EPIDERMAL NEVUS SYNDROME WITH FIBROUS DYSPLASIA

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A 10-year-old girl developed epidermal nevus, shortness of limb, scoliosis, hemiatrophy with history of multiple pathological fractures. Bone biopsy revealed fibrous dysplasia. This is an extremely rare combination of epidermal nevus with fibrous dysplasia.

Key words : Epidermal nevus, Fibrous dysplasia.

The epidermal nevus syndrome is a congenitally acquired syndrome consisting of deformities of the skin, the skeletal system, the central nervous system and cardio-vascular system. Associated anomalies include epidermal nevi, areas of hypopigmentation, other nevi and cafe-au-lait spots, kyphoscoliosis, vertebral defects, hemihypertrophy, short limbs, phocomelia, angiomas of skin, patent ductus arteriosus, coarctation and hypoplasia of the aorta, ocular abnormalities with brain tumor, hydrocephaly, mental retardation and convulsive disorders.[1-3] Although it appears that most cases of epidermal nevi appear sporadically, accumulated data suggest that in some cases atleast, autosomal dominant transmission may be present.

The nature of the verrucous lesions ranges from large unilateral hypertrophic deformities of the epidermis to whorled brush-stroke-like scaly lesions involving various areas of the skin surface. Association of fibrous dysplasia with epidermal nevus is not known. The triad known as Albright syndrome includes fibrous dysplasia, abnormal skin pigmentation and precocious puberty.[4] A case of epidermal nevus syndrome with fibrous dysplasia is reported.

Case Report

A young girl of ten years, born of non-consanguinous union was seen for brownish black, verrucous, linear lesions on the right side of the body since birth and limping on left side since six years with a history of repeated fractures on minor trauma since 2 years. The skin lesions were distributed between C2 and T10 segments on right side. On the right upper extremity, these were located along the long axis of the limb and had an interrupted pattern. However, on the trunk these had a transverse orientation and were continuous in pattern. Hypertrophy of the skin lesions was not uniform throughout. In addition, she had hemiatrophy of the right side of the tongue, right hand, right leg and right side of the chest. The physical and mental mile-stones were normal.

Muscle testing revealed wasting of the quadriceps and calf muscles of the right lower extremity, and deltoid and forearm muscles on the right upper extremity. There was genu-valgum on the right side and genu varum on the left side. Left lower extremity was shortened by 1 cm.

Routine investigations were normal. X-ray of the extremities revealed multiple fractures of various bones with gross osteoporosis. Serum calcium, phosphorus, alkaline phosphatase, uric acid and creatinine were within normal limits. Biopsy from the left femur showed a fibrous dysplasia.

Comments

Our patient had characteristic features of epidermal nevus syndrome, namely epidermal nevus, shortness of limb and scoliosis. Solomon[1] observed hemihypertrophy in his series of cases. However in our patient, hemiatrophy affected nearly all the muscles on right side of the body except muscles of the face and neck. In addition

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association of multiple repeated fractures after minor trauma was interesting. Cause of these fractures turned out to be fibrous dysplasia. Association of epidermal nevus with fibrous dysplasia has not been reported so far. However, fibrous dysplasia with abnormal macular skin pigmentation and precocious puberty has been reported by Albright et al.[4] In our case these abnormalities i.e. macular skin pigmentation and precocious puberty were absent. Thus this seems to be a rare combination of extensive epidermal nevus with fibrous dysplasia.

References

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