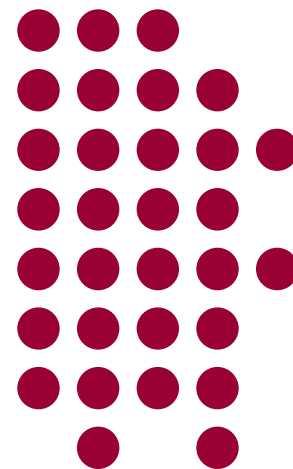


The Fragile X Behavioral Profile

Brenda Finucane, MS, CGC
Executive Director, Genetic Services



What is Etiology?

- **Etiology: Underlying cause**
- **Developmental disorders are symptoms for which there are many different etiologies**
- **Genetic and / or medical factors play a role in the etiology of most developmental disorders**

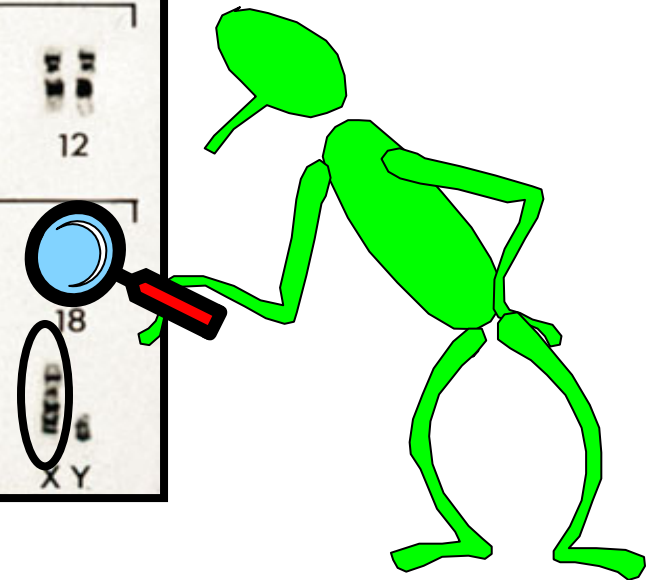
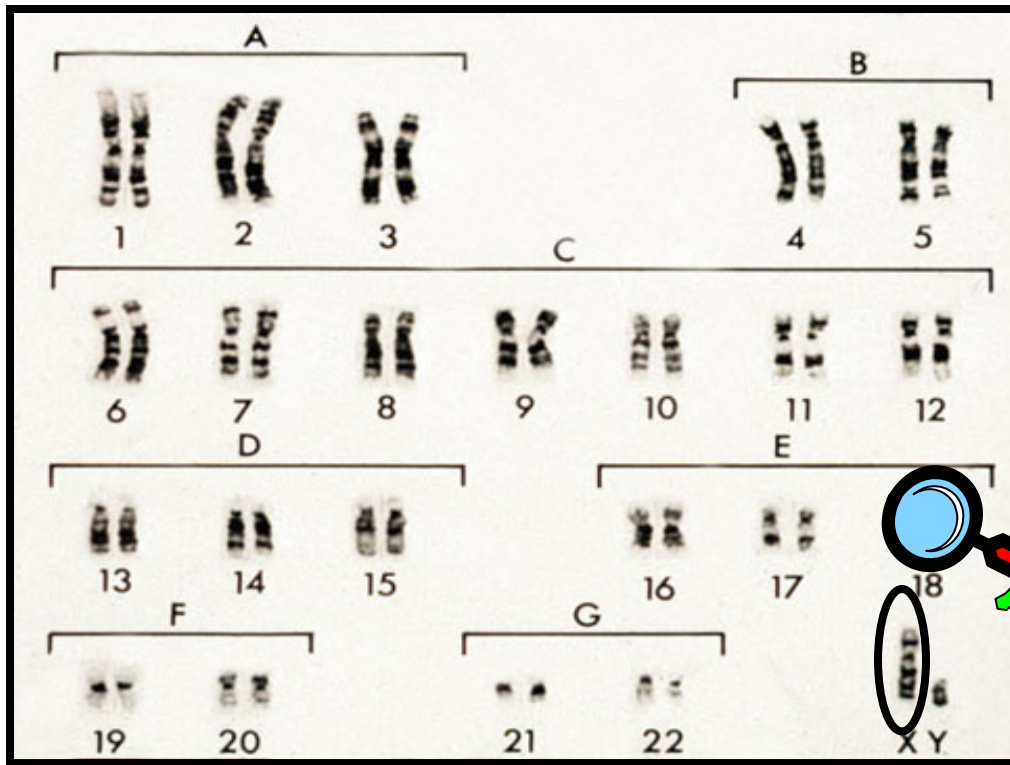
Why is Etiology Important?

- **Genetic counseling for families**
- **Alleviates guilt, misconceptions**
- **Anticipation of medical needs**
- **Insight into behavior, learning styles**
- **Support groups**

Psychiatric Diagnoses

- **based upon observed, recognizable patterns of human behavior**
- **diagnosed using criteria found in the DSM (Diagnostic & Statistical Manual)**
- **symptom diagnoses which do not emphasize etiology**
- **not diagnosed using laboratory tests**

Fragile X Syndrome



Fragile X Syndrome

- Most common hereditary cause of developmental disabilities in all populations
- Occurs in both males and females, although males are more commonly affected
- Diagnosed through DNA blood testing
- Physical features often subtle
- Majority of those affected are undiagnosed

Intellectual Functioning

- Majority of males function in the moderate range of intellectual disability
- <5% of males have IQs above 70
- Much more variability among females, ranging from severe intellectual disability to above average IQ

Fragile X and Autism

- Up to 21% of young boys with fragile X syndrome meet diagnostic criteria for Autistic Disorder
- A majority of males and many females with fragile X syndrome have symptoms consistent with an autism spectrum disorder

Autism and Fragile X

- Autism: recognizable pattern of behavioral symptoms
- Numerous known and unknown causes
- Fragile X: most common known single gene cause of autism
- Prevalence of fragile X among children with autism estimated to be around 1 in 20

Behavioral Characteristics

- Hyperactivity
- Hand-flapping
- Hand-biting
- Tactile defensiveness
- Perseverative speech
- Sensory hyperarousal
- Gaze aversion

Fragile X Syndrome

- characteristic speech patterns
- poor auditory processing
- sensory integration disorder
- deficits in sequential processing

versus Down Syndrome.....

- relative strength in sequential processing
- auditory processing not impaired
- relatively few maladaptive behaviors

Fragile X Syndrome

- imitation / modeling
- shy but friendly personality
- good sense of humor
- excellent long-term memory
- respond well to structure / routine

Hyperarousal in Fragile X Syndrome

- Over-reaction to sensory stimuli
- Enhanced electrodermal response to sensory stimuli which correlates inversely with FMRP levels
- Enhanced cortisol release after stressors
- Delayed physiological recovery after hyperarousal

Hessl et al., *Journal of Child Psychology and Psychiatry*, 2006

Scharfenaker and Stackhouse, *Natl Fragile X Fdn Quarterly*, 2006

Boccia and Roberts, *Behavior Research Methods, Instruments, and Computers*, 2000

Gaze Aversion

- Gaze aversion: distinctive form of poor eye contact (active turning away versus “looking through you”)
- Majority of males with fragile X syndrome exhibit gaze aversion, starting in early childhood
- Forced eye contact in males with fragile X syndrome triggers abnormal cortisol response
- Gaze aversion may serve as an avoidance behavior



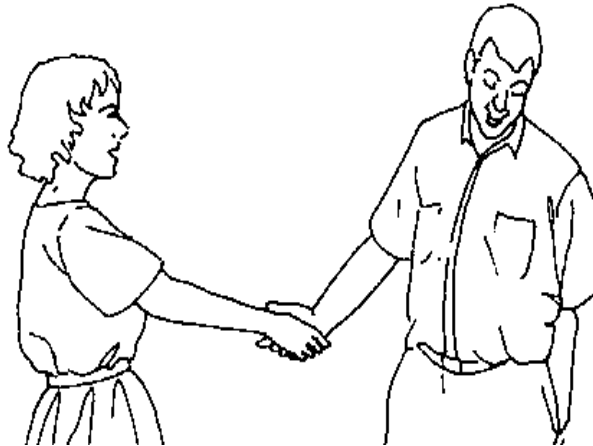
American Journal on Mental Retardation 93:406-411, 1989



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Gaze Aversion

- Verbal prompting and forced eye contact often exacerbate gaze aversion in males with fragile X
- When eye contact goals are de-emphasized, eye contact often improves



Anxiety / Hyperarousal in Fragile X Syndrome

EARLY SIGNS:

- self-injury (hand- and wrist- biting)
- mouthing objects, clothes
- hand-flapping

ESCALATE TO:

- task refusal
- leaving the area
- verbal aggression (often imitated phrases)

Anxiety / Hyperarousal in Fragile X Syndrome

ESCALATE TO:

- property destruction (threatened & real)
- breaking personal objects (glasses)
- door slamming
- throwing objects

ESCALATE TO:

- physical aggression (threatened & real)
- punching
- slapping

Anxiety / Hyperarousal in Fragile X Syndrome

TRIGGERS:

- forced eye contact
- personal space issues
- tactile defensiveness
- emotional tone of peers and staff
- changes in routine
- auditory stimuli
- changes in environment

Anxiety / Hyperarousal in Fragile X Syndrome

OTHER POTENTIAL CONTRIBUTORS:

- auditory processing deficits
- delayed emotional processing
- hypersensitivity to negative correction
- performance anxiety
- poor concept of time
- strong associations (positive & negative)

FRAGILE X SYNDROME: POSITIVE ATTRIBUTES

- Friendly but shy
- Communicative – usually verbal
- Good symbol recognition
- Simultaneous learners
- Sense of humor
- Good visual memory
- Responsive to structure and routine

FRAGILE X SYNDROME: POSITIVE ATTRIBUTES

- Rise to the occasion – dressed for success
- Enjoy typical peers
- Recognizable response to anxiety
- Outburst triggers often identifiable
- Interest in community / vocational activities

FRAGILE X SYNDROME: CHALLENGING ATTRIBUTES

- Anxiety in novel situations
- Hyperactive / impulsive
- Distractible
- Over-reactive
- Resistant to change
- Low muscle tone, poor fine motor
- Sensory defensiveness

FRAGILE X SYNDROME: CHALLENGING ATTRIBUTES

- “Out of seat” behaviors
- Poor numerical, sequential processing
- Simultaneous processors
- Low tolerance for frustration
- Don’t like being singled out
- Attachment to inanimate objects
- Poor auditory processing

ATTRIBUTES THAT CAN GO EITHER WAY

- Imitation (verbal and nonverbal)
- Self-talking
- Vicarious learning
- Peer-orientation

Fragile Syndrome

Elwyn Fragile X Center
www.elwyngenetics.org

National Fragile X Foundation
www.fragileX.org

FRAXA Research Foundation
www.fraxa.org