LEND | Leadership Education in Neurodevelopmental Disabilities and Related Disorders

ASD Module Script

Module 4: Etiology

Etiological Assumptions: Investigating Causes of ASD: Welcome to the module on contemporary evidence regarding the etiology of ASD. Understanding current etiological assumptions can help families make decisions about vaccinations and other environmental factors that could be affecting their child.

Etiology: Genetics: Here is what we already know. The symptoms of ASD are neurologically based, and scientific evidence indicates a genetic predisposition for ASD. Contemporary etiological assumptions result largely from family and twin studies as well as association with genetic disorders of known etiology.

Etiology: Genetics: Family Studies: First we will look at family studies. The term broader autism phenotype reflects the concept that each symptom of autism exists along a continuum with typical behavior. Symptoms may include differences in language and psychiatric symptoms, such as anxiety or mood disorders. These may occur with greater frequency in family members of people with ASD. Understanding there is a gradient of symptoms is an important step in understanding how multiple genes must interact to result in ASD. Further, current findings indicate that when a first-born has ASD, recurrent risk for the subsequent siblings is 4% if the child is a girl and 7% if the affected child is a boy. This is ten-fold higher than what would be expected in the general population. If the second child has autism, subsequent siblings have a 25%-35% risk for ASD.

Etiology: Genetics: Twin Studies: Now we will look at twin studies. An identical twin of a child with ASD has a 70-90% risk of having ASD. Almost all identical twins of a person with ASD have some symptoms, although fraternal twins have no greater rate of autism than other siblings. This strongly supports the genetic component of ASD. These rates also suggest an interaction of multiple genes, and that unknown environmental factors may influence gene expression.

Etiology: Genetics: Associated Genetic Disorders: ASD is associated with other genetic disorders that have known etiologies. It is understood that brain functioning in certain genetic disorders, such as Fragile X, place an individual at greater risk for ASD. An important point about Fragile X is that a child does not need the full-blown syndrome to have autistic symptoms. Microscopic gene deletions or duplications are also related to behaviors symptomatic of ASD. Combined, these etiologies account for 5-10 percent of diagnostic etiologies, so it is important that all children receive a medical workup after diagnosis.

Etiology: Genetics: To wrap up genetic etiology, it is important to emphasize:

- Evidence strongly supports a genetic component of ASD
- Current research suggests an interaction of multiple genes
- Probable that unknown environmental factors influence gene expression
- More research is necessary

Etiology: Brain Structure and Functioning: Some studies have found neurobiological differences in individuals with ASD, including irregularities in head circumference, brain volume, cell size and structure, protein levels, and white matter in the cortex & cerebellum. Now with FMRI technology, researchers are studying changes in brains that correspond with mental operations and how they compare between neuro-typical brains and brains with ASD. However, more data is needed to understand how these irregularities are linked to ASD. FMRI studies show differences in facial and object processing – suggesting important differences in brain function in individuals with autism; however, this is NOT a diagnostic tool.

Etiology: Neurochemical: There are neurochemical-based theories of etiology due to irregularities in levels of neurotransmitters, listed here (dopamine, norepinephrine, serotonin, endorphins, and opioids in the GI tract). Abnormalities in these levels are present in individuals without ASD, so over all, the evidence is limited, but it does relate to how children with ASD are sometimes medicated for symptoms (e.g. common co-existing anxiety or mood problems).

Etiology: Environmental Exposures 1: Before continuing on to theories about environmental factors and ASD etiology, the debate about vaccines should be addressed. There has been significant controversy about the alleged association of measles, mumps, and Rubella (MMR) vaccine and ASD. In 1998, a controversial and well-known theory proposed by a researcher named Wakefield linked Thimerisol in the MMR shot to ASD. Subsequently, epidemiological data as well as the work of several researchers has not supported the association of the MMR vaccination with autism.

Etiology: Environmental Exposures 2: As stated earlier, it is likely that environmental factors interact with genes to cause symptoms of ASD. To date, the only established environmental risk factors are medications that were prescribed to expecting mothers in the ‘50s and ‘60s that are now known teratogens (substances that increase the risk of birth defects).

Etiology: Environmental Exposures 3: Prenatal infections, such as Rubella increase risk for CP, ID, visual impairments, & ASDs. This risk has been eliminated in the U.S. due to appropriate vaccinations for women. Other exposures to infections in early childhood or neurologic injury after meningitis can also result in ASD symptoms. Regardless of what causes differences in development, whether infection or even injury, a person can be diagnosed behaviorally for ASD.

Etiology: Gender and ASDs: When investigating the cause of ASD, people often consider the excess of identified males compared to females. The gender ratio for ASD is 4:1, male to female if IQ > 50 and 1:1 if IQ is < 50. This may be explained by genetic or hormonal susceptibility or the application of diagnostic criteria.

Etiology: ASD and ID Comorbidity: ASD may be associated with intellectual disability, as well as other conditions. Thus far, most studies report that up to ¾ of individuals with ASD also have intellectual disabilities. However, with more inclusive criteria for ASD, it is likely that increasing numbers of individuals with typical cognitive abilities and with significant intellectual disability will be identified as having an autism spectrum disorder. This overlap of symptoms can complicate the diagnostic process, which is why a qualified professional and comprehensive evaluation is necessary.

Etiology: ASD and Epilepsy Comorbidity: Overall, epilepsy is reported in about ¼ of individuals with ASD and most commonly present in infancy and adolescence. While the implications of this finding are unclear, it is significant to note that uncontrolled epilepsy can affect general health, learning and functioning.

Summary: Etiological Assumptions: You have now completed Module 4: Etiology. In this module we evaluated contemporary evidence regarding the etiology of Autism Spectrum Disorders. Please proceed to the post assessment.