Rett Syndrome

Neurodevelopmental disorder

• Genetic mutation in the MECP2 gene on the X chromosome • Random mutation (sporadic cases) • Rarely inherited (X-linked dominant pattern)	**Causes**
• Normal early growth followed by loss of motor and communication skills • Hand-wringing or washing movements • Breathing irregularities (e.g., hyperventilation) • Seizures • Scoliosis	**Symptoms**
• Symptomatic and supportive care • Physical therapy and occupational therapy • Medications for seizures and breathing abnormalities • Management of scoliosis	**Current Therapy**
• Not preventable due to genetic cause • Senetic counseling for families with a history of Rett Syndrome	**Prevention**
• Lifelong condition • Variable severity; some individuals may live into adulthood with supportive care • No cure; research ongoing into potential therapies	**Prognosis**

Affected Population: Predominantly females
(1 in 10,000 births)