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Case Report

Maffucci syndrome: Case report and review of diagnostic signs of the rare disease ☆,☆☆

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ABSTRACT

Maffucci syndrome is a non-hereditary congenital condition that affects the skin and skeleton. Enchondromas (benign cartilage enlargements), bone abnormalities, and venous anomalies (hemangiomas) are all symptoms. Enchondromas occur as a result of mesodermal dysplasia and have the potential to become cancerous. They are most commonly found on the phalanges and long bones. Venous abnormalities commonly manifest themselves as soft lumps or tumors on the distal extremities. A 19-year-old boy presented with swellings on his fingers and left foot since the age of 5, along with a few bluish soft tissue swellings on his left heel. Multiple expansile lytic lesions and soft tissue swellings with phleboliths were seen on X-ray. Histology confirmed the diagnosis of hemangiomas and enchondromas. Soft tissue swellings were found to have hyper echoic areas, as well as modest marginal blood flow on Doppler, which could indicate hemangiomas. Maffucci syndrome was identified, and treatment with a multidisciplinary approach was initiated. Maffucci syndrome is a rare genetic illness reported in the literature less than 200 times. The enchondromas and hemangiomas have a strong link to malignant changes, with chondrosarcomas accounting for 30% of the associated malignancies. On X-ray, enchondromas are easily identified as osteolytic lesions with cortex thinning and endosteal scalloping while color Doppler ultrasound detects the presence of hemangiomas. Phleboliths are easily identified as small calcifications on X-rays. Radiographic examinations should be considered in patients presenting with bone or soft tissue swellings for an early diagnosis of Maffucci syndrome.

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Introduction

Maffucci syndrome is an extremely rare congenital disease characterized by enchondromas and venous abnormalities like soft issue hemangiomas and lymphangiomas [1]. Since its first description in 1881, by Italian physician Angelo Maffucci, less than 200 cases have ever been reported in literature. Maffucci syndrome has been linked to somatic mutations in the isocitrate dehydrogenase 1 and 2 (IDH1 and 2) genes. It normally appears at the age of 4-5 years old and has no sexual or ethnic predisposition, or a genetic pattern of inheritance [2–4].

Case presentation

A 19-year-old male patient complained of swellings on the fingers of both hands and the left foot, as well as a few bluish soft tissue swellings on the left heel (Fig. 1). The swellings began to form at the age of 5 years and had gradually increased in size. For bony lesions, an X-ray of both hands was done. Ultrasound and fine needle aspiration biopsy was performed for soft tissue swellings.

X-ray

Both hands and feet were X-rayed (Fig. 2). Multiple radiolucent, expansile, and osteolytic lesions with bone deformities were found in the phalanges and metacarpals of both hands (Figs. 2A and B), as well as the first metatarsal of the left foot (Fig. 2C). Cortex thinning and endosteal scalloping were also observed, with few areas of complete absence of cortex. Furthermore, well-defined, round calcifications in the soft tissue masses characteristic of phleboliths were observed (Fig. 2D).

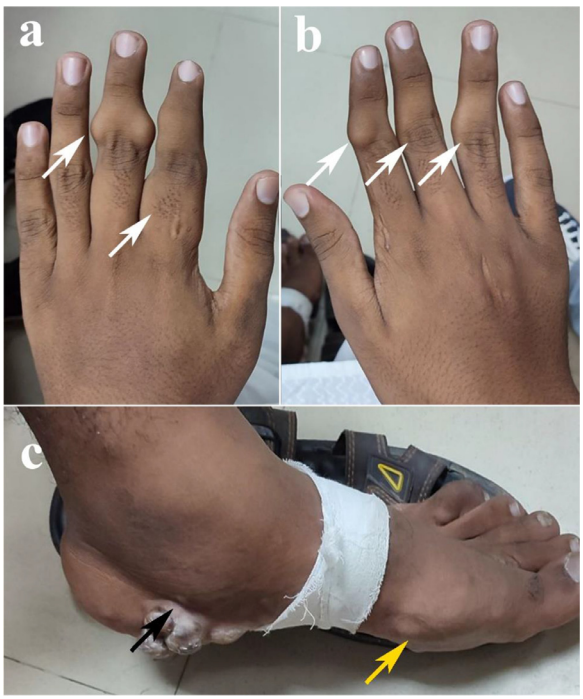

Fig. 1 – Arrows indicate bony swellings on index and middle fingers of left hand. (A) Bony swellings on index, middle and ring fingers of right hand (B). Bony swelling on medial side of foot just posterior to big toe (yellow arrow) and soft tissue swellings on inferior-medial side of medial malleolus (black arrow).

Gray-scale and Doppler ultrasound

A high frequency linear probe was used to perform an ultrasound of soft tissues on the medial side of the left foot. Ultrasound revealed multiple well-defined hyper echoic areas with mild marginal blood flow on color Doppler, indicating hemangiomas (Fig. 3).

Fine needle aspiration biopsy

Biopsy of soft tissue from swellings on the medial side of left foot and bony swelling from right hand was taken. The histologic sections of soft tissue revealed histology diagnostic of hemangioma (Fig. 4A), whereas sections from the bony swellings revealed chondroid tissue diagnostic of enchondromas (Fig. 4B).

Thus, on basis of the clinical, radiological, and histopathologic tests, Maffucci syndrome was diagnosed. The patient and his family were informed of his current condition and possible complications and the potential of malignant transformation [2]. The enlarged soft tissue swellings on the medial side of the left foot were surgically removed, and regular follow-up visits were recommended.

Discussion

Maffucci syndrome is distinguished by the presence of multiple (more than 3) enchondromas with vascular anomalies. There have only been 200 cases of Maffucci syndrome reported in the literature. Maffucci syndrome affects less than 1 in every 100,000 people [2,5]. Malignant transformations occur at a rate of 52%-57% in Maffucci syndrome. Chondrosarcomas account for 30% of the malignant lesions in Maffucci syndrome. The median time for symptom onset is 4 years. It has an equal gender distribution and no racial preference [4,5]. Bony lesions mostly appear in the metaphysis of short tubular bones such as metacarpals and phalanges of hands (89.3%), and infrequently in the short tubular bones of feet (35.7%) and long tubular bones of upper and lower limbs. Generalized mesodermal dysplasia occurs when the bone grows and enlarges in length but the cartilages lags behind and grows asymmetrically resulting in enchondromas formation. Multicentric radiolucent areas with significant cortical thinning is the pathognomic sign on X-ray [2,4].

Hemangiomas in Maffucci syndrome appear as bluish soft subcutaneous nodules that can be blanched with pressure. Phleboliths, which appear as distinct round calcifications in soft tissues on X-ray, are an important diagnostic criteria for Maffucci syndrome. These calcifications are prone to forming micro thrombi. Hemangiomas occur at a rate of 32% in the hands and 23% in the feet [2,4].

Maffucci syndrome is a rare condition, but it should be considered in the differentials of congenital hemangiomas.

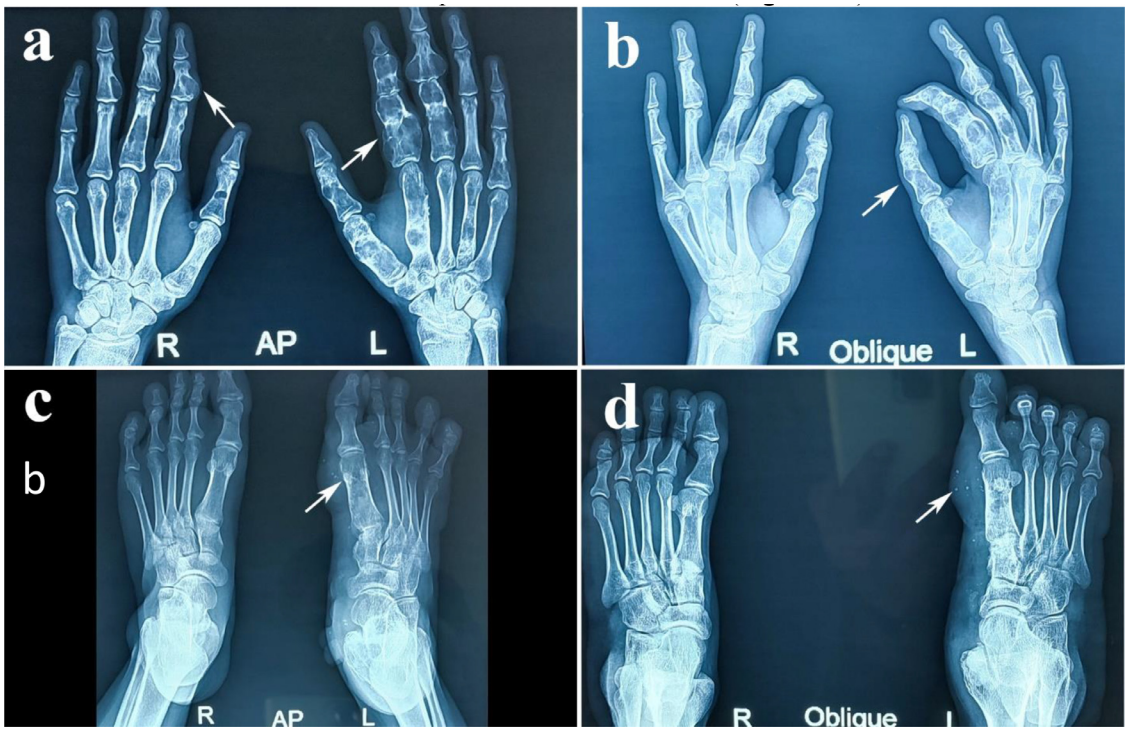

Fig. 2 – X-rays of hand and feet showing bony and soft tissue lesions. A) X-ray both hands AP view showing multiple osteolytic lesions in phalanges and metacarpals of both hands as indicated by arrows. B) X-ray both hands oblique view showing multiple osteolytic lesions in phalanges and metacarpals of both hands, indicated by arrow. C) X-ray both feet AP view showing similar findings as in hands. D) X-ray both feet oblique view showing microcalcifications in the soft tissue concerning for phleboliths, indicated by arrow.

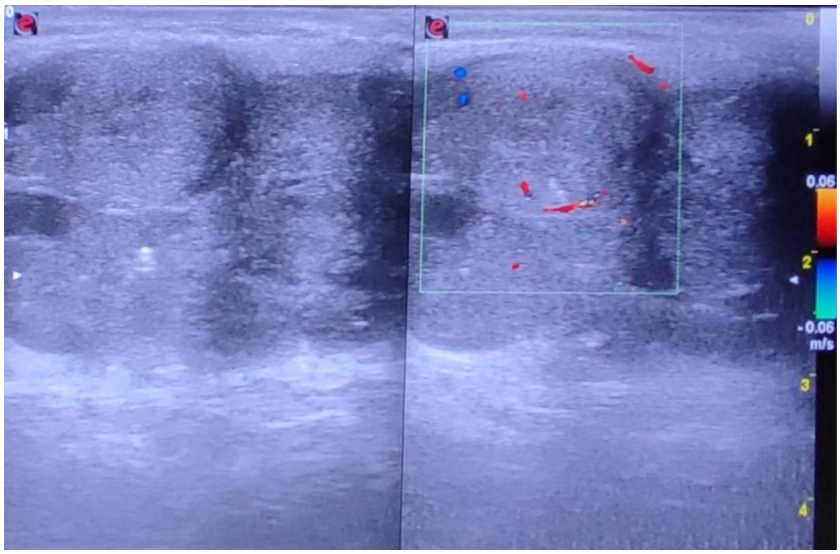

Fig. 3 – Doppler ultrasound of soft tissue swelling.

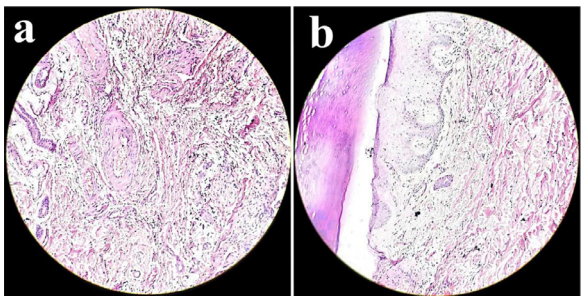

Fig. 4 – Hemangioma: anastomosing vascular channels lined by plump endothelial cells. (A) Enchondroma: chondroid tissue containing lobules of chondrocytes. (B) Chondroid tissue containing lobules of chondrocytes concerning for enchodroma formation.

The presence of radiological signs, as well as clinical and pathological evidence of enchondromas and hemangiomas, is critical in the diagnosis of Maffucci syndrome [2]. Maffucci syndrome can cause a variety of complications, including pathological fractures, skeletal deformities, vascular overgrowth, limb-length discrepancies, and malignant transformations.

Maffucci syndrome must be differentiated from Ollier disease, another condition characterized by enchondroma. Ollier disease is characterized by enchondromas that are characteristically unilateral and by the absence of hemangiomas. Other differentials of Maffucci syndrome are blue rubber bleb nevus syndrome, Gorham's syndrome (bone and soft tissue angiomatous lesions) and Kaposi's sarcoma [4].

The management aim of Maffucci syndrome is symptomatic relief and early recognition of disease so that complications can be prevented. For complications like vascular overgrowth, enlarged enchondromas, and limb-length discrepancies, surgical correction and removal of enchondromas is the only treatment possibility. Due to risk of malignant transformation, regular follow-up is an absolute necessity [5,6].

Conclusion

Enchondromas and hemangiomas are the main features of Maffucci syndrome which can be identified on the basis radiological investigations. It is important to consider radiological investigations in patients manifesting with bony and soft tissue swellings to make an early diagnosis so that timely management may prevent bony deformities and other complications. Due to risk of malignant transformation in patients of Maffucci syndrome, regular follow-up must be assured.

Authors' contributions

MTK conducted literature review, drafted initial document, created images, and amended the final draft. SA oversaw the research and helped with revision. MUM and RR revised the manuscript and edited images. The final version of the manuscript was approved by all authors.

Availability of data and materials

All relevant data and material has been provided with the manuscript.

Ethics approval and consent to participate

Dr. Sadaf Arooj, the Head of the Department of Diagnostic Radiology at the Punjab Institute of Neuroscience in Lahore, Pakistan, provided ethical approval. Written informed consent was obtained. This research follows the most recent version of the Helsinki Declaration.

Consent for publication

The patient and his parents gave written informed consent for the publication of this case and the necessary radiological pictures.

Patient consent

I would like to declare that I have taken a written consent from the patient for publication of his case as case report to the Radiology Case Report Journal.

If required, the consent form can be made available.

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