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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.

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 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.

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 (Pamela Sankar and Nora L. Jones, 2007). The interviews took between 30 minutes and 70 minutes, 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

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.

Table 2: Overview of categories and themes

Categories	Themes
Risks and gains	Explaining genetics Genetic testing Direct benefits
Relieving and inducing guilt	Difference de novo – inherited Guilt & blame Autism traits in undiagnosed parents
Acceptability of reproductive technologies	Reproductive choice Acceptability of technologies Comorbidities & quality of life
Quality of life versus burden on family	Burden of caring for disabled child Capacity of families Role of ethics committee

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 have to take blood and test all sorts of things.
(clinical geneticist #1)*

So she acknowledges that parents of children with a diagnosis of autism, and autistic individuals themselves, should have access to a professional who could explain the genetics of autism. However, this does not necessarily mean that in all cases a genetic test should be performed. She then continues by stating for which probands such a test may be useful:

Children for whom this is very meaningful are those with an additional intellectual disability or physical problem, if you suspect it might be a syndrome. And then the other group, those with an obvious familial component. (clinical geneticist #1)

Several respondents named these two cases, autism with comorbidities such as ID and familial autism as cases in which genetic testing would be useful. The reason they gave is first that in those cases, chances were higher that something genetic would be found. Indeed, respondents also thought that in sporadic occurrences of autism without ID, genetic testing would not be useful, because they assumed that in these cases no genetic explanation would be found, and so it would not be relevant for treatment plans or be useful for reproductive planning.

The possibility of finding genetic aberrations associated with syndromes was named as an explicit advantage of genetic testing in case of ID or physical comorbidities. One respondent stated that “sometimes something clearly genetic is found, of which you think this can actually means something for the genetic counselling of the parents in short time. For example, sometimes Fragile-X is found.” (pediatrician). Indeed, a subset of respondents thought genetic testing of children with autism and ID would be useful in all cases, to pick out that subset of individuals who have a very specific known genetic variant, as is exemplified by the following quote:

I think that each child that has a diagnosis of autism should be genetically tested, because you want to rule out certain things. Especially those genetic things that convey a heightened risk for comorbidities next to autism, and you can only effectively do so by starting up those tests. (clinical geneticist #2)

So this respondent, although she admitted further that only in a minor set of cases would the genetic findings be useful for further action benefitting the child, stated that for those children where something is found, genetic testing *is* useful. Hence, the idea here is that, although not many children will benefit directly from genetic testing, as variants associated with a syndrome are rarely found, those who have a genetic variant that is associated with a specific phenotype and a specific therapy will benefit significantly, and this warrants testing.

Relieving and inducing guilt

Next to benefits for the children, respondents also thought that genetic counselling and testing would convey benefits to the parents, as it would give a biological explanation for the autism and relieve them of feelings of guilt: "I think it will bring comfort to many parents, that they know they cannot help it." (educational specialist #2). But, respondents stated, this relief of guilt becomes problematic if genetic mutations are found that are familial in nature, rather than de novo:

The advantage of looking for a gene, it can mean a lot for these people if they are given a cause, or a reason why something goes wrong, a reassurance that it is not the way they raise the child, or because they have smoked or drank, the question of who is to blame. That is the positive side of the story. Except if you are dealing with a condition with an inherited susceptibility. (educational specialist #1)

So rather than to relieve guilt by giving a biological explanation -what people originally believed to be the benefit of genetic testing- finding a genetic cause may in some cases induce guilt and may actually interfere with family relations. A subset of respondents believed that familial genetic findings would disturb family relations, especially those that were already under much strain due to the challenges and subsequent diagnosis of their child. Some respondents stated that people often already had some ideas of 'where it came from' based on character traits of a parent or other family member, and that genetic findings may confirm that:

I see that often in families, that parents will apply their informal genetics. Your dad, he is also really strange, and such things, even without there being genetic research people will.. they know that it is genetic so it they will be looking in the family, who is a bit odd. That is how it goes. But yes than the genetic testing will be a confirmation of the hypothesis of course. (psychologist #1)

Hence, one respondent clearly stated that, unlike for cases where there is a clear indication to suspect a 'syndromic' cause of a child's autism, testing in case of autism when there are other cases of autism in the family, or autistic traits, is more complex with regard to counselling: 'what do you do in a family where the father has autistic traits and the other children also have autistic traits'. She also states that she used to suggest doing a microarray on all children with a diagnosis of autism, but that she came to find this problematic:

[...] for example, a father has [autism] traits, the mother not at all and the child has an obvious autism spectrum disorder. I perform an array on the child, it shows a deletion that can be a susceptibility factor. Then we have a problem. Because, I

discussed it in advance with those people, because you are almost sure that if we look at the parents it will be passed on from the father, and in this case that was so, in this case it was OK because we really explained beforehand, but I think you must explain very well in advance, otherwise there is the possibility to induce blame, look it is from your side, and it still is a bit difficult for that father because, yes, my life is not worth less than that of my wife who doesn't have it. (child neurologist #1)

In this quote, several issues related to the often familial nature of autism are brought up. First, there is the danger that the parent in which the genetic variant originated feels guilty or is blamed by the other parent for passing on the condition. Second, there is the fact that a parent may also have an autistic phenotype, but is potentially happy with his or her life. Indeed, whether having such phenotype, especially in milder cases, leads to dysfunctioning and hence to an ASD diagnosis is not only related to the severity of the phenotype itself, but also to other external factors. Moreover, as genetic variants are in most cases susceptibility factors, a certain genetic finding can lead to different levels of severity in different family members. What it *means* to share a genetic finding with one's autistic child for a parent is as of yet poorly understood. One of the respondents explicitly named the necessity for genetic counselors and all those who discuss genetics with families with a child with autism, or to autistic individuals themselves, to admit the paucity of knowledge we still have: "I think we really should be aware of the fact that we are sorcerer's apprentices and that very little is known and that we cannot draw grand conclusions with regard to genetic counselling, except for the group with clear syndromes." (psychiatrist #1)

Reproducing autism: The acceptability of reproductive technologies

An important aspect of genetic counselling is to inform people about the recurrence risk should they decide to have another child. If a genetic factor that contributes sufficiently to the autism in their child has been found, in principle, prospective parents have the choice either to accept the possibility that their next child will also have autism, not to reproduce further, or to opt for Preimplantation Genetic Diagnosis (PGD) or Prenatal Diagnosis (PD). Respondents overall did not consider autism as a determining factor in the choice between embryo selection and prenatal diagnosis, and those who considered these two options ethically different named burden for the woman (hormonal stimulation versus termination of pregnancy) or the value people would attribute to the moral status of the embryo as factors that would influence that decision.

The acceptability of using reproductive technologies and prenatal genetic testing was for most of our respondents linked with the question whether the genetic variant was related also to ID or to

comorbidities that would influence *quality of life* (QOL). One respondent was categorically against using such techniques to detect embryos and fetuses carrying autism genes, to prevent the birth of individuals with autistic traits, regardless of whether they had intellectual disability or not:

(...) yes, we are still dealing here with people who have the potential to function in a society, albeit with difficulties, but there are certainly examples of people who can live a fulfilling life (psychologist #2).

So this respondent, who clearly stated that selection against autism was not acceptable, bases this opinion on the fact that they can have quality of life and function in society. Hence, for this respondent, the difficulties people with autism may experience is not enough to endorse, as a society, the use of these techniques to prevent the birth of these people.

Respondents also considered the fact that it is difficult to predict, based on genetic findings, what would be the quality of life for a specific child. As one respondent stated:

[...] the point is, how many suffering and dysfunction there will be, that does not completely correlate with autism. There are children with autism who function better than children without autism and without any developmental delay" (psychologist #3).

Hence, this correspondent stresses that the finding of a genetic mutation coding for autism in a child is not sufficient to predict wellbeing or the potential to function well later in life. Related, a subset of respondents also mentioned the fact that it may be very difficult or impossible to predict the severity of the phenotype based on genotype alone, although some respondents believed that there are specific genetic variants for which one could be sure that the phenotype would include intellectual disability, for example in the syndromic variants of autism. But, as one respondent stated 'Autism itself is very variable, it is the comorbidities that will be the defining factors.' (clinical geneticist #2). Hence, this respondent emphasized the idea that autism in itself is not a reason enough to allow the use of prenatal or preimplantation diagnosis to prevent the birth of children with an ASD phenotype.

Quality of life versus burden to family

Especially those respondents who were in everyday contact with children with autism with severe ID acknowledged that raising such a child could be very *burdensome*, and that preimplantation and prenatal screening should be allowed:

I do not know where to draw the line, only the two extremes are clear, those with normal intelligence, yes that would bother me if they would do PGD for that, the

severely intellectually disabled, those who dysfunction completely, I would think it is normal [that the parents have access to PGD] (child psychiatrist #2)

For the group of children with severe ID, the rationale behind allowing parents access to PGD was, on the one hand, the fact that the quality of life of these children was considered very low. One respondent, who works on a daily basis with children on that side of the spectrum stated that they were often very scared and overwhelmed by the world surrounding them. But on the other hand, whether people would be entitled to such technologies or whether they were given this option would not only be based on how severe the phenotype associated with the familial genetic variant was, but also with the capacity of the family to deal with the birth of (another) child with the same problem, as the following respondent states:

Happiness is... a child with autism and severe developmental delays, who has siblings without autism, you could say that the child with autism is the most happy. A child that has multiple severe disabilities, of who you can say here it is clear, terminate a pregnancy, because nobody can cope with that, that doesn't mean that the child herself cannot be completely happy. If you have a developmental level of three months, you can be happy. But it is about other things, it is about being supported, often there is no network to support these families (educational specialist #3)

In this and other quotes this respondent states that it is not the happiness or quality of life of the child itself alone that would be a factor to allow technologies to screen out autistic individuals, but also the *burden* it would put on the family, including brothers and sisters. She also acknowledged that in our present-day society, which she considered to be very individualistic, there is a lack of network to support families with children with special needs, and this increases the burden of caring for such children. Another respondent acknowledged that the burden that specific families could cope with would be different for each family and that this is something an ethics committee should consider when evaluating such request:

But also, I do not know if this exists already, that they also look at these kind of things, what is the capacity of this family, what kind of support do they have, how are they dealing at the moment with their children with autism. Some families have three children with autism, they are often coping reasonably or even very well. Surviving as a family, happy moments together... They have of course more difficult moments than other families, but in some families there is only one child

with autism and the family falls apart. And that doesn't mean that in that case the autism is more or less severe. (psychologist #1)

Opinions were divided about whether *ethics committees* could help in deciding when to allow to the parents the use of reproductive techniques to avoid autism or not. Although many respondents thought that requests for prenatal or preimplantation diagnosis for autism should be assessed by ethics committees, others thought that the parents themselves were capable of making the decision, as demonstrated by the following quote:

Yes because... an ethical committee it doesn't really sound appropriate to the problems we are dealing with here, it is something interpersonal, I believe in the power of a human being to take decisions for themselves, also in difficult cases. That there should be another person warning them and telling, 'think carefully', after they have decided.. no, that is not possible. (educational specialist #2)

This respondent, who was in everyday contact with children with severe ID, believed these parents have the capacity to decide for themselves, and that ethics committees would only get in the way of that.

Discussion

Genetic counselling and testing: who is tested?

When parents or probands first receive a diagnosis of autism, the primary point of contact is often a psychiatrist or child neurologist, not a genetic counselor. A first question to be answered is who should be sent to see a genetic counselor and who is applicable for further testing. Should autism professionals already make a first triage based on criteria such as severity of the phenotype or whether there are other family members affected? Is it worthwhile for anybody confronted with a diagnosis of autism to talk to a genetic counselor and to receive information regarding expectations, limitations and complexities related to the genetic testing in the case of autism? Our respondents did not tackle this question. However, with regard to genetic *testing* the general opinion was that this was useful in cases where there were comorbidities such as intellectual disabilities and in familial cases. 'Familial' can mean that another family member has a genetic variant associated with autism. It can also mean that other family members have a diagnosis, or that they exhibit autistic trait, distinctions which were not made by our respondents. Also, it seems farfetched to genetically test even the autistic child with normal intelligence who is able to function in standard education with some support. The fact that in many cases, the ones who request these tests will be parents who want to be able to make more informed reproductive choices complicates the matter. Ethicists have

claimed that children have the right not to know their genetic makeup until they reach the age when they can decide for themselves whether they want to know this, unless genetic information helps to make treatment decisions and is beneficial to the child (Davis, 2010; Hens et al., 2013). How this right should be matched against the rights of parents to have information that influences their reproductive decision-making is unclear, especially in the case of 'mild' autism, where children will potentially grow up to be autonomous adults.

Advantages and disadvantages of genetic testing

Ethical issues related to genetic testing of minors were not explicitly mentioned by our respondents. Professionals named several advantages for genetic counselling and genetic testing. They stressed the possibility that genetic tests allow the detection or ruling out of so-called monogenic forms of autism. Thus they confirm some of the aims described in the literature. For example, authors state that certain monogenic or syndromic forms have a specific plan of action, and prepare parents better for a child's challenges, and perhaps begin behavioral interventions at an early age. (McMahon et al., 2006; Pellicano and Stears, 2011; Scherer and Dawson, 2011). Other benefits that were not named by our respondents include natural history studies of specific genetic etiology, connecting with other families having children with same genetic condition and contacting support groups.

Another reason why genetic counselling and testing are useful after an autism diagnosis in a child, as mentioned in the literature, is that parents want to know the cause of their child's autism (Scherer and Dawson, 2011). This is also confirmed by our interviewees who stated that knowing that the cause of their child's autism is in her genes can relieve parents of guilt. This is especially so if the genetic variant has occurred *de novo* (Jordan and Tsai, 2010; Marchant and Robert, 2008; Rossi et al., 2013; Scherer and Dawson, 2011). However, in case of an *inherited* variant, our respondents and some authors have argued that rather than taking away guilt, knowledge of who contributed the genes that lead to autism may induce guilt in the parent who carries the variant (Marchant and Robert, 2008; McMahon et al., 2006), and can lead to stigmatization and potential conflicts within families (Gershon and Alliey-Rodriguez, 2013).

Such mechanisms also play a role when families start to apply 'their informal genetics' as one respondent stated. Indeed, family members who may share some autistic traits but who were never diagnosed can be confronted with genetic information that may or may not suggest that they have the same phenotype as their child. An autism diagnosis should, according to its definition in the DSM-V, only be given if the characteristics of the person lead to problems in functioning in everyday life (American Psychiatric Association, 2013). Finding genetic variations in undiagnosed parents, who may share behavioral characteristics with their children, may seem to suggest that testing for autism

can already be done on a biological basis, which is premature and as of yet counter to the definition of what *is* autism. Counselors should therefore be careful, when confronted with this situation, in explaining the meaning of genetic information, but also what it means to have a behavioral diagnosis.

Reproductive issues

A third and core aim of genetic counselling as identified by respondents is to allow parents with the wish to have another child to understand for themselves what the chances are that their next child will have the same phenotype, or, in cases when a specific variant can be found, to give them access to reproductive technologies to avoid the birth of a such a child through Preimplantation Genetic Diagnosis (PGD) or Prenatal Diagnosis (PN), an option confirmed by several authors (Amor and Cameron, 2008; Daley et al., 2013; Gershon and Alliey-Rodriguez, 2013; Jaarsma and Welin, 2013; Marchant and Robert, 2008; McMahon et al., 2006; Pellicano and Stears, 2011; Rossi et al., 2013; Walsh, 2010). Whether or not such technologies would be acceptable for autism depended for many respondents on the specific phenotype associated with the genetic variant. Indeed, respondents referred to potential comorbidities associated with a genetic variant, such as ID etc., that affect quality of life of persons with autism, but many also believed that autistic traits alone may not be sufficient to warrant such technologies. Hence, they expressed the opinion that people with autism may have a valuable life and a high enough quality of life. In fact some of them expressed concern that the desire to rule out autism as such may be in some cases an expression of wanting a perfect child, rather than to avoid suffering. This seems to be the opinion of some parents of autistic children themselves: in an interview study, Tabor et al found that parents of autistic children are concerned about the fact that genetic information is used for reproductive planning, and that this may potentially harm their children, other children and society at large (Tabor et al., 2011). Moreover, as one respondent stated, intellectual disability in itself may be not be a good indicator for quality of life. Indeed, whether quality of life and the harm to potential offspring can ever be used to defend reproductive technologies, to define on which grounds to allow them, and which threshold of severity to use is a discussion at the core of reproductive ethics (Smajdor, 2014).

An issue left untouched in our interviews is how genetic tests influence reproductive decisions of the children themselves. Indeed, some of the children tested may grow up to be adults desiring to reproduce themselves. The fact that they will know that they carry a genetic variant associated with their autism may be beneficial, in that they may themselves consider using reproductive technologies to avoid transmitting autism. However, autism as a condition is tightly linked with one's identity, and to some adults with autism it is even a valuable or inseparable part of their identity. How these

adults, when reaching reproductive age, will feel about the genetic knowledge that was acquired when they were children is largely uncharted territory.

Some respondents have named the *capacity* of parents of raising (another) child with the same phenotype as a point to consider whether to allow PGD or PD for specific autism genes. Along the same line, Gershon and Alliey-Rodriguez argue that the acceptability of such choice depends on the specific history of the family (Gershon and Alliey-Rodriguez, 2013). There is uncertainty as to who should decide on whether to allow such request: some respondents argued very clearly that parents themselves, with their experience, are in the best position of deciding whether to opt for this or not, whereas others thought that ethic committees or guidelines could help. Also, as some respondents mentioned, a family's capacity of accepting another child with developmental problems depends heavily on the support or network they have, for example, whether close relatives are willing to help out. But also society at large influences capacity, and factors such as the availability of education or institutionalized care for the person when she grows up. Hence, the inclusion of the criterion of capacity to assess the acceptability of a termination of pregnancy or PGD in certain cases may on the one hand reflect a desire to care for and help specific families in need, but on the other hand there is the risk that collective responsibilities will too easily be bypassed. But an assessment of familial capacity on criteria related solely to intrinsic characteristics of that family is unrealistic as well, as it is almost impossible to isolate intrinsic and extrinsic factors. Giving people access to reproductive technologies if their capacity to support children with certain challenges has been exceeded should, in any case, go hand in hand with enabling a society that increases this capacity by providing facilities for such children.

Genetic counselling and genetic testing has certain benefits for families confronted with an autism diagnosis. 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 of this syndrome can enable prevention of these health issues. Relief of guilt and the possibility of reproductive choice are primarily benefits to the parents, although indirectly they may also affect the wellbeing of the person diagnosed. Moreover, more and more autistic people see the fact that ASD has a biological basis as a proof that that autism is a healthy genetic variation (Jaarsma and Welin, 2012). Indeed, whether or not a difference can be attributed to genetic variation or genes does not convey any proof that it is a disease or disability.

Limitations

Our study has several limitations. We have queried 15 Belgian autism professionals, who are at different stages involved in the diagnostic process. We chose this approach, rather than just querying genetic counselors or clinical geneticists, because in Belgium, child psychiatrists and psychologists often are the primary contact after initial ASS diagnosis. They will often be the first ones to discuss the possibility to also consult a genetic counselor with parents or autistic adults. Also, the vast majority of the referrals for genetic testing will be done for children, and, as became clear from the interviews, for children with a more severe phenotype. Hence, most of the respondents answered the questions with that population in mind. Although some respondents explicitly made the difference between autism with or without ID, for example in the discussion on reproductive technologies, answers may be biased towards the cases with ID. A follow-up study focusing on the issues related to the autistic child without ID seems warranted to filter specific ethical questions out. The issue of how to counsel the autistic adolescent or adult with normal or high intelligence, who desires to gain more insight in biological mechanisms behind her condition, also raises a number of different ethical questions which we have only hinted at in our study, and which warrant further investigation. As we only interviewed Belgian professionals, the answers are influenced by the local situation: in Belgium, genetic testing and (specialized) genetic counselling is reimbursed by the social security system and done at the eight official Belgian Centres for Human Genetics. We used a qualitative method, which allows us to distill important themes and categories, but which is less suited for comparison between the different individuals and professions. Hence, we consider the questions raised in our study as the basis for further studies, both qualitative and quantitative, with genetic counselors and clinical geneticists in Europe. For example, a large number of people from same profession (e.g. only psychologists or only geneticists or only pediatricians) could be queried and their opinions could be compared. Also, we believe that more research is needed that involves adult autistic people to learn how genetic knowledge about themselves that was gathered when they were minors influences their lives and whether they think such knowledge is always helpful or beneficial. Indeed, a comprehensive approach querying the opinion of different stakeholders may help answering ethical and practical questions regarding genetic testing and counselling for autism.

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