Searching for Autism Genes

Autism is a common disorder with a strong genetic basis. Genetic variants that modulate risk have been identified but fail to explain etiology in the vast majority of cases. Towards an improved understanding Dr. Brett Abrahams and his colleagues are undertaking a multipronged approach to identify additional risk factors and explore how they may interact. They are performing array CGH tests in families to interrogate the genome for rare structural variants that may be contributory. Such methods have proven informative in up to 20% of individuals. Dr. Abrahams is also interrogating the protein coding portion of the genome for point mutations that may confer risk. Subsets of cases to be evaluated by each approach are already known to carry a first contributory mutation; the new efforts aim to identify additional co-occurring variants that may shape outcomes. Finally, they are testing whether variants that play a role in syndromic autism might contribute to idiopathic cases in which the molecular basis remains unknown.

Critical for each of these efforts is the recruitment of local children and families to participate in research. Molecular overlap between the genetic factors that contribute to autism risk and clinically distinct conditions including intellectual disability, epilepsy, and schizophrenia suggest that ongoing efforts are likely to provide insight to a variety of disorders that impact the lives of children and families affected by this condition.

Einstein Montefiore Partnership: RIC and EHIT

As an example of the power of collaboration, the ICTR Research Informatics Core and Emerging Health IT (EHIT) recently enhanced the usefulness of Clinical Looking Glass (CLG), a software tool developed by EHIT for clinical data capture. Cancer investigators can now access data from the New York State Cancer Registry (NYSCR) via CLG. We interviewed Alexandre Peshansky, RIC’s senior bioinformatics analyst, regarding the Cancer Registry project.

What is the New York State Cancer Registry (NYSCR)? AP: The NYSCR collects and processes information on cancer cases in the State and produces reports on cancer incidence and mortality statewide. It is a comprehensive database of information on cancer cases in New York and very useful to health care professionals and researchers to track outcomes.

What information is now available through CLG? AP: Data in the NYSCR is now up to date and accessible through the CLG tool. In addition to thinking of CLG as a search tool, it acts as a clinical data warehouse that is updated frequently, and into which additional patient information can be added. Of course, CLG’s mainstay is the clinical information that it captures from the Montefiore electronic medical record, but the added NYSCR enhancement will be important for our cancer investigators.

What was RIC’s role in this project? AP: EHIT recognized an unmet need within CLG. After discussions with Cancer Center faculty, RIC collaborated with EHIT to meet this need. The RIC developed complex SQL query, which allowed the CLG to periodically download data from the newly reformatted NYSCR, thus providing up-to-date data for researchers.

If researchers need additional help in evaluating the data, who can they turn to? AP: The RIC can work with researchers on an individual basis to ensure they are receiving appropriate information, and will perform data analysis needed for the investigators’ grants.

Who can access this tool and how can they access it? AP: All Einstein and Montefiore investigators with access to CLG can access the NYSCR data. In order to gain access to the CLG investigators can visit: http://explorecagnostics.montefiore.org/. Once investigators are signed into CLG they can access NYSCR through Smart Reports.