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SCIENTIFIC LETTER

Congenital fiber-type disproportion myopathy: A case study

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Abstract Congenital fiber-type disproportion myopathy causes impaired muscle maturation or development. It is characterized by moderate to severe hypotonia and generalized muscle weakness at birth or during the first year of life, especially in the lower extremities. It is inherited as an autosomal recessive, dominant and X-linked. It is diagnosed by clinical data confirmation, generalized hypotonia and a muscle biopsy in which muscle fibers type I are smaller in caliber, 12% smaller than those of type II and type I fibers are more common than type II. Treatment is multidisciplinary.

The following describes the case of a patient who was born in the ''Dr. José Eleuterio González'' University Hospital in Monterrey, N.L, who presented clinical and muscle biopsy compatible with this myopathy.

Introduction

Within congenital myopathies classifications, there are myopathies with alterations in muscle maturation and/or development. One of these myopathies is congenital fiber-type disproportion, which is characterized by generalized muscle weakness at birth or during the baby's first year, mainly in the extremities.[1,2]

It was first described by Brooke and Engel[1,3] in a study of the morphology of children's biopsies. It is a rare

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entity, which occurs in 1 in every 50,000 live births.[4] All body musculature is affected. However, the muscles in the lower extremities are usually more altered than the upper extremities; thus the electromyography reports a myopathic process, although in others there is a neurogenic component.[11,13] Some cases with a longer evolution time show mild sclerosis[5] and there have been reports of important respiratory alterations, severe brain damage and heart diseases.[1,7,8] In a few cases an association of skeletal and articular alterations has been observed.[9,10]

Clinical case

The mother of the baby boy was a 25-year-old woman, previously healthy, secondary school completed, without drug addictions, living together with her partner. The father was a 30-year-old man, healthy, without any relevant history. The newborn had two other brothers, a 4-year-old and a 7-year-old, both healthy.

The product of a fourth pregnancy, the mother had had two C-sections performed and suffered a miscarriage when she was 9 weeks pregnant. She completed a normal pregnancy, with folic acid, iron intake, as well as supplementary multivitamins for the first month of pregnancy, prenatal control with 8 visits to the University Hospital without presenting abnormalities.

Born by elective C-section due to the history of two previous C-sections at 38.4 weeks of gestation, with an Apgar score of 2/6 because it was found to be acrocyanotic, flaccid, no respiratory effort, and with a heart rate of 80 bpm. He is given 2 positive pressure ventilation cycles, improving hear rate, but not respiratory effort, and was therefore intubated and left under mechanical ventilation and moved to the Neonatal Intensive Care Unit.

During physical examination: Weight: 3070 g, located in the 50th percentile, size: 50 cm, in the 50th percentile, cephalic perimeter: (CP) 36 cm, in the 50th percentile, myopathic facies with an elongated face, symmetrical eyes and a bilateral ptosis, presenting red reflex, permeable nostrils, proper ear implantation, an ogival palate and a tented upper lip. Neck: retrognathial, short, round. Thorax: no respiratory effort, without deformities, normal lung fields, holosystolic murmur grade 3–4 detected. The abdomen was soft, palpable without pain, and free of visceromegaly, an umbilical cord, 2 arteries and a vein. Male genitalia: Tanner 1, right cryptorchidie, with the right testicle in the inguinal canal and the left testicle in the scrotal bag. The extremities: generalized hypotonia, no response to painful stimulus, absence of muscle tone, deep tendon reflexes absent, rigid right clubfoot.

Upon admission, gasometry and asphyctic profile on admission and at 24 h (due to perinatal asphyxia suspicion) were normal and a transfontanellar ultrasound (TFUS) and brain MRI were performed and were also normal (Fig. 1). No infection-compatible alterations were found, as well as the metabolic panel (with electrolytes) and the metabolic screening were also normal. An electrocardiogram was performed due to the presence of ICT in 0.62 (Fig. 2) and due to the holosystolic murmur grade II/VI detecting an interventricular communication of 2 mm, mild tricuspid insufficiency,

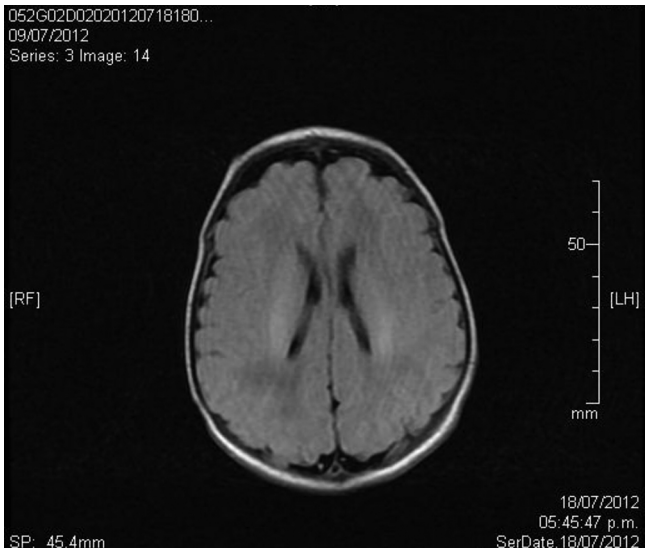

Figure 1 Normal MRI.

a persistence of a patent ductus arteriosus of 1.4 mm and an oval foramen without hemodynamic repercussions.

We referred the patient to the Genetics Department who evaluated the patient's genealogical tree, without finding any history of neuromuscular diseases in any of the first-degree relatives or extended family, thus classifying this as an isolated case. Furthermore, they requested a genetic mutations study; however, the family did not have the economic resources to have it done.

Due to the newborn's hypotonic diagnosis, a muscle biopsy, lab tests and normal imagining of the right quadriceps were performed, confirming the congenital fiber-type disproportion diagnosis.

The sample was divided into two parts; the first part was cut by freezing it and processed following standard

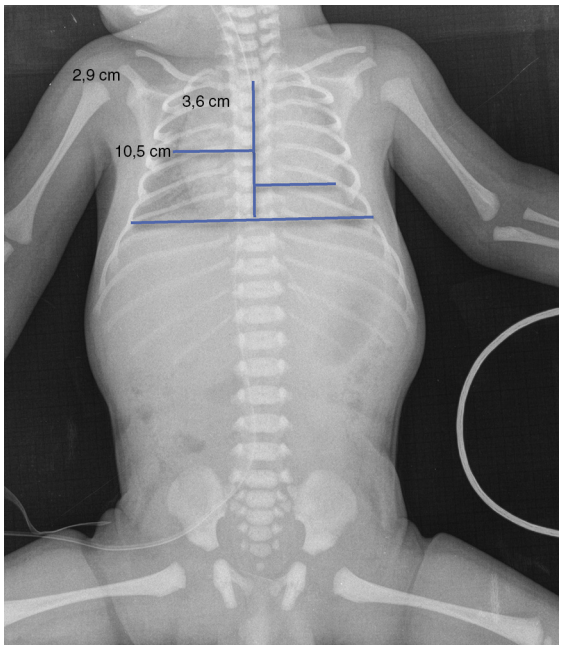

Figure 2 Cardiothoracic index at 0.62.