already in this research, there is a big variety between people with MFS in terms of the characteristics they have and how severe these characteristics are. In advance it was expected that the respondents who had more inward problems, for example cardiac- or visual problems, would experience more difficulties regarding work than those who had more outward problems. This was expected because these inward problems have an effect on the body itself, what is often ones ‘instrument’ at work.

From the analysis it turned out that this distinction is difficult to make. First, because all respondent had both inward and outward characteristics¹⁵. Secondly, because only four respondents referred to MFS having no influence on their working life at all¹⁶. Except for these four, the other fifteen respondents made remarks on MFS in relation to work. It turned out that MFS had an effect on the amount of work of fourteen respondents. Both those six respondents that were unable to work as a consequence of MFS (see table 3) and those respondents who worked for fewer than 30 hours per week said that they would work more hours if they could. This points out that working full time was regarded as normal by these respondents. This already

¹⁴ These adaptations might result in embodiment, which will be addressed in the chapter in identity.

¹⁵ Eight of them had severe outward characteristics, while thirteen had severe inward problems. Of these thirteen respondents one had severe ocular problems, while the other twelve had severe cardiac problems.

¹⁶One could expect that these four only have milder characteristics, but two of them had already been operated on cardiac problems and the other two had outward characteristics (one more severe than the other).

illustrates that MFS and the progression of it is a biographical disruption. Without MFS they probably would have reached this norm, but MFS forces them to change this perspective.

“Working is the biggest sacrifice I made.” (R15, stopped working at the age of 50)

“Well, if I would not have Marfan, I would still be working. Yes I would work fulltime then. (...) Even with cardiac problems you should be able to work, but I am just unlucky that my whole aorta is affected.” (R3, considered unable to work).

Furthermore, some respondents were influenced by MFS in choosing their job. Out of eleven respondents who had a job, five told that they had taken the limitations set by MFS into account when they chose their jobs.

“When I am searching the job advertisements, for example for shop workers, then I seek out what kind of shop is it? Does it involve a lot of standing, how much does it affect my back, how much does it affect my feet? I also pay attention to dayshifts, evening shifts and nightshifts. I must not respond to those adverts too much, because I know it is not good for me. I really need my eight hours of sleep.” (R5, currently unemployed, looking for a new job)

In addition, some respondents told of how they tried to make it as ‘easy’ as possible for themselves within their job. By making some adjustments, they tried to make it physically less demanding. One can point out that, by mentioning these adjustments, these respondents ‘admit’ that they would perform their job in another way when they would not have MFS. Working without these adjustments is regarded as normal, but by having these adjustments the respondents succeed in combining the norm of having work and living with MFS.

“For example, I worked in a supermarket for two years and that was pretty heavy. (...) After a while I thought I have to make sure that my colleagues take the heavy boxes, then I will take the light boxes. The boxes with toilet paper for example, those are light and then I still could work in their tempo. That is how I tried to deal with that.” (R14)

“My employer initiated some adjustments for me. I will get a new chair, my bureau is on the right height now and I got my own office. I asked whether I could get some more files than usual, so I can spread the papers and every file is less heavy than usual.” (R7, works on average 15 hours per week)

Remarkably there was a contradiction in the meaning given to having ones own business. Two respondents said that although it is nice to have your own business, it made them work harder than was good for them. Two other respondents, on the contrary, saw it as a blessing that they could work for their own company: it made it possible for them to work within their boundaries.

“As I told you, we had our own company and combined with the house, I worked for about 70 hours a week. Sometimes I say I went way too deep. I went on for too long and too deep. I think if I would have been in paid employment, I would have quit long before. Because it is your own company, I just kept on going.” (R15)

“ I really needed to give up a lot of working after my second heart operation. So, for me it is ideal that we have our own business and I can choose how many hours I work. That is my luck I think. I do not think I would be employed somewhere else (laughs).” (R12)

Family life

In this last section on MFS as a biographical disruption, it will be analyzed whether MFS has an influence on the family life. This issue is interesting, because it is an important part of most people's life. Once again with Bury (1982) in mind, family life may be regarded as a part of one's daily life that is surrounded by expectations of what it should look like. These expectations might be undermined by MFS.

The focus here lies on having children. From the nineteen respondents, five respondents had one or more children. Two of them did not know that they had MFS when they became a parent, two of them became parents 'by accident' and finally the last one consciously chose to get children 'despite' MFS. At the moment she got pregnant she felt that she could live a good life with MFS, so why would this be different for her child? Fourteen respondents had no children, but for six of them this subject was not yet a closed book¹⁷. Despite variations in how their family-life looks like, all respondents turned out to share a lot of considerations which will be analyzed below.

First of all, most respondents did not want to take the risk of passing MFS on to their offspring. They did not want to do this to their children. An important issue mentioned by almost all respondents is the uncertainty about the severity of MFS when it is inherited. Almost no respondent would take the risk that his or her children would come off worse than they did themselves. This uncertainty about the inheritance was an important consideration.

"Sometimes I use to say as a kind of joke: my kind should not be multiplied. Because you know you have an inheritable disease and you do not want to give this to someone else." (R1, unplanned parent of two children)

"We chose to try IVF because the risk that you pass MFS on to your baby is 50%, but the severity of the MFS is uncertain. If I look at myself and how much I am bothered by it now, I think: okay if I have the possibility to save my children from this, I need to take this chance." (R2, in a trajectory of IVF)

For seven of the ten women another important consideration was whether they were physically strong enough to be pregnant and, after that, raise a child. Two of them said they were not strong enough to raise children, because they could not carry them and did not have enough energy to play with them. Three women stated that their aorta was that wide, that they would not survive a pregnancy. One woman got pregnant unplanned, but because of both inheritance and aortic problems she would not risk to get pregnant for a second time. Finally, one woman, aged 21, did not want to wait too long anymore with looking for the possibilities because of possible

¹⁷ Two of these respondents were yet undergoing a trajectory of IVF. They wanted to try first to get a child without MFS and if these trajectories of IVF would not result in a pregnancy, they would reconsider the issue of inheritance again. The other four were thinking about it, but the decision was to be made in the future.

deterioration of her cardiac problems. For these women MFS contained not only an ethical burden, but also a physical burden to get children.

“Well, you have seen my partner, he is considerable older than I am, so that is not a good idea. Besides this, rationally you know that there is 50% risk you might pass it on. You never know in what severity and I doubt whether I could handle it myself. My sister has got children and if I look at her...I could never do that. That is way too heavy for me.” (R4, no children)

“ No, I would not have survived a pregnancy with an aorta like this.” (R15, no children)¹⁸

Whereas the above consideration is solely a consideration of the women, both three men and three women stated that they have certain responsibilities towards their (future) children. On the one hand this meant for two of them that they stopped with certain risky activities. On the other hand this meant that they wanted to grab every possibility to get a child without MFS. At the moment two respondents were on a trajectory of IVF and two of them stated that if they wanted children in the future, they would try it first via IVF. These four all stated that if you have the possibility to save your future child from MFS, it is your responsibility to take this chance.

“I still do what I want, but with the knowledge that I have certain responsibilities towards my children. If I would not have them, I would have worked and I would have been a thrill seeker. I would have lived life to the max, but now I take responsibility not only for myself but also for them.” (R3, two children)

“If I would not have Marfan, we probably would have tried it several years ago in a normal manner... But it is something that I wish I did not have to do to a child. If you have a choice, I think you have to try to save a child from it. Not that I am not happy that I am here, but you never know the severity when you pass it on. I think it would be terrible if your child blames you for passing it on, while you were aware of it and you have the chance to save it from Marfan.” (R13, in a trajectory of IVF)

Finally, and strongly related to the considerations already mentioned, were issues of guilt. On the one hand, two of the five respondents who had children and who did pass it on, felt guilty towards their children. One of them consciously chose to have children, because at that moment she did not feel bothered by MFS herself. During her second pregnancy she needed a cardiac operation and then she realized what the effects might be on her and her children. The other women also got difficulties, a dilatation of her aorta, during her second pregnancy, but she never knew before she had MFS.

“ When I was pregnant of my son, Marfan really started to bother me and then I thought: What have I done? What have I passed on to my children? (...) With 27 weeks my aorta ruptured, I got operated and then I thought: Marfan means that it is possible that your children have to live without their mother.” (R3, two children)

“For me that is the worst part, that I passed it on. Towards my son I think: what heavy burden have I given to him?” (R12, two children)

¹⁸ This statement was based on her general perception due to her general level of health.

On the other hand, those respondents who had no children said that they would have felt guilty if they had passed it on to their children. They stated that although their own life was worth living, they would feel guilty when they passed it on to their children.

“Most people are worse off than I am, so my children would probably be worse off than I am, so I do not want to do that to them. I could never forgive myself if I get a child with Marfan. Not that I have such a bad life, but it can give severe limitations and I do not want to pass that on.” (R6, no children, possibly in the future)

“If my future child will have Marfan and in an severe way, I think I will feel very guilty. I can imagine I would blame myself for passing it on.” (R16, no children, possibly in the future)

It turns out that MFS is an ethical burden, for both men and women, and a physical burden, for women, to get children. In this aspect of life, MFS turns out to be a biographical disruption. This is underlined by the statements of most respondents it was a tough decision, which took several years. Furthermore, the whole trajectory of IVF is a trajectory of several years. Especially for most of the women who decided not to have children, this was a tough decision. It meant for them that the picture they had in mind of a future with children did not become reality.

“Back then it was very tough for me. I nearly could not talk about it, because I always thought: one day I will be a mother. You feel you are not only in this world for yourself and I think I could have been a good mother. And now you do not get a chance. I followed their advice. They will never say you are not allowed to get children, but who am I to take this risk? But that was far from easy.” (R5, no children)

Conclusion

MFS might be regarded as a biographical disruption because it affects both the working life and the family life of most respondents. It meant for them that as a consequence of living with MFS, they could not reach what they regarded as a normal life. The typifications they used for this normal life were ‘working fulltime’ in ‘a job freely chosen’ and ‘without any adjustments’. Regarding their family life, most respondents thought a normal life consisted of children and having children ‘unconsciously’. Instead of living in line with these typifications, the respondents had to adapt themselves to the risk and the limitations that are related to living with MFS. For some of the respondents the diagnosis of MFS made them aware of these risk and limitations which made it a fateful moment, while for others it meant that they could categorize the consequences they had already experienced. One can conclude that although respondents were not always conscious about it, MFS affected different aspects of their life which resulted in another life than the respondents had expected based on their typifications. For some aspects in life, it took most respondents several years to accept that one could not live in line with these typifications, while for other aspects the respondents came up with new typifications. In sum, MFS affects the directions of the lives of the respondents.

Off the Line: Analysis of the Effects of MFS on Social Interactions and Relations

The previous chapter analyzed the longer-term effects of MFS on the lives of the respondents and the way MFS influences the direction of their lives, while this chapter focuses on the interactions with other people. Goffman (1967) developed an analytical framework for everyday encounters where he points at face to face or mediated contact between two or more participants. According to Goffman, everybody acts according to a particular 'line': "That is, a pattern of verbal and nonverbal acts by which he expresses his view of the situation and through this evaluation of the participants, especially himself" (Goffman 1967:5). Although a person does not always consciously take a line, the participants in the conversation will assume he has chosen this particular line and they act according to the expectations and values they attach to this line. So, the actor must always take into account the impression he gives with his line, because the participant reacts on this impression.

If people consciously choose a particular line, they always want to give an image of themselves that they are capable to act as desired in the particular situation. They will try to act as the situation desires them to act. They may claim positive esteem and status for themselves by acting in a line that others assume he has taken. This is called *face* by Goffman: "Face is an image of self delineated in terms of approved social attributes" (Goffman 1967:6). Besides trying to present a positive image of themselves, people also attach feelings to the face of the other participant in the encounter. When the face presented by the other participant is not in line with the expectations of the viewer, this viewer may feel bad or feel hurt. While on the other hand, when the face of the other participant is better than the viewer expected, he might feel good.

As said, a person will always try to act in accord with a line he thinks is suitable for the situation. The face that is attached to this line is a social construct: it depends on the rules of the group and the specific situation which line is expected and which face is appropriate. People maintain face when they choose a line that is appropriate for the situation: other participants will have positive judgment about the line and face. If this happens, people are mostly not aware of face, because it "is diffusely located in the flow of the events in the encounter" (Goffman 1967:7). On the other hand, people can be out of face when the line they choose, is not in accord with the line that is expected in this situation. When this happens, people often tend to feel ashamed because it damages their image of being an effective social actor.

As in all theories of Goffman, intersubjectivity plays an important role. People are aware of the expectations of others, and try to act in accord with these expectations. At the same time,

these others are also aware of these expectations and they too try to act in the expected way. These expectations are dependent on the specific situation and on the participants involved.

This chapter will analyze whether and how the respondents feel that MFS affects their social life. Do they feel they are sometimes out of face as a consequence of MFS and if so, is this in particular situations? How do they cope with this? And what are the consequences?

Social relations

In the previous chapter it already emerged that the respondents experienced several limitations which influenced the direction of their life. The final result of those limitations is that some of the respondents felt that they lived in a smaller world than they would without MFS. This section will explore this social smaller world in more detail. Compared to the previous chapter, this chapter explores this issue in more depth and analysis in more detail situations in which respondents experienced life in a smaller world.

To start with, nine respondents recounted that as a consequence of MFS they had to quit certain social activities, or that they sometimes had to cancel social appointments. This gave some of them the feeling that they had less contact with friends than they desired. If they could, they would have more contact with their friends and some stated that they would have more friends if they would not have MFS.

“Sometimes, when you are very tired, you have to leave your social life for what it is and say: sorry, but I am not going to make it.” (R4)

“As a consequence of my visual impairments, I did not really like going out when I was young. When I saw a nice girl, it was possible that I would not recognize her the next time. In advance I was already thinking how I should deal with that. I will not say it was all because of Marfan, but it was a major reason that I did not like it and that I never had a relationship. The chance to meet someone is smaller. Or at least less easy.” (R8)

Moreover, four respondents had severe visual impairments and for three of them these impairments had social consequences. According to these respondents, it was harder to communicate because they could not see expressions very well, or they did not even recognize people. This could lead to misunderstandings between themselves and the other person, or even irritation by the other person. To take it back to Goffman (1967), the respondents felt that the line of their behaviour was not in accord with the situation. This was hard for them to change, because they could not see the situation and as a consequence it is hard for them to maintain face and show behaviour in the right line.

According to Goffman, people who are out of face feel ashamed and may become insecure because they doubt whether they can be a good participant in the future. This turns out

to be true or used to be true for some of the respondents. Eight respondents recounted they were not always sure how they were expected to behave in certain situations or how they should express their thoughts and feelings. For most of these eight respondents, this used to be true especially in their youth and during their adolescence. As a consequence of this insecurity, they referred to how they kept themselves 'in the background' in many social situations and how they were reserved when they met new people. Almost all respondents who referred to having felt uncertain had severe external deviations¹⁹.

"At Secondary School I used to withdraw myself, I fell behind on the others. Other people used to go out and do fun things, but I did that a lot less. My circle of friends was smaller, I think." (R14)

"When I met new people, I used to feel uncertain. I tried to withdraw myself into a form of social isolation." (R16)

Another effect of MFS on the social relations of the respondents is the issue of guilt. In the previous chapter the issue of guilt was already addressed in relation to having children. In this chapter it turns out that some of the respondents, mostly women, felt guilty towards their partner, family or friends. These respondents felt guilty because they thought they were not the partner, child or friend they should be. They did not live up to their own expectations of their role and this made them feel guilty. One can say that if they could, they would behave according to another line and they would maintain face in accord to this other line. Unfortunately they are not able to behave according to this particular line, for example as a consequence of fatigue.

"That bothers me, that it affects me so much. I mean, you want to be a good partner in some way. Not somebody who is tired, or is not able to do something. I mean, that is an extra burden on your partner." (R5)

Whereas for these six respondents the relation with friends, family or a partner is a source of guilt because they cannot behave in accord with a particular line, for five other respondents this 'group' of relations was a source of reassurance. These respondents suggested that within this group they can be themselves.

"I know my friends already for a long time. At least one friend I know all my life, he does not know me any different. I can be myself when I am with him. The same is true for my family." (R6)

If one interprets this data in line with Goffman's theory on interaction rituals, one can conclude that these respondents know that they cannot always act according to the line of behaviour that is desired in a particular situation. But when they are with their family, friends or partner they do

¹⁹ In the next chapter, on the analysis of stigmatizing experiences, the issue of uncertainty will be further addressed.

not feel ashamed for this being out of face, because these people know about the cause of being out of face.

This might be related to the distinction Goffman makes between strangers and the people we know. To be able to act in line with the 'right' line, one needs to know what the right line is. When people meet strangers, they need to find out what line of behaviour is regarded as the right line by these strangers. To estimate what these new people regard as the right line of behaviour, people try to categorize these new people based on the signs and gestures these new people use. Some people feel stigmatized because they feel they are categorized on basis of their attributes, but these people try to categorize other people as well. By categorizing strangers, one tries to estimate what line of behaviour is the 'right' line of behaviour.

That the respondents feel more at ease of being out of face with family, friends and other relatives is because they are aware of themselves being out of face and they know that those people understand what is the cause of being out of face. When the respondents meet new people it is harder for them to know what these new people regard as the right line of behaviour and consequently the respondents are unsure whether they are in or out of face. Furthermore, these new people do not know why the respondents do not always show the right line of behaviour and thus this behaviour is not legitimized.

On the other hand, it explains at the same time why the respondents have feelings of guilt toward the people that are closest to them. Because these people are so close to them, the respondents know exactly what line of behaviour is suitable when they are together with these people. Unfortunately, they are not always able to show this line of behaviour and this results in feelings of guilt.

New activities and social relations

Although the above analysis may suggest that MFS has only negative consequences on the social lives of the respondents, this is not true for two reasons. First, MFS was not reported as affecting the social lives of all the respondents. Four respondents stated that their current and their past social lives would be the same if they did not have MFS²⁰. Second, it turns out that MFS is a source of new activities and new social relations as well. Nine respondents recounted that they undertook certain activities now that they would not undertake if they had no MFS. Some of

²⁰ One of these four had both severe inward and severe outward attributes, two had severe cardiac problems and one had both milder inward and both milder outward characteristics.

these can be seen as adaptations to their old activities²¹. Most of these new activities involved contacts with new people as well.

“Without Marfan, I would never have been in touch with wheelchair basketball. And it is great, because I found my circle of friends in it. Or at least, expanded my circle of friends.” (R2)

“I swim a lot nowadays. I quit basketball and tennis. The same is true for alpine skiing. Now I do another kind of skiing, which I probably would not have done that otherwise.” (R10)

Another source of new activities and new contacts is the patients association CMN. Besides organizing every year a Day of Contact, CMN organizes every year a family day, a weekend for women and a weekend for men. Ten respondents said that these organized days were a source of new contacts. Besides visiting these organized gatherings, four respondents did or had done voluntary work for the association. Three of them stated that this voluntary work offered them a lot of rewarding contacts with other volunteers.

“ I have been a member of the association for already 30 years. I know people via this association and these contacts are very precious to me. Besides information, this is one of the most important aspects of the association for me.” (R8, volunteer at CMN)

“I became a member because I wanted to get in touch with other people with Marfan. I wanted to know how they deal with things. (...) I went to the Wild Women Weekend and I had so much fun.” (R7)

Whereas these ten respondents praised the social aspect of these organized days, six other respondents slightly disagreed with them. According to three of them, people were so close that it is hard for others to fit in. Furthermore, three respondents had visited this Day of Contact in the past, but they did not want to be part of the group. They felt that a lot of other members were complaining about how bad it all was, while they did not feel like that. These respondents did not want to dwell upon the situation too much and just deal with it.

“You know it is a certain group. Some people know each other for years and it really is a specific group. I am not in it and I do not feel like I want to dive into this group. I do not think it is easy to fit in this group.” (R14)

“I like to get my information there, but I do not want to be part of it. I do not feel like I fit in. I do not want to fit in, I guess. I do not want to be a patient. I feel like they separate themselves by pointing on their limitations and I do not want that.” (R18)

“I feel like people there complain about so many things. I do not like that. Just act normal... (...) I am not someone who dwells upon these kind of things.” (R19)

²¹ For example, three respondents now did voluntary work instead of having a paid job. Another example is of a respondent who decided to play wheelchair basketball when he was not able to play ‘normal’ basketball anymore.

Meeting new people and other daily interactions

The respondents were asked how they felt when they met new people and whether they were influenced by MFS when they met new people. Nine respondents said they liked getting to know new people and according to them, MFS did not play any role in this. Furthermore, two respondents said that nowadays they liked meeting new people and they do not have any problems with that, while in their youth this had been harder for them. When they were younger, they were more insecure and they did not know exactly how to act when they met new people.

As adults they had adapted to the situation:

“I think in the past my life would have been different, if I did not have Marfan. It would have been easier to make new contacts and to maintain these contacts. But nowadays I would not say that. I think it does not make a difference now anymore.” (R13)

Although the next chapter will analyze this in more depth, three respondents felt that they were judged on their appearance when they met new people. In Goffman’s words: they felt that their appearance was judged by others as an important aspect of their line of behaviour, despite whether this was relevant for the situation or not. As a result these respondents felt insecure and were extra aware of their own appearance.

“On these moments I am very self-conscious. For example I am aware of my posture. I am not aware of Marfan, but I am aware of myself. So, when I meet new people, I am very self-conscious: how do I look, what impression do I give, what is my appearance?” (R14)

Besides feeling judged on their appearance, two of these three respondents also felt that it was more easy for them to interact with people who have MFS or another deviation than with people who have no disease at all. Three more respondents stated that it was easier for them to interact with people who were more or less similar to them. Once again, this can be analyzed via Goffmanian theory on interaction rituals. They feel more at ease with people who show certain similarities to themselves, because they know these people are not able to act according to the right line themselves, or because they expect these people to understand why they behave according to another line. Because these people are like-minded, respondents feel they do not really lose face.

“I think it is easier to interact with people who show similarities to you. (...) I think it is easier to get accepted when you are closer to the norm. It is easier then to find people who are similar to you. The more you deviate, the more tough it is to find people who are similar to you.” (R1)

Conclusion

In conclusion, one can state that the right line of interacting plays an important part in the social lives of the respondents. On the one hand there are respondents who feel that MFS does not bother them very much. One can interpret from their accounts that they, unconsciously, feel they can show in most situations the desired line of conduct and as a consequence they maintain face. They do not feel that MFS hinders them in their interaction with others. On the other hand there are others who feel they are not always able to show the right line of conduct, as a consequence of pain or fatigue. This leads to feelings of uncertainty, miscommunication and feelings of guilt towards the persons who are closest to them. At the same time, some respondents told of how they felt that when they are with the people closest to them it was not a problem that they could not show the right line of behaviour. The same is true for the contacts they have with people who are in the same situation.

So, the ones who are not bothered by MFS in their behaviour are not aware of their line of behaviour. The ones who are limited by MFS are more conscious of their line of conduct and their social lives are more influenced by MFS. This might be explained by the severity of the problems of the four who stated that their social lives would be the same without MFS. Three of them were already operated on their heart. This might be labelled as severe inward problems, but despite these operations they did not feel that their cardiac problems influenced them during their daily life. One of these three respondents recounted that he could not always join all activities, but he adapted himself to this situation and found other activities. As a result he did not feel his social life was really bothered by MFS. The fourth respondent has both milder inward and milder outward attributes. Although she felt pain and fatigue, according to her this did not influence the way she interacted with other people.

On the other hand, those respondents who were more conscious of their line of behaviour and the effect MFS has on their social life, had severe outward characteristics or severe problems of fatigue and/or pain. The respondents who were hindered by severe fatigue or pain had to cancel social appointments sometimes, while the respondents with severe outward problems were, at least when they were teenagers, more inclined to keep themselves 'in the background'. As a consequence of not joining certain activities, they were aware of themselves being out of face.

The most important finding of this chapter is that some respondents could not live in line with their typification of good social actor. On the one hand, they could not show the line of behaviour that is regarded by themselves as the right line. They felt that their behaviour was not suited for the role of a good partner, friend or child. On the other hand they felt that they could

not always show the right line of behaviour that others expected of them. This was especially true in relation to interactions with strangers.

Threatened Identity: Analysis of Experiences of Being Stigmatized as a Consequence of MFS

According to Goffman (1963) stigma is a discrediting characteristic. This characteristic renders this person different from the norm and as a consequence other people will negatively judge the beholder of this characteristic. This attribute may be visible on the spot (discredited) or may be hidden (discreditable). This distinction between discredited and discreditable characteristics is made to illustrate that stigma is not about the attribute itself, but about the social relations in a specific situation. The stigmatized person is mostly aware of the fact that he differs from normal and this might result in feelings of shame and eventually even in self-stigma (Goffman 1963).

This chapter will analyze whether the respondents feel stigmatized on basis of their MFS characteristics. This will be analyzed on the basis of the characteristics that Goffman ascribes to the stigmatized: feeling (treated) different(ly), feeling ashamed and insecure and feeling unequal to 'normals'. Furthermore, the chapter will analyze how they deal with their discreditable attributes.

Feeling (treated) different(ly)

To start with, almost all respondents show one or more of the characteristics that Goffman ascribes to the stigmatized person. For some this only holds true in the past, while for others these feelings are still current. Thirteen respondents clearly stated that they felt themselves to be different. The reason for this being different is mostly not MFS itself, but the consequences of MFS. Most respondents with more severe external characteristics felt different because of their appearance. This was true for six respondents. Also three respondents who had less severe outward attributes felt different because of their appearance. In the following quotes, one sees that these respondents take over the judgments of others, resulting in self-stigma. Furthermore they show feelings of insecurity and the feeling not be accepted.

“I always had that idea: I am different. I always thought people would think I am weird, because I look differently, because I am tall and skinny. (...) In my youth maybe only ten incidents happened of people making comments. For me this was a confirmation: indeed I am weird, I am not supposed to be here, I am ugly and I have to hide.” (R16)

“Before I was operated on my sternum, I felt so insecure. Especially on the beach, I always felt people were staring at me. When you are a child and other children are staring at you and talking about you... That happened to me several times and that affects you so much... You start to feel very insecure. You feel that you are different and you are looked at as if you are not normal, as if you are some kind of freak.” (R6)

Another reason why some respondents felt different is that they were more consciously living their life compared to other people. They felt to be more consciously about their body, about certain activities and about things that other people take for granted, like a sunny day.

“Do I feel different? Maybe...you know, you cannot deny it, so do I feel different? Yes, I think so. I think I am more concerned with myself. Well, not with myself, but with my body. In a practical way. I mean, with my artificial valve I hear literally every beat of my heart. I think about that sometimes and I think that makes me feel different, yes.” (R11)

“I do realize it. I feel that other people think more easily about certain things, while I myself have to take more issues into account. Things are not naturally: the future, children... When you are young there is nothing you want more than being average.” (R13)

Respondent 11 from the first quote above did not show any other signs of feeling stigmatized except from feeling different. This feeling different was not experienced as negative by this respondent. Although he did not feel stigmatized himself, he said that he could imagine that other people felt that way. He thought that for the ones with more severe problems, MFS could be a label that marks one for the rest of his or her life. On the other hand, respondent 13 showed in her interview more characteristics of the stigmatized: she felt different in her youth because of her appearance, she showed feelings of insecurity and she wanted to be judged on her personality and not on the fact that she has something that is deviant.

Furthermore, her quote above is in line with the analytical framework of Giddens (1991) in his analysis of modernity and self-identity. According to Giddens humans know what they are doing and why they are doing it. In a process of reflexivity people monitor the social conventions that they produce and reproduce in everyday life. This reflexive monitoring happens non-consciously²² because people take all these social conventions for granted. When these social conventions are breached, “the fragility of the natural attitude” becomes evident (Giddens 1991:37). Only then people start to ask themselves existential questions, because they get more anxious when the taken-for-granted character of ‘the everyday’ is breached. This is parallel to the biographical disruption described by Bury (1982) and Schutz’s (1973) notion of becoming aware of one’s typifications when they are no longer applicable. What Giddens adds to this is his notion of increased reflexivity. People get more reflexive on their actions, when they can no longer take for granted the social conventions. This is what can be interpreted from the quote from respondent 13. As a consequence of MFS she can no longer take some social conventions for granted. According to herself this makes her more reflexive about the course of life than other people.

Besides whether they felt themselves to be different, the respondents were asked whether they felt to be treated differently. Six respondents clearly stated that they felt that they were treated differently in some situations. Two of them stated that they felt treated like they were pitiful. They felt that people saw them as a person with a certain condition, instead of seeing

²² See Giddens (1991:35) why he calls this non-conscious instead of unconscious.

them like an average person. Although processes of identification are explored in more depth in the next chapter, here it is noted that from the following quote one can discern that stigma is a threat to identity.

“First people see what you are taking with you and only after that they see who you are. So to speak, they do not ask what is your name, but they ask why do you have that chair?” (R17, at secondary school she used to have her own chair)

Three respondents described that in their youth they were treated differently as a consequence of their height²³. Furthermore, nine respondents felt that if people who are close to them know that they have MFS, they start to think for them. For example parents who told their children that they are not allowed to do certain things because this was dangerous for them. These respondents felt that other people reacted overprotective towards them²⁴. They did not like this because it made them feel that they were different from the persons who do not have MFS and that people acted towards them on basis of the fact that they had MFS. This is exactly what Goffman means by stigma and it is a threat to their identity: others identify them as a pitiful person with MFS while they do not like this. This is not in line with their self-image.

“You know...at a certain age you notice as a child that your parents are overanxious. This obstructs you with certain things.” (R19)

“Often I do not tell people I have Marfan because I am afraid they will start thinking for me and will say you are not allowed to carry this because it is too heavy for you.” (R9)

One last remark on the feeling of being different and feeling to be treated different, is that this was relatively more apparent with the respondents who had no family members with MFS. Out of the thirteen persons who clearly stated to feel different, eight had a spontaneous mutation of their genes and four out of six who felt to be treated differently had no family members with MFS. Finally, four respondents who did have family members with MFS told that they expected that they would feel themselves more an exception when they would be the only one in their family, while three respondent who were the only one expected to feel less different when they would have family with MFS.

²³ People always thought that these respondents were older and stronger than they actually were. One respondent stated that because of his height, he always drew more attention than the other children and as a consequence, at primary school when something happened he was the first to be punished.

²⁴ This mostly happened in the youth of these respondents, but some told that currently some friends also reacted overprotective.

Feelings of shame and insecurity

Another attribute that Goffman ascribes to the stigmatized, is feeling insecure and being ashamed. This is a result of stigma, because people often are aware of the norms and values in a situation and thus know that they are different from the ‘normals’. This makes it hard for them to interact with the ‘normals’ on an equal level. “The standards he has incorporated from the wider society equip him to be intimately alive to what others see as his failing, inevitably causing him, if only for moments, to agree that he does indeed fall short of what he really ought to be. Shame becomes a central possibility, arising from the individual’s perception of one of his own attributes as being a defiling thing to possess, and one he can readily see himself as not possessing” (Goffman 1963:7).

In the previous chapter the issue of insecurity is already dealt with, because some of the respondents felt insecure whether they showed the right line of behaviour. Here it will be analyzed whether the respondents felt uncertain as a consequence of stigmatizing attributes. Eight respondents told that they felt unsecure because of their appearance. Most respondents had more than one characteristic that made them feel uncertain. Three times this uncertainty was caused by a scoliosis, for four people their deviating sternum was a source of uncertainty. For four respondents another source of uncertainty were their thin limbs and finally two respondents felt uncertain as a consequence of their height.

“In the summer, when people ask me to come along at their sailing trip and everybody is in bikini or swim short, I do not feel at ease. I am so tall and thin... (...) I am not sure whether I feel insecure as a consequence of Marfan, I think everybody basically is uncertain. But when I have to get undressed, then I am. That bothers me. My arms are thin as matches.” (R18)

“In a group with people I do not know that well, I feel uncertain. I will always try to look for someone familiar. I think it is tough. For example, going to a congress bothers me. I feel uncertain because I am so tall, because I am different than other persons.” (R7)

Not only do the above quotes illustrate Goffman’s notion of the stigmatized feeling insecure, they also illustrate his point of stigma being dependent on the situation. Respondent 18 only felt uncertain when she had to get undressed, while respondent 7 felt less insecure when she is with people she is familiar with.

Related to the issue of uncertainty, is the issue of feeling ashamed. This too is a consequence of being familiar with the norms and values that are current in a social context. The respondents were directly asked whether they felt ashamed for having MFS. Only one respondent felt ashamed for having MFS, while thirteen respondents said they were not ashamed for having MFS. The most common argument was that it was not one’s own fault that he or she had MFS. They were not to be blamed and as a consequence they did not feel ashamed for having MFS.

Although these respondents told that they were not ashamed for having MFS, six respondents recounted that they were ashamed for one or more of their characteristics. They were all ashamed as a consequence of an outward characteristic.

“Well, now I start to think about it, but then it comes from deep... but eventually it is all related to Marfan, I am ashamed for my feet. Not because I have size 47, but because I have got hammer toes. You will never see me in flip-flops.” (R11)

The fact that the respondents who told that they felt ashamed did this because of their outward characteristics, confirms Goffman's point about the differences between discredited attributes and discreditable attributes. According to Goffman feeling ashamed is a lot more common for discredited attributes than for discreditable attributes, because the former are visible on the spot. As a consequence of this visibility the respondents with severe external characteristics are at greater risk that other people identify them different than these respondents identify themselves.

Outsiders

Another point that Goffman makes about the stigmatized is that it is hard for him to get in contact with the ‘normals’ like they are equals. Again this is a result of the fact that the stigmatized often is aware of the norms and values and knows that he differs from these norms and values. Closely related to this is Becker's concept of outsiders. According to Becker (1963) an outsider is someone who breaks the social rules of a group. It depends on the rules in a situation who's an outsider and who's not.

Three respondents told it was hard for them to find connection with others. This might have been in the past, with other children at school, or currently with colleagues for example. This points to Goffman's argument that it is hard to get in contact with others like they are equals.

“At primary school I could not join them, I was not allowed to participate in games. Actually I was excluded. Those things happened and it made clear for me that I was different. (...) Dealing with other people and making social contacts, I was not good at that. That was a burden for me.” (R15)

From the above quote it is clear that the respondent finds it hard to get in contact with others and that the respondent is aware of the fact that she is not part of the group. This is in line with both Goffman (1963) and with Becker's (1963) notion of outsiders. Although only three respondents said that it was hard for them to connect with others, eight respondents recounted experiences in which they felt themselves to be outsiders or in which they had to do their best not to be an outsider. In this last case, being part of the group was far from taken for granted.

“Well...you do not meet the expectations and the acceptance of others. You fall out of place because you do not look like them and thus you will not be part of them.” (R1)

“At primary school, as a consequence of my visual problems, I always had to sit in the front of the class. I did not like that. And I was not normal, I was not part of them, you know. Maybe misfit is a word that is too harsh, but I felt it that way.” (R8)

Discredited and discreditable

Goffman makes a distinction between attributes that are visible on the spot, called discredited attributes, and discreditable attributes that are hidden and that only will be stigmatizing when they are revealed. In the case of MFS the outward problems like scoliosis and abnormal length can be regarded as discredited, while the inward problems like cardiac problems can be regarded as discreditable. Some external attributes can be regarded as discreditable because they can be hidden by clothes²⁵.

It already turned out that the respondents with more severe external characteristics relatively more often felt different compared to the ones without severe external characteristics. This was also true for respondents with severe external attributes that could be regarded as discreditable because they could be hidden. Furthermore, these attributes also made them feel ashamed and uncertain. There was one exception from these findings. This respondent had a severe scoliosis and some other external attributes, but he had no feelings of being different or being ashamed and uncertain. This might be explained by the fact that he had been successful in his life, ‘despite’ MFS. For years he had his own company, he was married, he lived in a house which was bigger than average and he thought that one’s own attitude towards others defines how people treat you.

On the other hand, if one takes a look at the respondents who’s main MFS attribute was an inward problem, these feelings were less apparent. Those of them who had also outward problems did have these feelings, but less strong and less often compared to the respondents with severe external problems. It turned out that having MFS itself is for some respondents of this group an attribute that will be stigmatizing when it is revealed. Seven respondents made remarks on their job perspectives when it would turn out that they have MFS. Among these seven respondents, only one had severe external attributes, while the other six had less severe external attributes and mostly severe inward problems. Having MFS was regarded by them as a discreditable characteristic: they expected that if they would reveal at work that they had MFS, their career perspective would be negatively affected by it. In other words: when MFS would be revealed, these respondents expected to be identified as an unsuitable employee.

²⁵ One could think about a deviating sternum, thinness or deviating feet.

“There was a time when I used to think that it would not be good, especially for my career, to tell everybody that I was sick. Over time this had less influence on me because I became self employed and now it is not an issue for me anymore.” (R11)

“Once at a job interview I have honestly told what I have. Well, I still hear myself talking... it sounded pretty heavy if you tell everything. When I left that interview, I already knew I would not be hired. With my current employer I sometimes give little hints, but actually I think that is not good. I think I can forget a transfer to another office. (...) I am afraid that they will see me as a weak link.” (R9)

These two quotes show that having MFS is regarded by seven respondents as a stigmatizing attribute in relation to work. Furthermore, four other respondents saw MFS as a discreditable attribute. Two of them expected that they would be treated differently if they told other people they had MFS, while two of them had experienced this different treatment. For example, one respondent told that at the school of her children she had told that they had MFS. As a consequence, her children were treated ‘with panic’: with activities outside of school, there always had to be an extra person to keep an eye on her children and with school swimming her daughter needed to wear a special cap. At the same time, she knew that there was another child at that school with MFS. The mother of that child had not told that her child had MFS and this child was not treated differently. Remarkably, this child was, according to the respondent, in a worse condition than her own children.

Management of stigmatizing attributes

From the above analysis it would seem that having MFS itself and some characteristics can be regarded as discreditable attributes. Herman (1993) describes four different tactics of information control about stigmatizing attributes: selective concealment, therapeutic disclosure, preventive disclosure and political activism (see earlier theory chapter). Here it will be analyzed which tactics were used by the respondents regarding their discreditable attributes.

To start with, it turned out that for nine respondents clothing served, or had served in their youth, as a mask for their external attributes. This was true for those who regarded themselves as skinny, those with a deviated sternum and those with scoliosis. These respondents tried to cover their characteristic by wearing shirts with long sleeves and trousers with long legs. Furthermore, some avoided wearing open shoes or tight shirts. Some had specific tactics for specific body parts:

“My toes are ugly, but no-one ever said anything about it, because your toes do not stand out. But in the summers I often polish them red, because then you do not see them at all. So, yes I pay attention to it. (...) If I put polish on my nails you cannot see anymore that they are very weird.” (R17)

For some respondents, hiding their attributes consisted only avoiding some clothes, like skirts or short trousers, but others put much effort into hiding their attributes and presenting the ‘right’ image.

“I want to hide that I am so skinny. So bony and so skinny. I try to clothe in a certain way that people think ‘you are thin, but not skinny’. Often people do not notice that I am so skinny, but that is an effect of the way I dress myself. I put a lot of effort into that.(...) I buy very expensive clothes which make me look good. I turn my wallet inside out for that (laughs). The same is true for make-up. I do not use that much, but just in such a way that I look more healthy.” (R18)

The above quotes are both about the hiding of certain characteristics, but also having MFS itself is experienced as a stigmatizing attribute. It turns out almost all respondents used one or more of the tactics described by Herman. Nine respondents turned out to make a distinction between ‘safe others’ and ‘risky others’. The issue of trust plays an important role in this.

“I tell somebody if I think the friendship is worth it. And I have to trust someone. So, if it is someone of whom I know he easily tells it to other people, then I will not tell him. These are the two considerations: whether I trust that person and whether it is important to me that this person knows this.” (R13)

Out of the four tactics described by Herman, especially selective concealment and preventive disclosure are used a lot. Here the motives of the respondents to tell about MFS will be lined out. To start with, four respondents clearly stated that if they decided they did not want to say something, then they did not. Although the situation sometimes required a certain amount of openness, these respondents stated that they were in control. Furthermore, four respondents nowadays almost never told others that they had MFS, because past experiences told them that people started to treat them with pity. They did not like it and as a result of these experiences they did not tell people anymore, or only rarely.

“I do not tell people spontaneously. If people ask about it, I will tell probably, but I never initiate it. I am not a victim, so I do not want to behave like it and I do not want to be treated like it.” (R19)

These two ‘motives’ can be headed under selective concealment: these respondents mainly conceal the fact that they have MFS and only when they think it is alright to tell, then they talk about it. The motives that now follow can be headed under the tactic of preventive disclosure. To start with, five respondents said that they would disclose the fact that they have MFS in situations in which it is necessary. For example, when they requested a certain facility, or when it was needed that other persons in a situation could take the limitations in account.

“If someone does not know about it, he cannot take it into account. You are the one then to tell him. You should do that yourself.” (R4)

Strongly related to this motive is the motive displayed by eight respondents: they explained it when it was useful for themselves or for the people they told. For example, when they can share the experiences they have with some medication or operation with a person who will have this in the future. In these cases the respondents were not afraid that the revealing of MFS was an threat to their identity.

“Well, it might be that you get in situations in which you have to speed up the tempo, or you have to give extra energy. Then it is more useful to tell it in advance that you have certain limitations and that you will take them into account yourself, than that you have to quit at the moment itself. Then you are of no use.” (R14)

It has already been mentioned that the issue of trust was important to a lot of respondents. This was especially true in relation to the last two motives mentioned. Five respondents said that they disclose about MFS if people start to ask questions about it and they feel they can trust these people. The last motive is strongly related to this: three respondents said that they will tell about it when it comes up in a conversation. How much they tell about it depends on their relation with the person they converse with.

Conclusion

In this chapter it turned out that almost all respondents showed one or more characteristics that Goffman ascribes to the stigmatized person. Feelings of being (treated) different, feelings of shame and insecurity and the feeling of being an outsider appear to be very common for the respondents. It turned out that these feelings were more common for those with severe external attributes and for those who had no family with MFS.

These results are in line with Goffman’s (1963) framework. Severe external attributes are visible on the spot and consequently can be regarded as discredited attributes. These cannot be hidden and as a result were more stigmatizing than inward problems, which could be hidden. In addition, the factor of presence of MFS in the family is also explainable with Goffman’s framework. An attribute is stigmatizing when it is different from the norm. If someone has family members who have MFS as well, then having MFS is normal within this family. If someone is the only one in the family with MFS, then living with MFS is both within the family and outside the family (thus in other social contexts) different from the norm. With this in mind, it is not a surprise that the respondents who are the only one in their family have more feelings of being (treated) different, feelings of shame and feelings of being an outsider compared to respondents who have family members with MFS.

Furthermore, it turned out that for most respondents not only the severe external attributes but all the external attributes were regarded as stigmatizing, despite of the fact that

some were hidden by clothes and thus were a discreditable attribute. Finally, MFS itself was regarded as a discreditable characteristic, especially in relation to someone's career.

In sum, one can conclude that all respondents experienced that, in one way or another, MFS has an influence on who they were. This might be in their own eyes, or in the eyes of other people. If it was in their own eyes the respondents felt that they differed from other people and from their own typification of what one should be like. If it was in the eyes of others, the respondents felt that they were treated differently because of MFS or its consequences and that they differed from the typifications of others. In both ways, MFS was a threat to their 'desired' identity.

New Typifications Lead to Normalization: Analysis of Processes of Identification as a Consequence of MFS

In the previous chapters it was noted that all respondents experience several limitations in both day to day and longer-term social interactions. MFS turned out to have an effect on the directions of the lives of the respondents. Furthermore, all respondents made remarks that can be labelled as experiences of a stigmatized person. For a lot of respondents MFS threatened their own typification of a ‘normal’ person, or the respondents felt that it threatened their identification as a ‘normal’ person by others. This may give the impression that all respondents live a tough life. When one could read the whole interviews, one would get another impression. Although ten respondents recounted that MFS was a determining factor in their life, most respondents were satisfied with their life and they did not experience their life as ‘tough’. This chapter will explore this apparent paradox between on the one hand the stories of the respondents which give the impression that MFS makes their life tough, and on the other hand the statement of most respondents that they live well. In developing an understanding of this paradox, the theory of Merleau-Ponty (1962) on embodiment will be used. Furthermore, this chapter will analyze whether these respondents identify themselves as MFS patient.

Embodiment

In the previous chapter it turned out that almost all respondents showed one or more characteristics that Goffman ascribes to the stigmatized person, but not all of them felt stigmatized. It turned out that some of the respondents who showed attributes of the stigmatized, did not experience this attribute as a negative experience. For them, MFS did not feel as threatening their identity. This might be explained by the theory of Merleau-Ponty (1962) on embodiment.

In short, embodiment is a process in which a person becomes less and less conscious of his physical differentness: embodiment means “taking one’s body image into account unconsciously” (Iwakuma in: Corker & Shakespeare 2006:81). According to Goffman (1963) and Becker (1963), the stigmatized person is aware of the fact that he is different than the group of which he cannot be part. In the process of embodiment, a person gradually loses this awareness. This process is complete when one is totally unconscious of his differentness and his own bodily image. When a person fully embodied his attribute that makes him different, this attribute became part of his identity (see the theory chapter). One can state that when an attribute is become part of one’s identity, it means that this attribute is regarded as normal by this person.

When one analyzes the interviews with the respondents with this in mind, it turns out that a lot of respondents have embodied MFS and some or all of their attributes.

The data from fifteen of the respondents seemed to reflect such processes of embodiment. MFS has become normal for them and only at some specific situations did they consciously think about MFS. In previous chapters it is already suggested that the respondents experienced several limitations and had to make some adjustments as a consequence of MFS. Most respondents recounted that they made these adaptations unconsciously and as a result of these adjustments they could live their life the way they wanted. As a result, they were even more unconscious about MFS. These respondents learn to live with it and as a result MFS became an unnoticed part of their life: instead of being a threat to their identity, MFS became part of it.

“At a certain moment it starts to belong to your life. It becomes part of your life and you do not know any better anymore.” (R12)

These respondents were only aware of MFS in specific situations. Six respondents stated that they were only consciously thinking about MFS when they, or in some cases their children, visited the hospital for their check-ups. Furthermore, two other respondents told of being conscious of MFS in relation to other medical settings.

“No, I am never really conscious that I have Marfan. Oh yes, when I have to go to the hospital. In the days before a hospital visit, I always feel everything (laughs). Yes, then I am very conscious about having MFS.” (R3)

“Well I am not really conscious of Marfan, but I am sometimes conscious of the consequences, like my height and the trouble that gives in an airplane. But I am conscious about it when I go to the dentist or my GP. When there is someone new, I always ask whether they are familiar with Marfan and what they know about it. If necessary, I give them a brochure.” (R10)

Although having MFS is embodied by most respondents, for some respondents there is a difference between the embodiment of the inward and the outward characteristics. Five respondents said that having MFS and the consequences had become part of their life, but these five still had difficulties with their external characteristics. They tried to hide their external attributes, or wanted to correct them if possible. The following quote illustrates that embodiment is a process. This respondent knew since a few months that he has MFS and now he had an explanation for why he looked like he did, he could accept this. As one can discern from this quote, the respondent thought he could learn this over time.

“I do not really think about it. Taking medication has become part of my morning routine, I do not pay much attention to it. (...) Well, you know situations like a swimming pool, the beach, terraces, hot weather, in the bathroom, getting undressed, you name it. In those situations I am very aware of my appearance. I have to work on that and I am the only one who could do that, right?” (R16)

Furthermore, two respondents with visual problems stated that they were used to these visual limitations, but they were not used to the insecurity they experienced during their whole life as a consequence of their MFS-characteristics. In other words, they embodied their visual impairment but they did not embody the social consequences of MFS. Finally, two respondents stated that they could never really 'release' it. Although they were used to reactions of others and used to the limitations they experienced and these had become part of their life, these respondents felt they could never set themselves free of the thoughts about it.

“Taking medication is one of those things that draw attention to the fact that you have Marfan, while you are trying to free yourself from it. All those minor details that force you to face the facts. Yeah, that is the hardest and most frustrating thing, that your pointed to it every time. You can never set yourself free of it.” (R1)

These two respondents were siblings and their shared history might explain this feeling that they could never set themselves free of it. Their father had died as a consequence of cardiac problems caused by MFS when both respondents were young. In contrast to other respondents who had lost a relative, these two respondents were adolescents when their father died. According to Erikson (1971,1980), the development of identity really gets started when people become adolescents. These two respondents experienced the death of their father in this period. It might be that this has had such a major impact that these two respondents are always aware of the consequences of MFS.

Identification

When the process of embodiment is completed, an attribute has become part of one's identity (see earlier theory chapter). The analysis above demonstrates that having MFS was fully embodied by the majority of respondents in this study, while some of them embodied only certain attributes that are a consequence of MFS. Now it will be further analyzed whether the respondents identify themselves as a MFS-patient. With this it is meant that the respondents felt themselves to be part of a larger group and whether they felt that MFS is defining for who they are.

According to Traas (1990) three aspects are important regarding identity: personal history, one's relation to the world around him and finally the dynamic of a person being able to make their own decision in a changing world. Erikson (1971, 1980) too points to this relational aspect of identity. According to him identity is developed in the interaction from a person with his social surrounding. Finally, both Erikson and Honneth (1996) pay major attention to the role of acceptance and recognition in the establishment of identity.

First, it will be analyzed whether the respondents show any signs of identification as an MFS patient. Furthermore it will be analyzed whether the respondents regard MFS as an ‘illness’ and whether they identify themselves as a ‘patient’ (later on it is explained what is meant with these concepts). Finally it will be analyzed what the role of personal history is.

Fourteen respondents made remarks that are related to identification. In these remarks five regularities could be discerned. To start with, five respondents clearly stated that MFS was not their identity. They did not want to be seen as an MFS-patient, because they were more than MFS; MFS is what they had, not what they were. Furthermore, they did not feel themselves to be part of the group of MFS-patients as a whole and they did not want to be part of this group either. They dissociated themselves from this group.

This might be explained by the relation between categorizing and control. According to Giddens (1991), Beck (1992), Douglas (1962) and Lupton (1999), people make use of categorizing to get a sense of control over things that are unknown to them. When people are confronted with situations they are unfamiliar with, in this case bodily changes, people feel insecure. By labelling these new phenomena, people categorize them, which results in a sense of control. These five respondents might refuse to identify themselves as a patient, because they might feel that by categorizing themselves, they lose control over the image other people have of them: they expected that other people attach certain meanings to the label of MFS. In other words: when they identify themselves with MFS they feel this as threatening the identity they ascribe to themselves.

“I bond myself to people with who I am and not what I have. (...) I want to enjoy life and I want that people judge me on who I am. You can say: okay but who you are is a weak person, who looks tired without make-up. But no, I want to get everything out of life and I have my qualities beyond Marfan.” (R18)

“I am a member of the association, but I wanted to stay anonymous. I did not want to walk around there like a Marfan-patient and I do not want to be seen like that. Although it dominates your life, it does not dominate the way I think.” (R12)

In contrast with the five respondents mentioned above, another group of respondents did identify themselves with the larger group of MFS-patients. These respondents used phrases like “we are” and “us”. Four respondents identified MFS-patients as a group and they regarded themselves to be part of this group.

“I have got a high and small palate. But I think we all have that, such teeth.” (R5)

“I do play golf. This sport is almost made for people with Marfan: you have to be flexible in your joints, well...that is what we are, all of us, you can walk instead of run, you are outside... They call it a sport for elder people, but we qualify ourselves perfectly for it.” (R11)

A third trend is that four respondents acknowledged that MFS has such a major impact on their life, that it has become part of who they are. Although in total ten respondents told that MFS had a major impact on their life, these four stated that this had an effect on their personalities: without MFS they would have been a different person. This points to the importance of personal history, as mentioned by Traas (1990). The consequences of MFS in the past, made who they were nowadays. Mostly this had to do with the reactions of other people, which in its turn points to the relational aspect of identity establishments as pointed out by Traas and Erikson (1971,1980). This confirms the findings in the first analysis chapter that MFS has an impact on the direction of the lives of most respondents.

“Look, I am not thinking that often that I have Marfan, but of course it made me who I am. In that sense, it affects you every day. Not that you are consciously thinking about it, but it made you who you are. Your character and such things.” (R6)

“I think it is important to show who you are. And I have Marfan. One way or another, whether I want it or not, I do have it. (...) It determines who you are and what you are able to do and it is determining for me too, yes.” (R17)

Fourthly, four respondents identified themselves with other MFS-patients on an emotional level. By this is meant that these respondents felt that they were connected to other MFS-patients because they share certain experiences. Although they were aware that every person with MFS is unique, they felt that they were connected to other MFS-patients because they had something in common.

“I think that is a good thing of Marfan, there is a connection between the patients as a whole.” (R10)

“That is funny, you immediately have a connection. And it feels like that too. You know, although you do not know the person, because he has MFS it feels like family. Come on, it is alright, it feels good. I does not make any sense (laughs), but this is how I feel it.” (R5)

Finally, four respondents doubted whether they were the way they were as a consequence of MFS, or that it was just their character. They felt that one should not regard everything as a consequence of MFS. Although these respondents acknowledged that MFS affected their life and although they made remarks that gave the impression that they identified themselves with MFS, they felt they had a personality that was not completely due to MFS²⁶. Again this might be related to the notion of categorization and control. It appears that these respondents felt that if they identified themselves as MFS patients, they were out of control of what their personality looked like. By stating that not every personal-characteristic is due to MFS, they felt that they were themselves more in control of who they were.

²⁶ One of the respondents who made a statement like this, also stated that he was more than MFS (first category mentioned above).

“Sometimes I think: is that a consequence of Marfan, or is it because of the personality you have yourself. It is hard to tell, you cannot just say ‘this is because of this and this is because of that’. I think it is an interaction between those two.” (R14)

“At such a ‘wild women weekend’ I try to compare myself to the other women to discover what is Marfan and what is my own personality?” (R9)

Illness and the patient

From the above analysis it turned out that five respondents distanced themselves of identification as a MFS-patient, while the other respondents showed signs of identification as MFS-patient. Now it will be analyzed whether the respondents regard MFS as an illness and whether they identify themselves as a patient. Important here is the difference between ‘disease’ and ‘illness’. Within medical sociology there is a major research area which is concerned with the social construction of illness. A basic premise of social constructionism regarding illness is the conceptual distinction between disease and illness (Conrad and Barker 2010). The concept of disease refers to the biological condition, while the concept of illness refers to the social meaning of the condition (Eisenberg 1977). “Social constructionists emphasize how the meaning and experience of illness is shaped by cultural and social systems. In short, illness is not simply present in nature, waiting to be discovered by scientists or physicians” (Conrad and Barker 2010: S68). The next section will analyze whether the respondents experience MFS as a ‘biological condition’ or that they bestow meaning to MFS and regard it as an illness.

The respondents were directly asked whether they regarded MFS as an illness. In the Dutch language there is no difference between illness and disease, these concepts are both translated in the word ‘ziekte’. To discern whether the respondents themselves made a distinction between disease and illness, they were asked about their motivation to regard MFS as a ‘ziekte’ or not. This question, whether they regarded MFS as an illness, was a difficult question for many of the respondents. Eleven respondents did regard MFS as an illness and used different arguments for this standpoint. One respondent said that if she would not have had MFS, she would not end up in the medical circuit. Furthermore, five respondents argued that MFS had several impetuous consequences for their life, that it was an inevitable part of their life. Three respondents argued that because it is called a syndrome, it is a collective noun of different illnesses. One respondent said that she could feel really sick as a consequence of MFS, while finally one respondent said it was an illness because it leads to several complaints, it sometimes make you feel not healthy and one could die from it.

“To start with, I think it is an illness because it bothers me all the time. And I think an illness gets worse over time and this is true for Marfan. Furthermore, it is so predominant in my life, although I try to make it as liveable as

possible. I think I succeed in that because I am happy and I can do what I want. But within in my boundaries, so in a certain amount it restricts me. Always and everywhere. So yes, I think it is an illness.”(R17)

On the other hand, seven respondents did not regard MFS as an illness. One respondent argued that it is not an illness because an illness is curable, while MFS is not. Five respondents said it was not an illness because, although they experienced several limitations, they never felt ‘sick’ as a consequence of MFS. Finally, one respondent regarded it more as an explanation for how she looked and how she behaved.

“I told them something went wrong with the development of their genes and as a consequence at several point weird things had happened which could hinder them sometimes. But you are not sick of it. You are sick when you have fever and when you feel nauseous.” (R3, answering the question how she explained her children what MFS is)

Finally, one respondent did not think it was useful to answer this question. According to him, it did not matter what name you gave to it, he felt what he felt and a label would not change this. This was a dilemma faced by more respondents. They were aware of the connotation of a label like ‘illness’ and some felt that this label was too heavy for their situation. From this difficulty, one can discern that the respondents were aware of the difference between illness and disease, but the Dutch language causes a problem here. Some did regard MFS an illness, because they noticed several consequences of it and they bestowed meaning to this. On the other hand, some respondent did not regard it as an illness because of the meaning they attach to the Dutch word ‘ziekte’ and according to these respondents MFS was not an illness, because it had not the same meaning as the word ‘ziekte’. Finally, some respondents regarded it as a biological condition and labelled this with the word ‘ziekte’, but it appeared that they regarded it as a disease²⁷.

Furthermore, a lot of respondents doubted between different labels to subscribe their situation. Most used labels were ‘ailment’, ‘construction error’, ‘deviation’ and ‘handicap’. The label of ‘handicap’ was a controversial one. On the one hand, some respondents used this word because they felt there was no remedy for their situation and there was not much that they could do. On the other hand, some respondents strongly disliked this word, because it made them feel passive. This contradicted how they experienced living with MFS, because they felt that they were in control of how their life looked like, despite of the limitations faced. These respondents dealt with MFS in a proactive way: they felt they should make the best of it despite MFS. Although MFS influenced the direction of their life, they felt that within these borders they could decide what their life looked like. Again this is explained by the notion of control: some respondents felt

²⁷ Here one can discern the distinction Schutz (1973) made between first-order constructs and second-order constructs. Where the respondents used the word ‘ziekte’ (first-order construct), the researcher made the distinction between ‘illness’ and ‘disease’ while interpreting the statements of the respondents (second-order constructs).

that they were out of control and labelled their situation consequently as handicap, while others refused to use this label because it made them feel that they were out of control while they were not.

The label of ‘construction error’ was used by some respondents to make sense to their situation. “Sense making being understood to refer to the process of interpretation and meaning production whereby people reflect on and interpret phenomena and produce intersubjective accounts (Leiter 1980)” (Currie and Brown 2003:564). By labelling MFS as a construction error, it made sense to these respondents why they had the appearance they had and why they behaved the way they did. By objectifying their situation as a medical fact, their situation made sense to them and they felt they were not to be blamed for their situation. This label was mostly used by people with severe external attributes.

Before turning to aspects of personal history, this last section will analyze whether the respondents identified themselves as ‘patient’. The respondents were directly asked whether they felt themselves to be patient. The purpose of this question was to research whether the respondents regarded themselves as a person with a biological condition or as a person with a biological condition which affected their life. To discern their interpretation of the word patient, they were asked about their motivation to call themselves a patient or not. Although seven respondents stated that they did not regard MFS as an illness and ten respondents never felt ‘sick’ as a consequence, only two respondents clearly stated that they did not feel like a patient. Nine respondents identified themselves as a patient, six only felt themselves to be a patient when they visited a medical institution and two said they did not want to feel like a patient.

The nine respondents who did call themselves a patient, did this because of all the hospital visits that are related to MFS or because they always had to take MFS into account when they planned an activity. Furthermore, two respondents said they only felt themselves a patient in periods that they were really bothered by fatigue and pain. It appears that the motivation to call themselves a patient is in line with the distinction between disease and illness: most respondents were intended to call themselves a patient only when they were ‘confronted’ with MFS. In their daily life MFS was regarded as a biological condition, but when they needed to visit a hospital for example MFS was more than a biological condition. This points again, to the embodiment of MFS by these respondents. Two respondents stated that when they started to take medication, this made them feel more like a patient than before. Nowadays taking medication did not have that effect on them anymore. Taking medication had become part of their life and thus was embodied.

Personal history

According to Traas, personal history is an important aspect of identity establishment. Now it will be analyzed which personal factors of the respondents correlate with the different ‘standpoints’ regarding identification with MFS or with the identity of a patient in general. First, the factor of relatives with MFS will be analyzed. Table 4 shows the distribution of those with and those without relatives with MFS over the different categories of identification (as discerned in the analysis above).

	Family with MFS	Spontaneous Mutation	Unknown
I am more than MFS	2	2	1
Identification with the group: "We/ Us"	3	0	1
It made me who I am	1	2	1
We have got a bond together	4	0	0
What is caused by MFS and what is my personality?	2	0	2
No remarks regarding identification with MFS	0	5	0

Table 4: The relation between presence of MFS in the family and different categories of identification. (Some respondents fell in more than one category, thus the sum is more than 19).

It appears that those who had relatives with MFS were more inclined to identify themselves with the group of MFS-patients than those who have a spontaneous mutation of their genes. The second and fourth category, which point at identification on a group-level, are mostly filled by respondents who had relatives with MFS. Furthermore, the five respondents who did not make any remarks regarding identification were all respondents who did have a spontaneous mutation of their genes. Although one cannot conclude from the unspoken, this again suggest that those without relatives with MFS are less inclined to identify themselves with MFS.

If one analyzes whether this factor of presence of MFS in the family affected the feelings of illness, it turned out according to table 5 that people with a spontaneous mutation were slightly more inclined to regard MFS as an illness than those who had relatives with MFS.

	Family with MFS	Spontaneous Mutation	Unknown
MFS is an illness	5	6	0
MFS is not an illness	3	2	2
Label does not matter	1	0	0

Table 5: The relation between presence of MFS in the family and regarding MFS as an illness.

If one takes a look at the influence of age of diagnosis, which can be regarded as another factor of one’s personal history, it turns out that all respondents who said that MFS had a major influence on their personality were diagnosed at an early age (4-7). In the other categories of

identification with MFS there is no regularity to discover and age of diagnosis seems not to affect feelings of illness or identification as patient in general.

Another factor that is part of one's personal history is whether one has been operated on or not. It is hard to make this distinction because seventeen respondents told of how they had been operated on at least one time. Thirteen of these were operated for cardiac problems, while four respondents had other operations. Although the group of respondents with other operations is too small to draw serious conclusions, it appears from table 6 that relatively more people with other operations identified themselves as a patient, while respondents with cardiac operation were more inclined to regard MFS as an illness.

	Cardiac operation	Other operation
Identification as a patient	7	3
No identification as patient	6	1
MFS is an illness	8	2
MFS is not an illness	4	2
Label does not matter	1	

Table 6: The relation between sorts of operation and identification as a patient and between sorts of operation and regarding MFS as an illness

One final factor that is part of one's personal history is employment status. Table 7 shows that those respondents who had their own company were less inclined to identify themselves as a patient compared to those respondents who were considered unable to work or were in wage labour. Furthermore, those with their own company were less inclined to regard MFS as an illness compared to the other two groups of respondents.

	Own Company	Considered Unable to work	Employed in wage labour
Identification as a patient	1	4	5
No identification as patient	1	0	0
Only with hospital visits	5	1	2
MFS is an illness	2	4	4
MFS is not an illness	4	1	3
Label does not matter	1	0	0

Table 7: The relation between employment status and identification as a patient and regarding MFS as an illness

Acceptation and recognition

From the above it turned out that the most compelling factor, at least within this sample, in the identification with the larger group of MFS-patients is the presence of MFS in the family. This is in line with the notion of both Erikson (1971, 1980) and Honneth (1996) that acceptance and

recognition play an important role in identity establishment. When one has relatives with MFS, within this family MFS probably normalised and this person will feel recognized and accepted. For those respondents who identified themselves with the group of MFS-patients as a whole, it turns out that the patients association (CMN) has the same function. These respondents said that they feel recognized and accepted by the others when they visited for example a day of contact.

Conclusion

At the beginning of this chapter it was stated that the apparent paradox of the impression of a tough life and the experiences of having a good life would be solved. One can conclude that a lot of potentially negative experiences of the respondents are ‘embodied’ by them. As a consequence these experiences and their attributes became part of their identity and thus became normal for the respondents. Although most respondents embodied one or more of their characteristics, not all respondents identified themselves with MFS. It turned out that different categories of identification with MFS might be discerned. The notion of control appeared to play an important role for some of the respondents. These respondents did not want to identify themselves with MFS or doubted about the influence of MFS on their personality. The data of these respondents seemed to reflect that they were afraid of losing control over their ‘image’, when they applied the label of MFS-patient to themselves.

Furthermore, it turned out that almost all respondents were aware of the difference between disease and illness. Some respondents regarded MFS as a biological condition, while others attached meaning to living with MFS. Their motivation to call it an illness was mostly because they experienced several limitations and consequences of MFS. Labelling MFS as a ‘handicap’ turned out to be related to notions of (loss of) control, while the label of ‘construction error’ served for some respondents to making sense of their situation.

Almost all respondents identified themselves as a patient, but most of them only identified themselves as a patient when they were ‘confronted’ with MFS, for example when they visited the hospital, which points to the embodiment of their situation.

The presence of MFS in the family appeared to be an important factor in relation to the different categories of identification. Those respondents who had relatives with MFS were more inclined to identify themselves on a group level with other MFS patients. In the previous chapter it already turned out that these respondents showed less signs of feeling (treated) different than those who did not have relatives with MFS. From these two results one can conclude that recognition and acceptance plays an important role in the identification process. This is confirmed by the statements of some respondents that they enjoy visiting the Day of Contact of

the patients association because they feel accepted and recognized on these days. Within these groups (the family with one or more relatives with MFS and a groups of MFS-patients) these respondents do not differ from the norm, because this norm is 'defined' by MFS.

Most important finding in this chapter is that most respondents learn how to deal with the effects of MFS, as described in the previous chapters, on their life. This happens on the one hand by embodying their situation, while on the other hand some try to stay away from the label of MFS to prevent it from becoming a major aspect of their identity. Although all respondents acknowledged that MFS influences the direction of their life, by 'inventing' new typifications this situation has become the norm and they embodied their situation.

Conclusion

The aim of this research was to map out and explore the experiences of people with MFS and to discern how they understand their condition. The key themes around which the content of the interviews with 19 respondents were analyzed were the long-term effects of MFS, the effect of MFS on the social life of the respondents, stigma and outsiders and finally identification.

To start with, within the motives of the respondents to participate in the research one could discern three categories. First, most respondents felt it was useful for next generations when the knowledge about MFS would be expanded. The second motivation was to show that MFS had a lot of effects on different aspects of their life and that MFS is not ‘just a label’. The third motive was exact the contrary: these respondents wanted to show that living with MFS was not that bad at all. The latter two categories balanced each other out.

Although the respondents from the third category initially stated that MFS had not that much effect on their life, during the interviews it turned out that MFS had an influence on different aspects of the lives of almost all respondents. In what follows, the different understandings of these effects of MFS will be outlined.

First, MFS was regarded as a source of limitations. As a consequence of pain and fatigue most respondents felt that they were not able to live a life that they regarded as normal. According to the respondents a normal life consisted a ‘freely chosen job’, ‘a job without adjustments’ and preferably ‘a fulltime job’. Furthermore, when they would have a normal family-life, this normal life consisted ‘children’ and ‘getting children would go unconsciously’. MFS was both an ethical and a physical burden to have children for most respondents.

Second, MFS turned out to be a source of guilt for twelve respondents. On the one hand, they were not always able to show the right line of behaviour which resulted in feelings of guilt towards their partner, friends and family. They could not always meet their own expectations of a normal friend, child or parent. The typification used for this normal friend, child or parent was that of an active, participating person. On the other hand, because of inheritance and insecurity about the severity of inheritance respondents showed feelings of guilt towards their (future) children.

Third, most respondents regarded MFS as a source of insecurity. On the one hand, as a consequence of pain and fatigue, respondents were not always able to participate and thus show the right line of behaviour. Especially with strangers the respondents did not always know when they were in or out of face. This resulted in feelings of insecurity. On the other hand, especially the respondents with more (severe) external attributes felt that they were judged on basis of their appearance. This made them feel insecure and made that they kept themselves more at the background. This was especially true in their youth

Fourth, all respondents showed signs of the stigmatized person as described by Goffman (1963). Although not all respondents regarded feeling to be different as a problem, most respondents understood MFS as a stigmatizing source. They showed feelings of being (treated) different(ly), shame, insecurity and feelings of being an outsider. These feelings were more common for those with severe external attributes and for those who had no family with MFS. According to themselves they differed from the norm, which was 'being healthy' and 'looking good, without deviations'. This differing from the norm of 'being healthy' made that not only the attributes were regarded as stigmatizing, but MFS itself was regarded as being potentially stigmatizing. Especially in relation to work, some respondents thought that disclosure of MFS would have a negative effect on their career perspective.

Fifth, almost all respondents identified themselves as a patient. This meant that MFS was more to them than just a biological condition. Instead they felt that MFS had an effect on their life. Most respondents did not feel themselves patient in daily life, but when they were confronted with MFS. For example, when they start taking medication or when they visit the hospital. Furthermore, some respondents regarded MFS as an biological condition which had not much effect on them (disease), while other did call it an illness, because it has effect on different aspects of their life.

Sixth, the label of MFS itself had two, contradictory, effects on the respondents. On the one hand, the diagnosis with MFS felt for some respondents as legitimating who they were. By objectifying MFS as a medical condition, it made sense to them why they were the way they were. On the other hand, the risks that are connected to MFS made that other respondents regarded the diagnosis as a fateful moment: a key personal event that irreversibly changes the ways in which people perceive themselves and the world in which they live (Alaszewski 2006:3). This was mostly related to age of diagnosis and the cause of diagnosis.

Seventh, MFS meant for some respondents a source of identification and new contacts. This was mostly due to the contacts they met via the patients association (CMN). At meetings of CMN they found acceptance and recognition which amplified their identification with MFS. On the other hand, some respondents wanted do dissociate themselves from the group of MFS patients. Identification with MFS meant for them a loss of control over their image.

Finally, several factors turned out to play a role in the way the respondents experienced living with MFS. External attributes were more often a source of insecurity and shame than internal characteristics. This is explained by Goffman's (1963) distinction between discredited and discreditable attributes. Furthermore, the age of diagnosis influenced partly the identification with MFS. Although those who were younger than ten when they got the diagnosis stated that

this diagnosis itself had no effect on them because they were so young, these respondents were more inclined than those diagnosed at a later age to acknowledge that MFS had a major influence on their personality. In addition, the factor of employment turned out to have an effect on the identification as patient and regarding MFS as an illness. Those respondents who had their own company, were less inclined to call themselves a patient and to regard MFS as an illness. Finally, and most importantly, is the factor of presence of MFS in the family. The respondents who had family member with MFS showed less signs of feeling (treated) different(ly) than those who did not have relatives with MFS. Furthermore, they were far more inclined to identify themselves with MFS on a group level, while they were slightly less inclined to regard MFS as an illness. Probably this is because within their family MFS is part of the norm and thus they are not different from the norm.

Although MFS was regarded as a source of limitations, guilt, insecurity, stigma, disturbance of what is regarded as normal and, for some, a source of identification, this was not how most respondents experienced living with MFS in their daily life. This was due to the fact that most respondents embodied one or more of their characteristics of MFS. As a result they became unconscious about living with MFS. Although not true for all respondents, most of them began answering some question by saying “Well...now that you are asking...”. Sentences like “but usually I do not realize that”, “that has become normal to me” and “it is funny that you are asking that” were used several times by most respondents. That embodiment is a process is illustrated by the statements of many respondents that in their youth and adolescence, they were far more insecure than they were nowadays.

On a conceptual level, one can conclude that MFS resulted for most of the respondents in a spoiled identity. First, these respondents had typifications in mind of what a good friend, partner or child should look like. As a consequence of fatigue and pain, most respondents could not always live up to their own expectations of their role. They were forced to come up with new typifications for their own role.

Second, all respondents showed one or more characteristics of the stigmatized person as described by Goffman (1963) and which were related to MFS. This means that all respondents experienced that MFS had in one way or another effect on their identity. Most of them felt to be treated differently because of MFS. This was especially true for external characteristics. This means that the respondents felt that other people negatively judged them on basis of their characteristics and that other people saw these characteristics as part of their identity. Third, MFS did not only influence the respondents in their role of social actor (friend, partner or child), it also

conflicted with the typifications the respondents had in mind for different ‘aspects’ in life. MFS turned out to effect the direction of the lives of the respondents in important aspects. If the respondents could have lived the life they had typified, they would all have a fulltime job, most of them would have had children ‘unconsciously’ and they would work in a job they had freely chosen. For most respondents who could not live in line with these typifications, it felt they were not living the life they had in mind. As a result most of them felt that these ‘failures’ were part of who they were and thus had become part of their identity.

Finally, MFS resulted in a spoiled identity because most respondents were in different situations insecure whether they showed the right line of behaviour and whether they were in or out of face. Again, as a consequence of MFS they could not live up to their typification of a successful conversation partner.

So, MFS resulted in a spoiled identity because most of the respondents could not live in line with their own typifications and because they felt they did not meet the expectations and typifications of other people. Most respondents felt that these ‘failures’ made them who they were. In other words they felt that the direction of their life in certain aspects was strongly affected by MFS. Although almost all respondents experienced one or more of these breaches with their typifications, not all respondents were bothered in the same way by it. All respondents showed signs of the process of embodiment. Those respondents who were not bothered that much by MFS spoiling their identity had found new typifications for their life. Within the borders of the direction set by MFS, they developed new norms. They could live in line with these new typifications and as a consequence they were not conscious anymore about living with MFS. This ‘spoiled’ identity had become part of their identity, because as a consequence of new typifications, the ‘failures’ were not ‘failures’ anymore.

For reasons outlined in the methods chapter, the results of this research cannot be generalized to all MFS-patients. In sum, one can conclude that for these respondents MFS has an effect on different aspects of their life. MFS is understood as a source of limitations, guilt, insecurity, stigma, disturbance of a normal life and a source of identification. Although MFS is regarded as a disturbance of ‘normal’ life, which contained children and a (fulltime) job, their current situation has become normal for most respondents. These respondents have embodied living with MFS and as a consequence they do not feel living with MFS is a tough life. Instead, they adapted themselves to their situation and fatigue, pain and a ‘deviant’ body had become part of their typification of a normal life.

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Hopefully, after they read this thesis they will feel I have done right to their stories. Furthermore, I want to thank Contactgroep Marfan Nederland for giving me access to their members and for their cooperation in the recruitment of the respondents. In particular I want to thank Ine Woudstra who made it possible that my appeal was distributed as soon as possible. Finally, I want to thank my two supervisors Patrick Brown and Christian Bröer. Especially you Patrick, you have been a great support and I want to thank you for your enthusiasm, your comments, your references and for giving me the opportunity to graduate under your supervision. Christian, thanks for your comments to improve this thesis and I know how busy you are with supervising, so I'm very thankful that you wanted to be my second supervisor.

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Attachments

Attachment 1: Criteria and characteristics of Marfan Syndrome

For the proband, diagnosis requires the presence of major criteria in at least two organ systems and involvement of a third organ system. For a family member, diagnosis requires the presence of one major criterion in family history and one major criterion in an organ system and involvement of a second organ system.

	Major Criteria	Minor Criteria
Skeletal system Two components of the major criterion or one component of the major criterion plus two of the minor criteria must be present	Pectus carinatum Pectus excavatum requiring surgery Reduced upper-to-lower segment ratio or arm span-to-height >1.05 Positive wrist and thumb signs Scoliosis of >20° or spondylolithesis Reduced extension of the elbows (<170°) Medial displacement of the medial malleolus causing pes planus	Pectus excavatum of moderate severity, Joint hypermobility Highly arched palate with dental crowding Facial appearance (dolichocephaly, molar hypoplasia, enophthalmos, retronathia, and down-slanting palpebral fissures)
Ocular system The major criterion or at least two of the minor criteria must be present	Ectopia Lentis	Abnormally flat cornea (as measured by keratometry) Increased axial length of globe (as measured by ultrasound) Hypoplastic iris or hypoplastic ciliary muscle causing decreased miosis
Cardiovascular system One major criterion or one of the minor criteria must be present	Dilation of the ascending aorta with or without aortic regurgitation and involving the sinuses of valsalva Dissection of the ascending aorta	Mitral valve prolapse with or without mitral valve regurgitation Dilation of main pulmonary artery in the absence of valvular or peripheral pulmonary stenosis or any other obvious cause, at age younger than 40 years Calcification of the mitral annulus at age younger than 40 years Dilation or dissection of the descending thoracic or abdominal aorta at age younger than 50 years
Pulmonary System One of the minor criteria must be present	None	Spontaneous pneumothorax Apical blebs (Shown on chest radiograph)
Skin and Integument The major criterion or one of the minor criteria must be present	Lumbosacral dural ectasia by computed tomography or magnetic resonance imaging	Stretch marks Recurrent or incisional herniae
Family History One of the major criteria must be present	Having a parent, child, or sibling with either: Presence of a mutation in FBN1 known to cause Marfan syndrome, or Presence of a haplotype around FBN1, inherited by descent known to be associated with Marfan syndrome in the family	

(Fusar-poli et al. 2008:245)

Attachment 2: Research appeal Contactgroep Marfan Nederland to recruit respondents

Alkmaar, February 8, 2011

Dear members of Contactgroep Marfan Nederland,

My name is Floortje Koks, I study sociology at the University of Amsterdam and like you I have Marfan syndrome. In the waiting-room for the different physical inspections in the last couple of years, I noticed the diversity of the group of Marfan patients. When I needed a subject for my master thesis, it turned out that little research had been done on this subject in the Netherlands. That's why I decided to graduate on this subject.

In my research I want to map out the social experiences of people with Marfan syndrome. For example, I am curious whether having MFS has an influence on your daily life. To find an answer on this question and other questions, I am looking for Marfan patients aged 18 years or older who want to be interviewed by me. These interviews will last about an hour and they will look like a normal conversation in which you can tell what you want. I won't ask the questions in a strict order, but I will ask certain questions to let you talk about subjects I am interested in. I would like to record these interviews on a voice-recorder, so I can listen more concentrated to your story. If you have got objections to this recording, I won't do it. In my theses I will only make use of quotes from the interviews and this could, on request, happen anonymously.

In short: Do you want to participate in my research? You would be very helpful to me and possibly the results of my thesis might be of use for the different Marfan policlinics in the Netherlands.

If you want to participate, please send an e-mail with your contact-details to Floortje.Koks@student.uva.nl, so we can make an appointment for an interview. The interview can take place in every desired location. If you think it is more easy to get directly in contact with me by phone, you can reach me at 06-17364962. If I am not able to answer your call, please leave a message at my voice-mail and I will call you back as soon as possible. I count on it that you won't give my number to thirds.

As you will understand, I have got a deadline for my thesis, so if you want to participate, let me now as soon as possible.

Thanks in advance for your cooperation!

Best regards,

Floortje Koks

Attachment 3: Topic- and question list

Introduction

- What's your age?

- What are your hobbies?
- What do you do in your daily life?

- How old were you when you were diagnosed?
- What was the 'inducement' for your diagnosis?
- Which characteristics of MFS do you have?
- Are you the only one in your family with MFS or do you have relatives who have MFS as well?

Daily life

- Do you 'notice' in your daily life that you have MFS?
 - If so: What do you notice/ how do you notice this?
 - Can you give examples?
 - How do you think about this? / What do you feel about this?
- Do you feel restricted sometimes by MFS?
 - If so: In what way?
 - Can you give examples?
 - How do you think about this? / What do you feel about this?
- Are there things that you do, because you have MFS?
 - If so: Can you give examples?
 - Why do you do these things? / What has having MFS got to do with these things?
- Are there things that you don't do because you have MFS?
 - If so: Can you give examples?
 - Why (don't you do these things)?
 - How do you think about this? / What do you feel about this?
 - Do you 'miss' things?
- Do you have a job?
 - If so: What kind of job?
 - Did (having) MFS influence your choice for this particular job?
 - How much do you work?
 - Did (having) MFS influence the amount of hours you work?
 - Do you 'notice' during your work that you have MFS?
 - If so: Can you give examples?
 - How do you think about this? / What do you feel about this?
 - If not: Why don't you have a job?
 - Did (having) MFS influence your choice not to work?
- Do you have children?
 - If one has consciously decided about having or not having children: did MFS play a role in the considerations?
 - If so: in what way?

Social life

- Does MFS directly or indirectly influence your social life?
- If so: Can you give examples?

- How do you think about this? / What do you feel about this?
- How do you feel about meeting new people?
- Does having MFS influence this?
- If so: Can you give examples?
- Are you satisfied with your social life?
- Why/ why not?
- Do you think your social life would be different if you hadn't MFS?
- Why/ why not?

Stigma/Outsiders

- Do you sometimes feel that you are treated differently?
 - If so: Can you give examples?
 - How do you think about this? / What do you feel about this?
 - Does this have an effect on you?
 - What do you think is the reason that you are treated differently?
 - Do you think MFS has something to do with it?
 - Do these experiences/ this experience influence the way you enter a situation similar to this?

If someone has bodily/ outwardly attributes:

- Do you think these attributes influence the way other people see you?
- Why/ Why not?
- How do you think about this? / What do you feel about this?
- Do you try (sometimes) to hide these attributes?
 - If so: In what kind of situations?
 - Why?
 - How?
- Are there situations you try to avoid because of these attributes?
- Are there situations you try to avoid in general?
 - If so: what's the role of MFS?
- How do you feel in a group with people you don't know that well?
- Does MFS play a role in these feelings?
- Are you ever ashamed because of MFS?
 - If so: why?

Self-image/identification/embodiment

- Do you feel yourself different?
 - If so: Why?
 - (Where do you compare yourself with?)
 - How do you think about this? / What do you feel about this?
- Do your MFS-attributes make you feel different (in some situations)?
 - Why/ why not?
- Are there situations in which you're consciously aware of your attributes/ condition?
 - If so: In what kind of situations? Can you give examples?

What do you do in these situations?

- Do you regard MFS as an illness?
 - Why/ why not?
 - If not: How would you call it?
- Do you regard yourself as a patient?
 - Why/ why not?
 - If not: How would you call yourself (in relation to MFS)?
- Do you take medicine?
 - If so: Does this influence the way you experience having MFS?
Does this influence the way you see yourself (in relation to MFS)?
- Did the official diagnosis have an influence on you/ Did it effect you?
 - If so: how/what/why?
- Did the diagnosis effect the way you see yourself?
 - If so: how/what/why?

- Do you often have to explain what MFS is?
- Can you give examples of situations in which you 'had' to explain what MFS is?
- Why did you 'had' to do this in these situations? Did people ask about it or did you 'feel the need'?
- How do you feel about that?

Patient Association Contactgroep Marfan Nederland (CMN)

- Are you a member of the CMN?
 - If not: why not?
 - Will you become a member in the future?
 - If not: Why?
When did you become a member?
What are the benefits of being a member?
Do you visit the contact day?
Why/ why not?
- Do you 'need' contact with other people who have MFS?
- Why/ Why not?
- What can they mean to you/ give you?

Finally

What is your motivation to participate in this research?