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An investigation of autistic opinions about autism-related genomic research

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Abstract

Genomic studies of autism are a heavily debated, often controversial, topic within the autism community. Autistic voices have historically been poorly represented within research which concerns them directly. Although recent efforts have been made to involve the voices of those with lived experience, there is a dearth of knowledge of Autistic opinions of genomic autism studies. This study sought to address this. This research has collected the views and opinions of late-diagnosed Autistic adults on genomic research as part of the PEAPOD (Personal Experiences of Autism and Perceptions Of DNA-based research) study. It consisted of 20 semi-structured interviews with Autistic adults residing in the UK, conducted by an Autistic researcher. The study used multiple data collection methods including using video conferencing software and social media messaging. Interviews were transcribed and thematically analysed. Four themes were identified including a diverse understanding of genomic research, what is genomic research seeking to achieve? who is doing what? (and for whom?), and reflecting on the Autistic voice. We conclude more is needed to address the concerns of the Autistic community surrounding genomic studies and suggest recommendations for how this can be achieved.

Lay abstract

What is already known about the topic?

There has been a notable, often negative, reaction to genetic studies into autism by the autism community. This includes the Spectrum 10k genetics study at Cambridge University, which was paused as a response to the community's reaction, both over social media and in person. There is currently very little known about the opinions of Autistic people about autism-related genetic research. This gap in the knowledge influenced the Personal Experiences of Autism and Perceptions of DNA-based research team at the University of York to ask Autistic adults, who were late diagnosed, their opinions on genetic studies of autism.

What this paper adds?

In the research literature currently, there is very little of what Autistic people think about genetic studies which directly involve and impact them. This research consisted of 20 semi-structured interviews with Autistic adults residing in the UK, conducted by an Autistic researcher. The study used multiple methods of data collection including video conferencing software and social media messaging. Interviews were transcribed, where appropriate, and thematically analysed. Multiple opinions are represented within this research, covering several topics, including the impact of historic and current practice on trust; ethical considerations such as the use of data in future studies; and representation of the Autistic voice within research.

Implications for practice, research, or policy.

This paper suggests recommendations for future genetic research into autism. This includes considering how current data usage and sharing practices can be adapted to suit the needs of the Autistic community and improving the science communication between researchers and the Autistic, and autism, community. However, this data was from a small group of late-diagnosed Autistic people in the UK, and so results will not represent the whole community.

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A note on language:

This paper will use identity-first language. However, if a participant employed person-first language, this has not been changed to respect their self-determination (Botha et al., 2021). Within this paper, 'Autistic community' will refer to Autistic people solely whilst 'autism community' refers to Autistic individuals, family members, allies and professionals within this field.

Keywords

autism, qualitative, genetics, genomics, co-production

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Introduction

There have been multiple, large-scale genomic studies of autism in the past two decades. The MSSNG project, started in 2014, was designed to create the world's largest autism whole genome sequencing database using both Google technology and the Autism Genetic Research Exchange repository of clinical and genetic information. It is a collaboration between Autism Speaks, Verily, DNAstack and SickKids. The aim on the project website is to 'provide the best resources to enable the identification of many subtypes of autism, which may lead to better diagnostics, as well as personalised and more accurate treatments' (2024). Likewise, AIMS2 (Autism Innovative Medicine Studies-2-Trials) was launched in 2018, aiming to identify biological markers for Autistic characteristics, which could, in turn, identify appropriate treatments for co-occurring conditions (AIMS2, 2020).

Genomic studies of autism are controversial within the Autistic and wider autism community, with concerns surrounding data protection, privacy and the aims of the research itself (Autistic Self-Advocacy Network (ASAN), 2022). There are also concerns about the organisations involved and their underlying philosophies. For example, problematic historical behaviour has led to members of the wider autism community to argue that Autism Speaks does not represent them (Willingham, 2013). Autism Speaks removed the term 'cure' from their own mission statement in 2016 and have amended their research priorities (Autism Speaks, 2024). However, whether these changes have created a neurodiversity-affirming organisation, which centres the Autistic voice, is unclear, and there remains a prevailing mistrust of Autism Speaks within the community (Luterman, 2020).

In August 2021, the project Spectrum 10k was launched at the University of Cambridge, in collaboration with the Wellcome Sanger Institute and the University of California at Los Angeles (UCLA). It was directed by Professor Baron-Cohen, director of the Autism Research Centre. The aim of the project was to 'investigate the genetic and environmental factors that contribute to autism and related physical and mental health conditions to better understand wellbeing in Autistic people and their families' (Spectrum10k, 2022). The plan was to collect both questionnaire data and DNA samples from participants. In response to this project, the 'Boycott Spectrum 10k campaign' held in-person protests in Cambridge (Pring, 2021) and published a statement of concern from the autism community, alongside a petition (Aucademy, 2021). Complaints were also made to the Health Research Authority regarding research transparency (Dattaro, 2022). Protestors identified both a lack of clarity as to how data would be stored by the research team and used in the future and an uncertainty as to the aims of the study (Dattaro, 2022). As Baron-Cohen stated : 'There's no way that we can ever say that a future political leader or a scientist won't use the research for eugenics' (Hare, 2021). The Spectrum 10k project paused recruitment in September 2021 to consult more fully with the autism community, a reaction deemed insufficient by several autism advocates at the time (Natri, 2021).

Past incidences of autism research have been dehumanising, objectifying or stigmatising for Autistic individuals (Botha, 2021; Botha & Cage, 2022). Alongside this, the Autistic voice is often excluded from knowledge production and from determining research priorities (Fletcher-Watson et al., 2018; Milton, 2014). Research which does not involve the Autistic voice has been criticised as lacking epistemological integrity (Woods et al., 2018), by disempowering the community and negatively impacting Autistic autonomy (Milton et al., 2014). These factors have contributed towards a lack of trust between researchers and the Autistic community (Milton, 2014). There is a paucity of research into Autistic opinions of genomic autism studies. The first study to explore the perceptions of Autistic adults on genetic testing for autism was only recently conducted, in which 49% of survey respondents ($n = 461$) felt genetic testing on autism should not be done and 40% believed genetic testing was harmful (Byres et al., 2023), demonstrating concerns shared by advocates. The scarce research we have on this topic suggest varying opinions within the community as this field of research (Gallion et al., 2024). Our research sought to expand on these concerns through qualitative investigation.

The Personal Experiences of Autism and Perceptions Of DNA-based research (PEAPOD) project was designed in response to the Spectrum 10k boycott. Recognising the heterogeneity of the Autistic population, PEAPOD conducted research with sub-groups within the community, in the hope of representing diverse voices. The current research focused on the opinions of late-diagnosed Autistic people who may have re-assessed their understanding of themselves, and autism, through a new perspective, following their diagnosis (Lilley et al., 2022). The objective for this research was *to document and understand late-diagnosed Autistic peoples' opinions on genetic studies into autism.*

Method

Community involvement

Our research was led by an Autistic researcher (RE) and benefitted from the involvement of two Autistic experts by experience, who were involved in the development and design of this research.

Participants

The target population were Autistic individuals who resided in the UK and were medically diagnosed late (18+ years old). Participants were recruited using opportunity sampling via social media including Facebook and X (then Twitter) in May 2022. We focused on adults who were diagnosed at the age of 18 or older, based in the UK, because we were interested in knowing how a later diagnosis shaped their views of genomics studies into autism. Participants reported how long it had been since their diagnosis (Range, 1 week to 11 years).

Volunteers were asked to complete a Qualtrics questionnaire, providing their name, the date, their preferred method of data collection and contact details. They were prompted to confirm both that they matched the inclusion criteria and the length of time since their diagnosis. In total, 28 people volunteered within the week, after which recruitment stopped. Volunteers were excluded for not being UK based (n = 3), not responding to further emails (n = 2) or declining to continue (n = 1). Twenty-two people were contacted requesting an interview, two of which did not respond. In total, 20 participants contributed data towards this study and were compensated for their time and expertise. All participants gave informed consent to take part prior to participation. Ethical approval for this study was granted by the Research Ethics Committee at York University.

Data collection

An interview guide, with key questions and prompts, was sent to participants ahead of time if requested and is available as supplementary material. A range of data collection options were provided to accommodate differing communication preferences, including video conferencing software (n = 10), written responses to prompts (n = 7), telephone calls (n = 1), email conversation (n = 1) and social media direct messaging (n = 1). Participants were advised that interviews would take between 30 and 60 min. Of those which could be timed, interviews took between 26 and 124 min (M = 53). Where appropriate, interviews were transcribed and anonymised by an external service. Additional data were gathered from participants which will be presented separately in a forthcoming paper.

Table 1. Summary of themes and sub-themes.

Theme	Sub-theme
1. A diverse understanding of genomic research	A. Current understanding across the community
	B. An emotive and controversial topic
2. What is genomic research seeking to achieve?	A. The potential to further understand and help people
	B. The downfalls and limitations of genetic research
3. Who is doing what? And for whom?	A. Ethical considerations
	B. Poor handling of historic and current research
	C. A bias in research
	D. Autism and Autistic representation within research
4. Reflecting on the Autistic voice	A. Differing opinions across the community
	B. Use of social media
	C. Autistic identity may influence opinions of genetic researchB. Use of social media

Data analysis

A thematic analysis was conducted inductively following Braun and Clarke's thematic analysis framework (2022). The thematic analysis included the following steps: familiarisation, coding, developing themes, reviewing themes, defining and naming themes and writing the report (Clarke & Braun, 2013). This method of analysis is flexible and applicable to multiple theoretical and epistemological positions, including the critical realist framework employed in this research (Braun & Clarke, 2013). A critical realist paradigm acknowledges the importance of human perception and opinions and was therefore deemed appropriate for this research (Fryer, 2022). Codes and themes were developed by the lead researcher (RE) and checked with the second researcher (KA) in a meeting. Following this meeting, codes were iteratively collapsed or expanded upon.

Results

A total of 4 themes were finalised, representing 11 sub-themes (Table 1). Due to the nature of the discussion, some quotes were coded multiple times, and there was some natural overlap between sub-themes which are referenced in the below narrative.

A diverse understanding of genomic research

There was a demonstrable range in *current understanding across the community* when it came to autism-related genomic research. Some participants believed they had limited knowledge about the processes involved in autism genomic research but were aware of the genetic component of autism or had heard of notable studies: *'I have only heard of Spectrum 10k'* (003). A range of aims were suggested, including identifying a gene or multiple genes linked to autism, aiding in earlier diagnosis and preventing misdiagnosis. Such aims were generally spoken about positively, e.g. '*I'd love to be able to know if my grandad or my uncle … were Autistic, because I reckon they were'* (011). Several individuals were concerned about the 'end goal' of genomic research: '*I believe the intention is to find a cause for autism, and that would probably lead to either a cure, or a way of testing for autism. Neither of these seem like important aims to me …*' (010). There were also concerns that genomic research implied that autism is inherently wrong as it '*aims to understand what is seen as a problem*' (005). It was acknowledged that this topic would have '*strong implications for the Autistic community'* (001), as genomics were *an emotive and controversial topic*. There was an acknowledgement that this topic was an upsetting one to discuss: '*I know I will get upset by it …'* (003). A couple of participants noted their introduction to genomic autism studies was Spectrum 10k *'because it caused a fair bit of controversy'* (004).

What is genetic research seeking to achieve?

The potential to further understand and help people were discussed as positives of genetic research. Firstly, several participants noted an interest in research in general, believing the expansion of '*knowledge for knowledge's sake'* (005), to be a worthy pursuit: '*I generally think that academia and research and improving our understanding of things is a good thing'* (012). It was also suggested that genetic research could lead to an earlier diagnosis, although this was coupled with the caveat that this would need to lead to earlier support provision, inclusive of support for co-occurring conditions which impacted daily life, as described below:

> *I know that a lot of us experience gastrointestinal and connective tissue/joint issues, and so if there were definite benefits to be had in those areas which did not aim to do anything to change our neurology, then that I could understand.* (017)

One participant remarked that '*… if someone could say, "Oh we can turn that down a little bit," that would be brilliant'* (004), reinforcing that attempts to identify the genetic basis of specific traits may be viewed as beneficial by some. However, *the downfalls and limitations of genetic research* were also discussed. For example, there were several participants who were concerned that the identification of genetic markers for autism would lead to an in utero test, such as that for Down syndrome, which most participants were not in favour of. Many participants believed this test would lead to pregnant individuals seeking abortions which, although they emphasised the individual's right to choose, '*I am VERY pro-choice, support abortion, and people have every right to choose …'* (003), did cause them upset, as they saw it as Autistic lives being assigned less value than neurotypical lives within society: '*Just because an Autistic person might not be 'high functioning' and needs constant support, that doesn't mean they're not enjoying life' (014).* There were concerns this could impact society's wider perception of autism and stall progress:

> *I worry that genetic testing and these studies, if they publish results and make a big deal of it, will push these positive happenings backwards.* (001)

There was a concern that genomics research had an 'end goal' with negative consequences for the Autistic community (see 1A): '*… I don't believe that once people find a genetic marker that's where it would stop'* (005). One participant questioned whether the lack of transparency with research aims was purposeful and that genomic research was kept '*shrouded in mystery'* (001) for it to continue indefinitely. Another participant reasoned that without an established end goal, genomic research would continue without creating change, reiterating the need for practical support within the community. Some participants believed that an earlier genetic test for autism may be superfluous, as it would be unable to identify the support levels needed and may not lead to the provision of support. In addition, the complexity of autism led some participants to question whether any of the stated aims of research projects *could* be achieved. One participant argued that there was enough of an understanding surrounding autism now to implement beneficial support, but this hadn't occurred, leading them to ask how much knowledge was 'enough' for this to begin:

> *… another genetic test … we do anything with that? No, we did nothing with that." So, there's a good series of evidence that you'll do another genetics test and nobody will do anything about it …* (007)

Several participants noted their preference for research which would focus on social, psychological and environmental elements within autism, rather than genomics as: '*I'm not convinced that genetic studies into autism will actually benefit Autistic people*' (017). It was often stated that genomic research, when not coupled with social and environmental factors, was a reductive approach as described by participant 006: '*You can't take the social and the Sociology out of this …*'. It was suggested real-life change would not be an outcome of research focused solely on genetics:

> *… what is the point of the study? If it is to help Autistic people then there's a better way to do it … because you're not solving Autistic people's problems, you're just unravelling a bit more genetic code.* (007)

There were appeals, within this research, for social perceptions and governmental structures to change, including: '*built environments, public spaces and services [that] we use not to be designed in ways that hurt and harm us*' (012). Without an explanation as to how genetic research can achieve such outcomes, participants were disinterested in this field of research; '*I don't care about my genetics, I care about having my needs met …*' (009). Although this participant later reflected on this, contributing:

> *… my current knowledge as a non-scientist makes me less drawn to it, but this could as much be an issue of public education about this type of research. I would like to know more about how it could impact me on a daily basis and in concrete tangible ways (for the better), and this might help me feel more engaged with it.* (009)

Who is doing what? And for whom?

There were several comments about *ethical considerations*. Several participants questioned how genomic studies could be considered ethically viable '*How these projects are ever given ethical clearance should be a major cause for concern by the entire breadth of academia*' (006). There were further comments regarding the need for protection for Autistic participants. Participant 007, for example, said that they would require further protections to be embedded within policy before they were comfortable about their involvement in research:

> *… you need much better rights and protection for Autistic people in society, and that needs to be legislated for. I mean, there is the Autism Act, which is effectively useless, there is the Autism Strategy, which as far as I know, nobody reads …* (007)

Others, however, did not share these concerns:

> *They put it to someone, an ethics committee, and that ethics committee goes yes or no. If someone explains it to me, I can either get involved or not.* (004)

One participant also reflected on this and said '*Scientists have a moral and ethical duty to consider how their work will be used*' (006). There were also concerns raised about how some Autistic people cannot provide consent to take part in such studies as: '*… decisions are being made on their behalf …*' (006). As participant 014 put it '*If they can't communicate for themselves, leave them alone*', describing any other way of dealing with a situation such as this as a '*minefield*'. Another major issue was the perceived lack of control regarding the future use of any DNA contributed beyond the initial research '*we don't know what those other studies will want to do with it*' (017). Several individuals stipulated that their involvement would only take place if:

> *… there were incredibly strict regulations in place about the limited use of the DNA for that purpose only, and it being subsequently destroyed and no option for the data to be used for any other purpose that I had not explicitly consented to.* (017)

Other conditions for taking part included providing full details of data usage and storage, data security, and confirmation that the research did not intend to use the information derived from the study to allocate, or deny support nor contribute towards a treatment or cure for autism. There were also concerns that genetic testing would negatively … *affect a specific Autistic person's comfort* (022) by not taking into account the sensory sensitivities of participants. Several participants noted that their perspective was influenced by the *poor handling of historic and current research*. As the '*track record of the medical profession'* (007), within autism research has a historically negative tint. Distrust was targeted towards research institutions and specific members of autism research teams. Participants reported that both historical and current research practices have significantly impacted trust between research teams and the Autistic, and wider autism, communities. Several individuals remarked that these issues were still evident in current autism research. Spectrum 10k was mentioned here as an example of poor handling:

> *There was insufficient information provided to people to reassure us [as] to the purpose of the data, who would store it, and what end would come from that kind of research that couldn't just come from a lot of surveys. (015)*

Participants also reported a feeling of being alienated by research institutions, reflecting on '*power structures'* (005) in which Autistic voices were '*often disregarded'* (010) in favour of parents because: '… *once you make autism a disorder, and disordered, our voices are forever disordered. If it's disordered, then it doesn't have to be taken into consideration'* (006). A sense of distrust was understandable to some, but not felt by all, further illustrating the heterogeneity even within this small representation of the Autistic population: … *by and large I suspect that the vast majority of research is all right and well-intended* (004). To attend to the diversity of the community, and potentially reduce alienation, it was reflected that there was a need for nuanced and open conversation between researchers and representatives across the community. It was added, however, that this may not be possible using social media platforms: '… *you quickly realise you're in an echo chamber and it's just reinforcing all the other things that it told you'* (004) (see 4B).

There were several comments made about *bias within research*. Participants reflected on their distrust of research institutions themselves. Several participants felt that research funding is '*skewed towards one type of research'* (021). Participant 021 acknowledged that, although funding pools were separate, a significant proportion of autism research was in the biological field, including genomics. Others were concerned genetic research was being funded over research which developed lifelong support:

> *What is genetic research going to do for me in terms of supporting me through my older years? Nothing. Absolutely nothing … why not build some residential care, supported living accommodation for Autistic people, Autistic elders, that will take our needs into consideration?* (006).

Participants were concerned about certain individuals within research teams, for example: 'I don't think he's [Baron-Cohen] covered himself in glory with past exploits, which has allowed people to become significantly cynical' (007). Participants believed researchers were influenced by the prospect of financial gain: '… all research is funded by somebody or something. The people who put money in seldom don't have an agenda' (007). For this participant, this was also true for Autistic researchers: 'I think a team of Autistic people have got more chance of having more integrity [with] that sort of stuff, but I don't think they're immune to [this] sort of influence' (007). Several participants discussed autism and Autistic representation within research. This included how autism was frequently discussed through a medical, deficit-based perspective, which most participants were against. There was a call for co-productive practices which included the Autistic voice in a respectful, accessible and meaningful sense. Co-production was described as 'in principle a good thing, but then it can be quite problematic or tokenistic' (021). Many participants hoped that Autistic inclusion within a research team would lead to a higher representation of the community's priorities and a use of language that was less medical:

> … *the autism focus means that we keep being 'othered' – why does research not frame things around different 'neurotypes' including neurotypical AND neurodivergent, to make it clear that we aren't some weird 'other'?* (009).

One participant stated they would only take part in research '… *if the entire study was run by Autistic people with a clear aim of improving Autistic people's lives* …' (005). Often, participants felt less value was attributed to their contributions, in comparison to those of professionals or parents: '… *responses from the Autistic community are blatantly ignored by the researchers - shows me that they don't care to listen to the actual community they claim to support, and if that is the case, how can I trust them?*' (017). However, a couple of participants expressed that the involvement of Autistic individuals within research was not necessary for them to take part: *'I think I would still take part … if I felt the research was beneficial to Autistics'*. (010). Overall, the lack of involvement of the Autistic voice was reiterated by participants, some of whom called for further inclusion, including employing Autistic researchers: '*Autistic people should be designing the research and driving what's needed …'* (012).

Reflecting on the Autistic voice

The collected data show *differing opinions across the community*. One participant empathised with Autistic people who may feel like a minority in their opinion within the community: '*I feel for Autistics that do have very different world views … It must be lonely for them'* (010). Several participants also speculated there to be a difference of opinion between groups within the wider autism community, such as Autistic individuals and parents, with regard to genomic studies. There was a general assumption by participants that parents of Autistic people may be more in favour of genomic studies, for example: '*I think parents …might welcome it so that they know maybe earlier*' (001). This pattern was observed in another branch of the PEAPOD that focused on parents of non-speaking or minimally speaking Autistic children (Asbury et al., 2024). There was a concern that the deficit-based, medical-focused narrative within genomic studies was capitalising on parental fear and the need to care for their children: '*… if you sell genetic treatment - you are preying on their biggest fears and their wildest hopes*' (006). There was an acknowledgement that '*parenting an Autistic child especially when you're neurotypical is going to be hard*' (005), but this was coupled with a hope, from this participant, that parents and caregivers could communicate with members of the Autistic community, rather than relying on professionals solely for their knowledge about autism. However, there was an acknowledgement that this communication wasn't as frequent nor as open as it could be: '*… I get the impression parents don't particularly want to engage with Autistic adults …*' (005).

Participants reflected on the *use of social media* for communication within the Autistic, and neurodivergent, community. Most participants used social media platforms such as Facebook and X (previously Twitter). Often participants chose to observe online debates rather than actively engage in these conversations. One reason they did not engage was because they were unsure what they could contribute '*I don't know enough about it to get stuck in*' (005) or were worried about facing social media toxicity: '*No, I generally don't join in with any discussions on social media. I just get really nervous …*' (018). Positives discussed of social media included the ease of gaining and sharing information, although it was acknowledged this information was often second-hand, without references to primary sources, frequently focusing on opinion rather than scientifically based debates, e.g.'*… lots of the stuff that's out there just isn't right. It's just people's opinions. And some of them are just meaningless*' (004). There was an acknowledgement that public understanding of genomics studies into autism may be limited by the studies which stirred controversy and were therefore spoken about more (see 1A). Although the opinions expressed by advocates over social media aligned frequently with participants' views, there was an appreciation that social media could act as an '*echo chamber*' (017), by presenting a singular narrative. Participants were aware that other voices, particularly from marginalised intersections within the community, were largely unrepresented on these platforms and across research itself, for example, '*I've seen … Black Autistic people, saying, look, the Autistic world doesn't represent me on a GOOD day*' (011).

It was suggested that views on *Autistic identity may influence opinions of genetic research*. Most participants reflected that autism was '*more than a diagnosis. It's an identity*' (004). This included recognising not only the challenges of living in a neurotypical world but also the benefits of neurodiversity '*there's so many amazing advancements made in science, technology, design, academia, directly due to neurodivergent brains, that will be lost just through to judgemental stuff based on biased opinions about a collection of genetic differences*' (006). Discussing one's Autistic identity was frequently coupled with the concern that genomic tests for autism would reduce *'the fluidity of people and human beings and the… rich diversity of people' (006).* One participant acknowledged that earlier in their journey of self-understanding, following their diagnosis, they would have been supportive of genomic research which sought to stop or lessen their Autistic characteristics: '*15 years ago … I just wanted my head to stop doing what it did … because I didn't understand why it was doing what it was doing*' (006). It was suggested by a couple of participants that those who were earlier on in their understanding of themselves as Autistic, and processing what that meant for them, may have a more medical-based viewpoint and therefore align themselves more easily with the perceived aims of genomic studies, as supported somewhat within this data: '*Originally, I probably would have taken a cure for Autism … That's not where I am now. I realise that there isn't a version of me that's possible without autism*' (015).

Discussion

This research has gathered the subjective realities of participants in relation to genomic studies of autism and demonstrates the diverse knowledge, understanding and views within the Autistic community.

The negative rhetoric

The underlying, medical-model, deficit-based narrative behind genomic studies was raised as a concern for the participants within this research. Several participants were worried that research with this narrative would influence public opinions on autism, increase the stigma already faced by the Autistic community (Han et al., 2021) and negatively impact the progress achieved by neurodiversity-affirming advocates. As demonstrated in previous research, Autistic individuals feel as though research priorities do not reflect nor coincide with the needs of the Autistic community (Pellicano et al., 2014; Roche et al., 2020). The use of the medical model rather than the social model as a framework for understanding within research (Chown et al., 2017; Poulsen et al., 2022), the latter of which a large portion of the autism and Autistic community prefer (e.g. Anderson-Chavarria, 2022), is indicative of this misalignment.

Transparency

Within this paper, a need has been established by participants for research aims, procedures and data storage information to be circulated in more transparent and accessible ways to interested parties. Social media platforms such as X have been used to share, exchange and collate information (Saha & Agarwal, 2016) and might be capitalised on for science communication but have been rife with misinformation (e.g. Aragon-Guevara et al., 2023). There seems to be a notable opposition to genomic research on various social media platforms. No participants, when asked, had seen posts which discussed genomic studies of autism positively. Further consideration is needed as to how we as researchers use social media to spread information and engage the communities we serve.

The perceived focus on genetics and the representation of community priorities

Within this research, the usefulness of genetic research, which was not multi-disciplinary in nature, was questioned. It was unclear for many participants what genomic research sought to achieve and how it could create tangible changes in areas of community importance (Frazier et al., 2018). Many participants emphasised the need for social, physical, educational, occupational, environmental and emotional support for Autistic people across the lifespan, an idea shared in current research (Frankish & Horton, 2021), looking to create tangible benefits for Autistic individuals and their families (Pellicano, 2020). As these community priorities had not been resolved by genetic research thus far, participants were unsure if further genomic progress would lead to their preferred outcomes for the community. Participants also wished to reiterate that autism was more than a medical diagnosis but also an instrumental part of a person's identity and culture, inclusive of social and environmental factors, an idea also identified in the literature (Woods, 2017; Anderson-Chavarria, 2021; Cresswell & Cage, 2019). There is a clear call for holistic autism research which generates a more comprehensive understanding of autism itself (Bolton, 2018).

Ethical considerations

The exclusion of marginalised sub-groups within decision-making is an ethical issue (Cascio et al., 2021), with the under-representation of service users' voices also being epistemologically problematic (Chown et al., 2017). There is a need for future research to include the meaningful inclusion of the Autistic voice, at all stages of the research process, to inform and empower the Autistic community, with changes at individual and systemic levels to achieve this (Pickard et al., 2022). This includes developing more effective, collaborative work between researchers, Autistic people and their allies (Nicolaidis et al., 2019; Pellicano, 2020). This also includes involving intersectional Autistic identities which have been less represented within research historically (Botha and Gillespie-Lynch, 2022; Malone et al., 2022). It is important to note here that some Autistic people will not wish to take part in research, regardless of any inclusive practices implemented, as trust between research institutions and this community may be irrevocable for some (Pellicano et al., 2014).

Recommendations

Further efforts need to be made to make research transparent. This involves clearly describing research aims, the practicalities of taking part, perceived benefits and data management procedures. It should also be communicated how funding is allocated within universities, by charities, and the government, within the field of autism. There needs to be transparency surrounding how participants, for whom parental consent is sought, are supported to provide additional assent to any studies, to respect the autonomy of the people involved. Research transparency is aided by co-productive practices which involve Autistic people on a meaningful level throughout the research process (Gower et al., 2019). Future research should work to reframe Autism as value neutral (Botha et al., 2020) and employ a neurodiversity-affirming perspective. If there are repeated concerns, made by the community, including within this research, which do not reflect how genomic research is conducted, this may be indicative of a lack of knowledge transfer between research institutions and interested groups. .

There is a notable conflict between open science movement principles, which determine best practice (McKiernan et al., 2016), and the preferences of some Autistic people regarding how their data are stored and used in research, for example: '*then some (like Spectrum 10K) also say they will share their data with other studies, and we don't know what those other studies will want to do with it*' (017). There is a need to address this conflict, incorporating the principles of open research practices, in which collaboration, engagement and accessibility are key tenets, to co-produce a way forward (Hobson et al., 2023). Whilst any changes implemented to resolve this conflict may add delays to research or may not be able to be put into practice, it would aid in re-establishing trust between researchers and the Autistic community if there were an open conversation about accommodations which could be made.

Limitations

We focused on a specific sub-group within the Autistic community with the intention of this project being one in a series. Unfortunately, due to word restrictions, and the volume of data gathered, the relationship between identity and perceptions of autism was unable to be explored fully and warrants a separate paper from this study. Due to the lack of demographic information, it is harder to determine how applicable the data are to a wider population. It is likely that the opinions we gathered do not represent those of all the Autistic community and we acknowledge that this is one sub-group of individuals. We hope to conduct an equivalent study of other sub-groups within the community including Autistic people with learning difficulties, which is an under-represented group within autism research (Russell et al., 2019). In future research, demographic information will be sought. It would also be useful to engage with Autistic people with genetic disorders as this research may impact them most, as opposed to this study's population, who may derive fewer benefits from this field of work.

Conclusion

A great deal of work is needed to reconnect the Autistic and research communities, to ensure the Autistic voice is heard, included and respected through inclusive research practices. This could start with re-establishing trust through transparent and accessible communication. There is a need to prioritise the wants of the community so that research teams can truly support the people they purport to serve. This is perhaps particularly important in relation to genomic studies of autism in which the weight of history is against us. Spectrum 10k, as an example, is currently working to realign its efforts with the Autistic community, to clarify its research scope and to reassure and involve the Autistic community in future consultations (Dattaro, 2021; National Autistic Society, 2022). How, and to what extent, these consultations will shape the research moving forward is yet to be known.

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References

Aims-2-Trials. (2020). *About us*. Aims 2 trials. https://www.aims-2-trials.eu/about-aims-2-trials/

Anderson-Chavarria, M (2021). The autism predicament: models of autism and their impact on autistic identity. *Disability & Society*, *37*(8), 1321–1341. https://doi.org/10.1080/09687599.2021.1877117

Aragon-Guevara, D., Castle, G., Sheridan, E., & Vivanti, G. (2023). The reach and accuracy of information on autism on TikTok. *Journal of Autism and Developmental Disorders*, 1–6. https://doi.org/10.1007/s10803-023-06084-6

Asbury, K., Toseeb, U., & Barrow, N. (2024). What do parents of nonverbal and minimally verbal autistic children think about genomic autism research? *Autism*, *28*(7), 1838–1846. https://doi.org/10.1177/13623613231213431

Aucademy. (2021). Boycott Spectrum 10k. https://aucademy.co.uk/2021/09/03/boycott-spectrum-10k-please-sign/.

Autism Speaks. (2024). Research. Available: https://www.autismspeaks.org/research.

Autistic Self-Advocacy Network. (2022). ASAN statement on genetic research and autism. 1-13. https://autisticadvocacy.org/wp-content/uploads/2022/03/genetic-statement-concerns.pdf

Bolton, M. J. (2018). With the silence of a thousand cries: Extremes of autistic advocacy. *Disability & Society*, *33*(6), 980–984. https://doi.org/10.1080/09687599.2018.1454381

Botha, M. (2021). Academic, activist, or advocate? Angry, entangled, and emerging: A critical reflection on autism knowledge production. *Frontiers in Psychology*, *12*. https://doi.org/10.3389/fpsyg.2021.727542

Botha, M., & Cage, E. (2022). "Autism research is in crisis": A mixed method study of researcher's constructions of autistic people and autism research. *Frontiers in Psychology*, *13*. https://doi.org/10.3389/fpsyg.2022.1050897

Botha, M., Dibb, B., & Frost, D. M. (2020). "Autism is me": An investigation of how autistic individuals make sense of autism and stigma. *Disability & Society*, *37*(3), 427–453. https://doi.org/10.1080/09687599.2020.1822782

Botha, M., & Gillespie-Lynch, K. (2022). Come as you are: Examining autistic identity development and the neurodiversity movement through an intersectional lens. *Human Development*, *66*, 93–112. https://doi.org/10.1159/000524123

Botha, M., Hanlon, J., & Williams, G. (2021). Does language matter? Identity-first versus person-first language use in autism research: A response to vivanti. *Journal of Autism and Developmental Disorders*, 53(2), 870–878. https://doi.org/10.1007/s10803-020-04858-w

Braun, V., & Clarke, V. (2022). Conceptual and design thinking for thematic analysis. *Qualitative Psychology*, *9*(1), 3–26. https://doi.org/10.1037/qup0000196

Byres, L., Morris, E., & Austin, J. (2023). Exploring autistic adults' perspectives on genetic testing for autism. *Genetics in Medicine*, *25*(8), 100021. https://doi.org/10.1016/j.gim.2023.100021

Cascio, M. A., Weiss, J. A., & Racine, E. (2021). Making autism research inclusive by attending to intersectionality: A review of the research ethics literature. *Review Journal of Autism and Developmental Disorders*, *8*, 22–36. https://doi.org/10.1007/s40489-020-00204-z

Chown, N., Robinson, J., Beardon, L., Downing, J., Hughes, L., Leatherland, J., Fox, K., Hickman, L., & MacGregor, D. (2017). Improving research about us, with us: A draft framework for inclusive autism research. *Disability & Society*, *32*(5), 720–734. https://doi.org/10.1080/09687599.2017.1320273

Clarke, V., & Braun, V. (2013). *Successful Qualitative Research: A Practical Guide for Beginners*. SAGE Publications.

Cresswell, L., & Cage, E. (2019). Who Am I?': An Exploratory Study of the Relationships Between Identity, Acculturation and Mental Health in Autistic Adolescents. *Journal of Autism and Developmental Disorders*, *49*, 2901–2912. https://doi.org/10.1007/s10803-019-04016-x

Dattaro, L. (2022). *Spectrum 10K gets green light from ethics agency*. Spectrum News. https://www.spectrumnews.org/news/spectrum-10k-gets-green-light-from-ethics-agency/

Fletcher-Watson, S., Adams, J., Brook, K., Charman, T., Crane, L., Cusack, J., Leekam, S., Milton, D., Parr, J. R., & Pellicano, E. (2018). Making the future together: Shaping autism research through meaningful participation. *Autism*, *23*(4), 943–953. https://doi.org/10.1177/1362361318786721

Frankish, H., & Horton, R. (2021). A way forward to improve the lives of autistic people. *The Lancet*, *399*(10321), 215–217. https://doi.org/10.1016/S0140-6736(21)02735-5

Frazier, T. W., Thompson, L., Youngstrom, E. A., Law, P., Hardan, A. Y., & Eng, C. (2014). A twin study of heritable and shared environmental contributions to autism. *Journal of Autism and Developmental Disorders*, *44*(8), 2013–2025. https://doi.org/10.1007/s10803-014-2081-2

Fryer, T. (2022). A critical realist approach to thematic analysis: Producing causal explanations. *Journal of Critical Realism*, *21*(4), 365–384. https://doi.org/10.1080/14767430.2022.2076776

Gallion, T., Williams, Z. J., Niarchou, M., Duncan, L., Hooker, G., & Taylor, K. A. (2024). Attitudes of autistic adults toward genetic testing for autism. *Journal of Genetic Counseling*, 1–15. https://doi.org/10.1002/jgc4.1918

Gowen, E., Taylor, R., Bleazard, T., Greenstein, A., Baimbridge, P., & Poole, D. (2020). Guidelines for conducting research studies with the autism community. *Autism Policy Practice*, *2*(1), 29–45. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7099931/

Han, E., Scior, K., Avramides, K., & Crane, L. (2021). A systematic review on autistic people's experiences of stigma and coping strategies. *Autism Research*, *15*(1), 12–26. https://doi.org/10.1002/aur.2652

Hare, J. (2021). Does The Spectrum 10K Autism Study Endanger Human Rights? *Each Other*. https://eachother.org.uk/does-the-spectrum-10k-autism-study-endanger-human-rights/

Hobson, H., Linden, A., Crane, L., & Kalandadze, T. (2023). Towards reproducible and respectful autism research: Combining open and participatory autism research practices. *Research in Autism Spectrum Disorders*, *106*, 102196. https://doi.org/10.1016/j.rasd.2023.102196

Lilley, R., Lawson, W., Hall, G., Mahony, J., Clapham, H., Heyworth, M., Arnold, S. R., Trollor, J. N., Yudell, M., & Pellicano, E. (2022). 'A way to be me': Autobiographical reflections of autistic adults diagnosed in mid-to-late adulthood. *Autism*, *26*(6), 1395–1408. https://doi.org/10.1177/13623613211050694

Luterman, S. (2020). *The biggest autism advocacy group is still failing too many autistic people*. The Washington Post. https://www.washingtonpost.com/outlook/2020/02/14/biggest-autism-advocacy-group-is-still-failing-too-many-Autistic-people/

Malone, K. M., Pearson, J. N., Palazzo, K. N., Manns, L. D., Rivera, A. Q., & Martin, D. L. M. (2022). The scholarly neglect of Black autistic adults in autism research. *Autism in Adulthood*, 271–280. https://doi.org/10.1089/aut.2021.0086

McKiernan, E. C., Bourne, P. E., Brown, C. T., Buck, S., Kenall, A., Lin, J., McDougall, D., Nosek, B. A., Ram, K., Soderberg, C. K., Spies, J. R., Thaney, K., Updegrove, A., Woo, K. H., & Yarkoni, T. (2016). How open science helps researchers succeed. *eLife*, *5*, e16800. https://doi.org/10.7554/eLife.16800

Milton, D. (2014). Autistic expertise: A critical reflection on the production of knowledge in autism studies. *Autism*, *18*(7), 794–802. https://doi.org/10.1177/1362361314525281

Milton, D., Mills, R., & Pellicano, E. (2014). Ethics and autism: where is the autistic voice?*Commentary on Post et al. J Autism Dev Disord*, *44*(10), 2650–2651. https://doi.org/10.1007/s10803-012-1739-x

MSSNG. (2024). *About*. Autism Speaks. https://research.mss.ng/

National Autistic Society. (2022). *Spectrum 10K (updated 14.06.22).* The National Autistic Society. https://www.autism.org.uk/what-we-do/news/spectrum-10k

Natri, H. M. (2021). Spectrum 10K and the questionable past, present, and future of genetic autism research. https://doi.org/10.13140/RG.2.2.14973.28642

Nicolaidis, C., Raymaker, D., Kapp, S. K., Baggs, A., Ashkenazy, E., McDonald, K., Weiner, M., Maslak, J., Hunter, M., & Joyce, A. (2019). The AASPIRE practice-based guidelines for the inclusion of autistic adults in research as co-researchers and study participants. *Autism*, *23*(8), 2007–2019. https://doi.org/10.1177/1362361319830523

Pellicano, E. (2020). Commentary: Broadening the research remit of participatory methods in autism science – a commentary on Happé and Frith (2020). *Journal of Child Psychology and Psychiatry*, *61*(3), 233–235. https://doi.org/10.1111/jcpp.13176

Pellicano, E., Dinsmore, A., & Charman, T. (2014). What should autism research focus upon? Community views and priorities from the United Kingdom. *Autism*, *18*(7), 756–770. https://doi.org/10.1177/1362361314529627

Pickard, H., Pellicano, E., den Houting, J., & Crane, L. (2022). Participatory autism research: Early career and established researchers' views and experiences. *Autism*, *26*(1), 75–87. https://doi.org/10.1177/13623613211019594

Poulsen, R., Brownlow, C., Lawson, W., & Pellicano, E. (2022). Meaningful research for autistic people? Ask autistics! *Autism*, *26*(1), 3–5. https://doi.org/10.1177/13623613211064421

Pring, J. (2021). *Autistic campaigners' anger over Spectrum 10k protest lock-out and 'score tactics'*. Disability News Service. https://www.disabilitynewsservice.com/Autistic-campaigners-anger-over-spectrum-10k-protest-lock-out-and-scare-tactics/

Roche, L., Adams, D., & Clark, M. (2021). Research priorities of the autism community: A systematic review of key stakeholder perspectives. *Autism*, *25*(2), 336–348. https://doi.org/10.1177/1362361320967790

Russell, G., Mandy, W., Elliott, D., White, R., Pittwood, T., & Ford, T. (2019). Selection bias on intellectual ability in autism research: a cross-sectional review and meta-analysis. *Molecular Autism*, *10*(9). https://doi.org/10.1186/s13229-019-0260-x

Saha, A., & Agarwal, N. (2016). Modeling social support in autism community on social media. *Network Modeling Analysis in Health Informatics and Bioinformatics*, *5*(8). https://doi.org/10.1007/s13721-016-0115-8

Spectrum 10k. (2022). University of Cambridge. Available: https://spectrum10k.org/.

Willingham, E. (2013, November). *Why autism speaks doesn't speak for me*. Forbes. https://www.forbes.com/sites/emilywillingham/2013/11/13/why-autism-speaks-doesnt-speak-for-me/?sh=361c37e23152

Woods, R. (2017). Exploring how the social model of disability can be re-invigorated for autism: In response to Jonathan Levitt. *Disability & Society*, *32*(7), 1090–1095. https://doi.org/10.1080/09687599.2017.1328157

Woods, R., Milton, D., Arnold, L., & Graby, S. (2018). Redefining critical autism studies: A more inclusive interpretation. *Disability & Society*, *33*(6), 974–979. https://doi.org/10.1080/09687599.2018.1454380