Improving Uptake of Referrals for Genetic Testing for Children Diagnosed with Autism Spectrum Disorder
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Background: As few as 3% of qualified families accept referrals for genetic testing for their children recently diagnosed with Autism Spectrum Disorder (ASD) despite myriad benefits and few risks. Benefits of undergoing genetic testing include receiving crucial information regarding related physical or medical conditions (and thus providing the ability to access early interventions), information which can impact family planning, as well as a sense of concreteness for parents. Primary risks include possibilities of feelings of guilt or disappointment.

Aim: To develop and test an intervention to improve uptake of referrals for genetic testing for children recently diagnosed with ASD.

Hypothesis: We predict that the intervention will have a higher percentage of accepted referrals than the control group.

Participants/Methods: This study will utilize a two-phase, qualitative-quantitative combined approach.

Phase 1:
- Semi-structured interview with parents who were previously referred for genetic testing
- Discovers parents’ understanding of genetic testing and their experience with their referral
- Code interview responses and extract common misconceptions as well as what parents generally did or did not like about their referral conversations
- Create an intervention (eLearning video encompassing the information provided from Phase 1 interview analyses)

Phase 2:
- Recruit and randomize clinicians into either the control or intervention group
- Intervention group clinicians show the video during referral conversations with families (inclusion criteria: child age 2-58, diagnosed with ASD in the past year); control group refer families for genetic testing without any additional components
- Access the child of each participating family’s medical chart 6 months post-referral to determine whether the family made an appointment for genetic testing; compare results between groups

Analysis: Phase 1 results will be qualitatively analyzed using the coding chart in Figure 2a. Phase 2 results will be analyzed using a Fischer’s exact test in addition to multiple regression analyses to determine the impact of possible confounding variables.

Interview Questions:
1. What was your initial reaction to your child’s ASD Diagnosis? ☐
2. Who mentioned a genetic test for your child? ☐
3. Before any specific conversation with a clinician regarding genetic testing, what was your prior knowledge of the subject? ☐
4. Did you ultimately agree to genetic testing for your child? ☐
5. If you were to redo the process of the proposal for genetic testing, what would the ideal scenario look like? ☐

Figure 1. Stem questions to be asked in every Phase 1 interview. Additional follow up questions are listed in the manuscript.

Interview Coding

Figure 2a. Full coding chart for Phase 1 interview analyses

Figure 2b. Example of a preliminary and secondary codes (used in Phase 1 analyses)

Figure 3. Stem questions to be asked in every Phase 2 interview. Additional follow up questions are listed in the manuscript.

Figure 4. Plan for the eLearning video to be used in Phase 2, including goals and leaving space for specific plans which will utilize Phase 1 data.

Discussion and Conclusions:
- If the hypothesis is supported, we will conclude that further education for clinicians and families that utilizes information gleaned from those who have undergone the process themselves is an effective way of increasing uptake of genetic testing referrals.
- If the hypothesis is not supported, we will investigate the impact of confounding variables such as race of parents, age of children, and type of clinician. We will also propose a future study which investigates the parents from the present study to determine the potential reasons for the lack of success in the present study.