Maffucci Syndrome- A Rare Case Report.

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Abstract

A 30 year male presented with multiple swellings of both hands which he noticed at the age of 4. On examination both his hands had firm, non-tender, multiple swellings, making it difficult for him to make fists. He had multiple diffuse bluish compressible swellings on both forearm. X ray confirmed multiple enchondromas in both hands. Large tumors in both hands were curetted. Soft tissue swelling from forearm was also excised. Both the specimens were sent for Histopathological Examination. One piece of forearm swelling was harder and was x-rayed which confirmed a calcified tissue as phelebolith. Histopathological report confirmed the presence of spindle cell haemangioendothelioma with enchondromas; the two pathological entities associated with Maffucci syndrome.

Introduction

Enchondroma with multiple angiomas (Maffucci syndrome) was first reported by Maffucci in 1881 after a 40-year-old woman died from complications following amputation of an arm. Maffucci reported a thorough autopsy that described all the main points of the syndrome named after him. In 1941, Carleton et al proposed the eponym Maffucci syndrome. Maffucci syndrome is a rare genetic disorder that affects both males and females. Maffucci syndrome is characterized by benign enlargements of cartilage (enchondromas); bone deformities; and dark, irregularly shaped hemangiomas. No racial or sexual predilection is apparent in Maffucci syndrome. No familial pattern of inheritance has been shown, but Maffucci syndrome manifests early in life, usually around age 4-5 years, with 25% of cases being congenital. Maffucci syndrome appears to develop from mesodermal dysplasia early in life. Patients apparently are of average intelligence, and no associated mental or psychiatric abnormalities seem to be present.

Case Report(s)

As the swellings increased in size, hands appeared ugly and routine hand function started deteriorating. These forced him to take medical advice. On examination both his hands had firm, non-tender, multiple swellings, making it difficult for him to make fists. He had multiple diffuse bluish compressible swellings on both fore arms. X ray confirmed multiple enchondromas in both hands (Fig. 2,3). He was operated upon large tumours in both hand, were curetted (Fig.4). Soft tissue swelling from forearm was also excised. This improved his hand appearance and function. Tissues were sent for histopathological examination. One piece of forearm swelling was harder and was x rayed which confirmed a calcified tissue as phelebolith (Fig.5). Histopathological report confirmed the presence of spindle cell haemangioendothelioma with enchondromas(Fig. 6,7,8) the two pathological entities associated with Maffucci syndrome there were no malignant changes in the bone or soft tissue material.

Discussion

The syndrome of multiple enchondromatosis and subcutaneous haemangiomas was first described by Maffucci in 1881. Since then nearly 200 cases have been described in medical literature. Its etiology is unknown and does not result from gross chromosomal abnormality. Maffucci described all the essential features of the disease, onset in childhood or adolescence, lack of family history. Multiple enchondromas leading to shortening and deformity of extremities, multiple haemangiomas and malignant changes in skeletal lesions. It occurs equally in men and women in all races and distal parts of limb are commonly affected. Sarcomatous deneration has been found to in 18% cases in review of literature. Surgical intervention is required in these cases in the form of reconstruction and corrective surgery and amputation of grossly affected parts to improve function is required. Therapy for vascular lesions is primarily surgical with varying degrees of success. Radiation is not recommended.

Conclusion

A case of Maffucci syndrome in a male of 30 years is described. Histopathologic examination did not reveal
any malignant changes in either bone or vascular lesions. Pathologic calcification of haemangioma was found. Surgery was performed to improve shape and functions of fingers.

References

Illustrations

Illustration 1

Clinical Picture showing multiple swellings of hand, fingers and forearm with discoloration of skin.

Illustration 2

X Ray showing Enchondroma of 2nd and 4th Ray.
Illustration 3

X Ray showing Enchondroma of proximal phalanges of thumb and index finger

Illustration 4

Intra-operative picture showing curettage of enchondroma.
Illustration 5

Microphotography showing osteoclastic giant cell rimming of the bony trabeculae of the overlying bone in Enchondroma.

Illustration 6

Microphotograph of spindle cell Haemangio-Endothelioma showing vascular channels lined by endothelial cells and stroma contains spindle cell with mitotic activity.
Illustration 7

Microphotograph of Enchondroma showing benign cells arranged in lobulated appearance.

Illustration 8

Calcified Phlebolith.
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