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Hennekam Syndrome: A Case Report

Mariem Labrahimi*, Madiha Benhachem, Aziza Elouali, Ayad Ghanam, Maria Rkain, Abdeladim Babakhouya

Department of Pediatrics, Mohammed VI University Hospital, Faculty of Medicine and Pharmacy, University Mohammed Premier, Oujda, Morocco
Email: *mariem.labrahimi@gmail.com

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Abstract

Hennekam syndrome is a rare autosomal recessive disorder that was first described in 1989. It is characterized by congenital lymphoedema of the limbs, genitals, and face; intestinal lymphangiectasia; variable mental retardation; and typical craniofacial dysmorphism. The syndrome is genetically heterogeneous, which explains the variability of the clinical phenotype. We present a case report of a 16-month-old male infant who was born to consanguineous parents. The infant exhibited generalized lymphoedema, typical dysmorphic facial features, psychomotor development retardation, and respiratory distress due to upper airway obstruction caused by the absence of the lingual frenulum. To our knowledge, this anomaly has not been previously reported in the literature.

Subject Areas

Pediatrics

Keywords

Lymphangiectasia, Lymphoedema, Hennekam Syndrome

1. Introduction

In 1989, Hennekam *et al.* reported a new, rare syndrome that involves congenital lymphoedema, intestinal lymphangiectasia, facial dysmorphism, and mental retardation [1]. To date, 50 cases of this syndrome have been reported in the literature [2].

The characteristic signs of Hennekam syndrome are lymphangiectasia, lymph edema, facial anomalies, and mental retardation [3]. Malformations of lymphatic channels block the lymph circulation, and accumulation of fluids affects multiple

body parts including the face and limbs, as well as internal organs. Facial features are characterized by a flattened appearance of the face, a broad depressed nasal bridge, hypertelorism, epicanthal folds, a small mouth, and other anomalies [3].

This case report describes an infant who presented with the typical clinical manifestations of Hennekam syndrome in order to make clinicians more conscious of this rare disease.

2. Case Summary

A male infant aged 16 months, born to parents who are first-degree relatives. He was admitted for respiratory distress associated with edematous syndrome. The mother was 26 years old, and the father was 31 years old. The pregnancy was estimated to be at term, and the baby had a normal birth weight of 3 kg and a normal height and head circumference. There were no reports of lethargy during feeding, chronic vomiting, or seizures.

His particular family history included deaths among aunts, uncles, and cousins at young ages for unknown reasons. During the first month, the infant had generalized hypotonia and chronic snoring.

At the age of 6 weeks, he presented with generalized lymphoedema that started in the lower and upper limbs, followed by facial swelling and bilateral hydrocele. Shortness of breath was present along with these symptoms, particularly during inspiration.

During the clinical examination, the patient was found to have facial dysmorphism, characterized by a flat face, hypertelorism, a wide nasal bridge, small dysplastic ears, an ogival palate, microretrognathism, and bilateral clubfoot. These physical features had been present since birth, but they did not lead the parents to seek consultation.

He was hemodynamically stable, tachypneic, and showing signs of respiratory distress, including bilateral wheezing. Peripheral oxygen saturation was 95% in ambient air. He had neither a choanal atresia nor a cardiac murmur.

Examination of the oral cavity revealed hypoplasia of the lingual frenulum, which has resulted in the tongue being pushed back towards the soft palate. There was no hepatosplenomegaly or ascites, and the diuresis was normal. The weight was 4800 g.

The biological assessment indicated hypoalbuminemia at 22 g/l and hypoproteinemia at 38 g/l. The liver function tests, electrolyte panel, complete blood count, and 24-hour urine protein test were all normal. Nasopharyngoscopy showed compression of the lower portion of the trachea. The abdominal ultrasound showed a small peritoneal effusion and the skeletal X-ray revealed leg bone deformities, while the facial CT scan and thoracic angiogram were normal.

3. Discussion

Hennekam syndrome is a rare autosomal recessive disorder that was first described by Hennekam *et al.* (1989). It is characterized by the presence of lym-

phoedema, intestinal lymphangiectasia, an intellectual disability and dysmorphic facial features.

The facial features include a flat and broad forehead, a wide nasal bridge, and hypertelorism. Other dysmorphic features have also been described, such as bilateral epicanthus, dysplastic ears with a narrow ear canal, a small mouth, an ogival palate, gingival hypertrophy, and a smooth philtrum [3]. Most of these abnormalities were present in our patient, as shown in **Figure 1**.

The range of developmental disorders observed in patients varied, with some having normal psychomotor development, while others exhibited varying degrees of mental retardation. This variation was also observed within families. Generally, patients had limited intellectual ability or mild mental retardation [3].

Gastrointestinal loss of proteins and essential minerals such as calcium and magnesium may be the cause of growth delay and convulsions [3] [4].

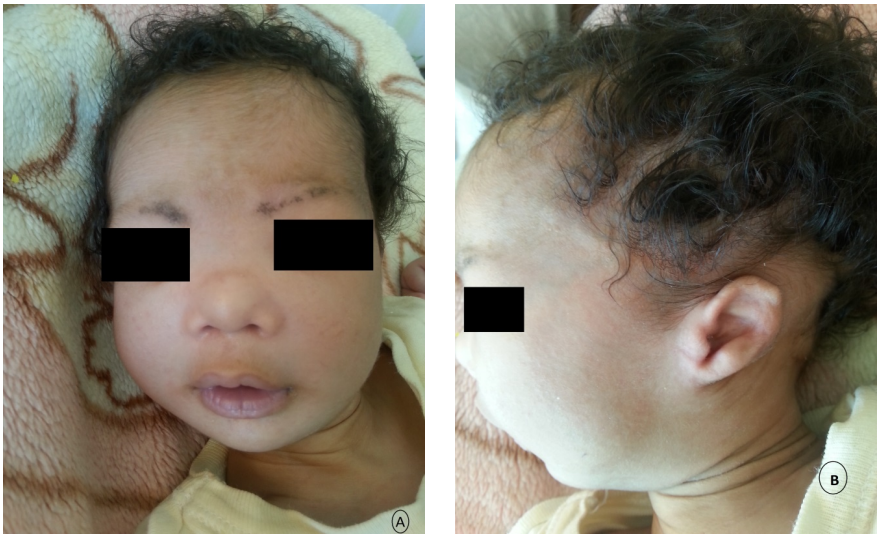

Figure 1. Dysmorphic face with flat midface, hypertelorism, and broad nasal bridge of front view (A) and side view (B).

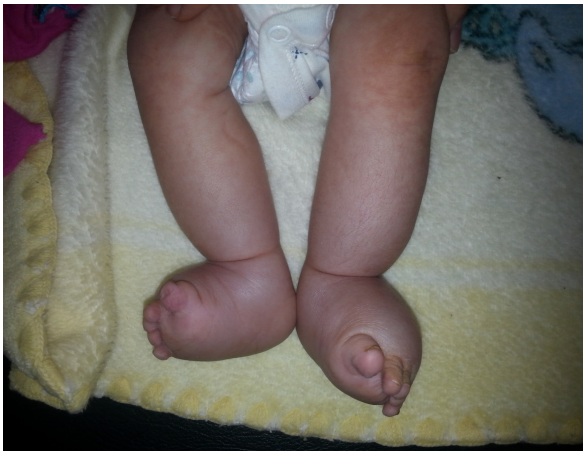

Figure 2. Bilateral lymphoedema of both legs and bilateral clubfoot.

Skeletal radiographs can detect bone deformities such as horizontal clavicles, scoliosis, spina bifida occulta (Cormier-Daire *et al.* 1995), hypoplastic iliac wings (Angle and Hersh 1997), and hypoplasia of the distal phalanges of the fingers and toes [1] [4]. In addition, to these abnormalities, our patient had bilateral clubfoot, as shown in **Figure 2**.

In summary, the medical team diagnosed the patient with Hennekam syndrome based on clinical phenotypic characteristics, including lymphoedema, bone deformities, and facial features. Despite the lack of a known curative treatment, complete decongestive therapy for lymphoedema has been shown to be beneficial.

4. Conclusion

Hennekam syndrome is a rare disorder, and its manifestations can be highly variable. The diagnosis of this syndrome is primarily based on clinical observations and the treatment will vary according to symptoms [3]. Our case report is unique in that it describes novel symptoms that have not previously been documented in the medical literature.

Conflicts of Interest

The authors declare no conflicts of interest.

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