Genomic Medicine in Pediatric Patients – Obstacles and Future Directions

Panel Discussion

Summary/Moderator: Jeff Botkin

What makes children different?

- Health conditions impacting children often different from adults
  - Children may reflect precursors to adult-onset conditions
  - Many serious health conditions in children are uncommon
- Children may be found to be at risk for diseases years or decades in the future
- Children cannot consent to participation in research
  - Older children can assent
  - Need to engage parents and children in education process
- Children and parent-child relationship may be more sensitive to psychosocial impacts

Obstacles to Current Research

- Incongruity between adult and pediatric datasets
  - Clarification of how pediatric/adult conditions are selected for analysis
- Is there a need to increase overlap between pediatric and adult phenotypes being targeted?
- Is the eMERGE network large enough with respect to pediatric participants?

New Approaches to Existing Data

- Copy Number Variants (CNV)
  - Good analytic tools available
  - Do we need better data on "frequencies and boundaries" of CNV's in Database of Genomic Variation?
  - Can eMERGE contribute to data on pathogenicity?

Prospective Directions

- Development of custom chip with broad spectrum of clinically relevant variants and CNV's
- Is the field ripe for a tool of this sort?

Panelist Questions and Issues

- Additional clarity on the "paths" for phenotyping adults versus children
- Should eMERGE consider more GxE data collection for children?
- Questions about potentially unique issues with pediatric participants
  - Return of results
    - Adult onset conditions
    - Results of uncertain clinical significance
  - Return of incidental findings
  - Genetic analysis of parents to clarify a child's results

Other questions…

- Is there a relationship between eMERGE and the newborn screening sequencing projects?
- Are there target conditions for genomic analysis that may have early clinical utility in the healthcare of children?