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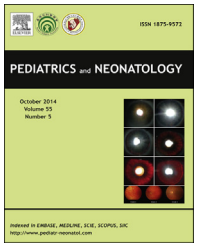

CASE REPORT

Infantile Systemic Hyalinosis Complicated with Right Atrial Thrombus and Pericardial Effusion in an Infant

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Infantile systemic hyalinosis (ISH) is a rare multisystem fatal autosomal recessive disorder that involves widespread deposition of hyaline on connective tissues and certain internal organs. The major manifestations include painful articular contractures, hyperpigmentation, subcutaneous nodules, gingival hypertrophy, failure to thrive secondary to protein-losing enteropathy, and osteolytic bone lesions. In this paper, we report a 12-month-old girl with ISH presenting with recurrent diarrhea, failure to thrive, and refractory infections. A molecular study identified a homozygous missense mutation, c.134T > C; p.L45P, in exon 1 of the anthrax toxin receptor 2 (*ANTRX2*) gene. Our patient passed through an eventful course that included septic shock, central line infections, right atrial thrombosis, and pericardial effusion. She incurred acute bronchiolitis due to respiratory syncytial virus infection, which led to her death. In conclusion, this case report highlights that severe and life-threatening morbidities and complications can be encountered in ISH, to which some management options can be applied.
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1. Introduction

Infantile systemic hyalinosis (ISH) [OMIM 236490] is a rare autosomal recessive disorder that usually manifests in infancy and leads to early childhood death.[1,2] Infantile systemic hyalinosis is characterized by widespread deposition of hyaline, which may produce distinctive features of progressive joint contractures, skin papules, gingival hypertrophy, chronic diarrhea, and growth retardation.[3,4]

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Patients with ISH are at increased risk of severe infections secondary to impaired immunity.[1,5] The gene responsible for ISH has been mapped to chromosome 4q21.21. The cause of ISH is mutations in the anthrax toxin receptor 2 (*ANTRX2*) gene, also known as the capillary morphogenesis protein 2 (*CMG2*) gene.[6] The purpose of this case report was to highlight the severe and life-threatening morbidities and complications encountered in ISH, to which some management options can be applied.

2. Case Report

A 12-month-old Saudi girl was born at full-term to a couple who were second cousins. This couple has two other healthy older children. There was no family history of birth defects, learning difficulties, or genetic disorders. The infant's prenatal and neonatal history was unremarkable. Our patient was delivered by spontaneous vaginal delivery. Her birth weight was 2.1 kg (<3rd percentile), and her head circumference and length were within the 25th percentile. She was apparently normal at birth.

In the 2nd month of life, she appeared to have growth retardation and required frequent hospitalizations for hypoalbuminemia, dehydration, and recurrent infections ranging from upper respiratory tract infection, pneumonia, urinary tract infection, and gastroenteritis, to full-blown septic shock. At the age of 4 months, she presented with painful restriction of the upper and lower extremities at several large and small joints. Despite aggressive physiotherapy, joint contractures ineluctably emerged and affected the large and small joints of the upper and lower limbs. She also developed gum hypertrophy, small ear lobe nodules, hyperpigmented macules on the bony prominences, periosteal reaction in the long bones, and a perianal fleshy mass. These symptoms and signs prompted us to determine a diagnosis of ISH. The DNA from lymphocytes was used to amplify the 17 coding exons and their corresponding flanking sequences of the *ANTXR2* gene. The polymerase chain reaction (PCR) products were analyzed by sequencing in the forward and reverse directions. Sequence analysis identified a variant of a missense mutation, c.134T > C; p.L45P, in exon 1 of the *ANTRX2* gene. Serum albumin was persistently low and the globulin level was normal. A cause for the recurrent infections could not be identified by extensive immunological workups, which included immunoglobulins, complements, lymphocyte subsets, T and B lymphocyte markers, and neutrophil functions.

At 8 months of age, she presented with fever, diarrhea, vomiting, and refusal to feed. She became severely ill and dehydrated. Her albumin level was 6 g/L. She received intravenous broad-spectrum antibiotics and albumin infusion. One week later, a femoral central line was established for the infusion of total parental nutrition (TPN). Two weeks after central line insertion, our patient developed central line infection that required another course of broad-spectrum antibiotics. Echocardiography showed a vegetation/clot extending from the inferior vena cava, the right atrial junction, and the right atrium (Figure 1). However, the tip of the central line catheter was in the optimal position in the inferior vena cava and not in direct contact

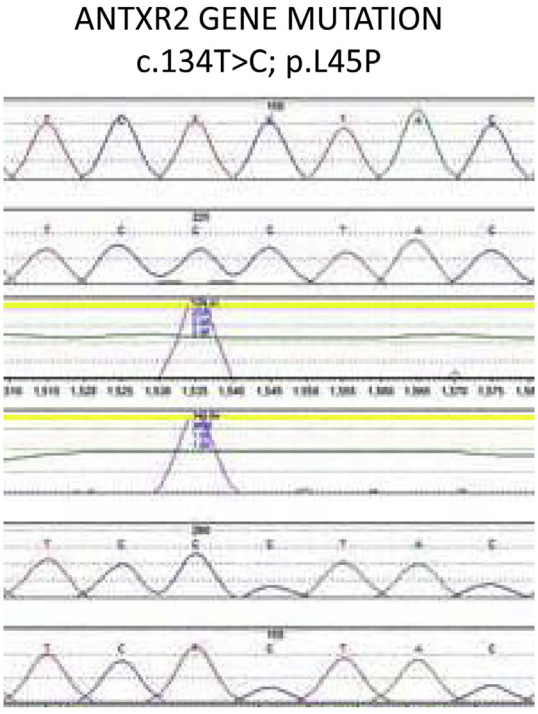

Figure 1 The electropherogram for the *ANTXR2* gene mutation c.134T > C; p.L45P.

with the right atrial wall. Prothrombin time, activated partial prothrombin time, and international normalized ratio were prolonged. Antithrombin II1, protein C, and protein S levels were within normal limits. These coagulation studies were compatible with disseminated intravascular coagulopathy, which was treated with vitamin K, fresh frozen plasma, and a continuous infusion of heparin. She subsequently developed pericardial effusion (Figure 2). Pericardiocentesis led to immediate hemodynamic improvement. The aspirated fluid was chylous; it contained normal protein and was free of red blood cells. She remained in the pediatric intensive care unit (PICU) for 4 months, during which time the vegetative blood clot enlarged progressively, despite vigorous administration of intravenous heparin. She unfortunately experienced acute bronchiolitis caused by respiratory syncytial virus infection and required mechanical ventilation. She died because of respiratory failure.

3. Discussion

Infantile systemic hyalinosis is a rare autosomal recessive disorder with a distinctive phenotype caused by mutations in the *ANTRX2* gene.[1,6] Our patient manifested most characteristic signs of ISH and had a homozygous missense mutation of c.134T > C.[1] This mutation predicts an amino acid change of leucine to proline (p.L45P).[1,6] Certain clinical features of ISH, such as coarse facial features, gingival hyperplasia, and progressive contractures, may resemble features of lysosomal storage disorders such as Farber disease and mucolipidosis.[7] Infantile systemic hyalinosis can be differentiated from lysosomal storage disorders by the

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