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BRIEF REPORT

Brief Report: Pediatrician Perspectives Regarding Genetic Evaluations of Children with Autism Spectrum Disorder

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Abstract

Despite current guidelines, few children with autism spectrum disorder (ASD) receive genetic evaluations. We surveyed Utah pediatricians to characterize the knowledge, beliefs, current practices and perceived barriers of pediatricians regarding genetic evaluation of children with ASD. We found over half lacked knowledge of current guidelines and many held beliefs about genetic evaluation that did not align with guidelines. Barriers were lack of insurance coverage for genetic evaluation/testing and long wait times to see geneticists. Pediatricians with beliefs aligned with guidelines and those aware of the role of genetic counselors were more likely to adhere to guidelines. Efforts to educate pediatricians are needed along with system level solutions regarding availability of geneticists and reimbursement for genetic testing.

Keywords Autism spectrum disorder · Genetics · Chromosomal microarray · Primary care provider

Autism spectrum disorder (ASD) is a behavioral phenotype consisting of deficits in social communication skills, restrictive interests and repetitive behaviors (APA 2013). Advances in genomic technology are providing an increasing understanding of the underlying etiologies of ASD, including the identification of hundreds of genetic variants that increase the risk for isolated ASD as well as genetic syndromes in which ASD is a recognized component (Miles 2011). These discoveries have significantly improved the diagnostic yield of genetic testing for children with ASD. For example, the diagnostic yield of a chromosomal microarray (CMA), which detects chromosomal deletions and duplications is estimated to be between 8 and 21% (Schaefer et al. 2013). A comprehensive clinical genetic evaluation (including a family history, physical examination and appropriate genetic testing) has a reported diagnostic yield of 30–40% and is expected to rise with further advances in genetic technology (Schaefer 2016). Determining whether there is a genetic etiology for a child's ASD has several benefits: it empowers families with knowledge regarding the underlying cause, may lead to earlier identification and management of associated medical complications, and provides more accurate recurrence risk counseling (Schaefer et al. 2013). Based on increasing diagnostic yield and the potential benefits of an etiologic diagnosis, the American College of Medical Genetics (ACMG) recommends that "a genetic evaluation should be offered to every person with an ASD (or his or family)" (Schaefer et al. 2013). According to the American Academy of Pediatrics, the pediatrician plays an important role in the etiologic evaluation of a child with ASD which may entail ordering genetic tests or referring to a specialist, such as a geneticist, who directs the laboratory evaluation (AAP 2012). Regarding genetic testing for children with ASD, the American Academy of Pediatrics (AAP) recommends that CMA be "offer(ed) to all patients" and the ACMG designates it as a "first tier" genetic test (AAP 2012; Schaefer et al. 2013). Despite these recommendations, few families of children with ASD are referred for genetic evaluation and counseling or receive genetic testing (Cuccaro et al. 2014; Kiely et al. 2016).

The reasons why few children with ASD receive a genetic evaluation are unclear. However, studies focusing on the perspectives of parents of children with ASD have found that most parents have an interest in pursuing a genetic evaluation for their child and have a favorable view of genetic testing (Li et al. 2016). Additionally, most parents believe that there is a genetic component to their child's ASD and those whose children received genetic testing felt it was helpful (Selkirk et al. 2009). Parents of children with ASD whose

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children had received a genetics evaluation were more likely to have had the evaluation facilitated by a primary care provider (PCP) (Wydeven et al. 2012). Given that PCPs have an important role in initiating genetic evaluation for children with ASD, it is important to understand primary care pediatricians' knowledge and perceptions regarding genetic evaluation for children with ASD. This information can provide the basis for quality improvement efforts to increase the proportion of children with ASD who receive recommended genetic evaluation and counseling. The objectives of this study were to assess the knowledge, beliefs, practices and perceived barriers of primary care pediatricians in Utah regarding the genetic evaluation of children with ASD and to identify factors associated with pediatrician self-report of performing appropriate genetic evaluations in children with ASD. We also wished to describe pediatricians' experiences regarding exposure to marketing and advertising of genetic testing services for children with ASD and their perspectives on these services as this is a relatively new development in the genetic evaluation of children with ASD.

Methods

Participants

We recruited Utah primary care pediatricians (n = 295) from the most comprehensive list of primary care pediatricians available, maintained by the Utah Pediatric Partnership to Improve Healthcare Quality. We crosschecked this list with a separate one maintained by the Utah Chapter of the American Academy of Pediatrics in order to confirm correct contact information. While all prospective participants were identified within the list as primary care pediatricians, we excluded those who reported that they either did not consider themselves PCPs or that they did not care for children with ASD.

Measures

In the absence of a validated tool, we designed and pilot-tested a survey to assess pediatricians' perceptions regarding the genetic evaluation of children with ASD. The survey required approximately 10 min to complete and included 22 questions in six domains: (1) demographics, (2) current knowledge regarding genetic aspects of ASD, (3) current practice regarding the genetics evaluation of children with ASD, (4) perceived barriers in obtaining genetic evaluation for children with ASD, (5) beliefs regarding appropriate genetic referral and testing for children with ASD, and (6) exposure to marketing and advertising of genetic testing services for children with ASD. Survey questions ("Appendix") were created by a multidisciplinary research team with clinical interest/expertise in ASD: two general pediatricians (PC, PY), a genetic counselor (KD), a genetic counseling student (AR) and a geneticist (LB).

Study Procedures

The survey was administered electronically using REDCap™ software. Survey recruitment emails sent to prospective subjects described the purpose of the study, emphasized that participation in the study was voluntary, that responses would remain *anonymous*, and included a link to the online survey. No incentives were offered for study participation. Follow up recruitment emails were sent up to three times to non-responders. The Institutional Review Board of the University of Utah exempted the study from review.

Data Analyses

Descriptive statistics summarized pediatrician responses to questions within the survey. We then identified a cohort of pediatricians within the study pool who self-reported performing an *appropriate genetic evaluation* for children with ASD, defined, as "always" initiating a discussion regarding the genetic aspects of ASD with families of children with ASD *or* offering a genetics referral for *all* children with ASD at some point during their care. We based this definition on the practice guidelines from the AAP and ACMG reviewed earlier (Schaefer et al. 2013; AAP 2012). We then compared this cohort with the rest of study participants (*not* performing appropriate genetic evaluation) to identify pediatrician-level factors (independent variables) associated with performing an appropriate genetic evaluation of children with ASD (dependent variable) using Chi square in univariate analysis. Pediatrician level factors (independent variables) for these analyses included: practice location (urban versus suburban, rural/frontier), number of patients with ASD cared for (1–20, > 20), years since completing residency (≤ 10, ≥ 11), participation in ASD-related educational activities with Maintenance of Certification (MOC) credit (y/n), having a clinical interest in the care of children with neurodevelopmental disabilities (y/n), having knowledge of clinical guidelines regarding genetic evaluation of *all* children with ASD (y/n), having a belief that all children with ASD be offered either genetic referral or testing (y/n), perception of few barriers to obtaining genetic evaluation for children with ASD (< 2 out of a possible 7), having knowledge of the roles of a genetic counselor (y/n), having seen ASD genetic testing advertisements (y/n) or having been marketed to by a representative of a commercial ASD genetic testing company within the past year (y/n). We then identified associations between the independent variables (pediatrician-level factors) and the dependent variable (appropriate genetic evaluation) using multivariable logistic regression in order

to control for potential confounding variables and to report adjusted odds ratios. We performed analyses with STATA 14.0 (StataCorp, College Station, Tex).

Results

Of the 295 survey invitations sent to pediatricians, 119 (40%) submitted a completed survey. Among these, 108 met inclusion criteria (considered themselves PCPs and reported caring for children with ASD) and were analyzed. Table 1 shows the characteristics of the pediatricians within the study sample. Nearly 60% of pediatricians reported caring for between 11 and 30 patients with ASD. Nearly all practiced in urban (30%) or suburban (65%) locations, and almost half had been in practice more than 15 years. Almost all (92%) reported participation in educational activities related to ASD, and 27% had received MOC credit for an ASD-related educational activity. Nearly two-thirds reported having a clinical interest in caring for children with neurodevelopmental disabilities.

Table 1 Characteristics of study participants

Demographic	Total response	Response rate n (%)
Number of children with ASD under PCP care*	**108**	
1–10		25 (23%)
11–20		38 (35%)
21–30		26 (24%)
> 30		9 (8%)
Not sure		10 (9%)
Primary practice location	**108**	
Urban (city)		32 (30%)
Suburban (residential district)		70 (65%)
Rural or frontier (countryside)		6 (6%)
Years since PCP finished residency	**108**	
Less than 5 years		20 (19%)
5–10 years		17 (16%)
11–15 years		18 (17%)
More than 15 years		53 (49%)
Educational activities regarding ASD PCPs have participated in within the last 2 years (select all that apply)	**108**	
Workshop(s) or learning collaborative for Maintenance of Certification credit		29 (27%)
Grand rounds or other didactic presentations		58 (54%)
Reading of journal articles		82 (76%)
No training		9 (8%)
Other		4 (4%)
The care of children with neurodevelopmental disabilities such as ADHD, ASD, and Intellectual Disability is the PCP's clinical interest	**108**	
Yes		68 (63%)
No		40 (37%)

Bold values indicate statistical significance at $p < 0.05$

Knowledge

With regards to pediatricians' *knowledge* of the genetic aspects of ASD, 44% correctly identified the diagnostic yield of CMA (Schaefer et al. 2013), 47% correctly identified the ASD recurrence risk for younger siblings (Ozonoff et al. 2011) and 55% had knowledge of clinical guidelines regarding genetic evaluation of *all* children with ASD (Schaefer et al. 2013). Fourteen percent of pediatricians correctly answered all three knowledge questions.

Beliefs

Figure 1 shows pediatricians' *beliefs* with regard to genetic evaluation (referral and testing) of children with ASD. Approximately one-quarter of respondents believed that *all* children with ASD should be offered genetic referral (21%) and testing (24%). The remainder either believed that *some* should be offered referral (49%) and testing (45%) or were undecided (30% and 31% respectively).

Practice

Regarding pediatricians' *practice* in the care of children with ASD (Fig. 2), 9% reported consistently initiating a discussion regarding the genetics of ASD, while 24% never initiated this discussion. Eighteen percent reported offering a genetics referral to all families of children with ASD, while 60% reported selectively referring to genetics when indicated by physical exam or review of the history. A majority of pediatricians reported that they either never ordered genetic testing (34%) or did so only after a recommendation by a specialist (36%). A small minority (12%) would order testing upon a family's request (Fig. 3). Among those who had ordered genetic tests, 56% typically obtained CMA, 36% obtained DNA for Fragile X Syndrome and 12% obtained a karyotype (data not shown). A majority of pediatricians estimated that fewer than 25% of their patients with ASD had been referred to genetics or received genetic testing or (Fig. 4).

Barriers

When asked to identify any of seven potential barriers in obtaining specialist consultation for a *genetic evaluation* for

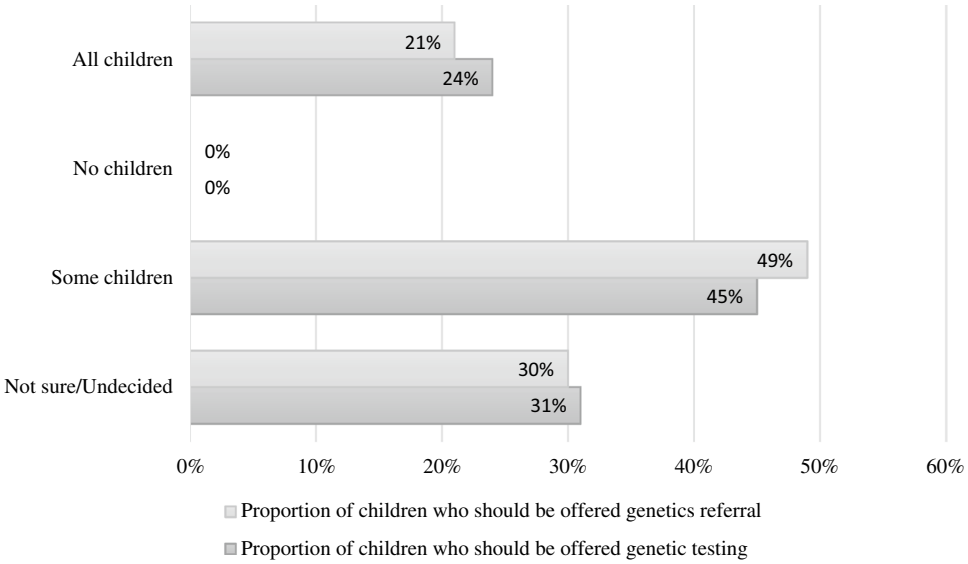

Fig. 1 Beliefs of pediatricians regarding the proportion of children with ASD who should be offered genetics referral and genetic testing (n = 108)

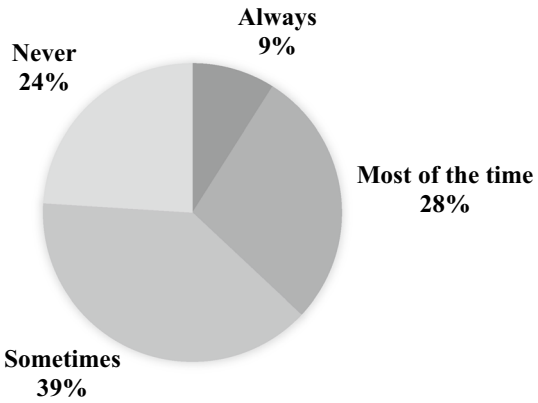

Fig. 2 Proportion of pediatricians who initiate a discussion regarding genetic aspects of ASD with families of children with ASD (n = 108)

children with ASD, 95% of pediatricians reported at least one (Fig. 5). Common barriers cited were long wait times to see a geneticist (74%), lack of insurance coverage (55%), and not knowing which children with ASD to refer (38%) Common barriers to obtaining *genetic testing* in children with ASD were lack of insurance coverage (71%), prohibitive cost of testing (55%), lack of confidence in ordering the correct test (49%), difficulty obtaining authorization for testing from insurers (47%), difficulty in interpreting the test results (40%) and lack of confidence in counseling families about the health implications of genetic testing results for parents and other family members (36%) (Fig. 6).

When queried about the role of genetic counselors in ASD, the majority of pediatricians correctly identified that these professionals counsel families about genetic testing (72%) and recurrence risk (78%); however, a minority (38%) viewed genetic counselors as being available to counsel PCPs about genetic testing and interpretation. Overall, 34% of pediatricians reported that genetic counselors had played an important role in the interpretation of genetic testing for patients with ASD, while 18% reported no experience with a genetic counselor in this area (data not shown).

We also assessed pediatricians' exposure to marketing and advertising of genetic testing for children with ASD. When asked how often representatives of commercial genetic testing companies marketed their products or services in person, 37% reported that they had been approached in the last year and 30% had been exposed to advertisements for genetic tests for children with ASD. Some 26% had utilized ASD-related products or services of the genetic testing companies and among those, 46% were satisfied with the services, 18% were dissatisfied, and 36% were undecided (data not shown).

Table 2 summarizes the assessment of pediatrician factors associated with performing an appropriate genetic evaluation (self-report of "always" initiating a discussion regarding the genetic aspects of ASD with families of children with ASD *or* report of offering a genetics referral for *all* children with ASD at some point during their care). In univariate analysis, pediatricians whose knowledge and beliefs aligned with clinical guidelines regarding genetic evaluation of *all* children with ASD and those who correctly identified all of the roles of a genetic counselor were more likely to report performing appropriate genetic evaluations of children with ASD within their practice. Two of these factors (*belief* that all children with ASD receive genetics evaluation and awareness of all of the roles of a genetic counselor) remained significant in multivariate analysis.

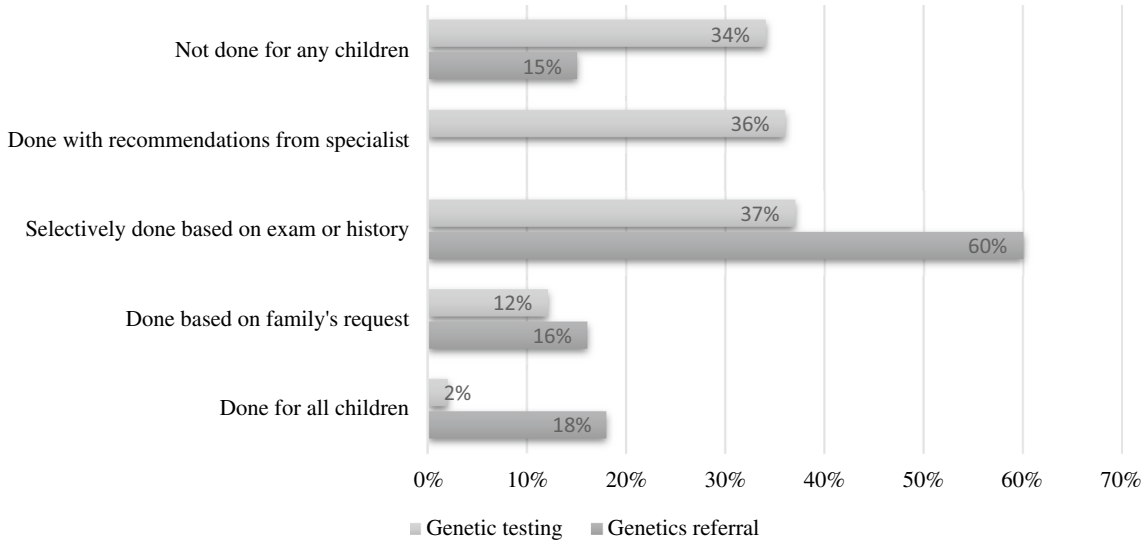

Fig. 3 Pediatricians' practices regarding genetics referrals and genetic testing (n = 108) for children with ASD

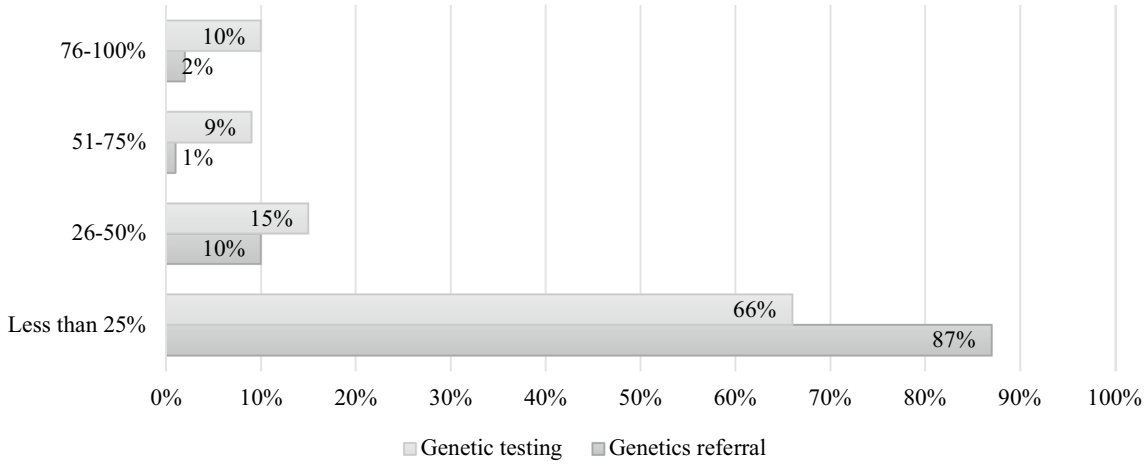

Fig. 4 The proportion of children with ASD in pediatricians' care who receive genetic referrals (n = 91) and genetic testing (n = 105)

Discussion

Although our study should be considered preliminary given its scope (limited to pediatricians within Utah), this study is the first to evaluate pediatricians' knowledge, beliefs, and practices regarding genetic evaluation of children with ASD, as well as perceived system level barriers to access and use of genetic services. We found that there were significant gaps in pediatricians' *knowledge* of clinical guidelines recommending genetic evaluation of all children with ASD. In addition, *beliefs* of pediatricians within our study often did not align with these guidelines. Finally, pediatricians report significant *barriers* to the access and use of genetic services. Taken together these provide insight into why pediatricians reported poor adherence to implementing clinical guidelines into their practice and suggest potential solutions. Our findings align with one previous study of community and specialty pediatricians which found variability in physician knowledge and underutilization of genetic testing in children with developmental delay/intellectual disability and ASD (Peabody et al. 2015). A previous survey of parents of children with ASD showed that they rarely access genetic counseling, referral, or testing for their children, despite a desire for these services (Cuccaro et al. 2014). In our study only 9% of pediatricians surveyed consistently initiate discussions with families of children with ASD regarding the genetic aspects of ASD, and only 18% offer genetics referral to all patients with ASD, which suggests that focusing

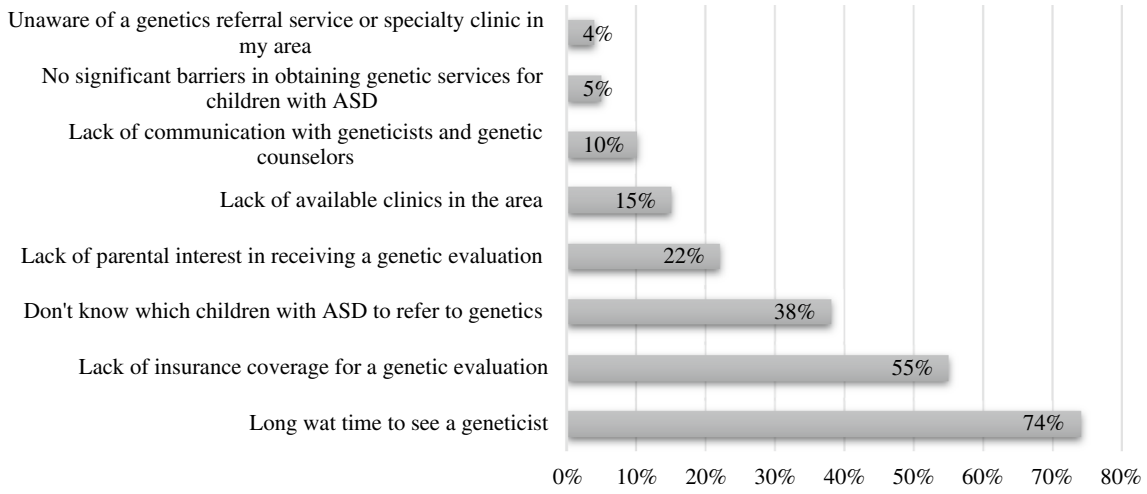

Fig. 5 Pediatrician identified barriers in obtaining genetic evaluation for children with ASD (n = 108)

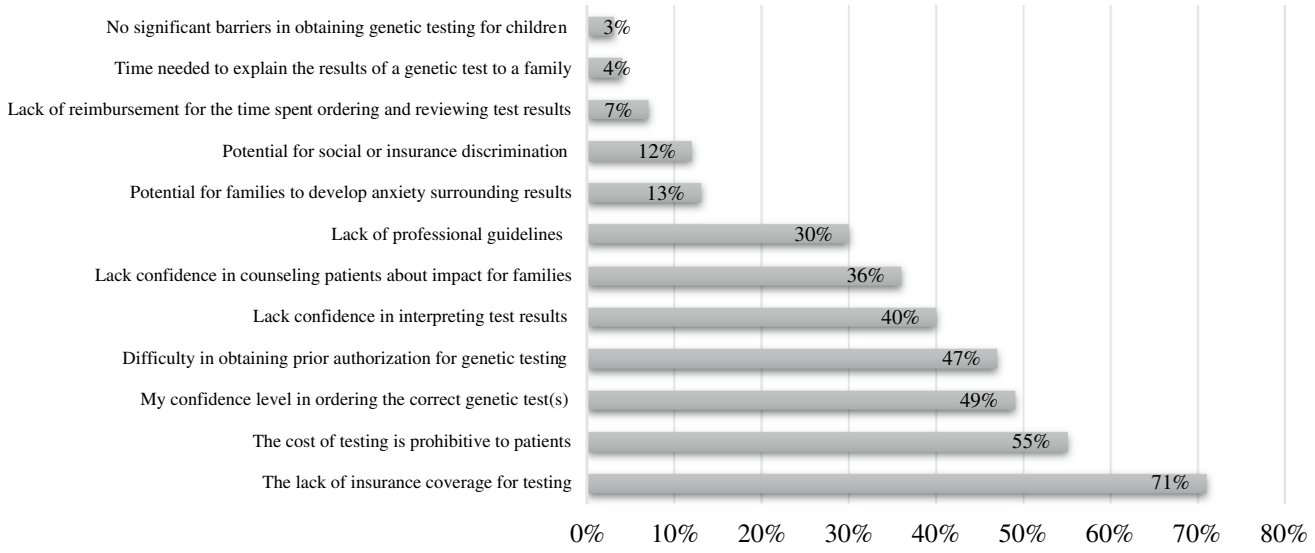

Fig. 6 Pediatrician identified barriers in obtaining genetic testing for children with ASD (n = 108)

interventions on primary care pediatricians may increase the proportion of children with ASD who receive appropriate genetic evaluations.

Our findings need to be interpreted within the context of several limitations. First, our study population of pediatricians from Utah may not be representative of all pediatricians with regards to knowledge, beliefs, practices and barriers faced. For example, the wait time for a genetic evaluation in Utah may differ from wait times in other states. Second, the low-response rate may have enriched the final study group with pediatricians who have an interest in neurodevelopmental disabilities. However, if that is the case we would expect such a cohort to have greater knowledge of, and adherence to clinical guidelines around genetic evaluation of children with ASD, in which case the findings might actually underestimate gaps in knowledge and practice of the larger population of pediatricians. Lastly, we were not able to verify if survey responses accurately reflected the true clinical practice of the participants. Future studies using administrative data from healthcare systems will provide a clearer picture of the nature and extent to which children with ASD receive appropriate genetic evaluations. Despite these limitations, our study indicates that efforts to increase the proportion of children with ASD who undergo genetic evaluation will need to extend beyond simply increasing knowledge of guidelines and will need to consider pediatricians' beliefs

Table 2 Pediatrician factors associated with self-report of appropriate genetic evaluation of children with ASD, univariate and multivariate analysis

Pediatrician factor		Univariate analysis			Multivariate analysis	
		Appropriate genetic evaluation n (%)	Not appropriate genetic evaluation n (%)	*p*	Adjusted OR (95% CI)	*p*
Practice location	Urban (n = 32)	7 (22)	25 (78)	0.924	1.22 (0.26–5.80)	0.797
	Rural/suburban (n = 76)	16 (21)	60 (79.0)			
Number of patients with ASD (n = 98)	1–20 (n = 63)	10 (16)	53 (68)	0.135	2.56 (0.62–10.65)	0.195
	> 20 (n = 35)	10 (29)	25 (71)			
Years since residency	< 10 years (n = 37)	6 (16)	31 (84)	0.352	0.68 (0.16–2.91)	0.599
	≥ 11 years (n = 71)	17 (24)	54 (76)			
ASD educational activity with MOC credit	Yes (n = 29)	8(28)	21(72)	0.333	1.87 (0.45–7.81)	0.390
	No (n = 79)	15 (19)	64 (81)			
Clinical interest in care of children with developmental disabilities	Yes (n = 68)	16 (24)	52(76)	0.460	0.91 (0.25–3.38)	0.892
	No (n = 40)	7 (18)	33 (82)			
Knowledge of clinical guideline	Yes (n = 59)	18 (31)	41 (69)	**0.010**	1.53 (0.39–6.06)	0.158
	No (n = 49)	5 (10)	44 (90)			
Belief to offer all children with ASD referral or testing	Yes (n = 31)	16 (52)	15 (48)	**< 0.001**	10.19 (2.66–39.06)	**0.001**
	No/undecided (n = 77)	7 (9)	70 (91)			
Barriers to obtaining genetic evaluation	< 2 of 7 (n = 30)	4 (13)	26 (86)	0.210	0.52 (0.10–2.66)	0.432
	≥ 2 of 7 (n = 78)	19 (24)	59 (76)			
Knowledge of all roles of a genetic counselor	Yes (n = 24)	9 (37)	15 (63)	**0.028**	5.54 (1.24–24.69)	**0.025**
	No (n = 84)	14 (17)	70 (83)			
Saw ASD genetic testing advertising	Yes (n = 32)	9 (28)	23 (72)	0.261	0.96 (0.20–4.68)	0.965
	No (n = 76)	14 (18)	62 (82)			
Marketed to by ASD genetic testing company	Yes (n = 40)	10 (25)	30 (75)	0.471	0.55 (0.11–2.88)	0.482
	No (n = 68)	13 (19)	55 (81)			

Bold values indicate statistical significance at $p < 0.05$

n = 108 unless indicated

as well as address system level barriers to obtaining genetic evaluation.

It is not entirely clear why over half of pediatricians in this study who had knowledge of clinical guidelines calling for genetic evaluation of all children with ASD did not believe that all children with ASD should be offered genetic evaluation. One possibility is that pediatricians, while aware of guidelines, may have reservations regarding the recommendation of universal genetic evaluation and testing for children with ASD. For example, one recent study indicated that clinicians tend to refer children with ASD to genetics based on the presence of distinctive physical features, epilepsy, family history of ASD and severity of developmental delay rather than taking a universal approach (Barton et al. 2017). Other studies have shown that children with ASD and co-morbid intellectual disability (ID) were more likely to receive genetic testing and counseling compared with those without ID, supporting the notion that referrals and testing are likely skewed towards the more severely affected children with ASD (Shea et al. 2014; Tchaconas and Adesman 2017; Kiely et al. 2016). These studies seem to indicate that providers may reserve genetic evaluation and testing for children in whom they believe have a higher probability of a positive finding. Previous studies further suggest that the diagnostic yield of genetic testing could indeed be higher among children with co-morbid ID or dysmorphic physical features, though another study found no association between CMA result and co-morbid features. Additionally, some providers perceive that positive results from genetic testing in children with ASD often do not result in changes in clinical management, leading them to question whether the benefits of testing justify the cost (Barton et al. 2017). In the case of CMA testing in children with ASD, however, a recent study found that that almost 30% of patients with positive results led to changes in clinical management including making medical referrals, ordering specific diagnostic imaging or ordering laboratory testing (Coulter et al. 2011).

A previous nationally representative study of children with ASD found that only 34% received genetic testing, concluding that more research is needed to identify barriers to the use of genetic testing in this population (Kiely et al. 2016). The pediatricians in our study identified numerous barriers to obtaining testing for children with ASD that should be addressed in order to increase the number of children who receive appropriate genetic evaluations. The most prevalent reported barriers included the long wait times to see a geneticist, lack of insurance coverage for genetic evaluation and testing and lack of confidence in knowing which genetic test(s) to order. The barriers we have identified in our study are similar to barriers identified by pediatricians when considering genetic evaluation and testing in other patients, regardless of ASD status (Saul et al. 2017). Long wait times for genetic consultations pose a delay and are frustrating for families; however, it is unclear why it would lead to not pursuing a genetic evaluation at all. In a previous study, 77% of parents whose children with ASD who saw a geneticist also obtained genetic testing. This may indicate that insurance coverage and cost of testing is less of a barrier than pediatricians perceive, but perhaps the barrier lies more in the process required to obtain these services (e.g., preauthorization).

Our findings suggest that genetic counselors might play an important role in facilitating genetic evaluations for children with ASD. Through pedigree interpretation and genetic variant analysis, genetic counselors can assist families and PCPs in interpreting genetic testing for the child with ASD and identify the recurrence risk for subsequent children. Pediatricians in our study who fully understood the role of genetic counselors were more likely to report obtaining appropriate genetic evaluations. Pediatricians reported lack of confidence in knowing which children with ASD to refer, which should receive testing, which tests to order, how to interpret the testing and how to explain results to families. These are all areas in which genetic counselors can effectively collaborate with pediatricians. With a shortage of medical geneticists, genetic counselors can also assist in triaging which patients with ASD need consultation with geneticists and which patients could be tested prior to a formal referral. Although we could find no examples in the literature evaluating this approach, it is one that should be considered as a means of facilitating timely genetic evaluations or ones that otherwise might not be accomplished.

Commercial genetic companies, whose advertising and marketing reached about one-third of our cohort and whose testing was used by about one-fourth, are a departure from the norm in that they offer genetic testing that bypasses the traditional evaluation done by a geneticist. These companies typically utilize genetic counselors rather than geneticists to review the genetic tests ordered and the results of testing with families and/or physicians and further study of this method is needed to ensure the appropriateness of the testing that is performed. About half of pediatricians in our study who utilized these services reported satisfaction, suggesting that commercial genetic companies may aid in completion of genetic testing bypassing perceived barriers including long wait times to see a geneticist.

Conclusions

In this survey of primary care pediatricians, we have identified gaps in knowledge, beliefs that often do not align with clinical guidelines, and barriers in obtaining appropriate genetic evaluation of children with ASD. Quality improvement efforts to increase genetic evaluation and testing of children with ASD should focus on provider education that includes data supporting the rationale for universal testing in order to address beliefs about the need for such testing, and address system level barriers that may in part be overcome by greater reliance on genetic counselors.

Author Contributions All authors contributed to the design of the study and were involved in creating the survey and writing the manuscript. AR and PSC collected the data. PSC analyzed the data.

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Compliance with Ethical Standards

Conflict of interest The authors declare that they have no conflicts of interest.

Ethical Approval All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

Informed Consent Informed consent was obtained from all individual participants in the study.

Appendix: Pediatrician survey

The following questions are about your practice:

1. Do you consider yourself a primary care pediatrician?

   - ☐ Yes
   - ☐ No, If no then EXIT

2. Do you provide primary care for children with ASD?
   - ☐ Yes
   - ☐ No, If no then EXIT

3. Approximately how many patients with ASD are under your care?
   - ☐ 1 to 10
   - ☐ 11 to 20
   - ☐ 21 to 30
   - ☐ > 30
   - ☐ Not sure

4. Please indicate your primary practice location by checking one of the options below:

   - ☐ Urban (city)
   - ☐ Suburban (residential districts)
   - ☐ Rural or frontier (countryside)
   - ☐ Other (please specify): _____

5. How many years since you finished residency training?

   - ☐ Less than 5 years
   - ☐ 5 to 10 years
   - ☐ 11 to 15 years
   - ☐ More than 15 years

6. Have you participated in any of the following educational activities regarding ASD within the last 2 years (Please indicate all that apply.)?

   - ☐ Workshop(s) or learning collaborative for Maintenance of Certification credit
   - ☐ Grand rounds or other didactic presentations
   - ☐ Reading of journal articles
   - ☐ No training
   - ☐ Other (please indicate): _____

7. Do you consider the care of children with neurodevelopmental disabilities such as ADHD, ASD, and intellectual disability one of your clinical interests?

- ☐ Yes
- ☐ No

The following questions address your current knowledge regarding the genetic aspects of ASD:

8. What is risk of ASD in a child who has an older sibling with ASD?

   - ☐ <1%
   - ☐ 1 to 10%
   - ☐ 11 to 25%
   - ☐ 26 to 50%
   - ☐ 51 to 75%
   - ☐ 76 to 100%

9. A chromosomal microarray (CMA) is a genetic test that aids in the identification of a specific etiologic diagnosis in children with ASD. What is the rate of success of CMA in identifying a specific etiologic diagnosis in children with ASD?

   - ☐ <1%
   - ☐ 1 to 5%
   - ☐ 6 to 20%
   - ☐ 21 to 50%
   - ☐ >50%

10. According to current clinical guidelines, a genetics evaluation should be offered to all children with ASD.

    - ☐ True
    - ☐ False

The following questions address your current practice regarding the care of children with ASD:

11. I initiate a discussion regarding genetic aspects of ASD with families of children with ASD for whom I am the primary care provider:

    - ☐ Always
    - ☐ Most of the time
    - ☐ Sometimes
    - ☐ Never

12. My practice for ***referring*** children with ASD for a ***genetics consult*** (a visit with a geneticist/genetic counselor) is described by the following (Please indicate all that apply):

    - ☐ I offer a referral to **all** children with ASD at some point during their care

- ☐ I refer upon a family's request
- ☐ I selectively refer when indicated on my exam or review of the child's history
- ☐ I do not refer any children with ASD (**If this response selected, skip to question 14**)
- ☐ **Other** (please specify) _____

13. What percentage of families of children with ASD in your care have you offered a referral to a geneticist/genetic counselor?

- ☐ Less than 25%
- ☐ 26 to 50%
- ☐ 51 to 75%
- ☐ 76 to 100%

14. My practice for ordering ***genetic testing*** for children with ASD is described by the following (Please indicate all that apply):

- ☐ I order genetic testing for all children with ASD.
- ☐ I order genetic testing upon a family's request.
- ☐ I order genetic testing when indicated on my exam or review of the child's history.
- ☐ I order genetic testing based on recommendations from a specialist (geneticist, genetic counselor, neurologist or developmental pediatrician)
- ☐ I do not order genetic testing for children with ASD. (**If yes, skip to question 17)**
- ☐ **Other** (please specify) _____

15. The genetic test(s) that I typically order for children with ASD include (Please indicate all that apply):

- ☐ Karyotype
- ☐ Chromosomal microarray (CMA)
- ☐ DNA for Fragile X syndrome
- ☐ **Other** (please specify) _____

16. What percentage of children with ASD in your care have undergone ***genetic testing***?

- ☐ Less than 25%
- ☐ 26 to 50%
- ☐ 51 to 75%
- ☐ 76 to 100%

The following questions address possible <u>barriers</u> you perceive in obtaining genetic services for children with ASD:

17. Please identify any of the following factors that are significant barriers in obtaining a ***genetic evaluation*** of children with ASD (Please indicate all that apply.).

- ☐ Not applicable – I do not refer children with ASD for a genetic evaluation.

- ☐ The long wait time to see a geneticist.
- ☐ The lack of parental interest in receiving a genetic evaluation
- ☐ The lack of available genetics clinics in the area
- ☐ I am unaware of a genetics referral service or genetics specialty clinic in my area
- ☐ I do not know which children with ASD are appropriate to refer to genetics
- ☐ The lack of communication with geneticists and genetic counselors.
- ☐ The lack of insurance coverage for a genetic evaluation
- ☐ I don't face any significant barriers in obtaining genetic services for children with ASD
- ☐ **Other** (please specify) _____

18. Please identify any of the following factors that pose a significant barrier in ordering ***genetic testing*** for children with ASD (Please indicate all that apply.).

- ☐ Not applicable – I do not offer children with ASD genetic testing.
- ☐ The lack of insurance coverage for such testing
- ☐ My confidence level in counseling patients about health impact for extended family members and for future pregnancies
- ☐ My confidence level in ordering the correct genetic test(s)
- ☐ My confidence level in interpreting genetic test results
- ☐ The lack of available professional guidelines concerning the ordering of genetic testing
- ☐ The potential for families to develop unwanted anxiety surrounding the results of the genetic test
- ☐ The potential for social or insurance discrimination as a result of a positive genetic test.

- ☐ The amount of time needed to explain the results of a genetic test to a patient
- ☐ The lack of reimbursement for the time spent ordering genetic tests or counseling patients on the results of genetic testing.
- ☐ The level of difficulty in obtaining prior authorization for genetic testing.
- ☐ The cost of testing is prohibitive to patients.
- ☐ I don't face any significant barriers in obtaining genetics testing for children with ASD
- ☐ **Other** (please specify) _____

The following questions address your beliefs about genetic referral and testing for children with ASD:

19. A **genetics referral** should be offered to which proportion of children with ASD?

- ☐ All children with ASD
- ☐ No children with ASD
- ☐ Some children with ASD
- ☐ Not sure/undecided

20. **Genetic testing** should be offered for which children with ASD?

- ☐ All children with ASD
- ☐ No children with ASD
- ☐ Some children with ASD
- ☐ Not sure/undecided

21. In my experience, genetic counselors have played an important role in the interpretation of genetic testing for patients with ASD.

- ☐ Yes
- ☐ No
- ☐ I have not had experience with a genetic counselor in this area.
- ☐ Not sure/undecided

22. My perception of the role of a genetic counselor in the care of children with ASD is… (Please indicate all that apply):

- ☐ Someone who orders genetic testing for patients
- ☐ Someone who counsels patients about genetic testing
- ☐ Someone who counsels patients about recurrence risk

- ☐ Someone who counsels primary care physicians about genetic testing and test interpretation
- ☐ I am not familiar with the role of genetic counselors in the care of children with ASD

The following questions address your exposure to marketing and advertising of genetic testing for children with ASD:

23. How often have representatives of commercial genetic testing companies marketed their ASD products or services to you **in person**?

- ☐ Once in the last year
- ☐ Multiple times over the last year
- ☐ I have not been approached in the last year

24. How often have you seen advertisements from commercial genetic testing companies regarding ASD?

- ☐ Once in the last year
- ☐ Multiple times over the last year
- ☐ Not at all in the last year

25. How often have you utilized the products or services of commercial genetic testing companies for a child with ASD?

- ☐ Once in the last year
- ☐ Multiple times over the last year
- ☐ I have not utilized products or services in the last year

26. How satisfied have you been with the genetic testing products or services you have received through the commercial companies you have been exposed to through advertising or representatives?

- ☐ I felt satisfied with the products or services
- ☐ I did NOT feel satisfied with the products or services
- ☐ Unsure/undecided
- ☐ I have not used genetic testing products or services through advertisement or through representatives

References

AAP. (2012). *Caring for children with autism spectrum disorders: A resourse toolkit for clinitians* (2nd ed.). Elk Grove Village: American Academy of Pediatrics.

APA. (2013). *Diagnostic and statistical manual of mental disorders: DSM-5* (5th ed.). Washington, DC: APA.

Barton, K. S., Tabor, H. K., Starks, H., Garrison, N. A., Laurino, M., & Burke, W. (2017). Pathways from autism spectrum disorder diagnosis to genetic testing. *Genetics in Medicine*. https://doi.org/10.1038/gim.2017.166.

Coulter, M., Miller, D. T., Harris, D. J., et al. (2011). Chromosomal microarray testing influences medical management. *Genetics in Medicine, 13*(9), 770–776.

Cuccaro, M. L., Czape, K., Alessandri, M., Lee, J., Deppen, A. R., Bendik, E., et al. (2014). Genetic testing and corresponding services among individuals with autism spectrum disorder (ASD). *American Journal of Medical Genetics Part A, 164A*(10), 2592–2600. https://doi.org/10.1002/ajmg.a.36698.

Kiely, B., Vettam, S., & Adesman, A. (2016). Utilization of genetic testing among children with developmental disabilities in the United States. *The Application of Clinical Genetics, 9*, 93–100. https://doi.org/10.2147/TACG.S103975.

Li, M., Amuta, A., Xu, L., Dhar, S. U., Talwar, D., Jung, E., et al. (2016). Autism genetic testing information needs among parents of affected children: A qualitative study. *Patient Education and Counseling*. https://doi.org/10.1016/j.pec.2015.12.023.

Miles, J. H. (2011). Autism spectrum disorders—a genetics review. *Genetics in Medicine, 13*(4), 278–294. https://doi.org/10.1097/GIM.0b013e3181ff67ba.

Ozonoff, S., Young, G. S., Carter, A., Messinger, D., Yirmiya, N., Zwaigenbaum, L., et al. (2011). Recurrence risk for autism spectrum disorders: A baby siblings research consortium study. *Pediatrics, 128*(3), e488–e495, https://doi.org/10.1542/peds.2010-2825..

Peabody, J., DeMaria, L., Tamandong-LaChica, D., Florentino, J., Acelajado, M. C., & Burgon, T. (2015). Low rates of genetic testing in children with developmental delays, intellectual disability, and autism spectrum disorders. *Global Pediatric Health*. https://doi.org/10.1177/2333794X15623717.

Saul, R. A., Trotter, T., Sease, K., & Tarini, B. (2017). Survey of family history taking and genetic testing in pediatric practice. *The Journal of Community Genetics, 8*(2), 109–115. https://doi.org/10.1007/s12687-016-0291-3.

Schaefer, G. B. (2016). Clinical genetic aspects of ASD spectrum disorders. *International Journal of Molecular Sciences*. https://doi.org/10.3390/ijms17020180.

Schaefer, G. B., Mendelsohn, N. J., Professional, P., & Guidelines, C. (2013). Clinical genetics evaluation in identifying the etiology of autism spectrum disorders: 2013 guideline revisions. *Genetics in Medicine, 15*(5), 399–407. https://doi.org/10.1038/gim.2013.32.

Selkirk, C. G., McCarthy Veach, P., Lian, F., Schimmenti, L., & LeRoy, B. S. (2009). Parents' perceptions of autism spectrum disorder etiology and recurrence risk and effects of their perceptions on family planning: Recommendations for genetic counselors. *The Journal of Genetic Counseling, 18*(5), 507–519. https://doi.org/10.1007/s10897-009-9233-0.

Shea, L., Newschaffer, C. J., Xie, M., Myers, S. M., & Mandell, D. S. (2014). Genetic testing and genetic counseling among Medicaid-enrolled children with autism spectrum disorder in 2001 and 2007. *Human Genetics, 133*(1), 111–116. https://doi.org/10.1007/s00439-013-1362-8.

Tchaconas, A., & Adesman, A. (2017). Diagnostic evaluation of children with autism spectrum disorders: Clinician compliance with published guidelines. *Journal of Developmental and Behavioral Pediatrics, 38*(1), 29–38. https://doi.org/10.1097/DBP.0000000000000349.

Wydeven, K., Kwan, A., Hardan, A. Y., & Bernstein, J. A. (2012). Underutilization of genetics services for autism: The importance of parental awareness and provider recommendation. *The Journal of Genetic Counseling, 21*(6), 803–813.