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Pharmacogenetic Testing In Patients with Autism Spectrum Disorder Evaluated in a Pediatric Precision Medicine Clinic

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Background & Objectives

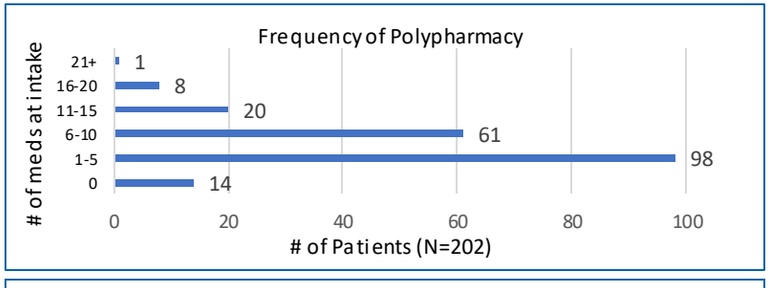
- Children with autism spectrum disorder (ASD) are more likely to be diagnosed with co-occurring mental health disorders and are also at increased risk for poor clinical response and adverse drug reactions
- No studies have investigated pharmacogenetic outcomes with patients with ASD
- **Primary Objective:** Characterize the demographic, clinical, and genetic profiles of children with ASD presenting for personalized medicine services
- **Secondary Objective:** Investigate the relationships between clinical phenotype and pharmacogenetic profiles

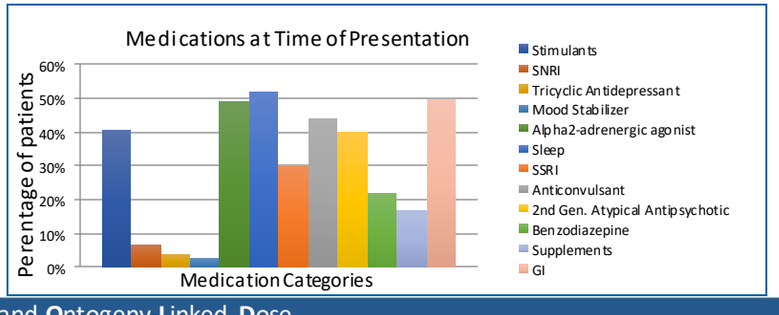
Methods

- This is a retrospective, observational cohort study utilizing the GOLDILOKs® Clinic and Autism Clinic REDcap databases, electronic medical records, and previously completed pharmacogenetic testing results
- **Inclusion Criteria:** Evaluation in the GOLDILOKs® clinic with documented diagnosis of ASD, autism, pervasive developmental disorder, or Asperger syndrome.

Results

Sample (n = 202)	Number (N)	Percentage (%)
Sex		
Female	63	31.19
Male	139	68.81
Race		
White	168	83.17
African American/Black	18	8.91
Asian	4	1.98
Other	10	4.95
Latino		
Yes	4	1.98
No	198	98.02
Referral Reason		
Adverse Drug Reaction	98	48.51
Poor Medication Response	131	64.85
Genotype Results Review	5	2.48
Genotyping Requested by Physician	29	14.36
Genotyping Requested by Family	25	12.38
Other	44	21.78
Referral Source		
Primary Care Provider	34	16.38
Subspecialist	158	78.22
Self-Referral	10	4.95





Conclusions

- At the time of referral, most patients were taking 1-10 medications (mean 6.15, SD 4.42, median 5)
- Sleep medications, medications targeting GI disorders, alpha2-adrengeric agonists, anticonvulsants, and stimulants were among the most common medications taken at the time of evaluation
- Most patients were referred by subspecialists
- Primary referral reasons were adverse drug reactions and poor medication response
- 12.9% of patients did not have results for PGX testing.
- Out of those with testing, 28.7% had changes in interpretation in CYP2D6 results since initial testing, primarily from extensive to intermediate metabolizer.
- The majority of recommendations made following testing were psychiatric in nature (68.6%).

Next Steps

- Conduct analysis with inclusion of a control group to evaluate for statistically significant differences
- Characterization of pharmacogenetic testing results and medications at the time of presentation with consideration of patient age, sex, and phenotype
- Multisite expansion of project to include individualized therapeutics clinics across the country for better generalizability
- Disparities remain evident with minority presentation for personalized medicine. Next steps include identifying modifiable factors to further address this.

LOVE WILL.

GOLDILOKs®: **G**enomic and **O**ntogeny-**L**inked **D**ose **I**ndividualization and c**L**inical **O**ptimization for **K**id**S**

PGX: Pharmacogenomic

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