

- Signs & Symptoms
- High arched palate
  - Muscle Weakness
  - Hypotonia (Lack of muscle tone)
  - Contractures
  - Scoliosis may develop in childhood.
  - Feeding difficulties
  - Respiratory Distress

Pathophysiology

This disease is caused by defects or deficiencies of myotubularin, a protein thought to promote normal muscle development. It is inherited in an X-linked recessive pattern, meaning it primarily affects boys, who inherit the disease through their mothers. Girls affected the condition is typically much less severe.

- Diagnostics/Labs
- Detailed Family history
  - Thorough clinical evaluation
  - Muscle biopsy
  - Molecular genetic testing

## Centronuclear Myopathy

- Treatment/Medication
- Ventilator
  - Trach - Bivona CPAP-Trach
  - G-tube
- Meds:
- Glycopyrrolate
  - Pediatric multivitamin with iron
  - Acetaminophen

- Nursing Implications
- Oxygen
    - o Nasal Cannula
    - o Continuous Positive Airway Pressure
    - o Ventilator
  - Intravenous Catheter treatments
  - Fluids and Nutrients
  - Positioning
  - Suctioning Airway as needed with Trach
  - Ensure client has properly sized and shaped tube.
  - Transport patient with portable oxygen, an Ambu bag, suction equipment, and extra tracheostomy tube.

- Patient Teaching
- Range of motion impairment
  - Gastrostomy Device, Care
  - Oral intake Inadequate
  - Preterm education
  - Infection
  - Tracheostomy care

D.O.B. 11/17/22

Gestational Age 32w4d Adjusted Gestational Age 43w4d

Birthweight 3 lbs. 1570 oz./ grams

Current weight 7 lbs. 3380 oz./ grams

APGAR at birth: 2/7/9

Priority Nursing Diagnosis

Impaired physical Mobility