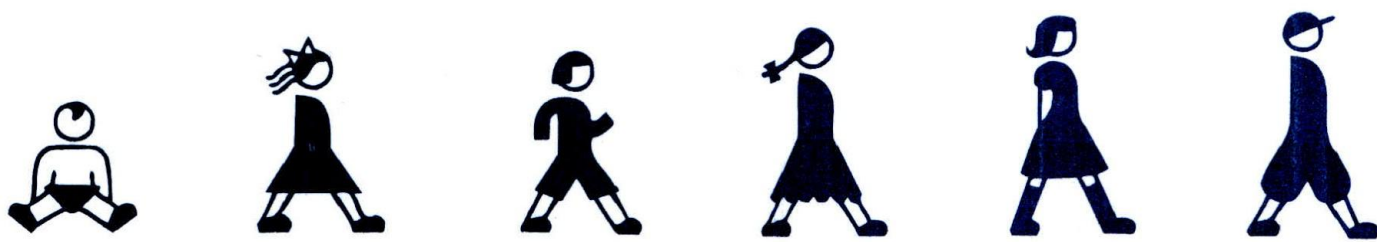


Snyder-Robinson Syndrome

Medical Management

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A H W A T U K E E P E D I A T R I C S P . C .

Physical Phenotype

- Mid face flatness / retraction
- Prominent lower lip/prognathia
- High arched palate / possible cleft

Physical Phenotype (cont.)

- Hypotonia
- Asthenic body, pectus excavatum and kyphoscoliosis
- Long hands, big toes, osteoporosis, fractures and hip luxation

Other Physical Findings

- Kidney stones
- Nephrocalcinosis, mixed RTA
- Speech abnormalities
- Unsteady gait
- Seizures
- Intellectual disability

Neurology

- Not all Snyder-Robinson Syndrome patients will have the same presentation
- Seizures -EEG
- Hypsarrhythmia- infantile spasms
- Lennox Gastaut – daily, multiple seizures; varied in presentation
 - Tonic and myoclonic (most common),
 - Atonic / drop attacks (1 – 4 sec, head nodding, sagging at the knees)
 - Tonic-clonic
 - Atypical absence

Ophthalmology / Vision

- Ophthalmic evaluation
- Cortical Visual Impairment
- Retinal pigmentation
- Retinal scar
- Photophobia

Pulmonology

- Reactive Airways Disease
- Pneumonia, chronic pneumonia
- Laryngo, tracheo, bronchomalacia
- Mucociliary dysfunction?
- Chylothorax (1 patient)
- Pneumothorax

Gastrointestinal

- Gastroesophageal reflux disease (GERD)
- Failure to thrive
- Slow eating
- G-tube
- GI bleed

GI / Fat Metabolism ?

- Chronic constipation and /or secretory diarrhea, dysmotility, ileus
- Disorder of fat metabolism?
 - Endoscopy and/or colonoscopy
 - Ultrasound / echocardiogram

Genitourinary

- Neurogenic bladder
- Undescended testicle (cryptorchidism)

Electrolytes

- Mixed RTA/ electrolyte abnormalities
- Glucose abnormalities
- Hypokalemia (decreased potassium)
- Bicarb loss (decreased CO₂)
- Sodium loss
- Calcium loss
- Magnesium loss

Renal

- Kidney disease
- Renal cysts
- Mixed renal tubular acidosis
- Nephrocalcinosis
- Calcium metabolism abnormality?

Osteoporosis –
demineralization of the
bones

Mish-Mosh

- Hematologic abnormalities
 - Anemia of chronic disease
 - Iron deficiency anemia
 - Thrombocytopenia
- Temperature regulation

Development

- SRS patients will present with varying types of developmental delay
- Speech Therapy
- Physical Therapy
- Occupational Therapy
- Music Therapy

Development

- Aquatic Therapy
- Equestrian Therapy
- Habilitation
- Attendant Care
- Respite Care
- Nursing Care

Tying up loose ends

- Raise awareness
- Education