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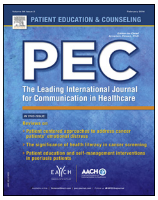

Autism genetic testing information needs among parents of affected children: A qualitative study

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ABSTRACT

Objective: Leading health agencies recommend physicians to provide information regarding genetic testing for autism spectrum disorders (ASD) to parents of affected children. How to effectively provide this information, however, is unclear for physicians. This qualitative study examined the information needs regarding ASD genetic testing among parents of affected children.

Methods: Semi-structured, in-depth interviews were conducted with 42 parents who had at least one child with ASD. Content analysis was utilized to analyze the interview data.

Results: The majority of parents (83%) reported they had never received information regarding ASD genetic testing from their doctors. Nevertheless, most parents (86%) expressed an interest to learn about this information. Their preferred topics included: cost (60%), benefits (48%), accuracy (38%), test procedure (29%), potential physical harms from the test (29%), confidentiality (12%), previous utilization by other affected families (2%), and eligibility criteria for this genetic testing (2%). Moreover, parents mentioned various methods to facilitate their learning, including Web-based approaches (43%), workshops/seminars (36%), brochures and flyers (31%), and videos (10%).

Conclusion: To promote parental informed decision-making regarding ASD genetic testing, educational materials should be developed based on our findings.

Practice implications: Application of these needs assessment findings will subsequently improve the delivery of healthcare services.

1. Introduction

The prevalence of autism spectrum disorders (ASD) in the United States is 1 in 68 children [1]. Affected children often show certain symptoms of these disorders before the age of 18 months [2,3], and by the age of two years a formal diagnosis of ASD can be made [1,4–6]. Yet, many affected children are not diagnosed until the age of four or more years [1]. For children affected with ASD, early diagnosis is critical as early interventions can significantly improve their language skills, intellectual capabilities and day-to-day living abilities [7–10]. ASD genetic testing, a standard diagnostic evaluation for ASD [11,12], can help with earlier and better diagnosis of ASD which may further facilitate better outcomes among affected children [13–15]. Alongside the early diagnosis, ASD genetic testing can help clarify the etiology of ASD and develop biologically-based approaches for the treatment of ASD-related complications [13,16,17].

According to the American College of Medical Genetics and Genomics practice guidelines [11] (p. 406), "[G]enetic testing should be discussed with all patients and families with ASDs", and a genetic consultation is recommended for families affected by ASD. The Autism A.L.A.R.M. Guidelines established by the American Academy of Pediatrics, the Centers for Disease Control and Prevention, and First Signs (a non-profit ASD organization) also suggest the need for physicians to provide ASD affected families with the updated information related to ASD (i.e., ASD genetic testing) [2]. Although parents of children with ASD may

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receive this information from genetics professionals, not everyone has access to genetic services [18]. This leaves the onus of offering the information of ASD genetic testing on pediatricians or primary care providers. Several studies, however, have shown that non-genetics physicians have insufficient competencies to present and explain genetic testing information to patients [19–21]. Yet, prior to referral, if the referring professionals (pediatricians or family medicine practitioners, in most cases) provide the affected families with information on genetic testing and evaluation, it may result in better follow-up by parents which may subsequently improve overall outcomes for their children [11].

To the best of our knowledge, no literature exists to guide physicians on how to effectively and comprehensively provide ASD genetic testing information to affected families. As parents of children with ASD – often the lifetime guardians of affected children – actively engage in decision-making for their children, it is anticipated that physicians should communicate adequately with these parents. To bridge this gap and provide evidence-based recommendations for ASD genetic testing education in the future, we conducted a needs assessment study to examine the information needs and wants regarding ASD genetic testing among parents of children with ASD by addressing the following questions: (1) Have parents of children with ASD ever received ASD genetic testing information from their doctors? (2) Are these parents interested in receiving ASD genetic testing information? (3) What type of information these parents need and which delivery methods are preferred by them to receive this information?

2. Methods

2.1. Study design and recruitment

As this study is a preliminary investigation, it is necessary to comprehensively understand parents' information needs regarding ASD genetic testing, which cannot be gathered through closed-ended survey questionnaires. Therefore, a qualitative method was used to conduct this study. All study procedures were approved by Texas A&M University's Institutional Review Board. The details of the study design, procedure, and data analysis have been described elsewhere [19,22].

In brief, semi-structured interview questions were developed and reviewed for the content validity by a panel of experts. Our participants were recruited from eight ASD communities and four professional conferences across Texas. Parents with at least one ASD affected child were eligible to participate. We also utilized a snowball sampling method to recruit the sample with various socio-demographic characteristics. Data was saturated with the final sample of 42 interviewees.

At the beginning of each interview, we explained the purpose of this study to participants and obtained their informed consents. Participants were then asked to complete a short survey, which included questions regarding their demographic characteristics and basic information of the affected children. Twenty-one individual interviews were conducted face-to-face, 16 by telephone and five by Skype live-cam (average interview time ~1 h). With participants' permission, interviews were recorded and field notes were taken. Each participant received an incentive of $50.

2.2. Data analysis and validation/trustworthiness

All interviews were recorded digitally and transcribed verbatim. With the assistance of Nvivo 9.0, two authors (M.L. and A.A.) used the content analysis approach [23] to code, categorize, discuss, and compare the reoccurring themes across the transcribed data. In particular, these two authors (M.L. and A.A.) independently identified the themes and subthemes of the data. They also met frequently to discuss and compare their coded themes and subthemes to reach an agreement. The final themes and subthemes were discussed and checked with the interviewer of this study (L.S.C.) for establishing trustworthiness. Field notes taken during interviews were also utilized to validate and supplement the data analysis.

3. Results

Interviews were conducted with 42 parents (32 mothers and 10 fathers with an average age of 44 years, SD = 8.5; range = 24–58). Half of the participants identified themselves as non-Hispanic White, 11 were Asian, six were Hispanic, three were Black, and one was multi-racial. More than two-thirds (78%) of our participants had obtained a baccalaureate or higher degree. Participants' annual household incomes were: ≤$25,000 ($n$ = 6), $25,000 to <$35,000 (n = 4), $35,000 to <$50,000 (n = 4), $50,000 to <$75,000 (n = 8), ≥$75,000 ($n$ = 19), and one participant declined to answer this question. Most interviewees (76%) had one child diagnosed with ASD. The diagnoses were Autistic disorder (n = 20), Pervasive developmental disorders-not otherwise specified (n = 14), Asperger syndrome (n = 6), and other/unsure (n = 2).

3.1. Did the parents of children with ASD receive any information regarding ASD genetic testing from their doctors?

The majority of the parents (n = 35, 83%) reported that they had never received any information regarding ASD genetic testing from their physicians. In particular, 12 participants had taken their affected children for testing, but nine parents told us they had not received any instructions regarding the procedure or any general information on ASD genetic testing. A mother, for example, reported that her daughter underwent ASD genetic testing at the age of six or seven years, but she did not recall receiving any information from her doctor before the test.

Conversely, seven participants (n = 7, 17%) were aware of ASD genetic testing. Yet, only two had obtained this information from their doctors. The remaining five parents studied about the ASD genetic testing from research papers, magazines, online resources, or Autism Speaks. Moreover, regardless of how parents acquired ASD genetic testing information, they all felt that the information they had received was limited and did not address their questions and concerns completely.

3.2. Were parents of children with ASD interested in receiving information regarding ASD genetic testing?

The majority of our participants (n = 36, 86%) expressed their interest in learning about ASD genetic testing information. For instance, one mother was interested to know about ASD genetic testing, but had no idea where to obtain the information. She stated:

> I don't have any resources of how would I find this [ASD genetic testing information]. And where would I find out this information? (#31, female, White, 42 years-old, high school diploma or less).

Participants disclosed the reasons behind their interest in learning about ASD genetic testing, and their most frequently cited factor was curiosity (n = 8, 24%). For instance, one mother wanted to explore what ASD genetic testing was and how it would benefit affected families. Furthermore, two parents believed the information was useful for their families and other parents in a similar situation. Lastly, one interviewee wished to be educated about ASD genetic testing to help develop educational materials for other parents.