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Maffucci's Syndrome Associated with Chondrosarcoma and Aneurysm: Case Report[1]

Hyoung Gun Lim, M.D., Won Jong Yoo, M.D., Yeon Soo Lim, M.D., Mi Sook Sung, M.D., Myung Hee Chung, M.D., Hae Giu Lee, M.D., So Lyung Jung, M.D., Jea Na Kim, M.D.[2]

Maffucci syndrome is a rare congenital non-inherited condition characterized by multiple enchondromas and cutaneous hemangiomas. It is associated with increased risk of malignancy, including chondrosarcomas, and because of generalized mesodermal dysplasia, aneurysms can develop. We present a case of Maffucci syndrome associated with intracranial chondrosarcoma and aneurysm.

Index words : Maffucci syndrome
Enchondroma
Hemangioma
Chondrosarcoma
Aneurysm, intracranial

Maffucci syndrome is a congenital, non-hereditary form of mesodermal dysplasia associated with multiple enchondromas and soft tissue hemangiomas, and usually occurs in childhood or adolescence. Malignant transformation of the enchondroma has not infrequently been reported. Review of 170 reported cases of the syndrome noted the occurrence of six intracranial chondrosarcomas and two intracranial aneurysms (1 - 3). To the best of our knowledge, this is the first report in which Maffucci syndrome has been associated with intracranial chondrosarcoma and aneurysm.

Case Report

A 32-year-old male patient was admitted with diplopia at the left lateral gaze and headaches, which had first occurred three weeks earlier. At physical examination, multiple non-tender bluish subcutaneous masses were palpable in the left hand and foot. Plain radiography demonstrated multiple cystic lesions in the left phalanges, suggesting enchondromas (Fig. 1A), and T2-weighted MR imaging of the foot revealed multiple subcutaneous hyperintense masses with central hypointensities representing hemangiomas with phleboliths (Fig. 1B). MRI of the brain showed that in the left parasellar region, a homogeneous mass, hypointense on T1-weighted images and hyperintense on T2-weighted images, was present (Fig. 1C), and after intravenous infusion of gadolinium-DTPA, reticular and septal enhancement was observed (Fig. 1D). Enhanced coronal CT depicted a marginally enhanced bulging mass in the left lateral aspect of the clivus, and angiography revealed mild lateral displacement of the left internal carotid artery by the mass. A small aneurysm of the left internal carotid artery was discovered incidentally near the origin of the ophthalmic artery (Fig. 1F). Two weeks later, transzygomatic craniotomy was performed and the tumor was partially removed. Histopathologic examina-

[1]Department of Radiology, College of Medicine, The Catholic University of Korea
[2]Department of Pathology, College of Medicine, The Catholic University of Korea
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Address reprint requests to : Won Jong Yoo, M.D., Department of Radiology, Holy Family Hospital, The Catholic University of Korea, 2 Sosa-dong, Wonmi-gu, Puchun, Kyunggi-do 420-717, Korea.
Tel. 82-32-340-2185 Fax. 82-32-340-2187 E-mail: wjyu@hfh.cuk.ac.kr

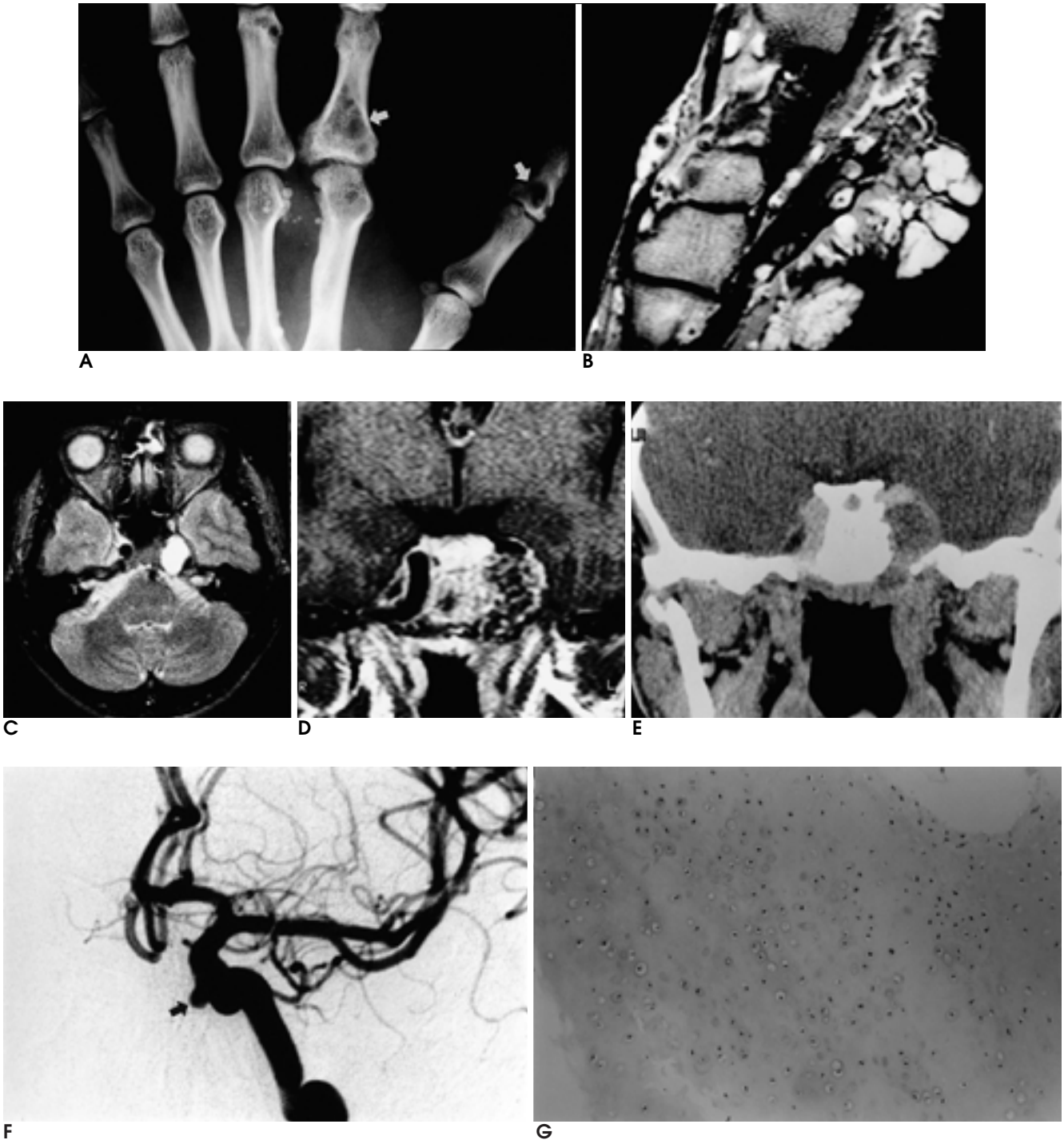

Fig. 1. A 32-year-old male with multiple soft tissue masses in the left hand and foot, headache, and diplopia at the lateral gaze.
A. A plain radiograph shows multiple enchondromas (arrows) in the phlanges and hemangiomas with phleboliths in the left hand.
B. Sagital T2-weighted MR image of the left foot shows multiple variable sized hyperintense masses in the subcutaneous tissue with signal voids, representing hemangiomas with phleboliths.
C. Axial T2-weighted MR image shows the homogeneous hyperintense mass in the left lateral aspect of the clivus.
D. Coronal T1-weighted MR image with gadolinium-DTPA shows irregular septal enhancement of the mass.
E. Contrast enhanced coronal CT scan shows inhomogeneous low density mass and bony destruction in the left lateral aspect of the clivus with marginal enhancement.
F. Angiogram of the left internal carotid artery shows saccular aneurysm(arrow) near the origin of the ophthalmic artery.
G. Grade 1 chondrosarcoma involving the left parasellar region. It shows increased cellularity, mild hyperchromasia, and pleomorphism of the nuclei. (H & E, ×100).

tion showed that the specimen had a chondroid matrix, with chondrocytes in the lacunae, relatively increased cellularity, and moderate cytologic atypia of the tumor cells (Fig. 1G), indicating chondrosarcoma. Immunohistochemical study was positive for S-100 protein and negative for epithelial membrane antigen and cytokeratin. Postoperatively, the patient was relatively well.

Discussion

Maffucci syndrome was first described by Maffucci in 1881. It is characterized by generalized mesodermal dysplasia and a strong propensity for malignant transformation. The syndrome occurs during childhood or adolescence, there is a lack of familial history, and the clinical features include multiple enchondromas leading to the shortening and deformity of the extremities, multiple hemangiomas, and malignant transformation of the skeletal lesions (4).

Enchondromas in Maffucci syndrome represent abnormal cartilaginous development (endochondral ossification). They are depicted at plain radiography as multiple cystic radiolucencies in long and flat bones, with the metacarpals and phalanges of the hand most frequently involved.

The typical vascular lesion in Maffucci syndrome is a hemangioma, which is typically classified as cavernous, capillary, or the mixed venous/capillary type. The distribution of hemangiomas is more extensive on one side of the body, commonly in the hand and foot ; in our case, left-side development occurred. In dilated vessels and vascular spaces, calcification may occur, with the formation of phleboliths.

The reported incidence of malignancy in this syndrome varies. Lewis and Ketcham (4) reported an overall incidence of approximately 23%, and sarcomatous transformation of enchondroma of 15%. Kaplan et al. (5) reported an overall malignancy rate of 37%, with 30% of patients developing chondrosarcoma.

Because this syndrome involves a generalized mesodermal dysplasia, a wide variety of non-cartilaginous tumors can arise from fibrous, bone, and vascular tissue, and non-mesodermal tumors such as astrocytoma, glioma, and pituitary adenoma may also occur (4, 5). However, the most common serious complication of Maffucci syndrome is chondrosarcoma, which probably arises from an enchondroma. The majority of chondrosarcomas are of low histologic grade, but because of the difficulty in differentiating between low-grade chondrosarcoma and atypical enchondroma, MRI and biopsy should be performed. The prognosis of Maffucci syndrome in patients with chondrosarcoma is considered poor. The choice of treatment for chondrosarcoma is surgical resection; for intracranial chondrosarcomas, radiotherapy and chemotherapy are usually not indicated (2).

Several plain radiographic findings suggesting malignant transformation have been described (6): areas of endosteal or cortical erosion, frank cortical destruction, an associated soft-tissue mass extending beyond the area of cortical disruption, and a zone of developing lucency within a previously mineralized cartilaginous lesion.

The MR findings of cartilaginous tumors such as enchondroma and chondrosarcoma are low signal intensity on T1-weighted imaging, high signal intensity on T2-weighted imaging, lobular margins, internal septations, and punctate signal voids representing calcification or ossification (7). Differentiation between enchondroma and low-grade chondrosarcoma on the basis of MR imaging is sometimes difficult. In a review of 27 cases of low-grade chondrosarcoma with histopathologic correlation (8), low-grade chondrosarcomas showed fibrovascular septal enhancement, high-grade chondrosarcomas showed inhomogeneous or homogeneous enhancement, and for osteochondromas, enhancement was peripheral. It was concluded that septal enhancement may help identify low-grade chondrosarcomas. Aoki et al., however, in there MR studies of five chondrosarcomas and three enchondromas, stated that all tumors showed marginal and curvilinear septal enhancement (9). Whether septal enhancement does in fact suggest low-grade chondrosarcoma is thus somewhat controversial, though the chondrosarcoma in our case showed curvilinear septal enhancement. In a review of 13 chondrosarcomas and ten enchondromas, Janzen et al. (7) stated that on STIR imaging, high signal intensity of marrow adjacent to the tumor (abnormal peritumoral marrow signal) was present in all 13 chondrosarcomas but none of the enchondromas, and that abnormally high signal strands of soft tissue adjacent to the tumor or overlying cortical surface (abnormal soft tissue signal) were present in eight of 13 chondrosarcomas but none of the enchondromas. The strands were more common around high-grade than low-grade chondrosarcomas. They concluded that peritumoral marrow and soft tissue signal abnormality around a chondroid tumor on STIR imaging is strongly suggestive of chondrosarcoma.

In this review of 170 cases of Maffucci syndrome, 27

of the patients involved had intracranial lesions, while only six presented with chondrosarcoma. The origin of this was the sphenoid ridge (n=3), clivus (n=2), or cerebellopontine angle (n=1). Only two patients who underwent MRI showed solid enhancement (2).

Because Maffucci syndrome is a form of generalized mesodermal dysphasia, aneurysms can develop, though whether these represent a category of mesodermal dysplasia, or are a coincidental finding, is a mater of debate. Aneurysms have been reported in only three cases of Maffucci syndrome; two of these, one of which was an unruptured left paraophthalmic aneurysm and was found incidentally (3), were associated with the internal carotid artery, and the other with the superior mesenteric artery (10).

This report has described a case of Maffucci syndrome associated with intracranial chondrosarcoma and aneurysm of the internal carotid artery.

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연골육종과 동맥류를 동반한 Maffucci 증후군: 증례 보고[1]

[1]가톨릭대학교 의과대학 방사선과학교실
[2]가톨릭대학교 의과대학 임상병리과학교실

임형근 · 유원종 · 임연수 · 성미숙 · 정명희 · 이해규 · 정소령 · 김진아[2]

Maffucci 증후군은 연골종증과 혈관종들로 특징지워지는 선천적이고 비유전적인 드문 질환으로 두개내 연골육종과 같은 악성종양이 생길 가능성이 있다. 또한 이질환은 중배엽성 이형성증과 관련되어 동맥류가 생길 수도 있다. 이에 저자들은 연골육종과 동맥류를 동반한 Maffucci 증후군 1예를 보고하고자 한다.