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Case report (Pages: 15495-15498)

Syndromic Congenital Chylothorax – a 7q21.13q31.31 Duplication

* Raquel Monteiro Costa [1], Ana Isabel Martins [2], Joaquim Sá [3], Ana Rodrigues Silva [4], Raquel Henriques [4]

[1] Pediatrics Resident, Pediatrics Department - Centro Hospitalar Tondela-Viseu, Viseu, Portugal.

[2] Pediatrics Resident, Pediatrics Hospital – Centro Hospitalar e Universitário de Coimbra, Coimbra, Portugal.

[3] Genetic, Genetics Department – Centro Hospitalar e Universitário de Coimbra, Coimbra, Portugal.

[4] Neonatology, Neonatology Department – Centro Hospitalar e Universitário de Coimbra, Coimbra, Portugal.

Abstract

Congenital chylothorax is a rare cause of respiratory distress in the newborns. It has a high mortality rate and its prognosis depends on the time of the diagnosis, etiology and therapy.

The chromosomal gain, duplication of 28 Mb, including more than 200 genes, in the long arm of chromosome 7 (seq [GRCh37] 7q21.13q31.31, chr7:g.89783721_117877082dup) is very rare and is established as the likely etiology in this clinical case.

Phenotypic reports of chromosomal imbalances are an important source for genetic counseling.

Key Words: Chromosomal gain, Congenital chylothorax, Genetic syndrome.

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**** Corresponding Author:***

Raquel Monteiro Costa, Pediatrics Resident, Pediatrics Department - Centro Hospitalar Tondela-Viseu, Viseu, Portugal. Email: raquelmonteiro91@hotmail.com

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1- INTRODUCTION

Congenital chylothorax is defined as the accumulation of lymph in the pleural space (1, 2). It is a rare cause of respiratory distress in the newborns. This pathology usually presents before birth (1, 2). In few cases, it is detected after birth with a chest radiography and it is confirmed by the biochemical examination of the pleural fluid (2). Its etiology is mostly unknown. It may result from structural changes in the lymphatic system, trauma or genetic / chromosomal changes (3, 4). First-line treatment is the thoracocentesis and hypolipidemic diet with medium chain triglycerides (3, 4). And high mortality rates are described (4).

Here, a case of a newborn with a congenital chylothorax, diagnosed postnatally, with a rare genetic etiology is presented.

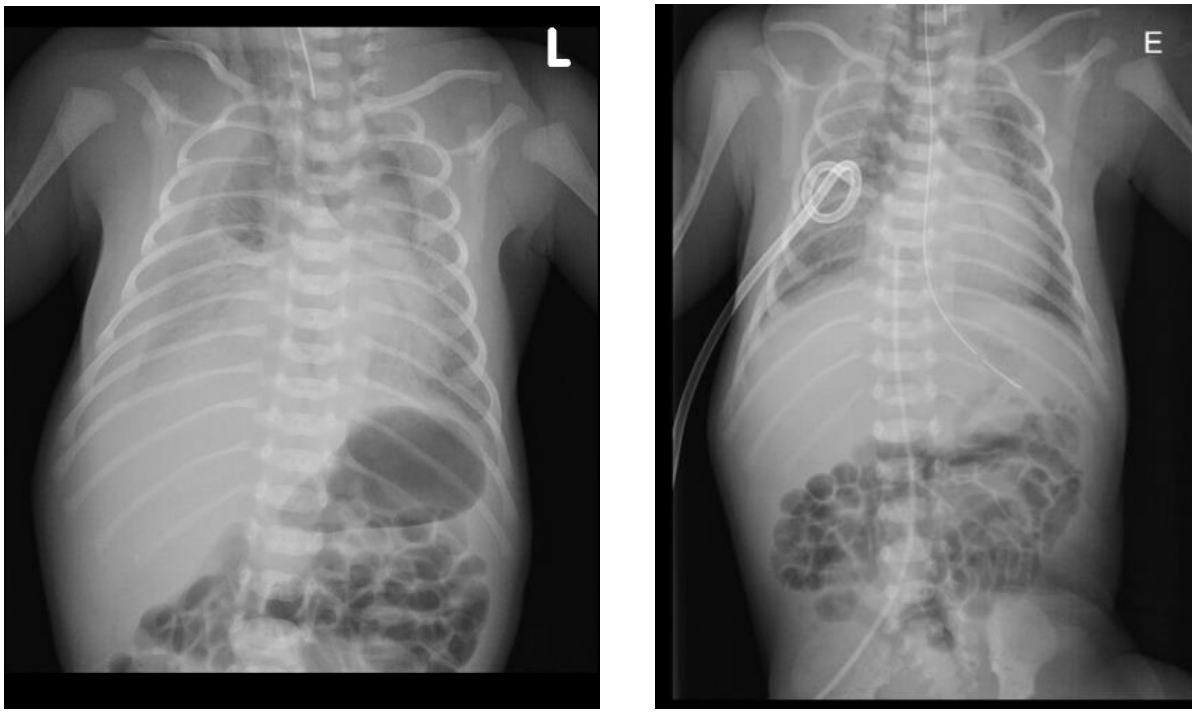
2- CASE REPORT

We report the case of a first newborn from healthy and non-consanguineous parents. The family history was negative for genetic disorders. The pregnancy was uneventful up to 38 weeks, when a moderate right pleural effusion with mediastinal deviation was diagnosed by prenatal ultrasound. The Delivery was done by a cesarean section at 38 weeks with a weight of 2670 g, a length of 50 cm and a head circumference of 35 cm. The APGAR Scores were 6/9/10 (resuscitation with a positive pressure cycle and oxygen therapy). The patient persisted with respiratory distress motivating mechanical ventilation. Thoracentesis was performed with drainage of citrus fluid with the following characteristics: pH alkaline 7.65, leukocytes 8020/uL with 100% mononuclear cells, protein 2.1 g/dL and lactic dehydrogenase 174 U/L, gram stain showed no bacteria and the culture was sterile. Its study was compatible with chylothorax. Extubation occurred after 24 hours and the newborn was placed under non-invasive ventilation. A hypolipidemic diet was instituted with medium chain triglycerides. At 12 days of life, the chest tube was removed without further reappearance of pleural effusion. During hospitalization, serial echocardiographic assessments showed patent ductus arteriosus, requiring diuretic therapy. Medical therapy was started (ibuprofen and paracetamol) without success. He was then proposed for percutaneous correction. He was also evaluated by the Genetic Medical Team due to some dysmorphisms – retrognathia, transverse palmar crease, low-set thumb, sandal gap, long fingers – hypotonia and hyperlaxity. The whole exome sequencing found a likely pathogenic variant, a duplication of 28 Mb which includes more than 200 genes, in the long arm of chromosome 7: seq [GRCh37] 7q21.13q31.31, chr7:g.89783721_117877082dup. Before that, a next-generation sequencing (NGS) panel for Rasopathy principal genes was performed which was negative.

At one month of age, he was transferred to a pediatric service for multidisciplinary care (Genetics, Pediatric Pneumology, Pediatric Cardiology, Pediatric Palliative Care, Pediatric Neurodevelopment and Physical Medicine and Rehabilitation). The patent ductus arteriosus was closed percutaneously and diuretic therapy was suspended. To evaluate the risk for future pregnancies, the parents' chromosome analysis was performed and revealed a normal karyotype. The child was discharged under non-invasive ventilation during sleep due to hypercapnic respiratory failure, maintaining multidisciplinary and pediatric care follow-up.

3- DISCUSSION

Congenital chylothorax, although rare, is the most common form of pleural effusion in the neonatal period (1). Its prognosis depends on the time of the diagnosis, etiology and therapy (2, 4).



(a) **(b)**

Fig. 1: Chest radiograph before (a) and after (b) thoracentesis.

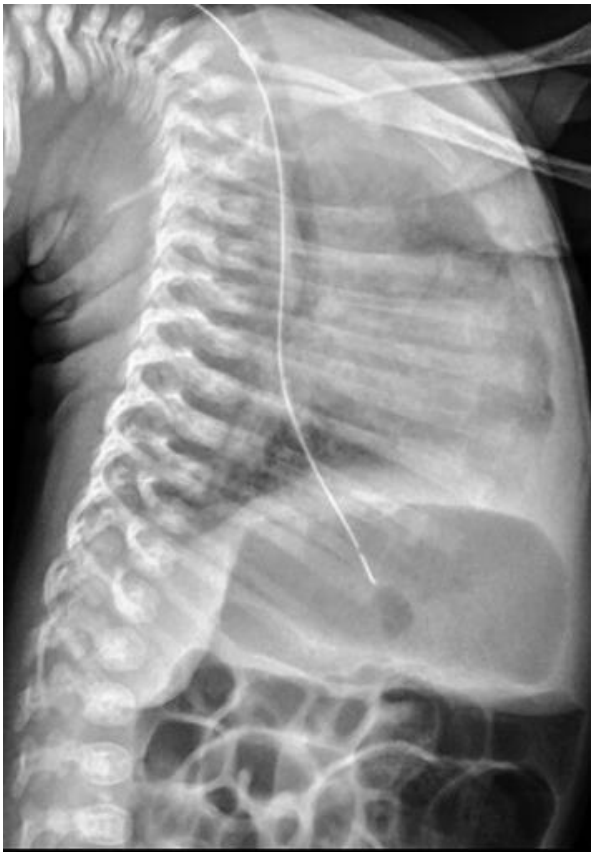


Fig. 2: Chest radiograph showing hypotonia and hyperlaxity

The vast number of genes establish this gain as pathogenic. This gain is very rare, there are only two other patients reported, a 3-year-old male child patient with severe neurodevelopmental delay, craniofacial dysmorphisms and pulmonary stenosis (5) and an 8-year-old boy with marked developmental retardation, facial dysmorphism, short stature, strabismus and hyperextensible metacarpophalangeal joints (6).

Our patient showed complex congenital anomalies consisting of minor nonspecific craniofacial features and heart and skeletal impairment as well as chylothorax.

In this case report, the detected chromosomal gain is established as the likely etiology and the overall prognosis is dependent on this etiology.

4- CONCLUSION

Phenotypic reports of chromosomal imbalances are an important source for genetic counseling especially to the parents of newborns. The genetic study, although not essential for the initial approach and stabilization, should be performed as soon as possible because of its importance in the prognosis.

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