Research Days

Pharmacogenetic Testing In Patients with Autism Spectrum Disorder Evaluated in the Children's Mercy Hospital GOLDILOKs© Clinic

Rachel Goodson

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☐ **Medical Student**
☐ **Resident/Psychology Intern (≤ 1 month of dedicated research time)**
☐ **Resident/Ph.D/post graduate (> 1 month of dedicated research time)**
X Fellow

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Other authors/contributors involved in project: Jennifer Wagner, MD; Sarah Soden, MD; Jean Dinh, PharmD, PhD; Vincent Staggs PhD; Tracy Sandritter, PharmD, BCPPS

IRB Number: 00001522

Describe role of Submitting/Presenting Trainee in this project (limit 150 words):
Submitting/presenting trainee is the primary investigator on this project. With the support of my mentors & collaborators, I have written the IRB and am leading data collection and data analysis.

Background, Objectives/Goal, Methods/Design, Results, Conclusions limited to 500 words

Background: Autism Spectrum Disorder (ASD) affects 1 in 54 children in the United States. Children with ASD are more likely to be diagnosed with co-occurring mental health disorders. There is currently little research guiding medication choice and dosing in patients with ASD. Children with ASD are at increased risk for reduced clinical response and adverse reaction (including due to polypharmacy). Pharmacogenomics is an approach for medication decision making that leverages an individual's genetic information and clinical presentation to make informed choices, but no studies have specifically investigated the outcomes of PGX for patients with ASD. Understanding the clinical and genetic predictors of drug response variability and the outcomes of personalized medicine can advance the clinical care of youth with ASD and set the stage for prospective investigations into further pharmacokinetic-pharmacodynamic studies, medication efficacy, and further pharmacogenomic research.

Objectives/Goal: The primary aim is to characterize the demographic, clinical, and genetic profiles of children with ASD who present for personalized medicine services. The secondary aim is to investigate the relationships between clinical phenotypes (e.g. IQ, adaptive scores, autism severity; comorbid diagnoses) and pharmacogenetics profiles of these patients.

Methods/Design: This retrospective, observational cohort study will utilize the GOLDILOKs© (Genomic and Ontogeny-Linked Dose Individualization and cLinical Optimization for Kids) Clinic

and Autism Clinic REDcap databases, electronic medical records, and previously completed pharmacogenetic testing results. The data extracted from the GOLDILOKs© Clinic REDcap includes demographic information, medications at time of visit, any adverse drug reactions, testing completed, results of said testing, and recommendations provided by the clinic. From the electronic medical records, when available we will extract information about the patients' autism spectrum disorder diagnoses including testing results and clinical phenotype where able. Inclusion criteria includes an evaluation in the GOLDILOKs© Clinic and a diagnosis of Autism Spectrum Disorder, Autism, Pervasive Developmental Disorder, or Asperger's Syndrome, as documented in the GOLDILOKs© Clinic database.

Results: A sample of 202 patients was identified based on the inclusion criteria noted above. See table 1 for their demographics and reasons for presenting to the clinic. Data analysis is in the beginning stages and will focus on characterizing patients with ASD presenting for personalized medicine evaluation as well as using baseline characteristics to predict the likelihood of actionable clinical recommendations.

Conclusions: Results from this study will directly inform personalized medicine referral practices and pharmacogenomic testing decisions. Improved decision making has the potential to enhance clinical response and minimize adverse drug reactions in patients with ASD and actionable pharmacogenetic differences. Moreover, results from this study will provide a framework for future autism/clinical pharmacology research and clinical collaboration, and potentially serve as pilot data for prospective investigation of personalized medicine decision making and outcomes.