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Dehydrated Hereditary Stomatocytosis Causing Severe Ascites Leading to Pulmonary Hypoplasia and Respiratory Insufficiency in a Neonate

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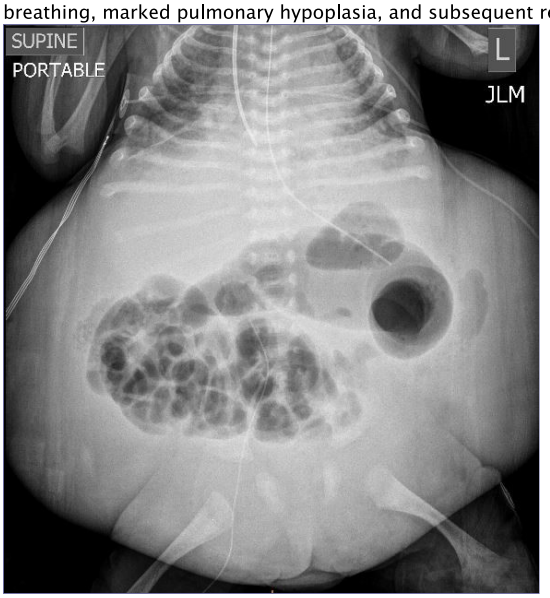
Abstract:
Introduction: Dehydrated hereditary stomatocytosis (DHS) is a rare, autosomal dominant disease primarily causing chronic anemia. However, DHS has been associated with perinatal ascites and edema. We report a unique case of DHS associated with severe ascites leading to pulmonary hypoplasia with resultant respiratory insufficiency in a neonate.
Case Presentation: EM is a 32.5-week EGA infant who was born with severe congenital ascites (following repeated prenatal paracentesis) identified on prenatal ultrasound. Initial chest-abdomen imaging is shown in Figure 1. He required CPAP at delivery but was weaned to high flow nasal cannula with intermittent episodes of prolonged intubation during repeat paracentesis. Paracentesis demonstrated simple ascites rather than chylous fluid. Lymphangiograms demonstrated transient lymphatic leakage with resolution. Genetic evaluation revealed a pathogenic mutation in the *PIEZO1* gene for autosomal dominant DHS. This disorder has reduced penetrance and variable expressivity and symptoms range from absence of clinical features to lethal perinatal edema. During his first year of life EM developed severe chronic obstructive pulmonary disease with hypoxemia and a requirement for supplemental oxygen, restrictive lung disease secondary to thoracic dystrophy, and chronic lung disease of prematurity. He remained on supplemental oxygen for the first year and a half of life.
Discussion: DHS is a rare form of autosomal dominant hemolytic anemia reported in 26 individuals among 7 families worldwide. Clinical presentation varies significantly and includes chronic anemia, neonatal edema and ascites, and lymphatic malformations. Symptoms may develop in utero, during the perinatal period, or later in adulthood. Pulmonary hypoplasia and respiratory insufficiency caused by pre/perinatal ascites have not been reported. DHS is caused by gain-of-function mutations in the *PIEZO1* (16q24.3) gene, as was demonstrated in our patient. Mutations result in increased red cell cation membrane permeability, particularly potassium, leading to depletion, dehydration, and shortened red cell survival. Expression of *PIEZO1* in lymphatics may explain pre/perinatal edema and ascites. EM developed severe prenatal ascites leading to abnormal abdominal musculature and impaired diaphragm movement and fetal breathing causing pulmonary hypoplasia with subsequent respiratory insufficiency at birth. The pathophysiology in this case is similar to that of prune belly, where abnormal abdominal musculature is a known cause of impaired lung growth and development and subsequent respiratory compromise. Respiratory complications are primarily managed with supplemental oxygen and ventilatory support, if needed, until the lungs mature. This case highlights a previously unreported extremely rare disease, DHS, leading to abnormal fetal breathing, marked pulmonary hypoplasia, and subsequent respiratory insufficiency.



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