



Etiological Assumptions

Investigating causes of ASD

Etiology

Genetics

- ASD is assumed to result from *several causes*, but with a strong genetic component
 - Family & twin studies
 - Association with genetic disorders of known etiology

Etiology

Genetics: Family Studies

- Rates among relatives are also higher than general population for diagnosis or features consistent with the broad ASD phenotype
 - e.g., speech and language differences, anxiety, mood disorders
- *Recurrence risk for siblings is 4% if the child is a girl and 7% if affected child is a boy*
 - *If second child has autism, subsequent siblings: 25%-35% risk for ASD*

Etiology

Genetics: Twin Studies

Monozygotic (identical) twins:

70 – 90 % risk

Dizygotic (fraternal) twins:

no greater risk

than subsequent siblings

Etiology

Genetics: Associated genetic disorders

- Some single gene diseases are known to present greater risk for ASDs:
 - Tuberous sclerosis
 - Seizure Disorder
 - Fragile X
- Microscopic gene deletions or duplications

Etiology:

Genetics

- Evidence strongly supports a genetic component
- 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

- Irregularities in
 - head circumference
 - brain volume
 - cell size/structure
 - protein levels
 - White matter
- FMRI studies have shown differences in facial/object processing

Etiology:

Neurochemical

- Dopamine
- Norepinephrine
- Serotonin
- Endorphins
- Opioids in the GI tract

Etiology:

Environmental Exposures

The vaccination debate

- Thimerisol in MMR shot
- **No causal association**

Etiology:

Environmental Exposures

- Teratogens associated with increased ASD risk
 - Thalidomide
 - Valproic acid
 - Mesoprostal
- No other known chemicals

Etiology:

Environmental Exposures

- Prenatal infections
 - Rubella: increases risk for CP, ID, visual impairments, & ASDs
 - This risk has been eliminated in the U.S. due to appropriate vaccinations for women
- Infections in early childhood
- Neurologic injury after meningitis

Etiology:

Gender and ASDs

- 4:1 ratio of boys to girls for ASDs if IQ > 50
- 1:1 ratio of gender if IQ is < 50

Etiology:

ASD & ID Co-morbidity

40-75%

of children diagnosed with an **ASD** have **ID**

Nebel-Schwalm, M. S., & Matson, J. L. (2008). Differential Diagnosis. In J.L. Matson, *Clinical Assessment and Intervention for Autism Spectrum Disorders* (pp. 91-114). Burlington, MA: Elsevier. Towbin, K.E., Mauk, J.E., & Batshaw, M.L. (2007). Autism Spectrum Disorders. In M.L. Batshaw, *Children with Disabilities* (pp. 325-343). Baltimore, MD: Paul H. Brookes.

Etiology:

ASD & Epilepsy Co-morbidity

- Epilepsy occurs in 1/4 of cases of ASD
 - Literature has ranged from 5% to 46%
- Conversely, ASD is reported to occur at an increased incidence (32%) in populations of people with epilepsy



Summary: Etiological Assumptions