2017 Southeastern Pediatric Research Conference

June 9, 2017
Georgia Aquarium
Case Study 4a - Predictive diagnostics
Patterns of Visual Engagement Identify Distinct Subgroups of 24-Month-Olds with Autism Spectrum Disorder

Julia Yurkovic, Scott Gillespie, Warren Jones, Ami Klin, Sarah Shultz

3rd Place
Basic Science Poster
Presented on June 9, 2017

2017 Southeastern Pediatric Research Conference
Children’s Healthcare of Atlanta Data Warehouse

Todd Sharp, MD, MBA

2017 Southeastern Pediatric Research Conference
HOW TO ACCESS

Someone from the study team familiar with the Research Protocol Review Form (RPRF) can provide assistance. The RPRF should be submitted with a copy of the protocol and any supporting documentation.

Please allow 2-4 weeks for each request. Study team members will return the Radiology Protocol Review Form (RPRF) and our protocol approval status. The RPRF protocol review, required for this study, includes a description of what information is required.

IMPORTANT CONTACTS

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MRI: 78 Studies
CT: 40 Studies
Nuclear Medicine: 53 Studies
Ultrasound: 15 Studies
Interventional Radiology: 3 Studies
Impact of this system

- Self-monitoring of anemia in chronically ill patients (~5.5 million in U.S)
- Self-screening of anemia in the general healthy population
- Inexpensive anemia testing in global health settings (> 1 billion affected in LRS)

- Inflammatory Diseases
- Neonates/Toddlers
- Elderly
- Chronic Infections
- Aging Age
The test that keeps on giving

**RE-ANALYSIS**

- **CSER Research Program at HudsonAlpha**
  - Previously unidentified genotypes
  - New analytical methods, newly published information
  - Whole genome sequencing 11% provided a diagnosis when reanalyzed one year later

- **Undiagnosed Disease Network (UDN)**
  - Baylor using Exomes 5% identified new diagnoses when re-analyzed a year later
  - HudsonAlpha—redoing whole genome on previous exams – 16% increase in diagnosis

- **Pharmacogenomics**
  - Available for the rest of the patient’s life
  - Better choices by the right drug at the right dose at the right time
  - Real-time always up to date
Impact of Diagnostic Error

- Institute of Medicine just reported 5% of Diagnoses are errors. On average physician makes 62 misdiagnoses per year!
- 10% of deaths and 6% to 17% of admissions due to misdiagnosis.
- Leading reason for malpractice claims.
Whole Exome Sequencing

Whole Genome Sequencing