Two Hennekam Syndrome Cases Presenting with Massive Pericardial Effusion and Hydrops

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Introduction
Hennekam syndrome is a rare autosomal recessive syndrome first described in 1989, characterized by congenital lymphedema of the limbs, genitalia, and face, intestinal lymphangiectasia, mild growth retardation, variable mental retardation, seizures and craniofacial dysmorphic features. We want to report two cases with Hennekam syndrome who were two months and 13 years old. First case was presented with hydrops and massive ascite in neonatal period and diagnosed when he was 2 months old. Second case was 13 years old girl applied with chest pain and massive pericardial effusion in her echocardiography. We aimed to review the literature in light of these 2 cases.

Case 1:
At birth the patient presented with severe respiratory distress due to non-immune hydrops. Generalized edema was seen especially prominent in his genitals and periorbital region. After resolution of the hydrops fetalis his facial features became apparent: flat face, prominent forehead, somewhat down slanted palpebral fissures, epicanthal folds, hypertelorism, a broad, depressed nasal bridge, a bulbous nasal tip, small mouth, low-set ears, micrognathia and high-arched palate (Figure 1). Bilateral pleural effusion was seen in his chest X-ray and massive ascite in abdominal ultrasonography. Repeated examination of fecal excretion of alpha-1 antitrypsin showed positive, suggesting intestinal lymphangiectasia. Repeated intravenous supplementation of albumin has given according to his albumin level. His symptoms get better with a special fat-free diet with added medium chain triglycerides and fat soluble vitamins but ascites continued.

Figure 1

Case 2:
13 years girl patient applied to emergency with chest pain, edema in her limb appearance in left side. Previous doctors told that lymphedema in her left side of the body could be due to allergy. She had used multiple ointments, attacks of diarrhea continued for one or two days but resolved spontaneously. Dysmorphic features were: small mouth, thin limb, low-set ears and long philtrum. There was left hemifacial, left sided upper and lower extremity edema (Figure 2). She had normal mental development. Massive pericardial effusion was seen in echocardiography.

Radionuclide lymphoscintigraphy scan showed abnormal drainage of the lower and upper (significantly left side) limb (Figure 3). Gastroduodenoscopy showed snowflake appearance of the duodenum and intestinal biopsy revealed lymphangiectasia (Figure 4). His symptoms improved with a special fat-free diet with added medium chain triglycerides and fat soluble vitamins.

Discussion
Hennekam syndrome is a rare autosomal recessive syndrome originally described by Hennekam et al. In 1989. Since the first report 31 patients have been described including two cases in the Turkey. Generalized maldevelopment of the lymphatic system is the main characteristic of the syndrome, preferentially affecting intestines, limbs, and genitalia, but it can also affect the pleura, pericardium, thyroid gland, and kidneys (Van Balkom IDC). Intestinal lymphangiectasia has been present in most (but not all) patients. As documented by intestinal biopsy, this is characterized by dilatation of the intestinal mucosal lymphatic channels in the lamina propria, with enlargement of the villi and thinning of the lymphatic vessel walls. Gastroduodenoscopy shows snowflake appearance of the duodenum. Intestinal biopsy revealed lymphangiectasia.
Lymphedema is usually apparent at birth or in early infancy and manifests both in the face or limbs. There can be an expressed asymmetry in involvement of the limbs. The lymphedema may be static for a prolonged period of time, but can also be progressive. Pleural lymph vessel anomalies have been reported in 8/28 cases, pericardial lymph vessel anomalies in 8/26 and ascites 16/29. In literature like our case 2, one boy with massive pericardial effusion was reported. Also another case with non-immune hydrops: generalized lymphangiectasia including pleural/pulmonary lymph vessels was reported.

Conclusion
Massive pericardial effusion and non-immune hydrops were rare clinical presentation of Hennekam syndrome. In addition to 31 cases in literature; these two cases cause of massive edema even as fetal hydrops were presented in order to take attention to this rare autosomal recessive disorder.