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Attitudes of Healthcare Professionals Towards the Utilization of
Genetics Professionals Following the Diagnosis of Autism Spectrum Disorder

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Abstract

Autism spectrum disorder (ASD) is a group of neurodevelopmental disorders characterized by social communication deficits and repetitive behaviors. A diagnosis of ASD can be made by various healthcare professionals, including developmental pediatricians, child neurologists, child psychiatrists, and child psychologists. As there is a significant genetic component to ASD, many professional organizations recommend that individuals diagnosed with ASD undergo some form of genetic testing; notably, psychological organizations do not make any such recommendation. Nevertheless, current literature has shown that most patients are not referred to genetics.

This study compared the utilization of genetic professionals by physicians and psychologists. It was hypothesized that the rate of referrals to a genetics provider would be dependent on the type of professional training; we compared physicians, who attended medical school, and psychologists, who obtained a doctorate in psychology. A Fisher's exact test was performed to compare the rate of referral of physicians versus psychologists. It was found to be statistically significant ($p < 0.05$), with a higher rate of referral among physicians, suggesting that greater focus on the genetics of ASD in psychology doctoral programs and the creation of standardized genetic testing guidelines following a diagnosis of ASD should increase the rate of referrals to genetics.

Key words

autism, autism spectrum disorder, genetic evaluation, genetic testing, genetic diagnosis, autism diagnosis, etiology

Introduction

Overview of ASD

Autism spectrum disorder (ASD) is a group of neurodevelopmental disorders characterized by deficits in social communication and nonverbal communicative behaviors, lack or understanding of relationships, restricted or repetitive patterns of behavior or interests, and hyper- or hypo-reactivity to sensory aspects of their environment. These symptoms occur independently of global developmental delay or intellectual disability, although ASD frequently co-occurs with intellectual disability (American Psychiatric Association, 2013). In the United States, it is estimated that 1 in 59 children have been diagnosed with ASD, with a higher prevalence in males and in children with certain genetic disorders (CDC, 2018).

Autism spectrum disorder is a multifactorial condition with a wide phenotypic spectrum that is influenced by both genetic and environmental factors. ASD is associated with several genetic variants including large chromosomal abnormalities, single nucleotide polymorphisms (SNPs), microdeletions and microduplications, and copy number variations (CNVs); it can be inherited in a dominant or recessive manner and can result from both hereditary or *de novo* mutations (Miles, 2011; Yoo, 2015). Studies have also revealed the existence of high-risk ASD genes and susceptibility loci. In addition, numerous genetic and metabolic conditions have reported associations with ASD (Devlin & Scherer, 2012).

Diagnosis of ASD

In 2006, the American Academy of Pediatrics (AAP) Committee on Children with Disabilities issued a policy statement endorsing developmental surveillance at routine visits; subsequently, a 2007 AAP statement specifically recommended ASD screening at 18 and 24 months of age. Caregivers such as parents or teachers are often the first to notice behavioral and

social abnormalities in children and are encouraged to bring up any developmental or behavioral concerns at these well-child visits. At this time, developmental and/or ASD screening tools can be administered (AAP, 2016). After a positive result on an ASD-specific screening tool, the individual should be referred for early intervention, audiologic evaluation, and comprehensive evaluation (Plauché Johnson & Myers, 2007).

According to the Centers for Disease Control and Prevention (CDC) (2018), a primary care physician may choose to refer a child suspected of having ASD to a specialist such as a developmental pediatrician, child neurologist, child psychiatrist, or psychologist to make the diagnosis. Although assessment by a single healthcare professional is sufficient in making the diagnosis of ASD, it is considered best practice when the ASD evaluation is conducted by a team of specialists (Westman Andersson, Miniscalco, & Gillberg, 2013). However, a number of factors influence who inevitably makes the diagnosis, such as finances, insurance, and physician availability. For instance, developmental pediatricians often book appointments several months in advance due to high demand; sometimes, a diagnosis is needed sooner rather than later to receive early intervention and other services, and a different specialist is consulted (Rudy, 2018).

Genetics of ASD & Current Genetic Testing Guidelines

An estimated 20-25% of individuals with ASD have an identifiable genetic etiology, such as visible chromosome abnormalities on karyotype (5%), monogenic disorders (5%), and copy number variants (10-20%) (Miles, 2011). Genetic testing may have a higher diagnostic yield in affected individuals who are female, have dysmorphic features, and/or have concurrent conditions such as intellectual disability or seizures as opposed to individuals with isolated ASD (Plauché Johnson & Myers, 2007). Parents who have a child with ASD have a 18.7% chance of having another affected child; this recurrence risk increases to 33-50% if they have two or more

children with ASD (Ozonoff et al., 2011; Lauritsen, Pedersen, & Mortensen, 2005; Simonoff, 1998). These recurrence risk numbers do not account for ASD with a genetic diagnosis, which has a recurrence risk of up to 50% depending on inheritance.

Due to the high percentage of ASD attributed to discernible genetic factors, several professional organizations endorse specific genetic tests. The American Academy of Neurology (AAN) and the Child Neurology Society (CNS) currently recommend chromosome analysis and testing for Fragile X syndrome for individuals with ASD and intellectual disability, dysmorphic features, and/or a positive family history of Fragile X syndrome. AAP recommends chromosomal microarray and testing for Fragile X syndrome for all affected individuals (Barton et al., 2017). The American Academy of Child and Adolescent Psychiatry (AACAP) states that clinical discretion should be used to offer genetic testing (Volkmar et al., 2014). Yet according to the American College of Medical Genetics (ACMG) 2013 Practice Guidelines, first-tier testing for ASD should include karyotype, chromosomal microarray, and Fragile X syndrome testing for males; however, if the individual presents with a recognizable syndrome, genetic testing for that syndrome should be considered first. ACMG Practice Guidelines also include a second-tier testing strategy which includes *MECP2* sequencing for females (or *MECP2* duplication testing for indicated males), *PTEN* testing if the head circumference is over 2.5 standard deviations above the mean, and brain magnetic resonance imaging (MRI) if seizures, regression, or microcephaly is present. If first-tier testing returns with negative results, clinicians can consider whole exome sequencing (WES) as an appropriate next step (Schaefer & Mendelsohn, 2013); however, the current diagnostic yield for finding a Mendelian disorder in a child with ASD is quite low.

Psychological associations such as the American Psychological Association (APA) do not have specific practice guidelines for genetics as it relates to ASD management. Of the three approved Clinical Practice Guidelines provided by the APA, none are specific to ASD (S. Rose, personal communication, April 17, 2019). Although an APA clinical guide for diagnosing ASD makes mention of a recognizable genetic component for ASD, they do not recommend genetic testing due to cost and lack of effect on medical management (Durand, 2014).

While many organizations acknowledge the utility of genetic testing in ASD and recommend specific tests, there is currently no clear consensus between organizations on what should be offered to patients. Furthermore, a 2017 survey of 15 healthcare providers noted that it was difficult to adhere to genetic testing guidelines due to the lack of uniformity (Barton et al.).

Parent Perspectives on Genetic Testing

Several studies have demonstrated that many individuals with ASD have not met with a genetics professional nor undergone some form of genetic testing (Amiet et al., 2014; Selkirk, McCarthy Veach, Lian, Schimmenti, & LeRoy, 2009; Vande Wydeven, Kwan, Hardan, & Bernstein, 2012). In fact, one publication noted that the majority of parents of children with ASD have never heard of genetic testing (Chen, Xu, Huang, & Dhar, 2013). Yet many parents whose children did have chromosomal microarray reported that the results were moderately-to-extremely helpful (Reiff et al., 2015). Reported benefits of genetic testing included (but were not limited to): family planning; contributing to research; finding the etiology for their child's condition; guiding treatment/management of disease; obtaining early intervention and services; assisting with health insurance coverage; and connecting families with a "genetic ASD" to a support group (Reiff et al., 2015; Hayeems, Babul-Hirji, Hoang, Weksberg, & Shuman, 2016; K. Gallagher, personal communication, May 14, 2018; Chen, Xu, Huang, & Dhar, 2013).

On the other hand, families who received variants of uncertain significance or negative results were less likely to find genetic testing helpful. Nonetheless, it was speculated that families were more likely to have a positive experience regardless of test results if testing was accompanied by genetic counseling (Reiff et al., 2015).

Even with a diagnostic yield of only 25%, there are clear benefits to a genetics evaluation. There is a general consensus between clinicians and researchers working in the field of pediatric autism spectrum disorder that genetic testing can be useful for patients and their families for medical management and coordination of care. Most professional organizations recommend that individuals with ASD receive some form of genetic testing, but given that many parents report that their children with ASD have not had testing, or in some cases, have not even been offered testing, little data exists on how many individuals are actually referred to and meet with a genetics professional. As such, our survey explored attitudes of healthcare professionals towards the utilization of genetics professionals as a component of the ASD diagnosis. Our study specifically compared two groups of healthcare professionals: physicians, who received their education from a medical school (M.D. or D.O.); and psychologists, who obtained a doctorate in psychology (Psy.D. or Ph.D.). We hypothesized that the rate of referrals to genetics would correlate with the referring professional's training and that physicians are more likely than psychologists to make this referral.

Methods

Study Participants and Design

Approval for this study was obtained from the Institutional Review Board at Sarah Lawrence College. Data collection occurred from November 10, 2018 through February 24,

2019. Study participants were healthcare providers who diagnose autism spectrum disorder in Canada and the United States of America. Recruitment was done by email, which provided information about the objectives of the study, risks of participation, and the survey link. Potential participants were informed that clicking the survey link would be considered informed consent; however, they could exit the survey at any time to withdraw their participation and consent, and their answers would be excluded from data analysis.

Several approaches were utilized to solicit study participants:

1. Emails were sent to medical and psychological professional organizations requesting distribution of the survey using a listserv or direct emails to its members.
2. Emails were sent to the following:
 - a. Autism Speaks: All individuals with websites or emails listed in the Evaluation & Diagnosis section.
 - b. Autism Junction: All individuals with websites or emails listed in the Diagnostic/Assessment section.
 - c. Association of University Centers on Disabilities (AUCD): All directors and co-directors of University Centers for Excellence in Developmental Disabilities (UCEDDs) and Leadership Education in Neurodevelopmental and Related Disabilities (LEND).
 - d. New York State Psychological Association: All individuals with websites or emails listed under Assess/Eval Neuropsychological Conditions and Autism and Asperger's Syndrome.
3. The survey was also shared and distributed by personal and professional contacts of the principal study investigators.

Instrumentation

This survey was distributed online and hosted by SurveyMonkey. To determine eligibility, survey participants were initially asked if they diagnose children with autism spectrum disorder; if they answered “No,” they were not permitted to continue the survey. Responses were anonymous, although some demographic information (such as gender and state/province) was collected at the end of the survey.

Participants were asked questions in a variety of formats including: multiple choice - single best answer; multiple choice - check all that apply; and rating statements using a traditional 5-point Likert scale. Many of the questions also included an option to provide a free response under “Other.” Four questions used skip logic: based on the answer to one question, participants were directed to specific follow-up questions regarding the original answer. A copy of the survey is supplied in Appendix A.

Data Analysis

Survey responses were grouped into two categories, physicians and psychologists, based on the answer to question 3 – “What type of healthcare professional are you?” Responses of “Yes” and “Sometimes” to question 9 – “Do you routinely refer your patients with ASD to a genetics professional?” – were also grouped together.

Using R, a Fisher’s exact test was performed to compare physician and psychologist responses to question 9. A p -value of < 0.05 was considered statistically significant. Answers to other survey questions were analyzed on a qualitative and descriptive basis with no statistical methods applied.

Results

Participant Characteristics

In total, 104 responses were received. Twenty-nine responses were excluded: 27 did not complete the survey and 2 did not fit the criteria of a healthcare provider who clinically diagnoses children with ASD. Of the 75 responses which remained for analysis, 34 (44.3%) respondents were physicians and 41 (55.7%) were psychologists. Eighteen (24.0%) participants identified as male; 56 (74.7%) identified as female, and 1 preferred not to answer. Most of the participants were between the ages of 35-44 and 55-64 (Table 1). On average, study participants had 20 years of experience in their healthcare specialty with a minimum of 2 years and a maximum of 45 years. Twenty-one respondents practiced in New York, with 7 in California and 4 in Texas; other represented states had 3 or fewer respondents per state, and several US states and Canadian provinces were not represented in the sample.

Physicians reported diagnosing patients in private practice (5; 14.7%), local hospitals (9; 26.5%), and schools (2; 5.9%); over half (52.9%) made diagnoses in university-/community-based clinics, academic institutions, and child development centers. Thirty-four (82.9%) psychologists diagnosed most of their patients in a private practice, and the remaining psychologists diagnosed patients in local hospitals (4; 9.8%) or university-/community-based clinics (3; 7.3%) (Table 2).

Continuing Education

Over 80% of all participants reported keeping up to date with evolving practices in their respective fields by attending courses and conferences, reading research papers in their field of expertise, and regularly consulting with colleagues in related fields; over half (52.0%) of all participants regularly checked their specialty's practice guidelines relating to ASD (Table 3). There was no difference in continuing education between physicians and psychologists.

Characteristics of Study Participant's Patient Population

Nearly half (47.1%) of physicians diagnosed over 50 children per year compared to 17.1% of psychologists. Only 1 physician diagnosed less than 10 children per year, while 12 (29.6%) psychologists diagnosed less than 10 children per year. The ratio of male to female diagnoses was approximately 2:1 for both physicians and psychologists; however, the average age at diagnosis reported by psychologists was higher than the average age of diagnosis reported by physicians (Table 4).

Referrals Made Following a Diagnosis of ASD

When asked about referring to a genetics professional, 26 (76.5%) physicians and 18 (43.9%) psychologists routinely or sometimes referred to a genetics professional (Table 5). A Fisher's exact test to compare physician and psychologist responses was performed and found to be statistically significant ($p = 0.005$) (Figure 1).

Participants who responded that they routinely refer to a genetics professional were subsequently asked additional follow up questions regarding the referral process and follow-up. Ten (76.9%) physicians provided their patient with specific contact information for a genetics professional, and 5 (38.5%) contacted the genetics professional directly to set up an appointment. Eleven (84.6%) physicians followed-up with their patient to ensure they attended their appointment; all did this in person, and some also followed-up by phone (18.2%), email (18.2%), or mail (9.1%). Nine (69.2%) physicians followed-up with the genetics provider (Table 6).

Five (55.6%) psychologists provided the patient with specific contact information, and 2 (22.2%) personally contacted the genetics professional; 4 (44.4%) psychologists suggested a genetic evaluation or provided non-specific referral information such as the National Society for Genetic Counselors (NSGC) website or a list of genetics providers in the area. One (11.1%) psychologist followed-up with the patient by phone and email to ensure they attended their

appointment. Two (22.2%) psychologists directly followed-up with the genetics professional (Table 6).

Reasons for Referring to a Genetics Professional

All respondents were asked to rate statements regarding their rationale as to why they did or did not refer to a genetics professional using a 5-point Likert scale. Responses of “Strongly Agree” and “Agree” as well as “Strongly Disagree” and “Disagree” were combined for analysis purposes (Table 7).

Of the 13 physicians and 9 psychologists who routinely referred to a genetics professional, the majority of both groups agreed that their motivation was to obtain genetic testing and determine if there was an underlying genetic condition associated with ASD. Ten physicians and 7 psychologists agreed they would refer to a genetics professional for the family to receive information on recurrence risks. Seven psychologists would make a referral in order for the patient to learn about the genetics of ASD. Six physicians would also be inclined to refer for this reason, but another 6 physicians selected “Neutral.”

When asked if contribution to research was a motivation for the referral, 5 physicians (38.5%) and 5 psychologists (55.6%) agreed with the statement. The remaining physicians and psychologists reported they were neutral about or disagreed with this reason. There was no strong leaning towards agreement or disagreement for both groups on whether the referral was made to help with medical management.

Eight physicians and 4 psychologists indicated they referred to a genetics professional to be consistent with their respective organization’s guidelines; 5 physicians and 3 psychologists responded “Neutral,” and 2 psychologists disagreed with this statement.

Study participants who “Sometimes” referred their patient to a genetics professional (13 physicians and 9 psychologists) were also asked to rate statements regarding their motivation for doing so. All physicians and psychologists within this group agreed they would make a referral if their patient had dysmorphic features. The majority of physicians and psychologists would make a referral if the patient had abnormal imaging or laboratory results, if the family requested a genetics evaluation, if the patient had health problems in addition to ASD, and/or if the patient had a family history of ASD. One psychologist would not refer if their patient had additional health problems; 1 physician and 1 psychologist would not refer for a family history of ASD.

Reasons for Not Referring to a Genetics Professional

Eight physicians and 23 psychologists indicated that they would not refer to a genetics professional (Table 8). While both groups agreed that ASD has a genetic component, 1 physician and 7 psychologists believed that genetic testing would not affect the medical management of a patient with ASD; 1 physician and 8 psychologists had no opinion about this factor. Five physicians (62.5%) reported that they do not refer to genetics because they order their own genetic testing. 22 psychologists responded that they do not order their own genetic testing; one psychologist responded “Neutral.”

More than half of physicians had a genetics department accessible (5; 62.5%), and 87.5% knew where to refer their patient. One physician selected that the reason she did not refer to a genetics professional was because the patient already had a diagnosis; the 7 other physicians disagreed with this reasoning. The main reasons the psychologists did not refer to a genetics professional were that the patient already had a diagnosis (12; 52.2%), the clinic in which the psychologist worked did not have a genetics department accessible (16; 69.6%), and/or the psychologist did not know where to refer the patient (15; 65.2%).

Most of the respondents did not cite the patient's inability to afford another appointment or that the patient had too many appointments as a reason to not refer to a genetics professional.

Genetic Testing Ordered by Survey Participants

Study participants were asked if they ordered any genetic testing directly (Table 9). Almost half (16; 47.1%) of the physicians selected "Yes," with 10 (29.4%) physicians selecting "Sometimes," and 8 (23.5%) physicians answering "No." Thirty-three psychologists (80.5%) responded that they do not order genetic testing on their patients with ASD; 8 psychologists responded "Yes" or "Sometimes" despite not having the necessary licensure to order testing.

Respondents who selected "Yes" were asked to specify what type of genetic testing was most commonly ordered. All respondents ordered Fragile X syndrome testing and chromosomal microarray (CMA). Five physicians chose to order whole exome sequencing (WES), 3 physicians selected karyotype, and 2 physicians opted for a Next Generation Sequencing (NGS) panel. One physician selected "Other" and commented that he ordered whole genome sequencing (WGS) and occasionally DNA methylation studies for select patients.

With the exception of 1 physician, all physicians indicated that they understood how to interpret genetic testing. Eight psychologists stated that they knew how to interpret genetic testing results, whereas 10 psychologists responded that they did not know how to interpret genetic testing results. The remaining psychologists gave no indication on whether or not they were comfortable interpreting these type of results.

Discussion

Autism Spectrum Disorder (ASD) is a multifactorial condition affecting 1 in 59 children in the United States; approximately 20-25% of individuals with ASD have a genetic etiology

discoverable by current testing methodologies (CDC, 2018; Miles, 2011). Ideally, the diagnosis is made by a comprehensive evaluation from a team of medical and psychological specialists; however, if this is not possible, a single physician or psychologist is sufficient to make an ASD diagnosis (Westman Andersson, Miniscalco, & Gillberg, 2013). Exposing a genetic etiology can have implications for medical management and family planning; accordingly, several professional organizations recommend that individuals diagnosed with ASD are evaluated by a genetics professional and undergo genetic testing (Barton et al., 2017; Schaefer & Mendelsohn, 2013). Studies have shown that many families have not heard of nor been offered genetic testing for their child and agree that there are many benefits to genetic testing (Amiet et al., 2014; Reiff et al., 2015; Selkirk, McCarthy Veach, Lian, Schimmenti, & LeRoy, 2009; Vande Wydeven, Kwan, Hardan, & Bernstein, 2012). Our research revealed that there is currently no data as to how many children with ASD are being referred to a genetics professional, receiving a genetic evaluation, and/or undergoing some form of genetic testing. To investigate this further, we surveyed healthcare professionals who clinically diagnose ASD to explore attitudes regarding the use of genetics professionals following an ASD diagnosis. We hypothesized that there would be a difference in the rate of referral based on the healthcare professional's education.

Our survey found that physicians are more likely than psychologists to refer to a genetics professional after making the diagnosis of ASD; this result was statistically significant. Compared to psychologists, physicians were also more likely to follow-up with their patients to ensure that they attend their genetics appointment. It is likely that the rate of referral to a genetics professional was higher for physicians due to their medical training. Genetics is a component of the medical school curriculum, and physicians are taught about the various genetic implications for different diseases and conditions; thus, physicians should have a better understanding of the

importance of genetic testing. In contrast, based on the current APA Office of Program Consultation and Accreditation guide, genetics is not a clearly delineated or defined component of the coursework required for a psychology doctoral program (APA, 2018). Hence, psychologists are less likely to have had formal training that covers genetic factors underlying psychological conditions such as ASD and the repercussions of having a known genetic etiology for an ASD diagnosis. This may explain why a higher proportion of psychologists believed genetic testing would not affect the medical management of ASD or responded “Neutral” regarding this statement. While it is true that finding a genetic etiology does not always impact medical management, it does help to clarify recurrence risk and inform family planning (Chen, Xu, Huang, & Dhar, 2013).

In spite of this disparity, the rationale as to why respondents did or did not refer to a genetics professional was surprisingly similar among both groups. Many respondents who made the referral wanted their patient to see a genetics professional to rule in/out genetic conditions, obtain genetic testing, and help with family planning and medical management. This finding is consistent with the outcomes of parental surveys (Reiff et al., 2015) and clinical practice guidelines; the 2013 ACMG Practice Guidelines cite the primary roles of the geneticist to be determining the etiology, improving care, and providing genetic counseling (Schaefer & Mendelsohn, 2013). Of the respondents who “Sometimes” referred to genetics professionals, most agreed that dysmorphic features, additional health problems, abnormal laboratory testing or imaging, or a family history of ASD were sufficient reasons for referral.

Interestingly, all respondents who did not refer to genetics believed that ASD had a genetic component. Several of the surveyed participants indicated that the lack of referral to a genetics professional was contingent on accessibility, in contrast to a 2017 study that cited

insurance coverage as the main deterrent to the use of genetic testing (Barton et al.). Unlike physicians, most of the psychologists practiced in suburban areas, which are less likely to have large hospitals with multiple specialties, specifically a genetics department. In addition, the majority of psychologists diagnosed their patients in a private practice, compared to the majority of physicians who diagnosed most of their patients in local hospitals or academic hospital settings, where they are more likely to have direct access to a genetics professional. According to the National Society of Genetic Counselors (NSGC) 2018 Professional Status Survey, 61% of genetic counselors work in university medical centers or hospitals, with many providing direct patient care to pediatrics patients (NSGC, 2018).

A noteworthy finding in our study was that approximately half of the surveyed physicians diagnosed 50+ patients with ASD compared to 17.1% of the surveyed psychologists. This was surprising as previous research had shown that due to the long wait time for an appointment with a developmental pediatrician and low number of these physicians, many children with suspected ASD instead are diagnosed by child psychologists (Rudy, 2018). At the end of 2016, there were less than 800 developmental-behavioral pediatricians in the United States (American Board of Pediatrics, 2017) compared to an estimated 106,500 psychologists in 2012 (American Psychological Association, 2014). This finding may also be the result of a small sample size as well as the fact that our survey included 23 developmental pediatricians, representing 67.6% of our physicians, which causes an inherent bias to our results.

Due to the large number of practicing psychologists as well as shorter appointment wait times (Rudy, 2018), it was expected that patients would have easier access to psychologists, resulting in a younger average age of diagnosis. Yet results of the study revealed that the average age of diagnosis was actually higher for psychologists as compared to those diagnosed by

physicians. A possible reason for this disparity is that children with co-morbidities such as developmental delay or dysmorphic features are brought to the attention of physicians sooner than children with isolated ASD (Rudy, 2018), who may be more likely to receive official diagnoses from psychologists. Females may also receive an earlier evaluation by a physician due to the estimated 4:1 ratio of male to female patients often referenced in literature (CDC, 2018). Interestingly, in our study, this ratio was approximately 2:1, which may simply be an outcome of the small sample size.

Patients benefit when they save time by receiving genetic testing from the diagnostician with proper pre- and post-test counseling (Allen, Stoll, & Bernhardt, 2016); however, it cannot be ascertained if any of the physicians had formal training in returning genetic testing results or providing counseling about the benefits and risks of genetic testing.

There are no genetic testing recommendations in psychological association practice guidelines, and ordering genetic testing is only authorized by physicians; despite this, 2 psychologists indicated that they ordered genetic testing on their patients with ASD. One of these psychologists reported working in a private practice and the other reported working in a local hospital; neither left comments in any free-response section of the survey. Therefore, we cannot make a conclusion about the type of genetic testing ordered or how they accomplished this. Additionally, 17 of the 39 participants who indicated that they regularly check their specialty's practice guidelines were psychologists; 7 of the 9 psychologists who routinely referred to genetics also cited that they wished to be consistent with the guidelines in spite of our research not identifying recommendations for genetic testing following a diagnosis of ASD published by any North American psychological organization.

It was surprising to find that not only do psychological organizations lack genetic testing guidelines, but they also do not have practice guidelines specifically for ASD. Considering that psychologists make up a significant portion of diagnosticians, having a standardized method to promptly identify, diagnose, and treat these individuals can facilitate optimal care and subsequently enhance quality of life.

Study Limitations

A major limitation of our study is the sample size. Many of the professional organizations contacted were only willing to distribute our survey through their listserv if financial compensation was offered. Due to the financial limitations of a graduate thesis and no outside funding, we were unable to offer study participants financial compensation for completing the survey. As such, survey distribution relied primarily on personal and professional contacts as well as cold emailing individuals listed on Autism Speaks, Autism Junction, AUCD, and NYSPA. Most professional contacts were located in the metropolitan New York area and distributed the survey within closed networks or hospital systems; within these networks, healthcare providers had access to the same resources and therefore might have had similar survey answers. In addition, the metropolitan New York area has a high concentration of genetics professionals and thus creates bias in our data; approximately 5% of genetic counselors in the NSGC Professional Status Survey practice in New York, surpassed only by California at 12% (NSGC, 2018). Other geographic locations or centers located outside of large urban environments may have less association with a genetics department. With a more reliable means of distributing the survey and financial incentives, it is likely that we would have obtained a larger and more diverse sample.

Based on current literature, more patients receive their diagnosis from psychologists rather than physicians due to the considerable number of psychologists in North America (Rudy, 2018). As such, more psychologists were expected to be included in the sample population, especially given that hundreds of individuals from the aforementioned organizations were cold emailed. However, non-response is a common obstacle with cold emailing, due to lack of personalization and the possibility of the email getting flagged as spam by the server. Another important consideration is that, at least for Autism Speaks, individuals volunteer their contact information and self-identify as an ASD or developmental specialist; this also may be the case for other organizations. Therefore, we are unable to confirm the training and certification of these individuals.

Given the limited distribution of our survey, our sample population is not an accurate representation of the total population of healthcare providers that diagnose ASD. In addition, not all states and provinces were represented in our sample. Therefore, we are only able to make inferences and draw conclusions from our sample population rather than extrapolate our data to apply to the total population.

Practice Implications

It is clear that while the respondents of this survey recognize the benefits of utilizing genetic services for patients diagnosed with ASD, lack of access creates a substantial obstacle to referral. The rise in availability and acceptance of remote genetic counseling and telehealth services with the general public and healthcare providers may lead to more individuals with ASD referred for genetic evaluation. However, a particular challenge of telehealth is the inability to perform a physical examination, which is a key component of pediatric genetic counseling; findings from an in-person physical examination often have implications for the type of genetic

testing ordered. To combat this barrier, many pediatric telehealth programs use peripheral devices such as video cameras and specialty examination telescopes (Olson, McSwain, Curfman, & Chuo, 2018). Nonetheless, clinical genetic training programs could include courses and rotations that incorporate practical training for application of telehealth consultations. Through this training, future geneticists could become more comfortable with remote counseling and subsequently be more inclined to provide genetic evaluations to individuals who might otherwise be unable to see them.

Additionally, by providing further education to healthcare professionals about the potential genetic etiology of ASD, the rate of referral should increase, subsequently enabling more individuals to receive appropriate care. In particular, supplementary education should be provided to psychology programs and current psychologists emphasizing the importance of referring to genetics. Psychological organizations should create ASD practice guidelines that include information on the importance of genetic testing. These suggestions are especially crucial given that psychologists are more often the primary diagnostician of ASD due to their increased availability in comparison to physicians.

Research Recommendations

Further research should be conducted using a larger, more diverse sample size to assess whether the results from this study are truly reflective of those healthcare professionals who diagnose autism spectrum disorder. More specifically, future studies should focus on including additional participants from the particular physician specialties and states/provinces that were underrepresented in this study. Moreover, with greater research funding, this study might be expanded to include individual qualitative interviews with study participants to gain more in-

depth information about the research question, as Likert-style questions have inherent bias when pre-written statements are utilized.

A particular area of interest is psychologists who diagnose ASD. As they are unable to order their own genetic testing in their scope of practice, research may be conducted on the potential implications of authorizing psychologists to perform genetic testing, either specifically for ASD or for a wider range of conditions (ex. psychiatric disorders). The addition of genetic testing to psychologists' scope of practice would clearly require education and training to ensure that psychologists are capable of providing appropriate pre- and post-test counseling, order the correct genetic tests, and accurately interpret results. Researchers also must determine whether psychologists would even want to order genetic testing and be responsible for this information, and as counselors, how they would disclose different types of results to patients.

Most medical programs are composed of one first-year genetics course; however, there is little to no data on the specific content of these courses (Ormond, 2009). A 2006 study of physicians in specialties other than genetics found that many participants believed they were underprepared in genetics and that there was a lack of formal postgraduate genetics training (Burke, Stone, Bedward, Thomas, & Farndon). In spite of this, the surveyed physicians who ordered their own genetic testing responded that they were comfortable interpreting genetic testing; future studies may wish to evaluate the type of information physicians give their patients regarding the genetic contributions to ASD. For example, many of the surveyed physicians reported they would not refer to a genetics professional for a negative result, while they would for a positive result or a variant of uncertain significance. As such, it would be interesting to learn what information is provided to families, if any, regarding the limitations of current testing methodologies following a negative result

While our study looked into what genetic testing was most commonly ordered by physicians, it did not explore why this particular testing was ordered and if the physician utilized ASD genetic testing guidelines. Hence, future research should focus on the use of genetic testing following a diagnosis of ASD rather than referrals made to genetics professionals. Lack of consensus among providers and professional guidelines on what genetic testing should be provided is seen by providers as a deterrent for offering testing (Barton et al., 2017). With an increasing proportion of children receiving an ASD diagnosis per year and the rapid rate at which we are learning about genetics, it is crucial for professional organizations to collectively standardize what genetic testing should be offered and incorporate it into individual guidelines; these recommendations can then be translated into standard of care for individuals diagnosed with ASD.

Conclusion

Our study has shown that there is a significant difference in the rate of referral to genetics professionals between physicians and psychologists despite both groups acknowledging genetic contributions to the development of ASD; some possible reasons for the difference in the number of referrals include accessibility to a genetics professional and variation between groups in training and education, but there may be several other reasons that cannot be concluded due the limits of this sample size and population. It is important to continue investigating these reasons, as finding a genetic etiology for ASD has implications for family planning and medical management. We have proposed ideas to improve the number of genetics referrals such as increased use of telehealth services, the addition of genetics training to psychology program curricula, and incorporation of the utilization of genetic testing in ASD diagnoses to psychological organization guidelines.

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Compliance with Ethical Standards

Conflicts of Interest

Sydney Alexandra Lau and Tova Lejtman Wagner declare that they have no conflict of interest.

Human Studies and Informed Consent

All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000 (5). Informed consent was obtained from all patients for being included in the study.

Tables

Table I: Healthcare Provider Demographics

Type of Healthcare Professional	n	Percentage of Participants (%)
Pediatrician	3	4.0
Developmental Pediatrician	23	30.7
Pediatric Neurologist	6	8.0
Neurologist	1	1.3
Child Psychiatrist	5	6.7
Psychologist	41	54.7
Gender		
Gender	n	Percentage of Participants (%)
Male	18	24.0
Female	56	74.7
Prefer not to answer	1	1.3
Age		
Age	n	Percentage of Participants (%)
25-34	5	6.7
35-44	25	33.3
45-54	16	21.3
54-64	22	29.3
65+	7	9.3

Table II: Work Settings

	Physician		Psychologist	
	n	Percentage of Participants (%)	n	Percentage of Participants (%)
Private Practice	5	14.7	34	82.9
Local Hospital	9	26.5	4	9.8
School	2	5.9	0	0.0
Other	18	52.9	3	7.3

Table III: How Providers Keep Up to Date with Evolving Practices in Their Field

	n	Percentage of Participants (%)
Attend courses for continuing education credit	61	81.33
Attend conferences in my field of specialty	67	89.33
Regularly check specialty guidelines on ASD	39	52.00
Read research papers relating to my field of expertise	67	89.33
Regularly consult with colleagues in related fields	66	88.00
Other	2	2.67

Table IV: Patient Population Characteristics

	Physician		Psychologist	
Number of Children Diagnosed Per Year	n	Percentage of Participants (%)	n	Percentage of Participants (%)
Less than 10	1	2.9	12	29.3
11-20	2	5.9	6	14.6
21-30	6	17.7	7	17.1
31-40	6	17.7	4	9.8
41-50	3	8.8	5	12.2
50+	16	47.1	7	17.1
	Physician		Psychologist	
Average Age of Diagnosis	n	Percentage of Participants (%)	n	Percentage of Participants (%)
12-18 months	0	0	1	2.4
18-24 months	3	8.8	1	2.4
24-30 months	6	17.6	3	7.6
30-36 months	10	29.4	8	19.5
36-48 months	6	17.7	8	19.5
4-8 years	7	20.6	10	24.4
8+ years	2	5.9	10	24.4
Gender of Patient	Percentage (%)			
Male	64.0			
Female	36.0			

Table V: Routine Referrals to a Genetics Professional Following an ASD Diagnosis

	Physician	Psychologist
Yes / Sometimes	26 (76.5%)	18 (43.9%)
No	8 (23.5%)	23 (56.1%)

Table VI: How Patients are Referred to Genetics and Follow-Up

	Physician		Psychologist	
How do you refer patients?	n	Percentage of Participants (%)	n	Percentage of Participants (%)
Suggest patient sees a genetics provider; no specific resource given	0	0.0	2	2.2
Provide patient with general contact information	0	0.0	2	2.2
Provide specific contact information for a genetics professional	10	76.9	5	55.6
Contact specific genetics professional to set up appointment for patient	5	38.5	2	22.2
	Physician		Psychologist	
Do you follow-up with the patient?	n	Percentage of Participants (%)	n	Percentage of Participants (%)
Yes	11	84.6	1	11.1
No	2	15.4	8	88.9
How?				
By phone	2	18.2	1	100.0
In person	11	100.0	0	0
By email	2	18.2	1	100
By mail	1	9.1	0	0
	Physician		Psychologist	
Do you follow-up with the genetics professional?	n	Percentage of Participants (%)	n	Percentage of Participants (%)
Yes	9	69.2	2	22.2
No	4	30.8	7	77.8

Table VII: Reasons for Referring to a Genetics Professional

Physician (n=13)	To obtain genetic testing	To provide information on recurrence risks	To learn about the genetics of ASD	To help with medical management	To contribute to research	To rule in/out genetic conditions associated with ASD	To be consistent with organization guidelines
Strongly Agree	9	6	2	1	2	10	1
Agree	3	4	4	3	3	2	7
Neutral	1	3	6	5	6	1	5
Disagree	0	0	1	3	2	0	0
Strongly Disagree	0	0	0	1	0	0	0
Psychologist (n=9)							
Strongly Agree	8	3	2	2	1	6	2
Agree	0	4	5	4	4	3	2
Neutral	1	1	1	0	3	0	3
Disagree	0	0	1	2	1	0	1
Strongly Disagree	0	1	0	1	0	0	1

Table VIII: Reasons for Not Referring to a Genetics Professional

Physician (n=8)	Patient already has a diagnosi s	Patient has too many other appointme nts	My clinic does not have a Genetics department accessible	I do not know where to refer my patient	I do not know how to interpre t genetic testing	I order my own genetic testing	My patient cannot afford another appointmen t	I do not think ASD has a genetic compon ent	Genetic testing will not affect medical management
Strongly Agree	1	0	1	0	0	3	0	0	1
Agree	0	1	1	1	1	2	1	0	0
Neutral	0	0	1	0	0	1	3	0	1
Disagree	2	3	1	2	2	2	2	1	2
Strongly Disagree	5	4	4	5	5	0	2	7	4
Psychologist (n=23)									
Strongly Agree	2	1	13	7	6	0	2	0	4
Agree	10	6	3	8	4	0	4	0	3
Neutral	5	7	2	0	5	1	10	0	8
Disagree	4	5	2	3	3	0	2	5	5
Strongly Disagree	2	4	3	5	5	22	5	18	3

Table IX: Genetic Testing Ordered by Healthcare Providers

	Physician		Psychologist	
Did you order genetic testing on your patients with ASD?	n	Percentage of Participants (%)	n	Percentage of Participants (%)
Yes	16	47.1	2	4.9
No	8	23.5	33	80.5
Sometimes	10	29.4	6	14.6
What type of genetic testing did you order?	Physician		Psychologist	
	n	Percentage of Participants (%)	n	Percentage of Participants (%)
Karyotype	3	18.8	0	0.0
Fragile X Syndrome	16	100.0	2	100.0
Chromosome Microarray (CMA)	16	100.0	2	100.0
Next-Gen-Sequencing (NGS) Panel	2	12.5	0	0.0
Whole Exome Sequencing (WES)	5	31.3	1	50.0
Other	1	6.3	0	0.0

*Other: Whole Genome Sequencing (WGS), DNA methylation studies

Figures**Figure I: Does the Rate of Referral to Genetics Differ Between Physicians and Psychologists?**

Fisher's Exact Test for Count Data

```
data: dat
p-value = 0.005204
alternative hypothesis: true odds ratio is not equal to 1
95 percent confidence interval:
 1.380182 13.059472
sample estimates:
odds ratio
4.070203
```

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Appendix A

1) Do you diagnose children with autism spectrum disorder (ASD)?

- If no, survey will end
- If yes, continue

2) What type of healthcare professional are you? (check all that apply)

- Pediatrician
- Developmental Pediatrician
- Pediatric Neurologist
- Neurologist
- Child Psychiatrist
- Psychiatrist
- Geneticist
- Psychologist
- Other:

3) In what type of setting do you diagnose most of your patients with ASD?

- Private Practice
- Local Hospital
- School
- Other:

4) Approximately how many children do you diagnose with ASD per year?

- Less than 10
- 11-20
- 21-30
- 31-40
- 41-50
- 50+

5) Of your ASD patients, what is the average age of diagnosis?

- 6-12 months
- 12-18 months
- 18-24 months
- 24-30 months
- 30-36 months
- 36-48 months
- 4-8 years
- 8 years+

6) Of the children you diagnose with ASD, what percentage are female? What percentage are male?

- Sliding scale, with 100% female/male on either end and 50/50 in the middle

7) In general, to what healthcare professionals do you refer your patients to following a diagnosis of ASD? (check all that apply)

- Developmental Pediatrician
- Occupational Therapist
- Physical Therapist
- Speech Therapist
- Audiologist
- Cardiologist
- Neurologist
- Psychiatrist
- Geneticist/Genetic Counselor
- Psychologist
- Other:

8) How do you keep up to date with evolving practices in your field? (check all that apply)

- I attend courses for continuing education credit
- I attend conferences in my field of specialty
- I regularly check my specialties guidelines (e.g. American Academy of Pediatric's guidelines)
- I read research papers relating to my field of expertise
- I regularly consult with colleagues in related fields
- Other:

9) Do you routinely refer your patients with ASD to Genetics?

- Yes
 - Answer Q10-14; skip Q15-16
- No
 - Answer Q15; skip Q10-14, 16
- Sometimes
 - Answer Q16, skip Q10-15

10) Why do you refer to Genetics? (Likert scale)

- To obtain genetic testing
- To provide information on recurrence risks
- To learn about the genetics of ASD
- To help with medical management
- To contribute to research
- To rule in/out genetic conditions associated with ASD
- To be consistent with my organization's guidelines
- Other:

11) How do you refer your patients? (check all that apply)

- I suggest that my patient see Genetics but do not provide a specific resource/provider

- I provide my patient with general contact information for genetics providers such as the National Society of Genetic Counselors (NSGC.org) and/or hospitals with Genetics departments in the area
- I provide my patient with specific contact information for a genetics professional in the area
- I contact a specific genetics professional/department to set up the appointment for my patient
- Other:

12) Do you follow-up with your patient to ensure they attend their Genetics appointment?

- Yes
- No
 - Skip Q13

13) In general, how do you follow-up with your patient? (check all that apply)

- By phone
- In person
- By email
- By mail
- Other:

14) Do you follow-up with the Genetics provider?

- Yes
- No

15) Why do you not refer these patients to Genetics? (Likert scale)

- The patient already has a diagnosis; thus genetic testing will not be helpful
- The patient has so many appointments that I want to maximize the ones they need to go to
- My clinic does not have a Genetics department that is easily accessible
- I do not know of a Genetics professional to refer
- I do not know how to interpret the results of any genetic testing
- I order my own genetic testing
- My patient cannot afford another appointment
- I do not think ASD has a genetic component
- Genetic testing will not affect my patient's medical management
- Other:

16) Under what circumstances would you refer your patient to Genetics? (Likert scale)

- My patient has dysmorphic features
- My patient has other health problems in addition to ASD
- My patient has a family history of ASD
- The patient's family requested a referral
- My patient had abnormal laboratory testing / imaging studies
- Other:

17) Do you order genetic testing on your patients with ASD?

- Yes
 - Answer Q18-21; skip Q22-23
- No
 - Answer Q22; skip Q18-21, 23
- Sometimes
- Answer Q23; skip Q18-22

18) What type of genetic testing do you order? (check all that apply)

- Karyotype
- Fragile X Syndrome Testing
- Chromosomal Microarray (CMA)
- Next-Gen Sequencing (NGS) Panel
- Whole Exome Sequencing (WES)
- Other:

19) Why do you order these tests? (Likert scale)

- To obtain an etiology
- To provide information on recurrence risks
- To help with medical management
- To contribute to research
- To rule in/out genetic conditions associated with ASD
- To follow my organization's guidelines
- Other:

20) Please rate the following statement: (Likert scale)

- I feel comfortable interpreting genetic testing on my own

21) How likely are you to refer to a genetics professional after receiving a... (Likert scale)

- Positive result
- Negative result
- Variant of uncertain significance (VUS)

22) Why do you not order genetic testing? (Likert scale)

- I do not think genetic testing for ASD is useful for medical management
- I refer to Genetics for genetic testing
- I don't believe ASD has a genetic component
- My patient's insurance does not cover genetic testing
- I do not know how to order genetic testing
- I do not know what genetic testing is appropriate
- I do not feel comfortable interpreting genetic testing results
- Other:

23) Under what circumstances would you order genetic testing? (Likert scale)

- My patient has dysmorphic features
- My patient has other health problems in addition to ASD
- My patient has a family history of ASD
- My patient's family requested genetic testing
- My patient had abnormal laboratory testing / imaging studies
- Other:

24) Do you see your patient for follow-up after their diagnosis?

- Yes
 - Answer Q25-26
- No
 - Skip Q25-26

25) How many times a year do you follow-up with your patient?

- Once a year
- Twice a year
- Monthly
- Other:

26) Why?

27) What is your gender identity?

- Male
- Female
- Other:

28) What is your age?

29) How many years of experience do you have in your current specialty/specialties?

30) What state/province do you work in?

31) What type of area do you work in?

- Urban
- Suburban
- Rural