MAFFUCCI'S SYNDROME : REPORT OF A CASE

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ABSTRACT :

Waisayarat J. Maffucci's Syndrome : Report of A Case. (Region 7 Medical Journal 1996 ; 4 : 531-536).

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Maffucci's syndrome is a congenital, non-hereditary mesodermal dysplasia manifested by enchondromatosis and multiple hemangiomas. The syndrome is frequently associated with mesodermal and non-mesodermal tumor. The sarcomatous transformation of enchondroma is 15 - 20 percent. The over-all incidence of malignant tumor is 23 per cent. A case of Maffucci's Syndrome with mutiple hemangiopericytomas is presented for the first time in this report. Non - mesodermal tumor component, tubular adenoma of the breast is detected in the same patient.

บทคัดย่อ :

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กลุ่มงานพยาธิกายวิภาค, รพ. นครปฐม.

Maffucci's Syndrome เป็นกลุ่มอาการของ congenital non - hereditary mesodermal dysplasia ที่ปรากฏอาการด้วย enchondromatosis และ multiple hemangiomas มักมีความสัมพันธ์กับ mesodermal และ non - mesodermal tumor การเปลี่ยนเป็น sarcoma ของ enchondroma พบประมาณ 15 - 20% และสัมพันธ์กับมะเร็งอื่น ๆ 23% คนไข้ที่นำเสนอ 1 ราย มาด้วยอาการ ของ multiple hemangiopericytomas และ tubular adenoma ของเต้านม ซึ่งเป็น non - mesodermal tumor

Introduction

Maffucci's Syndrome is the syndrome that presented with multiple hemangiomas and enchondromatosis. It is first described by Maffucci in 1881.[1] It is congenital, non - hereditary, no familial incidence, no sex predilection and not the result of chromosomal aberration.[2] The importance of this syndrome is that the incidence of sarcomatous transformation of enchondroma is 15 - 20 per cent.[3] And the over - all incidence of malignancy association is 23 per cent.[4] The disorder was thought to be quite rare, but in the last 20 years, more than 100 cases have been reported in the world literatures. This case is the third case report in Thailand and is the first patient that presented with multiple hemangiopericytomas. Non-mesodermal tumor component, tubular adenoma of the breast is detected in the same patient.

Case Report

A 16 - year - old woman initially appeared at Nakhonpathom hospital with clinical of bleeding per nipple of left breast. Two years before she developed left breast mass. The mass was approximately 2 cm in diameter. It was movable and not tender. Two days before this admission there was an ulcer with oozing blood on the skin of left breast. Bloody discharge per left nipple was also developed. Historical review disclosed that the patient was essentially normal until the age of 3 years, when a bowing of her left knee and multiple subcutaneous nodules of her both hands had appeared. By the year of 13 years she developed left neck mass without tenderness. No other family members were known to suffer from this disorder and breast mass. The physical examination revealed left breast mass, measuring 15 cm in diameter with well - circumscribed border and oozing ulcer of the skin breast. There was no lymphnode enlargement. The right breast was normal. The right and left knee were deformed from bony tumor that were diagnosed as enchondromas from x - ray findings. Multiple subcutaneous nodules of both hands are soft and brownish in color. Multiple hemangiomas were diagnosed according to the x - ray findings. The left thyroid nodule , 4 cm in diameter was detected. The patient is recognized as having Maffucci's Syndrome according to physical examination findings. The operation of left simple mastectomy, left thyroid lobectomy and vocal cord nudulectomy were performed. (The vocal cord nodule was found on the operation day.) The specimens were sent to the Anatomical Pathology Department. The left simple mastectomy mass was 14 x 13 x 10 cm. It showed a huge gray - white mass, measuring 13 x 12 x 9 cm. The serial section showed gray - white tissue with focal areas of dilated spaces containing blood. The nipple was normal. The microscopic findings were tubular adenoma of the breast with scattered areas of hemangiopericytomas. (Figure 1) The left thyroid lobectomy was 4 x 4 x 3 cm. The sections revealed a well - defined nodule, 3 cm in diameter. The cut surface showed tan spongy tissue with cystic spaces. The microscopic findings were hemangiopericytomas. (Figure 2, 3) The vocal cord nodule was 0.8 cm in diameter which showing tan spongy tissue with cystic spaces on the cut sections. Its microscopic findings were hemangioma. (Figure

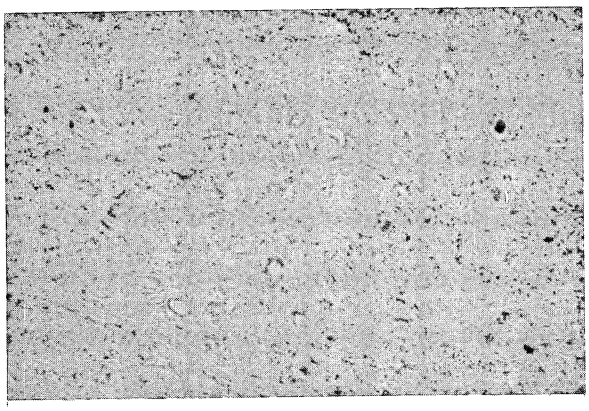

Figure 1 Breast : Tular adenoma (Low mangnification)

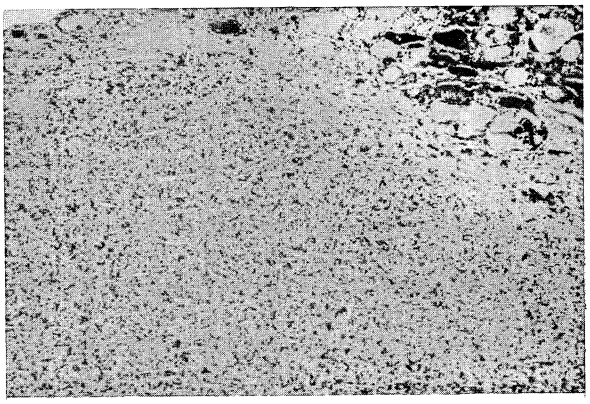

Figure 2 Thyroid nodule : Hemangiopericytoma (Low mangnification)

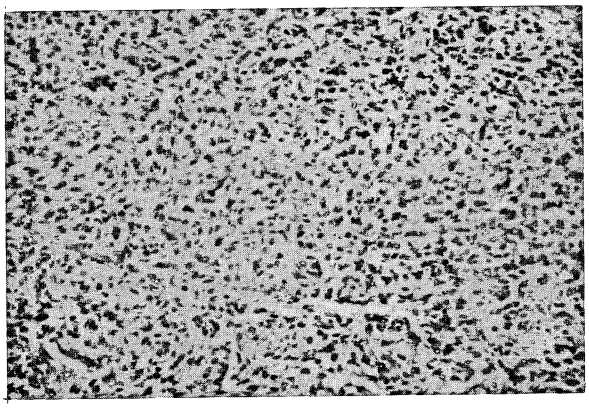

Figure 3 Thyroid nodle : Hemangiopericytoma (Medium magnification)

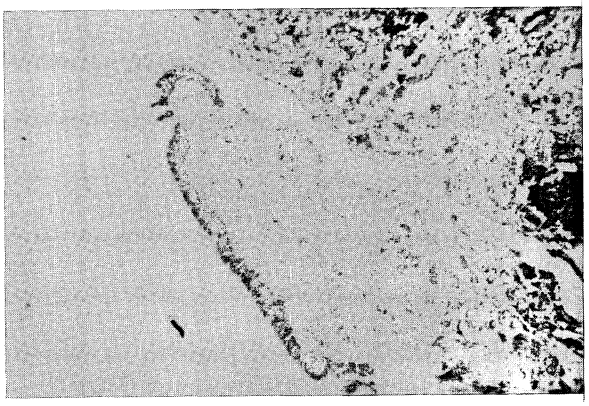

Figure 4 Vocal cord nodule : Hemangioma (Low magnification)

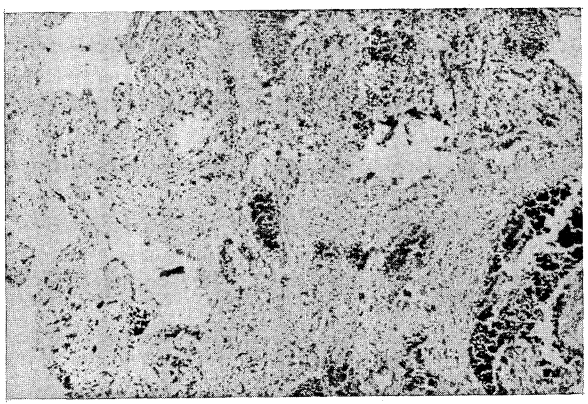

Figure 5 Vocal cord nodule : Hemangioma (Medium magnification)

4, 5) Immunoperoxidase study of the breast mass, thyroid and vocal cord nodules were performed and supportive the pathological diagnosis. So this patient was finally diagnosed as Maffucci's Syndrome presented with multiple enchondromas of both knees, multiple hemangiomas of both hands, hemangioma of vocal cord, hemangiopericytomas of the left breast and left thyroid and tubular adenoma of the left breast.

Discussion

Maffucci's Syndrome consists of dyschondroplasia and multiple soft tissue hemangiomas. The dyschondroplasia and hemangioma are mesodermal, and therefore, most authors believed that the syndrome is a congenital mesodermal dysplasia.[5] It is congenital but it is not hereditary without racial or sex predilection.[6]

The average age that symptoms are first noted is five years. Twenty - five per cent has symptoms at birth or in the first year of life, forty - five per cent has symptoms before the age of six years, and seventy - eight per cent has symptoms before puberty.[3] This patient the bone tumor of left knee and subcutaneous nodules of both hands were first noticed at the age of three years.

The predominant presenting features are usually the hemangiomas and the enchondromatosis. The enchondromas appear on radiographs as multiple irregular cystic lucencies in the growth region. Marked bony deformities may be seen and may be complicated by pathological fracture. In this patient, there were multiple calcified enchondromas in both hands, both distal end of the radious - ulna, both distal end of femurs and proximal end of both tibias.

The vascular lesions include hemangiomas, phlebectasias, vascular harmatomas, lymphangiomas and lymphangiectasias have been reported in Maffucci's Syndrome.[7] In this patient multiple hemangiopericytomas is presented for the first time in Maffucci's Syndrome. The hemangiopericytomas are found in the left breast mass and left thyroid gland. She also has hemangiomas in the both hands and the vocal cord. The huge tubular adenoma that is non - mesodermal tumor is detected in the left breast in the same patient.

The most serious complication of the syndrome is the high risk of malignancy association. The incidence of malignant transformation of enchondromas to chondrosarcoma is 15 - 20 per cent.[3] There are also reports of transformation of hemangiomas and lymphangiomas to sarcomas. The over - all malignancy associated with the disease is 23 per cent. Other reports associated-with nonskeletal malignancy include fibrosarcoma, angiosarcoma, malignant lymphangioma, glioma, astrocytoma, ovarian teratoma, pancreatic adenocarcinoma and liver carcinoma.[3,8] There was no associated malignancy in this patient. Although the disease has deformity, the patient can live a reasonably normal life. Periodic follow - up was recommended for early detection of associated malignancy.

Summary

A case of 16 - year - old Thai female with Maffucci's Syndrome presented with multiple

hemangiomas, hemangiopericytomas and enchondromatosis is reported. It is the first case of Maffuccis Syndrome that presented with multiple hemangiopericytomas. The symptoms are first noted on her both hands since three years old. The non - mesodermal tumor component, tubular adenoma of the breast is also detected in this patient. Periodic follow - up of the patient was advised because of the significant incidence of associated malignancy.

Acknowledgement

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Reference

1. Maffucci A. Di un caso enchondroma ed angioma multiple : Cont ribuzione alla genesi embrionale dei tumori. J Movimento Napoli 1881; 2 : 399-412, 565-575.
2. Anderson IF. Maffucci's Syndrome : report of a case with review of literature. S Afr Med J 1965 ; 39 : 1066-70.
3. Lewis RJ, et al. Maffucci's Syndrome : Functional and neoplastic significance. J Bone Joint surg 1973 ; 55-A : 1465-79.
4. Loewinger RJ, et al : Maffucci's Syndrome. A mesenchymal dysplasia and multiple tumor syndrome. Br J Dermatol 1977 ; 96 : 317-22.
5. Marberg K , Dalith F , Bank H. Dyschondroplasia with multiple hemangiomata (Maffucci's Syndrome). Ann Int Med 1958 ; 49 : 1216-28.
6. Ben - Itzhak I, Denolf FA , Versfeld GA , Noll Bj. The Maffucci's Syndrome. J Pediatr Orthop 1988 ; 8 : 345-8.
7. Caro WA, Bronstein BR. Tumors of the skin. In : Moschella SL, Hurley HJ, eds. Dermatology. 2nd ed. Philadelphia : Saunders, 1985 : 1533-8.
8. Schwartz HS, Zimmerman NB, Simon MA, et al. The malignant potential of enchondromatosis. J Bone Joint Surg 1987 ; 69-A : 269-74.