How and Why Parents Change Their Attitudes to Prenatal Diagnosis

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ABSTRACT

Parents who know they are carriers of a genetically transmitted disease because they gave birth to an affected child are being offered a number of reproductive options in a subsequent pregnancy. We describe the attitudes and motives of parents of children with cystic fibrosis (CF) who were initially determined to use prenatal testing but eventually refused it. Based on our observations and clinical impressions we built a model of change that entails a series of stages and depicts change as a process. This begins with the provision of comprehensive information and emotional support through health professionals (Stage 1). The parents then engage in intense confrontation with the impact of prenatal testing and are exposed to influences from many sides. The repeated comparing of the arguments results in uncertainty and distress (Stage 2). When pregnancy occurs a decision must be made and the process culminates in participating in a life-or-death decision. If the parents cannot cope with that responsibility then a change towards rejection of prenatal diagnosis is likely (Stage 3). This phenomenon seems to be the most decisive factor. A case vignette is used to illustrate this model which may be applicable to many individuals who face major and emotionally fraught decisions.

KEYWORDS

cystic fibrosis, preimplantation genetic diagnosis, prenatal diagnosis, theory of change

IN RECENT DECADES there have been major medical advances, including in the area of several genetically transmitted diseases. Parents who have already had a child with a serious genetic disease or couples who know that they carry a defective gene are no longer at the mercy of an inherited disorder. They can now choose from a number of options. The first section of this article addresses how change occurs in the decision-making processes of couples who already had a child with cystic fibrosis (CF), who wanted further offspring, and who were offered the option of prenatal diagnosis (PD). In the second section we refer to preimplantation genetic diagnosis (PGD) and present a case to illustrate the process of change in a couple's attitude to PGD.

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Prenatal diagnosis in CF

Over the past 30 years we have helped and supported hundreds of patients and their families but we have also seen many children suffer and too many, sometimes very young, children die. The experiences with these patients, and in particular with individuals with CF, have greatly affected our interest in reproductive options.

CF is an inherited disorder with a 1 in 4 risk for subsequent children. The defective gene causes altered membrane function in a variety of organs, ranging from the lungs to the sweat glands. Although predicted median life expectancy for babies born today is 40 years the disease remains incurable and requires lifelong, time-consuming therapies. Physical limitations and psychosocial restrictions contribute to progressively decreasing quality of life.

To most patients and parents the diagnosis of CF represents a severe blow because they have to face an unexpected and unwanted turn in their life. There is no doubt that this diagnosis not only affects the child but for ever changes the life of all the people involved. Notably, the changes that need to be made are involuntary ones. In the past, parents of a child with CF who wanted to prevent the subsequent birth of an affected child had to give up the desire to have (further) offspring genetically related to both of them. With the introduction of DNA analysis in combination with amniocentesis and chorionic villus sampling, a wide range of disorders, including CF, became identifiable in the first or second trimester of pregnancy. In the case of a negative result, PD relieves parental anxiety early in the pregnancy. In the case of an unfavourable result the parents may prepare for the birth of an affected child or choose selective abortion.

In 1996 we reported that only 45% of couples who had planned to use PD in a subsequent pregnancy acutally did so (Jedlicka-Köhler, Götz, & Eichler, 1996). In subsequent years we continued to document parents' decision-making processes. For the purpose of this article we report the thoughts, attitudes and behaviors of parents who in the beginning intended to prevent the birth of an affected child and later changed their mind.

Why did parents change from acceptance to rejection of prenatal diagnosis?

Information about PD was provided 4 to 8 weeks after the birth of the child with CF and follow-up lasted until the birth of the subsequent child.

Although attitudes towards PD were manyfold and most parents gave more than one answer, the following items represent a rank order with regard to their frequencies. The

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predominant reasons for favouring PD in a subsequent pregnancy were the strong desire to have a healthy child, the anticipation of an unbearable emotional burden and of the inability to master the demanding management of two affected children, respectively. Further arguments are given in the parents' words: 'We wish to know the child's condition early on'; 'it is inhuman to knowingly burden the child with a disease'; 'we could opt for selective abortion'; 'we want to have a surviving child'; 'the good health status of our first child with CF may not be predictive for the second child'; 'we could not bear to see our child die'; 'two siblings with CF carry the risk of continuous cross-infection'.

When the parents actually faced a subsequent pregnancy, again, most of them gave a variety of reasons as to why they now rejected PD. But despite multiple motives, in almost all cases the major concerns centred around the stress of the procedure *per se* and around fear and selective abortion. The couples were afraid that they would not know how to react to a positive test result, they feared that they would be unable to terminate the pregnancy and at the same time they would not want to have two affected children. There was no knowledge deficiency; they all had been aware that as a result of PD they would need to face the question of pregnancy termination. But now, that pregnancy had become real, the confrontation with this conflict had become inevitable and no one else but the parents would be responsible for the decision. The fear of not knowing what to do in case of CF, feelings of guilt and feelings of disrespect for the first-born in case of selective termination, and upcoming moral concerns turned what was once considered a beneficial option into severe distress. As a result of this emotional turmoil parents made the decision not to prevent the birth of a possibly affected child but preferred to leave it to fate. For parents who in the meantime had seen their first child die the decision about the second child was too difficult to make.

Some parents escaped the dilemma of decision making by reporting the pregnancy too late or by pretending to not have been informed about PD. But there were also a number of other arguments that had made the parents change their mind. They now had seen their child grow and develop favourably in many ways, indistiguishable from their peers in terms of temperament, vivacity, and cognitive functioning. They loved their children, they had gained experience with the disease and felt more confident now with the management of its complex treatment. Due to the good health status of most children the parents were now less scared of the threatening facts of the disease and experienced trust in and hope for further scientific progress. They compared survival statistics and average life-expectancy, they learned of patients who were 60 years and over, and they concluded that because the chances had increased over the past decades this development would certainly continue. Their beliefs were supported by treatments like lung transplantation and the potential of gene therapy. Some parents speculated that CF just would not happen twice in a row, first because the chances were 1 in 4 and they already had an ill child, and second because destiny or God would not be that cruel.

Many parents had met adolescents and adults with CF and perceived this as convincing evidence that a person with CF can live a full life. Apart from communication with other patients and parents the couple's families and friends came into play. Their attitudes, advice, suggestions and recommendations reaffirmed, questioned or destabilized the parents' viewpoints. In one family the 14-year-old daughter with CF was strictly against PD because she said she would not have been born had prenatal testing then been performed. She found it 'disgusting and unacceptable that her parents would kill a foetus only because he/she was like her'. In another family the mother did not carry out PD because she knew – and we learned about it only after the birth of the second child – that the father of that child was not her husband. Some parents stated that they no

longer wanted to know in advance whether the child would have the disease or not and some chose termination of pregnancy without undergoing PD because they had decided that they would not want to have another child at all. In several cases reluctance to undertake the risks involved in PD was the decisive factor.

Towards a model of change

We will now try to build a model of change that may help to explain why parents of children with CF differed in their intentions to use PD at different points in time. Our model is empirically driven, entails a series of stages, and depicts change as a process.

Stage 1: Information and counselling

At this stage, usually a few weeks after the diagnosis of CF has been communicated, the parents are given the essential genetic and medical facts regarding PD and other reproductive options. The goal is to increase the range of options available to the parents so that reproductive choices can be made on the basis of personal preference rather than as a result of confusion or lack of information.

Counselling is given to provide emotional support and to help the parents to consider the impact and the ramifications of all available choices, for example, what would it mean to have a foster child? To adopt a child? To have no further offspring? To have two ill children? To abort a child? To opt for selective termination? How would the ill child affect the development of a healthy child and vice versa? What would an ill child as an adult say to his or her parents knowing that they could have prevented his or her birth?

Stage 2: Absorption of information and developing new concepts

This stage lasts months or years and therefore gives most parents ample time to reflect on their initial attitudes and to consider further possibilities and alternatives. Now that the first feelings of shock have lessened they begin to search for additional information and they are both voluntarily and involuntarily exposed to information from many sides. Disease knowledge and management skills increase, subjective health beliefs and the subjective interpretation of medical facts (e.g., 'It's only a 1 in 4 risk') add to or replace the health professionals' information. The opinions of family members, friends, care givers, and other significant people come into play (e.g., 'How can you possibly think of NOT carrying out PD'; 'you have to accept a child the way it is'; 'do you really want to undertake the risk of miscarriage?'), exert considerable pressure on the parents, and make them fluctuate between contradicting standpoints. Other parents serve as models (e.g., 'They have not used PD and yet their child is healthy') and the comparison with patients older than their own child evokes hope or dispair depending upon their health status. Support groups, the Internet, chat rooms, and medical conferences serve as additional sources of information enabling exchange of attitudes and experiences. Scientific advances such as the prospect of gene therapy are being eagerly followed and sometimes perceived or interpreted by the parents as a potential cure that would make PD superfluous.

It is our impression that the major feature of this stage is an ongoing weighing and comparing of arguments and uncertainty with regard to a future decision (e.g., 'We will only know what to do when we must decide').

Stage 3: Pregnancy, facing reality, action required

While in Stages 1 and 2 decision making happens on a theoretical level, Stage 3 requires a decision. The parents *must* now choose between two undesirable situations, that is, the uptake of PD with all its consequences or the risk of having another affected child. They

clearly feel the weight of the decision they have to make and they find themselves assuming responsibility for a decision they do not feel qualified to make. The parents do not have the desire to 'play God' and the once considered beneficial effect of PD now becomes stressful. Being placed in this position overburdens them and they want to have the burden of responsibility removed from their shoulders by someone else. But there is no one to assume this potential life-or-death responsibility and there is no convincing argument that may diminish the upcoming sense of personal culpability. The solution the parents choose to escape from this dilemma is to delegate decision making to fate or, in some cases, God.

All stages: The impact of facts the health professionals do not know

Even if we know the patients well and even if we follow chronically ill children and their families for many years we need to address the fact that there may be life events health professionals will not know about. We already mentioned the father of the child with CF who then was not the father of the subsequent child. Other situations might include marital discord, financial difficulties, unemployment, and so on. The point is that all these events influence not only the parents' lives in general but also contribute to changes regarding further offspring and the use of PD. Therefore apart from the influential factors which we have integrated in our model of change we need to take into account also the existence of unidentified ones.

A suggested model of change

With regard to the intended, but then rejected, use of prenatal diagnosis in CF we suggest that change is the result of a process that is based on the confrontation with a variety of factors the parents cannot access at the time when they find themselves in favour of PD. With time, pros and cons are repeatedly compared. The process may take both predictable and unpredictable turns. Among the predictable factors are the perception and interpretation of information, the impact of the opinions of significant others, experiences with the disease status of the living chronically ill child, and the development of the parents *per se* and their relationship, respectively. Unexpected turns include unforseeable events and all the things that happen of which health professionals are unaware. The key is in how all these elements interact to create change. The process culminates in a life-or-death decision. If the parents cannot cope with that responsibility then a change towards rejection of PD is likely. This phenomenon seems to be the most decisive factor. We speculate that this model may be applicable not only to parents of children with CF but also to other individuals who face reproductive choices that entail prenatal testing and termination of pregnancy.

Whether or not the information we conveyed and the counselling we provided led to a change of parents' attitudes, viewpoints, and decisions is for speculation. In contrast to what health professionals usually do, that is, identifying problems and suggesting ways of resolving them, we had no such target. There was no behaviour that had to be changed and no therapy that had to be adhered to. And yet there were parents in need who presented with totally different arguments – sometimes with a lack of understanding for opposing parents' viewpoints – and who often wanted guidelines, help and advice.

We conducted the initial and many of the following sessions together because we considered this setting as a means to supervise each other in order to remain neutral and nonjudgemental and to avoid taking sides without recognizing it. Therefore we believe that change did not take place through a coercive attempt by us or by persuasion or any other such means. We believe that we have played an important role because we

provided emotional support, delivered state-of-the-art knowledge, were available whenever a discussion was required, and accompanied the parents through their complex and distressing decision-making process. However, we are unable to present any measurable strategies or to prove any effect of our interventions.

Preimplantation genetic diagnosis

In the remainder of this article we address a reproductive option that differs in technology from PD but also produces enormous stress. Preimplantation genetic diagnosis (PGD) became available in the early 1990s. It is an early form of prenatal diagnosis, in which embryos created *in vitro* are analysed for well defined genetic defects; only those free of the defects are placed into the womb (Sermon, 2002). Essential components of the procedure include genetic analyses, ovarian stimulation, and assisted-reproductive technology. Unlike prenatal diagnosis, PGD offers couples who are at genetic risk the identification of embryos that do not have the potential for a genetic disorder. In contrast to PD the procedure preceeds pregnancy. Because only unaffected embryos are transferred to the uterus the need of invasive fetal testing and termination of an affected pregnancy can be avoided. PGD is a rather invasive, complex and time-consuming procedure, only available for a limited number of genetic conditions and with a pregnancy success rate of about 24%.

PGD has stoked public controversy and concern and provoked ethical debates. The main ethical arguments against it include the selection of embryos and embryo destruction, incidental sex selection, and the potential for eugenics. Pros and cons need to be respected (Cameron & Williamson, 2003; Watt, 2004) but it also has to be recognized that PGD is not the endpoint. New developments and technologies are being worked on (Sermon, Van Steirteghem, & Liebaers, 2004) and each novel procedure will raise new questions and ethical considerations.

Attitudes of at-risk subjects towards preimplantation genetic diagnosis

Several studies have investigated populations of high-risk couples of different cultural background and religion. In an English–Spanish study all participating couples had already undergone PGD prior to the investigation because of cystic fibrosis, X-linked disorders or chromosomal disorders. The mostly Protestant or Catholic participants perceived the main advantage of PGD to be that only unaffected embryos were transferred into the mother's uterus and thus therapeutic pregnancy termination could be avoided (Lavery et al., 2002).

In Italy, attitudes and preferences to both methods, PD and PGD, were assessed in couples in which both partners were carriers of the β-thalassaemia gene. Two-thirds of the mostly Catholic couples (97%) came already for their second or further PGD, one-third had previously had selective abortion resulting from prenatal diagnosis. The willingness to undergo PGD was greatest in couples who had both previous experience with the procedure and with selective abortion (Chamayou et al., 1998).

A comprehensive list of advantages and disadvantages of PGD resulted from a study of 245 carriers of recessive orders including cystic fibrosis and spinal muscular atrophy. Two of the major advantages dealt with the fact that the child would not inherit the disorder (94%) and yet be genetically related to both parents (86%). The disadvantages were generally given lower ratings. The long waiting lists (60%), the dilemma of what to do with spare embryos (51%), the possibility that the treatment may not be successful (49%), and possible health risks (46%) were seen as the most important disadvantages (Snowdon & Green, 1997).

Case study: Lucas

Paul, a 46-year-old remarried business man, and Anna, 36, a lawyer, both successfully persuing a vocational career, faced an unexpected pregnancy. They had not planned to have children. In the beginning this surprising turn of their lives was kind of a shock but it did not last long. The couple soon became familiar with the idea of having a child and becoming a family and looked forward to it. Testing for Down's Syndrome due to advanced maternal age was rejected because the child was seen as a present, a gift from life, and abortion was absolutely unacceptable.

Shortly after his birth Lucas presented with 'floppiness', a muscular disease was suspected and finally the diagnosis of Werdnig Hoffman disease (WHD) was made. WHD is a rare, inherited progressive neuromuscular disorder characterized by muscle weakness, lack of motor development and poor muscle tone. Most children who fall into the severe category die before reaching the age of 2.

Lucas' life lasted little more than 4 months. Most of the time he lay relatively still with little or no movement, and only through his very awake eyes was he able to follow what happened around him. He developed difficulties sucking, swallowing and breathing, suffered from repeated respiratory infections and was near to suffocation several times. He spent many weeks in hospital but the endeavours of his parents along with the help of greatly committed health professionals enabled an intensive care-like setting at home, where Lucas eventually died.

During Lucas's lifetime his parents gave him the best of love and care. They enjoyed the time they had together and did everything that would relieve his suffering. According to earlier family-planning decisions and now being knowledgeable of their carrier status the parents were determined to not have a subsequent child. But the experience with Lucas, although on some level extremely painful, had enriched their lives to such an extent that they began to reconsider and to question their initial life plans. A few months after Lucas's death there were determined to have a second child but only if this would be free of WHD. By no means they would knowingly burden another person's life with all the pain and suffering Lucas had to go through.

The couple decided to undergo PGD which is not yet available in Austria. The first visit in a Dutch centre failed to result in a trustful relationship. A second try in a French centre proved successful and resulted in the first treatment cycle which, however, did not lead to pregnancy. While waiting for a second treatment cycle Anna became spontaneously pregnant. The couple then decided to undergo prenatal testing and that they would opt for selective abortion in case of an affected child. Prenatal diagnosis confirmed a baby without WHD. Now they are the happy and loving parents of a 2-year-old daughter who is free of the disease her brother died from.

Changes Lucas's parents went through

What made Lucas's parents change their mind several times? In the beginning they had not planned to have a family and they were against abortion for ethic and religious reasons. When they expected a baby they began to see the child as a gift from life or a heaven-sent blessing and this made them look forward to a new kind of life with the mother being determined to interrupt her vocational career for the next few years. Then they went with Lucas through months of extreme suffering and learned of the tortures a neuromuscular disease entails. In parallel, they experienced the joy of parenthood and a new dimension of love. Their relationship as a couple became even stronger. Based on these experiences, and after Lucas's death, they decided that they really wanted a child. In contrast to earlier attitudes towards embryo selection and selective pregnancy termination the parents put the child's wellbeing in the first place and concluded that their initial moral standpoints would not justify another person's disease. This viewpoint of Lucas's parents resembles that of adult patients with cystic fibrosis and their parents who stated that they would not wish any

future children or parents to have to relive their own experiences of cystic fibrosis (Conway, Allenby, & Pond, 1994).

Conclusions

Parents of children with life-threatening genetically determined diseases, who wish to have further children, are confronted with complex and emotionally fraught decisions. Often their initial views, firmly held, change over time. Contributing factors to the process of change include interactions between perception and interpretation of information provided, impact of the opinions of significant others, the experiences of the disease of the first child with chronic illness, and the nature of the parental and parent–child relationships.

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