Cancer Prone Disease Section

Mini Review

Maffucci syndrome

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Identity

Alias
Dyschondrodysplasia with haemangiomas
Enchondromatosis with multiple cavernous haemangiomas
Multiple angiomas and endochondromas
Kast syndrome
Haemangiomatosis chondrodystrophica

Note
It is a rare, developmental disorder characterized by the presence of multiple enchondromas (enchondromatosis), multiple haemangiomas or, less commonly, lymphangiomas.

Inheritance
It is a non hereditary disease.

Clinics

Note
Maffucci syndrome was first described in 1881 by Angelo Maffucci. In the 2002 World Health Organization classification Maffucci syndrome is considered as a subclass of enchondromatosis. It is characterized by the occurrence of multiple enchondromas of the bones combined with haemangiomas of the soft tissue. Both of which may undergo malignant change. Males and females are equally affected. Haemangiomas often protrude as soft nodules usually presenting on the distal extremities, but they can appear anywhere in the body such as in the leptomeninges, in the eyes, in the pharynx, in the tongue, in the intestines and in the trachea. Multiple enchondromas are benign cartilaginous tumours growing in the medulla of bones, predominantly in long bones.

The enchondromas bear the risk of secondary fractures in metaphyses and diaphyses.

Phenotype and clinics

Maffucci syndrome is characterized by enchondromas, resulting in bone deformities, and soft tissue haemangiomas. The disease appears to develop around the age of 4 to 5 years in 25% of cases. In 45% of cases symptoms start before the age of 6 and in 78% of cases symptoms developed before puberty. The bone and vascular lesions may be progressive. No association with mental or psychiatric abnormalities but in few exceptional cases, it is associated with ovarian tumor and nervous system abnormalities. A review of ninety-eight cases showed that hand, foot, femur, tibia and fibula were the most frequently affected sites. The diagnosis is made based on a combination of clinical, radiological and histological grounds. Enchondromas can be radiolucent or mineralized and can be intramedullary or periosteal in location. Phleboliths (associated with soft tissue calcification) in haemangiomas can be visualized in radiographs. Histologically, haemangiomas can be divided into two groups; capillary haemangiomas (narrow, thin-walled capillaries, thin epithelium separated by connective tissue) and cavernous haemangiomas (sharply defined and having deeper structures more often than capillary subtype). Another specific subtype called spindle cell haemangioma (features like cavernous haemangioma combined with Kaposi sarcoma-like features) can be seen. Distinction between enchondroma and low-grade chondrosarcoma in Maffucci syndrome is complicated as compared to solitary tumors and usually it is based on clinical and radiological grounds (cortical destruction, soft tissue extension) or the presence of mitosis.
**Neoplastic risk**

There is a high risk of development of malignant tumors in Maffucci patients, with an overall incidence of 23-100%. The majority is of mesenchymal origin and includes secondary central chondrosarcoma and angiosarcoma. Moreover, benign tumors such as pituitary chromophobe adenoma, uterine polyp, uterine fibroid, adrenal cortical adenoma and ovarian thecoma are described, as well as fibrosarcoma, glioma, mesenchymal ovarian tumour and carcinoma of the pancreas.

**Treatment**

Individuals having Maffucci syndrome need to have regular physical examinations in order to evaluate changes in the skin and bone lesions that may suggest malignancy. Haemangiomas could be treated with sclerotherapy, surgery or laser treatment to reduce the size of the lesions. Biopsy can be done in case of symptomatic enchondromas. Surgery can be done in case of borderline or low grade chondrosarcoma.

**Prognosis**

It depends upon severity of the disease but there is an increased risk for malignancy.

**Cytogenetics**

The molecular defect underlying Maffucci syndrome is still unknown. There was one case reported with inversion of p11 and q21 of chromosome 1 in a patient with Maffucci syndrome.

**References**

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