Enhancing access to genetic testing for individuals with ASD/IDD by providing education to clinicians in the primary care setting

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Background

- Genetic testing can help in diagnosing and managing autism spectrum disorders (ASD) and intellectual and developmental disabilities (IDD), with genetic causes identified in 10-40% of cases.[1]
- The American College of Medical Genetics and Genomics (ACMG) recommends that all individuals diagnosed with ASD/IDD be offered genetic testing.[2,3]
- Primary care providers are uniquely equipped to provide counseling on genetic testing for patients with ASD/IDD, however implementation of genetic testing remains low in this population.[4,5]

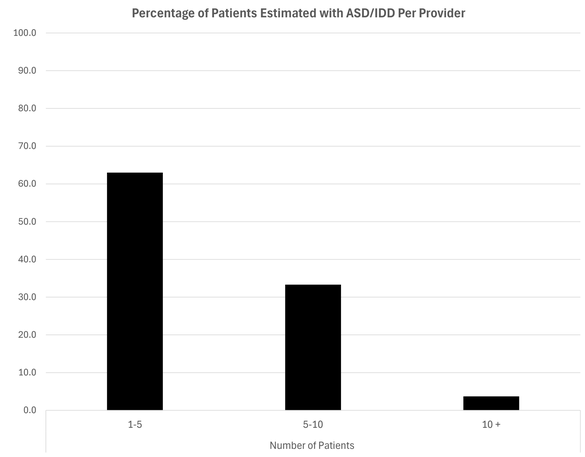
Objectives

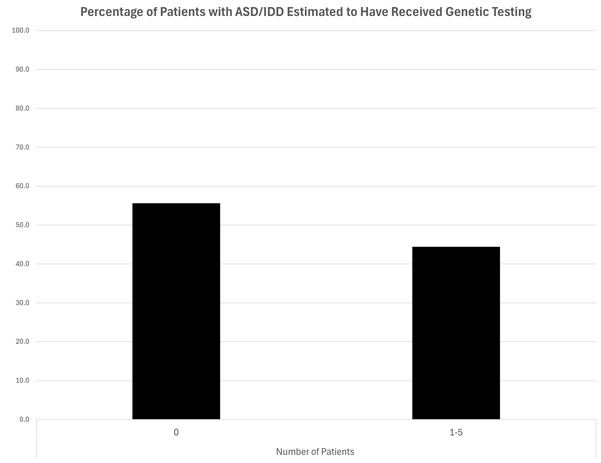
- To determine the barriers experienced by family medicine providers in regards to genetic testing for patients with autism and/or intellectual disabilities.

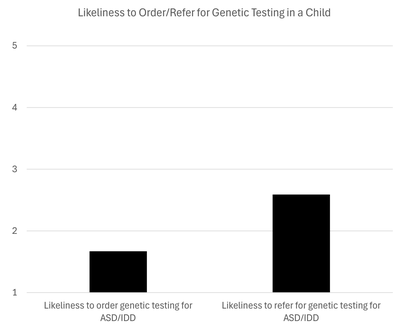
Methods

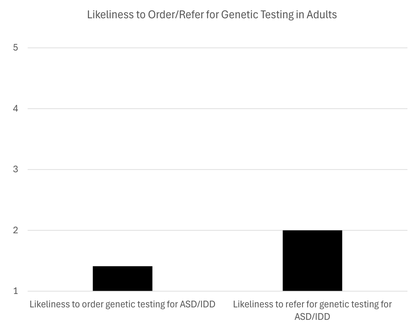
- We conducted a survey at a single site family medicine clinic to gather data on the current challenges faced by family medicine physicians regarding genetic testing for patients with ASD/IDD.
- Sixteen question survey included a mix of multiple-choice, Likert scale, and open-ended questions to gather quantitative and qualitative data with input from geneticists.
- Survey was developed and distributed via RedCap to a single site family medicine listserv.
- Performed quantitative analysis on closed-ended responses with statistical software (SPSS) and thematic analysis on open-ended responses to identify common themes and insights.

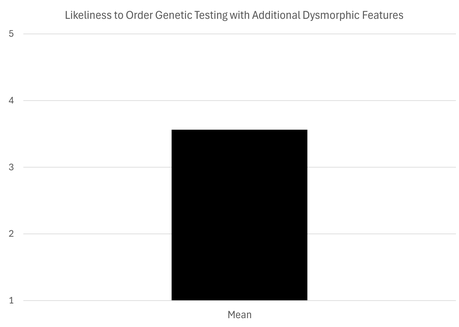
Results

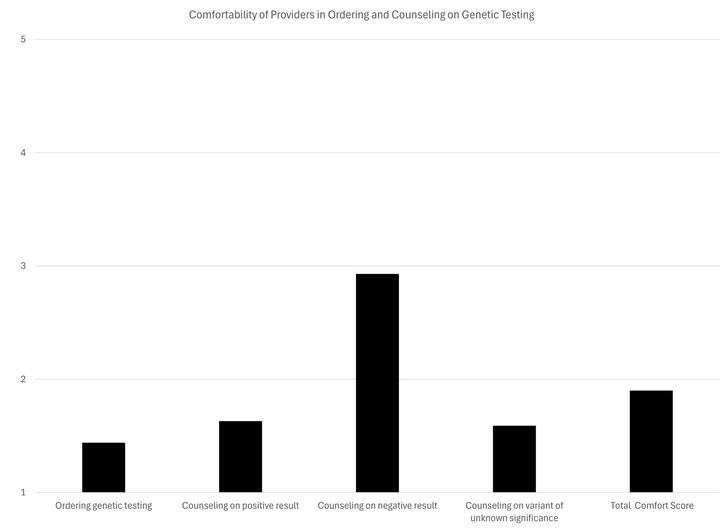












Conclusions

- There is a large gap in the number of patients per provider compared to the prevalence rate of people with ASD and more than half have not received genetic testing.
- Family medicine providers are more likely to order or refer for genetic testing in a child with ASD/IDD versus an adult and much more likely to order genetic testing if there additional dysmorphic features present.
- The total comfort score for ordering and counseling on genetic testing results is overall low in family medicine providers.
- There is a correlation between providers who think it is important for patients with ASD/IDD to receive genetic testing and to refer for genetic testing, especially in a child with ASD/IDD.

Implications

- With the growing prevalence of autism, there will be an increased demand for genetic testing in this population and family medicine providers can help to meet that demand.
- Increasing knowledge and education about the importance of genetic testing in this population could increase the likelihood of family medicine providers to refer or order genetic testing.

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