Abnormal Dentition in an 8 Years Old Female Child

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Patient with shortening distal phalanx 1st finger both hands, from the birth and familiar precedents of similar abnormality. At the age of 8 she presented dental alterations with hipertrofia gingival and dental incorporations, and backaches and pain in femoral region. Radiology: increase of bony widespread density, hypoplasia lower jaw, and acrosteolisis distal phalanxes. The jaw biopsy (dental piece and alveolar surrounding bone): bony fragments are constituted for coarse and irregular trabeculas with importantly bony resorption and newly formed. Bone densitometry: T +5.2 in column lumbar and neck femoral. Gamma scan bone: diffuse captation of the axial skeleton. Diagnosis: picnodisostosis.

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Clinical Case

We present the case of a 32-year-old woman, who at the age of 8 presented diffuse bone pain in the femoral an lumbar spine area with marked dental alterations, a delay in deciduous and definite dentition, gingival hyperplasia and dental inclusions on the maxillary and jaw bones (Figure 1), requiring a gingivectomy at the age of 11. Her development otherwise had been normal (155 cm, 58 kg), she had a female karyotype 46XX. She presented shortening of all of the fingers on both hands, which was more evident on the first finger (Figure 2A), due to shortening of the distal phalanges (Figure 2B), and nail anomalies characterized by clubbing, hyperpigmentation and parched striae. The patient had required several maxillofacial surgeries up until the age of 18 and is currently asymptomatic. She did not have any history of interest. Her paternal grandmother and her 5 brothers had hypoplasia of the

Figure 1. Orthopantomography: hypoplasia of the jaw. The left half of the jaw is radiolucent from the premolars to the second left molar, and in the region of the second right molar there are multiple radiopaque inclusions.

Figure 2. Orthopantomography of the hands. Shortening of the phalanges:

Dentición anómala en niña de 8 años

Presentamos el caso de una paciente con acortamiento de la falange distal del primer dedo de ambas manos desde el nacimiento y con antecedentes familiares de similares anomalías. A los 8 años presentó alteraciones dentarias con hipertrofia gingival e inclusiones dentarias, y dolores lumbar y en la región femoral. Las pruebas radiológicas evidenciaron aumento de densidad ósea generalizada, hipoplasia maxilar inferior y acrosteolisis de las falanges distales. Biopsia mandibular (pieza dentaria y hueso alveolar circundante): los fragmentos óseos están constituidos por trabéculas toscas e irregulares con importante resorción y neoformación ósea. Densitometría ósea: T +5,2 en la columna lumbar y el cuello femoral. Gammaografía ósea: captación difusa del esqueleto axial. Diagnóstico: picnodisostosis.

Introduction

Picnodystosis is a rare bone dysplasia that is transmitted in an autosomal recessive manner. It is due to a mutation in the 21, 1q chromosome, which inactivates cathepsin K, which plays an important role in the degradation of the bone matrix. The disease is characterized by a marked osteosclerosis in all of the axial and peripheral skeleton. The clinical characteristics and the radiologic alterations allow for a diagnosis.

This disease very likely affected the French painter Henri Toulouse-Lautrec (1864-1901).
distal phalanges of the hands and/or similar nail abnormalities than the patient; the family had a history of inbreeding.

The image tests observed a generalized increase in bone density with more trabecular and cortical bone land homogeneous osteosclerosis (Figure 3). Hypoplasia of the jaw (Figure 1) with a radiolucid left half from the premolars to the second left molar and multiple inclusions. There was also acroosteolysis and hypoplasia of the distal tip of the distal phalanges (Figures 2A and B).

**Laboratory Analysis**

Hemogram: 3 series of normal characteristics. Serum chemistry: calcium, 9.5 mg/dL; phosphorus, 2.8 mg/dL; alkaline phosphatase (AP) of up to 1477 U/L at age 14, and still elevated (AP, 393 U/L). Twenty-four-hour urine: urine calcium, 178 mg/24 h; urine phosphate, 603 mg/24 h; tubular phosphate reabsorption (TPR), 87%; hydroxiproline, 65 (interval, 5-40) mg/24 h; hydroxiproline/creatinine, 43 (5-40) mg/1 g creatinine; desoxipyridinolines/creatinine, 80 (3-18) nmol/1 mmol creatinine; erythrocye sedimentation rate (ESR), 6 mm/h; CRP <3.9 mg/dL; 25-OH-vitamin D, 66.7 (6-98) ng/mL; parathyroid hormone (PTH), 35 pg/mL.

**Pathology**

Jaw biopsy (tooth and surrounding alveolar bone): the bone fragments are constituted by gross and irregular trabeculae with an important resorption and neoformation of bone; with polarized light microscopy there is an evident absent of the brush-shaped border. In the interior of the wider trabeculae there are apposition lines forming a mosaic. There is also parched areas in which the stroma is more densely cellular and contains mineralized elements with the appearance of cementiculae.
Complementary Testing

Bone scan: diffuse uptake in all of the axial and appendicular skeleton. Bone densitometry: lumbar spine: 1617 g/cm² T +5.2 SD, Z +5.2 SD and femoral neck: 1769 g/cm² T +6.8 SD, Z +6.8 SD.

Analysis of the Complementary Tests

Laboratory findings reflected an increase in bone turnover, with an elevation of bone remodeling biomarkers, AP and hydroxiproline in urine, as well as an elevation of the hydroxiproline/creatinine ratio and the desoxypyridinoline/hydroxiproline ratio. The increased axial uptake of radioisotope reflected great bone metabolic activity with respect to the zones where uptake was normal. Bone densitometry showed an increased bone mineral density secondary to bone sclerosis.

Diagnosis

Picnodysostosis (Henri Toulouse-Lautrec’s disease). 1

Discussion

The term is derived from the greek pycnos, dense; dys, defective; osteos, bone. Picnodysostosis is a rare disease with a prevalence of 1:1 000 000 inhabitants. 1 It is transmitted in an autosomic recessive manner, and is found more frequently in inbred families. It is due to a mutation that inactivates the K⁺ cathepsin, an important protease in the degradation of collagen from the bone matrix, localized on chromosome 21, 1q. Osteoclasts, although quantitatively normal, a qualitatively deficient, leading to localized osteolysis. 2

Defective osteoclasts with large undigested-collagen filled vacuoles can be seen. Electronic microscopy shows a larger number of mineral particles and a chaotic alignment as a result of the altered orientation of the collagen fibers. 3 All of these changes lead to a considerable increase in bone fragility in these patients. 4

References