Neuroorthopedic Management of Rett Syndrome

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**Purpose:** This course will present an overview of the management of patients with Rett Syndrome. The speakers will discuss the genetics, pathophysiology, and diagnosis of Rett syndrome. The initial treatment strategies, surgical indications, and the orthopedic surgical treatment of patients with Rett Syndrome will also be presented.

**Target Audience:** Physicians, Occupational and Physical Therapists, Nurses

**Course Summary:** This course will provide an introductory level discussion of orthopedic impairments of children with Rett Syndrome, and the typical orthopedic surgical procedures that may be required in the care of those children. The impact of these deformities and surgeries on the patient and their family will be discussed within the context of the International Classification of Functioning, Disability and Health (ICF). A review of the genetics, pathophysiology, diagnosis, and prognosis of Rett Syndrome will be discussed. Surgical indications will be presented for the typical orthopedic deformities in patients with Rett Syndrome. Surgical management will be discussed thoroughly, including postoperative management.

**After completion of this course, participants will be able to**

**Learning Objective 1:** Discuss the pathophysiology and diagnosis of Rett Syndrome,

**Learning Objective 2:** Understand the orthopedic impairments of Rett Syndrome and the principles of non-operative care,

**Learning Objective 3:** Understand the typical orthopedic surgical procedures that children with Rett Syndrome typical undergo, including spine fusion and the typical outcomes that can be expected.
Outline:

I. Introduction (Mohan Belthur, M.D.)
   a. Definition of Rett Syndrome
      i. Epidemiology
      ii. Genetics

II. Clinical Features
   a. Pathophysiology
   b. Clinical Presentation
   c. Diagnostic Tests
      i. Pediatric Neurology Exam
      ii. Clinical Exam - Criteria
         1. Main
         2. Supportive
         3. Exclusion
      iii. Genetic testing – MECP2
      iv. Neuroimaging - Exclusion
   d. Differential Diagnosis:
      i. Cerebral Palsy
      ii. Autism
   e. Stages of Rett Syndrome
      i. Stage I
      ii. Stage II
      iii. Stage III
      iv. Stage IV

III. Medical Co-Morbidities
   a. GI reflux
   b. Constipation
   c. Long QT Syndrome
      i. Risk for sudden cardiac death syndrome

IV. Orthopedic Manifestations
   a. Scoliosis
   b. Foot deformities
   c. Lower Extremity Contractures
V. Scoliosis (M. Wade Shrader, M.D.)

a. Incidence

b. Pathophysiology
   i. Low tone
   ii. Non-ambulation
   iii. ???

c. Effect on longevity
   i. Recent literature suggests that PSF may prolong life

d. Nonoperative care
   i. Close monitoring
   ii. Seating/wheelchair accommodation
   iii. Bracing?

e. Operative care
   i. Surgical Details
   ii. Perioperative management
   iii. Results in the Literature

f. Parental/Family Impact of Scoliosis/PSF
VI. Neurological Assessment and Treatment of Rett Syndrome (Michael Kruer, M.D., Ph.D)