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To know or not to know, disclosure of a newborn carrier screening test result for cystic fibrosis

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ABSTRACT

Purpose: Most newborn screening (NBS) strategies for Cystic Fibrosis (CF) also identify carriers. However, it is unclear if parents want to be informed about their child's carrier status or not.
Methods: Focus group discussions with pregnant couples to explore their opinions about disclosure of a carrier result for CF of their newborn.
Results: All ($n = 30$) wanted to be informed when newborn screening would show their newborn being a CF-carrier. Their main reason was the implication of this knowledge for further family planning. Other family members could be informed and children within the family could be tested. Parents stated they have the right to know, but others also expressed that the choice of not being informed should be offered as well.
Conclusion: Most parents want to be informed when NBS for CF reveals that their child is a CF-carrier, but the choice of not being informed should also be offered.

1. Introduction

Newborn screening programs for Cystic Fibrosis (CF) aims to identify newborns with CF. Newborn screening for CF (NBSCF) is expanding throughout the world. Screening programs vary but most use a combination of Immunoreactive trypsinogen (IRT) followed by either a second IRT at the age of 4–6 weeks or a DNA mutation analysis consisting of one or more CFTR mutations [1,2]. Using a DNA-based program also automatically leads to the identification of unaffected infants that carry one copy of an altered gene for CF; healthy carriers of the disease.

Although the main objective of neonatal screening is early detection and treatment, resulting in considerable health benefits, and not primarily the detection of carriers, informing parents about their newborn's carrier status can be beneficial. The main rationale for informing parents about a carrier test result is the immediate consequence for parent's future reproductive choices. A newborn carrier test result implies that at least one of the parents is also a carrier. A blood test can reveal if both parents are carriers and consequently are a couple at risk to give birth to a newborn with CF (25% chance). If it is confirmed that parents are a couple at risk, they have multiple possibilities to prevent the birth of a newborn with CF in case of a next pregnancy (primary prevention). Parents may decide not to have any more children, or they may consider prenatal and preimplantation diagnosis of CF [3,4]. Secondly, extended family members can be informed and may consider to test themselves before getting pregnant [4]. Finally, other previously born children in the family still asymptomatic may be discovered by sweat testing or DNA analysis [5].

However, some health professionals consider detection of healthy carriers undesirable because it may cause confusion and anxiety for the parents, and therefore lead to problems in the child–parent relationship and/or early stigmatization [6]. Therefore, carrier identification can be judged a major problem when implementing community-based screening.

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Few studies have explored parents' opinion about disclosing a newborn carrier test result [7]. In new strategies for NBSCF, carriers are found during the screening program but disclosure for carrier status is not necessary for the identification of CF patients [8]. Opinions differ whether or not this knowledge should be revealed to the parents.

The aim of this study was to evaluate the opinions and reasoning of future parents about whether or not they wish to be informed when NBSCF reveals that their child is a CF-carrier.

2. Methods

This study was part of a large study in the Netherlands investigating two novel strategies for NBSCF. In the second strategy in all samples with an IRT above $\geq$ 50 μg/l a CFTR-mutation analysis (36 mutations) was performed and when a single mutation was identified an extended DNA analysis was performed by sequencing the entire CFTR gene. The result of this protocol was positive only when two mutations were detected, results showing only one CFTR mutation were considered screen negative. Therefore parents were not informed when a child was found to be a CF carrier. However, parents were given the possibility to request this information, as was written down in the information leaflet. As part of this study focus groups were organized with future parents to take their opinions into account before implementing newborn screening for CF in the Netherlands.

2.1. Study population

Two focus groups were organized after an information meeting for expecting parents in a hospital setting (Atrium Medical Centre) inside the study region where newborn screening for CF was performed, the other two focus groups were organized connected with a pregnant women yoga class in Zoetermeer outside the study region. Participants were asked to join the focus groups, all participated voluntarily but received a small gift certificate afterwards.

2.2. Study design

We held dual moderator focus groups, one moderator focusing on the group process and discussion and the second moderator taking notes and documenting nonverbal communication and looking after the environment and logistics [9–11].

The discussion leader of the focus groups started with an explanation about the disease CF, the newborn screening program, how carriers were identified and the consequences of being a carrier. Then participants filled in a short questionnaire individually. Five questions tested if the participant had understood the information. Next, participants were asked for their opinions about seven statements. Those seven statements were discussed in more detail in a group discussion. When all participants indicated having the same opinion during the discussion, the discussion leader would introduce counter-arguments.

2.3. Analysis

All focus groups were audio-taped and transcribed. Data were analyzed by two researchers (AR who was also present at the group discussions, and AV), who independently identified key findings under certain themes. Results were discussed with a senior researcher (JD) and a psychologist-researcher, and also discussion leader of two of the focus groups (SP). Data were de-identified to protect participant confidentiality .

3. Results

3.1. Participants

In total 30 expecting mothers/fathers participated; 23 women and 7 men. Parents participated voluntarily, 8 expecting parents decided not to participate, the reason was not asked for. Most parents were highly educated (44.8% beyond high school and 6.9% completed university). About half of the parents was married (51.7%), the other half was living together (44.8% of which 41.4% registered). One couple had an LAT-relationship (Living-Apart-Together). Twenty-two (75.9%) were expecting their first child, four expected their second child (13.8%), and three parents expected their third, fourth or fifth child respectively (all $n = 1$). All participants were Dutch, except for one woman from Australia. The median age was 29 years, with a range from 21 to 46 years, and the median pregnancy duration was 29 weeks, range 23–36 weeks.

3.2. Knowledge-items in short questionnaire

The first knowledge statement was that "Cystic Fibrosis also is called mucoviscidosis (or "taaislijmziekte", a much used terminology in Dutch), because very viscous mucus is produced in different parts of the body". This item was correctly answered with "true" by 26/30 (86.7%) of the parents, 10% thought this was not true and 3.3% had no idea. Next, all parents correctly knew that a healthy person might be a carrier of CF. 90% (27/30) of the parents understood that at least one of them must be a carrier, if their child turned out to be to be a carrier. The same percentage of parents understood that the diagnosis of CF is not sure after a positive heel prick test result, and further tests are necessary. All but one participant remembered that parents were not informed about their child being a carrier during the study on NBSCF in 2008 and 2009. We therefore concluded that participants had understood the information in the introduction well and were sufficiently able to participate in a group discussion about this subject.

3.3. Group discussions

We divided the discussion into seven themes and compared the opinions of the participants during the focus groups with their individual responses on the short questionnaire completed prior to the group discussion.

3.3.1. *"Wanting to be informed about their newborn's carrier test result for CF"*

Most participants (25/30; 83%) wanted to be informed about their child being a carrier. Their arguments were that, although a carrier is healthy, the child may use this information when deciding to have children. Parents indicated they would be capable enough to inform their child so that their child could make an informed decision about preconceptional or prenatal screening.

> "I think, that we should know, also for the baby's future, to inform him/her (the baby) that (s)he is a carrier." "I think that if that information is known, your child has the right to be informed. On the other hand, (s)he (the child) did not ask for that information and the child is healthy, and therefore the child has no benefit. However, as the child finds a partner who may also be a carrier, and they have a baby with CF, I can imagine that (s)he will say, did you know? I think that when that information is known, the child has the right to hear it."