INTRODUCTION

Huntington’s disease (HD) is a rare, severe, and complex genetic disease characterized by a triad of motor, cognitive, and psychiatric symptoms that progress over time. Features of HD include the onset of involuntary movements, impairment of voluntary movements, cognitive deterioration, as well as psychiatric and/or psychological symptoms, such as depression, anxiety, disinhibition, or apathy (Bates et al., 2015; Heiberg, 2008; McColgan & Tabrizi, 2018).

HD is an autosomal dominant disease caused by a heterozygous expansion of a CAG trinucleotide repeat in the HTT gene (MacDonald et al., 1993). Thirty-six or more CAG repeats are considered to be disease-causing. In general, 36–39 repeats are classified as reduced-penetrance HD-causing alleles, and 40 or more
CAG repeats are considered full-penetrance HD-causing alleles; 27–35 CAG repeats are considered intermediate alleles (McColgan & Tabrizi, 2018). Individuals are often diagnosed at middle adulthood (30–50 years old), the stage when a person who carries a CAG-expanded HTT allele develops unequivocal motor symptoms (Ross et al., 2014).

Predictive genetic testing is available to all adult individuals at risk of inheriting the mutation for HD. The Norwegian Biotechnology Act mandates the inclusion of genetic counseling as component of predictive genetic test. A predictive test for genetic disorders without prevention or treatment options can only be performed on individuals of 16 years or older. However, it has been recommended that those who are at least 18 years old may undergo the test (MacLeod et al., 2013). In Norway, the HD test protocol applied in practice includes a pre-test genetic counseling, an evaluation by a healthcare professional with an expertise in psychology, a second genetic consultation with blood sampling, and a genetic consultation on the result, as recommended by the European Huntington’s Disease Network (MacLeod et al., 2013). A follow-up consultation is automatically scheduled for those who tested positive for the HD mutation. Genetic consultation services are provided by a clinical geneticist and a genetic counselor, who thus play a key role in the predictive genetic testing process.

Studies indicate that more than 80% of those at risk for HD choose not to undergo a predictive genetic test, suggesting that arriving at a decision whether or not to get tested is quite complex (Baig et al., 2016). People's motivation to undergo predictive testing may include the need to eliminate uncertainty, to plan their life and career, and to determine the risk of their children (Ibiler et al., 2017). Individuals who decide to undergo predictive genetic testing may experience a range of psychologically challenging dilemmas prior to the test and after receiving the test result. Research shows that genetic testing indeed may have an adverse psychological impact on those at risk for HD (Crozier, Robertson, & Dale, 2015). Factors that may aggravate psychological distress among individuals who tested positive for the HD mutation include their inability to predict the onset of disease symptoms or the progression of their disease, as well as the fact that a cure for their disease is currently non-existent. Also, the disease can be stigmatizing for the affected individual and/or for his/her family (Crozier et al., 2015).

Many individuals have experienced stress and anxiety when they first learn that they carry the HD mutation (Gargiulo et al., 2009); such a test result has been shown to negatively impact one’s life decisions, particularly their long-term plans (Broadstock, Michie, & Marteau, 2000; Gong, Fanos, Korty, Siskind, & Hanson-Kahn, 2016). Interestingly, some individuals describe positive changes after learning that they have the HD mutation; for instance, they become more purposeful toward their future, and therefore, they reach milestones early, and they make active choices for their future in terms of education, career, romantic relationships, and family planning (Gong et al., 2016).

Ambivalent or mixed emotional reactions have been reported both by individuals who tested positive and by those who tested negative for HD mutation. Studies show that the mental health and quality of life of those who have and those who do not have the mutation do not significantly differ (Crozier et al., 2015; Licklederer, Wolff, & Barth, 2008). Clinical experience and some studies suggest that prior to getting tested, some people may have spent years believing and living as if they had the mutation; therefore, they may experience a struggle adapting to a future not having the disease (Mand, Gillam, Duncan, & Delatycki, 2013). Accepting the choices they made based on the perceived certainty of developing HD in the future may therefore constitute an emotional burden. Some people may also experience ambivalence over their own status as being not at risk for developing HD; they are relieved and happy that they cannot pass the mutation on but worried at the same time knowing that other family members may be at risk or have developed HD (Codori & Brandt, 1994).

1.1 Purpose of the study

Indications of complex psychological challenges affecting individuals at risk for HD both prior to and after genetic testing suggest the need for in-depth knowledge about the experiences of these individuals before they decide to get tested; also, knowledge about their reaction to the test result is warranted. Most studies exploring this subject have investigated participants receiving genetic counseling services, that is, those who have already undergone genetic testing; by contrast, the experiences and personal choices of those who have not yet received genetic counseling are less investigated. Consequently, the reasons affecting one’s decision to either get tested or not for HD mutation and the impact of the test results on one’s life remain largely unknown. There is a lack of knowledge about how people at risk for HD perceive and/or consider undergoing genetic testing and also which psychological and socio-ecological factors affect their perception of this process. Also, there is a lack of qualitative research investigating the psychological and emotional impacts of the test results in individuals with the HD mutation and those who do not have the mutation. Therefore, this study aimed to explore the experiences of the predictive genetic testing decision-making process in a diverse population of individuals who are or have been at risk of HD.

2 METHOD

2.1 Design

Data were extracted from a primary study involving a larger dataset collected through semi-structured interviews; the primary study is a component of a concurring project that investigates childhood experiences in families affected by HD (Kjøelsas, Tillerås, & Feragen, 2020). Given the focus of the original study, inheritance and predictive genetic testing were not covered in the interview guide. However, all participants shared substantial thoughts on these topics during the interviews; therefore, the interviewers asked them to elaborate their thoughts on these topics. Considering the importance of these topics to the participants, we decided to perform a
secondary data analysis (Thorne, 1994). Data were extracted from the original dataset and then qualitatively analyzed separately.

### 2.2 Study sample and recruitment

By using the convenience and snowball sampling methods, we recruited participants through the National Association for Huntington’s Disease in Norway, through the counselors at a National Resource Center for HD in Norway, through the departments of medical genetics in Norway, and through the Internet (websites and social media). To be included, prospective participants should be a member of a family where one of the parents had/has HD (original study), speak Norwegian, and be at least 12 years old. Individuals with communication skills or cognitive function indicative of difficulties in providing informed consent for participating in in-depth interviews were not eligible to participate in the study. Ethical approval for this study was obtained from the Regional Committee for Medical Research Ethics (Health Region South-East; Reference No. 2017/1613).

An information sheet about the study and a consent form were sent by post to those who expressed their interest to participate. The information sheet provided details about what their participation in the study would entail, as well as key ethical information, such as confidentiality and their right to withdraw. A total of 42 individuals consented to participate. However, six individuals were eventually excluded as they could not be reached for a discussion on matters regarding interview schedule, leaving us with a sample consisting of 36 participants. Moreover, three individuals were tested prenatally and were also excluded, further reducing the sample size to 33 individuals. Of these, 19 participants had undergone testing, had shared post-test narratives of the test process, and had described their reactions to the test result and to the follow-up they had received. The demographic characteristics of the participants are presented in Table 1.

### 2.3 Data collection

An interview guide based on relevant literature and based on feedback coming from clinical experts was created for the primary study. The participants were asked open-ended questions and were prompted to provide more details whenever appropriate. The broad interview topics were as follows: childhood narrative and family situation, relationship to parents, level of understanding or lack of understanding of HD as a child, openness about the disease, and support experiences (interview guide in Norwegian available in Data S1). These interview topics and sample items from the interview guide in English are presented in Table 2. The interviews took place (12/17-09/18) and were conducted face to face or over the telephone by different interviewers who are all qualified and who have been trained in counseling and/or in qualitative methods. On average, the interviews lasted approximately 60 min (range: 27–90 min).

### 2.4 Data analysis

The interviews were recorded and transcribed verbatim. In the primary study, the interviews were conducted by several researchers, including a licensed clinical psychologist and a specialist in clinical genetics; in addition, three final-year students in clinical psychology and one master in psychology were involved, all of whom were under the supervision of the clinical psychologist. The interviewers were not familiar with the participants prior to the study. Thematic analysis was performed according to the method described by Braun and Clarke (2006), as follows: (a) becoming familiar with the data, (b) identifying interesting features of the data, (c) searching for themes, (d) reviewing the themes, (e) defining and naming the themes, and (f) producing the report. The first author coded the material in tandem with authors KBF, SHK, and CvdL. The codes were organized under thematic headings. Analysis was seen as a recursive process, and detailed notes were written throughout. Themes were subsequently chosen for their prevalence and/or their apparent importance in relation to the research questions. Thematic analysis was performed according to the method described by Braun and Clarke (2006), as follows: (a) becoming familiar with the data, (b) identifying interesting features of the data, (c) searching for themes, (d) reviewing the themes, (e) defining and naming the themes, and (f) producing the report. The first author coded the material in tandem with authors KBF, SHK, and CvdL until a consensus was reached. Themes and data extracts were reviewed by referring to the transcripts and to the research questions, and quotes that were representative of the themes were selected. Illustrative quotes were translated into English, and the participants were given pseudonyms.

### 3 RESULTS

Three main themes were identified, as follows: ‘a life in preparation for disease’ (Theme 1), ‘factors influencing the test decision’ (Theme 2), and ‘the test result’ (Theme 3). These themes and their corresponding subthemes are presented in Table 3.

#### 3.1 Theme 1: A life in preparation for disease

Regardless of the participants’ decision of whether or not to undergo a genetic test to determine their mutation status and regardless of whether they tested positive or negative for the mutation, a substantial number of the participants said that they had lived in an anticipation of having inherited the disease. Being uncertain of whether they had the HD mutation, the participants consistently
describe their experience of having demanding thoughts, emotions, and perceptions, and they organized their life around the possibility or certainty of living with HD in the future.

### 3.1.1 Thoughts, emotions, and perceptions prior to testing

Prior to deciding whether to get tested, several participants mentioned their struggle with negative thoughts arising from the possibility and/or from an anticipation of having inherited the disease. Many admitted that they constantly think that they might develop the same symptoms as their parent, impacting their thoughts and emotions.

‘All the thoughts of possibly inheriting the disease were exhausting. I feel it probably guided my decisions. I mean, I went to school, studied, that kind of thing, but everything was sort of done with a lid on...’

*(Mathilde, adult female, mutation positive)*

Convinced that they had inherited the disease, the participants articulated how they searched for symptoms in their everyday life. For some, this habit started as early as in primary school, and it was initiated by their inner anticipations, perceptions, and/or attributions.

‘I was completely sure I had the disease. I was convinced I had all the symptoms—the bad concentration, difficulties swallowing, the movements—I had it all. (...)

*(Susanne, adult female, mutation negative)*

Others shared that their search for symptoms had been triggered by other people’s doubts, comments, or questions. Again, the participants described a variety of negative thoughts and emotions related to the possibility of having inherited the disease, ranging from minor or passing thoughts in everyday life to feelings of overwhelming negativity and hopelessness. Additionally, society’s lack of knowledge about HD could exacerbate the feelings of loneliness and isolation.

‘Not a lot of people know about the disease, only my closest family (...), it isolated me in a way. And it scares me a lot that I can get it too’.

*(Trevor, teenager, male, unknown mutation status)*

For some participants, anticipating that they have inherited the disease created the feeling of powerlessness. Anxiety, depression, fear, and hopelessness were also apparent in their descriptions of their experience of uncertainty arising from the possibility of living with the disease in the future. The brutality of such thoughts was vividly described by one of the participants, who found a potentially positive test result to be similar to a death sentence.
It’s like there’s a loop around your neck constantly; you don’t know if it’s there or not, and that’s what makes it so much harder. I have a 50% death sentence. (…) I feel indifferent, very numb. I’m not sad, but I am not happy either; not mad, I’m just very flat. It’s like I have no pulse; I don’t feel anything. I feel more like a robot than a human being’.  

(Jake, teenager, unknown mutation status)

While the anticipation of inheriting the disease instigated a negative outlook in the life for many, some participants had chosen to focus on making the most of their lives in the present.

’I guess we just chose to leave it be, and live our lives’.  

(Elinor, adult female, unknown mutation status)

3.1.2 | Organization of life prior to testing

Despite not knowing their genetic status, the participants have made life decisions that were influenced by their anticipation of having inherited HD. These life decisions included practical, personal, and/or emotional decisions. Many had focused only on goals they believed they could achieve, and they had restricted their choices in terms of education, romantic relationships, and having children. Moreover, they experienced a lack of motivation to achieve their goals, as well as hopelessness and fear of the test result; both experiences became a hindrance in building their own family.

’It’s scary; as if there is no hope (…). I don’t know how to explain it. I’m too afraid to get married and of having children. I just can’t bring myself to imagine having these things before I get a result and know what my future will be like. But I am too afraid to get tested’.  

(Frida, young adult female, unknown mutation status)

Some participants became hesitant to enter into romantic relationships because they do not know how their future would be like and because they have fear of being rejected due to their risk status.

’Everyone else is talking about a future, a house, a car, an education. I can’t have any of that. (…) If I meet someone I like, I would have to tell him about this before we start dating’.  

(Annie, female teenager, unknown mutation status)

Due to the possibility of having inherited HD, the participants had restrictive beliefs in terms of the milestones they could reach in their academic and/or professional lives, which intensified their lack of hope for their own future.

’I thought that I would probably get sick too, so what’s the point in spending a lot of time getting an education. I had to invest in something that gave me a secure job, with an easy education. I thought that I would probably not get that old’.  

(Andrea, adult female, mutation negative)
The irreversible nature of the choices described by the participants can be illustrated by the story of one of the participants, who later found out that she had not inherited the disease. She had chosen to have an abortion based on the fear that her child might be at risk for HD, a decision she had to live with after finding out that she does not have the mutation.

'We were pregnant with a child that we had to terminate, because we did not know if he could get sick. But of course, he wouldn't have been...'

(Mia, adult female, mutation negative)

Although many felt restricted by their genetic risk, some participants described how they managed to organize their adult lives in order to achieve certain milestones, such as having children, before the onset of the disease.

'I thought that if I do get sick, I wanted to get married and have children early. So that my kids would be grown-up by the time the disease started to appear. I was very sure of that'.

(Olivia, adult female, mutation negative)

### 3.2 Theme 2: Factors influencing the test decision

Apart from describing how their lives were influenced and organized in anticipation of having inherited the disease, the participants also talked about how they arrived at a decision whether or not to get tested for the mutation. The decision-making process was often experienced as complex, and it was influenced by several factors.

### 3.2.1 Personal and social influences prior to testing

The participants talked about how other people's presumptions and opinions on whether they should get tested had annoyed them, had been difficult to manage, and had influenced their decisions. These people could be their friends, family members, potential romantic partners, and/or healthcare professionals. In some cases, the same questions and opinions come from several people.

'Because people do ask [if you have been tested or not]. Say that ten people ask you. That does something to you'.

(Vilde, adult female, unknown mutation status)

Some participants shared that their families had social expectations regarding genetic testing, and they felt that these expectations could influence their own decision making. They also talked about the existing predeterminations or 'family norms' in relation to taking the test, and these norms served as an unwritten but influential guide that caused them to arrive to a decision that their families expected them to make.

'There is a pledge in the family. No one should get tested. And if you do, you can't say anything to anyone, because they believe that it will increase the probability of others getting ill too'.

(Anders, young adult male, mutation negative)

One participant recalled how a healthcare professional had influenced her decision by helping her recognize that she might not be ready to get tested at that point in her life and thus advised her to wait.

'I am so happy that I met one of the counselors when I was young. It was not a good time for me to get tested. If I had received a bad result at that time, I don't know what I would have done. So I waited until I was older. (...) That conversation was worth a lot to me'.

(Sara, adult female, mutation negative)

### 3.2.2 Practical influences prior to testing

In addition to norms and opinions conveyed in social interactions, the participants’ decision of whether or not to get tested was influenced by practical concerns and arrangements needed to make future plans. Hence, the decision was influenced by one’s personal goals or marital goals more than by other people’s opinion of the disease. Nevertheless, the decision-making process seemed intertwined with the norms and opinions conveyed by others.

'All of a sudden I had a problem, because when the question of testing first came up, I was so sure that I did not want to know. Then I realized little by little that if I wanted to have kids, I would have to know'.

(Louise, young adult female, mutation negative)

More specifically, several participants felt the pressure or the expectation to get tested so that their potential romantic partner would know the former's risk status, which is crucial when making decisions regarding having children.

'I ended up in a situation where I had one child and wanted other children, and we had to wait with the second one until I got tested, since my husband wanted to choose to get a healthy child if we could'.

(Milla, adult female, mutation negative)

One participant was certain that one of her parents had HD. However, this parent did not recognize his/her need for medical attention, delaying the initiation of care. To confirm the occurrence of HD
in her parent, the participant, who anticipated having a positive risk status, got herself tested.

'I got tested to know whether my [parent] had the disease. (...) If I had the disease, then my [parent] had to have it'.

(Laura, adult female, mutation positive)

3.3 | Theme 3: Test result

The participants who got tested and had received their test results described their experience of going through a range of psychological reactions and outcomes. Most of them talked about how they felt, how they perceived the consequences of the test result, how they will share and whether to share their test results with others, and how the follow-up they had received from healthcare services went.

3.3.1 | Handling the knowledge of disease

Most of the participants who had received a positive result (mutation positive) shared this outcome to be emotionally and psychologically difficult to handle. The test result had affected various aspects of their lives, and for some, learning that they had the mutation for HD led to powerful negative thoughts. One participant shared that her negative emotions were so overwhelming that it led to suicidal ideation.

'I considered suicide several times; I was devastated. There was so much going on at once'.

(Klara, young adult female, mutation positive)

Another participant expressed that she felt concerned for her children, who were now at risk for HD. She shared feelings of fear and sadness, and admitted that she was even searching for symptoms in her children's behavior. She also expressed a wish for her son to get tested, with a hope for a negative result so that she would not have to worry about him anymore.

'My son keeps saying that he will get tested. (...) And I see the changes in him, and it hurts, because I'm thinking, if I can get the answer that he does not have the mutation, it will calm me down. (...) I don't worry about myself. I worry more about him, because he's so young'.

(Laura, adult female, mutation positive)

Despite the emotional difficulties of receiving a positive test result, the participants said that having taken the test was crucial and was right for them. They experienced relief as taking the test has put an end to years of believing that they have the HD mutation.

'I was pretty convinced that I would get a bad result. I guess there were a few days and months [after she got the response] that were worse than others. But at the same time, I have kept it at a distance for a few years, either I had it or I didn't. I had that feeling for so many years, so when I finally had the answer, I pretty much felt that it was good just to be done with it'.

(Jenny, adult female, mutation positive)

One participant shared how she, after a while, became more positive and conscious about her priorities and about the things that mattered in her life; she recognized that this experience had also influenced others positively. Some participants felt being closer to their family because of the test result; for instance, they now tend to understand their affected parent’s experience better.

'I've got a bad result; it didn't matter to me because I felt that [my relationship] with mom fell into place, and that meant more. (...) I understood my mother more, and it changed my relationship with Mom'.

(Laura, adult female, mutation positive)

The participants who have the mutation had different opinions about whether they believed in the importance of sharing their test results or not. They also varied with regard to whom and how they would share this information with. Trust appeared to be an important factor when disclosing their mutation status.

'I try to be open about it. I am that type of person that can talk about things with people I know I can trust. And that feels good, but it used to be more difficult'.

(Mathilde, adult female, mutation positive)

One participant was given advice by healthcare professionals to keep her test result a secret, a piece of advice that contradicted her own feelings and her experiences of HD being perceived as normality in her family.

'When we had taken the test (...), [the genetic counselors] pointed out that we should keep quiet about this. Not to tell anyone, that we didn’t have to say it if I was going to apply for a job, my boss didn’t have to know, and the insurance companies did not need to know either if I wanted life insurance; I had no obligation to inform. I felt that I grew up thinking it was normal, with my uncle and my mom, but when it came to me, I had to keep quiet'.

(Laura, adult, mutation positive)
Most of the participants who tested positive were pleased with the post-result follow-up consultations conducted by the providers of healthcare and support services.

‘I have been very happy with the follow-up. I went to a couple of sessions with a psychologist, and that was beneficial, being able to talk to someone other than those at home’.

(Jenny, adult female, mutation positive)

A few participants also said that they coped with the difficult news by hoping for the development of a cure for their disease in the near future.

‘When the disease develops, it might take some time before I become really sick, and by that time, there might be, probably, a cure, before I become really sick’.

(Jonathan, young adult male, mutation positive)

Despite being pleased with the follow-up consultations with the healthcare personnel after receiving the test result, one participant shared that no options for follow-up consultations were offered to the participants’ relatives, partner, or immediate family.

‘There should have been a more comprehensive setup for us together where she [his wife] also had the chance to talk about her issues (...). I also think that more long-term follow-up is needed because your partner’s needs may change when you get things at a distance’.

(Henrik, adult male, mutation positive)

3.3.2 | Handling the absence of the disease

The participants who received the result that they did not have the mutation also felt that the absence of the disease affected various aspects of their lives. Most of them experienced joy and relief, and they and/or their children felt as though they have been spared.

‘I was so incredibly happy that my kids weren’t going to experience the same insecurity that I did. That was almost the most important thing’.

(Susanne, adult female, mutation negative)

However, many participants felt ambivalent about the test result; that is, their positive emotions were frequently contrasted by sadness, by disbelief, or by ‘survivor’s guilt’. Feelings of guilt toward siblings seemed to be particularly prevalent. Most of these participants expressed being surprised by their own conflicting reactions.

‘When I received the good news, I felt guilty for being free while my sister was not. (...) We had the same probability; (...) I would gladly have taken the disease, so that my sister wouldn’t have to’.

(Andrea, adult female, mutation negative)

A few participants said that they had been concerned about whether the test results were valid and that they found themselves still searching for symptoms in spite of mutation-negative test results.

‘Even though you get a good result... You still see things... I always thought I walked strangely, and I always thought it was because of Huntington. And I have a temper, I can get furious. Am I sick after all? A little bit clumsy, right? Dropping a few things. (...) you think ‘oh my god, maybe they got the wrong test.’ You can’t be sure. You are never truly free’.

(Sara, adult female, mutation negative)

Several participants who did not have the HD mutation had wished for additional follow-up consultations to help them deal with the complexity of their reactions after receiving their test results.

‘It took a long time for me to understand the test result, but I did eventually. (...). I have really wanted to talk to someone’.

(Julie, adult female, mutation negative)

One participant who had received a follow-up had initiated this process herself; that is, she took the initiative of asking for a follow-up consultation. However, she felt that it was difficult having to be the one asking for support.

‘I felt that I had to be the one to reach out to get further follow-up, and then it’s just not that easy’.

(Louise, young adult female, mutation negative)

The participants also shared their need for social support after receiving their test result; such a need could not be addressed by the follow-up consultations offered by the providers of healthcare services. One participant explicitly mentioned that peer support is important given that only a few people without a direct experience with HD could understand the magnitude and complexity of her situation.

‘I wanted to talk to someone that was going through the same thing as I was. Someone who is not going to become sick, but who has a mother or father who is, and who has a sibling who might be affected too. I missed that’.

(Olivia, adult female, mutation negative)

Some participants said that they did not want to share their test result since they felt no one would really understand its personal importance and impact.
When the good results arrived, I was so sure I would be happy, wanting to share it with everyone. But I had a completely different reaction (…), and I didn’t really want to share it with anyone. I believe I had a feeling that no one would understand how important it was, how big it felt.

(Louise, young adult female, mutation negative)

Another complicated aspect related to disclosing their own genetic status was the fear of revealing other family members’ risk status to others.

‘It’s a genetic disease. (…) I have a brother, and if I tell other people that I’ve been tested, then they know that ‘there’s a 50 % chance of him having it’. That’s difficult’.

(Anders, young adult male, mutation negative)

4 | DISCUSSION

This qualitative study shows that predictive testing for HD is a highly complex process that may impact the lives of those at risk long before they decide whether or not to get tested. The participants described that they lived as if they were preparing for the onset of the disease, even prior to deciding to take the test. The decision of whether to get tested or not seems to be influenced by social, practical, and personal factors. Regardless of the outcome, test results may impact the lives of people long after results have been received. Dealing with the test result was difficult regardless of the mutation status. The participants who had received a negative mutation status expressed a need for more follow-up than they had received.

4.1 | Organizing one’s life and preparing for the worst

The participants lived their lives as if they had the mutation before they decided to undergo predictive testing. Preparing for the worst may be a prevention-focused coping mechanism (Hazlett, Molden, & Sackett, 2011; McAllister, 2003), and pessimism has been shown to reduce some people’s anxiety levels (Norem & Illingworth, 1993). Further, people’s health expectations and/or how they believe a disease will progress have been found to mediate both the actions taken and the measurable outcomes of health and disease (Williams & Bond, 2002). In our study, the participants consistently reported negative beliefs about the possibility of having inherited HD; they also planned their future taking into consideration the limitations posed by their disease, and they even reported symptoms of HD despite having no idea of their mutation status.

Placebo and nocebo effects may help us understand the possible impact of negative assumptions held by the participants about living with HD in the future. Placebo effect can be defined as the positive effect produced by a substance without inert treatment, and nocebo effect can be defined as the negative effect produced by the negative beliefs and expectations of individuals (Colloca & Miller, 2011; Wolman, 1989). Placebo and nocebo effects are a widely recognized phenomenon in clinical studies, and they have been found to influence cognition in many medical conditions (including motor disorders) and perception of pain (Benedetti, Lanotte, Lopiano, & Colloca, 2007). Expectancies also appear to play a substantial role in the actions taken to ensure health and to address a disease. For instance, positive expectancies have been found to decrease a range of physical symptoms for both chronically ill and healthy individuals (Andersson, 1996; Beckham, Rice, Talton, Helms, & Young, 1994).

However, negative expectancies may have an unfavorable impact on one’s health status (Reed, Kemeny, Taylor, & Visscher, 1999). The present study did not investigate the long-term emotional, psychological, or somatic effect of the certainty of having inherited HD. Nevertheless, the participants shared powerful stories about the choices they had made based on this certainty, which could easily produce detrimental nocebo effects. Their descriptions of the symptoms of HD years before knowing they were found mutation-negative also illustrate the nocebo effect in this population. The present findings therefore illustrate the possible negative psychological impact that the belief of inheriting HD may have on individuals at risk, even in the absence of a confirmed diagnosis. Clinical geneticists and genetic counselors involved in the follow-up of individuals at risk for HD should explore the magnitude of this potential psychological impact and help address its consequences to prevent its possible damaging impact on the individuals at risk.

4.2 | Socio-ecological framework

An important finding in this study is that the participants’ experiences and decision were influenced by several factors related to their upbringing, including family and social relations, and by their interactions with healthcare professionals. Given the various relationships and situations that influence a participant’s cognition of the disease and the testing for HD, a socio-ecological framework (Bronfenbrenner, 1979) is an appropriate tool for the analysis and application of the findings. Generally, this model proposes that human behavior is the result of reciprocal interactions between the individual, the family, the social surroundings, and other everyday life contexts. This model may help us to understand the full range of factors that will determine a person’s decision-making processes, behaviors, and reactions. Similarly, in our study, factors expected to influence the participants’ perception of being directly or indirectly affected by HD ranged from personal variables, such as expectancy and self-efficacy, to the influence of family and personal relationships and to more distal influences, such as advice from healthcare practitioners. According to this framework, the immediate environment exerts the most significant impact on future decision-making processes and behavior.
This phenomenon was true for our participants. Their beliefs about the possibility of having inherited the HD mutation and their decision of whether to get tested or not seemed to be strongly influenced by their experiences with HD in their family environment, by the family 'mathematics' (the fear that if one sibling is found to be mutation-negative, the next one has more chances to be mutation-positive), by the norms upheld within a family about pre-symptomatic genetic testing, and by their experiences of how symptoms of HD had affected their family. Several other factors within the family environment could also have a strong impact on one's decision to get tested and on one's reactions to the test result. A qualitative study has found that individuals who grew up with a parent with HD had reported more adverse experiences, such as parental dysfunction, loss, and trauma, compared with individuals whose parent has another hereditary condition (breast/ovarian cancer (BRCA1/2)) or compared with controls with a negative family history of HD or BRCA1/2 (Van der Meer, van Duijn, Wolterbeek, & Tibben, 2012). These adverse experiences will in turn have an impact on a person's psychological makeup, such as his/her style of attachment or emotional regulation, which may influence his/her level of post-test distress regardless of the test result received (van der Meer, van Duijn, Giltay, & Tibben, 2015).

Individuals affected by HD through their immediate family environment could therefore be predisposed for a greater risk for and sensitivity to stress, which could lead to more adverse reactions to a challenging test process, or instigate the development of depression or anxiety, as a reaction to test results, regardless of mutation status (Heim, Shugart, Craighead, & Nemeroff, 2010; McLaughlin, Conron, Koenen, & Gilman, 2010; Van der Meer et al., 2012). Hence, the participants' experiences while they were growing up in a family affected by HD may significantly influence their decision to undergo genetic testing, as well as their reactions to the test result. Earlier research has shown that it may be important to identify the need for psychological intervention before a pre-symptomatic testing for HD is performed (Tibben et al., 1992).

According to the socio-ecological framework, the decision about whether or not to get tested is closely intertwined with the reactions, opinions, and presumptions of other people, including one's family circle, friends, or remote acquaintances; also, the decision would be influenced by the opinions of healthcare professionals. In this study, we found that other's opinions and values seemed to either reinforce or be in conflict with the participants' own beliefs and with their family experiences with HD; as a result, their thoughts and feelings about inheritance and about the test process were either reinforced or came into conflict with their values and beliefs. By considering the mutation status, other people (e.g., potential romantic partners) could also strongly influence the participants' important life choices, such as in the area of education or in having children. Recent research supports the idea that young people's experiences of HD depend on a range of factors similar to those in the socio-ecological framework (Kjøeelas et al., 2020). The findings highlight the need to evaluate the process of undergoing the test for HD not only as a personal choice but also as a lifelong process that is continuously influenced by a number of variables proximal and distant to an individual. Clinical geneticists and genetic counselors should be aware of the risk of such a predisposition when working with this vulnerable group. Moreover, healthcare professionals and other sources of support (e.g., family members and friends) for individuals at risk for HD should be aware of the complexity and the possible lifelong impact the decision to undergo predictive genetic testing can have on them.

### 4.3 Biographical disruption

Individuals who receive the news of being mutation-negative or mutation-positive may experience a biographical disruption (Bury, 1982). In our study, most of the participants had prepared themselves to know that they have inherited the mutation and thus they had created an identity as being mutation-positive. Many participants had constantly searched for symptoms and had even experienced symptoms of HD, some in young age, regardless of whether a future test will reveal a positive or a negative result. With this belief, they organized their lives accordingly, decided to make the most out of their lives as early as possible, or restricted their life choices. Hence, news of being mutation-negative compels these individuals to change their core identity (as mutation-positive initially) and redefine themselves (Duncan et al., 2007; Williams, Schutte, Evers, & Holkup, 2000). This group will also have to cope with the conscious or unconscious choices they have made prior to receiving the test results. Also, the quality of their relationships may change or may be challenged by the test results. For instance, individuals who do not have the mutation may experience survivor’s guilt (Tibben et al., 1992) or feel less connected to family members who do have the mutation (Duncan et al., 2008; Williams et al., 2000).

Understandably, test results could trigger powerful emotions, such as fear, anxiety, anger, or depression, regardless of whether the results were positive or negative. Having chosen not to pursue higher education or not to have children based on the assumption of having inherited the disease is expected to trigger challenging psychological reactions, such as loss and grief in sharp contrast to the more recognized and accepted feelings of happiness and relief. Unfortunately, suicide ideation and suicide rates are higher in HD population than in the general population (Robins et al., 2000; Solberg, Filkuková, Frich, & Feregen, 2018), and these issues must be addressed in the follow-up consultations with the individuals who were found positive and negative for the HD mutation.

Clinical experience and research have proven the occurrence of delayed grief reaction (Sobel & Cowan, 2003), whether individuals have or do not have the mutation. Further, individuals who receive a predictive test result of being mutation-positive may go through a liminal stage after the test, wherein they feel neither healthy nor ill (Gargiulo et al., 2017). Hence, a post-test follow-up should take time into account and if possible be tailored to individual needs. The progressive nature of HD, for those receiving the news of being mutation-positive, must also be taken into account, wherein
4.4 | Study limitations and strengths

The strengths and limitations of this study must be considered when interpreting our findings. First, the participants are believed to broadly represent the Norwegian population at risk for HD in terms of age, sex, and mutation status. No age restriction was set for the adult participants, and we consider the varied ages of our participants a strength. The diversity of participants also provided descriptions about some of the processes leading up to a test decision, pre-symptomatic genetic testing for HD at different time points in the participants’ lives, and reactions after the test for both mutation-positive and mutation-negative participants. Moreover, the involvement of researchers with different professional backgrounds (e.g., a psychologist and a clinical geneticist) in the conduct of the interviews and in data analysis is considered a methodological strength of our study. However, the interview guide was not developed to systematically explore the topic of genetic testing, which is a limitation of our study. However, one of the strengths of this qualitative research is the possibility of discovering and exploring areas that are of central importance to the participants. The present data show that a topic not covered by the interview guide was still raised by most of the participants. When the participants mentioned genetic testing, the interviewer explored this topic further. Furthermore, the time it took for the participants to arrive at a decision whether to undergo testing might have influenced the participants’ perceptions and experiences. Unfortunately, given that the interview guide was not developed to systematically explore genetic testing, the participants were not asked about how long before they decided to undergo or not to undergo genetic testing. Nevertheless, we believe that this study sheds light on several important aspects of genetic testing for individuals at risk for HD which are described and discussed in the present study.

4.5 | Clinical implications

Clinical experience and research, as well as the present findings, suggest that healthcare professionals should be aware of the need for support and should provide the same to help all individuals at risk of HD throughout the period of decision making. Our findings emphasize the importance of exploring factors that influence individuals’ socio-ecological framework in pre-test counseling. Individuals who are found to not have the mutation for HD should be scheduled for a follow-up consultation with a genetic counselor or a healthcare provider with an expertise in psychology; note that such consultations are usually only set for individuals who are found mutation-positive for HD. Our results highlight the complex and often challenging emotions that the participants experienced prior to the test and after receiving the test result, regardless of the outcome. Their reactions after receiving the result often persist, as well as change, over an extended period of time; thus, the changing needs of the tested individuals should be incorporated in follow-up routines. We therefore suggest that follow-up consultations should be conducted on a regular basis among those who underwent testing, regardless of the test results; moreover, when possible, the individual’s closest relatives, such as his or her life partner, should also be offered follow-up consultations.

4.6 | Research recommendations

Despite the growing attention given to the possible risks posed on individuals at risk of HD, more knowledge is still needed. Future research should further explore the experiences of those at risk while they were growing up in a family with a parent with HD; moreover, future research should contribute novel insights into issues associated with being at risk, into the choices available in predictive testing, and into the reactions toward and the consequences of the test results.

5 | CONCLUSION

Genetic counselors and clinical geneticists should keep in mind that individuals at risk for HD might have adapted their lives to the perceived certainty of being mutation-positive for years before deciding to undergo predictive testing. Having made significant and non-reversible life choices prior to genetic testing may strengthen one’s emotional reactions to test results, whether negative or positive. The current study suggests that individuals who receive the news of being mutation-negative should, in the same way as individuals who receive the news of being mutation-positive, automatically be scheduled for routine follow-up consultations soon after receiving the test results.

AUTHOR CONTRIBUTIONS

KBF, SHK, and CvdL designed the study; KBF and CvdL conducted the interviews; KHT, CvdL, SHK, and KBF analyzed the data; KHT, SHK, and CvdL drafted the manuscript; KHT, SHK, ED, KBF, and CvdL revised the manuscript critically and contributed to important intellectual content; and KHT, SHK, ED, KBF, and CvdL approved the final version of this manuscript.

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COMPLIANCE WITH ETHICAL STANDARDS

Conflict of interest

Kristine Hansen Tillerås, Siri Hagen Kjøelaa, Elisabeth Dramstad, Kristin B. Feraen, and Charlotte von der Lippe declare that they have no conflict of interest.
Human studies & informed consent

All procedures were performed in accordance with the ethical standards of the concerned committees on human experimentation (institutional and national) and with the Helsinki Declaration of 1975 as revised in 2000. Informed consent was obtained from all participants.

Animal studies

No non-human animal studies were carried out by the authors for this article.

DATA AVAILABILITY STATEMENT

The data are not publicly available due to Oslo University Hospital’s privacy restrictions, since data contain information that could compromise the privacy of research participants.

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SUPPORTING INFORMATION

Additional supporting information may be found online in the Supporting Information section.

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