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## Genetic Testing for Autism: The Autistic Adult Perspective

Thomas Scott Dent

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# GENETIC TESTING FOR AUTISM: THE AUTISTIC ADULT PERSPECTIVE

by

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Bachelor of Science  
University of Michigan, 2019

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## ABSTRACT

Many professional medical organizations recommend some level of genetic testing as standard of care for those diagnosed with autism spectrum disorder. However, genetic testing related to the diagnosis of autism is seen as controversial by many in the autistic community. While opinions about genetic testing have been well-documented from the perspective of the parents of autistic children, our understanding of the autistic adult perspective remains limited. We implemented a descriptive, web-based survey of autistic adults to assess their awareness of, attitudes towards, and interests in genetic testing for autism (n = 145). Our data demonstrated that half of our participants are unfamiliar with genetic testing for autism, and 86% are unaware of current medical guidelines recommending genetic testing for autism. A minority of participants held an overall positive opinion towards genetic testing (19%) and want genetic testing for themselves (17%). Furthermore, none of the commonly perceived benefits of testing were endorsed as benefits by a majority of participants. Regarding the potential development of a prenatal genetic test for autism, 16% are in support of its development and 74% are concerned that it could lead to an increase in terminations of pregnancies suspected to develop autism. These findings highlight a disconnect in perspectives about genetic testing between autistic individuals, their parents, and the medical community. Future research should continue to explore the autistic adult perspective on this topic and ensure that autistic voices are heard during the development of new tests and professional guidelines involving genetic testing related to autism.

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## CHAPTER 1: BACKGROUND

### 1.1 Defining Autism Spectrum Disorder

#### *1.1.1 Clinical Description and Etiology of Autism Spectrum Disorder*

Autism spectrum disorder (ASD) is a neurodevelopmental disorder characterized by deficits in social communication, restricted interests, and repetitive behaviors. ASD is among the most common developmental disorders and is estimated to affect 1.7% - 2.21% of the U.S. population (Dietz et al., 2020; Hyman et al., 2020). As of 2020, there were approximately 5.4 million autistic adults living in the United States (Dietz et al., 2020). The prevalence of ASD has markedly increased in the past three decades, both in the United States and globally (Salari et al., 2022). A specific cause for this increase is unknown; however, it is believed to be due to a combination of an increased awareness of ASD, implementation of public health measures such as pediatric screenings, and alterations to the diagnostic criteria (Salari et al., 2022). ASD is a life-long condition with symptoms presenting as early as 12 months of age and the current average age of diagnosis being 4 years of age (Tanner & Dounavi, 2021). Autism occurs in all socioeconomic, racial, and ethnic groups, and is four times more common in males than in females (Volkert et al., 2022).

The distinctive social features of ASD include deficits in social-emotional reciprocity, nonverbal communication, and developing or maintaining relationships (American Psychiatric Association [APA], 2013). The criterion is further supported by the presence of stereotyped or repetitive movements, inflexible adherence to routine,

highly restricted interests, and hyper- or hyporeactivity to sensory input (APA, 2013). These deficits result in a wide range of manifestations that vary across the lifespan. The earliest signs of autism may include poor eye contact, reduced facial expressions, delayed speech, and a lack of showing or sharing objects (Hodges et al., 2020). School-aged children with autism may display concrete or literal thinking, odd or excessive fixations on certain objects, difficulty understanding emotions, and a lack in conversational skills with their peers (Hodges et al., 2020). The transition period from adolescence to adulthood is typically associated with an improvement in social skills, adaptive functioning, and independence (Howlin 2021). However, autistic individuals may continue to experience a variety of challenges in emotional, behavioral, and adaptive functioning throughout their adult lives (DaWalt 2021).

In recent years, it has become increasingly apparent that autistic adults have unmet social and healthcare needs (Weir et al., 2022). Autistic adults also face levels of unemployment significantly higher than the general population, even for those with IQ scores comparable to or higher than the general population (Maslahati et al., 2022; Murphy et al., 2016). As a result of their functional impairments, unmet needs, and high unemployment, autistic individuals have scored significantly lower on quality-of-life assessments compared to the general population (Oakley et al., 2020). However, it is important to note that ASD is highly variable with regards to its range and severity of symptoms in affected individuals, and not all autistic individuals experience poor outcomes in society (Griesi-Oliveira & Sertié, 2017). Many autistic adults play to their strengths in perceptual processing, attention to detail, and enthusiasm for their interests to make remarkable contributions to society and attain a higher satisfaction in life (Motttron

et al., 2009; Schipper et al., 2015). Moreover, autistic adults comprise some of the most brilliant minds known to the arts, mathematics, sciences, and innovation (Happé & Frith, 2009).

### *1.1.2 Etiology of Autism Spectrum Disorder*

ASD is a complex, multifactorial disorder that results from both genetic and environmental risk factors (Park et al., 2016). The genetic contributions to ASD are highly heterogeneous with hundreds of genes implicated in conferring an increased risk (Satterstrom et al., 2020). Many of the proteins encoded by these genes converge into shared biological pathways for either neuronal development, axonal guidance, synaptic function, or chromatin remodeling (Fernandez & Scherer, 2017). The types of genetic variants associated with autism include copy number variants, single nucleotide polymorphisms, trinucleotide repeats, and epigenetic alterations which primarily affect synaptic function, brain development, and metabolism (Genovese & Butler, 2020; Rylaarsdam & Guemez-Gamboa, 2019).

Autism may either be found as part of a syndrome or as an isolated feature. Isolated, or nonsyndromic, autism comprises the majority of cases (75-80%) (Ivanov et al., 2015). The Simons Foundation Autism Research Initiative (SFARI) gene database currently contains over 900 genes associated with non-syndromic autism, and these are categorized based on the confidence rate for their role in ASD (Sauer et al., 2021). Category 1 genes have the highest confidence rate and currently contains 207 genes (Sauer et al., 2021). This is followed by 211 genes in category 2 and 506 genes in category 3 (Sauer et al., 2021). The vast majority of individuals with nonsyndromic

autism are believed to have variants in several of these genes, rather than in only one gene.

Syndromic autism is defined as a disorder with a clinically defined pattern of somatic abnormalities and a neurobehavioral phenotype that may include autism (Fernandez & Scherer, 2017). Syndromic autism occurs from either copy number variants (CNVs), monogenic conditions, mitochondrial disorders, or chromosomal aneuploidies. ASD-associated chromosomal aneuploidies are identified in up to 2% of autism cases with the most common being Down syndrome, sex chromosome aneuploidy (i.e., Klinefelter syndrome), and a supernumerary isodicentric chromosome 15 involving the Prader Willi / Angelman Syndrome region (Miles, 2015; Wassink & Patil, 2001). CNVs are found in 7-14% of patients with ASD and the most common findings are in the following loci: 16p11.2, 1q21.1, 15q13.3, 17p11.2, 22q11.2, and 16p13.1 (Moreno-De-Luca, 2013; Wiśniowiecka-Kowalik & Nowakowska, 2019). In individuals harboring these CNVs, autism co-occurs frequently with intellectual disability, epilepsy, dysmorphic facial features, and congenital heart defects (Genovese & Butler, 2020; Wiśniowiecka-Kowalik & Nowakowska, 2019). Monogenic disorders account for 5-10% of autism cases and these genes are primarily involved in regulating the expression of a large group of other genes (Wiśniowiecka-Kowalik & Nowakowska, 2019). Fragile X syndrome is the most common autism-related monogenic syndrome (1.5-3% of ASD cases), followed by Tuberous Sclerosis Complex (1%), Rett syndrome (1%), *PTEN* Hamartoma syndrome, and Neurofibromatosis Type 1 (Genovese & Butler, 2020; Wiśniowiecka-Kowalik & Nowakowska, 2019).

In recent years, researchers have identified a strong connection between autism and mutations in both the mitochondrial DNA (mtDNA) and nuclear genes involved in mitochondrial maintenance (Valiente-Palleja et al., 2018; Wang et al., 2022). An ongoing meta-analysis has revealed that mitochondrial disease is found in approximately 5% of autistic children and that abnormal mitochondrial biomarkers are found in 30-50% of autistic children (Balachandar et al., 2021; Rossignol et al., 2012). Considering the mitochondria's essential role in energy production for the brain, these new insights make mitochondrial dysfunction a promising field of study in clarifying the etiological landscape of ASD.

As previously mentioned, ASD is a multifactorial disorder and there is an apparent contribution of several non-genetic risk factors. The environmental exposures that have been linked to autism are believed to have a mutagenic effect that increases the likelihood of *de novo* deleterious mutations, especially in the context of prenatal development (Pugsley et al., 2021). This is supported by the finding that approximately 5-15% of ASD patients have *de novo* mutations that are presumed to be implicated in the pathogenesis of the disease (Iossifov et al., 2014). Prenatal contributions linked to ASD include maternal deficiencies in folic acid and zinc, maternal exposure to toxic metals (i.e., mercury and lead) and medications (i.e., valproate), and maternal immune response to infections (i.e., rubella) (Sauer et al., 2021). The increased *de novo* mutational burden in ASD probands may also occur prior to conception, as advanced paternal age and parental exposure to toxicants are known to increase the mutation rate in germline cells (Pugsley et al., 2022). Although numerous other non-genetic risk factors have been weakly associated with ASD, the high heritability estimates for ASD (70-90%)

demonstrate the relevance of genetics in the pathogenesis of the disorder (Eapen et al., 2013).

Due to its complex etiology and strong genetic contribution, ASD co-occurs frequently with other psychiatric and medical conditions (Bougeard et al., 2021). A recent review study revealed that between 54-94% of autistic individuals have at least one comorbid psychiatric condition (Hossain et al., 2021). Among these, the most common comorbidities are sleep disorders (50-73%), anxiety (42%), depression (37%), ADHD (30-50%), and OCD (22%) (Hodges et al., 2020; Hossain et al., 2021). Among medical and neurological comorbidities, the most common are developmental delays (83%), gastrointestinal disorders (46-84%), intellectual disability (31%), and epilepsy (10-30%) (Al-Beltagi, 2021; Hodges et al., 2020). Researchers have also demonstrated the negative impact that these comorbidities can have on autistic individuals and their family members, particularly in terms of economic burden and quality of life outcomes (Buescher et al., 2014; Oakley et al., 2021).

## **1.2 Genetic Testing for Autism Spectrum Disorder**

Due to the prognostic value of genetic testing in anticipating the potential comorbidities of ASD and guiding clinical management, five specialty organizations have created recommendations related to a genetics evaluation for ASD (Barton et al., 2018). While each of these differ in their approach, genetic testing is generally recommended for every autistic individual in a stepwise manner. For example, the American College of Medical Genetics (ACMG) recommends beginning this genetics evaluation with a dysmorphology assessment and documentation of a three-generation pedigree (Schaefer et al., 2013). If the clinical indicators suggest a particular genetic

condition, then targeted testing is warranted as the first-tier test (Schaefer et al., 2013). If not, a chromosomal microarray analysis (CMA) in addition to Fragile X syndrome testing for males are recommended as the first tier, followed by Rett syndrome (*MECP2*) sequencing for females, Rett syndrome (*MECP2*) duplication testing for males, or *PTEN* Hamartoma syndrome (*PTEN*) sequencing if the patient's head circumference is  $\geq 2.5$  standard deviations above the mean (Schaefer et al., 2013). Magnetic resonance imaging (MRI) of the brain can be an important tool in this etiological workup, but is indicated only in the presence of specific indicators such as microcephaly, developmental regression, and seizures (Schaefer et al., 2013). Other professional societies have a less regimented testing algorithm as their recommendation. The American Academy of Child and Adolescent Psychiatry, for example, recommends a medical exam with genetic testing that may include G-banded karyotyping, Fragile X syndrome testing, and CMA (Volkmar et al., 2014). The American Academy of Pediatrics (AAP) recommends CMA with Fragile X syndrome testing, followed by whole exome sequencing (WES) (Hyman et al., 2020).

While there is no one genetic testing algorithm that best suits every patient, genetic testing related to the diagnosis of ASD typically begins with a CMA due to its high diagnostic yield for ASD and other neurodevelopmental disorders (Genovese & Butler, 2020; Schaefer et al., 2013). CMA has been reported to reveal copy number variants (CNVs) in approximately 17-42% of individuals with ASD (Hyman et al., 2020). However, CNVs are often poorly understood in their relation to ASD, so a pathogenic CNV is only found in 5.4-14% of individuals with ASD (Genovese & Butler, 2020; Hyman et al., 2020). Fragile X syndrome is the most common monogenic cause for ASD



and is a low-cost testing option, so it is often included as a first-tier test during the etiological workup (Harris et al., 2020; Kaufmann et al., 2017). This test requires CGG trinucleotide repeat expansion analysis of the *FMRI* gene, which cannot be detected by CMA, so it is typically run concurrently with CMA (Hyman et al., 2020). The pathogenic yield of this testing is approximately 1-5% for autistic individuals (Harris et al., 2020; Schaefer et al., 2013). Whole exome sequencing (WES) is among the most comprehensive genetic tests and is able to detect single nucleotide variants and certain copy number variants in the exons of the nuclear genome. Traditionally, WES has been utilized after CMA and Fragile X syndrome testing have been nondiagnostic; however, there has been a recent push to make WES the first-tier testing option due to its slightly higher diagnostic yield (Moreno-De-Luca et al., 2020; Srivastava et al., 2020). The diagnostic yield of WES in clinical populations of ASD has been reported to be between 8-20% (Retterer et al., 2016; Tammimies et al., 2015). However, due to the broad coverage of WES, a far greater proportion are found to have variants of uncertain significance (VUS) (Hyman et al., 2020). For this reason, pre- and post-test counseling is highly recommended to manage expectations and provide appropriate interpretation of the results (Hyman et al., 2020). Using a combined genetic testing approach, biochemical testing, clinical phenotype, and family history, an underlying genetic etiology is now identified in 30-53% of autistic patients (Genovese & Butler, 2020; Lucas et al., 2022).

Concerns regarding discrimination based on genetic results are not specific to the autistic community (Feldman, 2012). Recognizing the possibility that certain entities may choose to discriminate, the United States Congress passed the Genetic Information Nondiscrimination Act of 2008 (GINA) to protect genetic testing recipients and the

morality of precision medicine. GINA prohibits health insurers from using genetic information to adjust premiums, deny coverage, or impose preexisting condition exclusions. Likewise, GINA prohibits employers with 15 or more employees from acquiring genetic results and using them to make decisions about hiring, compensation, and other conditions of employment. This federal law does not apply to life, disability, or long-term care insurance. The protections and limitations of GINA can be paramount to one's decision to receive genetic testing, so it is critical for providers to discuss GINA during pre-test counseling.

### **1.3 Perceptions and Awareness of Genetic Testing for ASD**

#### *1.3.1 Providers' Perspective about Genetic Testing for Autism*

From a clinical standpoint, the potential benefits of genetic testing for autism have been well described and primarily include: 1) understanding the etiology, 2) prognostic predictions, 3) early treatment interventions, and 4) informing reproductive decision making (Hens et al., 2016; Hyman et al., 2020). Identifying a genetic etiology can be important for several reasons. It can provide physicians and family members with more information about recurrence risks, guide patients and families to condition-specific resources, prevent unnecessary medical tests, and lead to the identification and treatment of comorbid health conditions (Hyman et al., 2020). A retrospective chart review of 500 autistic toddlers at Boston's Children's Hospital found that 72% of the toddlers with a pathogenic or likely pathogenic variant had subsequent medical recommendations implemented into their care (Harris et al., 2020). However, despite these potential benefits, the clinical utility of genetic testing for ASD has been brought into question by several providers in the medical community. It is not always clear what genetic

information can help to predict for autistic individuals and how this can impact their medical care (DeThorne & Ceman, 2018; Lucas et al., 2022). Due to a number of factors, including a relatively low diagnostic yield and high degree of uncertainty following particular genetic findings, some experts suggest that genetic testing does not provide an organic improvement in symptoms for the majority of autistic individuals who receive testing (Hanish et al., 2018).

Some researchers have also suggested that another factor preventing genetic testing for autism is the relatively low genetic literacy amongst healthcare providers who provide care for autistic individuals (Little & Gunter, 2021). Beyond this, the current medical guidelines on genetic testing for ASD present conflicting information about when a referral should be made, and which provider should make the genetics referral. (Li et al., 2016). As a result, it has been identified that the uptake of genetic testing is far lower than expected in practice (Barton et al., 2018). A collection of studies in the past decade have reported that approximately one third (22-41%) of autistic patients had received any type of genetic testing (Cuccaro et al., 2014; Kiely et al., 2016; Vande Wydeven et al., 2012; Zhao et al., 2019). Another study examining self-reported and medical record data from 1,280 autistic patients found that only 3% received a multi-step testing approach such as the ACMG recommendations described above (Moreno-De-Luca et al., 2020). While this may, in part, be explained by limitations in clinician knowledge and comfort with genetic testing, the low uptake of autism-related genetic testing has spawned a new area of research investigating parental awareness and perceptions of genetic testing for autism.

### *1.3.2 Parental Awareness and Perspectives about Genetic Testing for Autism*

Parental awareness of genetic testing for ASD is limited, with two studies reporting that only 37% and 53% of parents were previously aware of autism-related genetic testing (L. S. Chen et al., 2013; Zebolsky et al., 2020). In a study conducting semi-structured interviews with 20 parents of autistic children, 95% reported a lack of knowledge about the availability of genetic testing for autism despite 70% being aware that genetics was a cause for autism (Hanish et al., 2018). Despite the generally low awareness of testing among parents of autistic children, two studies involving parents of autistic children have demonstrated that 83-96% were interested in pursuing genetic testing for their child (Li et al., 2016; Wagener et al., 2020). Furthermore, in a large Turkish sample of 951 parents, 87% reported that they would pursue genetic testing if it could identify a cause for their child's ASD, and 84% of parents said that genetic testing was a key part of the diagnostic process (Ayhan et al., 2021).

To our knowledge, there have been at least nine studies in the past decade that have assessed parental perspectives about genetic testing for autism. While they each differ in their exact approach, they have all inquired about the perceived benefits and barriers towards pursuing genetic testing for their children. The most commonly reported perceived benefit with genetic testing is to better understand the cause of their child's autism (L. S. Chen et al., 2013; Lucas et al., 2022; Reiff et al., 2017; Reiff et al., 2015; Xu et al., 2016). A common follow up to this finding was the mention of how obtaining an etiological label for their child helped to reduce guilt within the family (Hanish et al., 2018; Lucas et al., 2022; Reiff et al., 2015). However, a subset of these parents mentioned how this genetic information played into, but did not entirely change, their

current narrative about their child's autism, thereby showing the resilience a family's narrative may have in the face of new or conflicting genetic information (Reiff et al., 2017).

Interestingly, parents were able to foresee benefit to the testing regardless of the outcome, as some mentioned a positive result may lead to better acceptance of their child's difference, whereas others mentioned a negative result may increase their family's hope for a better prognosis (Narcisa et al., 2013; Reiff et al., 2017; Reiff et al., 2015). Another commonly perceived benefit is that genetic testing may identify potential comorbidities and lead to targeted medical care for their child (L. S. Chen et al., 2013; Hanish et al., 2018; Lucas et al., 2022; Reiff et al., 2015; Wagener et al., 2020; Xu et al., 2016). Parents also agreed that identifying a genetic change could improve access to various supports and services such as early intervention (Hanish et al., 2018; Lucas et al., 2022; Narcisa et al., 2013; Reiff et al., 2015; Wagener et al., 2020; Xu et al., 2016). Obtaining genetic information may also inform family planning, both in deciding whether or not to have more children, and in preparing for a similar outcome if future children were to be at risk for autism (L. S. Chen et al., 2013; Hanish et al., 2018; Reiff et al., 2017; Reiff et al., 2015; Xu et al., 2016;). A final commonly perceived benefit is that testing may contribute to our understanding of autism genetics and consequently benefit other autistic children and family members in the future (Hanish et al., 2018; Reiff et al., 2017; Wagener et al., 2020; Xu et al., 2016).

As previously mentioned, the majority of parents in these studies have indicated that they would like to pursue genetic testing for their autistic child. Nevertheless, a considerable proportion of parents (13-38%) would reportedly decline the option to

pursue genetic testing for their child (Ayhan et al., 2020; L. S. Chen et al., 2013). To understand why, it is helpful to explore the perceived barriers and negative attributes of genetic testing for autism that have been reported from this same set of nine studies. One of the most commonly cited reasons to oppose genetic testing is a perceived lack of benefit for the child and family (L. S. Chen et al., 2013; Narcisa et al., 2012; Reiff et al., 2017). Another commonly cited reason is the psychological distress that may occur from awaiting genetic testing results and from receiving an uncertain result (VUS) (L. S. Chen et al., 2013; Narcisa et al., 2012; Reiff et al., 2017; Xu et al., 2016). Of note, even a negative result may be confusing for families who had previously been informed about the multifactorial inheritance of autism, and now think that a genetic causation has been ruled out (Reiff et al., 2015). This highlights the need for pre- and post-test counseling to set expectations and deliver appropriate interpretation of the results (DeThorne & Ceman, 2018; Reiff et al., 2015). While some parents find the identification of a genetic etiology to aid in their acceptance of their child's condition, others have reported concerns about putting a label on their child due to the stigma associated with genetic conditions (L. S. Chen et al., 2013; Narcisa et al., 2012; Reiff et al., 2017).

Furthermore, a genetic finding may infer a sense of immutability for their child's condition which may discourage a family from taking action to improve their child's quality of life (L. S. Chen et al., 2013). The tendency to attribute genetic contributions to individual differences without acknowledging contextual factors, such as stigma and discriminatory practices, is referred to as "geneticization" and may have an adverse impact on individual and familial perspectives about autism (DeThorne & Ceman, 2018; Scully, 2008). This may also lead to blame and increased tensions within the family

regarding the inheritance of genetic traits (L. S. Chen et al., 2013; Reiff et al., 2017; Reiff et al., 2015). Parents also expressed concern regarding financial barriers such as a lack of insurance coverage for testing, as well as the possibility that a genetic finding may negatively impact eligibility for and costs of insurance in the future (L. S. Chen et al., 2013; Johannessen et al., 2017; Wagener et al., 2020; Zebolsky et al., 2020). Considering the sensory concerns experienced by autistic individuals, parents have also reported that the process of obtaining a sample for genetic testing may be painful and stressful for the autistic child (Hanish et al., 2016; Xu et al., 2016). For this reason, 92% of parents in one study cited their preference for a saliva sample over a blood draw for their child's genetic test (Wagener et al., 2020). A final set of barriers perceived by parents of autistic children include the cost of testing, transportation and scheduling issues, mistrust in healthcare providers, and privacy concerns (L. S. Chen et al., 2013; DeThorne & Ceman, 2018; Hanish et al., 2018; Xu et al., 2016).

### *1.3.3 First Person Perspective about Genetic Testing for Autism*

Genetic testing for autism primarily occurs in the childhood years for autistic individuals and is most often under the discretion of the child's parents. For this reason, the majority of research exploring the views and impact of genetic testing for autism has been focused on the parents' perspective. However, to our knowledge, only one study to date has evaluated these metrics from the perspective of autistic individuals themselves (Byres et al., 2023). This gap in understanding has become increasingly apparent in recent years as the autistic community has voiced their concern about being underrepresented in autism research (DeThorne & Ceman, 2018; Leadbitter et al., 2021). Furthermore, considering the current clinical recommendations for genetic testing by

several major medical organizations, and the resulting high volume of autism-related genetic testing, it can be argued that the lack of autistic perspective on this topic is incongruent with its clinical application.

The first attempt at formally assessing autistic adults' perspective about this topic occurred very recently, when 461 autistic adults were surveyed regarding their experiences with, attitudes toward, and interest in genetic testing for autism (Byres et al., 2023). Of the 24 participants who had received genetic testing, there were mixed experiences regarding the tolerability of testing, and several of these individuals reported receiving insufficient explanation about the reasons for testing and the results from the testing. Regarding interests for genetic testing for autism, 48% of participants indicated they would not have wanted genetic testing as a child, and 56% stated they would not want autism related genetic testing for any of their current or future children. Participants were also surveyed regarding when genetic testing should be offered, to which 74% agreed that testing should only be offered if the autistic individual themselves is able to consent, regardless of age. To this end, only 28% agreed/strongly agreed that parents should be able to make the decision for their child. Furthermore, 35% agreed/strongly agreed that testing should be routinely offered to autistic adults and 26% agreed/strongly agreed that testing should be routinely offered to autistic children. Approximately half (49%) agreed/strongly agreed that genetic testing should not be done at all for autism (Byres et al., 2023).

This study also inquired about autistic adults' perception of the benefits and harms of genetic testing, to which 39% felt that there were only harms associated with genetic testing for autism, both to the individual and the community. Only 15% of



respondents felt there were solely benefits. Moreover, 41% agreed/strongly agreed that genetic testing was overall harmful to the individual, and 23% agreed that it was beneficial. Interestingly, although most participants believed that genetics contributed to their autism, 50% did not think testing would be able to provide them with more certainty about their diagnosis and 69% believed it would cause them to doubt their diagnosis. Just over half of individuals (53%) did not think a positive result would increase support from family members or increase access to medical, social, and/or educational support (54%). The greatest perceived benefit was that 38% of participants thought that genetic testing may have allowed for an earlier diagnosis of autism. These participants also had concerns for how genetic testing would impact the autistic community as a whole, as 60% agreed/strongly agreed that genetic testing is harmful to the community, while only 20% agreed/strongly agreed that it is beneficial. To this end, participants utilized free response text questions to express their concerns that genetic testing for autism may lead to increased societal discrimination of autistic persons and may provide a means for eugenic practices in the form of terminations of pregnancies at risk for autism (Byres et al., 2023).

When exploring relationships between variables, it was identified that participants' choice of self-identifying terminology was predictive of opinions about genetic testing for autism (Byres et al., 2023). Individuals who used identity first language ("autistic person") were more likely than individuals who used person first language ("person with autism") to see genetic testing as harmful to the autistic individual and less likely to see it as beneficial to the autistic individual and the autistic community. Furthermore, self-diagnosed individuals were less likely than professionally

diagnosed individuals to see genetic testing as harmful to the autistic community, and more likely to see genetic testing as beneficial to the community (Byres et al., 2023).

Beyond this research, what is currently known about the autistic perspective on this topic can primarily be found in online blog posts, statements from self-advocacy groups such as the Autistic Self Advocacy Network (ASAN), and through hearsay (DeThorne & Ceman, 2018). These sources of information tend to emphasize that autistic individuals view themselves to be in the best position to judge the nature of their disability and the purpose of genetic testing for their condition (DeThorne & Ceman, 2018). Some of the anecdotal pushback against genetic testing, as stated by members within the autistic community, can be attributed to a fundamental disagreement about whether or not autism is a disability. While the medical community tends to view autism as a disease, members of the autistic rights movement view autism as a form of natural variation (known as the neurodiversity paradigm) that may benefit the autistic individual and society as a whole (Armstrong, 2015; DeThorne & Ceman, 2018).

Moreover, autistic advocates of the neurodiversity paradigm fear that genetic testing could lead to societal discrimination against autism, and that the expense of autism-related genetic testing may divert funds away from improving the quality of life for autistic persons (DeThorne & Ceman, 2018). A final concern from within the autistic community is that genetic testing for autism could decrease the number of autistic children being born, either through elective terminations of pregnancies suspected to have autism, IVF processes that include genetic testing that can select against autism susceptibility traits, or parents making an informed decision to not have more children due to genetic markers suggesting an increased risk for autism (DeThorne & Ceman,

2018). While there is currently no prenatal genetic test that looks specifically for autism, prenatal genetic testing is currently able to identify genetic syndromes that are highly associated with autism (i.e., Fragile X syndrome), and it is possible that an autism-specific prenatal genetic test may be developed in the future.

#### **1.4 Rationale of Study**

While the past decade has seen significant developments in autism-related genetic testing, there continues to be limited research into how this testing is perceived by the autistic community. This gap in understanding has become increasingly apparent in recent years as the autistic community has voiced their concern about being underrepresented in autism research (DeThorne & Ceman, 2018; Leadbitter et al., 2021). Many in the autistic community have expressed their discontent towards autism-related genetic testing; however, these attestations are primarily found anecdotally - either online or through hearsay - and have only been documented in the literature once (Byres et al., 2023).

Formally evaluating the autistic perspective about autism-related genetic testing can foster a more holistic understanding of the bioethical considerations surrounding this topic and can help to bridge the gap in understanding between healthcare providers and the autistic community. The knowledge gained from this assessment can aid in healthcare providers' approach when communicating the benefits, risks, and limitations, and possible outcomes of genetic testing to autistic individuals and their caregivers. The present study hoped to broaden our understanding of the autistic perspective on autism-related genetic testing for the benefit of healthcare providers and the autistic community.

## **1.5 Purpose of Study**

The aim of this study was to assess how autistic adults view genetic testing related to the diagnosis of autism spectrum disorder (ASD). To accomplish this aim in a comprehensive manner, we outlined the following objectives:

1. Assess our participants' understanding of the genetics of ASD, and the availability of genetic testing related to an autism diagnosis;
2. Explore our participants' motivations and oppositions towards genetic testing for autism;
3. Evaluate the impact that autism-related genetic testing has had on our participants, for those who have received testing;
4. Assess which autism-related healthcare indications our participants find it appropriate to receive genetic testing; and
5. Explore participants' perspectives about the potential development of a prenatal genetic test that looks specifically for genetic markers of autism.

In this exploratory study, we expected to learn more about how autistic adults view genetic testing for autism. We predicted that our participants would express mixed feelings towards autism-related genetic testing with a slightly unfavorable perspective overall. We also expected that our autistic participants would express concern over the potential development of prenatal genetic testing for autism, and that our participants would share insights that we had not yet considered via the free response options.

## CHAPTER 2: GENETIC TESTING FOR AUTISM: THE AUTISTIC ADULT PERSPECTIVE<sup>1</sup>

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<sup>1</sup> Dent, T.S., Carpenter, L., Cobb, S., Drazba, K.T., To be submitted to *Journal of Autism and Developmental Disorders*

## **2.1 Abstract**

Many professional medical organizations recommend some level of genetic testing as standard of care for those diagnosed with autism spectrum disorder. However, genetic testing related to the diagnosis of autism is seen as controversial by many in the autistic community. While opinions about genetic testing have been well-documented from the perspective of the parents of autistic children, our understanding of the autistic adult perspective remains limited. We implemented a descriptive, web-based survey of autistic adults to assess their awareness of, attitudes towards, and interests in genetic testing for autism (n = 145). Our data demonstrated that half of our participants are unfamiliar with genetic testing for autism, and 86% are unaware of current medical guidelines recommending genetic testing for autism. A minority of participants held an overall positive opinion towards genetic testing (19%) and want genetic testing for themselves (17%). Furthermore, none of the commonly perceived benefits of testing were endorsed as benefits by a majority of participants. Regarding the potential development of a prenatal genetic test for autism, 16% are in support of its development and 74% are concerned that it could lead to an increase in terminations of pregnancies suspected to develop autism. These findings highlight a disconnect in perspectives about genetic testing between autistic individuals, their parents, and the medical community. Future research should continue to explore the autistic adult perspective on this topic and ensure that autistic voices are heard during the development of new tests and professional guidelines involving genetic testing related to autism.

## 2.2 Introduction

Several major medical organizations (American Academy of Pediatrics, American College of Medical Genetics, American Academy of Child and Adolescent Psychiatry) have published guidelines recommending that patients diagnosed with autism spectrum disorder (ASD) receive genetic testing (Barton et al., 2018; Hyman et al., 2020; Schafer et al., 2013). While there is no one genetic testing algorithm that best suits every patient, genetic testing related to the diagnosis of ASD typically begins with a chromosomal microarray (CMA) with a diagnostic yield of 5.4-14%, and Fragile X testing (*FMR1* trinucleotide repeat analysis) with a diagnostic yield of 1-5% (Genovese & Butler, 2020; Harris et al., 2020; Hyman et al., 2020; Schaefer et al., 2013). Whole exome sequencing is often used as a second-tier genetic test and is reported to have a diagnostic yield of 8-20% (Retterer et al., 2016; Tammimies et al., 2015). Using a combined genetic testing approach, biochemical testing, clinical phenotype, and family history, an underlying genetic etiology is now identified in 30-53% of autistic patients (Genovese & Butler, 2020; Lucas et al., 2022).

However, genetic testing related to the diagnosis of autism is seen as controversial by many in the autistic community. Autistic advocates of the neurodiversity paradigm fear that genetic testing could lead to societal discrimination against autism, and that the expense of autism-related genetic testing could divert funds away from improving the quality of life for autistic persons (DeThorne & Ceman, 2018). Genetic testing for autism primarily occurs in the childhood years for autistic individuals and is under the discretion of their parents or guardians. For this reason, the majority of research exploring the views and impact of genetic testing for autism has been focused on the parental perspective.

However, to our knowledge, only one study to date has evaluated these metrics from the perspective of autistic individuals themselves. This gap in understanding has become increasingly apparent in recent years as the autistic community has voiced their concern about being underrepresented in autism research (DeThorne & Ceman, 2018; Leadbitter et al., 2021).

From a scientific standpoint, the potential benefits of genetic testing for autism have been well described and primarily include: 1) understanding the etiology, 2) prognostic predictions, 3) early treatment interventions, and 4) informing reproductive decision making (Hens et al., 2016; Hyman et al., 2020). Identifying a genetic etiology can be important for several reasons. It can provide physicians and family members with more information about recurrence risks, guide patients and families to condition-specific resources, prevent unnecessary medical tests, and lead to the identification and treatment of comorbid health conditions (Hyman et al., 2020). However, despite these potential benefits, the clinical utility of genetic testing for ASD has been brought into question by some in the medical community. For example, some have argued that it is not always clear what factors genetic information can help to predict for autistic individuals, and how this can impact their medical care (DeThorne & Ceman, 2018; Lucas et al., 2022). Due to a number of factors, including a relatively low diagnostic yield and high degree of uncertainty following certain genetic findings, some experts suggest that genetic testing does not result in any meaningful benefits for the majority of autistic individuals who receive testing (Hanish et al., 2018).

Despite the publication of guidelines advocating for genetic testing by several major medical organizations, the uptake of genetic testing is far lower than expected in



practice (Barton et al., 2018). A collection of studies in the past decade have reported that approximately one-third (22-41%) of autistic patients had received any type of genetic testing (Cuccaro et al., 2014; Kiely et al., 2016; Vande Wydeven et al., 2012; Zhao et al., 2019). Another study examining self-reported and medical record data from 1,280 autistic patients found that only 3% received a multi-step genetic testing approach as recommended (Moreno-De-Luca et al., 2020). The low uptake in genetic testing may be due to a number of factors including conflicting information across guidelines about when a referral should be made and which provider should make the genetics referral, low genetic literacy amongst healthcare providers who provide care for autistic individuals, and individual parent and proband factors (Li et al., 2016; Little & Gunter, 2021).

Research suggests that parental awareness of the availability of genetic testing for ASD is limited, with two studies reporting that only 37% and 53% of parents were previously aware of autism-related genetic testing (L. S. Chen et al., 2013; Zebolsky et al., 2020). In a study conducting semi-structured interviews with 20 parents of autistic children, 95% reported a lack of knowledge about the availability of genetic testing for autism despite 70% being aware that genetics was a cause for autism (Hanish et al., 2018). Despite the generally low awareness of testing among parents of autistic children, two studies involving parents of autistic children have demonstrated that 83-96% were interested in pursuing genetic testing for their child (Li et al., 2016; Wagener et al., 2020). Furthermore, in a large Turkish sample of 951 parents, 87% reported that they would pursue genetic testing if it could identify a cause for their child's ASD, and 84% of

parents said that genetic testing was a key part of the diagnostic process (Ayhan et al., 2021).

To our knowledge, there have been at least nine studies in the past decade that have assessed parental perspectives about genetic testing for autism. While they each differ in their approach, they have all inquired about the perceived benefits and barriers towards pursuing genetic testing for their children. The most commonly reported perceived benefit for genetic testing is to better understand the cause of their child's autism (L. S. Chen et al., 2013; Lucas et al., 2022; Reiff et al., 2017; Reiff et al., 2015; Xu et al., 2016). Families have also reported that obtaining an etiological label for their child helped to reduce guilt within the family (Hanish et al., 2018; Lucas et al., 2022; Reiff et al., 2015). Another commonly perceived benefit is that genetic testing may identify potential comorbidities and lead to targeted medical care for their child (L. S. Chen et al., 2013; Hanish et al., 2018; Lucas et al., 2022; Reiff et al., 2015; Wagener et al., 2020; Xu et al., 2016). Parents also agreed that identifying a genetic cause could improve access to various supports and services such as early intervention (Hanish et al., 2018; Lucas et al., 2022; Narcisa et al., 2013; Reiff et al., 2015; Wagener et al., 2020; Xu et al., 2016). Obtaining genetic information may also inform family planning, both in deciding whether or not to have more children and in preparing for a similar outcome if future children were to be at risk for autism (L. S. Chen et al., 2013; Hanish et al., 2018; Reiff et al., 2017; Reiff et al., 2015; Xu et al., 2016). A final commonly perceived benefit is that testing may contribute to our understanding of autism genetics and consequently benefit other autistic children and family members in the future (Hanish et al., 2018; Reiff et al., 2017; Wagener et al., 2020; Xu et al., 2016).

As previously mentioned, the majority of parents in these studies have indicated that they would like to pursue genetic testing for their autistic child. Nevertheless, a considerable proportion of parents (13-38%) would reportedly decline the option to pursue genetic testing for their child (Ayhan et al., 2020; L. S. Chen et al., 2013). One of the most commonly cited reasons to oppose genetic testing is a perceived lack of benefit for the child and family (L. S. Chen et al., 2013; Narcisa et al., 2012; Reiff et al., 2017). Another commonly cited reason is the psychological distress that may occur from awaiting genetic testing results and from receiving an uncertain result (L. S. Chen et al., 2013; Narcisa et al., 2012; Reiff et al., 2017; Xu et al., 2016). Of note, even a negative result may be confusing for families who had previously been informed about the multifactorial inheritance of autism, and now believe that a genetic causation has been ruled out (Reiff et al., 2015). Furthermore, other parents have reported concerns about putting a label on their child due to the stigma associated with genetic conditions (L. S. Chen et al., 2013; Narcisa et al., 2012; Reiff et al., 2017). A genetic finding may also infer a sense of immutability for their child's condition which may discourage a family from taking action to improve their child's quality of life (L. S. Chen et al., 2013).

Parents also expressed concern regarding financial barriers, such as a lack of insurance coverage for testing, as well as the possibility that a genetic finding may negatively impact insurance eligibility and costs in the future (L. S. Chen et al., 2013; Johannessen et al., 2017; Wagener et al., 2020; Zebolsky et al., 2020). Considering the sensory concerns experienced by autistic individuals, parents have also reported that the process of obtaining a specimen sample for genetic testing may be painful and stressful for the autistic child (Hanish et al., 2018; Xu et al., 2016). A final set of barriers

perceived by parents of autistic children include transportation and scheduling issues, mistrust in healthcare providers, and privacy concerns (L. S. Chen et al., 2013; DeThorne & Ceman, 2018; Hanish et al., 2018; Xu et al., 2016).

The first attempt at formally assessing autistic adults' perspective about this topic occurred very recently, when 461 autistic adults were surveyed regarding their experiences with, attitudes toward, and interest in genetic testing for autism (Byres et al., 2023). Approximately half (48%) of participants indicated they would not have wanted genetic testing as a child, and 56% stated they would not want autism related genetic testing for any of their current or future children. The greatest perceived benefit was that 38% of participants thought genetic testing may allow for an earlier diagnosis of autism. Participants were also surveyed regarding when genetic testing should be offered, to which 35% agreed/strongly agreed that testing should be routinely offered to autistic adults, 26% agreed/strongly agreed that testing should be routinely offered to autistic children, and 49% agreed/strongly agreed that genetic testing should not be done at all for autism. Regarding overall perceptions of genetic testing, 41% agreed/strongly agreed that genetic testing was overall harmful to the individual, and 23% agreed that it was overall beneficial (Byres et al., 2023).

Beyond this research, what is currently known about the autistic perspective on this topic can primarily be found in online blog posts and statements from self-advocacy groups such as the Autistic Self Advocacy Network (ASAN) (DeThorne & Ceman, 2018). These sources of information tend to emphasize that autistic individuals view themselves to be in the best position to judge the nature of their disability and the purpose of genetic testing for their condition (DeThorne & Ceman, 2018). A common concern

held by the autistic community, as stated in several of these anecdotal sources, is that genetic testing for autism could decrease the number of autistic children being born, either through elective terminations of pregnancies at risk to develop autism, IVF processes that include genetic testing that can select against autism susceptibility traits, or parents making an informed decision to not have more children due to genetic markers suggesting an increased risk for autism (DeThorne & Ceman, 2018). To this end, a recent survey of 333 mothers of autistic children in Taiwan found that 66.6% would undergo prenatal genetic testing to detect autism susceptibility genes, and 53.1% would terminate the pregnancy if the fetus were expected to develop autism (W. J. Chen et al., 2020). While there is currently no prenatal test that looks specifically for genetic markers of autism, prenatal genetic testing is currently able to identify genetic syndromes that are highly associated with autism (i.e., Fragile X syndrome), and it is possible that an autism-specific prenatal test may be developed in the future.

In light of these aforementioned concerns and the limited data regarding the autistic adult perspective, this study was created to contribute to the burgeoning literature on the autistic adult perspective about genetic testing for autism. Formally evaluating the autistic perspective about autism-related genetic testing can foster a more holistic understanding of the bioethical considerations surrounding this topic and can help to bridge the gap in understanding between healthcare providers and the autistic community. The knowledge gained from this assessment can aid in healthcare providers' approach when communicating the benefits, risks, and limitations, and possible outcomes of genetic testing to autistic individuals and their caregivers.

## **2.3 Methods**

This study was approved by the Institutional Review Board at the University of South Carolina in July 2022 (Pro00121781).

### *2.3.1 Participants and Recruitment*

Participants included in this study were autistic individuals 18 years of age and older. The diagnostic label of “autism spectrum disorder” has only been in use since 2013, so the inclusion criteria was expanded to include participants with any of the following autism-related diagnoses: autism, autistic disorder, Asperger’s syndrome, pervasive developmental disorder (PDD), pervasive developmental disorder not otherwise specified (PDD-NOS), and childhood disintegrative disorder (CDD). Those who identify as autistic and do not have a formal autism-related diagnosis were excluded from participation in this study.

The primary means for recruiting autistic adults was through an advertisement message (Appendix A) and research flyer (Appendix B) posted in autism-related social media pages on Reddit and Facebook. The advertisement message and flyer were also uploaded to the following websites’ research pages: Medical University of South Carolina’s Project Rex, Autism Science Foundation, and Greenwood Genetic Center. The survey was open between August 2022 and January 2023.

### *2.3.2 Study Design*

We created an online, descriptive web-based survey using Qualtrics (Qualtrics, Provo, UT) that utilized multiple-choice, multi-select, Likert scale, visual analog scale, and free response questions aimed at understanding how autistic adults view genetic testing for autism (Appendix C). This survey used identity-first language as this is the

most preferred self-describing language type among members of the autistic community (Botha et al., 2021; Bury et al., 2020). In line with best practice recommendations, an autistic self-advocate was recruited for consultation to ensure that the project's aims and survey language use were sensitive to the preferences of the autistic community (Keating, 2021). To enhance the comprehensibility of our survey for our autistic participants, who may experience difficulties in reading comprehension, we avoided complex sentence structure, difficult vocabulary, figures of speech, and imprecise response options (Nicolaidais et al., 2020).

Participation in this study was voluntary, and the participants' completion of the survey served as their consent. Participants remained completely anonymous and no identifying information was collected on the survey. At the end of this survey, participants were given the option to click on a link to a different Qualtrics survey in order to provide their email address for monetary compensation in the form of a \$5 Amazon Gift Card to the first 100 participants. The survey took approximately 7 to 15 minutes to complete and included four sections:

- 1) Demographics and genetic knowledge. Demographic information (age, race, ethnicity, and gender) was collected, along with information about diagnosis, perceived severity of disability, and knowledge about the genetics of autism and genetic testing for autism.
- 2) Perceptions about genetic testing for autism. Participants were asked to indicate their opinion about the reasons to pursue and oppose genetic testing for autism via 'select all that apply' questions.

- 3) Experience with genetic testing. Participants were asked whether they have received autism-related genetic testing. If they had, a series of ‘select all that apply’ questions appeared regarding the ways in which the genetic testing may have positively and/or negatively impacted their lives. If they had not received autism-related genetic testing, they were asked to select the reasons why they haven’t, and whether they would be interested in pursuing it.
- 4) Overall opinions about genetic testing. Participants were provided with brief information regarding the current medical guidelines recommending genetic testing for autism and were asked for their agreement with these guidelines. They then completed multiple choice, multi-select, and open-ended free response questions regarding their overall opinion of genetic testing for autism.
- 5) Prenatal genetic testing for autism. In the final portion of the survey, participants were given a brief description of the current state of prenatal genetic testing as it relates to autism. To this end, they were informed that prenatal testing can currently detect genetic conditions associated with autism (i.e., Fragile X syndrome); however, there are currently no prenatal tests that look specifically for the genetic markers of autism. Participants were then given a series of questions relating to the hypothetical development of a prenatal test that looks specifically for markers of autism and had the opportunity to provide further perspective in the form of a free response question.



### *2.3.3 Statistical Analysis*

Analyses were applied after filtering out duplicate responses, incomplete (<75% of survey) responses, responses lasting fewer than 260 seconds, as well as surveys where the respondent selected “I do not have an autism diagnosis.” Fraudulent / spam responses were filtered out using a modified version of Qualtrics’ fraud detection feature; the authors came to a consensus about the minimal standard of rigor for this filtering process.

Descriptive statistical analysis was conducted using Microsoft Office Excel software. Responses to categorical questions were reported via count and frequency and then assembled into rank order to determine the highest rated items. The data from our Excel spreadsheets was transferred into Statistical Package for Social Sciences (SPSS) for comparative statistical analysis. Here, Chi-Square and Wilcoxon-Mann Whitney tests were employed to explore relationships between perceived severity of disability and 1) interests in genetic testing, 2) opinion of genetic testing for autism, 3) agreement with national criteria, and 4) opinions about the hypothetical development of a prenatal genetic test for autism. An inductive content analysis approach was used to analyze the qualitative data gathered from the open-ended, free response questions. Themes were created by the primary investigator based on the participants' responses and were subsequently reviewed and approved by the fourth author. These themes were then coded by the first, third, and fourth authors and the resulting thematic frequencies were reported. Quotations from the free response texts were extracted and used to supplement the quantitative data.

## 2.4 Results

### 2.4.1 Demographics

A total of 2,796 responses were recorded from this survey. After removing duplicate and spam responses (n = 2,552), “*I don’t have an autism diagnosis*” responses (n = 51), incomplete responses (< 75% complete) (n = 12), and responses lasting fewer than 260 seconds (n = 36), a total of 145 responses were remaining for analysis.

Respondents were primarily White (84%) and non-Latinx / Hispanic (67%). Ages ranged from 18 to 77 years old with an average age of 29.4 years old (SD = 9.1). A variety of gender identities were reported with 39% identifying as female, 29% as male, 21% as non-binary, 3% as transgender male, 2% as transgender female, and 6% as Other/Prefer not to answer (Table 2.1).

**Table 2.1** Participant Demographic Information

Characteristic (n = 145)	n (%)
<b>Gender</b>	
Female	56 (39%)
Male	42 (29%)
Non-binary	31 (21%)
Transgender male	4 (3%)
Transgender female	3 (2%)
Prefer not to answer	5 (3%)
Other	4 (3%)
<b>Race <sup>a</sup></b>	
Caucasian / White	122 (84%)
Native American / Alaskan Native	6 (4%)
Asian / Pacific Islander	5 (3%)
African American / Black	3 (2%)
Middle Eastern	1 (1%)
Prefer not to answer	2 (1%)
Other	8 (5%)
<b>Ethnicity</b>	

Not Latinx / Hispanic	97 (67%)
Latinx / Hispanic	20 (14%)
Prefer not to answer	10 (7%)
Other	13 (9%)
<b>Current autism diagnosis</b>	
Autism Spectrum Disorder (ASD)	94 (65%)
Autism or Autistic Disorder	29 (20%)
Asperger's syndrome	16 (11%)
Childhood Disintegrative Disorder (CDD)	2 (1%)
Pervasive Developmental Disorder (PDD)	2 (1%)
PDD-Not Otherwise Specified (PDD-NOS)	2 (1%)

<sup>a</sup> Participants were allowed to select more than one option

#### 2.4.2 Perceived Severity of Disability

To assess perceived severity of disability, participants were asked to indicate their agreement with the statement “*I view my autism diagnosis as a difference and not a disability.*” Of the 144 responses to this question, 41% (n = 59) disagreed/strongly disagreed, 40% (n = 58) agreed/strongly agreed, and 18% (n = 26) neither agreed nor disagreed with this statement. Responses to this question did not influence desire for genetic testing, opinion of genetic testing, agreement with testing guidelines, or opinion of prenatal testing. Awareness of the availability of genetic testing for autism was limited, with 50% (n = 48/96) participants indicating that genetic testing is available and 50% (n = 48/96) indicating it is not. When asked to estimate the percentage of autistic individuals with an identifiable genetic change that is suspected to be the cause of their autism, responses ranged from 0-100% (Mdn = 30%, IQR = 48%).

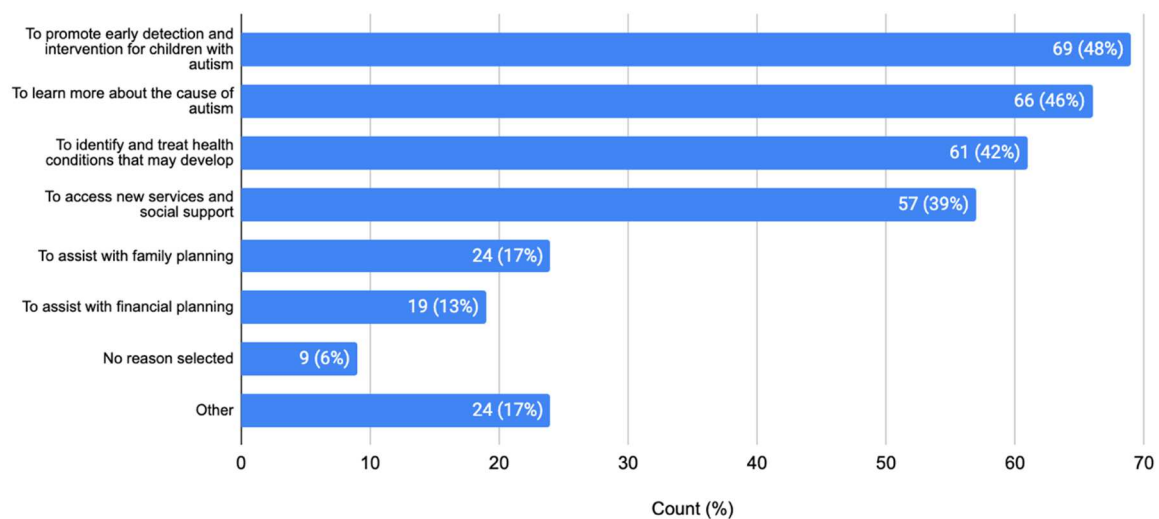
#### 2.4.3 Awareness of and Agreement with National Medical Guidelines

Awareness of the national (United States) medical guidelines recommending genetic testing for every autistic individual was limited, as 86% (n = 95/110) indicated that there are no such guidelines. Later in the survey, participants were given brief

information about the guidelines and were asked if they agreed with them. More than half (56%) (n = 80/143) did not agree with these guidelines, 21% (n = 30/143) agreed, and 23% (n = 33/143) were not sure.

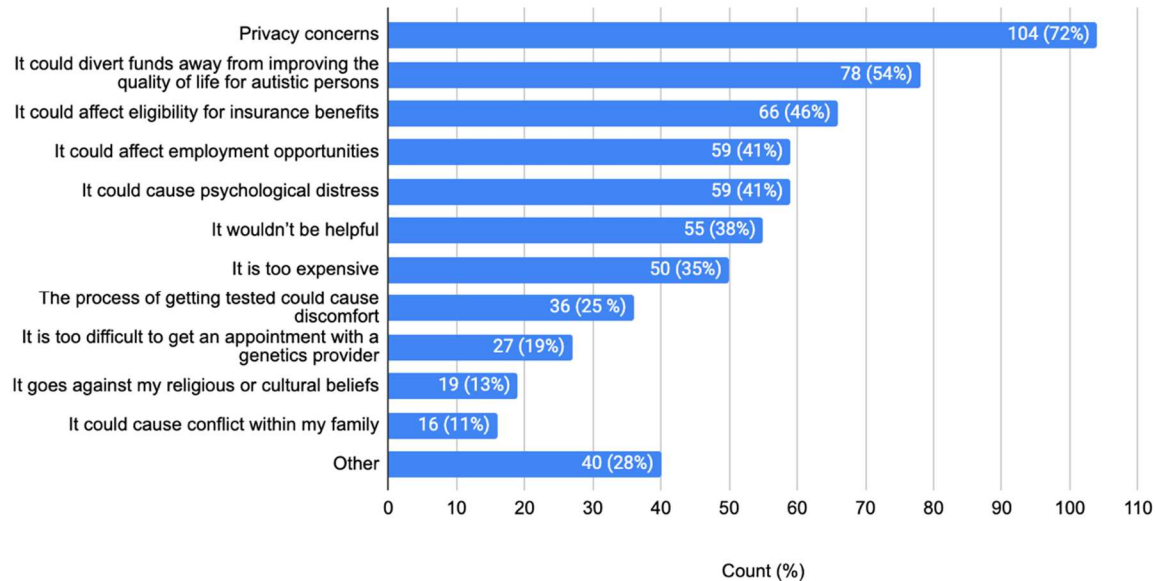
#### 2.4.4 Reasons to Receive and Decline Genetic Testing for Autism

The most commonly perceived reasons to receive genetic testing (Figure 2.1) were “To promote early detection and intervention for children with autism”, “To learn more about the cause of autism”, and “To identify and treat health conditions that may develop” (Figure 2.1). A free response text option was provided to those who selected “Other”, to which 7 participants mentioned “eugenics” and 7 mentioned either “none” or “no reason.” Of note, 6% (n = 9) of respondents did not select any reason to receive genetic testing for autism.



**Figure 2.1** *Perceived reasons to receive genetic testing for autism (n = 136)*

The most commonly perceived reasons to decline genetic testing for autism were “Privacy concerns”, “It could divert funds away from improving the quality of life for autistic persons”, “It could affect eligibility for insurance benefits”, and “It could affect employment opportunities” (Figure 2.2).



**Figure 2.2** *Perceived reasons to decline genetic testing for autism (n = 145)*

A free response text option was provided to those who selected “Other”, to which 24 individuals mentioned fears for eugenic practices, and 7 mentioned fears for how this genetic information could be used to “cure” or initiate unwanted treatment for autism.

Regarding these concerns, one individual made the comment:

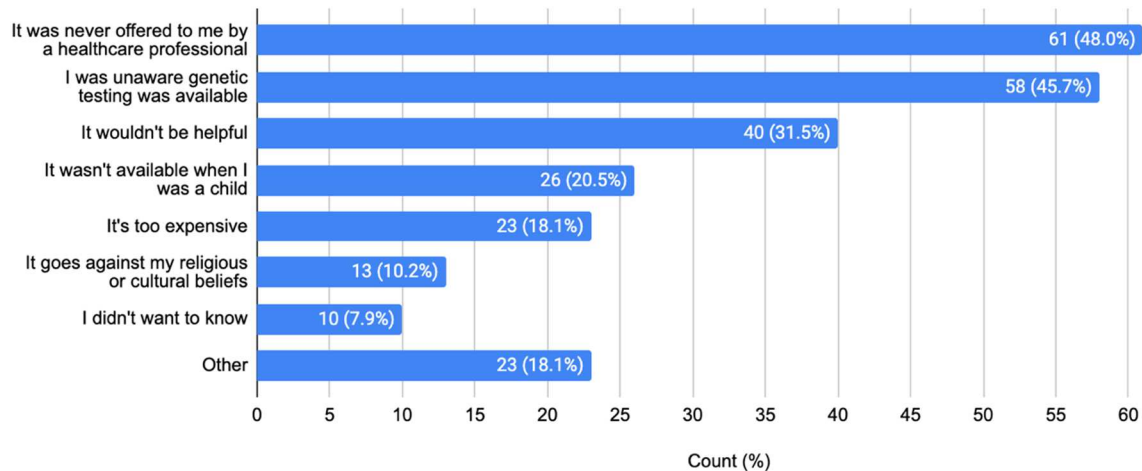
*“It sounds suspiciously like eugenics. I feel frightened to my core every time I see an article about a study trying to identify genetic causes for autism because I am afraid it will lead to prenatal testing and thus eugenic abortions. I am so afraid for the future of the autistic community if this happens. Autism is a disability, but it is also beautiful. A world without my people, without autistic people, is too bleak to fathom, and I fear that this will happen if prenatal tests for autism are created...” - Caucasian female, age not provided*

#### 2.4.5 Impact of Genetic Testing for Autism

Only 18 participants (12%) indicated that they have received genetic testing for autism, and 9 of these individuals received a positive result, 3 received a negative result,

2 received a variant of uncertain significance (VUS), and 3 indicated they did not know their result. Of these 18 individuals who received genetic testing, the most commonly perceived benefits were “I learned more about the cause of my autism” (n = 9), “It helped identify and treat health conditions that I developed” (n = 7), and “It enabled early diagnosis and intervention for my autism” (n = 6). The most commonly perceived detriments of genetic testing were “It caused me psychological distress (i.e., low self-esteem, anxiety, depression)” (n = 9), “It caused tensions or conflict in my family” (n = 7), and “The process of getting the genetic testing caused me discomfort (i.e., blood draw, hospital visit)” (n = 5). When these same 18 respondents were asked about the overall impact of genetic testing on their life, 8 indicated it had a positive/strong positive impact, 4 indicated a negative/strong negative impact, and 6 indicated neither a positive nor negative impact.

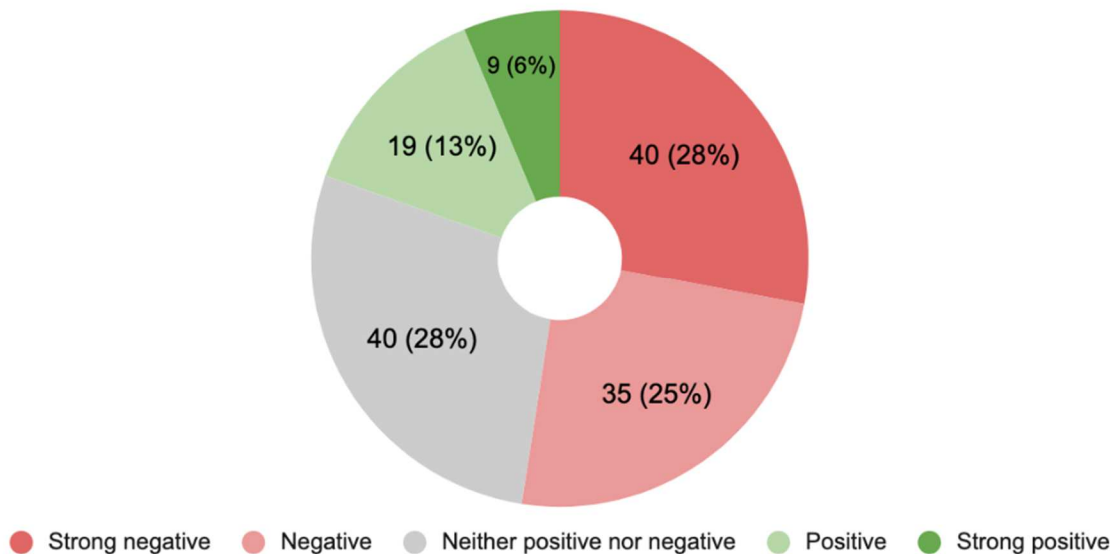
Of those who have not received genetic testing for autism (n = 121), the most commonly selected reasons for not receiving testing were “It was never offered to me by a healthcare professional”, “I was unaware that genetic testing was available”, and “It wouldn’t be helpful” (Figure 2.3). A free response text option was provided to those who selected “Other”, to which 7 individuals expressed concern for genetic testing leading to potential eugenics practices. Interest for genetic testing for autism was limited, as only 17% (n = 25) indicated they want genetic testing. Furthermore, 55% (n = 67) do not want testing, and 23% (n = 28) were not sure whether they would want genetic testing.



**Figure 2.3** *Reasons participants have not received genetic testing for autism (n = 121)*

#### 2.4.6 Overall Opinions of Genetic Testing for Autism

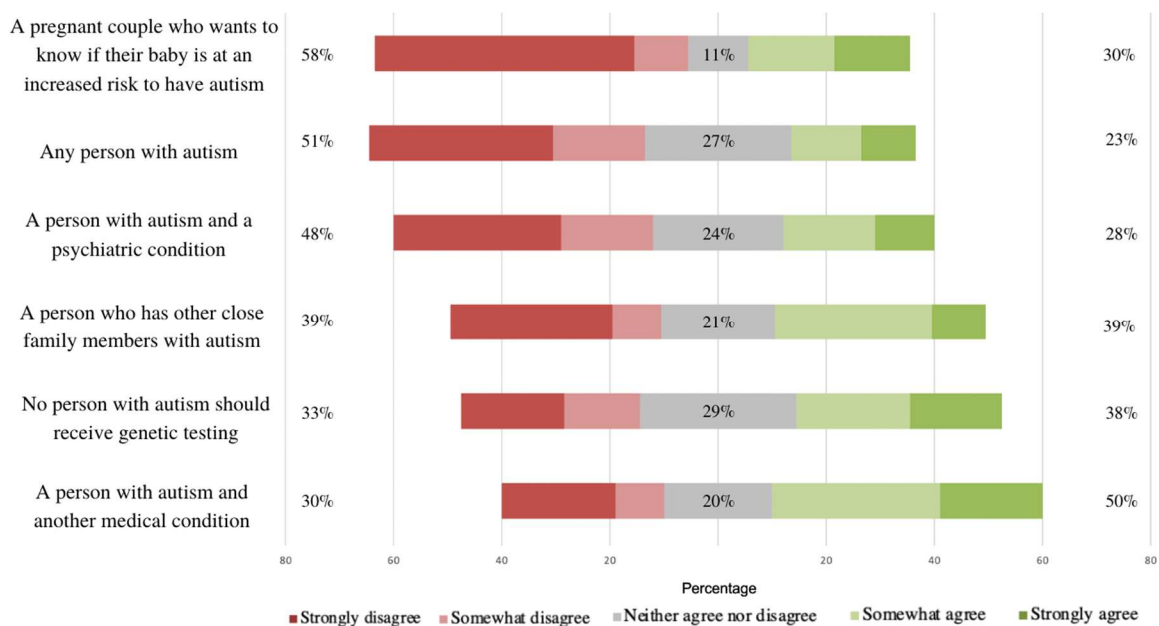
Overall opinions towards genetic testing for autism were varied; however, the majority of respondents (53%, n = 75) shared a negative/strongly negative opinion (Figure 2.4).



**Figure 2.4** *Overall opinions about genetic testing for autism (n = 143)*

#### 2.4.7 Who Should Receive Genetic Testing for Autism

When presented with a set of scenarios, participants were asked to indicate their agreement about whether they should receive genetic testing (Figure 2.5). The strongest opposition to a group being tested was for “A pregnant couple who wants to know if their baby is at an increased risk to have autism” to which 58% disagreed/strongly disagreed with this statement and 30% agreed/strongly agreed. The strongest support for a group being tested was “A person with autism and another medical condition” to which 50% agreed/strongly agreed they should receive testing.



**Figure 2.5** Participant agreement about who should receive genetic testing for autism ( $n = 143$ )

#### 2.4.8 Prenatal Genetic Testing for Autism

Regarding the potential creation of a prenatal genetic test looking specifically for genetic markers for autism in the future, 74% ( $n = 108$ ) of participants agreed that “It may lead to pregnancy terminations of fetuses expected to develop autism”, and 63% agreed with the statement “I am very concerned about the development of a prenatal test



specifically for autism.” A minority of the participants agreed with the potential benefits of a prenatal test, as 46% (n = 67) agreed “It may help families better prepare for their child’s needs” and 46% (n = 67) agreed “It may lead to earlier access to support services for autistic children and their family members.” Regarding support for the development of such a prenatal test, 67% (n = 97) are not in favor, 16% (n = 23) are in favor, and 17% (n = 25) are unsure.

#### 2.4.9 Free Response

Two open-ended, free response questions were asked towards the end of the survey: 1) “*Is there anything else you would like to say about genetic testing for autism?*” and 2) “*Is there anything else you’d like to say about prenatal genetic testing for autism?*” Themes from these free response questions can be found in Table 2.2.

**Table 2.2** Themes created from free response

“Is there anything else you’d like to say about genetic testing for autism?” (n = 87)		“Is there anything else you’d like to say about prenatal genetic testing for autism?” (n = 73)	
Themes <sup>a</sup>	Count (%)	Themes <sup>a</sup>	Count (%)
Eugenics and abortion concerns	39 (45%)	Eugenics and abortion concerns	42 (58%)
Genetic testing can lead to discrimination, stigmatization, or dehumanization of autistic people	20 (23%)	Against the development of a prenatal genetic test for autism	19 (26%)
Can be beneficial to the autistic individual	15 (17%)	Can be beneficial to the family	8 (11%)
Should be optional and with the autistic person’s consent	10 (11%)	Can be beneficial to the autistic individual	7 (10%)
Concerns for how the results would be used (e.g., to “cure autism”, ABA therapy, etc.)	7 (8%)	Non eugenics concerns (e.g., stigmatizes the child, leads to harmful treatment, etc.)	6 (8%)
Wouldn’t be helpful / not necessary	7 (8%)	Neutral / uncertain about the development of a prenatal genetic test for autism	6 (8%)
Can be beneficial to the family	5 (6%)	In support of the development of a prenatal genetic test for autism	4 (5%)
Resources should be used for other purposes	4 (5%)	Uncategorized	13 (18%)
Uncategorized	13 (15%)		

<sup>a</sup> More than one theme could be counted for each response

In response to this first question, multiple individuals brought up concerns for eugenics and stigmatization:

*“It has historically been used to justify eugenics. From what I know it seems pretty clear that there is a significant genetic link, but I fail to see how exploring the individual genes that may cause autism would be helpful in any capacity. As a scientist I understand wanting to know more specifics and the mechanism that causes ASD, but as an autistic person I have very little faith that this information would be used for anything other than the elimination of fetuses with autism genes. And while I do consider myself disabled and struggle in many aspects of my life, I would not be me if I didn't have autism...” - Caucasian female, 22 years old*

Some believed that the resources spent on genetic testing would be better spent on improving the quality of life for autistic people in other ways:

*“Resources should go to giving actually Autistic people a platform to educate society because we are constantly talked over by people who spread misinformation and care more about getting us to conform at the expense of our physical and mental health.” - Caucasian female, 27 years old*

While some individuals saw potential benefit to testing, these comments often came with reservations about how this genetic information could ultimately be used against autistic persons:

*“If the testing was done in the cultural context of providing support I would be in favor. However, it is very likely that testing will provide opportunity for eugenic*

*selection away from autistic traits, or intervention that can be harmful for the individual.” - Caucasian non-binary participant, 37 years old*

More than half of those who responded to the prenatal free response question expressed concerns about eugenics. To this end, one individual commented:

*“I think it's better to make autism understood and destigmatized, rather than trying to create tests to find it in a fetus. This is kind of insulting. Autism is a mindset, an outlook, a creative way of processing and seeing details that are missed by the neurotypical. To give parents a chance to stop that before birth is... it's just wrong.” - Caucasian non-binary, assigned female at birth (AFAB), 39 years old*

To this end, several participants made the comparison of prenatal tests being used to terminate fetuses expected to have other conditions:

*“Iceland has nearly eliminated people with Downs syndrome [sic], largely due to pressure to abort those pregnancies in prenatal screening. I think that people with developmental disorders like autism and Downs syndrome [sic] are valuable to have in this world and I think selective abortions for things that are simply undesirable (rather than incompatible with life) is immoral, especially when accompanied by significant pressure from healthcare providers.” - Caucasian transgender male, 23 years old*

Some commenters recognized the potential benefits of prenatal testing, but remained conflicted about the overall impact this technology would have:

*“I am very conflicted with the notion of prenatal genetics testing. Ideally, this would be used for early identification to help the family and medical providers to*

*prepare for the child's needs. Early testing could identify autism and eliminate the long process that is currently getting a diagnosis. It could also help the family and their doctors to find support and services in a much more timely manner and to be better prepared. However, while this is the ideal scenario, I believe that many parents would choose to abort or give up their child if they found out they had autism, which completely goes against everything I believe...*” - Caucasian female, 25 years old

Others were more direct in indicating their disapproval for such a prenatal test: *“I think it is a terrible, devastating idea. Autism looks so different in so many different people - and most medical providers and would-be parents have NO IDEA about what Autism can look like... Prenatal genetic testing for Autism is eugenics and Autistiphobic [sic] and begins with the life threatening and erroneous idea that Autism is a terrible disorder. There is no way to ‘prepare for their child's needs’ until they meet the child. Every single Autistic person is different and unique. Having the diagnosis without the child is ridiculous.”* - Caucasian female, 40 years old

## **2.5 Discussion**

To the best of our knowledge, this is the second study to investigate how autistic adults view genetic testing related to the diagnosis of autism. As predicted, although a range of perspectives were captured, the majority of participants expressed an overall negative sentiment with regards to their opinions of, and interests in, genetic testing for autism. This negative sentiment had previously been hypothesized by theoretical work based on anecdotal statements from the autistic community and has only recently been

examined in descriptive research involving autistic adults (Byres et al., 2023; DeThorne & Ceman, 2018).

Our findings share several parallels with the assessment conducted by Byres et al. (2023). A major concern recognized in both studies is that genetic testing may be more harmful to the autistic individual than it is beneficial. To this end, a large portion of participants agreed that testing could 1) stigmatize the individual, 2) result in unwanted therapies, 3) increase the psychological burden experienced by autistic individuals, and 4) divert resources away from improving the quality of life for autistic persons via more practical approaches (Byres et al., 2023). Furthermore, only a minority of respondents in each study recognized the benefits commonly perceived by the medical community and parents of autistic children, such as in 1) promoting early detection and intervention for autism, 2) learning more about the cause of autism, 3) identifying and treating comorbidities that may develop, and 4) accessing new services and social support (Byres et al., 2023). These concerns and unrecognized benefits likely contribute to the low interest in, and overall negative opinion towards, genetic testing revealed in both studies.

Our findings regarding autistic adult perspectives diverge from research regarding parental perspective about genetic testing. While a portion of parents do cite the potential for increased psychological distress and uncertain utility of genetic testing as reasons to oppose testing their child, the majority express an interest in genetic testing and endorse the commonly cited benefits as described above (L. S. Chen et al., 2013; Li et al., 2016; Lucas et al., 2022; Narcisa et al., 2012; Reiff et al., 2017; Wagener et al., 2020; Xu et al., 2016). Seeing that autism-related genetic testing most frequently occurs in the pediatric setting when the autistic child is unable to consent for themselves, these disagreements

highlight a conflict between the autistic individual's medical autonomy, and the parents or providers acting upon their perceived notion of what is best for the child. Furthermore, considering the current clinical guidelines recommending genetic testing for all autistic children and the resulting high volume of genetic testing, it can be argued that the lack of autistic perspective on this topic is incongruent with its clinical application.

Consequently, more research into the autistic adult perspective about genetic testing and how it relates to parents' and providers' decision making for autistic children is needed.

Disagreements between autistic individuals and their family members, support networks, and/or medical providers are not specific to genetic testing. Differing opinions between these stakeholders have been well-documented, especially with regards to interests in finding “cures” for autism and whether or not autistic children should receive therapies viewed as traumatic and ineffective by some autistic individuals (i.e., ABA, early intervention) (Armstrong, 2015; DeThorne & Ceman, 2018; Hens et al., 2016; Pellicano & Sears, 2011). A source of these disagreements may stem from differences in how stakeholders pathologize autism. While the medical community tends to view autism as a disease, members of the autistic rights movement tend to view autism as a form of natural variation (known as the neurodiversity paradigm) that may benefit both the autistic individual and society as a whole (Armstrong, 2015; DeThorne & Ceman, 2018). These differing perspectives may exacerbate the documented mistrust between autistic individuals and the medical community (Bradshaw et al., 2019; Moseley et al., 2020). Our free response data further demonstrate this point, as several individuals stated that any potential benefit of genetic testing is overshadowed by the mistrust they have for the medical providers using those results:

*“Genetic testing would lead to more kids being diagnosed at young ages, and thus being subjected to abusive ‘treatments’ like ABA. I wish neurotypical doctors, researchers, and psychologists would just leave us alone and stop trying to ‘fix’ us. All they do is make our lives worse. Please, if you’re a researcher reading this, stop trying to fix us, stop trying to ‘cure’ us, stop trying to ‘treat’ us. Just let us live our lives.” - Caucasian female, age not provided*

Another area of expressed concern is how genetic testing may impact the autistic community as a whole. To this end, Byres et al. found that participants were more likely to agree that genetic testing is harmful to the autistic community than to the autistic individual (2023). In response to this finding, the authors hypothesized that a primary cause for this discrepancy was their participants' concern regarding the advent of routine prenatal screening for autism, which would likely result in increased terminations of fetuses expected to develop autism (Byres et al., 2023). This theory is in line with the previous research that has discussed genetic testing as a means to empower informed family planning (L. S. Chen et al., 2013; L. S. Chen et al., 2015; W. J. Chen et al., 2020; Hanish et al., 2018; Reiff et al., 2015; Steinman, 2019; Xu et al., 2016). While this research was primarily focused on the parental perspective, the present study is the first, to our knowledge, to assess perceptions about the use of prenatal testing for autism from the perspectives of autistic individuals themselves. Our findings suggest that the majority of autistic adults are very concerned about the development of a prenatal genetic test for autism, primarily due to the potential for increased terminations and stigmatization of the child suspected to develop autism. Moreover, the participants' free response submissions

have revealed the intensity of this concern which, at times, is to the point where prenatal genetic testing is perceived as an existential threat to the autistic community:

*“If a reliable prenatal genetic test for autism were developed, I suspect that the situation of autistic people nationwide would be similar to the situation of people with Down syndrome in countries like Denmark” - Asian / Pacific Islander non-binary, 21 years old*

#### 2.5.1 Limitations

The individuals who participated in this study may not be representative of the autistic population as a whole. While it has been established that autism is a pan-ethnic disorder that affects different ethnic groups similarly (Yuan et al., 2021), the vast majority (84%) of the participants in this study identified as Caucasian/White. It is also recognized that the autistic community is highly diverse with regards to gender identity, especially pertaining to the large portion of autistic adults identifying as non-binary and transgender (Walsh et al., 2018). While our survey was well-represented by gender diverse individuals (21% non-binary; 5% transgender), only 29% identified as male. This is a significantly lower proportion than previously reported data suggesting that autism is four times more common in males than in females (Volkert et al., 2022).

Moreover, our survey did not include autistic individuals who are self-diagnosed. There is a growing body of evidence showing that autism is underdiagnosed, especially in females, and that self-diagnostic tools can be accurate and specific for correct autism self-diagnoses (Loomes et al., 2017; Ratto et al., 2018; Sizoo et al., 2015). Furthermore, individuals who responded to this survey represent a subset of the autistic population who are able to independently access the Internet and respond to an electronic survey. Autistic



individuals may prefer a variety of communication methods, so additional measures should be taken to better access and support individuals using alternative communication methods in future research.

Another limitation of this study is the possibility that few fraudulent (“bot”) responses were included in the final analysis, and that few legitimate responses were incorrectly filtered out. This was an open-source survey that offered compensation to its participants, which rendered the survey to be at a high risk for receiving fraudulent responses. In response to this, we utilized Qualtrics’ fraud detection features and modified the settings to best fit our subjective analysis of which responses were nonsensical and likely fraudulent.

A final recognized limitation of this study is that only a small portion of our respondents (12%) indicated that they had received genetic testing for autism. As a result, the majority of our respondents participated based on their conceptual understanding of genetic testing, which may not be as accurate or comprehensive as those who participated based on their own lived experience. However, we do recognize that recall bias may have resulted in fewer participants reporting they had received genetic testing than in reality, as some participants may have forgotten or never been told about testing that occurred during childhood.

### *2.5.2 Future directions*

The most pressing concern identified in this study is the disconnect in perspectives between autistic individuals, and that of their parents/guardians and the medical community. To address this conflict, efforts should be made to educate all stakeholders involved about the current capabilities and limitations of genetic testing for

autism, as well as the potential benefits and harms (Byres et al., 2023). Furthermore, efforts should be made to inform medical providers and parents of autistic children about the emerging body of evidence exploring the autistic adult perspective about genetic testing for autism. Not only will this transparency support a more comprehensive understanding of the bioethical considerations on this topic, but it will also be essential to all parties reaching a consensus about how and when genetic testing related to the diagnosis of autism should be offered.

## **2.6 Conclusion**

This descriptive, exploratory study was created in order to address the limited understanding we have concerning autistic adults' perspectives about genetic testing for autism. Our data suggest that autistic adults hold a variety of opinions about genetic testing, but with an overall negative perspective. While participants were generally aware that genetics may contribute to the cause of autism, they held limited understanding about the availability of genetic testing and the medical guidelines recommending testing for all autistic persons. Interest in genetic testing was also limited among our participants. We hypothesize that a variety of factors may contribute to this limited interest. To this end, our participants demonstrated limited recognition of the potential benefits of genetic testing and held overwhelming concern for how genetic testing could be used to promote stigma, initiate unwanted therapies, and divert resources away from improving the quality of life for autistic persons via more practical approaches. Another significant concern shared by autistic adults in this study was the potential for prenatal genetic testing to result in eugenics practices against the autistic community. Our free response data reveal

the intensity of this concern, which for many participants, is to the extent that genetic testing for autism is viewed as an existential threat to the autistic community.

To our knowledge, the present study is only the second to explore how autistic adults view genetic testing related to the diagnosis of autism. Both studies have revealed the disconnect in perspective about autism-related genetic testing between autistic individuals, their parents, and the medical community. Considering the large volume of genetic testing offered and performed for autistic children, this disconnect reveals the urgent need to further explore the autistic adult perspective about this topic. Similarly, efforts should be made to foster communication between all of the stakeholders, as this will prove essential to discerning when genetic testing should be offered, and how the results can be utilized in a meaningful way.

### CHAPTER 3: CONCLUSIONS

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## APPENDIX A: PARTICIPANT RECRUITMENT INVITATION

Do you have an opinion about genetic testing for autism? We'd love to hear it!

We're looking for autistic adults (18+) to take a short survey that will help us understand the autistic perspective about genetic testing for autism.

The survey will take about 10-15 minutes to complete and a **\$5 Amazon gift card will be given to the first 100 participants who complete the survey.**

Our study seeks to improve the healthcare provided to autistic people. Your participation will help to make the autistic voice heard in medical and scientific research.

Contact us at [uscautismsurvey@gmail.com](mailto:uscautismsurvey@gmail.com) if you have any questions about the study.  
Thank you!

Link to survey:

[https://uofsc.co1.qualtrics.com/jfe/form/SV\\_6Gv5oGtQQpwKiPk](https://uofsc.co1.qualtrics.com/jfe/form/SV_6Gv5oGtQQpwKiPk)

## APPENDIX B: PARTICIPANT RECRUITMENT FLYER

### Genetic testing for autism... what do you think?



We are looking for autistic adults (18+) to take a short survey (10-15 mins) asking your opinion about genetic testing for autism

#### Who can participate?

- Adults (18+) with any autism diagnosis (doesn't have to be ASD)
- You don't need to have had genetic testing to participate

#### Questions about the study?

Please contact us at  
[usautismsurvey@gmail.com](mailto:usautismsurvey@gmail.com)



#### Why should I participate?

- Your participation will increase the autistic perspective in medical / scientific research
- The findings from this study could improve the healthcare provided to autistic individuals
- **5\$ Amazon gift card** will be given to the first 100 participants!

#### QR Code to the survey:



## APPENDIX C: PARTICIPANT QUESTIONNAIRE

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### Start of Block: Introduction

Q1 Thank you for considering to participate in the study, **Genetic Testing for Autism: The Autistic Adult Perspective**. This survey will contain a series of multiple-choice, multi-select, and free response questions to better understand how autistic adults view genetic testing for autism. This survey will take approximately 10-15 minutes to complete.

**You must be 18 years or older with an autism diagnosis to participate.** Your participation is completely voluntary, and you may choose to skip questions or end your participation at any time. All responses gathered from this survey will be kept anonymous and confidential. The results of this study might be published or presented at academic meetings; however, participants will not be identified by their personal information. This survey presents minimal risk to its participants.

Completion of this survey will serve as your informed consent to participate. If you have any questions, please contact the principal investigator, Thomas Dent, at [thomas.dent@uscmed.sc.edu](mailto:thomas.dent@uscmed.sc.edu) or the faculty advisor, Katy Drazba MPH, MS, CGC, at [kdrazba@ggc.org](mailto:kdrazba@ggc.org)

If you are interested in participating in this survey, please click the blue arrow button below. If not, please exit the browser.

---

### End of Block: Introduction

---

### Start of Block: Demographics

Q2 How old are you?

	18	100
18-100		

Q5 What is your gender?

- ☐ Male
  - ☐ Female
  - ☐ Transgender Male
  - ☐ Transgender Female
  - ☐ Non-binary
  - ☐ Prefer not to answer
  - ☐ Other \_\_\_\_\_
- 

Q6 What is your race (select all that apply)

- ☐ African American / Black
  - ☐ Asian / Pacific Islander
  - ☐ Caucasian / White
  - ☐ Middle Eastern
  - ☐ Native American / Alaskan Native
  - ☐ Prefer not to answer
  - ☐ Other \_\_\_\_\_
-



Q26 What is your ethnicity?

- ☐ Latinx / Hispanic
- ☐ Not Latinx / Hispanic
- ☐ Prefer not to answer
- ☐ Other \_\_\_\_\_

End of Block: Demographics

---

Start of Block: Diagnosis and Identity

Q7 What is your current autism diagnosis?

If you have more than one autism diagnosis, please select the most recent one

- ☐ Autism or Autistic disorder
- ☐ Autism Spectrum Disorder (ASD)
- ☐ Asperger's Syndrome
- ☐ Pervasive Developmental Disorder-Not Otherwise Specified (PDD-NOS)
- ☐ Pervasive Developmental Disorder (PDD)
- ☐ Childhood Disintegrative Disorder (CDD)
- ☐ I don't have an autism diagnosis

*Skip To: End of Survey If What is your current autism diagnosis? If you have more than one autism diagnosis, please select... = I don't have an autism diagnosis*

---

Q8 Which of the following most accurately describes your agreement with this statement:

**"I view my autism diagnosis as a difference, and not a disability"**

- ☐ Strongly agree
- ☐ Somewhat agree
- ☐ Neither agree nor disagree
- ☐ Somewhat disagree
- ☐ Strongly disagree

End of Block: Diagnosis and Identity

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Start of Block: Genetics of autism and genetic testing for autism

Q9 This study is about genetic testing for autistic people. Genetic testing involves analyzing a person's DNA that is collected through spit/saliva or blood in order to find differences that might explain why a person has autism. These differences can be unique to the person (de novo) or inherited from a parent.

Please answer the following questions to the best of your ability. If you don't know the answer to a question, please take your best guess and do not use outside resources to answer.

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Q10 Is genetic testing available for people with autism?

- ☐ Yes
- ☐ No
- ☐ I don't know

-----

Q11 What percent of autistic persons have an identifiable genetic change that is suspected to be the cause of their autism?

0 10 20 30 40 50 60 70 80 90 100

0-100%



Q12 Are there national medical guidelines recommending that every person with autism receive genetic testing?

- ☐ Yes
- ☐ No
- ☐ I don't know

End of Block: Genetics of autism and genetic testing for autism

Start of Block: Perceptions about genetic testing for autism

Q13 In your opinion, what are reasons to get genetic testing for autism? (select all that apply)

- ☐ To learn more about the cause of autism
- ☐ To promote early detection and intervention for children with autism
- ☐ To identify and treat health conditions that may develop
- ☐ To access new services and social support
- ☐ To assist with family planning
- ☐ To assist with financial planning
- ☐ Other \_\_\_\_\_

Q14 In your opinion, what are reasons to decline genetic testing for autism? (select all that apply)

- ☐ It could cause psychological distress (i.e. low self-esteem, anxiety, depression)
- ☐ It wouldn't be helpful
- ☐ It is too expensive
- ☐ It goes against my religious or cultural beliefs
- ☐ It could cause conflict within my family
- ☐ The process of getting tested could cause discomfort (i.e. fear of needles)
- ☐ Privacy concerns (i.e. fear of how the genetic data will be used)
- ☐ It could affect eligibility for insurance benefits
- ☐ It could affect employment opportunities
- ☐ It is too difficult to get an appointment with a genetics or other provider that could coordinate the testing
- ☐ It could divert funds away from improving the quality of life for autistic persons
- ☐ Other \_\_\_\_\_

End of Block: Perceptions about genetic testing for autism

---

Start of Block: Impact of genetic testing for autism

Q15 Have you received genetic testing for autism?

- ☐ Yes
- ☐ No
- ☐ I don't know
- ☐ Prefer not to answer

---

*Display This Question:*

*If Have you received genetic testing for autism? = Yes*

Q17 What was the result?

- ☐ Positive result (a genetic change was found that is known to cause autism)
- ☐ Negative result (no genetic change was found that is known to cause autism)
- ☐ A variant of uncertain significance (a genetic change was found, but it is unknown whether it causes autism)
- ☐ I don't know
- ☐ Prefer not to answer

---

*Display This Question:*

*If What was the result? = Positive result (a genetic change was found that is known to cause autism)*

Q18 Did you receive a particular diagnosis based on this result?

- ☐ Yes (please specify) \_\_\_\_\_
  - ☐ No
  - ☐ I don't know
  - ☐ Prefer not to answer
-

*Display This Question:*

*If Have you received genetic testing for autism? = Yes*

Q19 How did genetic testing for autism positively impact your life? (select all that apply)

- ☐ I learned more about the cause of my autism
- ☐ It enabled early diagnosis and intervention for my autism
- ☐ It helped identify and treat health conditions that I developed
- ☐ It helped me access new services and social support
- ☐ It helped me make family planning decisions
- ☐ It helped me to plan financially
- ☐ Other \_\_\_\_\_

*Display This Question:*

*If Have you received genetic testing for autism? = Yes*

Q20 How did genetic testing for autism negatively impact your life? (select all that apply)

- ☐ It caused me psychological distress (i.e. low self-esteem, anxiety, depression)
- ☐ The process of getting the genetic testing was uncomfortable (i.e. blood draw, hospital visits)
- ☐ It caused tension or conflicts in my family
- ☐ It hurt my ability to get insurance benefits
- ☐ It hurt my ability to find employment
- ☐ The test result was not explained to me in enough detail, or wasn't explained to me at all
- ☐ Other \_\_\_\_\_

---

*Display This Question:*

*If Have you received genetic testing for autism? = Yes*

Q23 What was the overall impact of genetic testing on your life

- ☐ It had a strong positive impact on my life
- ☐ It had a positive impact on my life
- ☐ It had neither a positive nor negative impact on my life
- ☐ It had a negative impact on my life
- ☐ It had a strong negative impact on my life

---

*Display This Question:*

*If Have you received genetic testing for autism? = No*

Q24 Why haven't you received genetic testing for autism? (select all that apply)

- ☐ I was unaware that genetic testing was available
- ☐ It was never offered to me by a healthcare professional
- ☐ It wasn't available when I was a child
- ☐ It wouldn't be helpful
- ☐ It's too expensive
- ☐ It goes against my religious or cultural beliefs
- ☐ I didn't want to know
- ☐ Other \_\_\_\_\_

---

*Display This Question:*

*If Have you received genetic testing for autism? = No*

Q25 Do you want genetic testing for autism?

Please note, your responses are confidential, and will not be shared with a healthcare professional. You will not be offered genetic testing if you select 'yes'

- ☐ Yes
- ☐ No
- ☐ I'm not sure
- ☐ Prefer not to answer

End of Block: Impact of genetic testing for autism

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Start of Block: Final questions



Q26 Several leading physician groups recommend that every child with autism receive genetic testing. Do you agree with the national medical guidelines recommending that every child with autism should receive genetic testing for autism?

- ☐ Yes
  - ☐ No
  - ☐ I'm not sure
-

Q27 Out of the following scenarios, how much do you agree that they should receive genetic testing for autism?

	Strongly agree	Somewhat agree	Neither agree nor disagree	Somewhat disagree	Strongly disagree
Any person with autism	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
A person with autism and a psychiatric condition, such as anxiety or ADHD	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
A person with autism and a medical condition, such as seizures or movement disorders	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
A person who has other close family members with autism	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
A pregnant couple who wants to know if their baby is at increased risk to have autism	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

No person  
with autism  
should  
receive  
genetic  
testing for  
autism

☐ ☐ ☐ ☐ ☐

---

Q28 What is your overall opinion of genetic testing for autism?

- ☐ I have a strong positive opinion
- ☐ I have a positive opinion
- ☐ I have neither a positive nor negative opinion
- ☐ I have a negative opinion
- ☐ I have a strong negative opinion

---

Q30 Is there anything else you would like to say about genetic testing for autism?

\_\_\_\_\_

---

Page Break

Q29 The following questions are about prenatal genetic testing for autism. Prenatal genetic testing is when genetic testing is done on an unborn fetus to see if they have a genetic condition.

In the present day, there are prenatal genetic tests that look for genetic conditions associated with both autism and medical problems (i.e. Fragile X Syndrome). However, there are currently no prenatal genetic tests that look specifically for autism.

If a prenatal genetic test looking specifically for autism becomes available in the future, which of the following statements do you agree with about its potential application? (select all that apply)

- ☐ It may lead to pregnancy termination (abortion) of fetuses expected to develop autism
  - ☐ It may help families better prepare for their child's needs
  - ☐ It may lead to earlier access to support services for autistic children and their family members
  - ☐ It may help doctors diagnose and provide care to autistic children
  - ☐ I am very concerned about the development of a prenatal genetic test specifically for autism
  - ☐ I do not agree with any of these statements
- 

Q28 Are you in favor of scientists developing a prenatal genetic test looking specifically for autism?

- ☐ Yes
  - ☐ No
  - ☐ I'm not sure
-

Q29 Is there anything else you would like to say about prenatal genetic testing specifically for autism?

---

End of Block: Final questions

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Start of Block: Compensation

Q27 Thank you for participating in this survey! **If you would like to receive a \$5 Amazon gift card, please click on the link below.** This will take you to a new survey that will ask for your email address. This way, we won't be able to connect your email address to the responses you've provided on this survey.

[Click here for the link](#)

End of Block: Compensation

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