

CAN GENETIC RESEARCH INVOLVEMENT MOTIVATE PARENTS TO PURSUE CMA  
GENETIC TESTING FOR CHILDREN WITH AUTISM SPECTRUM DISORDER?

by

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## Introduction

Autism spectrum disorder (ASD), a neurodevelopmental condition characterized by impaired social behavior and restricted interests, continues to present (Heil & Schaaf, 2013). According to the newest autism prevalence estimate (Autism and Developmental Disabilities Monitoring) by the Centers for Disease Control and Prevention (CDC), one in every 68 children in the United States has been diagnosed on the spectrum (CDC, 2014). The prevalence of ASD increased roughly 123% from 2002 to 2010, but has remained unchanged between 2010 and 2012. This report also suggests that while a large margin of parents of children with ASD have concerns about their children's medical or educational records, less than half of these children receive developmental evaluations or diagnosis prior to age 3 (CDC, 2014).

While the etiology of ASD is still uncertain, the existence of a connection to genetic disorders is attested by a body of findings (Heil & Schaaf, 2013). ASD is found to be highly heritable, with the risk of a parent of a child with ASD having another child being affected by ASD being 7% if the first child is female and 4% if the first child is male (Schaefer & Mendelsohn, 2013). If multiple children have ASD, the risk of recurrence is estimated to be between 33–50% (Miller et al., 2010; Schaefer & Mendelsohn, 2013). Currently, chromosomal disorders such as Fragile X and Down's Syndrome have been identified as comorbid for ASD (Shen & Miller, 2014). However, it is believed that roughly half of ASD genetic causes involve *de novo*, or non-syndromic, mutations (Bauer & Msall, 2011). Of these, 8-21% of autism-linked genetic disorders exist in the form of a varying degree of point mutations known as copy number variants (CNVs) (Carter & Scherer, 2013; Heil & Schaaf., 2013). Genes that are considered CNVs for ASD can affect multiple points across numerous portions of the genome (Carter, M.,

& Scherer, 2013). The variety of distances and number of CNVs are believed to be related to the complex range of phenotypes appearing on the autism spectrum (Heil & Schaaf, 2013).

Chromosomal microarray analysis (CMA) has been reaffirmed by the 2013 revision guidelines of the American College of Medical Genetics (ACMG) as a first-tier genetic test for neurodevelopmental disorders, including ASD (Miller et al., 2010; Toruner, Dermody, & Tolia, 2012; Bauer & Msall., 2011). In this tiered approach, CMA is recommended as the first test for developmental disorders such as ASD. The diagnostic yield through CMA is found to be 15-20% in most studies, as well as almost 30% by prior screening with complex cases (Shen & Miller, 2014). In this procedure, a blood sample is taken from the patient, and the genetic material is then comprehensively scanned in comparison to a healthy genome control (Jiang et al., 2014). In identifying distinct variants from the control, this reveals mutations that may be related to the development of ASD in the patient (Toruner, et al., 2012).

The value of the results of CMA to ASD patients and caregivers in identifying these CNVs is outlined by the 2013 revised consensus statement of the AMCG (Schaefer & Mendelsohn, 2013). Obtaining a usable genetic diagnosis for autism is suggested to parents of affected children due to the possibility to influence treatment, management, and decision-making for family planning (Miller et al., 2010; McGrew, Peters, Crittendon, Veenstra-Vanderweele, 2012; Cuccaro et al., 2014). In addition to its prospective uses for those receiving the test, another outcome of undergoing CMA for neurodevelopmental disorders is this information's potential role to advance autism research (Miller et al., 2010; Schaefer & Mendelsohn, 2014).

In the 2010 consensus statement of AMCG, the use of genomic databases was recommended to increase the utility of findings from genetic tests. One such example was DECIPHER (Database of Chromosomal Imbalance and Phenotype in Humans Using Ensembl

Resources) hosted by the International Standard Cytogenic Array Consortium, which served as an as online password-protected storage system of genetic tests results (Firth et al., 2009; Miller et al., 2010). A more recent example that is specifically focused specifically on autism genomic results is SFARI Gene 2.0 organize by the Simons Foundation (Abrahams et al., 2013).

Platforms such as these are designed to share and present data from genetic testing patients among researchers (Miller et al., 2010). Professionals involved with this project can analyze a bank of test results recorded from patients to identify previously unidentified mutated genes which may contribute to the cause of ASD and other genetic disorders (Miller et al., 2010; Shen et al., 2014). By identifying and recording new candidate genes, the genetic causes that underlie ASD can be more accurately described and more greatly understood (Asadollahi et al., 2014). Thus far, the collection of this data has been useful in detecting and describing new CNVs, which can ultimately influence future diagnosis and treatment (Battaglia et al., 2013; Oikonomakis et al., 2016). However, further elucidation of suspected genetic components of ASD can only be driven by the contributions of results obtained from genetic testing data of affected individuals.

Parents can be regarded as the primary caregivers of children suffering from ASD and thus the main rational actor in testing decision making. When the phenotype of autism is diagnosed and is ruled to likely have an uncertain cause, genetic testing can be advised by a healthcare professional, typically the patient's pediatrician (Carter & Scherer, 2013; Jiang et al., 2014). Concerning the levels of parent interest, studies have consistently found that supportive attitudes to pursue genetic testing are high (Narcisa et al., 2013; Trottier et al., 2013; Chen, Huang, & Dhar, 2013; Amit, Couchon, Carr, Carayol, & Cohen, 2014). Despite positive perceptions as well as the potentially beneficial outcomes of a genetic diagnosis, actual

utilization of CMA among other genetic testing options are reported as being low among parents of children with ASD (Cuccaro et al., 2014; Peabody et al., 2016; Vande-Wydeven, Kwan, Hardan, & Bernstein 2012; Amiet, Couchon, Carr, Carayol, & Cohen, 2014; Selkirk, McCarthy-Veach, Lian, Schimmenti, & Leroy, 2009).

One possible factor of intention toward receiving CMA testing is the prospect of contribution to the genomic databases used in research, such as SFARI Gene 2.0. Regarding this outcome of testing, decision by parents to have a biological offspring undergo CMA may represent a form of participation in autism genetic research. Parent perceptions toward using their ASD-affected child's information in genetic research has been previously been found to be overall positive (Johannesen et al., 2016). Due to this information being passed on for study within a genomic resource for the use of researchers, electing to participate in research using CMA results can be considered an altruistic motivation. Throughout studies of autism genetic research, altruism is found as a consistent motivator (Trottier, et al. 2013; Singh, 2015; Johannesen, et al., 2016). For the Simon's Simplex Collection, it was discovered that at least two-thirds of parents participated to altruistically contribute to autism research (Singh, 2015). Trottier et al. (2013) found in an exploratory study that participation was motivated by an amalgam of both personal interest and altruism.

Previous studies have shown contributing to research motivates genetic testing intention among tests for a variety of conditions. Exploratory and pilot studies have identified furthering research as among the primary reasons given for testing for genetic causes of sudden cardiac death, smoking susceptibility, deafness, and a variety of cancers (Boudreault et al., 2010; Giordimaina, Sheldon, & Petty, 2014; Hallowell et al., 2010; Erskine, et al. 2014; Esplen et al., 2001). For quantitative studies involving genetic tests for conditions including BRCA,

Alzheimer's, and pancreatitis, testing intention has previously been associated with wanting to help advance research (Roberts et al., 2003; Phillips et al., 2000; Applebaum-Shapiro, Peters, O'Connell, Aston, & Whitcomb, 2001; Geller, Doksum, Bernhardt, & Metz, 1999). Regarding genetic testing for a variety of psychiatric disorders, a questionnaire-based study also found that positive belief in genetic researchers to use this information to advance knowledge was also associated with participants' intention to undergo genetic testing (Laegsgaard, Kristensen, & Mors, 2009). Because desire to help with research motivates intention to test for other genetic disorders, this may also be true for intention regarding genetic tests for ASD. Despite this trend among other conditions as desire to pursue genetic testing as being motivator, cross-sectional studies have recommended that each test be properly assessed for its own factors of motivation and parameters that determine parents' perceptions of testing (Holly, 2011; Sweeny, Ghane, Legg, Huynh, & Andrews, 2014). To date, no available quantitative studies have measured the intention to test an ASD-affected child as it is associated with desire to help with research or perspectives about research.

Recently, several qualitative studies have been conducted to measure parents' intention in genetic testing for ASD, two of which were hypothetical studies focused on CMA (Reiff, Bernhardt, & Easley; Xu, Mitchell, Richman, & Clawson, 2016; Chen, Xu, Huang, Shweta, & 2013). Each has included parents' responses that suggest an interest in testing based on an acknowledgement of its use in ASD genetic research and desire to help advance these efforts. In a sample performed on an underserved population, desire to help with research was among most prominent items mentioned (Xu et al., 2016). While impact of altruism to help advance research on ASD features as a reason that parents would test, the relationship between testing and this altruistic motivation has not been documented. Information is needed to fill the gap regarding

the extent of this factor as a motivator for genetic testing intention among parents of children with ASD.

To better understand the relationship of desire to help with genetic research on autism and test intentionality, parents across Eastern North Carolina were surveyed about their perceptions. This sample was collected from the surrounding region from which Xu et al. (2016) previously detected desire to help with research as a motivator for interest in testing among parents. This study sought to answer two questions: (1 *Is interest in CMA genetic testing associated with parents' desire to contribute to autism research?*) (2 *Does perception about the impact of testing on autism research relate to parents' CMA intentionality?*)

## **Methodology**

### **Participants**

Those involved with the study were required to be the biological parents of at least one currently living child with autism. Participants were also required to be residents in one of the 44 counties in Eastern North Carolina. Age was necessitated at 18 years old, however age of the child of the participants with ASD was not restricted. English literacy was also required to read and complete the survey.

### **Design and Recruitment**

This study was a component of a larger interactive online survey distributed by email. The survey was designed by a panel of experts using instruments found from questionnaire-based studies which previously measured genetic testing perceptions, motivators, and barriers. It was then revised based upon revision sessions among the panel and the suggestion of a focus group of parents of children with autism, which included 20 members who were one-on-one interviewed. The completed 44-item instrument was uploaded to the online survey platform

Qualtrics for parents to complete through convenience sampling. The link to complete the study was distributed to parents of children with autism through email contact to advocacy organizations within the designated 44 counties of Eastern North Carolina. These were located by online listing, social media presence, and personal contact. In several instances, a paper survey was hand-delivered to participants in the local community and subsequently collected. Participation and successful completion of the survey was incentivized by a \$20 Walmart gift card.

### **Outcome Variables**

Regarding the survey's sample, demographic factors were taken of the sample. These measures included race, gender, average age, household income, and level of education.

The primary outcome variable was based on whether the parent would hypothetically take the child to receive genetic testing, which was coded as the dependent variable. In the survey, participants (parents) were asked the likelihood of them taking their child to a genetic testing for autism if recommended by a healthcare professional. Responses were measured on a four-point Likert scale which ranged "A great deal," "Somewhat," "Slightly," and "Not at all." t was coded Yes (1) if a participant responded, "A great deal" or "Somewhat", and No (0) was entered if a participant responded "Slightly" or "Not at all."

The other outcome variables were parents' *motivation* and three *perceptions* toward ASD genetic testing. These were coded as four dependent variables. For parents' *motivation* involving helping with ASD research, the section was prefaced with the instructions "*Please check whether the following factors would affect your decision-making*" The response item "desire to help with research" was included to represent research as a possible reason to pursue genetic testing. Responses were measured on a Likert Scale which ranged "Very likely,"

“Likely,” “Unlikely,” and “Very unlikely.” It was coded Yes (1) if a participant responded, “Very likely” or “Likely”, and No (0) was entered if a participant responded “Unlikely” or “Very unlikely”.

To represent parents’ *perception* about the relationships of genetic testing outcomes and ASD genetic research, three items were included in the survey. These items were prefaced with the instruction for the participant to “*Please check whether you agree or disagree with the following statements:*”. Each sentence was based upon prior qualitative findings about parent perceptions of research outcomes related to genetic testing of their children. The first measure was based on positive belief about the influence of genetic testing upon ASD research, which stated “Genetic testing can positively impact autism research.” The second was of positive belief about the personal outcomes of ASD research, which stated: “Genetic Research improves my child’s quality of life.” The third was a negative perception about the personal outcomes of ASD genetic testing regarding ASD research which stated: “Genetic testing would only be beneficial for autism research.” Responses were measured on a four-point Likert scale which included “agree.” For each statement, a “1” was recorded if the participant answered, “strongly agree” or “agree”; a “0” was recorded if the participant answered, “disagree” or “strongly disagree”.

### **Statistical Analysis**

Frequency tables were used to measure the demographic variables of the sample. Fisher’s exact test and odds ratio was used to compare outcome variables examined in this study. This was done to assess the association between intention to test and responses related to both desire to assist with ASD research and perceptions about genetic research. Analyses were conducted using SPSS Version 23.

### **Results**

**Table I: Demographic Characteristics of Parents (N = 204)**

Characteristic	Percentage
Gender	
Male	11.87
Female	88.7
Ethnicity	
Caucasian	74.5
Black/African American	12.7
Native American	7.4
Hispanic	2.5
Other/Mixed	2.9
Education Level	
Did not Finish High School	2.5
High School or GED	11.8
Associate's Degree or Some College	45.6
Bachelor's Degree	25.0
Master's or Professional Degree	15.2
Income	
<\$20,000	13.2
\$25,000 - \$39,999	20.6
\$40,000 - \$59,999	25.5
\$60,000 - \$75,000	14.2
>\$75,000	25.5
Other	1.0

Descriptive statistics of the sample are presented in Table 1. A total of 204 (N=204) participants completely responded to all questionnaire items related to this study, 181 female and 23 male. All were parents of children with autism living in Eastern North Carolina. Majority of the participants were Caucasian and female. Most had completed some college and had a family annual income level above \$40,000. About 84.3% of the parents reported that they would take

their children to receive ASD genetic testing (Table I).

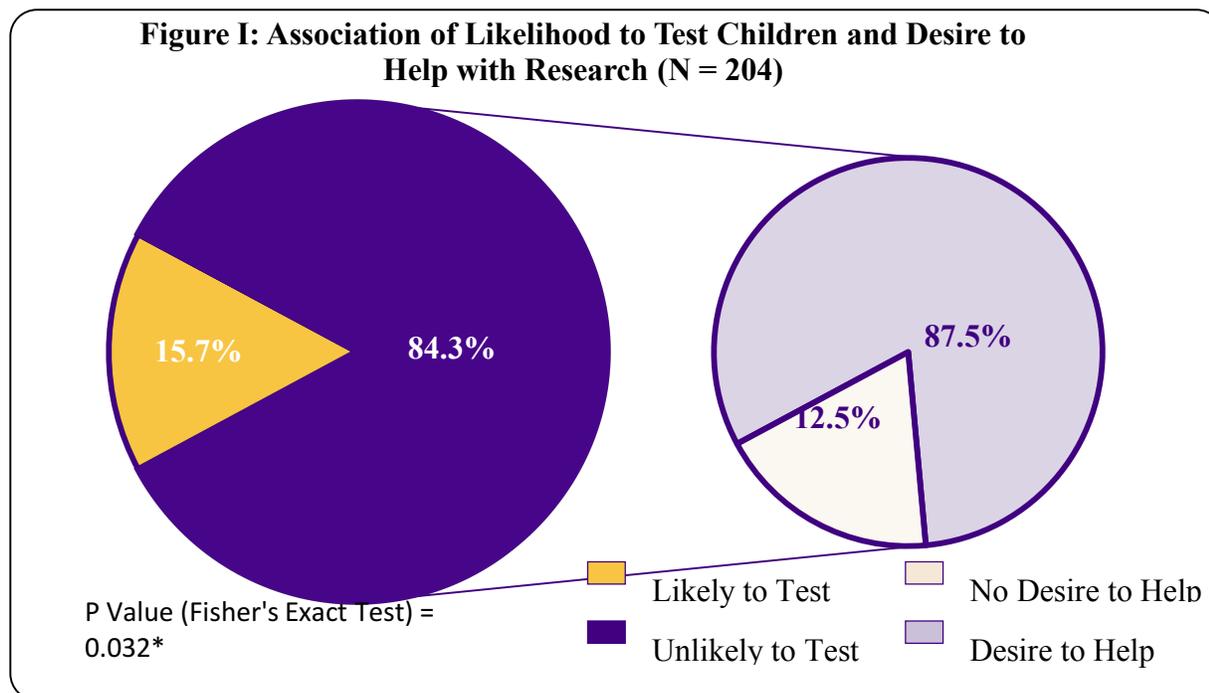


Figure I is a subset pie chart which displays the subdivision of those who desire to contribute to research within those who would likely test. The figure also provides an association between desire to help with research and likelihood to test their child ( $p = 0.032^*$ ) as determined by Fisher's exact test as well as with an odds ratio of 2.62.

**Table II: Association of Likelihood to Test Children and Perceptions of Research (N = 204)**

<i>Please check whether you agree or disagree with the following statements:</i>	p value*	odds ratio
“Genetic testing can positively impact autism research”	0.001*	13.0
“Genetic research improves my child’s quality of life”	0.002*	3.63
“Genetic testing would only be beneficial for autism research, not me or my family”	0.340	0.676

\*By Fisher’s Exact Test

The relationship between likelihood to test and perceptions about ASD research was measured in Table II. The text of the item is displayed along with the outcome of Fisher's test as well as the odds ratio measured. For perception about genetic testing benefiting autism research, this item was found significantly associated at P value of 0.001. The odds ratio was also found at a likelihood of 13.0. For the relationship between decision to test a child and belief that autism genetic testing favorably impacts the child's quality of life, these two measures were also significantly associated with a p-value of 0.002. The odds ratio was also measured at 13.0. Regarding parental belief that genetic testing would only beneficial to research, this result would not be significantly associated, with a p-value of .340. The odds ratio that was presented was also found to be .676.

## **Discussion**

This study provides insight about the possible decision making of parents of children with autism when pursuing genetic testing. The intention of parents within our study to have their children receive testing was found relatively high at 84.3%, which suggests that majority of participants are likely to consider having their ASD-affected child undergo CMA. This number is consistent with existing findings about the level of parent interest toward ASD genetic testing across quantitative studies, which ranged from 80% to 86% (Cuccaro et al., 2014; Amiet et al., 2014; Narcissa et al., 2012). While not exclusively for genetic testing, Vande Wydeven et al. (2012) also found parent interest in utilizing genetic services at 90%. For CMA specifically, quantitative studies have found among samples 69% and 85% (Chen et al., 2013; Xu et al. 2016). The high intention to test measured in this study furthers the need to investigate factors that are considered important by parents related to testing decision, particularly for use in elucidating reasons for low CMA uptake in the clinical setting.

Most significantly, parental intention toward CMA genetic testing of their children within this study was associated with the prospect of contributing to research. This highlights that this factor, which has been consistently described in qualitative studies on parent perception about CMA could indeed be predictive of test intention (Chen et al., 2013; Reiff et al., 2015; Xu et al., 2016). The finding that increase in intention to test is associated with desire to support research corroborates existing data with similar findings from quantitative studies about genetic testing intention (Roberts et al., 2003; Phillips et al., 2000; Applebaum-Shapiro et al., 2001; Geller et al., 1999). However, these studies represent findings on personal motivation for genetic testing rather than for that involving another family member, such as a biological child. While participants were not answering for a testing decision involving themselves, this provides further evidence of genetics research advancement as a motivator for genetic testing intention. These results represent the first quantitative findings for an ASD genetic test that considers research contribution as a factor of intention for genetic testing of a child.

Moreover, within this study, decision-making is also shown to be partly influenced by expectations about testing outcomes on ASD research. Data from this study partly supports statements made by parents of children with ASD in previous exploratory studies about CMA perceptions. Intention increased with belief that genetic testing could advance ASD research, as was brought up by parents in existing studies (Chen et al., 2013, Reiff et al., 2015; Xu et al., 2016). Intention also increases with belief that assisting these efforts would improve their ASD child's "quality of life." It was hoped that research would offer their child additional support at some future point by parents that Reiff et al., (2015) interviewed. However, negative attitudes about genetic testing for ASD, such as that testing outcomes would not be personally helpful, was challenged by lack of significance of this response item (Xu et al., 2016). This data would

suggest that greater parental belief and positive value of the outcomes of the elements surrounding testing could serve as a motivator for testing intention. This would also indicate that negative perceptions about outcomes do not have a direct impact on hypothetical parental decision for genetic testing.

Given that parents have demonstrated that genetic research is potentially a factor in test decision, health education studies have yet to explore possible value in affecting intention among parents. Enhancing knowledge and formation of beliefs on this subject may have a different effect on hypothetical testing decision beyond awareness. For this effort, it is necessary that educational interventions that can demonstrate the change of intention through providing information about research efforts through genetic testing for ASD.

## **Conclusion**

The implications of this study inform existing data on parental intention about genetic testing for ASD. Our results demonstrate an association between beliefs and motivations surrounding ASD research and testing decision, which at this point have not been assessed outside of qualitative findings. The possible outcomes of receiving positive information about research upon genetic testing intention are thus indicated by this study. Parents may wish to test their ASD-affected child at least partially because they want to advance research, they perceive in genetic testing their child benefits research, or because they believe research developments enhance the prospects of their children. Conversely, this study demonstrates that negative perceptions about genetic research outcomes from testing, such as that they are not personally useful, do not impact testing intention of parents. This would indicate that negative assessments not be a factor in decision alongside positive assessments of outcomes related to genetic research. Since unfavorable perceptions can be negated, it could thus be expected that providing

information related to the role of genetic testing in ASD research would have a positive impact on testing intention of parents.

Especially because research contribution is an outcome not immediately experienced by parents in having their child undergo CMA, the worth of these findings is predicated on explaining this aspect of testing to parents. It could be recommended that current methods of patient education be modified to incorporate sufficient material about ASD research prospects. Health educators seeking to inform parents about genetic testing for ASD should include details within programs about the prospects of CMA to benefit DECIPHER, DGV, and other genomic resource endeavors within ASD research. In the clinical setting, both physicians and genetic counselors should consider the aspect of assisting genetic research within their support provided to parents of patients referred for advanced-level ASD genetic testing. By providing sufficient information about all considerations related to testing decision, professionals can more effectively direct their guidance and support offered to families about CMA. By supporting parent understanding of this endgame outcome for their child's procedure, this can be expected to increase their likelihood to pursue genetic testing. With greater awareness about this possibility, parents will be able to make a more informed decision about their utilization of genetic services and in doing so potentially advance efforts to better understand ASD.

### **Limitations**

Several factors limit the capabilities of this study's findings. For one, accuracy of information collection may be lowered through use of an online survey. Responses were under the assumption that respondents were in fact parents of children with autism residing in the designated counties of Eastern North Carolina. This study's validity was also dependent on participants answering each question honestly and carefully to reflect their true beliefs. The

survey was also relatively long and items related to this study were not all located in the same section. In that respect, the responses could have reasonably been skewed and biased upon mental state and level of focus through each portion of the survey. Parents also provided responses from a hypothetical scenario of genetic testing and may not represent a participant's views during actual testing experience.

Through participant recruitment, the study also did not produce a diverse sample. Despite a higher area of income inequality and racial diversity within the area of recruitment, majority of active members of contacted parent organizations were considerably comprised of Caucasian mothers of mid-level SES. Incidentally due to our recruiting method, the demographics of those taking the survey were skewed toward middle income, middle education, Caucasian, and female. This may lower the generalizability of this study toward parents of minority backgrounds or lower SES.

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