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Genetic testing and counseling in the case of an autism diagnosis: A caregivers perspective

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ABSTRACT

The search for genes that can explain the development of autism is ongoing. At the same time, genetic counselling and genetic testing can be offered to families with a child diagnosed with autism. However, given the complexity of autism, both with respect to its aetiology as well as with respect to its heterogeneity, such genetic counselling and testing raises specific ethical questions regarding the aim and scope. In order to map these questions and opinions we interviewed 15 Belgian autism professionals. We found that they believed that genetic counselling and genetic testing have certain benefits for families confronted with an autism diagnosis, but also that direct benefit to the child is limited to those cases where a genetic finding offers a certain prognosis and intervention plan. In cases where autism is the result of a syndrome or a known genetic variant that is associated with other health problems, detection can also enable prevention of these health issues. Benefits of genetic testing, such as relief of guilt and reproductive choice, are primarily benefits to the parents, although indirectly they may affect the wellbeing of the person diagnosed. These benefits are associated with ethical questions.

1. Introduction

Autism Spectrum Disorder (ASD) is a condition, or a group of conditions, diagnosed through behavioural observation and —ideally- after assessment of a multidisciplinary team. The DSM-V describes the following characteristics: persistent deficits in social communication and social interaction across multiple contexts, restricted, repetitive patterns of behaviour, interests, or activities, and qualitative impairments in communication (American Psychiatric Association, 2013). These characteristics, as stated in DSM-V, must have developed in early developmental periods, must hamper social functioning and may not be better explained by, for example, intellectual disability (ID). Specific behaviors may be more or less present in different individuals, hence the current assumption that autism covers a 'spectrum' of phenotypes, and possibly also a spectrum of underlying causes. Due to the association of autism with certain known syndromes, such as fragile-X and tuberous sclerosis, the concordance in monozygotic twins and the fact that autism is often familial, it is now accepted that autism has at least partly a genetic cause, although environmental factors have been identified (Herbert, 2010).

Genetic counselling has been defined as a communication process dealing with the occurrence, or the risk of occurrence, of a genetic disorder in the family, and the possibility of performing a genetic test based on this information (Fraser, 1974). Given the complexity of the aetiology of autism, genetic counselling for autism is especially difficult. After a diagnosis of autism, often the proband or her parents is offered the possibility of genetic counselling and, if considered useful, genetic testing. Recent discoveries of autism genes enable, next to an empirical risk calculation based on family history, now also estimations based on test results (Gershon and Alliey-Rodriguez, 2013). In general, a positive genetic test of the proband for a certain condition can help explain the causes of a condition and may facilitate an accurate prognosis or an accurate treatment plan. With autism, only in a subset of cases will the outcome of the test straightforwardly point in the direction of an action plan, such as when a clear genetic syndrome is found (e.g. Fragile-X), or when a known genetic variant associated with autism is found. Indeed, given the complexity of autism genetics and the large phenotypic and genetic heterogeneity, genetic counselling to help families understand autism genetics and inheritance patterns

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is complex (McGrath et al., 2009; Mercer et al., 2006). At the moment, some authors claim that a genetic variant that offers a (partial) explanation for the cause of autism can be found in 40% of the persons with autism (Jeste and Geschwind, 2014). Of this 40%, 5–10% are monogenic causes (such as Fragile-X or SHANK variants) and 20–30% are genetic risk variants. One of the aims of genetic counselling is to help prospective parents and probands understand the risk that their (next) child has the same condition, and, based on that information, to help them decide to stop having children, to continue reproducing naturally or to use certain reproductive techniques such as Preimplantation Genetic Diagnosis (PGD) or Prenatal Diagnosis (PD). Given autism's etiological complexity, genetic counselling, especially in the light of reproductive choice, can be difficult. But the complexity of autism is not only related to its etiology. Many autistic individuals themselves have argued that they consider their autism not as a disorder or disease but as a variant of normality that has its own value. Thus they challenge the assumption that autism should be avoided or that genetic tests should be used to prevent the birth of autistic children (Jaarsma and Welin, 2012). Given this complexity, both in the etiology as well as in the conception of autism, we considered it useful to query values and opinions of Belgian autism professionals.

2. Methodology

We interviewed 15 Belgian professionals, who were selected based on the fact that, within their specific discipline (clinical genetics, psychiatry, neurology, psychology or educational science), they have autism as their primary expertise. This means that their patients or clients are primarily people with autism or parents of people with autism. The vast majority of the patient population of the professionals are children and their parents, although many of them see the children until well into adulthood. Two of the respondents, both educational scientists in a non-academic setting, were selected because their clients are children with autism and intellectual disability. All other respondents have clients or patients with autism, with or without intellectual disability. After a literature review, which we have published elsewhere (Hens et al., 2016a), we designed a topic-list. This topic list included topics on genetic research, clinical genetic testing and counselling and reproductive choice. In this paper we present our findings with regard to genetic counselling and testing, and reproductive choice. The findings with regard to genetic research were published in an earlier issue of this journal (Hens et al., 2016b). HP and KH made a list of 12 possible interviewees, making sure also to include also professionals with experience with autistic children with ID. Three extra respondents were added after the interviews were ongoing, based on the suggestions of some of the interviewees. An overview of the different professions of the respondents can be found in Table 1. KH used a semi-structured interview schedule, representing the topics from the topic list. Using such loose schedule promotes an interview in which respondents are encouraged to talk at length about their opinions and experiences, and allows for in depth exploration of certain topics according to the answers that are provided. The interviews took between 30 min and 70 min, and were transcribed verbatim by KH. They were then coded using NVIVO 10 software. We did not use an initial list of codes. In a first round of open coding we extracted and coded all pieces of text that were relevant for questions related to genetic testing and counselling into several subthemes. In the next two rounds of coding, the axial coding and the selective coding, we connected the different subthemes into broader categories. KH did the initial rounds of coding, the subsequent results and story were agreed upon by KH, HP and KD. The study was reviewed and approved by the KU Leuven Social and Societal Ethics Committee (file number G-2014 12 112).

KH conducted the interviews in Dutch, the native language of all interviewees, and translated selected quotes for this paper. Hence, some of the nuances may be lost in translation. We used *she* to denote all respondents in this paper, to ensure anonymity of the respondents. We will use both the terms “autistic persons” and “persons with autism”, as some individuals may prefer the former or others the latter (Kenny et al., 2015).

Table 1
Information about the interviewees.

		#
Profession	Psychologist	3
	Clinical geneticist	2
	Educational specialist	5
	Pediatrician	1
	Child neurologist	2
	Child psychiatrist	2
Setting	Academic	12
	Non-academic	3
Gender	Female	7
	Male	8

3. Results

We found that overall the respondents thought genetic counselling and testing could be useful after an ASD diagnosis, either for all subjects diagnosed, or for those with ID or with a familial history of ASD. However, respondents also acknowledged that only in a subset of cases there would be benefits for diagnosed children, especially if a known variant with a known action plan and prognosis was found. Other benefits include offering parents an explanation for their child's autism, although genetic knowledge could also induce guilt and blame, and the possibility of offering reproductive choice to parents. The use of reproductive technologies such as preimplantation genetic diagnosis and prenatal testing were thought to be only acceptable if there were additional factors that would decrease quality of life or whether the burden to parents of having an (extra) child with autism would be too high. An overview of categories and themes can be found in Table 2.

3.1. Small risks, big gains?

Genetic counselling for autism consists of explaining the genetics of autism to parents of a child with an autism diagnosis, or in a subset of cases, to an autistic adult, and give them information about recurrence risks. The process includes discussion of diagnostic testing, interpretation of genetic test results, assistance in coping with the diagnosis, discussion of medical treatments and prognosis, and subspecialty referrals. If deemed appropriate, a genetic test can be done on the proband and often also the parents and/or other family members to check if any known genetic or chromosomal aberration is possibly the cause of the autistic behaviour.

A first question that was explored during the interviews was whether all those who have received a diagnosis of autism for themselves or for their children should be referred to a genetic counsellor. Respondents did acknowledge that it would be useful that a clinical geneticist or a genetic counsellor would explain the genetics of autism, and the limits of current knowledge. In the following quote, a clinical geneticist states:

> *I think you should make a distinction between, a part of the counselling is explaining what we can and do know, and what we do not know. And that is … yes, that does not always mean that we*