Osteopathia Striata and Focal Dermal Hypoplasia

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Abstract. A 43-year old female, presenting with pain in the left knee, attributed to a stress fracture of the tibia, was found to have osteopathia striata and numerous other congenital anomalies, not only of the skeleton, but also of other tissue systems. Of the latter the most important was the presence of focal dermal hypoplasia, a disorder of the skin. Review of the radiological and dermatological literature has shown this to be the fourteenth case in which these excessively rare entities have been associated. Such an association is considered to be more than a coincidence. A radiological diagnosis of osteopathia striata, therefore, should be followed by a careful dermatological examination for focal dermal hypoplasia. Conversely, diagnosis of this rare dermatological disease should indicate the necessity for radiological survey of the skeleton.

Key words: Osteopathia striata - Voorhoeve’s disease - Congenital bone disease - Ecto-mesodermal dysplasia - Focal dermal hypoplasia - Goltz syndrome.

In 1972 Larregue et al. described, in nine of eleven cases with focal dermal hypoplasia syndrome (FDH), longitudinal striations in the metaphyses of the long bones [21]. This association has been confirmed in four other cases [9, 14, 17]. In 1970 Goltz et al. [13] and Ginsburg et al. [11] described in detail the coexistent bony dystrophy of FDH, but they did not mention osteopathia striata in their roentgenological review.

We have observed an adult woman with FDH, osteopathia striata, and multiple abnormalities of bone and viscera. Our patient presented with a stress fracture in the striated bone area and was observed also to have a generalized osteopenia.

Case Report

A 43-year-old woman was admitted to the psychiatric ward for toxicomania. She was referred subsequently to the rheumatological clinic because of pain at the left knee without a history of trauma. Her parents were normal, but her mother was of short stature (150 cm). She had a healthy brother. At birth the patient had diffusely scattered patchy ‘café au lait’ spots, which were thought to be due to neurofibromatosis of Von Recklinghausen. Extraction of several teeth due to malformation was necessary at the age of 16 years. A supernumerary toe and digit had been amputated during childhood. The patient’s general growth and development was otherwise normal, but she was always of short stature compared to children of the same age.

At the time of investigation her height was 148 cm. There were many unusual facial features: asymmetry with hypoplasia of the left side, large protruding ears, broad and high columella, sessile papillomata at the palato-pharyngeal arches, and a coloboma iridis. Multiple poikilodermal lesions were present on the trunk consisting of atrophic skin, pigmentation, and telangiectasia. Large circumscribed cicatrisable lesions and orange coloured fatty tumours were situated on the buttocks. Numerous papillomata were present at the perivulvar and anal region, which disappeared with 5-fluorouracil ointment. The breasts were asymmetrical, the left being larger than the right. The extremities showed syndactyly of the left second and third fingers, atrophy of the hypothenar muscles, and sequelae of amputation of the left third toe and right fifth metatarsal. Dystrophic nails and micronychia were also present.

Laboratory investigations showed no abnormality other than a moderately elevated hydroxyproline excretion in the urine, 60 mg/24 hours. The urinary excretion of other amino-acids was slightly reduced. The serum calcium, phosphorus, and alkaline phosphatase activity were normal on repeated measurements, as were the 24 hour urinary excretion values for calcium and phosphorus. The bone mineral content, measured by $\gamma$ photon absorptiometry, at 8 cm of the distal end of the radius was just below one standard deviation of the sex-age matched mean value, indicating osteopenia.

Other abnormalities found on investigation included bilateral conduction deafness and a right bifid ureter and renal pelvis. Histological examination of the skin showed a normal epidermis, dermal atrophy, and adipocyte proliferation under the epidermis, typical of FDH.
Fig. 1. X-ray of the knee showing osteopathia striata and callus of a stress fracture on the medial side of the left tibia

Fig. 2. X-ray of the left knee showing the stress fracture at the time of first visit

Fig. 3. X-ray of the hands showing osteopathia striata of the distal end of the radius, fusion of the right trapezium and scaphoid, brachymetacarpia of the fourth metacarpal bone, and dysplastic fifth middle phalanges

Fig. 4. Antero-posterior X-ray of the skull showing aplasia of the sinus frontalis, absent diploe, low left petrous ridge, and a hypoplastic left maxillary sinus