

Receiving the news of Down syndrome in the era of prenatal testing

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With recent developments in prenatal screening, more parents are expected to receive a Down syndrome diagnosis before the birth of their child, which can involve complex decision-making for many. To

understand what such a diagnosis means for parents who decide to continue their pregnancy and prepare for a child with Down syndrome (DS), the Dutch Down Syndrome Foundation surveyed 212 parents of children with DS born between 2010 and 2016.

The aim of this study was to describe their prenatal and postnatal experiences and better understand the way in which they had been informed about a possible and/or definitive diagnosis of DS in their [child](#). During the study period, noninvasive [prenatal screening](#) (NIPS) was available as part of a nationwide program, offered as a secondary test after high-risk first-trimester combined testing (ftCT) result, from April 2014 onwards.

Results show that:

- The majority of the mothers in our study received the indication for/diagnosis of DS after their child was born, reflecting the relatively low uptake of prenatal [screening](#) for DS in the Netherlands during the study period (2010-2016). However, this finding also reflects the fact that the majority of cases where a prenatal diagnosis of DS was made was followed by an elective termination of pregnancy.
- 24% of mothers of a child with DS had had some testing (of which 9% had invasive testing and 13% a false negative ftCT result)
- 76% of [women](#) had chosen not to have prenatal testing, giving, on average, 3.8 reasons per mother for this decision: 81% gave at least one value-related reason such as "a child with DS is welcome in our family"; 34% gave at least one test-related reason such as "the fCT is not a good screening test"; 36% gave at least one information related reason such as "I assumed that in my case the chance for a child with Down syndrome was small." Only 1% mentioned cost as a reason.

- The mothers that were offered ftCT, 54% recalled that during the counseling of ftCT, no information regarding DS was provided. Only 17% of the women received information they considered sufficient both in terms of medical information and about living with a child with DS.
- 3-26% of women judged the information received at possible or final diagnosis as completely insufficient; more women considered information provided prenatally as insufficient as compared to postnatal received information.

The study shows that most of the mothers of a child with DS born in the Netherlands between 2010 and 2016 have consciously chosen not to have prenatal testing. This may be considered as an informed choice, as the majority of these mothers felt that a child with DS was welcome in their family. Some mothers decided not to have testing because of the poor quality of the available screening [test](#) or they did not consider themselves to be at risk, which might reflect a lack of information. More than half of the women did not recall having received information on DS at all prior to choosing whether or not to have ftCT, which might as well indicate that the information provided at that moment was incomplete.

When receiving the diagnosis, not only is the information important, but the perceived atmosphere is crucial on how the conversation will be judged. The atmosphere is determined by level of empathy. Terms as "respect, warmth, and support" were used for positive-rated conversations, while words as "hasty, negative, cool, and distant" were used for negative-rated conversations.

From the results of this study, satisfaction was highest among those [mothers](#) who had received a confirmed postnatal [diagnosis](#) and lowest among women receiving a prenatal screening result indicative for DS, potentially related to the available options and difficult choices being discussed. As presented in this study, parents continuing their pregnancy

prefer to receive positive information on DS, focused on the future. Apparently, this is more likely to be discussed in postnatal situations.

Discussing prenatal options in a balanced way can be challenging, particularly when parents already have an unspoken preference toward one pregnancy option, as reflected in the results of this study. Careful exploration of preferences seems to be crucial, and adequate use and compliance to existing guidelines would be helpful.

More information: Neeltje MTH Crombag et al, Receiving the news of Down syndrome in the era of prenatal testing, *American Journal of Medical Genetics Part A* (2019). [DOI: 10.1002/ajmg.a.61438](https://doi.org/10.1002/ajmg.a.61438)

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