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Case report

McCune-Albright syndrome with loss of vision and its management: a case report

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Abstract

McCune-Albright syndrome (MAS) is defined by the triad of *café-au-lait* macules, endocrinopathies, and polyostatic fibrous dysplasia (FD) of the bone. It results from somatic *GNAS* mutations leading to mosaic Gαs activation. FD can result in bone deformities, fractures, and cranial nerve compression, including optic neuropathy. Optic nerve compression by FD may lead to visual disturbances or even blindness.

We report a 13-year-old boy with MAS and left eye visual impairment due to optic nerve compression by FD.

Endoscopic transnasal optic nerve decompression of the optic nerve was performed. Postoperatively, the patient exhibited slight visual improvement, though delayed intervention may have limited recovery.

The case report highlights endoscopic decompression as a safe and effective approach for FD-related optic neuropathy, though optimal timing remains debated. Prophylactic intervention in high-risk cases warrants further investigation to prevent irreversible vision loss.

Keywords

Endoscopic, transnasal, fibrous dysplasia, decompression, McCune-Albright syndrome, optic nerve.

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Introduction

McCune-Albright syndrome (MAS) is a rare, non-inherited disorder with an estimated prevalence of 1/100,000 to 1/1,000,000.[1] It is characterized by *café-au-lait* macules, endocrine dysfunction, and polyostotic fibrous dysplasia (FD) [1, 2]. Endocrine manifestations may include hypophosphatemic rickets, precocious puberty, acromegaly, hyperthyroidism, and Cushing's syndrome [1, 2].

FD is "a benign, presumably developmental lesion characterized by fibrous connective tissue with a whorled pattern and trabeculae of immature non-lamellar bone" [3]. It is caused by activating mutations in *GNAS*. While the most common mutations are R201H and R201C, rare variants such as R201G, Q227L, and R201S have also been reported [4].

The pathogenic *GNAS* mutation occurs in early embryonic development, affecting the cAMP pathway-associated G protein Gαs (Gs alpha subunit), leading to FD/MAS [4]. Somatic activating mutations in Gαs result in mosaic FD, where fibro-osseous tissue replaces normal bone and marrow. These expansile bone lesions can cause deformities, pain, fractures, and functional impairment. FD may affect any skeletal region, presenting as monostotic (single bone) or polyostotic (multiple bones) [5].

FD/MAS typically involves the skin, bones, and certain endocrine organs [4]. The earliest manifestation is often hyperpigmented *café-au-lait* macules, which usually appear in early childhood. FD can range from an asymptomatic monostotic lesion (discovered incidentally) to a severe polyostotic form affecting nearly the entire skeleton. In advanced cases, it may lead to progressive scoliosis, facial deformities, and hearing or vision loss.

Endocrinopathies associated with MAS include: precocious puberty, thyroid hormone abnormalities, elevated growth hormone (GH) levels, FGF23-mediated phosphate wasting, which reduces 1α-hydroxylase activity and increases renal phosphate excretion and neonatal hypercortisolism [6].

The expansion of fibrous tissue may compress cranial nerves, particularly those involved in vision and hearing. Optic nerve compression can lead to visual disturbances or blindness, while auditory canal involvement may cause hearing loss [7].

The treatment of patients with MAS is highly specialized, often requiring a multidisciplinary approach involving pediatrics, endocrinology, orthopedics, and pain management. While current therapies focus on symptom control, emerging treatments show potential for more targeted intervention [8]. For precocious puberty, girls may receive aromatase inhibitors or estrogen modulators, whereas boys are typically treated with androgen blockers or steroidogenesis inhibitors [9]. Non-autoimmune hyperthyroidism is usually managed with thionamides, though thyroidectomy remains a preferred long-term solution. GH excess often responds well to somatostatin analogs or GH receptor antagonists. Neonatal hypercortisolism is primarily treated with metyrapone, though adrenalectomy may be necessary [10]. FD is managed with pain relief and bisphosphonates [11].

Recent advances suggest that novel therapies, such as denosumab (a RANKL inhibitor) and burosumab (an FGF23 antibody), could revolutionize MAS treatment by addressing underlying molecular mechanisms. However, further research is needed to confirm their efficacy. Unlike traditional symptomatic approaches, these new strategies aim for targeted therapy, though the mosaic nature of MAS complicates the development of a universal cure [12].

Optic nerve decompression surgery has its risks, like postoperative blindness in 5-33% of cases or other types of visual impairments [13]. Optic nerve decompression may be performed through classic transcranial and transfacial approaches and through endoscopic approaches, which have many advantages when compared to traditional external approaches. Advantages include shorter recovery time, lack of diplopia, and decreased morbidity ratio [14].

We describe a 13-year-old boy with MAS who experienced left-eye vision loss due to optic nerve compression from FD. Endoscopic transnasal optic nerve decompression was performed.

Case report

A 13-year-old boy, a known case of MAS, was diagnosed at the age of 3 years. He had multiple

giant *café-au-lait* macules on his back, face and abdomen with irregular ("coast of Maine") borders, distinct from the smoother borders seen in neurofibromatosis type 1 (NF1). He experienced bone pain and recurrent pathological fractures. X-rays showed extensive bowing, mixed lytic-sclerotic bone lesions, loss of cortico-medullary differentiation, and healed fractures. Bilateral femur involvement with progressive varus deformity resulted in a "Shepherd's crook" appearance. He otherwise had normal development and intelligence, with early signs of puberty (pubic hair) without signs of precocious puberty. Thyroid, parathyroid, cortisol and GHs were normal. FSH/LH were also normal for his age. A *GNAS* mutation test was not performed, which can help to differentiate MAS from other overlapping conditions like NF1; the skin lesion of the *café-au-lait* spots, in our case with irregular ("coast of Maine") border, distinguishes it from the smoother border of NF1.

He is a product of a non-consanguineous marriage with uncomplicated pregnancy and birth. The family has no history of bone diseases. He was treated with calcium, vitamin D, and bisphosphonate. A bone biopsy was done to confirm the diagnosis of FD.

In the last year, the boy suffered from a gradual deterioration of vision in his left eye. On eye examination, the visual acuity in the right eye was 6/9 after myopia correction. However, left eye visual acuity was 6/60 with optic nerve atrophy without papilledema and non-correctable (**Fig. 1**). Magnetic resonance imaging (MRI) and computed tomography (CT) scans of ethmoidal sinus and optic nerves showed compression of the left optic nerve by immature bone (**Fig. 2** and **Fig. 3**). CT is preferred in visualization of bony defect [15]. Endoscopic endonasal optic nerve decompression was recommended with the diagnosis of optic neuropathy.

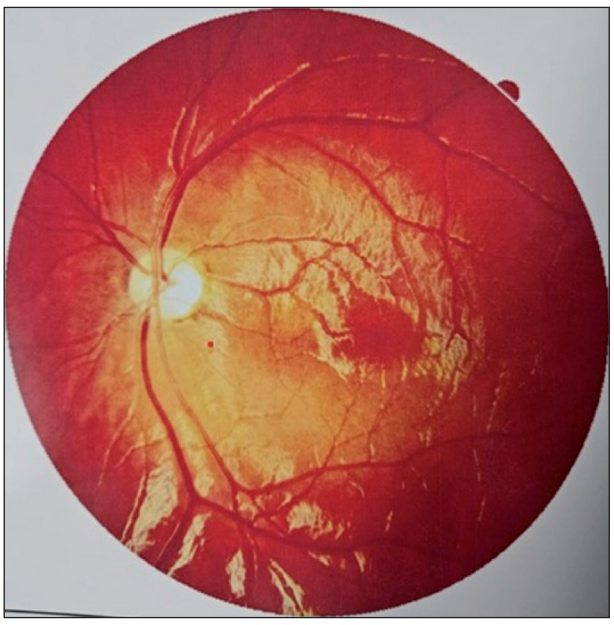

Figure 1. Left optic nerve atrophy.

Surgical method

The procedure was performed by an experienced rhinologist specializing in advanced endoscopic sinus and skull base surgery. General anesthesia was induced via orotracheal intubation without complications. The patient was positioned supine on the operating table with the head in a neutral position. The nasal cavity was prepared with an antiseptic solution and sterilely draped. Nasal decongestion was achieved by placing cotton pledgets soaked with oxymetazoline into the left nasal cavity, followed by submucosal infiltration of 1% lidocaine with 1:200,000 epinephrine for local anesthesia and vasoconstriction.

A rigid endoscopic approach was employed through the left nostril. Surgical access was optimized by partial resection of the left middle turbinate and conservative trimming of the mid-portion of the left inferior turbinate. The uncinate process was meticulously identified, sharply incised, and removed using angled forceps combined with a 4-mm endoscopic microdebrider, thereby exposing the natural maxillary ostium. This was subsequently enlarged to create a wide left maxillary antrostomy, providing complete access to the maxillary sinus. Under continuous irrigation, the posterior, superior, and inferior walls of the maxillary sinus were carefully drilled using a 4-mm high-speed diamond burr until the lamina papyracea was fully exposed. The drilling proceeded posteriorly toward the orbital apex with strict maintenance of a lateral trajectory to preserve the integrity of the orbital fat and periorbita.

Optic nerve decompression

Attention was then directed to the sphenoethmoid region housing the left optic canal (**Fig. 4**). The thickened bony plate overlying the optic nerve was identified and meticulously thinned and removed under constant endoscopic visualization using the high-speed burr (**Fig. 5** and **Fig. 6**). Particular care was taken to avoid injury to adjacent critical structures (for example, the internal carotid artery and orbital contents). The

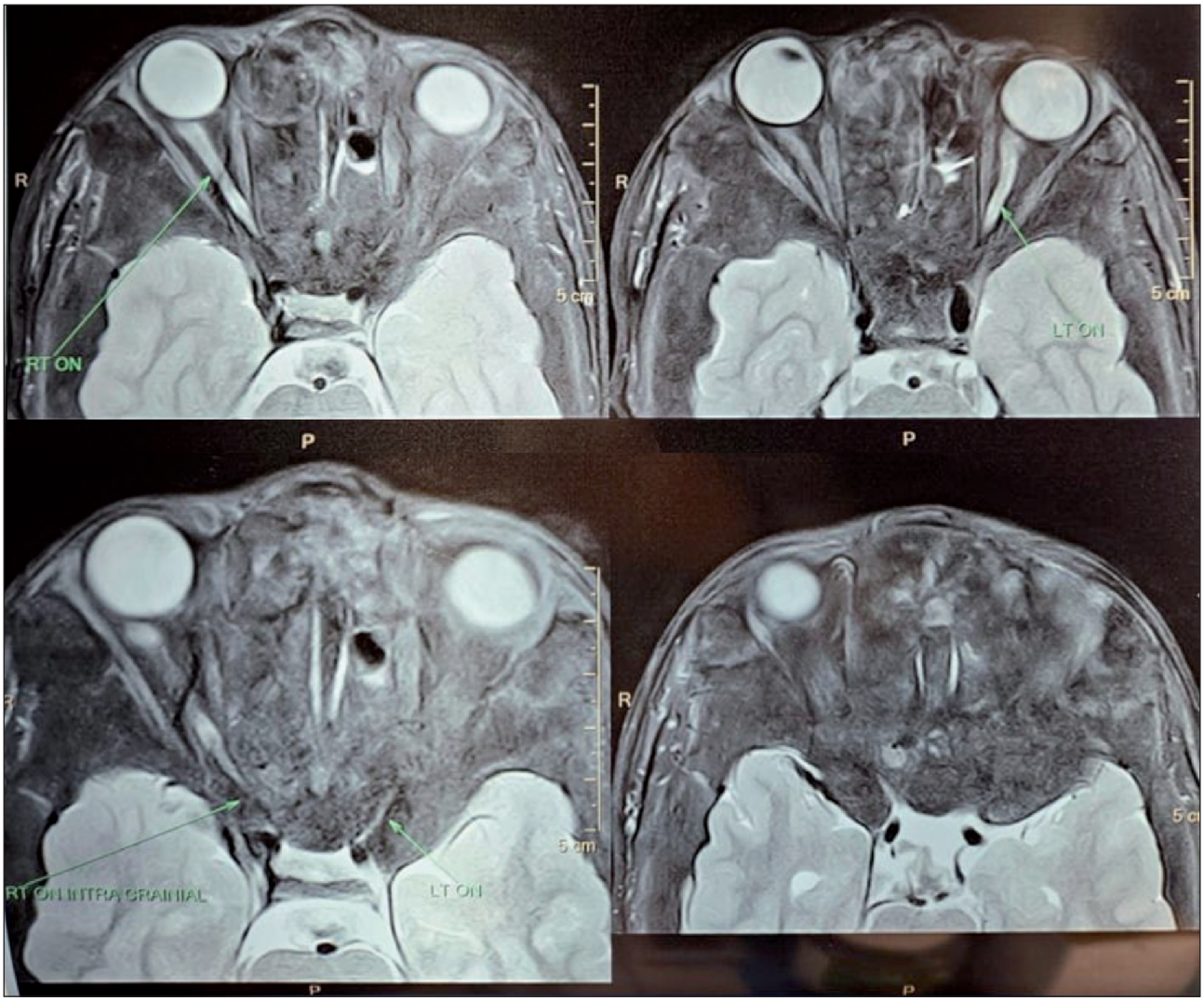

Figure 2. Orbital computed tomography (CT) scan: compression of the left optic nerve by immature bone.

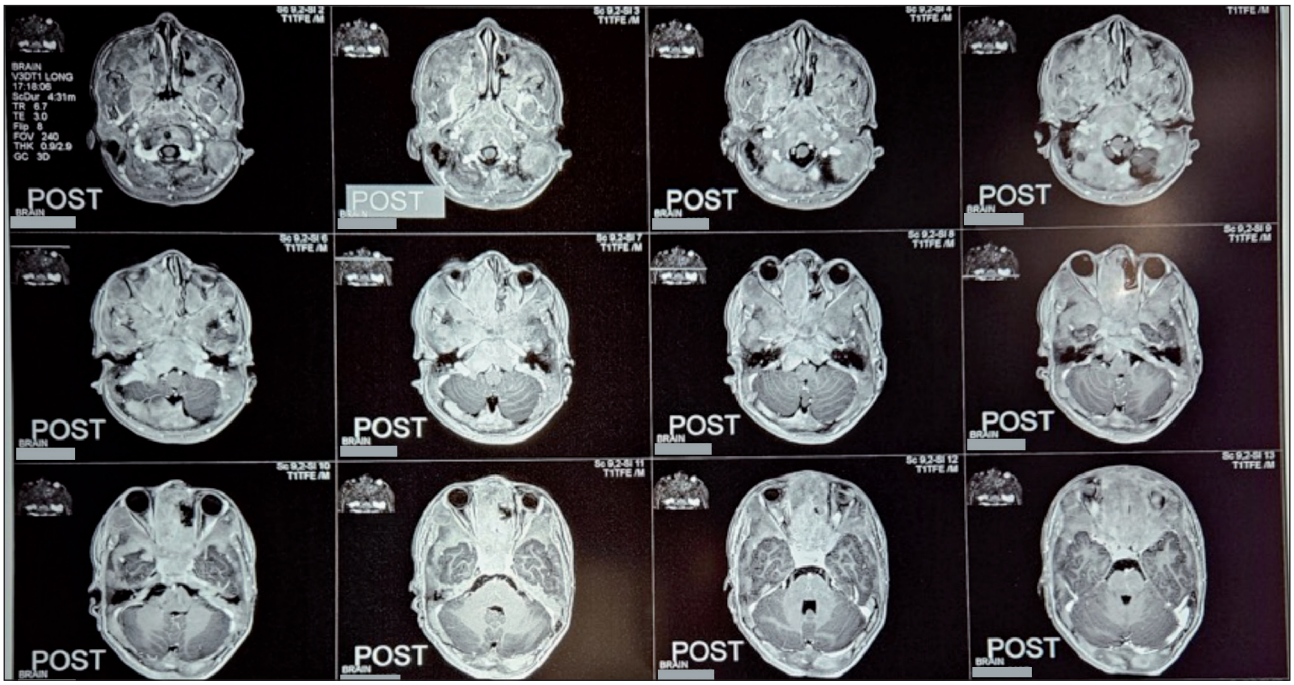

Figure 3. Magnetic resonance imaging (MRI) of optic chiasma: compression of the left optic nerve by immature bone.

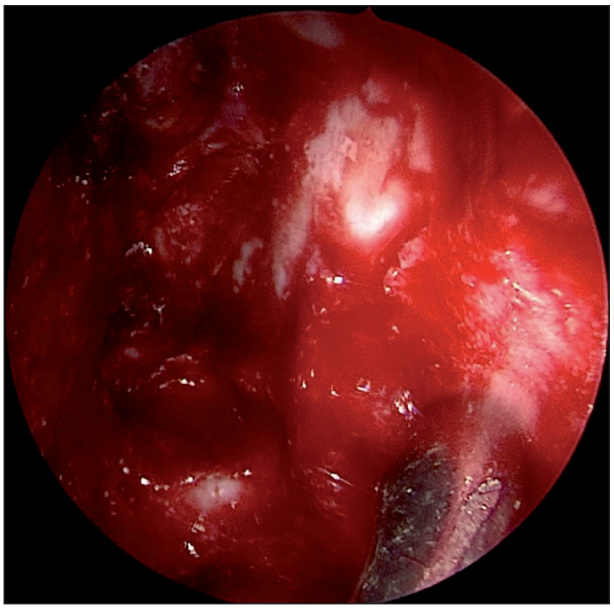

Figure 4. Open ethmoid sinus and sphenoid sinus.

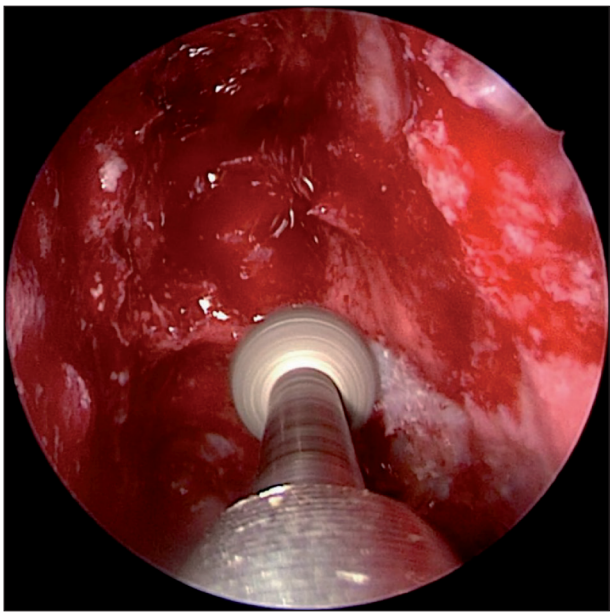

Figure 5. Fracture of the optic nerve bony canal.

optic nerve sheath was preserved at all times. This bony decompression was carried out longitudinally along the optic canal toward the orbital apex, ensuring circumferential removal of the encasing bone to fully decompress the left optic nerve.

No intraoperative optic nerve function monitoring (such as visual evoked potentials) was available due to resource limitations, so the adequacy of decompression was confirmed by direct anatomical assessment and endoscopic visualization.

Hemostasis was achieved primarily with bipolar electrocautery and reinforced with absorbable hemostatic agents.

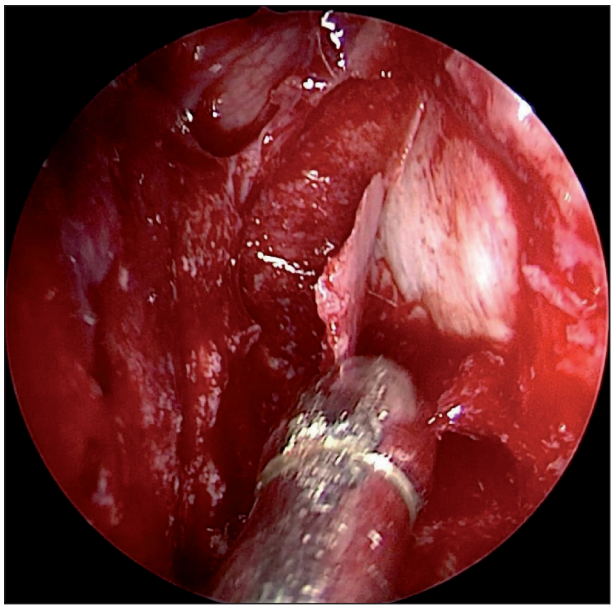

Figure 6. The endoscopic visualization reveals the left sphenoid sinus and orbital apex. The abnormal bone covering the orbital apex and optic nerve is carefully drilled away.

A bone specimen was obtained from the decompression site in the sphenoethmoidal region. This specimen was sent for histopathological analysis to confirm FD and exclude any other pathology. Intraoperatively, the bony anatomy appeared consistent with FD, with areas of dense sclerotic, dysplastic bone observed. The total operative time was approximately 3 hours, and the estimated blood loss was about 100 mL. There were no cerebrospinal fluid leaks, no vascular injuries, and no other intraoperative complications. The patient tolerated the procedure well without hemodynamic instability.

Postoperatively, the patient was given a short course of systemic corticosteroids to reduce optic nerve edema. A 7-day course of prophylactic broad-spectrum antibiotics was prescribed, and saline nasal irrigations were initiated multiple times daily to maintain nasal hygiene. The patient was instructed on standard nasal precautions (avoiding nose blowing, heavy lifting, and sneezing with the mouth closed).

Ophthalmology follow-up was arranged for postoperative visual assessment. At 6 weeks after surgery, the patient demonstrated slight improvement in left-eye vision; he was able to count fingers at a distance of < 1 meter, indicating a modest recovery of the optic nerve function.

The biopsy revealed slender, curved trabeculae with a “fishhook” pattern interspersed in fibrous

tissue, exhibiting the classic "Chinese characters" appearance – confirming FD.

Discussion

The diagnosis of MAS is primarily clinical, requiring ≥ 2 characteristic features (*café-au-lait* macules, polyostotic FD, endocrinopathies). The classical triad is more common in females, while the non-classical form involves only 2 features. *GNAS* mutation detection varies by tissue mosaicism and assay sensitivity – PCR-based sequencing identifies mutations in > 80% of lesional tissue but only 20-30% of peripheral blood lymphocytes. While a pathogenic *GNAS* mutation supports diagnosis, its absence does not exclude FD/MAS; thus, genetic testing rarely alters management in clinically evident cases [9, 16].

This patient exhibited 2 FD/MAS features: characteristic *café-au-lait* macules (distinguishing it from NF1) and polyostotic FD, confirmed by intraoperative bone biopsy, which revealed slender, curved, and irregularly shaped trabeculae with the distinctive "fishhook" pattern, intermixed with fibrous tissue.

Other researchers have reviewed the spectrum of MAS, noting that the non-classical form may include only 2 components of the triad (e.g., FD plus *café-au-lait* macules) without precocious puberty, particularly in boys. There are several key differences between precocious puberty in girls with MAS and its occurrence in boys. One distinction is that precocious puberty is far rarer in affected boys, who are more often diagnosed due to bone disease or *café-au-lait* pigmentation. Additionally, when precocious puberty does occur in boys, it tends to be subtle and indolent. Finally, in some reported cases, the activating Gαs mutation and resulting gonadal hyperfunction have been limited to testicular Sertoli cells in boys with MAS [16].

FD is a condition of underdeveloped and insufficiently calcified bone due to a benign intraosseous disorder that results from fibrous connective tissue replacement of medullary bone [17]. Lichtenstein, in 1938, first used the term FD. Three different phenotypes of FD were recognized, including monostotic FD, involving a single bone and affecting about 70-80% of cases; the second form is polyostotic FD, affecting multiple bones, in about 20-30% of cases; and the third one occurs when the polyostotic form is accompanied by endocrinal and skin findings and is called MAS [18, 19].

The most frequently affected craniofacial bone is the maxilla (58%), followed by the mandible (43%), frontal (33%), sphenoidal (29%), ethmoidal (24%), parietal (14%), temporal (5%), and occipital bones (3-4%) [13].

Typically, FD manifests itself throughout the first 30 years of life. Approximately 60% of patients with polyostotic FD experience symptoms before the age of 10. However, it's also not uncommon for symptoms to manifest in older age. Equal prevalence has been reported regarding the gender distribution of FD [14].

Visual impairment affects 20-35% of patients with craniofacial FD [20], which includes poor visual acuity, abnormalities in the central and peripheral fields, and loss of color vision. Immature bone tissue extension into the optic canal causes increasing optic nerve compression, which is the primary source of visual impairment in patients with craniofacial FD [14, 21, 22].

In the present case, the diagnosis was made around the age of 3 years, but symptoms of visual impairment appeared 9 years later. Various studies indicate that visual impairment in FD can result from optic nerve traction, spontaneous bleeding, or optic canal compression caused by a separate cystic lesion, such as a mucocele [14, 23, 24]. In this particular case, the visual impairment was attributed to optic nerve compression due to the expansion of bone lesions into the optic canal. The potential benefits of transcranial therapeutic interventions have been demonstrated in numerous studies; however, the role and appropriateness of less invasive approaches, such as endoscopic decompression, remain under investigation.

Although endoscopic decompression is used to treat traumatic optic neuropathy, it is still a debatable indication because several studies have not shown clear evidence of improvement [25].

In contrast to traumatic optic neuropathy, endoscopic decompression of the optic nerve for other benign diseases that cause canalicular segment compressive optic neuropathy has only been recorded in small case series or case reports [26]. In a similar study, a 19-year-old male with MAS underwent endoscopic trans-nasal trans-sphenoidal and trans-ethmoidal optic canal decompression due to compressive optic neuropathy. The successful outcome aligns with existing literature, which supports the efficacy of

endoscopic endonasal optic nerve decompression in preventing visual deterioration in patients with FD-related optic neuropathy [27].

Although they are uncommon, endoscopic transnasal decompression may result in nasal morbidity, carotid damage, deteriorating vision or blindness, and cerebrospinal fluid leakage (which can be prevented by avoiding nerve sheath incisions) [26]. None of these complications occurred in our patient.

The minimal improvement in the vision of this boy may be due to a delay in the operation after the start of vision deterioration; however, more follow-up time is needed to decide the degree of improvement, with a monthly visual field evaluation of both eyes and repeated optic nerve CT/MRI imaging at the onset of symptoms of visual deterioration.

The role of surgical intervention in optic neuropathy due to FD remains debated, particularly regarding the timing of decompression. Some studies advocate prophylactic surgery in cases of radiographic compression, while others recommend intervention only after the onset of visual symptoms [27]. A study where symptomatic visual decline prompted urgent surgical decompression reinforces the recommendations that optic nerve decompression should be performed as soon as vision is affected. Studies further suggest that image-guided navigation enhances the precision of endoscopic procedures, minimizing morbidity while optimizing surgical outcomes [28].

Our study supports the growing consensus that endoscopic decompression is a safe and effective treatment for optic nerve compression in MAS, particularly when performed before irreversible optic neuropathy occurs. Future studies should further investigate long-term visual outcomes and the potential role of prophylactic intervention in high-risk cases.

Conclusion

FD is a rare disorder that can cause compressive optic neuropathy, potentially leading to vision loss. Endoscopic transnasal optic nerve decompression is a safe and effective treatment for these patients. However, the limited visual improvement in our case could be due to delayed intervention. Further studies are needed to confirm the safety and efficacy of early surgical intervention for optic neuropathy related to MAS.

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Declaration of interest

The Authors declare that there is no conflict of interest. Funding: no funding.

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