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SCIENTIFIC LETTER

Gaucher Disease – A Rare Cause of Collodion

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To the Editor: A baby boy, twin 2 of a dichorionic diamniotic (DCDA) twin pregnancy was noted to have ascites, hydrocele and small pericardial effusion antenatally. At birth, the baby had tight shiny skin with absent skin creases (Collodion), blueberry muffin spots, everted eyelids, abdominal distension, hepatosplenomegaly and hydrocele.

Initial investigations at birth revealed persistent thrombocytopenia without coagulopathy with a platelet count of 26×10^9/L (normal 100–500 × 10^9/L) on day one of life and conjugated hyperbilirubinemia. Viral studies to rule out congenital infections, cranial and cardiac ultrasound were normal. An abdominal ultrasound confirmed mild to moderate hepatosplenomegaly. Ferritin level was 1021 μg/L (normal 10–336 μg/L).

Investigations for metabolic disorders showed a mild increase in ammonia of 55 μmol/L (16–50 μmol/L) and markedly raised glucosyl sphingosine of 2620 nmol/L (normal <10 nmol/L). Dried blood spot lysosomal enzyme analysis showed reduced β-glucocerebrosidase of 0.2 μmol/h/L (normal 2.5–8.5 μmol/h/L) and elevated chitotriosidase of 71 nmol/h/ml (normal 2–50 nmol/h/ml) confirming the diagnosis of lethal variant of Type 2 congenital Gaucher disease. Gaucher disease variant analysis revealed that he was homozygous for the pathogenic c.971G > A (p.Arg324His) variant in the GBA gene. He was provided supportive palliative care with high humidity, topical skin moisturizers, parenteral nutrition and antibiotics. He died from multiorgan failure at 22 d of life.

Gaucher disease (GD) is an autosomal recessive condition resulting from mutations in the glucocerebrosidase gene, causing deficiency of the lysosomal enzyme, beta glucocerebrosidase [1, 2]. The condition is characterized by the accumulation of lipid-laden macrophages known as Gaucher cells that infiltrate the bone marrow, spleen, liver and occasionally the lungs resulting in multiorgan dysfunction [1, 3]. In this case, the combination of hepatosplenomegaly, the timeline of presentation, thrombocytopenia, elevated ferritin and collodion membrane suggested perinatal GD.

The mainstay of treatment is enzyme replacement therapy (ERT) and substrate reduction therapy (SRT) which could be beneficial for other forms of Gaucher disease (*e.g.,* visceral manifestation of neuropathic GD) but not for the lethal perinatal GD [2]. This case illustrates the importance of considering lethal form of Gaucher's disease in a Collodion baby and providing palliative care in these cases.

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Compliance with Ethical Standards

Conflict of Interest None.

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