CASE REPORT

Olfactory dysfunction as first presenting symptom of cranial fibrous dysplasia

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SUMMARY

Fibrous dysplasia (FD) is a benign bone disorder presenting with a variety of clinical manifestations. This is the first reported case of anosmia as presenting symptom of FD. We present the case of a 72-year-old female patient with a progressive olfactory dysfunction. Clinical examination revealed evidence of chronic rhinosinusitis; therefore the patient was treated with a course of oral corticosteroids. The patient had no improvement in her olfactory ability and imaging studies were ordered. Bony lesions characteristic of craniofacial FD were found, causing obstruction of the central olfactory pathway. This case emphasises the need to conduct further investigations in patients with rhinosinusitis and olfactory dysfunction especially when they present no response to oral steroid treatment.

BACKGROUND

Fibrous dysplasia (FD) is a relatively common, benign disorder of bones attributed to a non-inherited mutation in the gene *GNAS1* on chromosome 20.¹ It presents in two clinical forms (monostotic or polyostotic) depending on the amount of the affected bones. Their prevalence ranges from 10% to 29% and 71% to 91% of all cases respectively.¹ Polyostotic cases, with multiple organ manifestations and café-au-lait skin spots, are referred to as cases of McCune-Albright syndrome. In the current literature, cases with the involvement of facial or cranial bones are described by the term craniofacial FD. Cranial presentations are rare, comprising 10–25% of FD cases.² The most commonly affected facial bones are the maxilla and mandible.²

The characteristic bone swellings are painless, progressively expanding lesions. They produce not only cosmetic deformities but also sino-orbital and audiological complications, peripheral nerve compression and central neurological manifestations. The involvement of different structures leads to a variety of clinical manifestations. The most common of these are facial asymmetry, visual loss, atypical headaches and facial pain.³ Olfactory dysfunction is rarely reported and to our knowledge this is the first case where it is the presenting symptom.

CASE PRESENTATION

We present the case of a 72-year-old woman with a 3-month history of olfactory dysfunction. In her medical history, no nasal trauma or surgery was reported. The patient had previous imaging studies of the nose and paranasal sinuses.

Clinical examination including a nasal endoscopy revealed inflamed nasal mucosa and, specifically polypoid middle meatal mucosa, (figure 1) findings consistent with chronic rhinosinusitis (CRS). The psychophysical olfactory testing was performed using the Sniffin' Sticks test battery, which involves tests for odour threshold, discrimination and identification. The sum of three subtest scores resulted in the threshold, discrimination, identification (TDI) score. As defined in large population studies, a score > 30 indicates normosmia, a score between 16 and 30 points indicates decreased olfactory function (hyposmia) and a score of less than 16 points indicates functional anosmia.⁴ The TDI score was 12, indicative of functional anosmia. More precisely the TDI scores were 2, 5 and 5, respectively. The patient received a 2-week course of oral corticosteroids (prednisolone, 25 mg/day), without showing any improvement.

INVESTIGATIONS

Owing to non-response to steroids, although endoscopic findings confirmed the diagnosis of CRS, a CT of the nose and paranasal sinuses was performed. The CT findings demonstrated bony lesions of the right temporal and sphenoid bone characteristic of craniofacial FD (figure 2). The sphenoid lesion completely distorted the area of the olfactory bulbs. Although CRS findings in the ethmoids and left frontal sinus were present, these could not be considered as a result of paranasal obstruction from the bony lesion of the sphenoid bone. Further investigations included MRI of the brain in order to evaluate the extent of soft tissue involvement in the brain (figure 3). The skeletal bones were investigated by plain X-rays and no other lesions were demonstrated.

Blood tests included measurements of serum alkaline phosphatase, calcium, parathyroid hormone and 1,25-dihydroxyvitamin D levels. All values were within the normal range.

DIFFERENTIAL DIAGNOSIS

Differential diagnosis of the masses affecting the olfactory cleft should include an adjacent meningioma as a possible source of hyperostosis, a low-grade intramedullary (central) osteosarcoma, Paget disease, ossifying fibroma and adamantinoma. An esthesioneuroblastoma, although it has different clinical and radiological appearance, should be included in differential diagnosis as it derives from the olfactory epithelium.

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Figure 1 Endoscopic view of the left nasal cavity (asterisk indicates nasal septum). Polypoid inflammatory distortion of the middle turbinate.

TREATMENT

Localisation of the lesion, the age and the general condition of the patient did not permit surgical removal of dysplastic bone or decompression of the olfactory structures.

OUTCOME AND FOLLOW-UP

The patient remains, in a close follow-up, without any change in the clinical status of the disorder.

DISCUSSION

FD can involve the skull base, paranasal sinuses and the orbit. According to Lustig *et al*⁵ the ethmoids are involved in 71% of cases. In another study by Van Tillburg,⁶ frontal bones were more commonly affected.

Several reports of cranial FD describe various initial clinical manifestations. Most common are facial deformities, atypical

facial pain and headaches.^{7–8} Additionally, cranial nerve dysfunction has also been reported including visual impairment and labyrinth involvement with sensorineural hearing loss and dizziness.^{9–11} In some cases FD can result in sinus obstruction and secondary sinusitis or mucocoele formation. Sinusitis was reported in 43% of the patients in Lustig *et al*⁵ series and in 8% of Rahman *et al*³ series. Interestingly, in our case, sinusitis seems to be a coincidental finding and not secondary resulted by FD.

Our patient presented with anosmia and the endoscopic findings were consistent with CRS. Olfactory dysfunction in CRS is mainly conducive due to nasal obstruction of the olfactory cleft. However, chronic inflammation of the olfactory mucosa can cause distortion of the olfactory epithelium and sensorineural loss in the long term.¹²

Imaging studies are not recommended as having fundamental importance in the diagnosis of CRS; therefore, a CT scan was performed after the treatment failure.

In a study by Mueller *et al*, with regard to CRS-related olfactory loss, the authors suggest that a confirmation of the diagnosis with a CT scan does not seem to be necessary.¹³ However, in their series a high percentage (33%) of patients with suggested history of CRS had no correlating radiological findings. As a short course of systemic steroids is proposed as first-line treatment to confirm or not the presence of CRS, a failure of this treatment should raise questions about the diagnosis requiring further imaging studies.

An interesting point of our report is the late occurrence of the disease. FD, especially its polyostotic form is usually diagnosed in children and adolescents and the progression stops after puberty.^{1–3} However, our patient was incidentally diagnosed in her eighth decade of life. Lesions of the cribriform plate with slow growth present as anosmia usually when they occupy the area of both olfactory bulbs. Thus only a few patients report olfactory dysfunction. This may be due to the fact that in most of the patients only one side of the nose is affected and olfactory function is maintained by the contralateral side.¹⁴ In addition, the decline in olfactory function is a long process, similar to what is seen in the elderly and an adaptation of the patient with this gradual decrease delays the diagnosis.

Imaging studies play a crucial role in FD diagnosis. CT findings may vary due to the osseous component of the lesion; however, their distinctive characters lead usually to a safe diagnosis.¹⁵ Three radiographic patterns have been described. In almost half of the cases, the ground-glass or mixed pattern is reported.¹⁶ In addition, the sclerotic and the cystic pattern are also presented. On the contrary, MRI lacks disease specific characters. However, it provides useful information, on the impact of bony changes on neighbouring soft-tissue structures, detection of sinus mucocoeles (high signal intensity on T2-weighted image) and exclusion of an adjacent meningioma as a possible source of hyperostosis.¹⁷ Owing to characteristic radiologic findings, imaging studies are considered diagnostic in many cases without the necessity for histological confirmation.^{15–17} Regarding the treatment of FD, no widely accepted protocol exists. Since FD has a benign nature, conservative management is the first choice of treatment. Surgery is not recommended except in cases of severe functional and aesthetic deformities or where the vital structures are compressed (decompression of the optic nerve).¹⁸ In all cases, the age, the general condition of the patient, the vitality of the structures implicated and the aggressiveness of the disease should be taken into account.¹⁹

Figure 2 CT scan axial section. Chronic frontal sinusitis in the left side is indicated with the black arrow. White arrow indicates the right olfactory bulb area. The lesion disrupts posteriorly both olfactory bulbs.

Figure 3 (A) MRI T1-weighted image coronal section. The lesion invades and compress the olfactory bulb area in both sides. (B) MRI T2-weighted image axial section the lesion (black area) disrupt the continuity of both olfactory bulbs (white arrow).

In the current literature, bisphosphonate therapy presents promising results of bone pain control and radiological improvement in cases of craniofacial FD.²⁰ Additionally, calcium, vitamin D and phosphorus supplements may be useful in patients with such a deficiency.

Patients with FD need a close follow-up in order to monitor deformities and the course of any functional impairment in the long term.

The presented case reports anosmia as the first presenting symptom of craniofacial FD. Although the loss of olfactory function is not a key symptom for investigation with a CT scan, our case confirms the need for imaging studies in selected patients with CRS without the response to steroid treatment.

Learning points

- ▶ Anosmia can be the presenting symptom of fibrous dysplasia.
- ▶ Chronic rhinosinusitis with anosmia and no response to steroids requires imaging studies.
- ▶ Surgery is limited only in severe functional or aesthetic deformities.

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Competing interests None.

Patient consent Obtained.

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REFERENCES

- 1 Feller L, Wood NH, Khammissa RAG, *et al.* The nature of fibrous dysplasia. *Head Face Med* 2009;5:22.
- 2 Rojas R, Palacios E, Kaplan J, *et al.* Fibrous dysplasia of the frontal sinus. *Ear Nose Throat J* 2004;83:14–15.
- 3 Rahman AMA, Madge SN, Billing K, *et al.* Craniofacial fibrous dysplasia: clinical characteristics and long-term outcomes. *Eye* 2009;23:2175–81.
- 4 Kobal G, Klimek L, Wolfensberger M, *et al.* Multicenter investigation of 1,036 subjects using a standardized method for the assessment of olfactory function combining tests of odor identification, odor discrimination and olfactory thresholds. *Eur Arch Otorhinolaryngol* 2000;257:205–11.
- 5 Lustig LR, Holliday MJ, McCarthy EF, *et al.* Fibrous dysplasia involving the skull base and temporal bone. *Arch Otolaryngol Head Neck Surg* 2001;127:1239–47.
- 6 Van Tilburg W. Fibrous dysplasia. In: Vinken PJ, Bruyn GW. eds. *Handbook of clinical neurology*. Amsterdam: North Holland Publishing, 1972:163–212.
- 7 Sharma RR, Mahapatra AK, Pawar SJ, *et al.* Symptomatic cranial fibrous dysplasias: clinico-radiological analysis in a series of eight operative cases with follow-up results. *J Clin Neurosci* 2002;9:381–90.
- 8 Camilleri AE. Craniofacial fibrous dysplasia. *J Laryngol Otol* 1991;105:662–6.
- 9 Alam A, Chander BN. Craniofacial fibrous dysplasia presenting with visual impairment. *MJAFI* 2003;59:342–3.
- 10 Morrissey DD, Talbot JM, Schleuning AJ II. Fibrous dysplasia of the temporal bone: reversal of sensorineural hearing loss after decompression of the internal auditory canal. *Laryngoscope* 1997;107:1336–40.
- 11 Grasso DL, Guerci VI, Di Emidio P, *et al.* Dizziness as presenting symptom of monostotic fibrous dysplasia. *Int J Pediatr Otorhinolaryngol Extra* 2006;1:138–41.
- 12 Konstantinidis I, Witt M, Kaidoglou K, *et al.* Olfactory mucosa in nasal polyposis: implications for FESS outcome. *Rhinology* 2010;48:47–53.
- 13 Mueller C, Temmel AF, Toth J, *et al.* Computed tomography scans in the evaluation of patients with olfactory dysfunction. *Am J Rhinol* 2006;20:109–12.
- 14 Welge-Luessen A, Temmel A, Quint C, *et al.* Olfactory function in patients with olfactory groove meningioma. *J Neurol Neurosurg Psychiatry* 2001;70:218–21.
- 15 Lisle DA, Monsour PA, Maskiell CD. Imaging of craniofacial fibrous dysplasia. *J Med Imaging Radiat Oncol* 2008;52:325–32.
- 16 Chen YR, Chang CN, Tan YC. Craniofacial fibrous dysplasia: an update. *Chang Gung Med J* 2006;29:543–9.
- 17 Khalil HS, Toynton S, Stevenon N, *et al.* Radiological difficulties in the diagnosis of fibrous dysplasia of the sphenoid sinus and the cranial base. *Rhinology* 2001;39:49–51.
- 18 Amit M, Collins MT, FitzGibbon EJ, *et al.* Surgery versus watchful waiting in patients with craniofacial fibrous dysplasia—a meta-analysis. *PLoSOne* 2011;6:e25179.
- 19 Schreiber A, Villaret AB, Maroldi R, *et al.* Fibrous dysplasia of the sinonasal tract and adjacent skull base. *Curr Opin Otolaryngol Head Neck Surg* 2012;20:45–52.
- 20 Chapurlat R. Medical therapy in adults with fibrous dysplasia of bone. *J Bone Miner Res* 2007;21:114–17.

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