

### Signs & Symptoms

- Respiratory distress
- less than 100 BPM or Absent HR
- Absent of newborn reflexes
- Paler cyanosis
- Flaccid limp muscle tone
- Irregular breathing
- Weak cry

### Diagnosis/Labs

Muscle X-ray

Approximately 80% of males affected, characterized by polyhydramnios, decreased fetal movement, and neonatal weakness, hypotonia, and respiratory failure. Motor milestones are significantly delayed, and most individuals fail to achieve independent ambulation. Weakness is profound and often involves facial and extraocular muscles. Respiratory failure is nearly uniform, with most individuals

- ECG
- MRI
- CBC
- CMP

### Genetics

Autosomal recessive

Genetic testing

Carrier status

Family history

Genetic counseling

## Myotubular Myopathy (X-MTM)

### Treatment/Medication

- Airway
- Breathing
- Circulation

### Nursing Implications

- Temperature
- Suction
- Resuscitations protocol

### Patient Teaching

- Tracheotomy maintenance

### Priority Nursing Diagnosis

Myotubular Myopathy

Respiratory Distress Syndrome

D.O.B. 11/17/2022

Gestational Age: 32<sup>6</sup> weeks Adjusted Gestational Age: 51<sup>4</sup> weeks

Birthweight 1 lbs. 7.1 oz./ 775.5 grams

Current weight 3 lbs. 7.6 oz./ 1557 grams