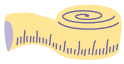
RETT SYNDROME

A **rare neurodevelopmental disorder** caused by a **mutation in the MECP2 gene**, resulting in insufficient or abnormal production of the MECP2 protein, which is **essential for normal brain function**. Primarily affecting **females**, it occurs in 1 in 8,500 births and follows an **X-linked dominant inheritance** pattern.

CLINICAL FEATURES

Growth:
- **Progressive microcephaly**
- Failure to thrive



Development:
- Initially, development is appropriate
- **Regression** occurs by 18 months of age

Neurologic:
- **Seizures**

- Stereotypies
  - Clapping
  - **Hand-wringing**

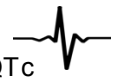
- Gait ataxia
- Neuropsychiatric disturbance
  - Agitation

- **Sleep disturbance**

Gastrointestinal:
- Constipation

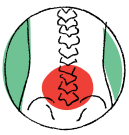
Extremities:
- Small hands and feet

Other:
- Prolonged QTc



- Bruxism (teeth grinding)

- Scoliosis



PROGRESSION THROUGH 4 STAGES

Early Onset	Rapid Regression	Plateau	Late Deterioration
6-18 months	1-4 years	2-10 years	After 10 years old
▪ Delayed development ▪ Reduced interaction	▪ Loss of speech, hand use, and mobility ▪ Repetitive hand movements	▪ Symptoms stabilize ▪ Behavior improves ▪ Motor issues persist	▪ Loss of mobility ▪ Muscle weakness ▪ Cognitive abilities often stable

DIAGNOSIS

- Identification of **key clinical features**

Genetic Testing
- MECP2 molecular genetic testing

SURVEILLANCE

- **ECG** to rule out prolonged QTc
- **EEG** if suspected seizure activity

MANAGEMENT

Treatment focuses on **symptom control** with a **multidisciplinary approach**
i.e., antiepileptic drugs for seizures, melatonin for sleep disturbance, selective serotonin reuptake inhibitors (SSRIs) for agitation, fiber and PEG-3350 for constipation.

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