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CLEIDOCRANIAL DYSPLASIA: A FAMILY REPORT

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ABSTRACT

Cleidocranial dysplasia is a rare autosomal dominant genetic bone disease. Its clinical expression can be surprisingly variable. Clavicular ossification anomalies, facial, pelvic and dental anomalies are the most common and can be considered as major signs. The incomplete penetrance of the disease and its good tolerance explain the frequency of discrete clinical forms of expression or undiagnosed ones. Through observation of a new Tunisian family, we will recall the clinical and laboratory characteristics of this disease.

KEYWORDS: Cleidocranial dysplasia, autosomal dominant transmission, CBFA1, RUNX2.

INTRODUCTION

The cleidocranial dysplasia (CCD), also called cleidocranial dysostosis, was described for the first time by Martin in 1765 and named as such by Pierre Marie and Sainton in 1897.[1] (CCD) is a dominantly inherited autosomal bone disease whose clinical manifestations can vary widely, explaining the diversity of circumstances of its discovery.

We report the case of a Tunisian family (father and two children) carrying CCD.

OBSERVATIONS

W. infant, from a non consanguineous marriage, is hospitalized twice in pediatrics at the age of six months for an acute respiratory failure requiring mechanical ventilation for seven days, adrenaline aerosol and corticosteroids with good evolution and at the age of three for severe laryngitis. Physical examination of the infant showed wide anterior and posterior fontanels with disjunction of the sagittal and coronal sutures. The thyroid function tests, trans-fontanellar ultrasound and karyotype were normal. The radiological assessment showed a complete disjunction of the sagittal, coronal and parietal-occipital sutures. (Fig.1), persistant wormian bone (fig1), slender ribs (fig2), absence of femoral nuclei (fig3) and a lack of ossification of the ischiopubic branches. (fig3). The endoscopic examination reveals a laryngomalacia.

Y., W's sister is 2 years old; she presented with large anterior and posterior fontanels with facial dysmorphia and flattening of the facial bones (fig4). The trans-fontanellar ultrasound, thyroid balance sheet and the karyotype were normal.

Mr. M., the father of Y and W, had a small size (1.55 m), macrocrania, facial dysmorphia with flat face bones, exaggerated frontal bossing and anarchic teeth implantation (fig5 and 6). He also had a bulging chest, kyphoscoliosis (fig7), coxa vara and bilateral clavicular hypoplasia (fig8 and 9).

The specific phenotype of this man, as found in two children, led to the diagnosis of dysplasia cleidocranial.

Legend of the figures

Figure 1: X-ray of the skull: total disjunction of the sagittal suture, coronal and parietal-occipital bone and persistence of wormiens.
Figure 2: lateral chest radiograph: slender ribs.
Figure 3: pelvic radiograph: absence of nuclei and femoral defect of ossification of ischiopubic branches.
Figure 4: facial dysmorphia Y. sister
Figure 5: macrocrania + chest puffing
Figure 6: Skull radiography: flattening of the facial bones + anarchic implantation of teeth
Figure 7: Kyphoscoliosis.
Figure 8: Hypoplastic clavicles;

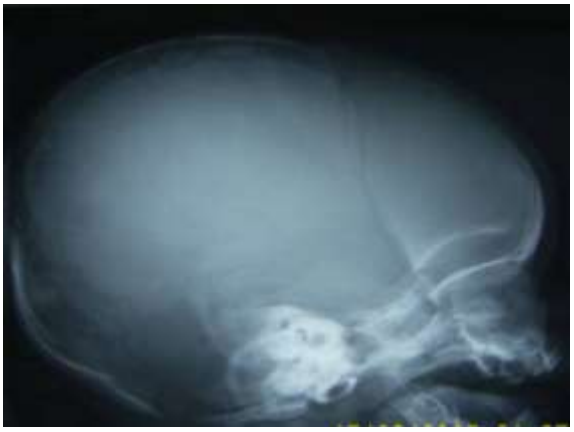

Figure 1: X-ray of the skull: total disjunction of the sagittal suture, coronal and parietal-occipital bone and persistence of wormiens.

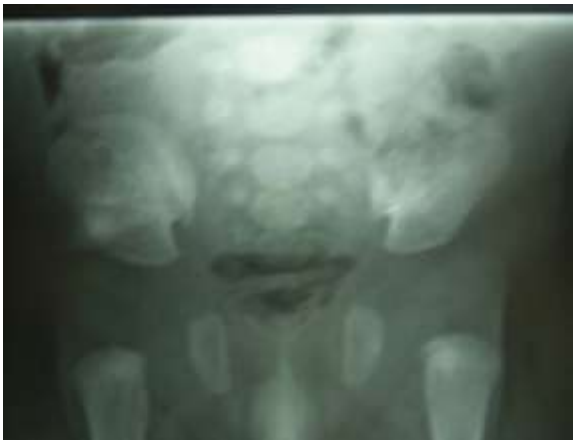

Figure 3: pelvic radiograph: absence of nuclei and femoral defect of ossification of ischiopubic branches.

Figure 2: lateral chest radiograph: slender ribs.

Figure 4: facial dysmorphia Y. sister

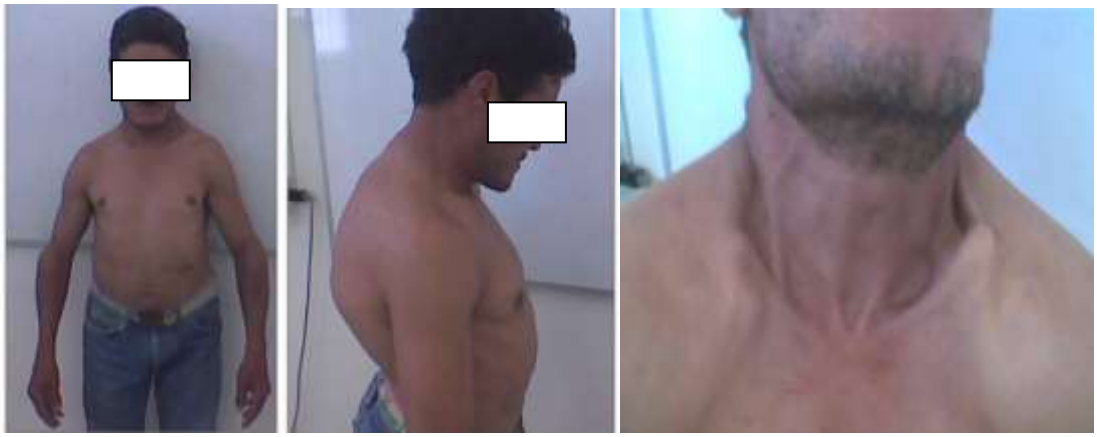

Figure 5: macrocrania + chest puffing

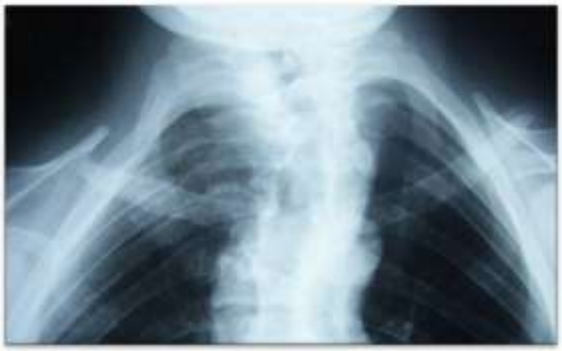

Figure 6: Skull radiography: flattening of the facial bones + anarchic implantation of teeth

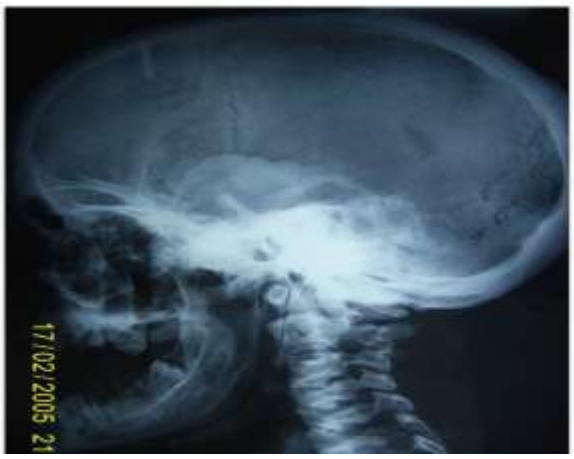

Figure 8: Hypoplastic clavicles.

DISCUSSION

The cleido-cranial dysplasia is a constitutional disease that primarily affects the bones with membranous ossification.[1,2] Its transmission is autosomal dominant with variable penetrance explaining the diversity of clinical forms, some of which are undiagnosed.[2] It results from a mutation of the transcription factor Cbfa1 and RUNX2 which plays a role in the differentiation of osteoblasts.[3,4,5] The persistence of fontanelles, clavicular hypoplasia, facial, dental anomalies and pelvis are the major signs of the disease; other locations are possible such as the spine (spina bifida), ribs, sternum, and members.[5,6] There is also a small final size.[7] A radiographic assessment allows the assessment that if clinical suspicion remains but confirmation genetics. Specific treatment is indicated only in case of orthopedic and orthodentique complications.[7] The prognosis of this disease is usually favorable but sometimes life-threatening can be engaged mainly at the young age when dysplasia combines laryngo tracheomalacia or.[6.7]

CONCLUSION

The incomplete penetrance of the genetic disease and good tolerance explain the frequency of discrete clinical forms of expression or undiagnosed.

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