FIBROUS DYSPLASIA OF MAXILLARY BONES. 2 CASE REPORTS

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Abstract

Fibrous dysplasia has been identified for more than a century, however the scientific and clinical interest for this pathology has been relatively low, probably because of its reduced incidence. The recent genetic investigations permitted to recognize the etiology of this malady, seen as uncertain and controversial for a long time. The study analyzes the cases of 2 patients with fibrous dysplasia at the level of the viscero-cranium bone, subjected to surgical interventions of modelling resections of the pathological bone tissue, the pieces being examined histopathologically. Analysis of the 2 cases attempted at elucidating the etiological character of this genetically-determined malady, with no traumatic, reactive or amartomatous origin.

Keywords: fibrous dysplasia, genetic determination, modelling resections

INTRODUCTION

Fibrous dysplasia represents a rare benign lesion, characterized by replacement of bone tissue and bone marrow by an osteofibrous tissue [1]. Even if it is a congenital-type lesion, it is frequently diagnosed in decades 2-3 of life [2], as well as at adult ages [3]. It shows no pathognomonic symptoms, the laboratory data being non-specific. The etiology of fibrous dysplasia is given by the mutation of gene GNAS1 from the arm of chromosome 20, causing an increase of AMPc and, consequently, development of the affected osteofibrous tissue. Identification of the DNA from the bone pieces taken over from the patients permits to individualize the presence of the respective mutation and, consequently, to confirm the diagnosis: fibrous dysplasia. The result of such an analysis plays an exclusion role, invalidating or confirming a presumtive diagnosis [4].

Fig. 1. Fibrous dysplasia of the upper right maxillary; extraoral image

MATERIALS AND METHOD

Case I

16 year-old female patient addresses the Clinics for the deformation of the left cheek region, of the alveolar process and of the left hemi-maxillary (Fig.1). Asymmetry was progressively installed along 3-4 years, especially in the last year. Such an insidious evolution was accompanied by no specific symptomatology. Intraorally, deformation was observed only at the level of the vestibule, palatinally the aspect being a normal one. The occlusion is open frontally, the superior incisors occurring in infragnation (Fig.2).
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Fig. 2. Fibrous dysplasia of the upper right maxillary; intraoral image

Radiological orthopantomographic and computer-tomographic examinations evidenced an extended radio-opacity area, with numerous bone trabecules (granular aspect). The lesion occupies the whole left upper hemi-maxillary, and also the zygomatic bone, being extended up to the basis of the skull (Fig.3).

Case II

51 year-old male patient addresses our Clinics for correction of his pronounced facial asymmetry. The medical history of the patient includes a surgical intervention, about 20 years ago, for left mandibular body fibrous dysplasia. Clinical examination showed that the whole right hemi-mandible is excessively deformed, having an almost double volume (fig 4). The soft parts remained unmodified, the insidiously-installed deformation having occurred only at skeletal level, without being accompanied by other symptoms, after the last surgical intervention.

Fig. 3. Fibrous dysplasia of the upper right maxillary, CT

Fig. 4. Fibrous dysplasia of the left hemimandible, extraoral image

The solution selected is surgical ablation of the pathological bone (modelling resection). The anatomo-pathological examination of the piece evidences a stromal fibroblastic proliferation containing bone trabecules with typical aspect, lined with osteoblasts, which confirms the diagnosis of fibrous dysplasia.
Computer-tomograph examination evidenced that the mandibular bone is replaced by a non-homogeneous tissue with a marble-like aspect. The radio-transparency zones alternate with excessively developed radio-opacity regions. The process affects the whole left hemi-mandible (Fig. 5).

Fig. 5. Fibrous dysplasia of the left hemimandible, CT

Laboratory analyses were within the normal limits, so that the medical staff decided to perform a surgical extirpation of the pathological tissue. The procedure meant at removing the soft spongious bone together with mesenchima tissues.

Histopathological examination – made on 2 pieces with sizes of 80/30/20 mm and 15/10/10 mm – confirms the diagnosis of fibrous dysplasia.

In the samples employed for histological examination, the DNA from the pieces of frozen bone was analyzed by the polymerisation reaction with specific allele oligonucleotides, the punctiform mutation of gene GNAS1 from chromosome 20 being thus discovered.

DISCUSSION

Fibrous dysplasia has been described for the first time by Von Recklinghausen, in 1891. The correct term for defining this disease is osseo-fibrous or fibrous osseodysplasia, being classified according to the number of bones involved in mono- and poly-ostoic position.

Monostoic dysplasia is the most frequent form of its manifestation – occurring in a ratio of approximately 70-80% [5].

The literature of the field evidences a prevalence for the feminine sex [6], in a ratio of 1/1. In the maxillo-facial region, fibrous dysplasia occurs more frequently on the maxillary, and not on the mandible (ratio: 2/1), and especially in the posterior region [7]. It may also affect the calvarya, zygomatic bone, or the base of the skull. Even if affecting bones by both ossification modalities, it appears as more frequently manifesting in the membranous than in the cartilaginous bones [8].

In the beginning, symptomatology is discrete, involving mild pains in the affected region while, with the growth of the tumour, ocular phenomena (reduced visual capacity, diplopia), nasal deformations, dental problems, or sensorial phenomena appear [9]. In the maxillary region, of interest is always the maxillary sinus, which is either occupied or pushed towards the other structures [10], whereas disappearance of the hard lamina is also observed. In the first case here presented, these aspects are shown by CT investigations, as well.

Usually, radiological examination evidences a mixed radio-opaque/radio-transparent image, with an aspect of matted glass or orange skin [11]. At maxillary level, alteration of the maxillary sinuses may be observed; frequently, at the mandible, the mandibulary canal appears as pushed, in the here discussed case the canal being included in the tumoral tissue. In most cases, no peripheric delimitation is present, the margins converging towards the healthy tissue [12].

Clinical and radiological examinations suggest the diagnosis, which is subsequently supported or invalidated by histopathological data. Discovery of gene GNAS1 mutation contributed
to a safer diagnostication of this lesion. In both cases, DNA identification was performed from the bone inclusions taken over from the patients, which permitted individualization of the specific mutation.

Fibrous dysplasia has no hereditary character, the mutation occurring after formation of the zygote at the level of the somatic cells, which leads to a somatic mosaic-like picture [13].

The treatment aims at preventing fractures, reducing pain and at re-establishing the bone facial contours. In most situations, it involves a surgical intervention, even if attempts of drug therapy, generally with bisphosphonates, have been made, once known that Pamidronate (Aredia) inhibits tumoral growth [14].

Nevertheless, the surgical treatment remains essential, involving modelling resections especially in the growth period, the disadvantage being that they permit recessions (up to 20%) (8). In cases of progression towards the skull base, when the risk of compression on the ocular foramen may appear, more extended ablations are recommended [15].

CONCLUSIONS

The two cases here analyzed confirm the existing literature data on the localization, imagistics and treatment of cranio-facial fibrous dysplasia. On the basis of modern investigations of DNA extraction from frozen bone samples, the specific mutation of gene GNAS1 from chromosome 20 was identified, which confirms the new currents in the literature of the field, explaining the genetic etiology of this quite rare pathology.

References