



RETT SYNDROME:

Comprehensive Care Quick Reference



Rett Syndrome (RTT) is a neurodevelopmental disorder almost always associated with a MECP2 gene mutation on the X-chromosome. In general, patients gain developmental skills at a near normal rate until 1-3 years when they lose skills in ambulation, speech, and hand use while developing repetitive, purposeless, stereotyped hand movements. RTT most commonly occurs in females but is increasingly recognized in males.

Review of Systems (Rett-related Items)

Genetics: Review genetic testing. Genetic counseling, including reproductive options.

Nutrition: Routes of nutrition, adequate calories, fluid, calcium, and vit. D

GI: Discuss chewing and swallowing, constipation, slow gastric motility, and reflux. Gallbladder disease may present early in RTT.

Pulmonary: Screen for sleep-disordered and awake-disordered breathing (i.e. tachypnea, air swallowing, and breath-holding +/- cyanosis). Frequent pneumonias may indicate aspiration and swallowing difficulty.

Development: Initial visit document developmental milestones (fine and gross motor, language). Screen for changes after the initial visit.

Neurology: Including sleep, dysautonomia, Rett spells. If seizures note frequency and changes in presentation. (Weight changes of >20% may require antiseizure medication dose changes). Abnormal movements (e.g. tremor, myoclonus, chorea, and dystonia). Use of FDA-approved medications for treatment of RTT?

Cardiology: If history of prolonged QTc interval, confirm Cardiology involvement. Annual EKG performed?

Musculoskeletal: Discuss the involvement of PMR and Ortho. Screen for mobility and use of mobility aids. Assess flexibility. Osteopenia risk with certain anticonvulsants, proton pump inhibitors, some contraceptives.

Urology: Screen for urinary tract infections and urinary retention (>8-10 hours daily to weekly) can be caused by dysautonomia, constipation, and anticholinergic medications. Kidney stone risk with poor hydration and carbonic anhydrase inhibitors (acetazolamide, topiramate, and zonisamide).

Endocrine: Regular menstruation? Increased seizures with menses? Signs of premature adrenarche?

Psychiatric/Behavioral: Screen for symptoms of anxiety and depression such as social withdrawal, screaming, irritability, and sensory processing difficulties.

Equipment used: Braces, wheelchairs, lifts, shower accommodations, bedside toilets, etc.

Therapies: Often include speech therapy involving augmentative and alternative communication (AAC) training, feeding therapy, physical therapy, occupational therapy, vision therapy, therapeutic horseback riding (hippotherapy), and swim/aqua therapy.

Ophthalmology: Regular visits and screening for cortical visual impairment.

Dental: Regular care. Bruxism present?

Education: Ask about IEP and teacher experience level with RTT

Social: Assess for sources of family stress (financial, social, care fatigue). Long-term care plans including plans for a will and establishing a legal trust/estate. Guardian wishes for DNR and advance care directives. Review available social support resources including disability parking permits, respite care, legal assistance for people with disabilities, etc.



Physical Exam Findings Expected with RTT

Gen: Short stature

HEENT: Dilated but normally reactive pupils

Abd: Abdominal bloating (from air swallowing)

Pulmonary: Tachypnea, air swallowing, breath holding +/- cyanosis

Musculoskeletal: Scoliosis, cool extremities, flushing of skin

Neuro: Absence of spoken language but socially engaging. Repetitive, purposeless hand movements. Tremors, myoclonus, chorea, and dystonia. Dyspraxic/impaired gait

Derm: Skin breakdown and calluses from repetitive hand movements and mouthing behaviors

Genitalia: Signs of premature adrenarche

Screening Labs

Yearly: Vit D, Chemistry panel (with Calcium, Phosphorus), Hgb

At least once: Lipid panel

Every 2 years: Urinalysis

PRN: T4 and TSH

Miscellaneous Screening Tests

Yearly: EKG, spine x-ray (if scoliosis noted), vision screening

Common Indications that may Trigger a Specialist Consult

Gastroenterology: Chewing/swallowing difficulties, reflux, ulcers, strictures, severe constipation

Pulmonology: Frequent respiratory infections, hypoxemia, sleep apnea concerns

Neurology: Seizures and suspicious spells, abnormal movements, Rett-specific treatments

Cardiology: Prolonged QT interval

Orthopedics: Scoliosis (Cobb angle >20o), decreased hip ROM, joint contractures

PMR: Equipment, muscle rigidity/spasticity, dystonia

Endocrine: Hypothyroidism, osteopenia/osteoporosis, menses suppression

Urology: Frequent UTI, urinary retention, kidney stones

Gynecology: Menses suppression

Additional Resources

Rett Comprehensive Care Guidelines with References: www.Rettsyndrome.org/Guidelines

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