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Genetic testing and genetic counseling among medicaid-enrolled children with autism spectrum disorder in 2001 and 2007

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Abstract

The rise in the prevalence of autism spectrum disorder (ASD) has resulted in increased efforts to understand the causes of this complex set of disorders that emerge early in childhood. Although research in this area is underway and yielding useful, but complex information about ASD, guidelines for the use of genetic testing and counseling among children with ASD conflict. The purpose of this study was to determine the frequency of use of genetic testing and counseling before the widespread implementation of clinical chromosomal microarray (CMA) to establish a baseline for the use of both services and to investigate potential disparities in the use of both services among children with ASD. We found that about two-thirds of children with ASD received genetic testing or counseling and the use of both services is increasing with time, even in the pre-CMA era. Being female and having a comorbid intellectual disability diagnosis both increased the likelihood of receiving genetic testing and genetic counseling. Initial discrepancies in the use of both services based on race/ethnicity suggest that troubling disparities observed in other services delivered to children with ASD and other mental health disorders persist in genetic testing and counseling as well. These results should incentivize further investigation of the impact of genetic testing and counseling on children with ASD and their families, and should drive efforts to explore

and confront disparities in the delivery of these services, particularly with the advancing scientific research on this topic.

Introduction

Autism spectrum disorder (ASD) comprises a set of neurodevelopmental disorders that manifest as deficits in social communication and repetitive behaviors or insistence on sameness in childhood (American Psychiatric Association 2000). Since the 1980s it has been widely understood that ASD is highly heritable, (Chakrabarti and Fombonne 2001; Constantino et al. 2010; Freitag et al. 2010; Ozonoff et al. 2011) as evidenced from twin studies and studies of the broader autism phenotype (International Molecular Genetic Study of Autism Consortium (IMG-SAC) 2001; Gupta 2007; Miller 2010). ASD has also long been known to be associated with several rare genetic syndromes, including Angelman Syndrome, Fragile X, Rett Syndrome, Tuberous Sclerosis, and Smith-Lemli-Opitz Syndrome (Abrahams and Geschwind 2008). At the same time, more recent genomic studies underscore the complexity of genetic mechanisms in ASD, suggesting that perhaps thousands of genetic variants are contributing to ASD, many of these changes in chromosomal structure (copy number variants or CNVs). CNVs can be de novo or inherited. Incomplete penetrance and variable expressivity (e.g., the same CNV may manifest phenotypically as ASD in one family member and as schizophrenia in another) further complicate the interpretation. ASD is not always caused by a single mutation in a single gene. A “two hit” hypothesis (Girirajan et al. 2010; Leblond et al. 2012) and a hypothesis of oligogenic heterozygosity (Schaaf et al. 2011) have been proposed.

Given the known heritability of ASD and association with known genetic syndromes, genetic testing has been used for decades in clinical research assessments of individuals with ASD (Folstein and Rutter 1977). The ultimate goal of genetic testing is to identify heritable predisposition to a disease, which could be important to tested individuals or their relatives in reproductive decision-making or to identify genetic causes that will influence decisions about treatment strategies and the frequency and nature of ongoing clinical follow-up (Mendelsohn and Schaefer 2008). Families of children with ASD may benefit from genetic testing and subsequent genetic counseling because it can contribute to disease management, including identifying potential or existing comorbidities (Schaefer and Mendelsohn 2008a). On the other hand, genetic testing and counseling can be stressful (Muhle et al. 2004) interpretation of genetic testing for a disorder like ASD with complex and heterogeneous genetic mechanisms will be challenging, and genetic testing results in ASD, even when they clearly implicate a known genetic cause, at this point do not inform intervention choices (Muhle et al. 2004; Schaefer and Mendelsohn 2008b). Translation of genetic testing results to an individual may also present unique challenges since risk is based on group averages, and implications in specific areas, such as recurrence risk, are highly variable depending on genetic testing results.

Current clinical recommendations for genetic testing of individuals with ASD are somewhat conflicting. Schaefer and Mendelsohn (2008b) suggest that genetic testing is an essential component of “appropriate management” of the care of individuals with ASD. The

American Academy of Pediatrics (AAP) recommends inclusion of genetic testing as part of medical evaluations of children diagnosed with ASD based on risk factors, such as co-occurring intellectual disability, and other findings (Johnson et al. 2007). Specific tests, including cytogenetic analysis (karyotype), chromosomal microarray (CMA), and Fragile X testing, have been named as primary components of the genetic testing battery for individuals with ASD in the recommendations from the AAP and other professional organizations. In the most recent guidelines, including those published by the American College of Medical Genetics in 2010, CMA testing for copy number variants has replaced G-banded karyotype as a first-line test in the initial postnatal evaluation of individuals with ASD (Freitag et al. 2010; Manning et al. 2010; Miller 2010; Shen et al. 2010).

However, others have argued for more limited application of genetic testing. El-Fishawy and State (2010) and Caronna et al. (2008) call for genetic testing only for individuals suspected as having “syndromic” ASD, meaning those with dysmorphisms, neurocutaneous findings, significant cognitive impairment, abnormal neurological exam or seizures (Caronna et al. 2008; El-Fishawy and State 2010). Recommendations from the American Academy of Neurology and the Child Neurology Society suggest that genetic testing for children with ASD should be completed only if there is a comorbid intellectual disability or one cannot be ruled out, if there is a family history of Fragile X syndrome or intellectual disability, or if dysmorphic features are present (Filipek et al. 2000). Similarly, as noted above, the 2007 AAP guidelines for the identification and management of ASD recommend genetic testing when there is comorbid intellectual disability or global developmental delay or when suggested by dysmorphic features, family history, or comorbid medical conditions (Johnson et al. 2007). However, in the second edition of “Autism: Caring for Children with Autism Spectrum Disorders: A Resource Toolkit for Clinicians,” released in late 2012, the AAP recommends offering genetic testing, including CMA and Fragile X testing to all patients with ASD (American Academy of Pediatrics 2013). The AAP recommends genetic counseling for families of all children with ASD to help parents understand recurrence risk. When a specific genetic etiology is determined by genetic testing, the recurrence risk may be higher or lower than the risk in idiopathic ASD, and pre-natal diagnosis may be possible (Johnson et al. 2007).

Recent empirical investigations of CMA testing in individuals with ASD have focused on diagnostic yield: the proportion of tested children who have significant findings on CMA (Roesser 2011; McGrew et al. 2012). Heterogeneity in array resolution and coverage affects the comparability of results across different laboratories and no uniform standards exist to establish the clinical importance of CNVs (Miller et al. 2010a). CMAs are rapidly evolving and are improving the ability to interpret genetic testing findings (Cooper et al. 2011; Kaminsky et al. 2011). Emerging data suggest that some specific abnormal findings may have implications for medical treatment (Saam et al. 2008; Coulter et al. 2011) and that CMA testing may be cost effective (Regier et al. 2010). A recent study found that 35 % of all pathogenic CNVs identified in one large laboratory identified conditions for which specific clinical actions are warranted (Ellison et al. 2012).

The purpose of the present study was to examine the extent to which Medicaid-enrolled children diagnosed with ASD receive genetic testing and genetic counseling services relative

to other children. Given that recent estimates suggest that approximately 45 % of children diagnosed with ASD are enrolled in Medicaid (Mandell et al. 2010a) genetic testing and genetic counseling practices in this system have important policy and practice implications. We had two specific aims within this study. First, we investigated genetic testing and counseling among children with ASD prior to recommendations for genetic testing via the use of CMA for children with ASD to determine the baseline for genetic testing and counseling practices before the field-shifting change of CMA. To inform current discussions regarding genetic testing and genetic counseling for children with ASD and their families and changes in both services over time, we compared children who received genetic testing and genetic counseling in 2001 or 2007 (the most recent year for which Medicaid claims are available for research purposes) with those who did not receive either service. Second, we examined whether racial and ethnic disparities in delivery of services to children with ASD observed in other areas (Mandell et al. 2002, 2009, 2010b) occurred in the delivery of genetic testing and genetic counseling. Racial and ethnic disparities in service delivery have been observed for many other services (Mandell et al. 2002, 2009, 2010b; Mandell and Novak 2005; Mandell et al. 2007, 2009). In addition, racial and ethnic minorities are much less willing than Whites to participate in biomedical and specifically genetic research which may further exacerbate racial disparities (Hilton et al. 2010). It is unclear if these disparities translate into clinical genetic testing and counseling services but, if so, it would further stress the need for targeted, concerted efforts to engage traditionally underserved populations.

Methods

Data source

Child-level data from Medicaid eligibility and encounter files were extracted from the 2001 and 2007 Centers for Medicare and Medicaid Services (CMS) Medicaid Analytic Extract data files of all Medicaid claims from 49 states and the District of Columbia. Maine was excluded because it did not submit Medicaid claims to CMS in 2007.

Sample

The study sample included all children who were 8 years old or younger and had two outpatient or one inpatient Medicaid-reimbursed claim associated with an ASD diagnosis [International Classification of Diseases, Ninth Revision (ICD-9) code 299.XX] in 2001 or 2007 ($n = 77,601$).

Variables

The outcome of interest was delivery of a genetic testing or genetic counseling service during the study year (either 2001 or 2007). Genetic testing included 32 CPT and ICD-9 codes. A full listing of these codes can be found at: <http://www.med.upenn.edu/cmhpsr/resources.html>. Genetic counseling consisted of the following CPT codes: 99211-5, 96040 and 96150-5 for genetic counseling or a genetic counselor. Initially four different outcome variables were considered: genetic testing only, genetic testing with genetic counseling, genetic counseling only, and any genetic testing or genetic counseling. Because the genetic testing only group was relatively small ($n = 543$ when 2001 and 2007 data were combined),

genetic testing only and genetic testing with genetic counseling were combined into “genetic testing with or without genetic counseling.” Although parents or guardians of children with ASD may receive genetic counseling, only CPT codes associated with the child with an ASD were included since billing convention requires a focus on only the individuals who receive genetic testing or about when genetic counseling information is being communicated. Independent variables were sex, race/ethnicity, comorbid intellectual disability (ICD-9 codes 317.XX–319.XX), state of residence, and year (2007 vs. 2001).

Analysis

The proportions of children with ASD receiving genetic testing with or without genetic counseling, genetic counseling only, any genetic testing or genetic counseling, or no genetic testing or genetic counseling were estimated by year. Separate unadjusted and adjusted (with all covariates) logistic regression models were used to estimate the effect of each independent variable on the relative odds of each dependent variable compared to the reference group of no genetic testing or counseling. State was entered into the adjusted analyses as a fixed effect.

Results

As shown in Table 1, most children with ASD received genetic testing or genetic counseling in both 2001 (62.9 %) and 2007 (70.8 %). Genetic counseling without genetic testing was much more commonly delivered to children with ASD than genetic testing. Adjusted odds ratio estimates suggest between 38 and 68 percent increases in the odds of all genetic testing and counseling outcomes over this 6 year period. Effects were strongest for genetic testing with or without counseling than for counseling only. Girls were more likely to receive genetic testing with or without genetic counseling, genetic counseling only or any genetic testing or genetic counseling (Table 2), although only adjusted odds ratio estimates for genetic counseling only and any genetic testing or counseling had 95 % confidence intervals excluding a null effect.

The odds that Latino children received genetic testing with or without counseling were more than twice that of White children. Latino children were also more likely to receive genetic counseling only or any genetic testing or counseling than non-minority children. Odds ratio estimates for other race/ethnicity groups all included the null, but the pattern in effect estimates suggested that African-American children had lower odds of genetic testing or counseling while Asian children and children of other races/ethnicities were more likely to receive genetic testing or counseling than non-minority children. Children with diagnosis codes for comorbid intellectual disability had higher odds of all genetic testing and counseling outcomes, with the strongest effect seen for testing, than children without those codes; however, confidence intervals all included the null.

Discussion

Most children with ASD received genetic testing or counseling in 2001 and 2007, and the use of both services increased markedly over time, even before the issuance of recommendation that CMA testing become standard practice in the diagnostic workup for

children with ASD. The increased use of genetic testing among children with ASD who have a comorbid intellectual disability preliminarily suggests adherence to a common component of most genetic testing and counseling guidelines.

Disparities were revealed, however, in the delivery of genetic testing and genetic counseling across racial/ethnic groups. Findings suggest unequal access to and/or delivery of genetic testing and counseling for children with ASD of different races and ethnicities. Specifically, Hispanic children with ASD were more likely to receive genetic testing and genetic counseling. Racial/ethnic disparities in the delivery of other healthcare services have been previously documented in children ASD (Mandell et al. 2009) as well as in children with other disorders (Alegria et al. 2010; Aratani and Cooper 2012). These studies also suggest, as was observed here, that specifically African-American children are less likely to receive services than their non-minority counterparts. The drivers of these disparities are multifactorial (Mandell et al. 2009). Previous research on racial and ethnic differences in attitudes around genetic testing has found that individuals from Latino and African-American racial and ethnic groups express a preference for both prenatal and adult genetic testing (Singer et al. 2004) which may, in part, explain the increased rate of genetic testing and genetic counseling among Hispanic children with ASD. Genetic testing and counseling are services that can be sought or refused by caregivers of children with ASD and further research is needed to understand if or how cultural or racial differences in the rate of acceptance of these services occur or how they affect families with ASD.

Understanding the impact of genetic testing and genetic counseling on families living with ASD is an important, but understudied area. The findings here, which indicate that the use of these services was on the rise even prior to recent changes in recommendations around the use of genetic testing in ASD diagnosis and also document racial/ethnic disparities in receipt of these services, underscores the need to better understand how these commonly used services affect clinical care for children with ASD and influence family reproductive decision-making. Some research has found that families want to know about the genetics of ASD because they find it beneficial to understand the cause of their child's ASD (Miller et al. 2010b). The goal of risk communication should be to inform families, which is an important reminder of the mission for genetic testing and counseling services. Providing thorough and easy-to-understand information on recurrence risk to families affected by ASD, particularly diverse families, however, is complicated by the lack of complete information on the genetic causes of ASD (Edwards and Elwyn 2001). It's also unclear how genetic counseling is currently presented to families with a child or children with ASD and the extent to which these practices might vary.

While this study is the first to report on the frequency of genetic testing and counseling in ASD using a larger claims database, there were limitations to this approach. First, our sample was comprised only of publicly insured children. A significant portion of children with ASD have Medicaid coverage with or without private insurance coverage (Semansky et al. 2011), but children with ASD with coverage from both systems may have received genetic testing or counseling through private insurance. In addition, the analysis included claims in two calendar years and it is likely that subjects may have received testing or counseling outside these 1-year windows, though the assumption is that these differences

would not be differential across the groups being compared. Claims data also present other measurement issues. Any genetic counseling services delivered within general medical visits or follow-up visits without referral to a geneticist or genetic counselor were likely not separately coded and therefore would have been missed here and, finally, the prevalence of intellectual disability is lowest in administrative databases, suggesting potential underreporting (Maulik et al. 2011).

With continued advances in genetic testing and genetic counseling, the recommendations regarding the roles of genetic testing and counseling in the diagnostic and treatment processes for individuals with ASD and their families will likely continue to evolve and include identification of individuals with genetic susceptibility in addition to etiologic determination in affected individuals (Schaefer and Mendelsohn 2008a; El-Fishawy and State 2010; Miller 2010). Continued work to identify the genetic variants which cause ASD is the obvious “first step” toward the development of a test that may help identify individuals likely to develop or have ASD or other neurodevelopmental and psychiatric disorders (Miller 2010). A new wave of endeavors, including several large scale, federally and privately funded projects and corresponding initiatives to develop and use new technology, efforts to correlate genetic information with detailed phenotypic information, and the broad exploration of these issues across developmental and neurological disorders, are expected to elucidate the genetic contributions to ASD and other neurodevelopmental disorders. Similar work completed in overlapping samples of individuals such as those with intellectual disability or schizophrenia may aid progress in identifying the genetic profile of ASD (Freitag et al. 2010). Continued efforts to monitor developments in the genetics of ASD and how they are communicated both to professionals and to families, particularly those in underserved and underrepresented communities, is crucial to working toward optimizing family and clinician engagement in the identification and care of individuals with ASD. Specific efforts to address racial and ethnic disparities especially if they persist over time are warranted. Along these lines, researchers should continue to investigate and support the impact of genetic testing and counseling on clinicians and families.

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Number and percent of medicaid-enrolled children with ASD age 8 receiving genetic testing or counseling in 2001 and 2007 ($n = 77,601$)

Table 1

	<u>Testing (with or without counseling)</u>		<u>Counseling only</u>		<u>Any testing OR counseling</u>	
	<i>N</i>	%	<i>N</i>	%	<i>N</i>	%
2001 ($n = 23,892$)	1,239	5.2	13,781	57.7	15,020	62.9
2007 ($n = 53,709$)	3,826	7.1	34,197	63.7	38,023	70.8

Table 2

Results of logistic regression predicting genetic testing and/or counseling among medicaid-enrolled children with ASD, age 8 in 2001 and 2007

	Genetic testing with or without genetic counseling		Genetic counseling only		Any genetic testing or genetic counseling	
	Unadjusted OR (95 % CI)	Adjusted OR (95 % CI)	Unadjusted OR (95 % CI)	Adjusted OR (95 % CI)	Unadjusted OR (95 % CI)	Adjusted OR (95 % CI)
Female (ref = male)	1.04 (0.95–1.15)	1.06 (0.97–1.16)	1.04 (0.99–1.09)	1.05 (1.00–1.10)	1.04 (0.99–1.09)	1.05 (1.00–1.10)
Ethnicity (ref = white)						
Black	0.89 (0.55–1.43)	0.89 (0.56–1.42)	0.93 (0.62–1.38)	0.93 (0.63–1.38)	0.92 (0.62–1.38)	0.92 (0.62–1.38)
Asian	1.13 (0.73–1.75)	1.09 (0.69–1.71)	0.84 (0.57–1.24)	0.82 (0.55–1.22)	0.86 (0.58–1.28)	0.84 (0.56–1.26)
Latino	2.21 (1.31–3.74)	2.10 (1.25–3.52)	1.58 (1.01–2.47)	1.52 (0.97–2.39)	1.64 (1.04–2.57)	1.58 (1.00–2.48)
Other	1.16 (0.77–1.75)	1.16 (0.79–1.72)	1.20 (0.86–1.68)	1.21 (0.87–1.66)	1.19 (0.85–1.69)	1.20 (0.87–1.67)
Comorbidities						
Intellectual disability	1.19 (0.91–1.56)	1.25 (0.97–1.62)	0.99 (0.78–1.27)	1.02 (0.80–1.31)	1.01 (0.79–1.29)	1.04 (0.82–1.32)
2007 (ref = 2001)	1.75 (1.29–2.36)	1.68 (1.25–2.25)	1.40 (1.08–1.82)	1.38 (1.06–1.79)	1.43 (1.10–1.86)	1.40 (1.08–1.82)