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HST.161 Molecular Biology and Genetics in Modern Medicine
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Lecture 16

Rett Syndrome

1/10,000 female births; one of most common causes of autism in girls

Nearly 100% de novo

Diagnostic criteria:

Necessary:

- normal pre- and perinatal history
- normal psychomotor development for first 6 mo.
- Normal head circumference at birth
- Postnatal deceleration of head growth in most individuals
- Lost of purposeful hand skills between 6 months and 2.5 years
- Hand stereotypies
- Evolving social withdrawal, communication dysfunction, loss of acquired speech, and cognitive impairment
- Impairment or deterioration of locomotion

Supportive:

- breathing disturbances during waking hours
- bruxisms (grinding and gnashing the teeth)
- impairment of sleeping pattern from early infancy
- abnormal muscle tone associated with muscle wasting and myotonia
- peripheral vasomotor disturbances
- progressive kyphosis ("hunchback" curve to the spine) or scoliosis (Sidewise, S-curve to the spine)
- growth retardation
- hypotrophic small and cold hands/feet

Exclusion criteria:

- evidence of a storage disorder including organomegaly
- cataract, retinopathy, or optic atrophy
- history of perinatal or postnatal brain damage
- confirmed inborn error of metabolism or neurodegenerative disorder
- acquired neurological disorder due to severe head trauma or infection

Videos portray:

- rigidity while walking
- other characteristics of walking abnormalities: feet turned outward, pauses while walking (requiring prompting to keep moving), jerky turning around
- motor tremors
- limited hand functionality; unable to grasp small things effectively
- complex hand stereotypies; often hand-to-mouth. Become more pronounced when excited, interested, or anxious.
- unprovoked crying spells
- in more severe cases, little interaction with others; no communication
- often more anxious and fearful than normal

Rett syndrome caused by mutations in X-linked MeCP2

Core features:

- motor syndrome
- behavioral syndrome
- cortical dysfunction
- respiratory dysfunction
- autonomic dysfunction
- somatic growth failure
- [others – see slides]

Growth failure:

- girls are often smaller in height and weight than their peers, especially as they get older