Coping with DCAA outside its community

What are the experiences and needs of DCAA family members living outside of Katwijk?

Author: H.F.J. Haasnoot (2786712)
H.f.j.haasnoot@student.vu.nl
Health & Life Sciences Thesis (6EC)

Patient Association HCHWA-D
Onsite Supervisor: Msc J.R. van Rijn

Vrije Univesiteit Amsterdam
VU-Supervisor: Assistant professor K.M.J.P. Pijpers (PhD)
Second Assessor: Assistant professor S. Duijs
Date: 30-05-2025

Table of contents

Abstract

Dutch type Cerebral Amyloid Angiopathy (DCAA) is an inherited disease in which carriers of the gene develop brain hemorrhages around the age of 55. The disease has a great impact on families that carry the gene. DCAA originated and still mainly occurs in the Dutch village of Katwijk, but there are also families outside of Katwijk who are affected by the disease. Because the disease is rare, families outside of Katwijk possibly feel isolated in their struggles with DCAA. To explore the experiences and possible needs of those people with DCAA outside of Katwijk, ten semi-structured interviews were conducted with a variety of people affected by DCAA, including risk carriers, gene carriers, patients, and someone who lost their partner to the disease. The interviews were analyzed thematically using open, axial and selective coding. The interviews showed that coping with DCAA outside of Katwijk happens in different ways and brings with it various problems. People often feel the need to talk about it, but mostly their environment cannot help them, because of lack of knowledge about DCAA. Lack of knowledge also causes that people are not treated the optimal way by the healthcare system. Help is sought from the DCAA patient association, but people are reluctant due to different social and geographical barriers.

Introduction

Dutch-type Cerebral Amyloid Angiopathy (DCAA) is a rare hereditary disease. DCAA is caused by an autosomal dominant gene, which means offspring of mutation carriers are 50% at risk. The mutation most likely occurred in a person from Katwijk, a Dutch village, centuries ago. Because the villagers tend to stay in Katwijk, most mutation carriers live in Katwijk. As a result of this, DCAA is also informally known as 'Katwijk disease'. Whole families are together engaged concerning DCAA because, beyond the psychological impact of the heredity, families also play a crucial role in caregiving and supporting each other. Feelings of fear and uncertainty, as well as survivor guilt can appear within these families. Because of the rarity, much of the knowledge about living with DCAA resides within DCAA families living in Katwijk, rather than in formal healthcare structures. Families with DCAA can consist of patients, gene carriers, non-carriers and risk carriers (those who could carry the gene, but have not been tested), and their partners, informal caregivers, and other family members. Although the disease still mainly occurs in Katwijk, a significant number of DCAA gene carriers have moved elsewhere, causing the disease to appear in other places as well. Virtually nothing is known about living with DCAA outside of Katwijk. Therefore, in this study, I will interview 10 members of DCAA families not living in Katwijk and analyze the gathered data to explore their experiences.

People with the mutation have a high chance of (repeated) cerebral hemorrhages, between the age of 45 and 65, on average at 55 (3). These hemorrhages can be fatal, or cause many different symptoms, leading to a diverse clinical picture for each individual patient. As the disease progresses, individuals may develop cognitive symptoms, such as memory problems, speech problems, physical failure, epilepsy, personality changes, psychiatric complaints, vision problems, overstimulation and difficulties with concentration or planning (4). Other symptoms may include brief, stroke-like symptoms, as well as seizures. In the later stages, many patients develop dementia due to ongoing damage in the brain (5). Currently, there is no treatment for the disease, but several studies investigate the disease and explore different possible ways to treat the disease (2). For more biomedical and clinical information, see appendix I.

The progressive character, the caused symptoms and the young age of patients of DCAA make living with the disease very difficult (6). Difficult choices must be made related to the disease, like for example whether to do preventive testing and choices about having children naturally or through predictive genetic testing. In previous studies, 14% of the study population with DCAA was affected by depression and anxiety (6). Research in other hereditary diseases showed that preventive testing can lead to severe psychological issues, including suicidality, see appendix I (8,9). This shows that appropriate and suitable care should be available for everyone within in a DCAA family.

Awareness of DCAA centers only in Katwijk and the region surrounding it. In other places in the Netherlands, awareness of DCAA is scarce. In Katwijk and surroundings, most DCAA family members have a GP that knows of the disease, a local healthcare provider offers a range of therapies within their DCAA expertise team (i.e. speech and physical therapy, social work and case managing) and the local hospital offers specialized care as an expertise center. The given healthcare is mostly comparable to patients of other

cardiovascular diseases or based on the current symptoms of the DCAA patient. Outside of the region, there is no specialized healthcare and especially patients have issues getting proper care. They tend to be treated like other patients who have suffered a hemorrhage or as patients with dementia, but care of both categories does not fit their needs.

In Katwijk, there also is a patient association based, which aims to improve the life of all DCAA family members. They provide information through their website, newsletter and social media. They organize peer support through informational meetings where members of DCAA families and others involved can meet each other, to talk about the disease and its challenges and in that way find support from each other, because they are all in the same situation. The association also aims to bridge the gap between their community and experts through 'theme-nights', advocate to improve healthcare and are a partner in scientific research. The association works together with many stakeholders, such as Leiden University Medical Center (LUMC) and the Dutch CAA Foundation, see appendix II. The work of the patient association is very valuable to DCAA family members in Katwijk and its surrounding area, but its reach remains limited beyond this region, despite the presence of patients living elsewhere. Therefore, this study, requested by the DCAA association, will be an exploration of the experiences and possible needs of members of DCAA family members, who reside outside of Katwijk or surrounding area. New data will be gathered through 10 semi-structured interviews with DCAA family members to identify the experiences and possible needs of the DCAA family members who reside outside of Katwijk.

Methods

Study design

A qualitative research design is used to explore the experiences of DCAA families and others involved, who reside outside of Katwijk. Semi-structured interviews are done to gain in-depth insights into the lived experiences of the interviewees. To collect rich and contextual data on the variety of situations, as described in the introduction, leading to a variety of possible experiences and needs of people within DCAA families, this research will investigate DCAA family members in different categories. Therefore, the inclusion criteria of the respondents were set on everyone who has to deal with DCAA in their family, not living in or near Katwijk. That includes symptomatic carriers, presymptomatic mutation carriers who are aware of their genetic status, family members at risk, and other family members/informal caregivers. The same topic guide was used in every interview.

Participation recruitment

To recruit the broad range of possible participants, convenience sampling was used. The DCAA patient association approached members with a different situation and background. They made a call to action via their social media and some potential participants were contacted personally by email by a DCAA patient experts connected to the association. In the call, they were informed of the study design and the researcher. When interest to participate was expressed, the researchers connected

with the potential participant. Possible respondents could sign up on the website of the patient association. Due to the time limit of three months, and the limits of the population because DCAA is a rare disease, the maximum number of participants was set on ten. Appointments were made via e-mail. The interviews were done to interviewees of different sexes, ages, phase of disease progression and place of residence (Tabel 1). Six participants were female and four were male. Two participants were symptomatic (patient), four were gene carrier, one was risk carrier, two were non-carrier, and 1 has lost her partner due to the disease. Five participants live in South-Holland, two in Utrecht, one in North-Holland, one in Gelderland, and one in Drenthe.

Participant	Sex	Disease state	Province of residence
1	Women	Risk carrier	Gelderland
2	Women	No carrier	Utrecht
3	Women	Partner deceased	Drenthe
4	Man	Gene carrier	South-Holland
5	Women	No carrier	North-Holland
6	Women	Gene carrier	Utrecht
7	Women	Gene carrier	South-Holland
8	Man	Patient	South-Holland
9	Man	Gene carrier	South-Holland
10	Man	Patient	South-Holland

Tabel 1. Participant Description: Sex, disease state and province of residence of the interviewees.

Data collection

After receiving informed consent (appendix V), semi-structures interviews were conducted. To lead the conversations, an interview guide was prepared based on the literature and previous knowledge of for example the patient association, see Appendix III. In the first interview, participant 1 shared her experiences extensively and the example questions turned out to be unnecessary to collect rich data. It was therefore decided that with the following interviews, it would be better to use a topic guide (appendix IV). 7 interviews were held at the residence of the participants, according to their preference. The interview with participant 7 was held in a meeting room of the patient association, according to the preference of the interviewee. The interviews with participants 1 and 9 were held online, according to the preference of the interviewees. The Informed Consent stated that the interview would take 45 to 60 minutes. Frequently, interviews lasted longer than 60 or shorter than 45 minutes. The interviews were recorded and transcribed and stored on a secured drive.

Data analysis

The transcribed interviews were coded and analyzed in an iterative process. The data was analyzed thematically, which is a method for identifying, analyzing, and reporting patterns (themes) within the collected data, according to *Braun and Clarke: Using thematic analysis in psychology (2016) (16).* With this flexible but structured approach,

meanings and insights from the interviewees could be explored. Three different types of coding were used, according to the book 'Qualitative research design' by Dimitri Mortelmans (17). The first way of coding was open coding, which involves breaking the data into smaller segments and give it descriptive labels, according to their meaning. This way, important concepts of the answers of the interviewed people emerged from the data. The open coding was done with the help of the computer program Atlas.ti (10). When every transcript has been coded open, the data was coded axial, in which relationships between the different codes that did emerge with the open coding were found. By making connections between the found concepts, underlying patterns in the data emerged. This was done manually. Every code has been printed and sorted by the researcher. Finally, using selective coding, the overarching themes out of the different relationships/connections found in the phase before opened up, so a combined identification of the experiences of the participants could be made.

Ethical consideration

The potential interview candidates were approached by the patient association. The reason for this is that the candidates mostly are already familiar with the patient association, so therefore trust has already been created. This trust relationship is important for the quality of data that will be gathered during the interviews and to safeguard the possible relationship of care between the patient association and its constituents after the research has been completed. The researcher has been aware of the sensitivity of the topic of the interview and therefore asked the questions carefully during the conversations.

Before the interview and recording started, informed consent has been obtained, see appendix IV. The interview started with an informative introduction, in which among others it was made clear that participation is voluntarily, the interview could be paused or stopped at any time, and that anonymity and privacy would be preserved. After transcribing the recordings, the data has been anonymized to ensure privacy of participants.

Results

Participant 1 is a young woman from a DCAA family. Her father and aunt are gene carriers, which makes her a risk carrier; she has not had herself genetically tested. Her father is DCAA patient and already had two hemorrhages, of which he recovered quite well. She states that the disease does have quite an impact on her life. Because she does not live in Katwijk, she feels a sense of distance to the patient association and to the specialized care. She is not actively involved with the patient association, because of emotional and geographical barriers. She follows developments about DCAA research through the social media of the patient association.

Participant 2 is a middle-aged woman, whose grandmother and mother both passed away at a young age due to cerebral hemorrhages, but both have never been tested on DCAA. She decided she wanted to get genetically tested because her children were thinking about starting a family. Her GP had never heard of DCAA, so she had to collect proof herself (a family tree), with help from the patient association, to convince the GP

to refer her for a genetic test. She had to wait several months for the test results, which she experienced as a difficult time, among others because she did not tell her children about the test. She is no gene carrier. During the time waiting on the test and after sharing the results, she struggled with the misunderstanding of the people around her. She did not join any event of the patient association, because her status was unknown, and she was afraid she would feel guilty if she turned out not to be a carrier.

Participant 3 is a middle-aged woman, whose partner passed away a few years ago after multiple brain hemorrhages, due to DCAA. One of her two sons is no carrier; the other one is risk carrier. She feels like an outsider of the patient association, due to the geographical distance, and religious differences with the citizens of Katwijk. After her partners death, she experienced a lack of support. She mentioned the challenges for partners, with at-risk children, and that there should be more attention for the psychosocial impact of the disease.

Participant 4 is a young man who recently got a positive result of his genetic test, after his brother also tested positive. His father had never shown symptoms until his death due to another disease, and therefore this participant and his siblings assumed they would not carry the gene. The diagnosis has changed his way of life. For example, he focuses on quality time with his wife and young children and spend less time on work. He experiences a lack of disease awareness in his local healthcare system, and therefore advocates for better information sharing, in which for example practical resources could be helpful. He is very open about his situation, and attends to join meetings of the patient association, but he does not come often due to his busy life.

Participant 5 is a middle-aged woman who recently got a negative result of her genetic test. Her mother passed away from the disease, after several cerebral hemorrhages. She gets psychological help because she is struggling with the result, because for many years, she was convinced that she was a gene carrier. This belief impacted her daily functioning and well-being that she decided to get tested. She barely has communication with others with similar situations. She does not visit Katwijk, due to geographical and emotional distance from its citizens. She mentioned that she would have liked local peer support during her mother's illness and her testing process.

Participant 6 is a young woman, who is a gene carrier of DCAA. Her father and uncle passed away from the disease. When she was younger, she was not really concerned about the heredity of DCAA. She did not think of the fact that the disease her father was suffering from could also affect her later. Now that she is older, and thinking about having children, she is more aware of the disease and its risks. She wanted to actively contribute to fundraising for DCAA, so she has started a fundraising campaign. In the past, her only connection with Katwijk was visiting her grandparents, but now she does not do that anymore and feels no connection with Katwijk. She mentioned that she oftend finds the mood at the patient association a bit too sad, therefore she does not often join their meetings. Especially when she feels good, she does not want to think about the disease, so she does not visit the patient association. Next to that, she finds Katwijk a bit far from where she lives and would like to have meeting with other people affected by DCAA closer to her own home.

Participant 7 is a woman who carries the DCAA gene, but has no symptoms, even though she is older than the average age for symptoms of DCAA. The diagnosis came as a shock. She had never heard of the disease, because she got the gene from her father, with whom she never had contact. She was tested in the hospital where she was working. She went through the testing process without psychological support, something she regrets in retrospect. At first, she was traumatized by 'Katwijk', but this changed over time and after she had met people from Katwijk. She has been declared unfit for her work in the hospital; therefore, she is now doing volunteering work.

Participant 8 is a physically active middle-aged man. He is DCAA patient. Twenty years ago, his mother passed away due to a brain hemorrhage. The family did not know about DCAA. Five years after his mother's death, he experienced his first brain hemorrhage, which was marked by severe migraines. Over time, he suffered multiple hemorrhages, which has left different restrictive symptoms. These include impaired vision and dizziness, which have significantly impacted his daily functioning and quality of life. For example, he is not able to race a bike anymore, and therefore he started running, to stay physically active. Furthermore, he was declared unfit for work, he sometimes has troubles to follow conversations, he no longer drives, and he stopped going on vacations.

Participant 9 is a young man; he is a gene carrier of DCAA. He regularly supports his father (participant 10), who is patient of DCAA. His grandfather and uncle both passed away from DCAA. When he was younger, his GP advised against testing, but the disease always had an impact on his life. Because he was aware of his risk, he has always been careful with alcohol, smoking, and drugs. He chose to study music, which he truly enjoys, keeping in mind that he could get symptoms at a young age. He chose to get tested because he and his girlfriend were thinking about children, he is now starting with preimplantation genetic testing. After his test result, he switched from freelance work to salaried employment, following advice from the patient association. In his supportive care for his father, he often struggles with where to turn for support. In his view, a central point for guidance would be a major improvement.

Participant 10 is a male middle-aged patient. He is the father of participant 9 and finds it deeply distressing that his son is gene carrier. During his first cerebral hemorrhage, he went to his GP, who did not appear to understand the implications of DCAA. The GP dismissed the possibility of a cerebral hemorrhage, sending him home without appropriate care. Initially he perceived his first hemorrhage as relatively mild, but now he is experiencing restrictive symptoms like fatigue and visual disturbances. These symptoms have led to difficulties in his daily life. He experiences misunderstanding from those around him. He is currently in the process of being declared unfit for work. He finds the healthcare system procedures confusing and is considering moving to Katwijk, because he believes the expertise knowledge about DCAA is located there, leading to better support, and understanding of his situation.

The stories of the 10 participants described above vary widely but have a common core. Figure 1 shows a schematic model of the common themes which opened up during the interviews. The themes will be explained in the following text.

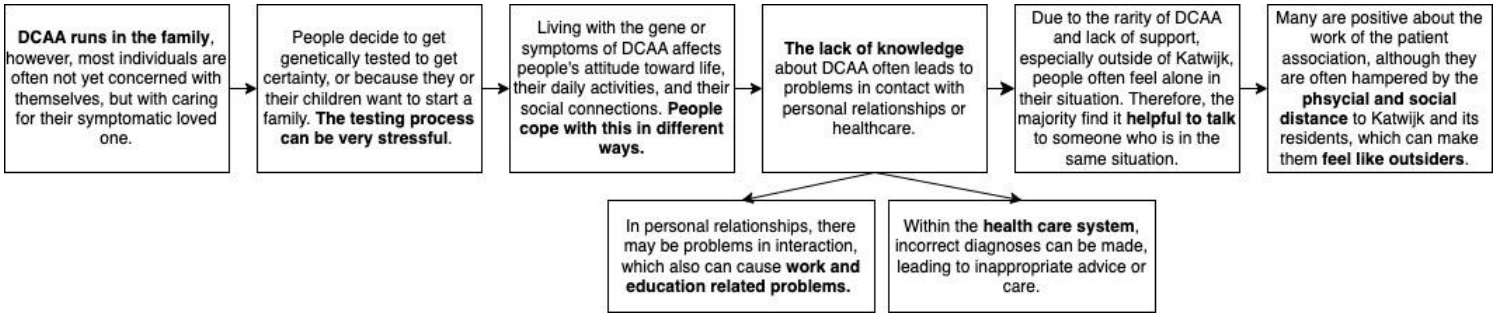


Figure 1: Schematic model of upcoming themes in the interviews.

1. **'I already knew it, but it wasn't talked about'**

For many participants, the impact of the disease can already be seen at sick (grand)parents or uncles and aunts, at least if there is still contact with the family, which is not always the case for people outside of Katwijk. Seeing family suffer from the disease also can cause anxiety because of the knowledge of the heredity of the disease.

However, the focus is often not there yet, and therefore the awareness of the disease lacks. Worries for one's own future usually follows the death of a parent or when individuals are confronted with DCAA through a family member suffering a hemorrhage or personally become symptomatic.

Although the disease often is visible, it is not spoken about in the families. The family is often very vague. Historically, there is a taboo surrounding DCAA in families who are aware they carry the gene. Some DCAA family members grow up in an environment where the disease is surrounded by secrets.

> "Back then, it was all a bit vague—nothing was really said out loud. It wasn't very well known at the time either, when it was actually happening. There was this sense of secrecy, like, 'If we don't talk about it, then it's not really there,' you know?" - participant 3

2. **'It's like full-on stress all the time'**

Not every at-risk DCAA family member decides to do predictive genetic testing. Some individuals at risk make a conscious decision to not know their mutation status. Some decide not to get tested because there is no treatment available. Others worry about practical implications of a positive test result, like the influence on mortgages and insurances. Some have a non-supportive environment, such as uncomprehending employers and GP's (described in more detail later).

Reasons for getting tested can be that people cannot live with the uncertainty, or the wish (of offspring) to start a family. Some people experience no difficulties during the testing

process (that can take up to a year), but others experience it as a harsh, long, and lonely process. Some people do not tell others about the testing process. Some experience a lack of available (online) information and support after receiving a positive or negative result.

"I felt emotionally burdened for eight months. Right up until the results came in, I was, well, incredibly tense — my stress levels were through the roof. I kept thinking, *Which way is this going to go? What will I do if I have the illness? What will that even look like? And how am I going to tell the kids?*" – participant 2

3. 'Get more out of it'

Many participants adjust their lives when they receive a positive result from the genetic test. Not everyone adapts their life, but many make different choices with the disease in mind, for example about their studies, how remaining lifetime is spent and about family planning and PGT. Participant 4, for example, mentioned that after his positive test result, he wanted to work less and do more fun things; he wants to enjoy the kids and finish the bucket list. Participant 1 is now in the middle of her studies, but she expressed a strong desire to begin living her own life as soon as possible, describing an internal feeling of being in a rush.

Also, people try to adjust their life to have as less stress as possible; they stop working too busy shifts and want to live healthy. Some also stop working nightshifts to achieve a healthier sleep pattern.

Furthermore, the daily life is adjusted due to DCAA because the symptoms after a hemorrhage, such as fatigue and visual impairment, cause that people can for example read, sport and work to a limited extent or not at all. Several participants also want to actively participate in the search for a medicine and therefore raise money for research and/or participate in research themselves.

"The only thing that bothers me are all the limitations I have — that really does affect me." – participant 10

4. 'It is too abstract for people to understand what it means'

The lack of knowledge about DCAA makes the disease very abstract for the environment of DCAA family members outside of Katwijk; this can lead to problems with study-related relationships if the participants are students, or with employers and colleagues if the participants have a job.

During the education and internship of participant 1 and 6, the people involved with their education and internship did not understand the emotional impact the individual themself or a parent undergoing predictive genetic testing, or a parent becoming symptomatic, which influences the possibility to focus on the internship or write a thesis.

Situations caused by DCAA can lead to unpleasant misunderstandings among colleagues and employers, because DCAA family members sometimes make (more or less forced) choices of which others do not understand why. Participant 2 adjusted her work schedule during the

stressful period of her testing process. Participant 9 was self-employed but became an employee in education. Participant 4 got a positive test result and decided to work less because he wants to spend time with his family. Participants 7, 8 and 10 had to stop working before reaching retirement age, due to DCAA.

"My manager says things like, 'I could die young too,' and 'We all get tired sometimes,' that kind of stuff." – participant 10

5. 'My GP indeed had absolutely no knowledge about the Katwijk Disease'

Lack of knowledge about DCAA from health care providers, such as general practitioners, pharmacists, neurologists, rehabilitation centers, psychologists, physiotherapists, et cetera can lead to many negative situations. For example, the GP of participant 2 resisted when participant 2 wanted to get genetically tested on DCAA, so she provided a family tree as evidence to convince the GP to refer her. Participant 7 entered the test trajectory without psychological guidance, after being referred by the neurologist at the hospital where she works, who apparently did not fully understand the impact of the test.

Furthermore, due to the young age of patients, health care providers often do not diagnose them with a cerebral hemorrhage, which can have disastrous consequences. Bad advices are given and inappropriate and inadequate care can be provided. The mother of participant 5 was once referred to psychiatry, when she had a hemorrhage. Participant 10 was sent home by his GP while having a cerebral hemorrhage, because the GP stated that he was too young to suffer from a cerebral hemorrhage. Participant 8 experienced resistance when he wanted to leave the rehabilitation center after he had recovered sufficiently from a hemorrhage, because it was mandatory to stay for a certain period. These misdiagnoses make people lose confidence in the healthcare system.

"The neurologist was surprised, like, 'guys, you look great, there doesn't seem to be anything wrong—you can walk, everything seems fine.' They did another MRI, and in the end, it turned out to be an aneurysm, a brain hemorrhage after all." – participant 8

"Brain hemorrhages at that age are really rare—they usually happen to older people, and they often can't do much anymore. But my dad is still really fit, he can basically do everything. And yeah, you have to keep explaining that over and over again, which is really exhausting. You figure out what works for yourself, sometimes you actually know better yourself, than the person who's supposed to refer you." – participant 1

Some participants visited a psychologist, but they indicated that they often did not receive adequate help there. The psychologist did not sufficiently understand the stress surrounding heredity and the *'sword of Damocles'* that hung over the heads of the DCAA family members. Some psychologists said that the DCAA members were coping to well to receive mental care, although that was something the participants certainly needed.

Some participants expressed a need to have an understanding of disease progression. However, MRI's are usually limited to when someone has a suspected hemorrhage or as a part of scientific research. Some feel like that leaves them lacking understanding of their health status.

Participants indicated that the communication between the different caregivers is not good, while it is important that all caregivers have a good understanding of the situation surrounding DCAA. The participants indicated a need for overarching care or something like an information leaflet. Participants have the feeling that good care is only available in Katwijk and that you have to live there, in order to access it.

6. 'No one can really put themselves in your shoes, and I think I miss that'

The misunderstanding from the environment and healthcare professionals outside of Katwijk can make people feel alone in their situation. Participant 6 mentioned that no matter how sweet and understanding the environment is, she wants to talk someone who also has it, or had it, or knows someone who had it. That is a need that other participants also share. The feelings of loneliness can be reinforced by symptoms such as prosopagnosia (the inability to recognize faces), which was the case with participant 8 and 10 (both symptomatic patients).

> "You don't recognize them, so then they can just — and that's hard — they can just ignore you, or at least that's what you think. And then, if you're already feeling lonely, you end up feeling even lonelier. And that understanding — when you try to explain, like, 'I have the Katwijk disease,' they don't understand it." – participant 10

Not every participant feels the need to talk about their situation, but most participants do. Often, they do not have anybody to turn to, because there is no one in their environment in the same situation, and the participants say that you only feel truly understood by people who are really going through the same thing. The environment of DCAA members outside of Katwijk almost always has absolutely no idea what DCAA entails, the family burden, the tensions surrounding testing, the fear of hemorrhages, the consequences of hemorrhages, etc. Participant 2 had not informed her children that she was at risk of DCAA and about her choice to get genetically tested. When the test showed that she was not a carrier, her children reacted very nonchalantly, while she herself had been in extreme tension for 8 months.

People dealing with DCAA often explain their situations to those around them, which can be difficult sometimes. The participants get their information from the association, or they look it up on the internet. The information about DCAA that participants find often is insufficient, therefore did several participants indicate that they would like to use something in the direction of an information leaflet or something in this regard, like described earlier. The environment of DCAA family members outside of Katwijk often read up on DCAA, but not always. Despite the ignorance, the environment often tries to provide good support. The lack of information and awareness often leads to unpleasant situations and a discrepancy between DCAA family members who have a need of support and the level of support people surrounding them are able to provide.

‘At the time that I was on the waiting list, people were very understanding, but well, they still really needed the explanation about what exactly is the Katwijk disease, and in the beginning, I found that complicated, because occasionally I had to search for words. Well, eventually it became easier for me, because at a certain point I just had my story ready.’ – participant 2

7. ‘It’s just not my home and not my place’

For understanding of their situation, participants often turn to the DCAA patient association, based in Katwijk. The patient association facilitates peer support through their meetings. DCAA family members experience that these meetings help them talk through their experiences with DCAA with people who understand and go through the same thing. Participant 3 mentioned how much she appreciated that the association also provided space to talk about the emotional side of DCAA, precisely because she missed that in her own environment. Most participants like to be able to tell their story at the patient association and to help others by offering a listening ear. Especially the podcast ‘sick heritage’ from the patient association about the experiences of one of the board members with DCAA, is often mentioned as very valuable (18).

"That’s something I really miss—when I’m here (in Katwijk) with *name of friend with DCAA* and the others, you don’t even have to say anything, you just understand each other." – participant 7

However, the experiences of the patient association vary widely. Although almost every participant is very positive about the patient association in general, there are a number of obstacles for DCAA family members outside of Katwijk that stops them from visiting the association meetings.

Some indicate that there is no connection with Katwijk or that they even have a trauma related to Katwijk because it is linked to DCAA. Some also feel a social distance to the residents of Katwijk and the patient association. Some participants stated they were welcomed with open arms, whereas others called the association a ‘club’, where people from outside of Katwijk do not feel welcome, they feel like outsiders. Some reasons mentioned for this is that they do not know anybody in Katwijk, they do not have family contacts. Also mentioned is the fact that Katwijk is a relatively religious community, which makes some topics of conversation sensitive and makes that the people that do not live in Katwijk ask different questions.

"So yeah, we attended one of those meetings in Katwijk once, but honestly, we really felt like outsiders. Everyone knew each other—it’s a small village, and they all speak Kattuks (the local dialect). People were looking at us a bit like, ‘Who are these guys?’ – participant 3

For many people, Katwijk is geographically too far. Visiting the meetings of the patient association costs a lot of time and money and therefore can be very exhausting. Moreover, those who have already had a hemorrhage often can no longer drive a car. Some therefore pick and choose the events they attend. Most choose to visit expert meetings that cover

topics of interest to them and to skip peer support meetings. Participants mention that at the theme-nights, there also is space to speak with peers.

Participants also indicated that the group attending the association evenings is too diverse. The fact that there are so many older people prevent young people from attending, because they are in a different stage of life, with different questions and needs, which makes that younger DCAA family members sometimes experience that there is nobody attending that they have questions and experiences in common with. Participant 2 mentioned that she experiences survivor guilt. As a non-carrier, she feels burdened to visit the association, because there also are so many people that do carry the gene or are already patient.

Furthermore, attending a meeting of the patient association can be perceived as very confrontational. Whereas in their daily lives outside of Katwijk people hear almost nothing about DCAA, attending a meeting organized by the DCAA association means specifically talking about DCAA and its consequences.

> "I think it's quite confronting when you see that other people can't do certain things anymore—like not being able to speak properly. And if you've recovered yourself quite well, you kind of want to avoid seeing that, because you know it could happen to you too." – participant 1

To lower these barriers, some suggested organizing association meetings with a more limited group, such as only young people, or only gene carriers etc. furthermore, many feel the need for meeting evenings in their own neighborhood.

Discussion

Experiences

The experiences of DCAA family members outside of Katwijk are that there is a lack of understanding from the environment and the healthcare. This lack of understanding often has a negative impact on the DCAA family members, making them feel lonely in their situation. Help is sought from the patient association, but many are reluctant due to various obstacles, which include: There is no social bond with the residents of Katwijk, there is often a large geographical distance to Katwijk, the group of visitors is too varied, and a visit is too confrontational. Because of the rarity of DCAA and the lack of previous studies on the topic, there is not much literature to compare the results with. However, the emotional burden associated with a hereditary disease that the results of this study show is consistent with previous research (5,7,8).

Needs

From the experiences arise different needs, which were mentioned by the people themselves. People need good information provision and help in sharing this information with their environment and their care providers. Many also indicate a need for peer support meetings, but locally or in more specific groups.

Limitations

Being from Katwijk, there is a possibility the interviewer was biased. However, not being a DCAA family member or being close to one, it is considered not to have influenced the results of the study. There also could possibly be bias through the fact that all participants are already in some way connected to the patient association.

Recommendation

Based on the results found, I make four recommendations. The recommendations may be applicable to the entire healthcare system, but since it is unrealistic that that this would be taken up (given all the current deficiencies described in this study), the DCAA patient association seems like the right and ideal candidate to take on the following tasks:

- Firstly, a comprehensive information leaflet should be made available about DCAA. This should cover topics like for example heredity, testing, symptoms, etc. This way, every DCAA family member, regardless of disease status, can make use of it to inform those around them and their caregivers. Ideally, this flyer should be distributed to the entire healthcare system; the best way to make this possible is if the DCAA family members themselves will contribute, as they have the most contact with the relevant healthcare system components.
- Secondly, there should be looked for a way to organize local peer support meeting everywhere in the Netherlands, instead of only in Katwijk. Due to the rarity of DCAA, these meetings will not be crowded, therefore other people who are dealing with a hereditary and/or chronic disease may possibly also be invited, if it turns out that they have similar experiences.
- Thirdly, the peer support meeting should be adjusted in a more specific way, in which only specific participants are invited; there should be meetings for e.g. only (non) carriers, only youth, etc.
- Fourthly, at every meeting of the patient association in Katwijk, attention must be paid to an open and positive attitude toward new people from outside of Katwijk.

Conclusion

It can be concluded that experiences are not so much dependent on the place of residence, but that the status of the person and the character play a greater role. How one deals with (the possibility of) having DCAA varies for everyone, but what is common is that everyone has to deal with a lack of understanding from their immediate environment and the healthcare system. Experiences run from dealing with it easily to struggling with different emotions, and from understanding in healthcare to misunderstanding causing even misdiagnoses and inappropriate care. Very occasionally it happens that the environment knows something about DCAA, for example through other patients or because they searched for information on the internet, and therefore can be a bit more understanding and supportive than usually occurs.

The stories covered throughout this study point to the need for a solution to the various problems. This investigation showed, among others, that people find it unpleasant that they often have to construct a story to explain their situation to others; I hope that this research

can contribute to the possible help that can be offered to people with this problem. There is a need to further investigate how people can properly explain their situation to those around them to avoid misunderstanding and how health care providers can be informed, to prevent misdiagnosis and inappropriate care of DCAA family members, living outside of Katwijk. Furthermore, it should be investigated whether the results found in this study also apply to chronic diseases and whether the same solutions recommend here can be used as well.

Acknowledgments

My thanks go to both my supervisors, who took the time to help me during my internship. I would like to thank Sanne van Rhijn, patient advocate of the DCAA patient association, for which this research was done. Thanks for the enthusiastic help offered during every part of the internship.

Also, I would like to thank Kevin Pijpers who supervised me on behalf of the Vrije Universiteit Amsterdam and offered a lot of help while writing my report.

Finally, of course, I thank all ten interview participants for making time free for the interview and for the trust to have open conversations about this serious and personal subject.

References

1. Maat-Schieman M, Roos R, van Duinen S. *Hereditary cerebral hemorrhage with amyloidosis-Dutch type*. Neuropathology. 2005 Dec;25(4):288-97. doi: 10.1111/j.1440-1789.2005.00631.x. PMID: 16382777.)
2. Website HCHWA-D, association Katwijk Disease, link: https://www.hchwa-d.nl/nl/over-hchwa-d
3. Voigt S, De Kruijff PC, Koemans EA, et al, *Cerebral hemorraghes in patients with Dutch-type hereditary cerebral amyloid angiopathy*, Internaltional Journal of stroke 2022 vol.17(6) 637-644, DOI: 10.1177/1747493021 1043663).
4. Cerebrale Hereditaire Angiopathieën (CHA), LUMC, available from: https://www.lumc.nl/afdelingen/neurologie/ziektes-en-aandoeningen/cerebrale-hereditaire-angiopathieen-cha/#:~:text=Op%20de%20CHA%2Dpoli%20van,ziekte)%20en%20RVCL%2DS.
5. Koemans EA, Chhatwal JP, van Veluw SJ, et al, *Progression of cerebral amyloid angiopathy: a pathophysiological framework*. Lancet Neurol. 2023 Jul;22(7):632-642. doi: 10.1016/S1474-4422(23)00114-X. Epub 2023 May 23. PMID: 37236210.)
6. Kaushik K, Waslam NG, van der Zwet RGJ, et al. *Quality of life, depression and anxiety in cerebral amyloid angiopathy: A cross-sectional study*. Eur J Neurol. 2024 Dec;31(12):e16476. doi: 10.1111/ene.16476. Epub 2024 Sep 22. PMID: 39308134; PMCID: PMC11555154.
7. Bakker E, van Broeckhoven C, Haan J, et al., *DNA diagnosis for hereditary cerebral hemorrhage with amyloidosis (Dutch type). Am J Hum Genet. 1991 Sep;49(3):518-21*. PMID: 1679289; PMCID: PMC1683137)
8. Almqvist EW, Bloch M, Brinkman R, et al, *A worldwide assessment of the frequency of suicide, suicide attempts, or psychiatric hospitalization after predictive testing for Huntington disease*. Am J Hum Genet. 1999 May;64(5):1293-304. doi: 10.1086/302374. PMID: 10205260; PMCID: PMC1377865.
9. Ng KP, Richard-Devantoy S, Bertrand JA, et al *Dominantly Inherited Alzheimer Network. Suicidal ideation is common in autosomal dominant Alzheimer's disease at-risk persons*. Int J Geriatr Psychiatry. 2020 Jan;35(1):60-68. doi: 10.1002/gps.5215. Epub 2019 Oct 22. PMID: 31642105; PMCID: PMC7232741.
10. Atlas.ti, link: https://atlasti.com/?x-source=pmax&x-campaign=pmax-en&x-id=21759486841&x-term=pmax-01&utm_source=google&utm_medium=pmax&utm_campaign=21759486841&utm_term=&utm_

content=&utm_adgroup=&device=c&placement=&matchtype=&network=x&gad_source=1&gbraid=0AAAAApc8-YCT7aGanw4QAKGYLdSDgNEp1&gclid=Cj0KCQjwzrzABhD8ARIsANlSWNMgF3AttChqh346XVAdYMeaEGR_HsaQCm1odwSMzGlNon7RHqczfpkaAtYKEALw_wcB

11. AURORA, link: https://www.lumc.nl/patientenzorg/meedoen-aan-wetenschappelijk-onderzoek/onderzoek-naar-hchwa-d/aurora/
12. BATMAN, link: https://www.lumc.nl/patientenzorg/meedoen-aan-wetenschappelijk-onderzoek/batman/
13. CLEAR_BRAIN, link: https://www.lumc.nl/patientenzorg/meedoen-aan-wetenschappelijk-onderzoek/clear-brain/)
14. BIONIC, CAFÉ, SCALA, link: https://www.radboudumc.nl/lopende-onderzoeken/archief/bionic-cafe-scala
15. Family Tree Research: https://www.lumc.nl/patientenzorg/meedoen-aan-wetenschappelijk-onderzoek/onderzoek-naar-hchwa-d/stamboomonderzoek/#:~:text=Voor%20het%20stamboomonderzoek%20zoeken%20we,korte%20vragenlijst%20over%20gezondheid%20ingevuld
16. Braun, V., & Clarke, V. (2006). Using thematic analysis in psychology. *Qualitative Research in Psychology*, *3*(2), 77–101. https://doi.org/10.1191/1478088706qp063oa
17. Mortelmans, D., *Handboek kwalitatieve onderzoeksmethoden*, Acco Uitgeverij, 1 oktober 2020
18. Podcast sick heredity, link: https://www.nporadio1.nl/podcasts/zieke-erfenis

Reflection

The link of this study to the major in clinical sciences I have followed is that I have found that peoples experiences are mostly determined by the clinical implications surrounding the disease. This has broadened my view on clinical implications and made me aware of the impact they can have individuals' lives.

During this internship, I grew tremendously in many areas. I have become very independent, because I worked independently on the research for most of the time. Of course, I learned a lot about the disease that was the focus of the internship: DCAA. As a resident of the village where the disease originated and still plays a major role, I had some prior knowledge, but during the internship it turned out that I still knew incredibly little about the impact of the disease. In addition to this new knowledge, I gained many new skills and I strengthen those new skills and academic skills previously learned, by applying them in practice to a real-life setting, during all the different steps I went through during the internship.

I practiced doing literature research for information related to the subject at the beginning of the internship and made an interview guide and topic guide based on this. Interviewing was almost completely new to me. Previously, during my study I have practiced interviewing family members, and only once did I interview a stranger about her illness, but for this internship I did ten of those interviews with unknown people about their personal and often intense experiences with DCAA. It was a bit scary, every time again, but also extremely valuable and interesting to do. I strengthened my skills in the different types of coding of transcripts, which I had also only done once before and to a much lesser extent in my studies. It was quite a lot of work, but I was happy with the guidance of my VU supervisor.

Its feels good to deal with a real problem, rather than an educational example. I am happy to have been able to describe the stories of these ten people, and with that a larger group of people, and I hope that it may be a step in the right direction, so that their needs can be met. I found it difficult to do justice to all their personal stories but did my best to paint the best possible picture of the situation.

I also noticed that my visit to the participants of the interviews created awareness in some participants. The questions I asked made people think about their coping with their situation and they were pointed to the work of the patient association. I also handed out leaflets and magazines of the patient associations, this was appreciated by the participants.

Appendix I: Extensive information about DCAA

Name

The disease is also called Hereditary Cerebral Hemorrhage with Amyloidosis – Dutch type (HCHWA-D). The mutation most likely occurred in a person from Katwijk, a Dutch village, centuries ago. Because the villagers tend to stay in Katwijk, most mutation carriers live in Katwijk. As a result of this, DCAA is also informally known as 'Katwijk disease'.

Biomedical

Dutch-type Cerebral Amyloid Angiopathy (DCAA) is a rare hereditary disease caused by the abnormal build-up of amyloid-beta protein in the walls of cerebral blood vessels. This abnormality is caused by a mutation in the amyloid B-protein (AB) E22Q gene (1). DCAA is caused by an autosomal dominant gene, which means offspring of mutation carriers are 50% at risk (7).

Clinical

The process begins early in life but usually remains unnoticed for decades, as the first symptoms often appear between the age of 45 and 65, most often at 55 (3). One of the key features of DCAA is lobar intracerebral hemorrhage – bleeding in the outer parts of the brain – which can occur repeatedly and tends to become more severe over time. These hemorrhages can be fatal, or cause many different symptoms, leading to a diverse clinical picture for each individual patient. As the disease progresses, individuals may develop cognitive symptoms, such as memory problems, speech problems, physical failure, epilepsy, personality changes, psychiatric complaints, vision problems, overstimulation and difficulties with concentration or planning (4). Other symptoms may include transient focal neurological episodes (TFNEs), which are brief, stroke-like symptoms, as well as seizures. In the later stages, many patients develop dementia due to ongoing damage in the brain. The progression of DCAA is driven by worsening amyloid accumulation, which weakens blood vessel walls and interferes with the brain's ability to clear waste. This leads to inflammation, vessel rupture, and increasing neurological problems (5).

Treatment

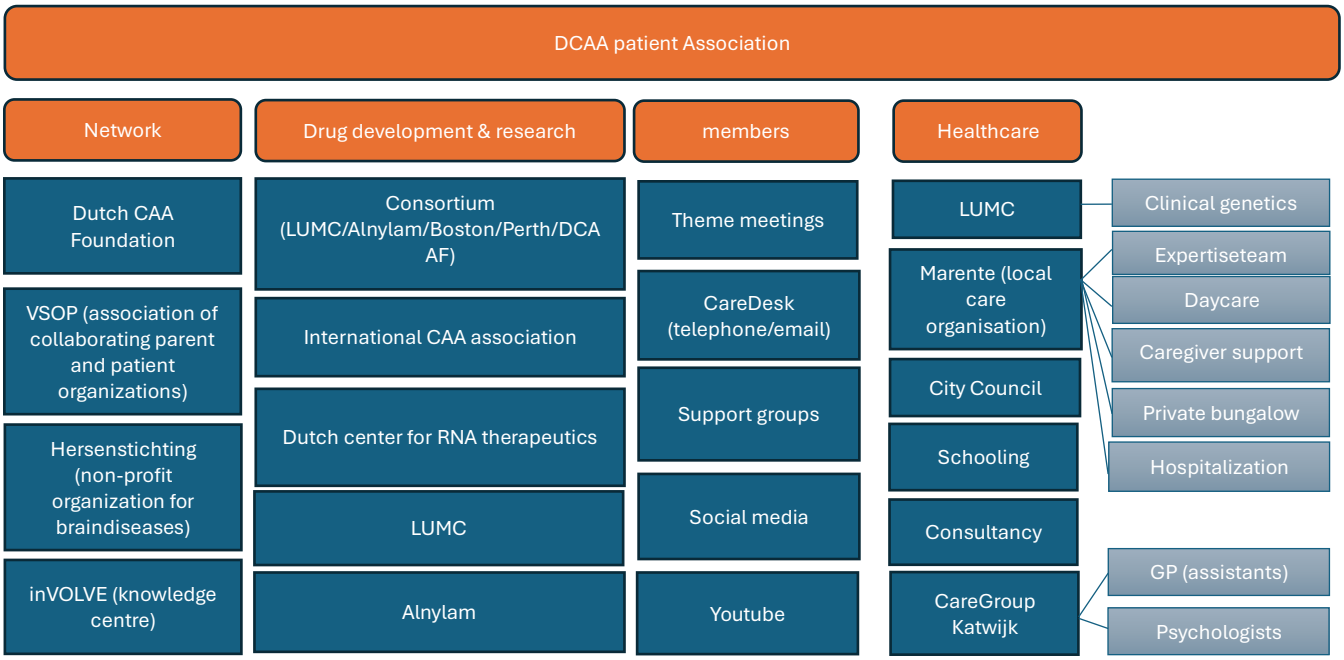
Currently, there is no treatment for the disease. Several studies investigate the disease and explore different possible ways to treat the disease, mostly done by Leiden University Medical Center (LUMC) (2). Some examples of current studies are:

- AURORA & AURORA Plus tracks disease markers (11)
- BATMAN tests minocycline for inflammation (12)
- Clear-Brain! studies sleep and vagus nerve stimulation for amyloid clearance (13)
- BIONIC, CAFÉ & SCALA identify biomarkers for early diagnosis (14)
- Family Tree Studies examines hereditary factors (15)

Preventive testing

Previous study showed that 14% of DCAA patients within the study population suffered from depression and anxiety (6). Because of the rarity of DCAA, not many studies have been done on the effects of DCAA on quality of life of DCAA family members, their need for support and their decision making, but studies from other hereditary disease can be used as an example. Previous studies on preventive testing in the hereditary diseases Huntington's Disease has shown that preventive testing could even lead to suicide, suicide attempts, or psychiatric hospilization (8). Also, with the hereditary autosomal dominant Alzheimer's Disease (ADAD), study has showed that depressive symptoms and suicidal ideation is common among ADAD at-risk individuals (9).

Appendix II: Stakeholder analysis



Appendix III: Topic Guide

- **Persoonlijke introductie**
  - Ontdekking
  - Ondersteuning/informatievoorziening (verbetering?)
  - Test
  - Omgang situatie in dagelijks leven
  - Familie
  - Patiënt/drager: werk (UWV keuring)
  - Toekomst

- **Wonen buiten Katwijk**
  - Ervaring
  - Obstakels
  - Reactie uit omgeving
  - Steun netwerk

- **Contact met zorgverleners**
  - Ervaring
  - Uitleg moeten geven (rol van vereniging)
  - Plaats van de zorgverlening
  - Hulp/ondersteuning nodig?

- **Vereniging**
  - Kennis (bijv. zorgloket)
  - Contact (en de behoefte daaraan)
  - Betrokken (inloop & thema-avonden/social media)
  - Drempel (rol vereniging)
  - Verbeteringen

  - Dingen niet aan bod gekomen?

Appendix IV: Example Questions

1. **Persoonlijke introductie**

Ontdekking

a. Kunt u me meer vertellen over of u zelf de Katwijkse ziekte heeft en wie in uw familie het heeft?
   - Heeft u uzelf laten testen op het gen? Waarom wel/niet?
   - Zo ja: hoe was het om de testuitslag te krijgen? Hoe gaat het nu? Hoe ging en gaat uw omgeving hiermee om?
   - Heeft u ervaringen met familieleden die ziek werden en zo ja, hoe was dat?
b. Wanneer bent u erachter gekomen dat het in uw familie zit en uzelf ook risico liep?
   - Hoe vond u de informatievoorziening op dat moment, kreeg u de informatie die u nodig had?
   - Waren er op dat moment bepaalde onderwerpen of situaties waarin u meer informatie of ondersteuning had willen ontvangen?
   - Heeft u ideeën over wie deze ondersteuning zou kunnen bieden?

Huidige situatie

c. Hoe gaat u op dit moment om met uw situatie?
   - Speelt de Katwijkse Ziekte een rol in uw dagelijks leven?
d. *Bij patiënt/gendrager:* Bent u (nog) aan het werk?
   - Ervaart u wel eens problemen op het werk door symptomen?
   - Zo ja, hoe gaat dat in relatie tot uw werkgever en eventueel het UWV? En als u een UWV keuring gehad hebt, bij welke vestiging was dit?

Familie

e. Op welke manier beïnvloed de ziekte uw familie en hoe gaat uw familie ermee om?
f. Waar maakt u zich op dit moment het meeste zorgen over met betrekking tot de Katwijkse ziekte?
g. Maakt u zich zorgen over de toekomst? Zo ja, waarover?

2. **Wonen buiten Katwijk**

Ervaring

a. Kunt u mij iets vertellen over hoe het is om de Katwijkse ziekte te hebben buiten Katwijk?
b. Denkt u dat uw ervaring erg verschilt met die van de Katwijkse families?
   - Zo ja, hoe wordt u ervaring beïnvloed doordat u niet in Katwijk woont?
   - Zijn er specifieke obstakels waar u tegenaan loopt, doordat u buiten Katwijk woont? (bijvoorbeeld doordat de ziekte in uw omgeving minder bekend is)

Steunnetwerk

c. Kunt u me iets vertellen over de eventuele steun die u krijgt vanuit uw omgeving?

d. Bent u wel eens ergens tegen aangelopen, waarbij u het gevoel had dat niemand u kon helpen?
   - Heeft u wel eens te maken gehad met onbegrip vanuit uw omgeving? Hoe gaat u daar mee om?
e. Heeft u op dit moment specifieke onderwerpen waar u graag ondersteuning of hulp bij zou willen krijgen?

3. Zorgverlening

Contact

a. Heeft u wel eens zorg nodig gehad? Heeft u contact gehad met zorgverleners? Zo ja, in welke discipline?
   - Hoe heeft u dit contact ervaren?
   - Bent u tegen dingen aangelopen?
   - Heeft u vervelende situaties meegemaakt? Hoe heeft u toen gehandeld? Wat zou u graag anders zien?
b. Heeft u zorgverleners wel eens uitleg moeten geven over uw situatie/de Katwijkse ziekte omdat zij hier geen weet van hadden?
   - Hoe heeft u dat aangepakt?
   - Kan de vereniging bijdragen aan een verbetering van het contact tussen u en de zorgverleners? Zo ja, hoe?

Plaats

c. Heeft u zorg bij u in de buurt ontvangen, of bijvoorbeeld in het LUMC in Leiden of de omgeving Katwijk?
   - Hoe ervaart u dat?
d. Kunt u in uw omgeving terecht voor uw behoeften, nu of in de toekomst? (denk bijvoorbeeld aan dagbesteding of voorzieningen die nodig zijn in huis?

4. De vereniging

Betrokken

a. Kent u de Vereniging voor de Katwijkse Ziekte?
   - Hoe is uw contact met de vereniging?
   - Wat vindt u van de vereniging?
b. In hoeverre bent u betrokken bij (activiteiten van) de vereniging?
   - Bezoekt u inloop- en of thema-avonden? Zo niet, wat is daar de reden voor?
   - Ervaart u een drempel om deel te nemen? Kan de Vereniging daarin iets voor u betekenen?
   - Bezoekt u wel eens de website of de social media van de Vereniging? Waarom? Kan de Vereniging iets doen om deze voorzieningen beter aan te laten sluiten op uw behoeften?
c. Heeft u behoefte aan contact met andere mensen uit families met de Katwijkse ziekte?
   - Het lotgenoten contact vindt plaats in Katwijk, neemt u daaraan deel?
   - Heeft u misschien een andere manier gevonden voor lotgenotencontact?
   - Hoe zou volgens u de Vereniging hieraan bij kunnen dragen?
d. Bent u op de hoogte van wat het Zorgloket van de Verenging kan betekenen?

Verbeteringen

e. Heeft zelf specifieke ideeën over hoe de Vereniging beter kan inspelen op uw behoeften en die van andere families buiten Katwijk?
f. Zou u zelf iets willen of kunnen bijdragen aan de vereniging?
   - Zo ja, op welke manier?

g. Zijn er nog dingen die u wilt delen die nog niet aan bod zijn gekomen tijdens dit interview?

Appendix V: Informed Consent

Onderzoeksleider: Hugo Haasnoot, student Gezondheid en Leven aan de Vrije Universiteit Amsterdam, in samenwerking met Vereniging Katwijkse Ziekte.

Doel van het onderzoek:
De Vereniging Katwijkse Ziekte wil er zijn ter ondersteuning van de mensen die te maken hebben met de Katwijkse Ziekte. Dat geldt voor zowel patiënten, als mensen die weten dat ze gendrager zijn, maar (nog) geen patiënt, risicodragers (mensen die hun genetische status niet weten), niet-dragers (mensen die weten dat ze geen gendrager zijn), partners, mantelzorgers en andere familieleden. Zij bieden onder andere lotgenotencontact en informatie-avonden en zetten zich in voor onderzoek en goede zorg.

Doordat de grootste groep van deze mensen in Katwijk zelf en omgeving wonen, heeft de Vereniging binnen de gemeente en omringende gemeenten het grootste bereik. Al hun activiteiten vinden in Katwijk plaats. Toch weet de vereniging dat er buiten Katwijk ook mensen wonen die te maken hebben met de Katwijkse Ziekte, maar dus buiten het bereik van de vereniging liggen. De vereniging wil ook hen graag helpen.

Wat houdt deelname in?

- Een interview van ongeveer 45 minuten.
- Vragen over uw situatie, ervaringen buiten Katwijk, contact met de zorgverlening en over de vereniging.
- Het gesprek wordt opgenomen en opgeslagen op een beveiligde harde schijf.
- Uw naam zal niet worden gebruikt in het verslag; uw deelname blijft volledig anoniem.

Vrijwillige deelname en privacy:

- Deelname is volledig vrijwillig.
- U kunt op elk moment besluiten een vraag niet te beantwoorden.
- U kunt het interview op elk gewenst moment pauzeren of stoppen.
- De verzamelde gegevens worden uitsluitend gebruikt voor dit onderzoek en worden vertrouwelijk behandeld.

Verklaring van toestemming: Ik heb de informatie in dit formulier gelezen en begrepen. Ik begrijp dat mijn deelname vrijwillig is en dat ik op elk moment kan stoppen zonder consequenties. Ik ga akkoord met het opnemen van het interview en de verwerking van de gegevens zoals hierboven beschreven.

Naam deelnemer: _____

Handtekening deelnemer: _____

Datum: _____-_____-_____

Naam onderzoeker: Hugo Haasnoot

Handtekening onderzoeker: _____

Datum: _____-_____-_____

HCHWA-D
VERENIGING KATWIJKSE ZIEKTE

VU VRIJE UNIVERSITEIT AMSTERDAM

Use of generative AI

I did not use generative AI.