Understanding Factors Influencing Genetic Testing for Autism Spectrum Disorders

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Background:

Autism spectrum disorders (ASDs) are among the psychiatric conditions with the highest genetic burden, with clinically identifiable pathogenic genetic changes explaining up to 35% of cases. Understanding the genetic underpinning can have an important impact on clinical management, including:

- Ending the diagnostic odyssey
- Defining more precisely recurrence risk
- Identifying risk of other mental health or medical conditions
- Connecting families
- Opening the door for tailored clinical management based on underlying areas of strengths and susceptibilities

This has led to the recommendation of genetic testing, specifically chromosomal microarray and Fragile X testing, as a key element in the evaluation of people with ASD, with growing support for exome sequencing as first tier testing. However, only a small proportion of patients with an ASD diagnosis receive such tests, highlighting a stark contrast between professional recommendations and clinical practice.

Objectives:

We aimed to develop a survey instrument to understand and address the factors influencing adoption of diagnostic genetic testing for ASDs, and subsequently share our experience as a team to develop clinical and educational interventions in response to those results.

Materials and Methods:

Rooted in implementation science, and guided by patients and families, a survey was developed and administered to explore knowledge, attitudes, and results of genetic testing among the autism community. Materials were available in both English and Spanish. Consolidated Framework for Implementation Research (CFIR) allowed for a structured and scientifically sound design in which to address the data.

CFIR Barrier Constructs		Specific Strategy for Local Healthcare System	Level
Innovation Characteristics	Complexity & Cost	Preautorization for our patients	System
		Psychiatric Genetic Counseling Service	System
	Relative Advantage	Genomic Psychiatry Consult Service	System
Outer Setting	Local Attitudes & Partnerships/Connections	Genomic Psychiatry Consult Service	System
		Psychiatric Genetic Counseling Service	System
Inner Setting	Work Infrastructure	Flowchart for testing protocol	System
	Recipient-Centeredness	Genomic Psychiatry Consult Service	System
	Relational Connections	Train the trainer	Clinician
	Communications	Grand Rounds	Clinician
	Deliverer-Centeredness	Growth of team	System
	Human-Equality Centeredness	Inpatient testing	System
Characteristics of Individuals	Opportunity	Translation of material in multiple languages	Patient
		Connection to advocacy groups	Patient
		Involve patients and families	Patient
	Capability	Psychiatric Genetic Counseling Service	Patient
		Buccal swab	Patient
		Explainer video	Patient
		Newsletter/ Marketing	System
	Need	Incorporation of testing into patient portal	System
		Patient results letter	Patient
Process	Engaging	Partnership with lab testing company	System
	Planning	Flowchart with testing process	System
	Doing	Patient result letter	Patient
	Reflecting & Evaluating	Quality improvement projects	System

Table 1. Healthcare strategies applied within CFIR constructs. Intervention: offering chromosomal microarray and Fragile X testing during healthcare encounters

Results:

Surveys were completed by 277 participants, for a 16% response rate. Most respondents were female caregivers of people on the autism spectrum, with a smaller proportion of people on the autism spectrum responding for themselves. Three main themes emerged from our descriptive, observational assessment: Education, Access and Communication. An implementation science lens enabled us to identify and design a multifaceted targeted strategy for each barrier culminating in the launch of successful clinical, educational, and communication interventions centered around the establishment of a Genetic Counselling and Genomic Psychiatry Clinical Consultation services.
Our team has been able to measure a higher frequency of individuals obtaining standard of care genetic testing, improved satisfaction and expansion of genetics-informed clinicians.

Disclosures:
The authors have no conflicts of interest to report

Conclusions:

Participant perspectives presented insights into barriers around education, communication, and access at various levels of care, suggesting possible solutions. Integration of implementation science to evaluate this data and facilitate evidence-based change is integral to creating a sustainable and reproducible process to bridge the gap between professional recommendations and clinical action. ASD care centered around an entwined genetic counselling and genomic psychiatry consultation services, provide the backbone from which to deliver a robust, successful and all-encompassing standard of care to every family living with autism and to bridge the gap between professional recommendations and clinical action.
Ongoing methodical quality improvement evaluation enables review of each CFIR barriers with the potential for further meaningful changes, ongoing assessment, and reproducibility across our institution and beyond.

"Timing is important. If genetic testing can provide a clear plan for medical treatment and other necessary treatments for the individual to help them to be what they want to be then I would be more apt to try. But genetic testing outstrips the process of treatment. Why bother finding out you have certain genes if there is absolutely nothing you can do about them/for them, except know you will pass them on to your own children if you have them?"

"Answering some of these questions brought up some unfavorable memories of me not getting to be able to comprehend the genetic testing that was done on my child"

"I only wish I knew what I now know thirty something years ago"

"Keep on working! ... specially to help the adults with ASD to be welcome, productive and accepted in the 'typical' world"

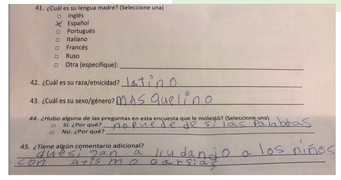

Figure 1. Excerpts from qualitative data obtain during surveys.

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