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Living with a Rare Disease as a Family: A Co-Constructed Autoethnography from a Mother

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Abstract: Research into family quality of life (FQoL) is becoming increasingly popular. However, studies into the interrelations between family and individual quality of life (QoL) are still scarce. The aim of this article is to illustrate how having a child with a (rare) chronical illness/disability (specifically, Neurofibromatosis Type 1) affects both the family as a whole and its members individually. The lived experiences are recounted by the Mother (first author) and have been further explored through the method of co-constructed autoethnography. Metaphors have been used to help understand the findings. Our findings show that each individual QoL not only influences the FQoL but has a domino effect on each other. Individual lives are intertwined, and accordingly their well-being cannot be seen as being distinct from these interrelationships. (F)QoL should be viewed as a 'praxis of care', where caregiving occurs to and by each member, and continuously changes over time.

Keywords: quality of life; family; care; autoethnography; family dynamics; neurofibromatosis; chronic disease; qualitative research; rare disease

Citation: Damen, I.; Schippers, A.; Niemeijer, A.; Abma, T. Living with a Rare Disease as a Family: A Co-Constructed Autoethnography from a Mother. *Disabilities* **2022**, *2*, 348–364. https://doi.org/10.3390/disabilities2020025

Academic Editors: Ann Alriksson-Schmidt and Ira Jeglinsky-Kankainen

Received: 1 April 2022
Accepted: 2 June 2022
Published: 13 June 2022

1. Introduction

1.1. (Family) Quality of Life

Families are an integral part of all cultures, the cornerstone of society, and harbor their own characteristics [1–3]. An important function of the family is to inform and to maintain the identity of their members [4]. Cummins [5] (p. 15) explains that a family consists of individuals to whom it is important to receive a 'homeostatic effect' in their personal experienced Subjective Well Being (SWB). This depends on both the genetical setpoint and the experienced life, which includes the setting of family life.

When a family is faced with chronic illness or disability, the individual family members and the family system not only have to deal with changes through the cycle of life [6] (p. 790), but also with the (changing) demands concerning the illness or disability which 'intensify life' [3,7–10].

In this article, we will use the definition of 'a family' as proposed by Poston et al. [11] (p. 319), namely, 'A family includes the people who think of themselves as part of the family, whether related by blood or marriage or not, and who support and care for each other on a regular basis.'

The way families function is affected by *every* single family member, and relates to factors such as the child's disease or disability and each family member's life skills, health issues, communication, and motivation. Turnbull et al. [3] (p. 77) emphasize that life in cases of people who have to deal with lifelong issues requires the long-term endurance of a marathon runner, which differs from a sprinter.

Individual QoL (Quality of Life) and family life have been topics of research for decades, in several disciplines [12]. While in the disability field, there currently consists strong consensus on individual QoL [13], the increasing political focus within the disability field has meant a shift towards a family-centered service delivery model [14–16]. The idea is that policies and services, which are there to improve individual and/or family well-being, can better respond to the needs with providing support and planning interventions [1,17]. The roots of the Family QoL (FQoL) framework can be found in the concept of individual QoL and it is a family centered and strength-based approach that is multi-dimensional and lifespan in nature [16,18–20]. However, even the focus of more family-oriented research still seems to be somewhat one-sided, often informed by and based solely on the experiences of the main caregiver [14], even though it is still not very clear how individual QoL is interrelated with FQoL. Nor is the concept QoL itself free from ambiguities. As Hunt [21] (p. 205) noted as early as in 1997: 'there is little agreement about the meaning of the term itself, there are rival factions each tendentiously urging the adoption of a different approach and a plethora of measures purporting to address quality of life'. Nevertheless, the concept of quality of life has been of paramount importance for evaluating the quality and outcome of health care. Yet despite its importance, there is still no consensus on the definition or proper measurement of (family) quality of life [22].

Zuna, Summers, Turnbull, Hu, and Xu [23] (p. 262) define FQoL as 'a dynamic sense of well-being of the family, collectively and subjectively defined and informed by its members, in which individual and family level needs interacts'. Brown and Brown [24] (p. 2195) explain FQoL as the extent to which family members individually experience their personal quality of life within the family context, as well as with how the family unit can pursue its possibilities and achieve its goals in their community and the society. A challenge is to show how these various viewpoints, including the person with the disability, intertwine with FQoL as a whole [25–28]. As part of FQoL, most scholars have been interested in the QoL of the child/person with the disease/disability, followed by the QoL of parents and their opinion about FQoL. However, research of the impact on FQoL, thus QoL of every family member, their interrelations, and their jointly experienced FQoL, is still scant and needs to be further explored [12,14,29] (p.162).

Although the concept of FQoL is mainly developed in the field of intellectual and developmental disabilities, we agree with Zuna et al. [23] (p. 242) that a theory of FQoL needs to consider factors of families with children with a variability of disabilities, including rare diseases (RD). Studying families living with RD can contribute to the refinement of the definition of the concept being sought.

Several methods are being used to measure FQoL. Two leading FQoL instruments within the disability field are now: the Family QoL Survey-2006 [30] and the Beach Center FQoL Scale [20,31,32]. According to Brown and Brown [26] (p. 4), these instruments can be strongly applied to all kind of families, thus also for chronic and rare diseases.

1.2. Rare Disease, NF1 and FQoL

Within the field of rare diseases (RD), even less research has been conducted into (F)QoL.

According to the European Union, a disease is rare when it affects less than 1 in 2000 citizens and can be characterized by a large variety of symptoms. These symptoms differ not only from disease-to-disease but also from patient-to-patient having the same rare disease [33]. Most of them are genetic. About the quality of life of someone living with a rare disease, it can be said that: 'Typically disabling, the quality of life of a person living with a rare disease is affected by the lack or loss of autonomy due to the chronic, progressive, degenerative, and frequently life-threatening aspects of the disease'. 'The fact that there are often no existing cures adds to the high level of pain and suffering endured by patients and their families' (Ibid).

Although it is common knowledge that rare diseases have a huge impact on a physical, emotional, and social level, there still is scant insight into the impact of having a rare disease both on daily life of the individual living with the disease and his or her family [34].

Neurofibromatosis Type 1 (NF1) is one of those rare diseases. NF1 is a dominant, neurological, genetic disorder, and its prevalence ranges between 1/300 and 1/6000 [35]. Furthermore, NF1 is a multisystem disorder, progressive, and incurable. The symptoms are highly variable and unpredictable. Living with NF1 also often entails cognitive, behavioral, psychiatric disorders, and psychological problems. It is complex and can be life-threatening, and thus can strongly affect daily life [36–38].

Persons with NF1 and their families will be uncertain about the development of their disease. One of these uncertainties is that they do not know whether they will be developing tumors on the inside or the outside of the body, whether tumors will become life-threatening and/or disabling and/or disfiguring, whether there will be cognitive impairments, behavioral problems, or just some brown spots (café-au-lait) on their skin, etcetera [39]. NF1 is 'a condition without parameters' [40] (p. 140). This refers to its huge variability and unpredictability, which creates a lot of uncertainty. Thus, it is important to obtain more insight into the FQoL of families who must live with it. This may help these families to enhance their individual and family well-being. Scholars are especially focused on 'Ending NF through research' [41] or disease management [35,42].

Within the field of NF1, there is growing interest in QoL of persons with NF1 [43–54]. Fewer research studies have focused on the impact on parents [40,55–59]. Pierre-Louis, Heinhuis, Riklin, and Vranceanu [60] mention, as some stressors for parents: the unpredictability of NF1, worries about educational, social, psychological, and medical needs of their child with NF1, worries about the sibling(s) without NF1, managing finances, multiple (medical) appointments, own health issues and employment, role changes, etcetera. However, it can be concluded that more research must be conducted on the impact of NF1 on the QoL of individual family members and the FQoL as a whole.

1.3. Aim of the Article

The main aim of this article is to illustrate in what way having a child with NF1 can affect family life, by specifically focusing on the interrelations within a family. As the family is the most fundamental social unit, these interrelations are essential to support and give a feeling of belonging and inclusion to all family members [61] (p. 290).

In doing so, we also intend to give more insight for the overall concept of FQoL, to gain better understanding of issues concerning family quality of life [12] (p. 1095), and thereby ultimately contribute to a unified theory on Family Quality of Life, which is still lacking [23,62].

The family that we report on has had to deal with many different health (care) providers, hospitals, schools, local government, and other care professionals. Experiences with formal professionals and organizations are beyond the scope of this article, although it is important to realize that they also (have/) had a huge impact on the FQoL. Accordingly, this article does not pretend to be exhaustive and does not aim to generalize its findings. The experiences are limited to those within the family through the eyes of the first author, using first-person pronouns, as they reveal the Mother's (first author) perspective, and were co-constructed through conversations with her family and the co-authors of this article.

2. Methodology

2.1. Co-Constructed Autoethnography

An autoethnographical approach was chosen as being ideally suited to the aim of this article, with a specific focus on ethical and relational aspects. According to Chase [63] (p. 549), a 'personal narrative' is a different form of communication. It is 'meaning making through the shaping of experience; a way of understanding one's own or others' actions; of organizing events, objects, feelings or thoughts in relation to each other; of connection and seeing

of consequences of actions, events, feeling, or thoughts over time (in the past, present, and/or future)'. Autoethnography reveals the identity of the author and acknowledges the subjective experience of being human [64–72].

Accordingly, use was made of personal memories, logbooks, photos, email correspondence, self-observation, self-analyses, comparisons, reflections, conversations, discussions, going forwards, and going backwards. During the process of data gathering, analyzing, and writing, the first author's experiences were consequently analyzed together with the other authors of this article through a co-constructed, dynamic, reflective, and dialogical process [73] based on and contributing to the insights of the research team and the rigor of the study. All authors are familiar with the concept of FQoL, autoethnography, and care ethics, and have personal experience with caring for a child with a disease or disability. Several external experts have also shared their thoughts. They are mentioned in the Acknowledgements. During the whole process, knowledge from different sources was used, e.g., disability studies, chronic/rare diseases, care ethics, family dynamics, NF1-specific, etcetera, enabling a broader view on the subject of (F)QoL.

The sharing of the narratives took place over an extended period. We had several meetings and sent each other updates. This way, joint knowledge, insights, and a deeper understanding of the experiences arose about the life of a family with a child with NF1, which we consider to also be 'useful for what happens to others elsewhere' [74] (p. 169).

2.2. Ethical Considerations

As Ellis states clearly [75] (p. 307): 'When we write autoethnographically about our lives, by definition we also write about intimate others with whom we are in relationship'.

This article includes the intimate others of the family of the first author, so accordingly we have taken several ethical considerations into account, which were mainly inspired by Tullis [76], who listed seven ethical guidelines for autoethnographers to work with (pp. 256–257).

First, she mentions the guideline of doing no harm to self and others, which meant for this article plenty of (self-) reflection during the whole process of gathering, analyzing, and writing. It also meant anonymizing as much as was possible the other family members, to the extent that they could not automatically be identified. The second guideline has to deal with obtaining ethical approval for conducting the research. This research, as part of bigger research on FQoL, was sent to the Medical Ethics Review Committee and they stated that the Medical Research involving Human Subjects Act did not apply to our study into FQoL. Nevertheless, all family participants gave their informed consent to this research, were given a chance to read and comment on this article, and gave their permission to share this narrative [77,78] as is also formulated in the third guideline. In this way, we showed our respect to the participants' autonomy. Indeed, all the family members were informed and involved during all phases of the project. In this way, we practiced process consent and we explored the ethics of consequences, as mentioned in guideline 4. Until the end of the process (publishing), we conducted several member checks (guideline 5). We also did not present publicly or publish anything about these family members that they have not seen (guideline 6), and we often spoke about the possible consequences of the afterlife of this publication for the family members (guideline 7). We cannot foresee all consequences of publishing this research, but we tried to be as careful and open as possible to give more insight into family life living with a rare disease.

2.3. Metaphors

We searched for a suitable way to express the experiences in this study [79]. The first author subconsciously used several metaphors during our meetings, so we consequently decided to focus on these metaphors analytically [14,80]. These metaphors, when well-chosen, serve as overarching themes and rich undercurrents that resound throughout the 'portrait' [81]. According to Lakoff and Johnson [82] (p. 247), 'which metaphors we have and what they mean depend on the nature of our bodies, our interactions in the physical

environment, and our social and cultural practices', which fits the use of autoethnography. Metaphors not only explain and shape our communication, but also shape the way we think and act. Lakoff and Johnson (Ibid) showed that metaphors structure our most basic understandings of our experience, and they are 'metaphors we live by'. Metaphors can shape our perceptions and actions without our ever noticing them.

2.4. Case

The nuclear family consists of the Father, the Mother, the Eldest (20, son), the Daughter (19), and the Youngest (15, son). They live together in a small village in the East of The Netherlands. Both parents are white and academically educated.

The first author's two decades of experiences since the Eldest was born were written down. At that point, the family awaited a new life phase for the Eldest: from Adolescence to Young Adulthood. The experiences that had a huge impact on the (F)QoL were explored. Thus, the family members gave access to their personal lives to gain more insight into what the consequences for a family living with a child with a chronic disease or disability can be—in this case, NF1.

The family has had little to no support from social instances during this period, which left the parents 'alone to figure out what to do and how to advocate on behalf of their child' and family (members) [25] (p. 20).

3. Findings

3.1. Gut Feeling

At the time the Eldest was born, we did not know that he had NF1. We had searched for eight years before the diagnosis for the Eldest could finally be made. As a mother, I felt something was wrong, but nobody confirmed my gut feeling. The Eldest was my first child, so I had no experience. Yet still, I had a gut feeling and had to find out why.

The Eldest cried a lot, both day and night. His head was somewhat big, he had some brown spots (café-au-lait) on his body, he had problems with his fine and gross motor skills, regularly threw up, had headaches, abdominal pain, ear infections, skin eczema, etcetera. Still, his growth was normal. He was very lively in his behavior and had a beautiful laughing smile. However, he could not stand sunlight, loud sounds, certain food structures, certain smells, showering, cutting his nails, hair, or wearing clothes around his neck. He bounced his head and had both anger and anxiety attacks. He could not be or sleep alone, so for more than ten years I slept in the same room as my Eldest, just to make sure the rest of the family got some sleep. After several years, I quit my job because of exhaustion and because the Eldest panicked when I was not around.

In the meantime, more problems with the Eldest became apparent: problems with sports, dressing up, opening cans, (un)tying shoelaces, opening/closing zippers and buttons, using cutlery, etcetera. He already wore glasses and arch supports. Socially, he was somewhat different, although he had friends. The Daughter surpassed him quickly with respect to physical agility. The Eldest compensated his clumsiness with a lot of humor, a memory like an elephant—and he was verbally strong.

My gut feeling kept pushing me. We visited several health professionals, but we received no answers. There seemed to be no problem other than that I 'worried too much'.

After the Youngest was born prematurely, the Eldest complained more about headaches, and the Daughter had speech problems. At that time, I thought our lives would just be temporarily hectic. Searches on the Internet resulted in my own diagnosis for the Eldest: Developmental Coordination Disorder (DCD), which explained a lot of his clumsiness.

I claimed a referral to a pediatrician and went there when the Eldest was almost 8. She was the first doctor who took the time to really listen to me and examined the Eldest thoroughly. She suspected NF1, which was confirmed by a DNA test in another hospital. The behavioral and physical problems had a cause, called: NF1! This period, which gave me a lot of frustration and exhaustion, as well as the feeling of not being taken seriously, came to an end. Thanks to my gut feeling! From this moment on, NF1 officially entered our

lives, with the Eldest being the only family member having it. However, instead of having more clarity and peace, a new hectic phase started. The years 2008 until 2020 have been exhausting for all of us.

3.2. Family Members

3.2.1. 'The Eldest'

As his headaches became increasingly worse and his whole physical condition was worrying, an MRI scan followed. This showed a hydrocephalus (high pressure in the brain) due to an aqueduct stenosis (constipation). The doctors panicked, but I was not surprised. On the memorable date of 25 April 2008, the Eldest had his first brain surgery in again another hospital. We thought his headaches would then be solved, but things went completely differently. During this period, the Eldest underwent twenty-three brain surgeries, because he appeared to be 'a problematic hydrocephalus patient'. He also has a huge inoperable plexiform neurofibroma (tumor) (among others) in his left buttock, growing from a big nerve in soft tissues. My Eldest often stayed in hospitals for (emergency) surgeries and innumerable clinical examinations. He also had to take puberty hormones and (experimental) medication due to his pain and his inoperable growing tumor.

Every day, he must deal with unbearable pain, chronic fatigue, sleeping problems, and side effects of the medication. Nothing seems to lessen the pain. This means that he sleeps and rests a lot and barely gets out of his room or the house. He has also been diagnosed with autism spectrum disorder (ASD) and DCD, which gives us all challenging situations. Due to his limitations, it is hard to socialize or even plan activities. Although he is terribly ill, it is barely noticeable on the outside. His symptoms are mainly invisible.

He must sit on a pillow, which he takes everywhere. He needs a hospital bed with an anti-decubitus mattress and recently also a wheelchair. Lifelong physical examinations are waiting for him because the disease is progressive.

His childhood has been exceedingly difficult and has left its scars. Not only physically, but also socially and emotionally. *'Mam, why me?' 'Mama, the pain is terrible.' 'This isn't happening. I can't live with this.'* My Eldest practically lives in hospitals. *'Mama, they know me here, that is not a good sign*.' He has traumatic memories of the many operations and examinations, uncountable number of different professionals, travel distances (travel sickness), the smell of hospitals, and so on. Most importantly: the feeling that he has not been taken seriously by a lot of people and the unpredictability of his disease.

Furthermore, he has barely been able to go to school. In the last 12 years, he has missed about 6 whole school years. *'Mama, I want to go to school, but I feel too sick.'* Very painful, even more because school friends do not contact him anymore.

At this moment, the Eldest is 20. An age at which you are supposed to discover the world and yourself, enjoy life with friends, and make plans. His outlook is totally different: he is too ill. When he feels capable, he is gaming on the Internet, watching football on TV, playing piano, or helping to prepare a meal. Sometimes, he can go to a football match with his dad or to a movie with the whole family, which gives us some quality time, structure, and distraction. Online gaming also gives many worries, due to bullying him with 'cancer-head'. Socializing is a problem; we do not know another person with the same symptoms, so he cannot share his problems with peers. He needs daily about 18 h rest; at 7.30 pm, he goes to bed: *'Mama, I really want to do things, but I feel terrible.'* To my mind, he barely has any quality of life. '*I rather stay at home, where I have everything, I need*.' The combination of being ill, being disabled, *and* having ASD is awfully hard. He needs structure and rest in his life, whilst having NF1 gives a lot of uncertainty and stress.

The Eldest also takes care *of me* by giving me hugs, making me coffee, setting the dining table, sometimes (un)packing the dishing machine. *'Mam, Can I help you? Shall we have lunch together?* He is my second memory: '*Mam, don't forget your appointment! Don't forget to pick up the Youngest from school*.' He obviously feels like a 'big brother'; he worries about the well-being of *his* siblings and watches over them, while keeping me informed. He warns his dad for working too hard and asks me often: *'Mam, are you okay? You look tired*.'

Since a couple of years ago, he is very patient and shows calmness. He knows much about football and actualities, and he often manages our technical equipment, such as computers and television.

It remains difficult to gauge his feelings. When I ask: '*Honey, how are you feeling*?' '*Tired*', is the main answer, or: '*Not good*.' '*How can I help you*?', I ask. '*Don't know*', he answers concisely. Sometimes, conversations go deeper, or shall I say: I talk, he listens? The Eldest does not talk much about his own feelings. When he feels bored, he finds it difficult to think of a distraction. He needs someone to give him clear tasks which he can carry out. There are difficult moments when he blames himself and feels guilty when we must cancel a family getaway because he is too sick. This hurts, because he is right when he sees our disappointment, but we also know that it is not his fault.

The Eldest is now officially an adult with a lot of life experiences and wisdom, and a lot of scars. There were moments that he thought nobody loved him and that he was not worth anything. This phase seems to be over. I say 'seems' because I am not sure about anything. His condition has become worse. The uncertainty about NF1 is always there.

The siblings of the Eldest have also had a lot of life experiences at a young age, which makes them wise beyond their years. They have experienced much stress, sadness, and incomprehension from others. Often, their needs were placed aside for the Eldest's needs, even though they deserved more time and attention.

The Eldest and I were away often, resulting in the siblings spending many hours with their grandparents. This was frequently unplanned and unexpected, which meant that they had no certainty in their daily life. As a result, questions about sickness and health, about sadness and happiness, about life and death, were asked regularly.

3.2.2. 'The Daughter'

My daughter is introverted, calm, and shy. She grew up with two boys who need(ed) much care and attention. This gave her a lot of stress, as well as emotional and physical problems. She is only sixteen months younger than the Eldest, so she has experienced the majority of his life. Before the diagnosis of NF1, the behavioral problems of the Eldest were the most challenging. Then, the physical problems came along, for the Youngest as well. At moments when the Eldest had to go to the hospital, she was the one who packed his suitcase with a little present in it. '*Mam, if you pack your stuff, I will pack his suitcase*.' She always took care of us, and in our absence, she felt responsible for the well-being of the Youngest and her Father: '*I will make dinner, mama. No problem*.' She also slept in the Eldest's room many times to keep an eye on him and I could sometimes sleep in my own bed. She always tried to reassure the Youngest and took on the role of mother. When the Eldest and I came back from hospitals, she became ill that same week. There was always the fear that the Eldest would die.

In my view, she had to mature too quickly, because of her experiences and the concomitant worries she had about each family member. '*Mam, how are you doing? Can I help you?*' She did not show her own feelings, because she did not want to bother us. My daughter was often completely exhausted and complained of migraine, stuffiness, and abdominal and back pain. She weekly missed a day from school because of her symptoms. I have also been to several doctors with her, trying to find the right balance for her well-being. We tried many things, such as emotional counseling, specialized sibling's courses, and several hobbies to help her relax. She even changed schools, and at her most recent school she finally had made some friends. '*Mam, when I'm at school, I don't think about the Eldest anymore*.' She seemed happy, although her health issues did not disappear.

It was hard to find time to spend alone with my daughter, because there was always a sick brother at home, or we as her parents were working or exhausted. Sometimes, she went alone with her parents for shopping, a walk, a play, a city trip, etcetera, although it should have been more. We have had to disappoint her so many times: '*Mama, I try not to look forward to promises, because they often have to be broken*.'

At a certain point, she started building a personal life outside of our family, planning her future, and seemed to enjoy life. I saw an independent young woman, which gave me some extra space and made me feel less guilty towards her. There was a short period of incomprehension regarding her parents and brothers. '*Mama, I don't know how to deal with my brothers, I don't understand them.*' '*I need personal space*.' She explained to me that she sometimes felt guilty towards the Eldest, because she could have fun with friends whilst he was sick at home.

Then, she hit a wall at age 16. This had been my biggest fear during her life. As a mother, I tried everything to prevent this during her life. However, apparently this had not been enough; she became very depressed due to a combination of her caring personality, her age, and growing up in this family. Two years on, she still battles exhaustion, physical pains, anxiety, sadness, and low self-esteem every day. She felt an outsider within our family. She is searching for her identity: '*People always asked me how the Eldest was doing, not how I was doing*.' However, our bond has become much stronger than the years leading up to her fall into depression. Her life has been overly complex, and now she tries to get it back on track, obtaining her diploma, then aiming for university and living on her own.

3.2.3. 'The Youngest'

The Youngest developed separation anxiety, which resulted in constantly wanting to be with mam, including the nights. When the Eldest and I went to the hospital for several nights, he knew how to reach me with innumerable phone messages. It did not matter for him whether others were at home; he needed me: '*Mam, I can't sleep without you! When are you coming home? Mammmmmmm?'* He has been very afraid that the Eldest would stay ill forever. This anxiety resulted in major stomach problems and severe headaches, no self-confidence, and a lot of sickness. He also missed many school periods and went to several doctors. For years, when the Eldest was sick, then the Youngest would be as well. Another thing was his motivation: *'Mam, the Eldest stays home, I want to stay home too'*. Uncountable times, I had to take care of two sick boys: the Eldest due to NF1 and the Youngest due to the NF1 of the Eldest.

For some years, the Eldest and Youngest slept in one room. Nowadays, they do not, except when the Eldest feels very sick. *'Mam, can I sleep with the Eldest? He doesn't feel well*.' Then, the Youngest picks up his mattress and lies down with the Eldest to keep him company and takes care of him with a wet wipe and some water. This way, the Youngest also keeps me updated.

For the future, the brothers have decided they work together as a professional football player and manager or as a YouTuber and vlog editor. They share some gaming 'friends', although they differ almost six years in age. The biggest challenge between the two brothers is finding a way to not irritate each other with their differing energy levels. The Youngest has too much energy, whilst the Eldest has too little energy, which clashes. Due to his age, the Youngest now understands better what the Eldest is sad or angry about. He has learnt to live with the uncertainty concerning the Eldest. Recently, my two sons can stay at home together while I am away for a couple of hours. For years, I could not leave them together, which felt very suffocating.

For some quality time with his dad, they both are actively involved in the local football club. He also loves to have a 'girl talk' with his sister and mam. If possible, the Eldest will go and see a football game of his brother. Sometimes, my three children play cards together, which makes me incredibly happy.

3.2.4. 'The Father'

The Father of our three children, my husband, has always worked. When we met, we were both career-oriented. He stayed that way, whilst I stayed at home to raise the kids. This agreement was based on the premise that it would be 'my time' after a couple of years, when the children would need less attention.

Due to his responsible job, he has a lot of stress, which he also brings home. Coming home, he needs rest, but at home there has been no opportunity for rest. For years, he avoided this by coming home late, whilst I went upstairs with the children at 7.30 pm. I tried not to bother my husband too much, so he could do his job well. This way, we barely had time together, although he did support me regarding my voluntary work.

During puberty of the Eldest and the Daughter, it became too much for me. My parents had given us much help during these years, but the Daughter and the Youngest did not want to stay that frequently with their grandparents anymore. We had to reset our roles, which meant a bigger involvement for him in taking care of our family at home. At first, this was particularly challenging for everyone, but finally it also resulted in a closer bond between him and our children.

My husband has the ability to put things into perspective, from a level-headed point of view, which gives me strength to go on. He tries to give each family member personal attention and tries to undertake some activities with our family as a unit. His hobbies involve doing voluntary jobs as a board member. The fact that we had to continuously adjust our expectations has also been frustrating for him, but he stayed positive about this: '*It is like it is . . . We have a special family, and we make a good team*'.

3.2.5. 'The Mother'

The moment we chose to have children, we envisioned them being able to experience the whole world, instead of this world of disease. Our lives are completely controlled by our family's circumstances. We have (had) no freedom of choice; we are just forced to go on and keep dealing with one worry after another. As a couple, we were surviving rather than living. We barely had privacy or time for each other. Our relationship is a matter of hard work, acceptance of the situation, and giving and receiving.

Through these years of survival, we continuously had to adjust our expectations. Many times, we did not know whether the health issues of the Eldest were due to NF1, his hydrocephalus, ASD, anxiety, or just a common flu, and whether we had to go to the hospital or not. It is hard to combine a family life with a stressful job and the many worries about our children. It is hard to keep each other informed about important issues concerning the kids and work. We managed it, although it has been a hell of a job. Somehow, we found some middle ground in respecting each other's need for personal time and raising a family together. Throughout the years, we have become incredibly resilient. Together, we have learned to prioritize: nothing is more important than our family.

Still, we both worry much about our children, not just the Eldest, although questions especially concern him. What more will NF1 bring him? We know that he will not be able to work because of the disabilities. Will he find some happiness in his life? Will he have friends? We are convinced that the Daughter and the Youngest will find their way. However, we are also afraid of what the future will bring them, having a brother with these kinds of limitations.

As the Mother within this family, I often feel like an acrobat: trying to juggle all the plates in the air and thus occupying several positions at the same time, to keep everything running. This means taking care of ten things at the same time and doing it right. Some things I already do very routinely, but often, unexpected issues show up at inconvenient moments. Making plans, rescheduling plans, cancelling plans. I go from 'hope' to 'no hope' to 'hope' again.

Unfortunately, as an acrobat I cannot divide myself into being at different places, doing different things and pleasing everybody at the same time. This feels like a shortcoming to me. Simultaneously, I must take care of myself, because otherwise all the plates will fall on the ground and shatter, and we will have an even bigger problem. Often, I feel stuck in the performance of an acrobat. Those plates can also be seen as different roles I have to perform as a mother, wife, secretary, manager, representative, doctor, nurse, caregiver, friend, taxi driver, teacher, housekeeper, activist, etcetera. I have experienced a lot of emotional and

physical problems due to exhaustion. My life consists of falling, standing, and surviving. All family members experience this, but they also depend on me.

Having been raised in the 1970s and the 1980s, I was told to make sure I could be financially independent. The reality is that I had to quit my job to take care of my family. Part of me has accepted this situation, but a part inside me still refuses to accept this. At moments, I felt like an outsider, because it seemed that everybody around me had a job. Although my days have been full of activities, it still felt inferior. Rarely, somebody asked about my days, while it seemed normal to talk about *their* jobs.

Being in charge is almost impossible when having to deal with chronic illness. Nothing seems to be logical or predictable; nothing is for sure. This takes a lot of energy from my family and me. It is challenging and frustrating at the same time. I am not sure where the last 15 years of our life have gone.

I have had many feelings of guilt towards the Eldest, that he had to suffer so much, towards my husband that he had to work that hard, and towards the Daughter and the Youngest that they had to experience all this worrying.

My experiences have changed me, as I am more flexible and more assertive. I care less about what others think of me. I am tired of meeting some people while keeping an open mind, because they barely show(ed) interest. I would rather spend my time with my nuclear family members, my parents, or other people who did not (do not) stay away. Meanwhile, puzzle pieces came together for me personally. I immersed myself in the world of NF and rare diseases, becoming an active volunteer at a national patient organization and a PhD researcher. This gave me valuable new friends and acquaintances. It takes time, but it mainly gives me positive energy and a feeling of belonging. This would not have been the case if my Eldest did not have NF1. Therefore, maybe I have to say: *'Thanks to the Eldest'*.

3.2.6. Extended Family and Friends

The Dutch saying, that you really learn to know your friends and family when you are in need, is absolutely true. The disappointment has been huge, especially when it comes to people from whom I expected more support. Although we always have been open about our life with NF1, several persons do not seem or even *want* to realize what it really means to have a chronically ill child with a rare disease. It seems as if the longer this disease takes, the less interest people show. Apparently, an adult ill child seems to be less interesting than a young child, even though this is still our life!

Any practical support is also more difficult for people to offer, except for some. Admittedly, I also barely ask for help, because I am afraid to bother others, who always make vary clear that they have a busy life. The people I have *always* been able to count on, both practically and emotionally, are my own parents. As grandparents, they have taken care of many things that we seldom (have/) had to ask for, such as taking care of the children, doing laundry, cooking, driving, buying groceries, gardening, etcetera, but also listening and empathizing with our family. Recently, due to aging and concomitant health problems, we try to lessen their burden, and also as the children are becoming more independent, the balance has changed.

3.2.7. Family as a Unit

It is difficult to undertake an activity with the whole family, due to different ages, and interests, but mainly the (im)possibilities related to NF1. Sometimes, we try to go to a movie, have dinner at a restaurant, or (if possible) go on vacation as a family, always considering the health of the Eldest and the presence of a good hospital. Going outside the house is becoming more burdensome or sometimes even impossible for him.

We try to put things into perspective with our sense of humor. During hard times, we often know how to use humor to make the other one smile, to make situations more bearable. Not everybody understands it or appreciates it, but we do not care as long it does not harm anyone else. Having this is important for us in order to carry on, to survive.

Furthermore, our two dogs play an important role, as they provide distraction, especially for the Daughter and the Youngest.

The fact that our nuclear family and its individual members have survived these years makes me both proud and sad at the same time. All the family members are marked by several scars. Our life is an unpredictable challenge. It is continuous hard work and we accept that we have other challenges as opposed to other families. Although scarred, I also think of our children as world citizens who have learnt much more about life and caring than many others ever will during their lives.

4. Discussion

This co-constructed autoethnography clearly shows that families and the world they live in are complex [23] and that any concept of FQoL that does not try to understand the interrelations within a family would be incomplete [14,83]. Daily life of the family in question seems to follow a pattern, showing a strong susceptibility to *domino-effects*. Whenever the Eldest becomes ill or must go to hospital, his brother and sister often follow suit and also become ill. This then impacts the Mother, as she feels more stressed or overburdened, which in turn affects the other family members [84,85]. About this impact, Sales states [8] (p. 40): 'Family should be recognized as an integral component of the patient care system' and their needs and difficulties should also be considered by professionals and researchers [86].

Living with NF1 has (had) a huge impact on the whole family and its members, which the Mother metaphorically describes as having *scars*. The many years of pain, uncertainty, guilty feelings, and suffering have created wounds and left many scars on every individual family member—literally and figuratively; old and new; visible and invisible, (re)opened and closed; strong and weak. The metaphor of the scar aptly reflects how NF1 has led to lived experiences which they rather would *not* have experienced but have also made each member more resilient.

This article also shows how each family member starts to relate to each other differently due to living with a rare chronic disease. As studies have already shown, in such cases it is often the Mother who mainly takes care of the child with a chronical illness/disability [87] (p. 20), while fathers often work as breadwinner [88,89]. In this case, the Mother made a choice to commit herself to mothering her children fulltime, which also meant a loss in terms of career and income, as well as self-esteem.

Relating to each other differently also means feeling guilty as a parent, as is the case with the Mother and Father, because in their view they structurally give too little attention and care to their other children. Brothers and sisters often turn out to have an ambivalent perspective on the involvement of their parents in the life of their relative who needs intensive care. An experienced lack of attention, a feeling of being hindered in building a peer relationship with that relative, worrying about their sibling with a chronical illness/disability, and having more care-taking responsibilities are often mentioned [90–98]. In this case, the Youngest and the Daughter can build a relationship with the Eldest but do feel hindered by the additional worries that they seem to take on. Ultimately, living with a rare disease has significant social impact for each family member, resulting in not being able to participate in society, loss of contacts and friends, and little sense of belonging, all important for developing one's 'horizontal' identity [9] (p. 2). Part of this is also caused by the fact that families are often confronted in their daily lives with incomprehension (and sometimes rejection) of the outside world about living with a child with a chronic disease/disability, as was experienced by the Mother in this case. Boelsma et al. [83] (p. 117) also found that 'a lack of acceptance and understanding can be impeding factors for the FQoL. These factors could also contribute to experiences of having a lack of social support.' Fortunately, for this family the grandparents have been important as 'social supporters' [99–102].

In addition to the emotional, financial, and social impacts [3,84,85,103], there is another, more implicit, moral impact. The Mother refers to her *'gut feeling'* which is constantly there,

especially in recognizing the needs and appeals that her Eldest son and the other family members make on her, as described by philosopher Levinas [104]. This family narrative also shows that 'care' is ultimately a praxis, which needs to be considered when researching FQoL. As the fields of care ethics and family ethics have persuasively argued, caring is innate to human relations and occurs *to* every person and *by* every person, including the person with the illness/disability [4,105–109]. What is more, care practices continuously change over time. There were times the Mother could not go out, which felt suffocating. However, now that the children are older and more independent, the Mother finally experiences more autonomy. However, she still views herself as an *acrobat*. This continuous adaption of changing situations and roles demands an enormous ability for all family members to be resilient [10]. Resilience can be viewed as 'the ability to withstand and rebound from disruptive life challenges' [110] (p. 527), [111]. For this family, certain factors contribute to this resilience. For instance, if the Mother did not have her voluntary work, she would have felt less satisfied with staying at home and more vulnerable to caregivers' burden [89,103]. If she were less satisfied with her situation, this would have influenced the experienced well-being and feelings of other family members. Humor is also an important strategy in this family to cope with adversity. This form of coping humor has been defined as 'the use of humor in which individuals seek to manage the effects of one or more life stressors. It involves efforts aimed at maintaining a humorous perspective in the face of adversity' [112–114].

5. Limitations and Uncertainties

Critics may argue that this narrative lacks completeness and depth, as it all centers ultimately on one family. However, this article deliberately chose to explore a single family unit with attention to each family member, in order to gain insight into the inherently complex, multi-layered context of such a family. This also included trying to grasp the interrelations between the individual family members and how this accounts for the FQoL of that specific family.

What is more, true completeness is not practically possible [115]. Therefore, can this incomplete story, told as it is here, be of value to other families living with rare diseases? How transferable is this story generally and how relevant is it to others?

We contend that it is not the aim of this research to generalize the findings to a larger population of all families living with NF1 or even other RD. Indeed, the aim of this study is to help us to 'understand the particularities of a case from multiple perspectives, leading to case reports that describe the meaning and context of that particular case' [116] (p. 1151), because, as Abma and Stake have so eloquently pointed, 'the paradox of case studies research . . . .(is that it) reveals the universal in the particular' (Ibid) (p. 1159).

6. Conclusions

To our knowledge, this is the first study to examine FQoL in a way that foregrounds all of the family's (private) experiences and subjectivity, as opposed to attempting to limit these [74]. This article shows the importance of viewing family dynamics as a continuously changing practice of care. In the case of this family living with the rare disease NF1, both the individuals within a family and the family unit are impacted emotionally, financially, socially, and morally, which is in part caused by the constant uncertainty they have to live with, and caused in part by the variability of symptoms, progressiveness, and the incurability of the disease.

Future studies into (F)QoL of living with rare chronic diseases should acknowledge this inherent and interrelated complexity, which, as Ablon [117] and Carrieri [29] have pointed out, will increase the more uncertain conditions become, as is the case with NF1. One way of doing this might be by placing the particular (the family) at the center, thereby also addressing the relational nature of knowledge [74].

Using co-constructed autoethnography and metaphors as an analytic tool has proven to be a particularly suitable approach in gaining more insight into the complex family

dynamics that characterize families living with rare chronic diseases. What is more, writing this article has been a care praxis of its own (Ibid).

Author Contributions: Conceptualization; I.D., A.S., A.N. and T.A.; methodology, I.D., A.S., A.N. and T.A.; validation, I.D., A.S., A.N. and T.A.; formal analysis, I.D., A.S., A.N. and T.A.; investigation, I.D.; resources, I.D.; data curation, I.D.; writing—original draft preparation, I.D. and A.N.; writing—review and editing, I.D., A.S., A.N. and T.A.; supervision, A.S., A.N. and T.A. All authors have read and agreed to the published version of the manuscript.

Funding: This research received no external funding.

Institutional Review Board Statement: According to the Medical Ethics Review Committee of VU University Medical Center (registered with the US Office for Human Research Protections as IRB00002991; FWA number: FWA00017598), the Medical Research Involving Human Subjects Act (Wet op Medisch Onderzoek, WMO) did not apply to our study into FQoL: Niet WMO-advies METc verklaring VUmc 2015.485.

Informed Consent Statement: Informed consent was obtained from all subjects involved in the study.

Data Availability Statement: The data generated and analyzed are not available publicly.

Acknowledgments: We would like to give a special thanks for their contributions to: Ivan Brown, Geert Van Hove, the anonymous reviewers, and the family members: the Eldest, the Daughter, the Youngest, and the Father.

Conflicts of Interest: The authors declare no conflict of interest.

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