

## CASE REPORT

# Maffucci Syndrome with Nasal Enchondroma: A Rare Entity

<sup>1</sup>Shruti Bansal, <sup>2</sup>Arpit Sharma, <sup>3</sup>JP Dabholkar

## ABSTRACT

**Introduction:** Maffucci syndrome is a congenital nonhereditary disorder in which patients develop multiple enchondromas and cutaneous, visceral, or soft tissue hemangiomas. Less than 200 cases of Maffucci syndrome have been published in the English literature. These lesions most commonly occur in the limb bones, especially in the hands and feet. Rarely, patients of Maffucci syndrome may present with mesodermal dysplasia involving head and neck region with nasal septum enchondromas reported only in two cases till now.

**Aim:** To report a rare case of nasal enchondroma in a patient of Maffucci syndrome.

**Case description:** A 15-year-old girl diagnosed with Maffucci syndrome 2 years back was referred to our ENT department with symptoms of left-sided nasal obstruction and epiphora. On thorough clinical examination and proper investigation, she was diagnosed to have nasal enchondroma. She successfully underwent endonasal endoscopic resection of nasal enchondroma.

**Conclusion:** Maffucci syndrome is a rare entity characterized by multiple enchondromas and hemangiomas, especially in the extremities. A manifestation in the head and neck region is rare and cartilaginous tumors of nasal septum are even rarer; however, differential of nasal cartilaginous tumors should be borne in mind in patients with underlying mesodermal dysplastic disorders like Maffucci syndrome.

**Clinical significance:** Patients of Maffucci syndrome with enchondromas are at high risk of undergoing malignant transformation, especially chondrosarcomas. These patients need vigilant and close follow-up. Despite their rarity, chondromas should be taken into consideration in the differential diagnosis of nasal tumors, especially those arising from the nasal septum.

**Keywords:** Maffucci syndrome, Mesodermal dysplasia, Nasal enchondromas.

**How to cite this article:** Bansal S, Sharma A, Dabholkar JP. Maffucci Syndrome with Nasal Enchondroma: A Rare Entity. Clin Rhinol An Int J 2016;9(2):94-97.

**Source of support:** Nil

**Conflict of interest:** None

<sup>1</sup>Medical Officer, <sup>2</sup>Assistant Professor, <sup>3</sup>Professor and Head

<sup>1-3</sup>Department of ENT and Head and Neck Surgery, King Edward Memorial Hospital and Seth Gordhandas Sunderdas Medical College, Mumbai, Maharashtra, India

**Corresponding Author:** Shruti Bansal, Medical Officer Department of ENT and Head and Neck Surgery, King Edward Memorial Hospital and Seth Gordhandas Sunderdas Medical College, Mumbai, Maharashtra, India, e-mail: drshrutibansal1706@gmail.com

## INTRODUCTION

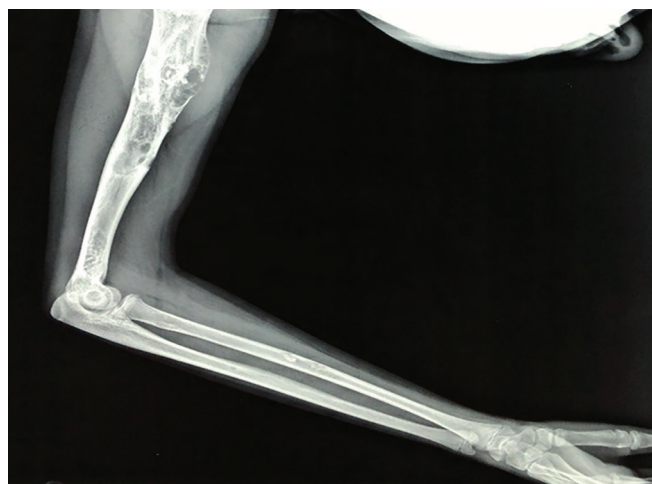
Maffucci's syndrome is an extremely rare disease characterized by multiple enchondromas and cutaneous hemangiomas.<sup>1</sup> It is a congenital nonhereditary mesodermal dysplasia causing enchondromas usually near to the growth plate cartilage of extremities. The first case of this syndrome was described by Angelo Maffucci in 1881<sup>2</sup> and eponym, Maffucci's syndrome was later proposed by Carlton et al.<sup>3</sup> Less than 200 cases have been reported in the literature till now.<sup>3</sup> A manifestation in the head and neck region is rare with even rarer involvement of nasal cavity. There are only two case reports that describe a primary involvement of the nasal cavities and paranasal sinuses (PNS).<sup>4,5</sup> It results from deregulated proliferation and differentiation of chondrocytes during physiological enchondral ossification.<sup>3</sup> Complications include spontaneous fracture, resulting in skeletal deformity and high risk of malignant transformation in up to 40% of the cases.<sup>3,6,7</sup> Chondrosarcoma is the most common neoplasm in this syndrome.<sup>1</sup> Classical clinical symptoms of cartilaginous nasal tumors include nasal mass, obstruction, headache, and epistaxis.<sup>8</sup> The treatment of choice is wide surgical excision.

## CASE REPORT

A 15-year-old girl presented to our department with chief complaints of left-sided nasal obstruction and increased watering from left eye. She was diagnosed as a case of Maffucci syndrome at the age of 13 years when she developed multinodular outgrowths on the dorsum and ventral surface of the foot (Fig. 1). She underwent a series of investigation for the same in the form of radiography (X-rays) and magnetic resonance imaging (MRI). She was found to have multiple expansile lytic lesions with coarse and punctate calcifications involving the small bones of foot and long tubular bone of humerus and femur suggestive of enchondromas (Figs 2 and 3). On detailed examination and investigations, she was diagnosed to have associated splenomegaly. Splenic biopsy was done and was suggestive of splenic hemangiomas. She was thus diagnosed as a case of Maffucci syndrome with characteristic features of multiple enchondromas and splenic hemangiomas.



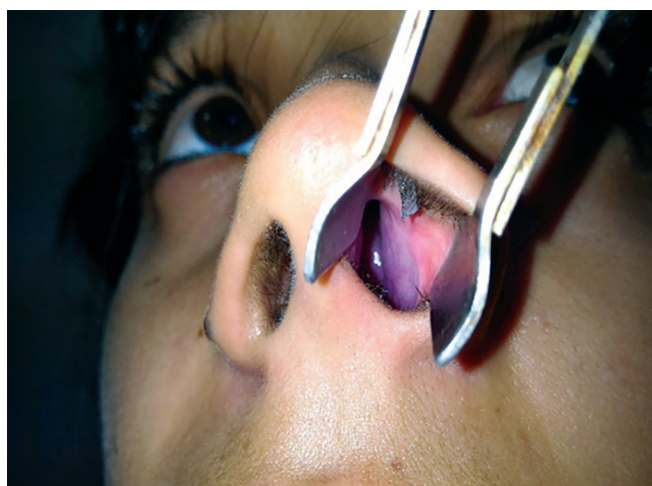
**Fig. 1:** Multiple nodular outgrowths on foot in a case of Maffucci syndrome



**Fig. 2:** Expansile lytic lesion with coarse and punctate calcifications in shaft of humerus



**Fig. 3:** Expansile lesions with ground glass appearance in proximal and distal phalanx of third and ring figure



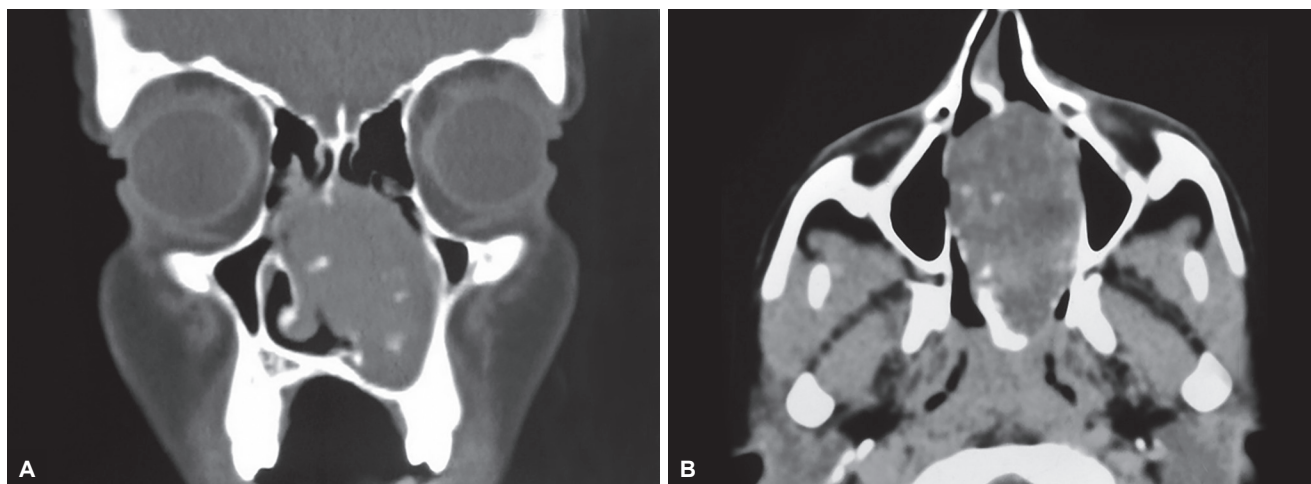
**Fig. 4:** Pale smooth nasal mass seen on anterior rhinoscopy in left nasal cavity

Two years later she presented to our Department of ENT with chief complaints of left-sided nasal obstruction and increased watering from left eye. On nasal examination we found a smooth firm nasal mass which appeared to be arising from nasal septum (Fig. 4). The endoscopic evaluation revealed a large mass in the left nasal cavity with no signs of inflammation or destruction of surrounding tissue suggestive of benign lesion arising from nasal septum which was suspected to be nasal enchondroma in a diagnosed case of Maffucci syndrome. To the best of our knowledge, this was one of the rarest case of nasal enchondroma in a patient of Maffucci syndrome with only two such cases reported in the literature till now.<sup>4,5</sup> We got a computed tomography (CT) of paranasal sinuses PNS, which confirmed our clinical findings. The CT-PNS was suggestive of well-defined expansile nasal mass approximately 3×2 cm in size in left nasal cavity with epicenter in nasal septum, pushing on left lateral nasal wall with smooth pressure erosions on left half of

hard palate and right-sided middle and inferior turbinate (Figs 5A and B). The tumor was resected via a transnasal endoscopic approach with the help of computer-assisted navigation. During surgery, the smooth and lobulated tumor was found to have infiltrated the nasal septum and the anterior wall of the sphenoid sinus. Consequently, the posterior part of the nasal septum and the anterior wall of the sphenoid sinus had to be removed. Specimen was sent for histopathological examination. Postoperative hospital performance report confirmed the diagnosis of nasal enchondroma. Since it was a benign cartilaginous tumor, no additional treatment was given.

## DISCUSSION

Maffucci syndrome is a rare genetic disorder characterized by benign enchondromas, bone deformities, and irregularly shaped hemangiomas. In 2011, the cause of Maffucci syndrome was discovered to be a mutation in a gene known as IDH1 (rarely IDH2).<sup>7</sup> There is no racial



**Figs 5A and B:** 3×2 cm well-defined mass in left half of nasal cavity with smooth pressure erosions of left maxilla, left half of hard palate, right middle and inferior turbinate

or sexual predilection in Maffucci syndrome. No familial pattern of inheritance has been shown, but Maffucci syndrome manifests early in life, usually around 4 and 5 years of age, with 25% of cases being congenital. The lesions most commonly occur in the limb bones, especially in the hands and feet; however, they may also occur in the skull, ribs, and vertebrae. Enchondromas may result in severe bone deformities, shortening of the limbs, and fractures. The affected individuals are at high risk of developing bone cancers called chondrosarcomas. Manifestation of Maffucci syndrome in the head and neck region is rare. Chondroma of the nasal septum is in itself a very rare entity. Till now, only two cases have been reported in the literature that describe cartilaginous tumor of the nasal cavities in a case of Maffucci syndrome. The symptoms of nasal septum chondroma are nasal obstruction, headache, and epistaxis. The treatment of choice for nasal chondroma is wide surgical excision.

The diagnosis of Maffucci syndrome is made by a detailed history, thorough physical examination, and radiologic assessment, i.e., plain radiographs, CT scans, and MRI. Surgical removal and microscopic study of the bony lesions confirm the presence of enchondroma and distinguish the tumor from chondrosarcoma.

Management of Maffucci syndrome is focused on the specific signs/symptoms in the particular affected individual. No intervention is needed for asymptomatic patients. Enchondromas can be surgically removed (resected) if necessary. A patient with Maffucci syndrome should be regularly monitored because of the risk of malignant transformation of an enchondroma or development of a tumor elsewhere.

## CONCLUSION

Maffucci syndrome is an exceedingly rare mesodermal dysplasia. Its manifestation in the head and neck region is

less common and enchondroma involving nasal septum and PNS is even rarer. A patient of Maffucci's syndrome needs vigilant observation for the development of sarcomatous lesions which is known to occur in almost 40% of cases. Malignant transformation of the associated enchondromas into chondrosarcomas is common, and should be considered whenever a change in the clinical course like pain or sudden increase in size occurs. No intervention is needed for asymptomatic patients. However, enchondromas can be surgically resected if necessary. Through this publication we take pleasure to report a very rare case of Maffucci syndrome presenting with enchondroma of nasal septum.

## CLINICAL SIGNIFICANCE

Patients of Maffucci syndrome with enchondromas are at high risk of undergoing malignant transformation specially chondrosarcomas. These patients need vigilant and close follow-up. Despite their rarity, chondromas should be taken into consideration in the differential diagnosis of nasal tumors, especially those arising from the nasal septum. These patients can be successfully treated with transnasal endoscopic resection.

## ETHICAL STANDARDS

The authors assert that all procedures contributing to this work comply with the ethical standards of the relevant national and institutional guidelines on human experimentation and with the Helsinki Declaration of 1975, as revised in 2008.

## REFERENCES

1. Verdegaal SH, Bovée JV, Pansuriya TC, Grimer RJ, Ozger H, Jutte PC, San Julian M, Biau DJ, van der Geest IC, Leithner A, et al. Incidence, predictive factors, and prognosis of chondrosarcoma in patients with Ollier disease and Maffucci



- syndrome: an international multicenter study of 161 patients. *Oncologist* 2011;16(12):1771-1779.
2. Gao H, Wang B, Zhang X, Liu F, Lu Y. Maffucci syndrome with unilateral limb: a case report and review of the literature. *Chin J Cancer Res* 2013 Apr;25(2):254-258.
3. Carleton A, Elkington J, Greenfield JG, et al. Maffucci's syndrome (dyschondroplasia with hemangiomata). *Q J Med* 1942;35:203-228.
4. Jacobi CM, Hiranya ES, Gay A, Holzmann D, Kollias S, Soyka M. Enchondroma of the nasal septum due to Ollier disease: a case report and review of the literature. *Head Neck* 2015 Mar;37(3):E30-E33.
5. Hyde GE, Yarrington CT Jr, Chu FW. Head and neck manifestations of Maffucci's syndrome: chondrosarcoma of the nasal septum. *Am J Otolaryngol* 1995 Jul-Aug;16(4):272-275.
6. Garzon MC, Huang JT, Enjolras O, Frieden IJ. Vascular malformations. Part II: associated syndromes. *J Am Acad Dermatol* 2007 Apr;56(4):541-564.
7. Pansuriya TC, Van Eijk R, d'Adamo P, van Ruler MA, Kuijjer ML, Oosting J, Cleton-Jansen AM, van Oosterwijk JG, Verbeke SL, Meijer D, et al. Somatic mosaic IDH1 or IDH2 mutations are associated with enchondroma and spindle cell hemangioma in Ollier disease and Maffucci syndrome. *Nat Genet* 2012 Nov 6;43(12):1256-1261.
8. Scierski W, Namysłowski G, Misiołek M, Widziszowska A, Hajduk A. Septal nasal chondroma – a case report. *Otolaryngol Pol* 2007;61(6):1021-1023.