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### SMA Type 1 Mimicry: A four-month-old in the NICU with Failure to Thrive

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## SMA Type 1 Mimicry: A four-month-old in the NICU with Failure to Thrive

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### **Introduction:**

Spinal muscular atrophy with respiratory distress type 1 (SMARD1) is a progressive autosomal recessive neuromuscular disease mimicking spinal muscular atrophy type 1. SMARD 1 invariably results in diaphragmatic paralysis and respiratory failure requiring mechanical ventilation. We report a case of a patient presenting with failure to thrive subsequently diagnosed with SMARD1.

### **Case Presentation:**

Patient is a 35.3-week gestation infant born to a 28-year-old mother with a prenatal course complicated by reverse end diastolic flow of the placenta and intrauterine growth retardation (IUGR). Infant required a one-month neonatal intensive care unit (NICU) hospitalization after birth for feeding difficulties and respiratory distress requiring supplemental oxygen. Infant was on room air and tolerating oral feeds at discharge. Infant required readmission to the NICU at corrected gestational age of 46 weeks for failure to thrive, feeding intolerance, and projectile vomiting. Infant was discharged 2 days later after weight gain on 200mL/kg/day of 24 kcal/oz feeds and a normal abdominal Ultrasound. She was readmitted from clinic at 50 weeks corrected gestational age for hypoxemia and respiratory failure requiring mechanical ventilation. She subsequently failed several extubation attempts. Serial chest radiographs were concerning for an eventration the diaphragm. Fluoroscopy of the diaphragm displayed bilateral diaphragmatic paralysis. Infant required nasogastric feeds for appropriate weight gain. At three months of life the infant developed generalized hypotonia, which continued to progress, while remaining cognitively intact with appropriate levels of alertness. An SMA genetic panel was negative. Next generation sequencing revealed mutations in the IGHMBP2 gene, consistent with a diagnosis of SMARD1.

### **Discussion:**

SMARD1 is characterized by worsening respiratory distress and failure in the first 13 months of life. The clinical manifestations of SMARD1 include a) respiratory distress between 6 weeks and 6 months, b) presence of diaphragmatic paralysis, c) distal muscular weakness, and d) IUGR. Patients present with distal muscular weakness that can later generalize. Progressive diaphragmatic weakness and paralysis is caused by mutations in the IGHMBP2 gene, with the loss of muscle strength occurring in parallel. IGHMBP2 is expressed primarily by motoneurons and myocytes, however the downstream events resulting from a dysfunctional IGHMBP2 gene to the clinical manifestation of this disorder remain unknown. Death before one year of age results from respiratory failure after a decision to withhold intubation or withdraw mechanical ventilation is made.

**Conclusion:** SMARD1 should be considered as a differential as a rare cause of neonatal failure to thrive, particularly in the setting of progressive hypotonia on neurological exam.