

De barsy syndrome: orthopedic case report and literature review

Volume 7 Issue 5 - 2017

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Introduction

This condition was first described in 1968 by De Barsy who reported a case of a patient with progeria, dwarfism, oligofrenia and degeneration of the elastic tissue of the cornea and skin, and since then, it is known as Barsy Syndrome or Barsy-Moens-Diereckx Syndrome. This is defined as the combination of progeroid aspect, cutis laxa, corneal opacity, intrauterine growth retardation and severe mental retardation (although some will learn to speak, intelligence is less than normal).¹ The orthopedic manifestations are dysplasia of hip development, hyper laxity of severe joints, athetoid movements, scoliosis and severe deformities of the foot. Epidemiology in Latin America is unknown because of its underdiagnosis and when confused with other connective tissue pathologies (Hutchinson-Gilford syndrome, gerodermic osteodiplasia, even Ehlers-Danlos), the life expectancy of these patients varies according to the degree of penetrance and in the world literature, there are very few reports (about 50), so the diagnosis requires a challenge.²

Clinical case

OGJA, male patient of 5 years and 6 months sent for first-time orthopedic evaluation from a telethon child rehabilitation center in the city of Irapuato, Mexico due to the antecedent of psychomotor retardation, asymmetry of the pelvic extremities and inability to stand and walk. It does not have a hereditary family history of importance, mother at the time of pregnancy 30 years of age, second pregnancy, no drug addiction during pregnancy, history of intake of folic acid, iron and vitamins, pregnancy at term 40 weeks gestation, Natural birth, weight 3000 gr, size 51 cm, do not report hypoxia at birth (does not remember APGAR scale), both graduates of the hospital at 24 hours. During development and birth it negates hyperbilirubinemia; Cephalic support at 1 year 5 months, sedestation 1 year and 8 months, currently does not achieve standing, ambulation not achieved, sporadic babbling, history of previous hospitalizations for bilateral congenital cataract surgery operated at 4 months of age, history of pneumonia Repetition, has been diagnosed with Barsy syndrome since the beginning of 2015 (Figure 1).



Figure 1 Clinical photo with covered face.

Physical examination:

Athetoid movements, Macrocephaly, cephalic support, adequate hair implantation, asymmetric eyes (hypertelorism), eruptive asymmetry of the upper and lower canines, cylindrical mobile neck, short, chest thorax, with limitation to the amplexation of the Respiratory movements, unstructured kyphosis, normal cervical lordosis, no spinal dysrhaphy, soft abdomen, hepatic dullness 5 cm below the costal border, 2 cm umbilical hernia not protruded. Without ambulation, Thoracic integral extremities, normal glenohumeral joint, elbow with hyperextension, hand with integral fingers, thumb in proper position, can perform fine and thick clamp. Asymmetric pelvic extremities with bilateral proximal femoral angular deformity, barlow and ortolani negative bilateral positive piston, bilateral positive yawning sign, positive drainage test for anterior and posterior of both knees, and ankle without alterations, preserved distal mobility. Brighton criteria of 8 points (touching the ground with the palms was not possible) (Figures 2 & 3).



Figure 2 AP clinic.



Figure 3 Lateral clinical image.

Radiographic findings

AP of proximal right humerus: Thinning of the humerus cortices, solution of bone continuity at the diaphyseal level transverse fracture in the process of consolidation.

Left forearm AP: Thinning of cortical radius and ulna, presence of bone, simple, transverse solution in ulnar diaphysis in consolidation process.

Pelvis AP projection: Cup shape, bilateral femoro-acetabular congruence, femoral diaphysis with angular deformity in varus. Femur cortical thinning is observed.

Knee AP: Joint congruence, with deformity in varus of right tibial shaft.

AP and lateral column: Rarefaction of the growth plates of the thoraco-lumbar vertebrae.

In all radiographic projections, metaphyseal rarefaction, proximal femur, distal, proximal tibia, as well as proximal and distal humerus, radius and cubit are observed.

Pulmonary perfusion scan: low uptake of right lower lobe and bilateral apices (Figures 4 & 5).



Figure 4



Figure 5

Laboratories

Normal red formula, normal white formula, normal 5-element blood chemistry, normal proteins, ESR 18, sodium 140, potassium

4.4, chlorine 117, phosphorus 2.7 Calcium 8.9, Magnesium 1.6. Antibodies mitochondrial 1:20 positive, normal complement tests.

Treatment

The treatment indicated for this patient was of non-surgical treatment, it was decided based on the non-orthopedic priorities that presented at the moment of its evaluation that are the heart disease and pneumopathy in study; Parents were advised of the need to continue rehabilitation therapy at their referral center with exercises consisting of progressive muscle strengthening, this in order to improve the hyper elasticity in the pelvic extremities, the patient despite the rehabilitation has not developed Seated or complete cephalic support in spite of the age, so that at the moment, performing some surgical procedure aimed at correcting the femoral deformity is currently discarded (Figures 6 & 7).

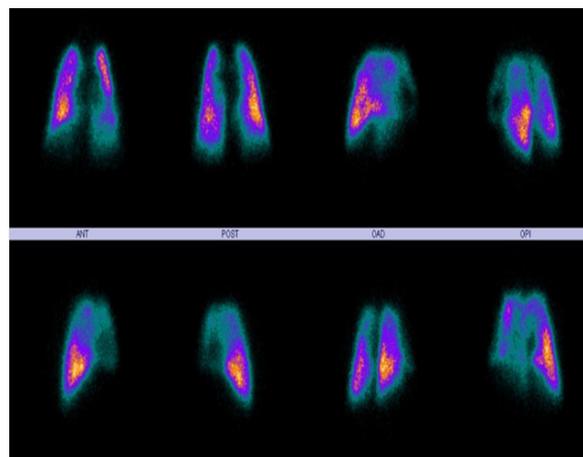


Figure 6 Pulmonary scintigraphy.



Figure 7 Extremidades pelvicas AP.

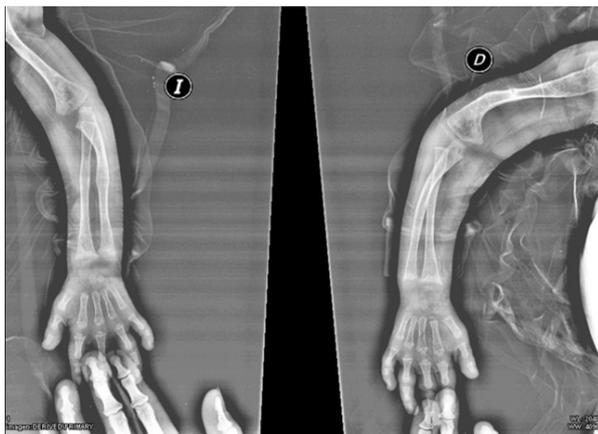


Figure 8 RX of thoracic limbs.



Figure 9 Heart rate cardiac.

Discussion

Cutis Laxa syndrome (CL) is a group of extremely rare diseases that affect the connective tissue, do not show predisposition by sex, and whose incidence is unknown since there is a subdiagnosis of this disease when mistakenly diagnosed with other connective tissue disorders.¹

Classification³

- A. Autosomal dominant Cutis laxa (ADCL) with symptoms that arise at any age and whose symptoms are variable, from cutis laxa to facial features with nasal bridge affection, vascular symptoms (aortic aneurysms), pulmonary (pulmonary emphysema); In part due to mutation of the gene elastin (ELN), fibulin-5 gene (FBLN5).
- B. Cutis Laxa Autosomic recessive (ARCL) which is divided into several subtypes according to the organic condition:
 - i. 1A: Linked to FBLN5 (fibulin 5): Loose skin, hernias and lung condition.
 - ii. 1B: Linked to FBLN4 (fibulin 4): Loose skin, aneurysms, aortic stenosis, and orthopedic condition (hyper elasticity, bone fragility).
 - iii. 1C: Linked to LTBP4: Lax skin, severe urinary problems (Urban-Rifkin-Davis syndrome).

- iv. 2A: Linked to ATP6Vv0A2 Cutis laxa that improves with age, severe psychomotor retardation, hip dislocation, hernia and myopia.
- v. 2B: Linked to PYCR 1: Cutis laxa, progeria, delayed growth, retarded psychomotor development, hyper articular laxity, triangular face.
- vi. 3: Barsity's syndrome, conditions a lax cutis with growth retardation, moderate-severe mental retardation, cataracts and hyper articular laxity, progeria, may present cardiovascular or pulmonary affection.

Alterations have been observed in both the number and size of the elastic fibers in the dermis and connective tissue.⁴ However elastin appears to be the only component affected in Barsity's syndrome, this has been demonstrated in tests for determination of elastogenesis and degradation of the same by immunocytochemistry found low levels of m-RNA suggesting that some elastin gene is Involved.⁵

Since the initial description of the De Barsity syndrome, cases have been described in the literature with different phenotypic alterations. The progeroid aspect predominates due to hypoplasia of the dermis in 100%; there is no predisposition for race, but if by gender (mainly men), there is also a broad forehead, dysplasia of auricular pavilions, impaired hair implantation, small mouth, teething alterations. 96% of patients will have postnatal growth retardation, 34% of patients will present umbilical hernia, psychomotor developmental delay in 76% of patients that can become severe in 48% of patients; Congenital cataract in 48% of the cases as well as myopia, divergent strabismus and blue sclera in various proportions.^{2,4,5}

The management of patients with De Barsity syndrome is multidisciplinary, genetic, ophthalmology, neurology; urology, cardiology, gastroenterology and orthopedics must be informed about the disease and its life expectancy (which is reported as a variable with mortality in the second decade Due to recurrent infections) and prioritize treatments that improve the patient's quality of life.⁶

There is little evidence on the orthopedic treatment of this syndrome, Robert Stanton in 1994 reported the case of two siblings with De Barsity syndrome and reported the main orthopedic findings: bilateral hip development dysplasia which was treated with open reduction, femoral osteotomy, and posterior immobilization, Scoliosis, carpal dislocations, which were treated by occupational therapy, as well as congenital vertical talus, which was treated with subtalar arthrodesis and soft tissue management; However reported complications due to hyperelasticity, which, because of this, required a prolonged time of post-operative immobilization in the management of hip dislocation. Of both cases reported, both with delayed psychomotor development required special education and rehabilitation, both of which exceeded adolescence.⁷

The anesthetic considerations that have been reported are from postoperative tachycardia and hyperthermia during the surgical procedure.⁸ Due to the lack of information should be personalized to the patient according to the clinical manifestations present, as well as the non-orthopedic priorities of the same, it would be worth remembering the surgical risk involved in performing an orthopedic surgical treatment with a view In angular corrections, guided growth, osteosynthesis and / or correction of fractures due to bone fragility. There are also no reports on the preventive management of fractures and dysplasia of hip development. Due to the few reports of cases of this syndrome, the natural history is unknown.

Acknowledgments

None.

Conflicts of interest

None.

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