Check for updates

Received: 19 July 2023 | Revised: 16 October 2023 | Accepted: 18 November 2023

DOI: 10.1111/cch.13212

RESEARCH ARTICLE

WILEY

Parents' views of benefits and limitations of receiving genetic diagnoses for their offspring

Robert Klitzman[1] | Ekaterina Bezborodko[2] | Wendy K. Chung[3] | Paul S. Appelbaum[4]

[1]Psychiatry and Director of the Masters of Bioethics Program; Department of Psychiatry, Columbia University, New York, New York, USA

[2]Department of Law Ethics and Psychiatry, Columbia University, New York, New York, USA

[3]Department of Pediatrics, Boston Children's Hospital, Harvard Medical School, Boston, Massachusetts, USA

[4]Psychiatry, Medicine and Law, Center for Law, Ethics and Psychiatry, Columbia University Irving Medical Center, New York, New York, USA

Correspondence
Robert Klitzman, MD, Professor of Psychiatry, Director, Masters of Bioethics Program, Vagelos College of Physicians and Surgeons, Joseph L. Mailman School of Public Health, Columbia University, 1051 Riverside Drive; Mail Unit #15, New York, NY 10032, USA.
Email: rlk2@cumc.columbia.edu

Funding information
The research was supported by funding from the National Human Genome Research Institute (NHGRI), Center for Excellence in ELSI Research (CEER) grant program, RM1HG007257 (Paul Appelbaum, PI) and NICDH Autism Center of Excellence grant P50HD109879 (Wendy Chung, PI) and SPARK: Simons Powering Autism Research.

Abstract

Background: Individuals with autism with intellectual disabilities (ID) are increasingly undergoing genetic testing, posing questions of how parents view/respond to such results.

Methods: Twenty-eight parents whose offspring had received genetic diagnoses of de novo pathogenic variants associated with autism were interviewed.

Results: Genetic diagnoses parents receive concerning their offspring's autism/intellectual disabilities can be 'double-edged' in several ways, having advantages, but also certain disadvantages and limitations. Benefits were medical/scientific (e.g., ending diagnostic odysseys and potentially contributing to research), emotional (e.g., relief, peace of mind and less self-blame), cognitive, social (e.g., validation and confirmation with others – it's 'not just in our head') and financial (e.g., estate planning), now and in the future (e.g., preparing for possible future symptoms, development and availability of treatment and setting realistic expectations). Limitations included a lack of medical treatments related to the genetic diagnosis, a sense of finality and heightened uncertainties which can increase anxieties (e.g., concerning additional symptoms associated with genetic diagnoses and offspring's ability to live independently in the future). Overall, parents were glad to receive the results, seeing the pros outweighing the cons. Parents responded to these tensions in various ways, having mixed feelings, recognizing the trade-offs and/or focusing on their offspring's present needs. Factors such as age of offspring at receipt of genetic diagnosis, parent's scientific background and prior views and degrees of self-blame affected these responses.

Conclusions: These data, the first to examine how parents perceive genetic diagnoses received for offspring with autism and ID through whole exome/whole genome sequencing, highlight practical medical and psychological benefits as well as limitations. These findings thus have important implications for future education, engagement of families and research. Providers should be aware of these issues, to inform and assist families, who are considering such testing, about these potential pros and cons and responses.

Permission to Reproduce Material From Other Sources: Not applicable to this paper.

© 2023 John Wiley & Sons Ltd.

KEYWORDS
anxiety, autism, decision-making, genetics, parents, patient attitudes, psychology

1 | INTRODUCTION

Professional guidelines have recently recommended that the evaluation of individuals with autism and/or intellectual disabilities (ID) include genetic testing, which most parents support (Johannessen et al., 2022; Srivastava et al., 2019). Critical questions arise regarding how stakeholders will view and respond to such results. One such individual wrote an Op-ed for CNN entitled, 'I now know what caused my autism, which changes everything – and nothing (Gravino, 2021)', in which she discussed her own personal responses. However critical questions arise of whether parents of children with autism would feel similarly or differently receiving genetic test results through whole-exome/whole-genome sequencing (WES/WGS).

Previous studies have probed parents' reactions to genetic testing for autism, yet used chromosome microarrays (CMA), included only small samples, and mixed together parents of children who had not and had received diagnoses or who had variants of unknown significance. For instance, among 21 parents of offspring with autism and/or congenital abnormalities, eight received pathogenic or likely pathogenic results, seven received a variant of uncertain significance and six had benign results (Hayeems et al., 2016). Parents as a whole perceived 'personal utility' of testing, but at times had 'inflated' expectations (e.g., that a cure was a 'short step away') (Hayeems et al., 2016). Among 57 children with autism undergoing CMA testing, six received pathogenic results (Reiff et al., 2015) Overall, among this study's respondents, test results provided a 'comforting' label and explanation, improved understanding of the offspring's behaviours, a sense of acceptance and permanence of the situation, but also of 'something … missing', that is, absent information about genetics, and about the relevance and potential use of the results (Giarelli & Reiff, 2015). Nine parents, two of whom had received genetic results, mentioned several benefits of testing, including providing a sense of personal control and hope (Trottier et al., 2013), but the analysis did not distinguish between responses from parents who did versus did not receive results.

Since these studies, genetic research has advanced rapidly, now using WES/WGS rather than CMA testing, with far more genetic variants involved in autism having been identified. Genetic diagnosis in autistic individuals has used karyotyping and CMAs and more recently WES/WGS to identify copy number variants as well as one or more single base-pair variants. (Finucane et al., 2021). As we have described elsewhere (Wynn et al., 2023), we found that among parents of children with autism who received a genetic diagnosis for their offspring through the SPARK (Simons Foundation Powering Autism Research for Knowledge) study (2023), those who received a genetic diagnosis (n = 148), compared to those who did not (n = 699), were more likely to view the autism as due to chance/luck (since the identification of a de novo variant was often presented to parents as reflecting a 'random' or 'chance' event), and less likely as due to trauma to the fetus, vaccination or lifestyle. Parents receiving genetic diagnoses also reported lower levels of optimism and stigma 1 month later, but not of self-esteem or perceived self-efficacy.

Questions arise, however, of whether parents may have other perceptions or responses than those assessed by the quantitative scales used in this study. Prior research has suggested several complexities related to genetic testing and counselling. Family members have reported, for instance, wide variations in experiences of pre-test counselling and extents of post-test counselling concerning autism (Yusuf et al., 2021). Patients in Ireland with rare diseases, for example, often experienced delays in obtaining timely genetic counselling to assist with psychosocial stresses. Self-directed web-based alternatives to genetic counselling, in which patients would not interact with a provider, are thus being considered, for instance, among *BRCA 1/2* negative women (Gaieski et al., 2019). Such a website might have advantages (e.g., reduced time), but also disadvantages (e.g., not being

Key Messages

- Genetic diagnoses that parents receive concerning their offspring's autism with intellectual disabilities (ID) can be 'double-edged' in several ways, having advantages, but also certain disadvantages and limitations.
- Parents receiving these genetic diagnoses perceived several benefits, including medical/scientific (e.g., ending diagnostic odysseys and potentially contributing to research), emotional (e.g., relief, peace of mind and less self-blame), cognitive, social (e.g., validation and confirmation with others – it's 'not just in our head') and financial (e.g., estate planning), now and in the future (e.g., preparing for possible future symptoms, development and availability of treatment and setting realistic expectations).
- Limitations perceived by parents included a lack of medical treatments related to the genetic diagnosis, a sense of finality and heightened uncertainties which can increase anxieties (e.g., concerning additional symptoms associated with genetic diagnoses and offspring's ability to live independently in the future).
- Parents responded to these tensions in various ways, having mixed feelings, recognizing the trade-offs and/or focusing on their offspring's present needs.
- Paediatricians and other providers should be aware of these issues, to inform and assist families, who are considering such testing, about these potential pros and cons and responses.

13652214, 2024, 1, Downloaded from https://onlinelibrary.wiley.com/doi/10.1111/cch.13212 by Columbia University Libraries, Wiley Online Library on [04/06/2024]. See the Terms and Conditions (https://onlinelibrary.wiley.com/terms-and-conditions) on Wiley Online Library for rules of use; OA articles are governed by the applicable Creative Commons License

able to respond specifically to patients' questions) (Gaieski et al., 2019). These issues are important, in part, because genetic counselling can affect the number of at-risk relatives to whom a patient with genetic test findings communicates these results. Relatives of patients with *BRCA 1/2* results, for example, may find the results helpful, but not always be told of them (Daly et al., 2016). Efforts are therefore being explored to enhance case-finding by increasing the number of relatives to whom patients communicate the results (Hodgson et al., 2014).

Yet autism, which may manifest itself largely in psychological and behavioural symptoms, may differ considerably from many other conditions, such as breast cancer, for which more objective medical diagnostic tests exist.

We thus conducted in-depth semi-structured interviews with a subset of the parents in the SPARK study to address their perceptions and experiences of genetic testing. In these interviews, themes arose concerning the perceived advantages and disadvantages of testing and needs to weigh these against each other. We explored these domains more fully and present these data here.

2 | METHODS

As described in the Supporting Information S1, we followed COnsolidated criteria for REporting Qualitative research (COREQ) guidelines (Tong et al., 2007) for conducting qualitative research. We invited the 148 parents of offspring who had received a genetic diagnosis associated with autism in SPARK to participate in a qualitative interview study. In brief, we then conducted in-depth qualitative interviews with 28 parents who received de novo results until 'saturation' was reached (i.e., 'the point at which no new information or themes are observed in the data') (Guest et al., 2006). Interviews were completed from April 2019 through June 2021, and participants were compensated with a $50 gift card. The Institutional Review Board of the New York State Psychiatric Institute approved the study.

The semi-structured interview questionnaire was drafted, drawing on the prior literature. Questions explored participants' views and experiences regarding autism, focusing on questions of ambiguity, responsibility and identity. The Principal Investigator (PI) for the qualitative study conducted all the interviews. Table 1 includes sample questions, asked of all participants.

The methods for the present study (see details in Supporting Information S1) adapted key elements from 'grounded theory' (Corbin & Strauss, 2014) and were thus informed by techniques of 'constant comparison', with data from different contexts compared for similarities and differences to see if they suggest hypotheses. This technique generates new analytic categories and questions and checks them for reasonableness.

TABLE 1 Sample questions asked of participants.

How have you viewed the genetic test results that you received?
How did you feel about receiving them?
Do you think that having the results is helpful? If so, how?
Do you have other thoughts on these issues?

3 | RESULTS

Twenty-eight parents (13 fathers and 15 mothers) who had received a genetic diagnosis of a de novo variant associated with autism for their offspring were interviewed within several months of receiving the genetic test result. Of these parents, 75% identified as White and 7% as Latino, and the race/ethnicity of 18% was not available. They resided throughout the US.

We interviewed both the mother and the father in six couples; these 28 parents had a total of 22 offspring with autism who had received a diagnosis. As seen in Table 2, 59% of these offspring were male and 41% were female. The mean age of the offspring with autism was 13.6 years, and they had a range of variants (see Table 3).

TABLE 2 Quantitative data on offspring with genetic diagnosis of de novo variants.

Variable	Number	Per cent
Gender:		
Male	13	59%
Female	9	41%
Age:		
0–5 years	2	9%
6–12 years	8	36%
13–18 years	9	41%
>18 years	3	14%
Vineland 3 adaptive behaviour: Composite scores and levels:		
Range	20–79	
Mean	58	
Median	64	
Adaptive behaviour level: Moderately low (71–85)	4	18%
Low (20–70)	14	64%
Unavailable	4	18%
Medical conditions: Major categories:		
Speech and language, intellectual disability (ID)/cognitive impairment, learning disability LD) or other developmental delay or developmental disability	17	77%
Birth or pregnancy complications: For example, premature birth, oxygen deprivation at birth and serious prenatal infection (e.g., rubella)	12	55%
Attention or behavioural disorders	10	45%
Mood, anxiety or obsessive-compulsive disorder (OCD)	7	32%
Sleeping, feeding/eating or toileting problems	6	27%
Neurological conditions	6	27%

13652214, 2024, 1, Downloaded from https://onlinelibrary.wiley.com/doi/10.1111/cch.13212 by Columbia University Libraries, Wiley Online Library on [04/06/2024]. See the Terms and Conditions (https://onlinelibrary.wiley.com/terms-and-conditions) on Wiley Online Library for rules of use; OA articles are governed by the applicable Creative Commons License

TABLE 3 Genetic variants found among offspring.

Variant	Number
CUL3	2
POGZ	2
ADNP	1
CASK	1
CTCF	1
EP300	1
GIGYF1	1
GRIN2B	1
KCNQ5	1
KMT2A	1
KMT2C	1
MED13	1
NRXN1	1
SCN2A	1
SHANK3	1
SYNGAP1	1
TAOK1	1
TBR1	1
TCF20	1
WDFY3	1
Total	**22**

In brief, as outlined in Figure 1 and described more fully in the text below, parents perceived both benefits (medical, scientific, emotional, cognitive and social, now and in the future) and limitations of these genetic diagnoses (lack of medical treatments related to the genetic diagnosis, a sense of finality and heightened uncertainties that can increase anxieties).

3.1 | Advantages of receiving genetic diagnoses

3.1.1 | Medical benefits

Medically, such results can end diagnostic odysseys and provide closure that assists parents both medically and emotionally.

> It just feels like there is a bit more closure or a little bit less of a mystery because it was always understood that there's a gazillion possible causes and nobody could narrow it down, and the treatment is just treating the symptoms. But having [the genetic] information just makes the picture more complete in my head. It seems more satisfying. [Father 4]

A genetic diagnosis could thus save time and effort otherwise spent in seeking medical answers.

> I felt relief because I spent a lot of time taking her to different specialists and doctors to find out what went wrong. The fact that there's a tangible syndrome is helpful after years and years of nothing. Half of those years were, 'No, this is not autism. This is cognitive developmental delay', which is really worse because you do not know why. [Mother 13]

New scientific understandings of the cause of an offspring's autism could eventually bring treatment and thus generate hope.

> It gives you some kind of glimmer of hope. Not that there's going to be a miracle cure one day, but at least we now understand what's going on in my son's head. We understand there's a source. [Father 2]

In contributing to scientific research, parents may perceive those potential benefits not only for themselves and their offspring but for others, too. Parents were excited about possible scientific advances and improvements in the future.

> It's comforting to know that progress has been made and with identification [of genetic variant] comes treatment or a way to eliminate the problem. Hopefully, other families and children will not go through what we had to go through. [Father 10]

Identification of genetic diagnoses through the SPARK study also aids research. Such research participation was gratifying as a form of altruism. 'I like to contribute to the database where we can. It's neat in that regard, to help contribute'. [Father 9]

3.1.2 | Cognitive benefits

The results also offered benefits cognitively. Parents commonly felt that knowledge was beneficial in and of itself, that, 'The more information, the better. I think it's good … More information is always better than less'. [Father 12].

Putting 'a name' to a condition can be helpful cognitively.

> It feels helpful having something to point to. To be able to *name* it and say, 'He has autism, and this is why', and 'His brain does not work the way that other people's brains work, and this is why'. [Mother 3]

3.1.3 | Emotional benefits

Having a label can also prove helpful emotionally, reducing troubling doubts and uncertainties. Genetic diagnoses and consequent sense of

13652214, 2024, 1, Downloaded from https://onlinelibrary.wiley.com/doi/10.1111/cch.13212 by Columbia University Libraries, Wiley Online Library on [04/06/2024]. See the Terms and Conditions (https://onlinelibrary.wiley.com/terms-and-conditions) on Wiley Online Library for rules of use; OA articles are governed by the applicable Creative Commons License

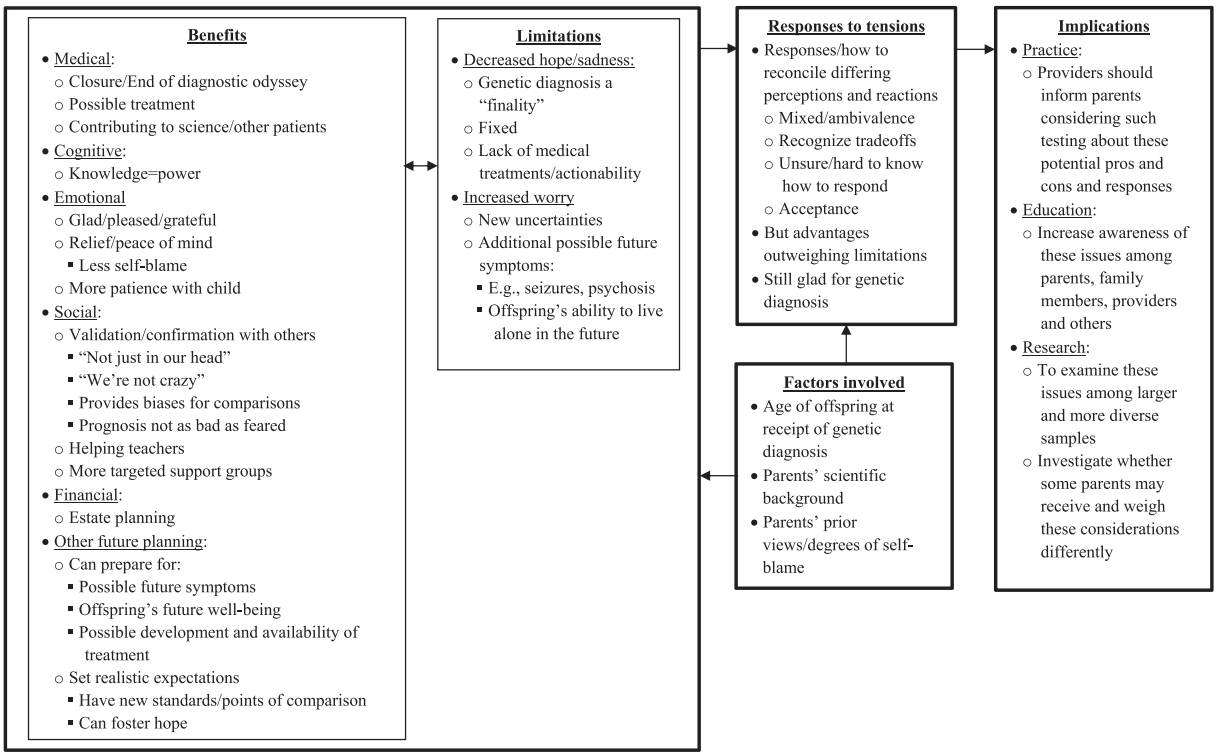

FIGURE 1 Views of benefits and limitations of genetic diagnosis.

closure gave parents relief and reduced anxieties. 'It was a relief in that we have a potential cause … It gives us the peace of mind that we have an answer to part of the question'. [Father 1]

Importantly, genetic diagnoses can also reduce parents' self-blame. 'The relief was knowing that there was a cause other than me for some of her characteristics'. [Mother 13]

The test results can decrease, too, parents' self-doubt and provide validation, alleviating self-questioning and mistrust parents may have had about their own assessments. 'I was just happy to know that we're not crazy and that there is something with [my daughter] that is unusual'. [Father 6]

A genetic diagnosis can also increase parents' patience and reduce their frustration with their child's specific traits.

> It makes us more understanding and gives us more *patience* because we know that this is the way it is. It helps us to understand what's going on in her brain a little bit more and why. [Mother 8]

Relatedly, parents may lower their level of irritation when their offspring exhibits difficult behaviour.

> I have less expectation now. I do not rack my brain trying to figure out why he's doing the things that he is … it's brought about more peace, for me … I was expecting [my son] to learn something because we have done things over and over again or to process more quickly, because he would say he does not remember things or forgets things that most people would have learned. That was frustrating for me. It's not frustrating anymore because I know he just cannot do it. The slow processing is a part of this syndrome. So, I've become much more *patient* in that regard. [Father 3]

3.1.4 | Social benefits

Parents also thought that the genetic diagnosis provided social benefits in interactions with healthcare providers, teachers and others. Parents often felt legitimated, especially if they had difficult prior diagnostic journeys and their sense of their offspring's condition was questioned by others.

> The result was validating … Our paediatrician kept saying, 'Oh, it's just the way she's made. I do not really see anything too concerning'. But then, we were seeing plenty of the opposite. But it leaves that little bit of doubt: 'Are we just overreacting? Should we stop comparing her to her sister?' The genetic results validate what is otherwise sort of a subjective assessment. [Mother 12]

Such an objective evaluation is helpful, given the perceived subjectivity of certain symptoms.

13652214, 2024, 1, Downloaded from https://onlinelibrary.wiley.com/doi/10.1111/cch.13212 by Columbia University Libraries, Wiley Online Library on [04/06/2024]. See the Terms and Conditions (https://onlinelibrary.wiley.com/terms-and-conditions) on Wiley Online Library for rules of use; OA articles are governed by the applicable Creative Commons License

> It just makes it more concrete, because before we had the genetic testing, I would say, 'He's just different'. And people would say, 'No, I do not see that'. I'd reply, 'But he is'. So, the result gave me something more *concrete*, to say, 'I'm not making this up in my head'. That was very reassuring. [Mother 1]

Genetic diagnoses can also help parents and offspring's interactions with teachers. One child's school did not believe he had a real disability. '[My wife] submitted information to the school … I think it will change their focus for him and help guide him in a way that adjusts to this [condition] some more'. [Father 3]

Due to genetic diagnoses, parents can now join more targeted support groups, focused on specific genetic diagnoses, and see that the prognosis need not be as bad as they may fear.

> The study did a good job of connecting the few people on Facebook who had children [with this genetic diagnosis], and we have been able to communicate. Seeing that the rest of these kiddos look like they are doing well and are cognitively on track is reassuring. [Mother 12]

3.1.5 | Financial benefits

Additionally, genetic diagnoses can prompt parents to financially plan more fully for their child's future.

> We have made estate planning decisions because of this information. We've been trying to focus on having plans in place for when we are no longer here and [my daughter] still likely needs to be cared for, and we have talked to our other kids, and kind of set them that [that's important]. [Father 6]

3.1.6 | Other benefits of future planning

Genetic diagnoses also help parents set realistic expectations and thus better plan for their offspring's future in other ways, providing more focused and specific knowledge of what to expect. 'At least we have a little bit of a parameter of what is common'. [Mother 1] The genetic diagnosis can confirm what they had long suspected, and thus solidify what to anticipate. 'We've come to terms with her non-verbal nature and some of her physical limitations … They are going to be here for the rest of her life'. [Father 7].

Genetic diagnoses can assist parents, too, in preparing for future symptoms and obtaining helpful tools and support. Parents can now proactively monitor for symptom development.

> The primary thing is the schizophrenia. That's part of the things we keep an eye out for. Since we did not have that information in advance, we were fumbling in the dark, trying to get assistance since he was one-and-a-half. We now might say, 'Ok, if he starts exhibiting certain behaviours, this is definitely something we have got to kind of keep our eyes open for'. [Father 12]

Such more precise 'benchmarks' or bases for comparison can also mitigate fears of the worst, thereby instilling a degree of hope. Parents may feel grateful that their child does not exhibit as many symptoms or appear as severely affected as others.

> Just knowing what other families are dealing with – it could definitely be a lot worse. There are kids way worse than [my son]. So, it makes me thankful. Not that I want other kids to be worse, but I think, 'Ok, this could have been way worse, you just need to move on, and be happy with what you have got'. [Mother 1]

3.2 | Limitations of receiving genetic diagnoses

Yet genetic diagnoses also have certain limitations, potentially decreasing hope, given the lack of medical treatments, and generating senses of finality and sadness.

> There is an element of sadness or grief, just putting the finality on it, knowing that this is *a life sentence* for her. There is no medication or magic pill that we can have to take it away from her. We already knew that, but I think just the more you dig in and find out, you just have to start coming to terms with that. [Father 7]

This sense of finality and diminished hope can be poignant, though felt to varying degrees by spouses at different points, especially initially. '[My wife] was more in tears: our son will never have a good life'. [Father 3].

Parents generally understand that science is advancing, but they nonetheless often wish for more insights or treatments.

> I wish there was more information out there that would be helpful and available to me: the gene affects *this*, so maybe *this type* of therapy would help. Maybe in the future. [Mother 17]

Learning about other possible future symptoms can also cause uncertainty and anxiety more than relief from being able to prepare for these potential problems.

> I believe that this condition typically might present seizures or epileptic issues. Our daughter has never experienced a seizure that we have noticed, that we have ever seen or caught. So, we do wonder and worry

13652214, 2024, 1, Downloaded from https://onlinelibrary.wiley.com/doi/10.1111/cch.13212 by Columbia University Libraries, Wiley Online Library on [04/06/2024]. See the Terms and Conditions (https://onlinelibrary.wiley.com/terms-and-conditions) on Wiley Online Library for rules of use; OA articles are governed by the applicable Creative Commons License

> about that. Is she more likely to have that? Is that going to be an adult issue for her? I do not know. [Father 7]

Fears can increase about the offspring's premature death. 'I worry about life expectancy, losing her before I die. I worry about her going before me. And life expectancy is one of those things that's part of this syndrome'. [Mother 13]

Not only knowing about such possible future symptoms but also now monitoring for them heightens uncertainties and anxieties, especially if these future symptoms may be somewhat subjective more than wholly objective and clear.

> That information [about possible future symptoms] is worrying to us. I know people live with schizophrenia. There are medications you can take. But to know that he could have yet another challenge tossed upon him later in his childhood has been in our thoughts. He likes to like tell stories or mutter to himself, so he'll say things. I'll just ask him … 'Did someone in the room say that, or are you just saying that?' He'll reply, 'No. Just telling a story. Leave me alone'. So, it has probably implanted the idea in my head. Sometimes I'm sure that I'm finding things based on my minimal knowledge of schizophrenia that do not actually exist – is this a potential issue I should be worried about? [Father 12]

Genetic diagnoses can exacerbate uncertainties, too, about an offspring's future abilities to live on their own.

> I'm just wondering how things are going to progress in the future, what the research is going to find out, if there is anything that can help. How is my son going to be when he's older? Is he going to be ok on his own? After the genetic test, everything's still up in the air. I have no idea what's going to happen, with either the [genetic diagnosis] or the autism. As he's getting older, we are definitely wondering more, 'Will he be able to be on his own? Is he going to end up living with us all of his life or our lives?' [Mother 1]

Due to the newness of information about a particular genetic diagnosis, the range of its future potential manifestations itself may not be entirely known, generating additional worries – so-called 'unknown unknowns'.

> It's concerning because we do not know a huge amount of what [the genetic variant] controls and what develops from the proteins it triggers. It can also affect the normal breakdown of proteins and cell growth and development. Could this potentially trigger tumor growth? We do not know. We need to get more science behind this to find out what it does. [Father 1]

Given the newness and current rarity of certain genetic conditions, specific support groups might also not yet exist. Online support groups exist for autism in general, but not necessarily for specific genetic diagnoses.

> We have not [joined any support groups for our offspring's condition] because this variant is extremely rare. There is only a handful of cases known in the world, so there is not any support at this point. I just wish there were more [cases]. [Father 4]

3.3 | Responses to tensions between benefits and limitations

Parents responded in several ways to these tensions between perceived advantages and limitations of testing, often having mixed reactions, seeing the genetic diagnosis as helpful in certain ways, but not others. The result can thus be 'double-edged', decreasing certain ambiguities but increasing others.

> [The test result has] relieved some uncertainty as far as what led to us having a [son like ours], but now it's the uncertainty of what to do – what general things can we expect down the road? I know there's no way to specifically say, 'at age 22 this is going to happen, at 36 that is going to'. So, it's been *doubled-edged* a little bit. [Father 1]

The genetic diagnoses can thus produce a 'mixed' picture.

> It's *bittersweet*, because I feel like we are on the front edge of everything. It would be nice to get this information and have it be 10 years from now – not today, when we are finding out, 'Oh, you are one of 12, and we know nothing about it. But keep in touch, and maybe someday, we'll get information'. So, it's a little bit of a mixed bag. When they said he's one of 12, we think this is great. We're on the cutting edge. But this is also not so great, because what can we do about it? [Mother 6].

These tensions between these advantages and limitations can yield feelings of ambivalence.

> I was *ambivalent* when I received the results. If it had changed what we can do for him, I would have been ecstatic. But just having that piece of information, if it does not change what you can do for that patient, then what good is the test? It does not change our opinion or mood or optimism in any way unless they figure out a particular treatment for this particular genetic abnormality. His functionality is not going to change. So, it

13652214, 2024, 1, Downloaded from https://onlinelibrary.wiley.com/doi/10.1111/cch.13212 by Columbia University Libraries, Wiley Online Library on [04/06/2024]. See the Terms and Conditions (https://onlinelibrary.wiley.com/terms-and-conditions) on Wiley Online Library for rules of use; OA articles are governed by the applicable Creative Commons License

> really has not impacted our planning. We're still terrified of what the future holds for him because there will come a time when physically we can no longer take care of him. [Father 8]

While parents may not have fully expected that the genetic test would have indicated a specific treatment, they often nonetheless had still hoped for such new targeted interventions. The definitiveness of this news of a lack of treatment can be hard. Many parents feel that genetic diagnosis 'didn't change anything'.

> We would have loved to have heard there's a cure or medication or nutritional supplement that would cure our daughter. We knew there was no magic cure. There wasn't going to be this 'magic moment'. I think we have already come to terms with that. So, it does not really change anything. I think we already knew it. [Father 7]

Parents may also respond to these pros and cons by managing their reactions and coming stoically to accept the vagaries that may ensue. 'It's a big worry for me. But I understand that we're just going to have to play things as they are dealt to us.' [Father 1]

Parents may respond, too, by focusing just on the child's immediate needs.

> You're always going to be worried about it. And it may be just there's a glimmer of hope that someday there's going to be [treatment]. But in the meantime, you still have to plan for what his current path is right now. [Father 2]

3.3.1 | Seeing advantages as still outweighing limitations

Nonetheless, parents all felt that the advantages of knowing outweighed the drawbacks, though the latter detracted somewhat from the former. 'I'm still definitely happier to have that information than not having that information'. [Mother 6]

Still, parents varied in which pros and cons they experienced most strongly and tended to feel that being prepared for future problems outweighed concomitant anxieties.

> It may give you more worries, considering things that might be an issue that we had never thought about, but at least we now know. At least, going forward, we will be better prepared. There may be other problems in the future – potentially schizophrenia, which is somewhat concerning – and more uncertainty about the future. But the fact that we have this information gives us a head start on being able to identify any issues, a jumpstart. [Mother 3]

Parents may feel sad about the loss of hope, but also appreciate the enhanced insight about their offspring. The resultant loss of hope can be painful, but still ultimately less important than the perceived benefits of testing.

> I always felt as though she had the potential to be some kind of normal, as they say. And [genetic diagnosis] wiped out just that little inkling and hope that I always held on to. That has just gone away, frankly. The hope has really dwindled down to nothing because she's 23 now. That hope was there more when she was 10 and earlier. Now it seems it's a concrete thing. And that's sad. [Mother 13]

Yet this same parent, appreciated, too, the advantages of testing, valuing the reduced frustration and increased insight on how to interact better with her offspring.

> [The genetic diagnosis] also helps me be more understanding about her weight problem, which is significant. I'm not quite as kind of in that blame mode about 'Do not eat as much'. [Mother 13]

To a large degree, these pros and cons may therefore effectively balance each other out, producing, in certain ways, little overall change. 'I wouldn't say I was more or less concerned either way'. [Father 2]

3.4 | Factors involved

Several factors, especially the child's age, may influence parents' views of these pros, cons and trade-offs. When an offspring is younger, the results and possible psychosocial interventions, even if no 'cure' exists, can feel more beneficial.

> If we had the knowledge a little bit earlier, it would've been nice, and maybe informed some of the earlier decisions that we made. We could've gotten him support at an earlier age. We did pretty well at three-and-a-half. But I look back and think, 'If we had started one-and-a-half to two, we could've helped him even more!' [Father 12]

Conversely, some parents of older children hypothesized that having known these results early on in their child's life would have made them more anxious. A mother of a 20-year-old daughter with autism reported,

> I would have definitely changed my expectations or had a major concern if she were 8 years old and we got those results … I would be more worried if she had

13652214, 2024, 1, Downloaded from https://onlinelibrary.wiley.com/doi/10.1111/cch.13212 by Columbia University Libraries, Wiley Online Library on [04/06/2024]. See the Terms and Conditions (https://onlinelibrary.wiley.com/terms-and-conditions) on Wiley Online Library for rules of use; OA articles are governed by the applicable Creative Commons License

> not been fully an adult yet … I would not know what to expect … I would fear the worst. [Mother 13]

Still, parents recognize that scientists may not have known these genetic variantsearlier.

> I wish I had [this genetic diagnosis] sooner, but as I understand it, this wasn't known until 2010. Having this diagnosis when a child is younger is now extremely helpful because you can find what your child needs. [Mother 8]

As suggested earlier, many parents had previously blamed themselves for their offspring's autism and now feel heightened relief at obtaining the genetic diagnosis. The degree of initial self-blame varied considerably among parents, moderating the degree of relief. Overall, mothers seemed somewhat more concerned than fathers about the effects of possible harms that may have occurred during the pregnancy (e.g., drinking a glass of wine before being aware of the pregnancy). Mothers, as the ones who are themselves pregnant, appeared more exquisitely aware of and sensitive to such phenomena and therefore may experience relatively more reduction in self-blame. Parents with the most self-blame appeared to feel the most relief, though also sometimes continued to question themselves.

4 | DISCUSSION

These data, the first to examine how parents perceive genetic diagnoses they receive for their offspring with autism and often with ID through WES/WGS, suggest how these diagnoses can be 'double-edged' and 'bittersweet', having clear advantages, but also certain disadvantages and limitations. The benefits were medical, scientific, emotional, cognitive, social and financial, both now and in the future. Perceived limitations of genetic diagnoses included lack of medical treatments related to the genetic diagnosis, a sense of finality and heightened uncertainties that can in turn increase anxieties. On balance, these parents were glad to have received the results, seeing the pros as outweighing the cons. Parents responded to these tensions in various ways, having mixed feelings or ambivalence, recognizing the trade-offs and/or focusing only on their offspring's present needs. Just as autistic adults may have mixed responses to genetic test results (Gravino, 2021), the present data show that parents do, too, but for varying reasons.

These data reveal cognitive and other psychological benefits of being able to put a label or name on a set of disturbing experiences. While 'labelling theory' has focused on how labels mark certain people as deviant in ways that exacerbate stigmatization and deviant behaviour (Bernburg, 2009), the present data highlight how naming a condition can also confer certain benefits. These data also highlight how genetic diagnoses can lead to personal, even if not medical, 'actionability'. Parents were glad to receive the genetic diagnoses, feeling 'validated', engaging more fully in financial planning for their offspring's future and feeling better prepared for that future and better able to obtain earlier psychosocial services.

These limitations and complexities are important to note because other parents, family members and patients will likely confront similar issues, and geneticists, genetic counsellors, other providers and the public at large should be aware of these dual sets of potential reactions, in order to best frame and understand such test results. Research participants often 'self-select' and may be more favourably inclined than the public at large towards medical science and appreciative of its benefits. Hence, while the parents here all valued the advantages of these test results more than the disadvantages, other parents and patients, in clinical rather than research contexts, may view these tensions differently and may weigh the advantages less and the disadvantages more. Such parents may therefore be more reluctant to have their child undergo genetic testing if a 'cure' for autism remains lacking, without fully appreciating other potential medical, scientific, emotional, cognitive, social or future benefits.

5 | LIMITATIONS

These data have several potential limitations. These findings are based on interviews with 28 parents, which is adequate for qualitative analysis (Hennink et al., 2017), but future studies employing larger samples can examine these issues further. The interviewees were largely, though not exclusively, White, yet future research can and should include more robust ethnic and racial diversity.

6 | CONCLUSION

These data have several important implications for future practice, education and research. These findings can help providers in understanding, as fully as possible, the potential pros and cons of genetic testing from parents' perspectives. Providers should be aware of possible mixed feelings and tensions and should address these, informing individuals considering genetic tests about these potential pros and cons, to help patients and families anticipate such responses. Parents pursuing genetic testing should be informed that they may experience a variety of complex emotions, including sadness and loss of hope, but that parents generally appear to conclude that the advantages of such testing outweigh the disadvantages.

Future quantitative studies should probe these areas further among larger, more diverse samples to examine whether perceived benefits and/or limitations are greater among certain groups, related to sociodemographic or other variables (e.g., parents' or offsprings' age, gender, socioeconomic status, race/ethnicity or scientific or other education), and whether individuals may differ in perceiving and weighing these considerations, and if so, why.

13652214, 2024, 1, Downloaded from https://onlinelibrary.wiley.com/doi/10.1111/cch.13212 by Columbia University Libraries, Wiley Online Library on [04/06/2024]. See the Terms and Conditions (https://onlinelibrary.wiley.com/terms-and-conditions) on Wiley Online Library for rules of use; OA articles are governed by the applicable Creative Commons License

AUTHOR CONTRIBUTIONS

Robert Klitzman, Paul S. Appelbaum, Wendy K. Chung and Ekaterina Bezborodko contributed to the conceptualization, writing, data analysis, revising and drafting of the article.

ACKNOWLEDGMENTS

The authors would like to thank Julia Wynn, MS, CGC and the SPARK study staff for facilitating our access to these participants and data collection and Rebecca Grinberg, Timothy Keith Hung, Batool Naqvi and especially Patricia Contino for their assistance with the preparation of the manuscript.

CONFLICTS OF INTEREST DISCLOSURE

The authors have no conflicts of interest to disclose.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available from the corresponding author upon reasonable request.

ETHICS APPROVAL STATEMENT

a. Research Involving Human Participants and/or Animals: The Columbia University Department of Psychiatry/New York State Psychiatric Institute Institutional Review Board approved the study.
b. Informed Consent: We obtained informed consent from all of the participants. We thank the study participants for giving their time and perspective.

ORCID

Robert Klitzman https://orcid.org/0000-0002-6827-8063
Wendy K. Chung https://orcid.org/0000-0003-3438-5685

REFERENCES

Bernburg, J. G. (2009). Labelling Theory. In M. D. Krohn, A. J. Lizotte, & G. P. Hall (Eds.), *Handbook of crime and deviance* (pp. 187–207). Springer. https://doi.org/10.1007/978-1-4419-0245-0_10

Corbin, J., & Strauss, A. (2014). *Basics of qualitative research: Techniques and procedures for developing grounded theory* (4th ed.). SAGE Publications, Inc.

Daly, M., Montgomery, S., Bingler, R., & Ruth, K. (2016). Communicating genetic test results within the family: Is it lost in translation? A survey of relatives in the randomized six-step study. *Familial Cancer*, *20*, 697–706. https://doi.org/10.1007/s10689-016-9889-1

Finucane, B. M., Ledbetter, D. H., & Vorstman, J. A. (2021). Diagnostic genetic testing for neurodevelopmental psychiatric disorders: Closing the gap between recommendation and clinical implementation. *Current Opinion in Genetics & Development*, *68*, 1–8. https://doi.org/10.1016/j.gde.2020.12.016

Gaieski, J., Patrick-Miller, L., Egleston, B., Maxwell, K., Walser, S., DiGiovanni, L., Brower, J., Fetzer, D., Ganzak, A., McKenna, D., Long, J., Powers, J., Stopfer, J., Nathanson, K., Domchek, S., & Bradbury, A. (2019). Research participants' experiences with return of genetic research results and preferences for web-based alternatives. *Molecular Genetics & Genomic Medicine*, *7*(9), e898. https://doi.org/10.1002/mgg3.898

Giarelli, E., & Reiff, M. (2015). Mothers' appreciation of chromosomal microarray analysis for autism spectrum disorder. *Journal for Specialists in Pediatric Nursing*, *20*(4), 244–258. https://doi.org/10.1111/jspn.12121

Gravino, A. (2021, April 22). *I now know what caused my autism, which changes everything – And nothing*. Retrieved July 17, 2023, from CNN.com Shortened link to article

Guest, G., Bunce, A., & Johnson, L. (2006). How many interviews are enough? An experiment with data saturation and variability. *Field Methods*, *18(1)*, 59–82. https://doi.org/10.1177/1525822X05279903

Hayeems, R. Z., Babul-Hirji, R., Hoang, N., Weksberg, R., & Shuman, C. (2016). Parents' experience with pediatric microarray: Transferrable lessons in the era of genomic counseling. *Journal of Genetic Counseling*, *25*(2), 298–304. https://doi.org/10.1007/s10897-015-9871-3

Hennink, M. M., Kaiser, B. N., & Marconi, V. C. (2017). Code saturation versus meaning saturation: How many interviews are enough? *Qualitative Health Research*, *27*(4), 591–608. https://doi.org/10.1177/1049732316665344

Hodgson, J., Metcalfe, S., Aitken, M., Donath, S., Gaff, C., Winship, I., Skene, L., McClaren, B., Paul, J., & Halliday, J. (2014). Improving family communication after a new genetic diagnosis: A randomised controlled trial of a genetic counselling intervention. *BMC Medical Genetics*, *15*, 33. https://doi.org/10.1186/1471-2350-15-33

Johannessen, J., Nærland, T., Hope, S., Torske, T., Kaale, A., Wirgenes, K. V., Malt, E., Djurovic, S., Rietschel, M., & Andreassen, O. A. (2022). Attitudes among parents of persons with autism spectrum disorder towards information about genetic risk and future health. *European Journal of Human Genetics*, *30*(10), 1138–1146. https://doi.org/10.1038/s41431-021-00966-y

Reiff, M., Giarelli, E., Bernhardt, B. A., Easley, E., Spinner, N. B., Sankar, P. L., & Mulchandani, S. (2015). Parents' perceptions of the usefulness of chromosomal microarray analysis for children with autism spectrum disorders. *Journal of Autism and Developmental Disorders*, *45*(10), 3262–3275. https://doi.org/10.1007/s10803-015-2489-3

SPARK: Simons Powering Autism Research. (2023). Retrieved July 17, 2023, from https://sparkforautism.org/

Srivastava, S., Love-Nichols, J. A., Dies, K. A., Ledbetter, D. H., Martin, C. L., Chung, W. K., Firth, H. V., Frazier, T., Hansen, R. L., Prock, L., Brunner, H., Hoang, N., Scherer, S. W., Sahin, M., Miller, D. T., & The NDD Exome Scoping Review Work Group. (2019). Meta analysis and multidisciplinary consensus statement: Exome sequencing is a first-tier clinical diagnostic test for individuals with neurodevelopmental disorders. *Genetics in Medicine*, *21*(11), 2413–2421. https://doi.org/10.1038/s41436-019-0554-6

Tong, A., Sainsbury, P., & Craig, J. (2007). Consolidated criteria for reporting qualitative research (COREQ): A 32-item checklist for interviews and focus groups. *International Journal for Quality in Health Care*, *19*(6), 349–357. https://doi.org/10.1093/intqhc/mzm042

Trottier, M., Roberts, W., Drmic, I., Scherer, S. W., Weksberg, R., Cytrynbaum, C., Chitayat, D., Shuman, C., & Miller, F. A. (2013). Parents' perspectives on participating in genetic research in autism. *Journal of Autism and Developmental Disorders*, *43*(3), 556–568. https://doi.org/10.1007/s10803-012-1592-y

Wynn, J., Karlson, A., Levine, A., Huber, B., Salem, A., Casey White, L., … Appelbaum, P. S. (2023). *Impact of a genetic diagnosis of a child's autism on parental perceptions*. [Manuscript submitted for publication]. Columbia University.

13652214, 2024, 1, Downloaded from https://onlinelibrary.wiley.com/doi/10.1111/cch.13212 by Columbia University Libraries, Wiley Online Library on [04/06/2024]. See the Terms and Conditions (https://onlinelibrary.wiley.com/terms-and-conditions) on Wiley Online Library for rules of use; OA articles are governed by the applicable Creative Commons License

Yusuf, A., Peltekova, I., Savion-Lemieuz, T., Frei, J., Jooba, R., Howe, J., Scherer, S., & Elsabbagh, M. (2021). Adaptation and validation of the genetic counseling outcome scale for autism spectrum disorders and related conditions. *Journal of Genetic Counseling*, *30*(1), 305–318. https://doi.org/10.1002/jgc4.1323

SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.

How to cite this article: Klitzman, R., Bezborodko, E., Chung, W. K., & Appelbaum, P. S. (2024). Parents' views of benefits and limitations of receiving genetic diagnoses for their offspring. *Child: Care, Health and Development*, *50*(1), e13212. https://doi.org/10.1111/cch.13212

13652214, 2024, 1, Downloaded from https://onlinelibrary.wiley.com/doi/10.1111/cch.13212 by Columbia University Libraries, Wiley Online Library on [04/06/2024]. See the Terms and Conditions (https://onlinelibrary.wiley.com/terms-and-conditions) on Wiley Online Library for rules of use; OA articles are governed by the applicable Creative Commons License