MINC Gene Scene

April is Autism Awareness Month

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According to the Centers for Disease Control and Prevention (CDC), autism spectrum disorders (ASD) are a group of developmental disabilities characterized by impairments in social interaction and communication and by restricted, repetitive, and stereotyped patterns of behavior. Symptoms of autism are typically apparent before age 3. When all forms of autism are considered, the incidence is approximately one in 110 individuals and it is three to four times more common in males.

Autism may be caused by a combination of both genetic and environmental factors. When a genetic cause cannot be identified, the recurrence risk after one child is diagnosed with ASD may be as high as 18 percent. When two siblings are diagnosed with an ASD, the recurrence risk increases to 25 to 35 percent.

Geneticists do not typically diagnose autism and there is no specific genetic test that can diagnose autism. However, genetic tests may help to identify an underlying cause for the ASD. A genetic cause can be identified in approximately 20 to 25 percent of individuals with autism.

Genetic causes of autism:

- Chromosome abnormality (5 percent)
- Microarray finding (10 percent)
- Fragile X syndrome (5 percent)
- *MECP2* gene changes (5 percent - females only)
- *PTEN* gene changes (3 percent - if head circumference >2.5 SDs)

Why is a genetics evaluation important?

- A diagnosis can help families acquire additional resources and services through their insurance company or school district
- Allows families with similar diagnoses to connect through support groups and online communities
- Families may be empowered by knowledge of the underlying cause of a relative's disorder
- Depending on the etiology, associated medical risks may be identified that lead to screening and the potential for prevention of morbidity
- Specific recurrence risk counseling—beyond general epidemiologic information—can be provided
- Targeted testing of at risk family members can be offered
- In a limited number of cases (e.g., metabolic disorders), targeted therapies may be or become available

Nursing Competency: **Uses genetic- and genomic-based interventions and information to improve clients' outcomes.**

National DNA Day is April 25, 2013 (60th Anniversary of the Discovery of DNA's Double Helix)

National DNA Day commemorates the successful completion of the Human Genome Project in 2003 and the discovery of DNA's double helix by James Watson and Francis Crick in 1953. This annual celebration encourages students, teachers and the public to learn about the latest advances in genomic research and explore what the information may mean for their lives.

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Sigma Theta Tau has a Genomic Special Issue of the *Journal of Nursing Scholarship* addressing genomics of common health conditions, emerging genomic science and technology, and the ethical, legal, social and nursing research issues associated with the integration of genomics into health care. The articles, as well as webinar presentations, are open access and available at the following link: http://www.genome.gov/27552093