Case report

X-linked myotubular myopathy and chylothorax

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Abstract

X-linked myotubular myopathy usually presents at birth with hypotonia and respiratory distress. Phenotypic presentation, however, can be extreme variable. We report on a newborn baby, who presented with the severe form of the disease. In the second week of life, he developed a clinically relevant chylothorax, needing drainage and treatment with octreotide acetate. Pleural effusions are frequently described in patients with congenital myotonic dystrophy. To our knowledge, the association of chylothorax and X-linked myotubular myopathy has not been described to date. As chylothorax could not be attributed to any evident condition in this child, perhaps it may be added to the clinical spectrum of X-linked myotubular myopathy.

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1. Introduction

Congenital myopathies often present with hypotonia and respiratory distress from birth, although their expression may be delayed. In most cases muscle biopsy is warranted for definitive diagnosis. In some instances associated systemic features point at the final diagnosis. In congenital myotonic dystrophy, e.g., the mother will show mild facial weakness and clinical myotonia. Likewise, hydrops and/or pleural effusions in a newborn with severe hypotonia is suggestive of congenital myotonic dystrophy.

2. Case report

A male newborn baby of two non-consanguineous Turkish parents presented from birth with generalized hypotonia and respiratory distress, requiring artificial ventilation. On day 11, 5 days after introduction of gavage feeding, he developed bilateral pleural effusions, requiring drainage (Fig. 1). Laboratory examination was compatible with chylothorax (5230 white blood cells/μl, 98% lymphocytes; chylomicrons were present; triglycerides 746 mg/dl). There were no central venous catheters in place who could have caused thrombosis, impairing lymphatic flow, neither could any other risk factor for chylothorax be identified. Because interruption of enteral feeding failed to control the chylothorax with daily losses approximating 400 ml, treatment with octreotide acetate, a long-acting synthetic analog of somatostatin, was instituted [1]. Chylothorax gradually resolved and enteral feeding with medium-chain triglyceride enriched formula was introduced without problems. Muscle biopsy disclosed typical features of X-linked myotubular myopathy (Fig. 2). The MTM1 gene was sequenced and revealed a C.1261-10 A>G mutation, confirming the diagnosis [2]. The baby died at home with palliative care at the age of 4 months. Autopsy was not performed.

3. Discussion

To our knowledge, this is the first report on a neonate with X-linked myotubular myopathy who...
developed a clinically relevant chylothorax. When not a postoperative complication, chylothorax probably results from a malformation of the thoracic duct, although in many cases no apparent abnormality can be found. Searching the literature for association between chylothorax and congenital myopathies, we found a case report that was very similar to ours. Chylothorax was described in a boy of healthy Turkish parents, who presented with lack of spontaneous movements at birth and who died at the age of 9 weeks [3]. Muscle biopsy confirmed congenital actin myopathy together with nemaline bodies and intranuclear rods. In that baby a mutation of the \(ACTA1\) gene in exon 4 was found. For the rest pleural effusions with or without hydrops are frequently described in cases of congenital myotonic dystrophy [4,5]. However, expansion of CTG repeats was excluded in our patient. An X-linked recessive transmission of chylothorax has been suggested [6]. As no other evident condition linked to chylothorax could be found, perhaps chylothorax may be classified under one of the systemic features of X-linked myotubular myopathy, as it is the case for pleural effusions and congenital myotonic dystrophy.

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**References**