CASE REPORT

Maffucci Syndrome

Widhodho T Karyomanggolo, Dyani Kusumowardhani

(Department of Child Health, Cipto Mangunkusumo Hospital-School of Medicine University of Indonesia, Jakarta)

ABSTRACT A case of Maffucci syndrome was found in a 6 year old Indonesian girl. Her first sign of bone malformation was noticed on her right arm at the age of 7 months, but since there were no other complaint, such as pain or dysfunction, no further investigation was done. The complaints that brought her to Cipto Mangunkusumo Hospital, Jakarta, were an incomplete cleft palate and right axillary masses which were then known as cavernous hemangiomas. On physical examination another hemangioma was found on the major labia fold. From bone survey, generalized enchondromatosis was recognized and bone biopsy was then scheduled, but when the incision was done another hemangioma was found, so biopsy was canceled due to risk of bleedings. Magnetic resonance imaging of the right humerus reaffirmed that the mass inside the bone was enchondroma and the masses in the axilla were subcutaneous hemangioma. Radiologic bone examination done 2 years later revealed that the enchondromatosis had increased in size and spreading. IQ test showed a borderline stage of intelligence, but there were no evidence of relationship with the disease. **[Paediatr Indones 1999; 39:237-241]**

Introduction

Maffucci syndrome is a rare and complex disorder of the mesodermal tissue development. The disease consists of dyschondromatosis or enchondromatosis and multiple hemangiomatosis, and was named after Angelo Maffucci who first described the condition in 1881.[1-3]

Dyschondromatosis or dyschondroplasia is a congenital disease with its essences being delayed and abnormality of the embryonal cartilaginous tissue ossification.[4] It is

Author's address: Widhodho T Karyomanggolo, MD, Department of Chiold Health, Medical School, University of Indonesia, Jakarta, Jalan Salemba 6, Jakarta 10430, Indonesia. Tel 62-21-3907742, Fax 3907743.

the result of cartilage failure to undergo the normal process of enchondral bone formation. Rounded masses or columns of uncalcified cartilage are produced within the metaphyses and diaphyses of certain bones, which are invariably shortened. Calcification with healing may occur in adulthood but deformity persists.[5]

In enchondromatosis, multiple enchondroma are found in the long bones shaft, distributed unequally in the skeletal bones and characteristically caused shortening of the affected bones. Cartilaginous mass represents cartilaginous cells persisted in the cortex, which in normal condition after its growth in the epiphyseal disc will transform into osteoblast and form solid bones.[6]

Report of the Case

A 6 year-old girl came to the Department of Child Health, Cipto Mangunkusumo Hospital, Jakarta with the complaints of cleft palate and masses in her right axilla. The masses encountered about a year before, without any pain and the sizes had not been changed. At the age of 7 months, her right forearm was enlarged and as she grew her arms, legs, and fingers grew bigger and dysformed. No pain had been felt and since there were no dysfunction of the extremities, the doctor she was first brought to did not do any further investigation.

On her first visit to our department she looked alert, active, with normal vital signs, weighted 18 kg (P10 NCHS) and her height was 95 cm (<P3 NCHS). On ENT examination an incomplete cleft palate was found on her soft palate. Her extremities were asymmetrically enlarged and dysformed, with the right side being bigger. (Fig. 1) Two cavernous hemangiomas were found in a row in her right axilla with the diameters of 0,5 and 1 cm. (Fig. 2) Another hemangioma was found on the labia majora fold with the diameter of 0,3 cm. There were no enlargement of the lymph nodes and no abnormality of the other organs.

Bone survey revealed that the humeral,. radial, ulnar, femoral, tibial, and fibular bones were shorter than normal, with irregular surfaces, widened ends, and sclerotic bands on the metaphyseal regions. Similar features were found on the phalanges, metacarpals, pelvic, and anterior rib bones. (Fig. 3) The peripheral blood, blood electrolytes, urine, and feces were normal. Mantoux test with PPD RT 23-2TU gave negative reaction.

The patient was consulted to the Orthopedic Surgery Division and bone biopsy was then scheduled. When the incision for biopsy was done on her right radial region, another hemangioma with the diameter of 2 cm was found on the soft tissue around the bone. Biopsy was then canceled due to risk of bleedings. The diagnosis of Maffucci syndrome was established. Magnetic resonance imaging (MRI) was performed on the right humerus and showed benign metaphyseal lesion with calcification inside it, which reaffirmed that the mass inside the bone was enchondroma, and subcutaneous hemangiomas in the right axilla. Higher intensity images also indicated that hemangioma was also present inside the bone.

A periodic follow up was done to the patient. During that time the dysformation of the bones had become more visual and the number of hemangiomas had increased. There were still no complaint of pain and dysfunction. Two years later another radiologic bone examination was done and revealed that the enchondromatosis had increased on size and can be found in more sites. IQ test was also done and showed a borderline stage of intelligence.

Discussion

This is the first case of Maffucci syndrome found in our hospital. In fact such cases are very rarely reported. Volkoy4 reported only 4% incidence of chondroma among all primary bone tumors and dysplasias in children. The accidentally finding of the disease in this patient is very interesting because the patient came with the complaint of cleft palate. So far no report has been published for cases of Maffucci syndrome associated with other congenital disorder. The MRI procedure was performed in this patient as a replacement of bone biopsy to recognize the masses inside the bones. The result of this procedure will become the base data of the lesions development.

Maffucci syndrome causes functional disorder in varied degrees, but the cartilaginous tissue malignant transformation is the most severe complication. Sun et al.[7] predicted the incidence of chondrosarcoma in Maffucci syndrome was 17,8%, while Leiber and Olbrich[2] mentioned that the tendency to transform into malignancy was 20%, and other than chondrosarcoma, it could also be angiosarcoma, lymphangioma, glioma, or even teratoma of the ovary. Schwartz et al.[8] said that malignant transformation is almost certain in Maffucci syndrome patients.

In their observation, Sun et al.[7] found that chondrosarcoma usually occurred after the age of 40 years. Signs that brought them to think of the possibilities of malignant transformation are pains, especially if not preceded by trauma, swelling, and rapidly growing soft tissue. As soon as the bone maturity is achieved, the cartilaginous lesions will stop growing and new lesions will not occur. If these do not happen, the presence of malignancy should be suspected.[9]

Our patient is only 8 year old now and so far she had never experienced any pain on her bones, but a long term follow up is important to be done. Besides giving us more important information about this very rare case, it will help the patient in having early medical aid whenever problems arise due to her disease. It is also important to remember that Maffucci syndrome patients are very likely to die from malignancy other than of the bones. That's why a periodic evaluation of the central nervous system and the abdomen can be beneficial in finding occult malignant lesions, especially if secondary chondrosarcoma have occurred.

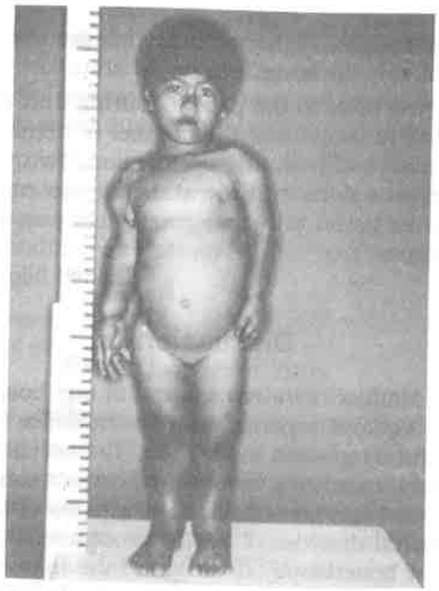

Figure 1. General appearance of the patient clearly shows asymmetrical extremities.

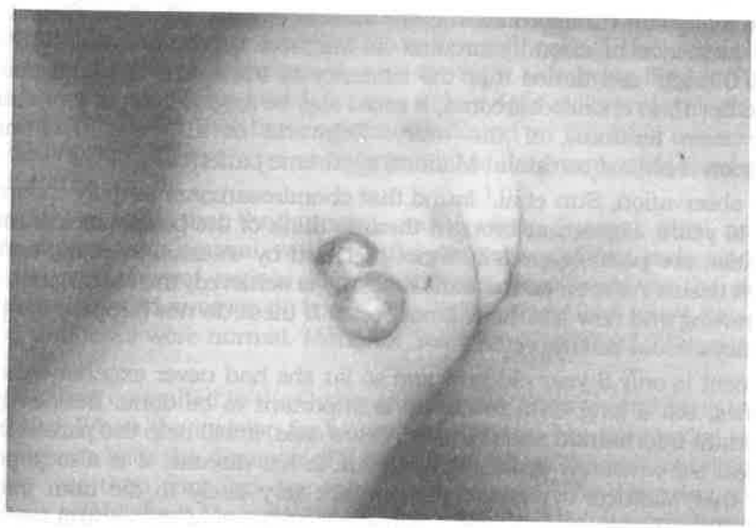

Figure 2. Cavernous hemangioma found in the patient.

The finding of a borderline stage of intelligence in this patient is interesting. So far there has been no report of intelligence degree in Maffucci syndrome patients or whether the disease may effect the patients intelligence. An examination on the central nervous systems anatomy and physiology may give information about the relationship between the disease and the patients intelligence.

Acknowledgment

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