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ORIGINAL RESEARCH

LQTS Parents' Reflections About Genetic Risk Knowledge and their Need to Know or Not to Know their Children's Carrier Status

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Abstract Long QT syndrome (LQTS) is a contributor to unexplained deaths in infants (SIDS), children, teenagers and young adults. A gene test result may allow for individual tailored treatment, but also pose a burden of knowing one's carrier status, with no treatment recommendation. Genetic risk knowledge in the case of LQTS can promote adjustment and coping, but also fear anxiety, ambivalence and moral dilemmas. This makes it challenging to respect both the right to know and the right not to know. The purpose of this study was to explore LQTS parents' perception of genetic knowledge, and their need to know or not to know about their children's carrier status. Qualitative, semi structured interviews were conducted with thirteen parents of LQTS-children. Results show that parents found it important to know the result of a gene test for LQTS including their children's carrier status. The risk was framed and incorporated into their everyday life and their life perspectives. Pertinent moral dilemmas concerned information disclosure to children and relatives. Parents thought that early and gradual disclosure to children would promote coping. Parents' moral dilemmas were rarely addressed during encounters with healthcare providers. The participants had several suggestions for improvement in that regard.

Keywords DNA test · Long QT syndrome · Parents · Child · Genetic knowledge · Risk information · Responsibility · Rights · Autonomy

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Introduction

There is growing evidence that Long QT syndrome (LQTS) is an important contributor to sudden, unexpected deaths in infants, children and teenagers (Schwartz 2004). LQTS is identified as an important contributor to SIDS (Arnestad et al. 2007; Schwartz 2004). LQTS is a genetic disorder related to mutations in several genes that encode cardiac ion channels. The most common characteristics of a predisposition to arrhythmia are prolongation of the ventricular action potential duration during cardiac repolarisation, measured as the QT interval on the electrocardiogram. LQTS is characterised by interval prolongation on the ECG (Modell and Lehmann 2006). The genetic mechanisms behind LQTS are heterogeneous, and various gene variants may contribute to LQTS (Modell and Lehmann 2006).

Eight major genotypes (LQT1-8) different mutations and polymorphisms are described, and the various LQTS genotypes identified correspond to mutations in different LQTS genes. In the LQT1 gene (KCNQ1 and KvLQT1) 62 % of events occur during exercise, especially swimming. In LQT2 (KCNH2; HERG) events, 43 % were connected to episodes of emotional stress such as fear and anger. Also LQT2 patients' events could be triggered by sudden intense auditory stimuli (alarm clocks and phones). LQT3 (SCN5A) was associated with attacks of arrhythmia occurring during rest and sleep (39 %). Thus, the physical manifestation and the implications for treatment will vary according to the genetic disorder. LQTS may be asymptomatic and lead to events of syncope or to sudden death, and LQTS may also cause sudden and unexpected death in individuals of young age (Modell and Lehmann 2006).

Two different forms of inherited LQTS are described. The first more common familial form is referred to as The Romano-Ward syndrome (RWS), with cardiac electro physiologic abnormalities of LQTS and normal hearing. The

second familial form is the Jervell Lange-Nielsen syndrome (JLNS) which is characterized by a high incidence of deafness and sudden death (Modell and Lehmann 2006).

In some families, LQTS may appear with a very low penetrance, and individuals with a normal QT interval may experience syncope and cardiac arrest (Priori et al. 1999). This poses considerable challenges for diagnosis, medication and counseling, and some of those who are treated will probably never experience symptoms (Hamang et al. 2009). Thus, some LQTS-carriers may deal with the burden of knowing about their carrier status, and with the knowledge of the side effects of medication. Moreover, they will be aware of the risk of a lethal disease (Hamang et al. 2009). Genetic testing can pave the way for individually tailored treatment for gene carriers of LQTS, depending on the accuracy of the test and—at best - it's potential to predict an effective treatment. Nevertheless, a gene test may also be inconclusive with respect to a treatment regime. Thus, there is a risk that the result of a test may yield knowledge of being a LQTS carrier, and knowledge of a potential unpreventable condition. In this case, the person will be aware of the risk of sudden cardiac death and the fact that there are no treatment options.

Ambivalence about the value and impact of DNA test information seems to influence patients who have experienced DNA testing and cascade screening for hypertrophic cardiomyopathy (HCM) and LQTS (Smart 2010). A negative test result for LQTS in a symptomatic patient does not give a clear and definitive answer, nor does it remove uncertainty. Another source of ambivalence can be the impact a positive DNA diagnosis for LQTS might have on a person's quality of life. Because a positive DNA result does not give certain knowledge about the prognosis, it might be better not to have to live with the knowledge that one is at risk for sudden cardiac death. DNA testing may not relieve the uncertainty about personal risks, and testing may have negative social and psychological consequences of its own (Smart 2010).

Parents of children who have tested positive for LQTS report a strong psychological reaction to the news of having an affected child experience, including high levels of anxiety and distress (Farnsworth et al. 2006; Hendriks et al. 2005a, b, 2008). LQTS patients seem to worry more about their children's risk of sudden death, than about their own death (Andersen et al. 2008; Farnsworth et al. 2006; ten Kroode et al. 2000). Parents' fear for their children is described as general and nonspecific (Farnsworth et al. 2006); accordingly, LQTS parents' awareness and uncertainty of a child's risk of sudden cardiac death may be associated with ambivalence and dilemmas. However, parents' distress seems to return to normal levels in the long term as they become more knowledgeable and adjust to the situation (Farnsworth et al. 2006; Hendriks et al. 2008). These findings indicate that parents actively and gradually acquire genetic risk knowledge while they also adjust and cope. Genetic risk knowledge in the case of LQTS has the potential to promote fear and anxiety, ambivalence and dilemmas, but also adjustment and coping.

LQTS patients report dissatisfaction and distrust in a healthcare system that lacks competence regarding LQTS (Andersen et al. 2008). It is vital that healthcare providers become educated about LQTS and about how patients and families experience the diagnosis (Andersen et al. 2008; Farnsworth et al. 2006; Hendriks et al. 2005a, b, 2008). Knowing how LQTS parents perceive and manage genetic risk is crucial for proper information disclosure and successful risk communication. Potential ambivalence and dilemmas may be highly influenced by parents' value judgments and moral deliberations. Hence, to understand and communicate well with parents we need more knowledge of how they perceive and manage genetic risk knowledge. Accordingly, the purpose of this study is to expand our knowledge about how LQTS parents' perceive and manage genetic risk, including their need to know or not to know their children's carrier status.

Methods

Participants

The participants were thirteen parents of LQTS positive children. The Norwegian Association for Children with Congenital Heart Disease contributed to the recruitment. The participants were recruited through an invitation and presentation of the study in the organization's magazine. Parents who wanted to participate contacted the organization or the researcher (first author) directly. Oral and written information was disclosed, and participants signed a written informed consent. The Norwegian Social Sciences Data Services (NSD) approved the project. According to the Norwegian Health Research Act approval from the Regional Ethics Committee (REC) is not necessary for persons who are not patients. Thirteen LQTS parents (27–53 years old; median= 36) were recruited between July of 2010 and June of 2011. Nine nuclear families were represented in the sample; two parents were brother and sister belonging to the same extended family. In four cases, both parents were interviewed. Eleven interviews were carried out individually. In one interview (of a couple), however, both participants were present.

All parents (n=8 mothers, n=5 fathers) had undergone DNA testing for LQTS, and seven had tested positive. In addition, in two of the families, the positive carrier status of one parent and one child were also associated with family history and ECG test results. Four of the LQTS-positive parents described themselves as healthy and without symptoms, though they took precautions in order to avoid seizures. Three of the LQTS-positive parents had experienced fatigue, dizziness, and fainting. One of these parents, who had

experienced serious arrhythmias, also had an implantable cardioverter defibrillator (ICD). Six parents had close relatives that had died of LQTS.

The families had a total of 21 children. Fourteen children had tested positive for LQTS, and 12 of them were being treated with beta-blockers. At the time of the interviews the age of the LQTS positive children who were put on beta-blockers were between 4 months and 18 years old; median=8. One child had implanted an ICD, and in one family, the child's carrier status was a gene mutation. Two children had a borderline test result, and in a third case, the test result was unclear. One parent was waiting for the infant's test result at the time of the interview. The time period between diagnosis of the fourteen LQTS positive children and the time of the interview varied greatly (4 months—8 years; median=3). One parent had tested herself during pregnancy 2 years prior to the interview, and her children 4 months before the interview session took place. In four cases however, the time of diagnosis of both children and parents was between 3 and 7 years before the time of the interview. In addition, in two cases, the time of parental diagnosis was about twenty and 30 years before the interview sessions took place. Six of the children had experienced symptoms related to LQTS after testing. Some parents talked about seizures; others described vague symptoms including side effects of medication.

Interview Protocol and Procedures

Qualitative interviews were conducted using a semi structured interview guide. The interview guide contained three main topics: 1) What they know about LQTS; 2) The significance of having knowledge about LQTS; 3) Information disclosed to others than the patient: to children, relatives. The interview sequence was mainly performed as described in the interview guide (Appendix 1). However, a flexible approach and open-ended questions were used. Hence, apart from the first topics in the interview guide and background information, the order of the topics was adjusted depending on the main issues and concerns of the parents. The first author conducted all interviews. Depending on participants' choices and on convenience, the chosen interview settings were the participants' homes or other appropriate places.

Data Analysis

The interview sessions were audio recorded, transcribed verbatim, and de-identified. The analysis was performed according to a four step analytical approach described by Malterud (Malterud 1993) and based on Giorgi's phenomenological method (Giorgi 1985). First the interview transcripts were read several times in order to gain an understanding of the text as a whole. Next, meaning units describing parental perceptions and management of genetic knowledge, and their need to know or not to know their children's carrier status were identified. Phrases, sentences and paragraphs were the units of analysis. The meaning units in each interview were summarized in order to get an overview of the material. Then the content of the meaning units was condensed and abstracted, and categories were developed according to the purpose of the study. Finally, the statements in each category were reformulated and transformed into general descriptions. The analytical process started at the same time as the collection of the data. We used a flexible approach, and the interviewer (MM) monitored the analytical process. The two authors (MM and BH) discussed the analysis together and disagreements were resolved through a process of discussion and consensus. Finally, the categories were validated against the original transcripts of the interviews.

Results

Three main categories and seven sub categories were identified describing parental perceptions and management of genetic risk knowledge, and their need to know or not to know their children's carrier status. Parents' value judgments and moral deliberations were intertwined in their descriptions of how they lived with the condition. The three main categories were: "To perceive and manage genetic risk," "Parents' desire to know about their children's carrier status," and "Genetic risk communication with healthcare providers."

Category 1: To Perceive and Manage Genetic Risk

Genetic Risk Knowledge is of Vital Importance

All parents stated that it had been of vital importance to know about their own, and their children's carrier status. This was described as a strong conviction, independent of factors such as the family history of the genetic disorder, time of diagnosis, side effects of medication, or a child's inconclusive test result. Participants emphasized the potential fatal consequences of the condition and their belief in the protective impact of medication. Hence, most participants described the decision to test as clear, without serious deliberations.

> *"It concerns a child, so I'm very happy that it is discovered. At least when I know how dangerous it can be if she's not taking medication. So I would never do anything other than what we've done."*

A crucial factor influencing many parents' views about the benefit of genetic testing, was their own and their children's pre-test history. All parents described their family history that had led to the decision to test. Eight parents described how

they made the decision after becoming aware of a family history of heart failure, misdiagnosis, late-term abortions, and sudden, unexpected deaths of young family members.

Several participants were concerned about the psychological burden on young relatives due to societal ignorance and delayed genetic testing. Participants recalled their own experiences as adolescents, and their concern for young, undiagnosed relatives. Parents described how they had been treated with the wrong medication and had been misdiagnosed or undiagnosed. They were also concerned about how undiagnosed adolescents and young adults had been subjected to suspicion and stigma. One parent described how an adolescent in her extended family had consulted a number of doctors before the genetic defect was randomly discovered.

> *"An adolescent who is sent back and forth, throughout puberty, a difficult phase of life. In addition, not being believed."*

One parent described a long-term deliberation before she made the final choice to test her children. Although several family members had suffered sudden cardiac death, neither she nor her children had ever had symptoms. Hence, in retrospect, she described her moral dilemmas connected to the choice of whether or not to test her children. Dilemmas were associated with the risk of overprotection, feelings of unfairness among siblings, and the insecurity connected to diagnosis and effect of treatment.

> *"There were both pros and cons; I have not had any symptoms. Then I wondered, 'Should I expose my kids to this? Am I going to be overprotective if one of them has the defect? On the other hand, if they take the medication, then they are a little more protected.' Yes, it took quite a long time before I could make up my mind. The reason really was that I was a little afraid that I would give the one that had it special attention, and then discriminate between my kids."*

Several statements described incidents where parents were uncertain whether the child's symptoms were heart related or whether they were caused by side effects of their medication. However, considering that medication would prevent unexpected, sudden death, parents were satisfied that they had chosen to test. In addition, parents of children with inconclusive test results relied on regular cardiologic check-ups. They hoped that through such check-ups they would gradually obtain more certain knowledge about their children's risk. They also hoped that further research would yield knowledge that could give certain, precise answers to their own, and their children's risk. One parent wished for reliable knowledge rather than living with the awareness of an inconclusive test result:

> *"They say that it may well be that she has the defect. She must take certain precautions, too. She has the defect, right, but it hasn't shown yet. I think that it would be good to know. With respect to her, it would be good to know whether she has the defect or not. Now we have to wait and watch for five years."*

Two parents and two couples were aware of the risk of having a LQTS positive infant before they became pregnant. Some parents shared their thoughts on having more children. The possibility of having another child with LQTS was not alone a hindrance to considering reproduction. Parents were concerned about their desire for siblings for their children, and they expressed concern over whether the family could handle caring for another child with special needs.

Response to the Diagnosis

How parents' responded to the diagnoses was highly influenced by their pre-test history. Some participants saw the positive test result as a relief. Finally, their symptoms were explainable; it was possible to initiate treatment and take precautions in order to prevent seizures. Other participants described increased distress, anxiety and fear after receiving the positive test results. Several statements illustrate the dramatic and overwhelming transition from the pre-test state of not knowing about the risk, to awareness of the risk of sudden, unexpected death.

> *"I was shocked, devastated. It was as, you know, we have two healthy children, we are a healthy family and life is great! And suddenly, it was a huge difference from what we were used to."*

A participant with a positive carrier status described the sudden and persistent fear of death after receiving the positive test result:

> *"I have never thought there was anything wrong with my heart, but suddenly I became very, very aware of my mortality. And I could die anywhere, anytime. The first symptom is deadly for 40 %, I've been told. And gosh, that's crazy. And I remember I just wanted to sit at home and crochet, for a while I spent a lot of time on the couch."*

Several statements illustrate parents' feelings of guilt for having transferred the LQTS gene to the children. One participant described a strong reaction with feelings of anxiety and guilt when he received the test result:

> *"When we got the test results, I was really shaky, I cannot remember, I've lost the memory about what really happened. Moreover, when I learned that I was the 'culprit': Panic! What is going on in my body? It was*

horrible. I have never had problems with anything; have been healthy as a horse. I was scared to death."

Handling Genetic Risk in Daily Life

Parents described how they actively adjusted and coped with the LQTS-related risk on a daily basis. Several statements illustrate how they tried to balance their attention to the risk in order to not be overwhelmed. Parents' ability to complete activities seemed to be crucial in order to cope with risk. They described how they performed a variety of everyday activities and tasks that required their continuous attention, presence and action. One father stated that they as parents had a very pragmatic attitude to knowledge, and that the family strived to live as normal a life as possible. Another parent with long-term experience of being a LQTS carrier described how she pushed her own limits in order to cope with her fear:

"And I love to hike in the mountains! So, it was like, okay: 'Let's go, if I die now then I die doing something I like,' and if I don't, then that's great! It is pretty far up there, and very steep, and it got to be something I just had to do. When I was standing at the top, and could see the whole world and know that okay I'm not dead yet, It was a turning point. Maybe I die on the way down, but that'll just have to be. It was such a very important milestone; from that time, it was important to change my frame of mind. Now we can no longer wait for death. And since then, I haven't done it either."

Sometimes the risk was the focus of parents' attention; in other phases of life however, there was a room to live a normal life. However, certain events could suddenly trigger fear and anxiety, and their perception and management of the risk seemed to change continuously. Factors such as their children's changing health condition, or unpleasant encounters with health care providers could easily shake their sense of stability.

"But we get little flash-backs, we get very scared sometimes. Every time she starts a new activity. So we've sort of learned to live with it, but none of us are really a hundred percent sure how dangerous it is for her."

Three participants used their own long-term experience as LQTS carriers when reflecting on their children's carrier status, and their potential capacity to manage their condition. One parent was a carrier himself and had a long-term, grave family history of LQTS. He expressed the values involved in living a good life with the LQTS diagnosis:

"It is important to know that even though you have this genetic defect, you can live a perfectly great life. You cannot live a hundred percent in some areas, and those are the areas that you need to identify. Nevertheless, I do not consider LQTS to be a disease that prevents my daily life. It often makes life extremely exciting. That's just something you have to take into account. It's scary to let your eight year old bike to school the first time, too."

Dilemmas were associated with potential consequences of genetic testing, and parents questioned whether the potential benefits of the medication would make up for the side effects. They were also concerned about how awareness of a positive test result would influence the care and upbringing of their children. Parents expressed moral deliberations including responsibility, conscience, duty and thoughts about what was the best option for their children's quality of life.

"She is a Scout, and she think it's great. And I have asked her: Would you like us to join? And she doesn't. I don't want her to be that girl who has her parents along all the time. Are we supposed to hang around her all the time because we are so damn nervous for her heart to stop?"

Hence, parents' moral dilemmas involved deliberations regarding their responsibility to do no harm, and their duty to promote the child's future autonomy.

Category 2: Parents' Desire to Know About their Children's Carrier Status

Disclosure, Restrictions and Quality of Life

There was broad agreement among parents that giving open and honest information to the children was essential. Parents were concerned that all children face adversity in life, and that it is fundamentally wrong to hide such essential information. All parents thought that gradual and adjusted information disclosure to the affected children would help them to cope with and manage their condition. One participant who was a carrier and who had experienced dramatic events in his own childhood stated:

"Today everyone is so worried about traumatizing kids. Many kids experience trauma in childhood; it's important to use it to turn it into something good."

Another participant emphasized the value of trustworthiness and honesty in the encounters with children:

"There were so many in the family who tried to hide the truth to protect the children. Children should not have to experience those kinds of things, but if they do, it is my

belief that they should get the truth. Sooner or later they'll find out that what they were told wasn't the truth."

Parents described how they used simple terms in order to explain the disease, including the precautions, to their children. However, several parents experienced dilemmas and doubts regarding content and timing of genetic risk information to children and adolescents. One parent said:

"Yes, that is the most difficult part! We still probably haven't cracked the code of how to talk about it. Because we have not told her how dangerous it is. She really does not know how dangerous it is. But what we say to her is that she has a heart condition and that she needs medication to get the heart to beat evenly."

Several parents had worries concerning the potential consequences of medication. They reflected upon whether or not medication would protect against fainting and sudden death or if serious side effects would occur. They were also concerned about whether or not medication would influence physical capacity and quality of life as their children matured.

Parents were concerned about the challenges such as overprotecting their children, or letting them explore their own boundaries. They dealt with dilemmas such as their desire to foster their children into autonomous human beings, while simultaneously protecting them from the risk of sudden, unexpected death.

"She's on the beta blocker but then, she lives quite normal. She bikes off where she wants to, you know, she is not at all overprotected."

Several parents were concerned about how children and adolescents could grow up and mature as autonomous human beings. One participant illustrated the parental dilemma using experiences from her own adolescence:

"Some were so sheltered that they were still at the fetal stage in a way, 'This is where I belong, with my parents and this is where I will always belong.' Others are in great opposition and aggressive to their parents. I think that is also very sad, that you end up with a situation where the kids oppose their parents because they have been overprotected. My parents tried to talk sense to me and make me think wisely and use my head. However, at the same time, in many cases they let me make the decisions. And I have told them so many times how grateful I am for that."

A recurring theme was communication barriers between parents and adolescents about risks related to LQTS. There was broad agreement among parents that abrupt, unprepared information disclosure to adolescents about the risk of sudden death could have devastating, long-term consequences for their quality of life. Parents were determined to get the special genetic counseling and follow up that adolescents need.

Concern for Children in the Extended Family

Parents expressed a strong responsibility for children and adolescents in their extended family. Hence, informing relatives was considered as a moral obligation in order to avoid serious harm. On the other hand, to inform about the potential genetic risk was also in conflict with their relatives' fundamental right not to know.

"I now feel it is a duty to notify the family if there is anything, if another family member could have such a disease, I wouldn't have the conscience not to tell. Imagine the scenario that a family member had a child that died within the next five years without actually realizing that we have the disease that caused the child's death. In such a case, my guilty conscience would have killed me."

Several parents described the competing notions of withholding or communicating genetic risk to relatives as intrusive value dilemmas. Family communication was sensitive, and several parents said that the issue was difficult to handle because a number of competing interests had to be taken into account. Parents described feelings of responsibility, conscience, anger, guilt, and vulnerability. They did not always want to know how relatives handled the information, whether they actually choose to test or not.

"I do not want to have some kind of family dinner where we discuss pros and cons of genetic testing. It would just be insane, I couldn't bear it. This is too awful for me. Now I have the gene, so now you have the information, and then just do what you want. Afterwards I have not asked them whether they tested or not."

Category 3: Genetic risk Communication with Healthcare Providers

To be Acknowledged

Parents' perception of genetic risk was closely associated to how they perceived and interpreted risk information from health professionals. Parents described their encounters with the healthcare service on various levels: genetic counseling, the cardiologist and the primary healthcare services. Several participants were concerned about the knowledge gap and lack of coordination between various levels of the service. Parents' evaluation of their communication with providers

varied, and some reported both positive and negative experiences. Some parents described encounters with professionals as supportive and humane. However, several parents were dissatisfied with the limited time spent in the specialist consultation, and some expressed profound distrust in the healthcare system. Hence, several statements illustrated how genetic risk communication that included parents' own issues and concerns was impossible within the narrow framework of a consultation. One parent who had a child with an inconclusive test result described a typical consultation:

> *"You notice very quickly. You arrive and they ask: 'You are okay?' Then he asks a little about family history, and you sit there and you have forgotten to take notes and you know that 'Well, end of conversation.' When they enter the room, you see they are ready to walk out again as soon as they arrived, they certainly have more rooms to visit."*

Contextual factors influencing patients' own perceptions and management of risk were rarely a theme during encounters with health care providers. Parents felt that there were limited opportunities to share their own daily experiences, their thoughts and worries about their children's individual level of risk.

> *"Perhaps the next step for us now is to get some guidance on how to talk about it without making her very scared."*

Several statements indicated that the conflicting advice from different professionals, especially regarding medication and activity for their children, was confusing. Several parents described their daily experiences and observations of the affected child. Parents described daily challenges and personal risk assessment that they wanted to discuss with health care providers.

> *"I think that it would be nice to have someone who could give pointers of how much to restrict children. Because it is like, you're not supposed to wrap them in foam and protect them from the rest of the world; you cannot do that their whole life anyway. Moreover, they have to learn to stand on their own feet. At the same time, then, it is something that needs to be addressed, then because it can be too much. It is very difficult I think, as parents to know where the boundaries are."*

Parents made considerable efforts, building up personal competence and systems in order to manage their children's risk. Parents made arrangements, including educating schoolteachers and providing facilities for worst-case scenarios. Several statements emphasized the importance of collaboration between professionals and parents in order to improve and facilitate genetic risk communication within families. Parents expected health professionals to assist and help them in conveying information to family and relatives.

Precisely, How Risky is LQTS on an Individual Level?

Parents were aware of the limited scientific knowledge base regarding LQTS and that the physicians' ability to predict individual risk was limited. However, they wanted a more open and honest dialogue with physicians about what they actually knew about LQTS, including their lack of knowledge. One participant said:

> *"I think they are terrible at taking the time to explain properly. If it is because they do not know or have little knowledge, then they ought to say just that. It would be more appropriate to say that, 'We cannot give you the answers and we cannot explain, because we do not know'."*

Parents also wondered exactly how dangerous LQTS actually was in their specific case. Parents wanted specific information about the diagnosis and an open dialogue with health care providers about their children's risk. One participant commented:

> *"I'd love to know what type of LQTS he had, and what specific things that can trigger seizures, and we know that they can find out in some case, 'You have type 1,' or 'He has type 1.' It is the same way if you have cancer. You would like to know if you have cancer of the foot or if you have it in your shoulder. And I think, 'Okay, you have a long q-time syndrome type 1, 2, 3, 4, 5,' and some people get that information. So then I think that they should either say what type he has, otherwise they should say, 'We do not know yet, because we haven't found out'."*

Several statements emphasized the lack of professional competence among general practitioners, including conflicting and inappropriate professional advice. Several statements indicated that communication with health professionals about the actual risks associated with LQTS was confusing.

> *"But I do not know how high the risk is. No one dares to tell me I think. Or in other words, I feel like, whenever I ask I get different answers."*

Several statements illustrated how parents got conflicting advice depending on which specialist they asked.

> *"I think it's very confusing. Since there are so many in our family who has the gene, and we've gotten different answers to the same question from the same doctor, so*

we're confused. One says: 'No, don't do it!' and then the other, 'No problem, just keep on!' So you get a little confused what you are allowed and not allowed to do."

Health anxiety was closely associated with the fact that health professionals could not exactly estimate individual risks associated with LQTS. Several parents did not receive the information that they considered to be useful. Knowledge about a positive test result was important in order to assess what they had to take into account in daily life activities in order to manage risks.

Discussion

The parents who took part in this study believed that it was of vital importance to know the result of a gene test for LQTS. These parents did not show ambivalence or report experiencing dilemmas connected to the basic question to test or not to test, to know or not to know. The alternative, being unaware that they or their children were at risk for sudden cardiac death did not appear relevant. Despite their uncertainty about personal risk, their willingness to know seemed to be fundamental. They wanted to get specific information about their condition.

However, several parents experienced ambivalence and dilemmas connected to what, how and when to disclose information to their children. They were aware that disclosure could have different outcomes, either be beneficial or have a potential to cause harm. They were especially concerned about adolescents' specific needs, and thought that they needed specific health services. Several statements revealed an intense need to discuss with health professionals how and when to inform their children and adolescents in order to promote their future autonomy and avoid harm.

Family communication about genetic risk was also a sensitive issue. Competing notions such as the duty to inform, relatives' right not to know, and guilt associated with non-disclosure caused dilemmas. Parents expected support from health professionals in order to facilitate family communication about genetic risk.

Parental perception of genetic risks seemed to change over time. Initially, after receiving the test results, several parents experienced the risk as dramatic and overwhelming. Over time however, parents seemed to adjust to the situation and several stated that they felt a sense of normality. However, several statements indicated that their sense of normality was fragile. Certain events could easily upset parents and dislocate their sense of coping and normality.

The participants described a variety of activities that they performed in order to resolve the dilemmas, to manage, and to cope with their own and their children's carrier status. Hence, parents were not passive recipients of genetic information. Instead, they managed risk within their daily activities and social relationships with their children, family and people in their surroundings. The risk was framed and incorporated in their everyday life and their life perspectives.

There was broad agreement among the parents that knowledge about their children's carrier status was of vital importance for the upbringing and promotion of their children's health. They thought that gradual and adjusted information should be given according to their children's maturity. Parents saw it as their responsibility to decide whether or not to test the children. Nevertheless, parents described dilemmas associated with their perception of genetic knowledge, and their need to know or not to know their children's carrier status. Despite this, there was broad agreement among parents that uncertainty was more burdensome than the state of knowing about the risks associated with LQTS.

Misunderstandings and lack of support and advice from healthcare providers comprised one factor influencing parents' perceptions of genetic risk. Parents pointed out several suggestions for improvement. In general, parents wanted more time during encounters with healthcare providers to discuss their own issues and concerns. Specifically, they wanted advice and pointers regarding medication and activity for their children. Parents wanted professionals' assistance and help in order to convey information to family and relatives. They wanted open and honest information about the limited knowledge base regarding LQTS. Parents also hoped to get specific and individualized information about their children's actual risk rather than inaccurate and conflicting advice depending on which specialist they asked. Parents did not feel that they had the opportunity to discuss their worries, dilemmas, and concerns during encounters with healthcare providers. Parents' suggestions may be valuable in order to promote better communication between LQTS parents and healthcare providers.

Parents highlighted their duty and responsibility to inform both children and relatives. They also emphasized children's and relatives' right both to know and not to know, and especially their respect for the (future) autonomy of others. Several statements indicated that parents' were very committed to prevent harm. However, our findings also challenge the view that ethical dilemmas associated with genetic testing of children can be understood and framed in terms of rights, responsibility and respect for individual autonomy alone. Our findings also challenge the view that the individual's perception of genetic risk is solely rational. Indeed, our findings indicate that both rationality and emotions were involved in parental perception of genetic risk. Feelings of good or bad conscience, anger and anxiety, responsibility, vulnerability for criticism, guilt, trust and distrust came in to play in parents' reflections about genetic risks and LQTS.

Results from a systematic review of family communication of genetic risk shows that the decision whether or not to

disclose risk information to relatives poses a dilemma between conflicting responsibilities (Gaff et al. 2007). Family members considered a desire to protect relatives from potential harm against a wish to provide them with information that might have important health consequences (Gaff et al. 2007). Parental decision to disclose or not disclose BRCA1/2 test result to children is complicated by moral and ethical considerations. Feelings of responsibility, the duty to inform or to protect the child from anxiety provoking information and feelings of guilt may influence parents' deliberations (DeMarco and McKinnon 2006). These findings are consistent with the results of our study.

The dilemma to test or not to test, to disclose or withhold risk information includes the ethical dilemma of balancing the obligation to respect the maturing child's future autonomy, without inflicting psychological damage. If there is no immediate medical benefit, genetic testing should be deferred until the child is sufficiently autonomous to make her or his own decision (Arribas-Ayllon et al. 2008). Genetic risk knowledge might have devastating social, emotional and psychological consequences, and thus be in conflict with the ethical principle of doing no harm. However, predictive genetic testing of children may also have psychosocial benefits, and facilitate the development of autonomy in the maturing child (Robertson and Savulescu 2001). Hence, genetic testing can be justified in terms of respect for autonomy (Andorno 2004; Arribas-Ayllon et al. 2008). Our findings however, illustrate LQTS parents' desire to discuss their moral dilemmas and deliberations with professionals. Hence, individualized genetic communication regarding LQTS parents' special needs may facilitate autonomy in the maturing child.

Parents are generally considered as best placed to judge what is in their own child's overall interests. It has been argued that a parental request for testing, after appropriate genetic counseling, should be respected unless there is clear evidence that the child will be harmed in an overall sense as a result of testing (Robertson and Savulescu 2001). The familial nature of genetic risk information creates a dilemma between balancing respect for autonomous decision making versus the responsibility of disclosing critical information (Hallowell et al. 2003). Consequently, parents' disclosure dilemma in the case of LQTS can be considered a question of balancing their parental responsibility of doing no harm without jeopardizing the child's current and future autonomy.

A biomedical model seems of limited use in understanding how affected individuals and families deal with genetic risks and knowledge about their children's carrier status. However, our findings seem consistent with an anthropological view, indicating that genetic information can be relevant on different levels (Rehmann-Sutter 2008). On one level, the user is confronted with the result of a test. The next level involves understanding the implications of genetic information for one's body. There is also a further level at which genetic information is reinterpreted as meaningful in social contexts. According to this perspective, it may be difficult to anticipate the personal and social reality of a test result on an individual level, and how ethical dilemmas can be modulated for the different persons involved (Rehmann-Sutter 2008).

Mol and Law (2004, p.45) describe three ways of defining the body: "The body we have" is an object for medical examination. "The body we are" or the "subject-body" is private, personal and a subject of self-interpretation and self-awareness. Also, integrated and as part of daily practice, we "do our bodies." Mol and Law (2004) describes "the body we do," by describing how people living with diabetes counteracted hypoglycemia. People described how they counteracted and avoided hypoglycemia. Several statements in our study also demonstrated how parents through a variety of activities were "doing genetics" and especially "doing genetic risks." Such doings were manifested in how they challenged their own boundaries in order to manage fear. "Doing genetic risk" was also expressed through descriptions of how they observed and handled their children's conditions in order to manage risk and avoid seizures in daily activities. Hence, parents did not seem to passively adjust to a state of normality. Instead, parents' perceptions and management of genetic risks were closely associated with their performance of daily activities.

The principle-based ethical guidelines concerning respect for individual autonomy become inadequate in the family setting (Arribas-Ayllon et al. 2008; Hallowell et al. 2003; McConkie-Rosell and Spiridigliozzi 2004). Given the difficulty in determining a psychosocial benefit, the discussion about genetic testing of minors ultimately tends to focus on who has the right to make the decision and whose right to autonomy is jeopardized, when there is no identified medical benefit: the parent's or the child's (Arribas-Ayllon et al. 2008; McConkie-Rosell and Spiridigliozzi 2004). Instead, a family-ethics model necessitates a cognitive shift where the personal voice of the family is regarded as equal in importance to the four basic principles of bioethics (McConkie-Rosell and Spiridigliozzi 2004). There is no universal right or wrong regarding when to test, and when to inform children.

Families should have access to multiple counseling sessions as their needs change through their child's developmental stages into adulthood. (McConkie-Rosell and Spiridigliozzi 2004) A family-centered approach may facilitate information sharing in families who have a child with a genetic condition (Gallo et al. 2010). Multiple counseling sessions could also promote trusting relationships between LQTS families and professionals. In this way, LQTS families would have the opportunity to share their unique family history, values and moral dilemmas. Important topics in such sessions would be parents' perception of their duties and responsibilities to their children and to members of the extended family.

Limitations of the study

Several weaknesses may be identified in our study. Qualitative data are not intended to be generalized to the population of interest. Moreover, given the small sample size and the limited setting in which the study was carried out, one should be careful about generalizing the results to other settings. In addition, due to the recruitment by a user organization there is a possibility that the participants were especially reflective, and interested in genetic knowledge. On the other hand, it is possible that the participants were those most in need of assistance.

All participants in this study had made the decision to go through genetic testing, and they were also aware of their own and their children's positive test result. Hence, they evaluated their choice in retrospect of the actual decision, procedure of testing, and after they had received and reflected upon their test result. In general, however, people might have a tendency to defend and justify their own choices. Such a need might be especially strong when it comes to existential issues. The time of the interview related to their genetic knowledge and their actual decision might have influenced the responses in this case, and consequently the results. Hence, a sample consisting of interviews conducted before genetic testing, of those considering the prospect of genetic testing, might have revealed other results or given more nuances on the topic. It is possible that such an approach would have uncovered more information about people's deliberations regarding genetic risk knowledge. However, our participants' arguments for their need to know seemed well founded in their ethical reasoning and in the rationale underlying their viewpoints.

Research Recommendations

More research is needed to explore how people reason regarding genetic risk knowledge. Under which conditions do they want to know, and under which conditions do they not want not to know about their genetic risks? What are people's reflections before they decide to test or not to test? Which factors contribute to fear, anxiety, ambivalence and dilemmas or to adjustment and coping?

Non-professionals and experts' perception of genetic risks seems to differ considerably. There is a need for a more in depth understanding of carriers' strategies in order to manage and cope with genetic risk. Daily activities and tasks seemed to be crucial in order to cope with genetic risks. In order to understand how individuals, children, adolescents, parents and families use activities in order to cope with genetic risks, more research is needed. An anthropological perspective may be useful in order to achieve these aims. Finally, more research is needed in order to explore more in-depth potential dilemmas associated with carriers desire to know or not to know.

Implications for Practice

Genetic counselors and healthcare providers in general, should acknowledge the complexity in LQTS parents' reasoning about genetic risk and positive test results. There is a need to support families in order to facilitate communication of genetic risk. A family-centered approach may be useful in order to achieve this. The findings of our study underline the importance of individualizing genetic information for the specific needs of the families with an emphasis on their moral deliberations, values and dilemmas. The findings also indicate that general practitioners' competence about LQTS varies and in some cases is limited. Hence, considering the rapid knowledge development in genetics, education of healthcare providers in genetic knowledge and public needs is urgent.

Conclusion

The LQTS parents saw it as important to know about their children's carrier status and to inform children according to their maturity. They showed a range of strategies in coping with and managing knowledge about genetic risk. Yet, they experienced a series of moral dilemmas associated with the state of knowing. Parents viewed information disclosure to relatives as a duty, even though this was a sensitive issue. Parents identified several suggestions for improving communication with health professionals. They also revealed a frustration over general practitioner's lack of competence about LQTS. Healthcare providers should support LQTS families in their efforts to disclose and convey information to children, adolescents and relatives. This study revealed a critical need for genetic counseling especially designed for adolescents and young adults. Healthcare providers should acknowledge parents' moral dilemmas associated with LQTS and adjust counseling sessions to meet the special needs of parents.

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Conflict of information Author Margrete Mangset and author Bjørn Morten Hofmann declare that they have no conflict of interest.

Informed Consent All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000 (5). Informed consent was obtained from all patients for being included in the study.

Appendix 1

Theme 1: What they know about LQTS

- Background information, personal and family history.
- What do you know about LQTS?
- Did you get any information about Long QT syndrome? From which sources?
- How was the information communicated and how did you perceive the information given?
- How do you perceive and consider the risks associated with LQTS? How do you evaluate the treatment options?
- How do you live with risk? Impact relative to daily life, satisfaction, confidence, insecurity?
- LQTS risk relative to other forms of risk. Driving a car, seat belts, bike in traffic area, sports, and outdoor activities?
- Do you have any thoughts about the uncertainty (risk) associated with living with Long QT syndrome?

Theme 2: The significance of having knowledge about LQTS.

For some forms of LQTS there are treatments that can prevent/avoid seizures – for other forms there is no effective treatment. A genetic test can reveal this—but it can also affect other family members as they can be carriers with no symptoms—and can transfer the gene defect to his/her children.

- Can you say something (in general) about the importance of having knowledge of LQTS?
- What impact has it had to know that your child (children) have the heart disease LQTS?
- How do you think it would have been for you not to know?
- What impact has it had that it was a genetically based cause?
- What impact do you think it would have had if the cause were non-genetic?
- How is the relevance of knowing or not knowing when it comes to reproductive planning?
- Do you think you have a right to know? A duty to know?
- What thoughts, considerations or objections have you had when it comes to test/not to test?
- Do you have any thoughts related to the consequences of testing yourself (or your children)?

Theme 3: Information to others than the patient, to children, relatives.

- Do you have any thoughts about informing your children about the risks of LQTS?
- Do you have any thoughts about informing other family members (relatives) about LQTS?
- Who do you think should disclose such information, parents, those who are carriers, health professionals, doctors?
- When should information be given? Only if the disease can be prevented or whatever treatment? Consent from the patient or independent from it?
- How should this information be disclosed?

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