CASE REPORT

Skull base fibrous dysplasia – diagnostic errors

Nicoleta Dumitrescu, Codrut Sarafoleanu
ENT&HNS Department, "Sfanta Maria" Hospital, Bucharest, Romania

ABSTRACT

Fibrous dysplasia is an uncommon benign bone tumor with a high destructive and invalidating potential. The skull base lesions are of particular interest for the specialists due to the deformities and dysfunctions that can appear. Therefore, a complete clinical and imaging evaluation (CT and MRI), followed by a correct diagnosis, is needed. An incomplete evaluation of the patients with craniofacial symptoms, with a normal or quasinormal ENT examination and nasal endoscopy represents a diagnosis error, which can have an important impact upon the prognosis of the disease. Considering the different histopathologic forms of fibrous dysplasia, the complications of this disease, it is important to have an early correct diagnosis and an appropriate therapeutic management considering the symptoms.

KEYWORDS: fibrous dysplasia, McCune-Albright syndrome, skull base, sphenoid

INTRODUCTION

Fibrous dysplasia is a benign disorder in which the normal structure of the bone and marrow is replaced by fibrous tissue[1,2]. It was first described by Lichtenstein in 1938[3] and it represents 5 to 7% of all benign bone tumors[4,5].

This lesion type can involve a single bone or multiple bones, in one-fourth of the cases being localized at the head and neck level[2]. In patients with skull involvement, the ethmoid bone is the most frequently affected, secondly being the sphenoid and frontal ones[6,7]. The skull base lesions are of particular interest for the specialists due to the deformities and dysfunctions that can appear.

Therefore, a complete clinical and imaging evaluation, followed by a correct diagnosis, is needed. The radiologic findings in case of fibrous dysplasia are variable, depending on the amount of fibrous tissue replacing the normal bone. There are numerous situations in which the imagistic features of a cranial tumor may not be conclusive for its benign or malignant form. Furthermore, there are cases when a benign tumor transforms in a malignant one.

The differential diagnosis is essential for a correct management and a favourable outcome of fibrous dysplasia. Sure enough, the final diagnosis is made by a thorough histopathological examination, even though the result may not be conclusive due to the inhomogeneous structure of the tumor[8].

In this paper, we present a case of an adult patient with skull base fibrous dysplasia, pathology first misdiagnosed as a chronic rhinopharyngitis due to an incomplete evaluation protocol.

CASE REPORT

A 39-year-old female patient was admitted in our ENT Department for left side hemicrania and mild left nasal obstruction with an approximately 3-month onset. The patient also presented an episode of diplopia and standing instability a month before the admission.

Her medical records showed that the patient was previously evaluated in another ENT Clinic, where, after a normal ENT clinical examination and nasal endoscopy, a biopsy sample was prelevated from the rhinopharynx mucosa. The histopathologic exam revealed a benign follicular hyperplasia (chronic rhinopharyngitis) and a second biopsy was indicated from the same region. We have to emphasise that the recommendation for the biopsy was made (wrongly?!) without having an imaging evaluation of the patient.

The nasal endoscopic examination, performed by us, revealed a hyperemic nasal mucosa with mucous secretions in both nasal cavities and hypertrophic inferior turbinates; at the rhinopharynx level – asymmetry of the posterior wall with a swelling covered by mucous discharge on its left side.

Corresponding author: Codrut Sarafoleanu, MD, PhD, Professor of Otorhinolaryngology
e-mail: csarafoleanu@gmail.com

The cerebral MRI showed a space replacing process in the left middle nasal cavity involving also the clivus, the left pterygopalatine fossa, the left sphenoid sinus and the rhinopharynx.

The endoscopic sinus surgery was taken in consideration. Preoperatively, the patient received parenteral antibiotic treatment associated with intra-venous steroid anti-inflammatory drugs.

For the preoperative decongestion of the left nasal cavity, we used vasoconstrictor infiltrations. Intraoperatively, we used nasal endoscopes and instruments specific for endoscopic sinus surgery. The left sphenoidotomy revealed a normal sinusal mucosa, without any tumoral mass or secretions at sphenoid sinus level. Our findings did not correlate with the MRI result. Further, we observed the protrusion of the left posterosuperolateral wall of the rhinopharynx covered by normal mucosa. We excised four biopsic pieces at this level and sent them to histopathological examination. The mucosa of the posterior ethmoid cells was hyperplastic, image that correlated with the MRI appearance, so we took a biopsic fragment from that level, as well.

The histopathologic results confirmed a nonspecific lymphoid hyperplasia.

The radiological monitoring, performed one week after surgery, involved a contrast enhanced brain MRI and skull base CT scan. They revealed an amorphous structure in the left half of the spongious bone of the sphenoid's body, the left greater wing and the base of the left pterygoid process, extending prior to the left superior canine and the anterior nasal spine of the maxillary bone; there was no change in calibre of the lacrimonasal duct, the greater and lesser palatine canals, but there was a narrowing at the level of the vidian canal and the pterygopalatine fossa on the left side. That "ground-glass" CT appearance (Figure 1) with MR tissue signal suggested the diagnosis of skull base fibrous dysplasia (Figure 2).

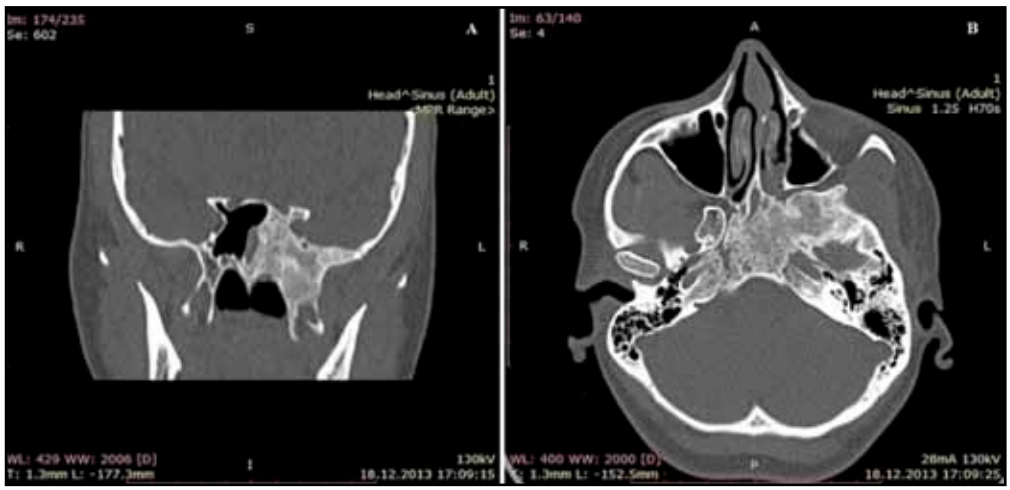

Figure 1 Skull base CT scan, coronal (A) and axial (B) slices – "ground –glass" appearance of the sphenoid's body, the left greater wing and the base of the left pterygoid process

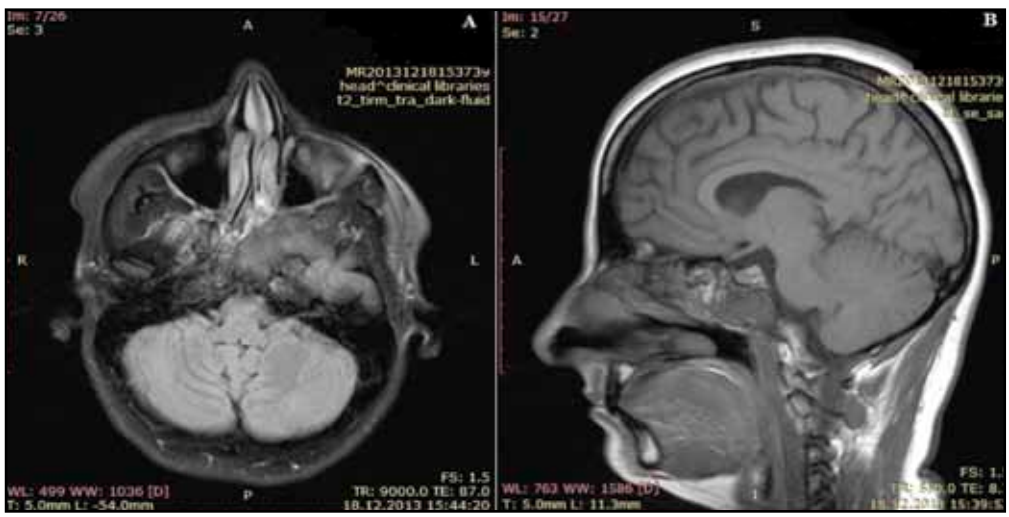

Figure 2 Contrast enhanced brain MRI – A. axial slice, T2-weighted image with high intensity signal showing an amorphous structure in the left half of the spongious bone of the sphenoid's body, the left greater wing and the base of the left pterygoid process; B. sagittal slice, T1-weighted image with low-intensity signal.

There was a normal aspect of the intracisternal cranial nerves, without perineural pathological contrast enhancement at this level. The embedding of the posterior part of the left carotid canal, at dysplastic bone level, may suggest a possible compression of the left abducens nerve in Dorello's canal.

DISCUSSIONS

The nasal and paranasal sinuses tumors are entities that may raise diagnostic problems, especially in their early stages. The incidence in Europe is about 1/100.000[9].

Among them, the fibrous dysplasia is a benign disease characterized by the replacement of the normal medulla of the bone with fibrous tissue caused by a defect in osteoblastic differentiation and maturation[10,11]. There are different types of fibrous dysplasia that may occur and may overlap: monoostotic (70-85%) type, which involves the femur, tibia or facial bones; polyostotic (15-30%) fibrous dysplasia involves multiple bones, mostly of the lower limb; the lesions can be localized or disseminated[8,12,13].

Fibrous dysplasia may be sporadic or integrated in syndromes like McCune-Albright Syndrome along with endocrine abnormalities (sexual precocity in female, hyperthyroidism) and café-au-lait spots[7] or Mazabraud Syndrome that goes with intramuscular lesions (muscular mixomas) near the affected bone[14].

The highest incidence of fibrous dysplasia is between 3 and 15 years[7], about 75% of patients presenting the disease before the age of 30 years[15]. One of the particularities of our case was the age of the patient.

It may affect in the same proportion males and females, with a female predilection in McCune-Albright syndrome[7,16].

The craniofacial fibrous dysplasia can be monoostotic in about 10% of cases and polyostotic in 50-100% of cases[17,18]. In case of skull base involvement, isolated lesions of the sphenoid bone is very rare[19].

From the clinical point of view, patients are usually asymptomatic, the disease being accidentally discovered during an imaging evaluation performed for a different pathology, like a head trauma for example. The type and the intensity of the symptoms depend on the site, the extension and the nature of the lesions, and they may manifest by nasosinusal, orbital, orodental, facial or neurologic syndrome, depending on the tumor extension. The most common symptoms of fibrous dysplasia are facial asymmetry (86%), mass presence (64%), blurred vision (24%), headache (20%), loss of visual field and sinusitis (8%), epiphora (7%), epistaxis and hearing loss (3%)[4,20]. In case of sphenoid bone involvement, the skull can become misshapen and cranial nerve syndromes may be present.

In our case, the patient complained about mild left nasal obstruction and left hemicrania, symptoms that may be also similar to those of chronic inflammatory pathology. The rhinosinusal tumors, with orbital and intracranial extension or bone destruction, often may have the same clinical manifestations, but rarely create difficulties in their diagnosis and treatment. In case of malignant tumors, the symptoms are represented by unilateral nasal obstruction accompanied by epistaxis, purulent or sanguinolent rhinorrhea and hyposmia and late diagnosis. In this case, the nasal endoscopic examination can reveal the presence of the tumor. But also, the same examination can show a normal or quasinormal nasal and rhinopharynx mucosa, these findings covering maybe a more profound bone pathology, as it was in the case presented. This is the reason why, in case of craniofacial symptoms, the nasal endoscopic examination has to be always associated with an imaging evaluation of the area.

Diplopia occurs when ethmoid and maxillary bones are affected. In our case, diplopia can also be present due to the embedding of the posterior part of the left carotid canal, at dysplastic bone level (a possible compression of the left abducens nerve in Dorello's canal). In both fibrous dysplasia and malignant tumor, diplopia may be accompanied by visual acuity loss, exophtalmia, periorbital oedema and epiphora, depending on the extension of the disease. Furthermore, a tumor of the nose or rhinopharynx can give symptoms of Eustachian tube dysfunction or chronic otitis media, symptoms that our patient did not present.

Considering the polymorphism of the symptoms, a thorough history (onset, symptoms, presence or absence of functional impairments, duration) and physical examination are needed.

The correct diagnosis of fibrous dysplasia has to be based on clinical symptoms and a complete radiological examination.

The classic radiographic evaluation can reveal a variety of features ranging from lucency to sclerosis, depending on the amount of fibrous tissue. In early stages, fibrous dysplasia may have ill- or well-defined borders, being unilocular or multilocular. In advanced stages, a mix of radiolucent and radiopaque images can be found, with mottled radiopaque patterns (groung-glass, orange peel or fingerprints) and ill-defined borders[18,21,22]. Although the plain radiological features of fibrous dysplasia are non-specific and it has to be later differentiated from an ossifying fibroma or Paget's disease, this simple and affordable investigation can lead the specialist towards the correct diagnosis of fibrous dysplasia.

A more complete radiological tool is the CT-scan. It is the investigation of choice for both diagnostic and follow-up, being able to differentiate the fibrous dysplasia from other osteodystrophies and to assess the

extent of the tumor (establishing the margins between affected and unaffected bone)[7,21,23]. The classical appearance of fibrous dysplasia on a CT-scan is the "ground-glass" pattern associated with bone asymmetry and cystlike changes[4,24]. The same characteristic aspects were found in the skull base CT-scan performed in our presented case.

On MRI, a low-intensity signal on T1-weighted images, associated with well-demarcated borders on both T1- and T2-weighted images, seems to be characteristic for fibrous dysplasia. Also, T2-weighted images have a higher intense signal and the area becomes more heterogeneous after gadolinium administration[4,21,25] .

The association of CT-scan and MRI can provide sufficient information for the diagnosis of fibrous dysplasia. The same protocol investigation was used in our case.

Single photon emission computer tomography (SPECT) was also suggested as being more sensitive in the diagnosis of fibrous dysplasia[26].

The differential diagnosis of fibrous dysplasia should be made with other benign or malignant tumors with craniofacial location and manifestations[27], as seen in Table 1.

The natural evolution of this disease, the prognosis of our patient are also very important. Having an early onset (infancy, early adolescence), it has been thought it will get inactive after puberty[28]. Recent publications show that fibrous dysplasia can progress into the adulthood period[25], similar with our case. The polyostotis fibrous dysplasia has a high rate of progression after puberty, beyond the 3rd or 4th decades of life[12].

Fibrous dysplasia spontaneous resolution does not occur, but the lesion can increase in size, involving multiple bones, eye and mouth cavities, the nose or paranasal sinuses. Craniofacial deformities, cranial nerve syndromes, protrusion of the eye-ball with / without blindness due to the compression on the optic nerve can be described. The fact that our patient already presented episodes of diplopia and standing instability can be considered as being a negative point in the disease natural evolution.

The appearance of visual or hearing changes, airways obstruction, paresthesia requires an immediate complete surgical evaluation (otorhinolaryngologist, neurosurgeon, ophthalmologist, oral and maxillofacial surgeon)[29].

Malignant transformation of fibrous dysplasia was reported in almost 0.5% of patients[30,31], most frequent being the osteosarcomas, followed by chondrosarcomas and fibrosarcomas. The time elapsed between the diagnosis and the malignant degeneration is usually counted in years. The malignant transformation seems to be higher in patients undergoing radiotherapy[32,33].

The periodical radiologic evaluation is essential in patients diagnosed with fibrous dysplasia. An increase in size of the lesion or the transformation of mineralized bony lesions into lytic ones are signs of malignancy.

Those cases with slow progress and with no functional impairment should be followed-up. Surgical treatment should be indicated only in patients with compression signs. Intravenous biophosphonate therapy for the osteolytic lesion can be performed[21,34].

CONCLUSIONS

Fibrous dysplasia is an uncommon benign bone tumor with a high destructive and invalidating potential. The positive diagnosis is made by performing a correct radiologic examination (CT and MRI), the bone biopsy being indicated only in cases with unspecific radiologic findings.

Craniofacial symptoms, like headache, nasal obstruction or diplopia, need a complete clinical and radiological evaluation. A simple ENT examination and nasal endoscopy can reveal a normal or quasinormal nasal and rhinopharynx mucosa, which can lead to a wrong therapeutical indication and diagnosis.

Regarding our case, we believe the prescription for

Table 1
The craniofacial tumors and some of their manifestations with which one can differentiate fibrous dysplasia

Tumors with craniofacial location	Manifestations
Cemento-ossifying fibroma	Better defined
Intraosseous meningioma	Abuts the intracranial structures
Paget's disease	Spares facial bones Has predilection for the skull vault
Sclerotic metastases	Smaller expansion Different demographics

a second biopsy was not a proper indication, because the relevant radiologic findings and the negative result of the first rhinopharynx biopsy were enough for the diagnosis of fibrous dysplasia.

Considering the different histopathologic forms of fibrous dysplasia, the complications of this disease, it is important to have an early correct diagnosis and an appropriate therapeutic management considering the symptoms.

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