Primary osteoma cutis – multiple café-au-lait spots and woolly hair anomaly

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Abstract. An 11-year-old girl with typical features of primary osteoma cutis is reported. She demonstrated multiple areas of subcutaneous, asymptomatic, slow-growing nodules, mostly localized at the extremities. Radiographic evidence of soft tissue calcification and histologic confirmation of ectopic bone formation was obtained. She also showed café-au-lait spots, woolly hair and intrauterine growth deficiency.

Primary osteoma cutis is an unusual condition characterized by de novo bone formation localized to the skin or subcutaneous tissues with no tendency to excessive and invasive growth or involvement of other structures. The lesions can be single or multiple, in various locations and of varying size [1, 2]. The nodules, which are usually covered by normal skin, are hard in consistency, generally asymptomatic and mostly localized at the extremities [1–3]. The events leading to true bone formation in the skin are poorly understood [3]. Inherited forms of this condition are very rare [4].

We report an 11-year-old girl with primary multiple osteoma cutis mostly localized at the extremities, who also had café-au-lait spots scattered over her body, other skin and hair manifestations and intrauterine growth retardation.

Case report

The patient was the first-born of unrelated parents and had one healthy brother. The family history revealed that her father was affected by migraine and psoriasis; her mother presented with multiforme dermatitis. The patient was delivered by cesarean section at 39 weeks. At the 4th gestational month, ultrasonography revealed intrauterine growth deficiency. Birthweight was 1650 g, height 47 cm and head circumference 32 cm. At birth she was hospitalized because of her low weight. Over the next 2 months she gained weight and height slowly and was treated with Vitamin D₃ at a dose of 1 µg/day.

She presented with frequent vomiting and a history of weight and growth retardation up to the age of 5 years, when she underwent tonsillectomy. Thereafter she grew and developed normally. She was 8 years of age when her mother first noticed a tender subcutaneous swelling on her left heel. At 9 years of age a new nodule, hard in consistency, was noted in the right thigh. In the meantime, a physician noted a few café-au-lait spots which were believed to have been present since birth. There was no history of trauma.

When the girl was first referred to the pediatric clinic of our institution, at 10 years of age, physical examination revealed her to be in good general health, with weight, height and head circumference at the 50th centile. Her skin was dry and wrinkled, especially at the extremities, with psoriatic lesions at the knees and elbows.

Firm, round, well-defined, nontender subcutaneous nodules, 1–2 cm across and covered by normal skin, were detectable in the left temporal region, dorsal surface of the left foot, right thigh, left ankle and left heel. There were café-au-lait spots on her right arm (1 × 0.5 cm), in the right lower lumbar region (3 × 1 cm), on the anterior surface of the left thigh (2 × 2 cm), and on the right lateral chest (3 × 0.5 cm), and some seven to eight were scattered over her back. There was a wide, flat, hyperpigmented area (11 × 12 cm) with regular margins situated between the upper posterior surface of the right thigh and the right flank, and a cutaneous fibroma (0.5 × 0.5 cm) in the intergluteal region. Her hair, which had grown poorly in the first years of life, was woolly, thin and curly in some areas (Fig. 1), especially in the occipital region. Complete physical examination revealed no other abnormalities. She was of normal intelligence. Puberal stage was P4, B4 with menarche.

Routine laboratory examinations, including calcium, phosphorus, alkaline phosphatase, blood urea nitrogen, immunoglobulins, creatine kinase, lactate dehydrogenase gamma-glutamyl transferase, thyroglobulins, anti-nuclear antibody, anti-smooth muscle antibody, antimitochondrial antibodies, cholesterol and triglycerides, lupus erythematosus phenomenon, osteocalcin, type 1 procollagen, parathormone, 1,25(OH)₂D₃, 25(OH)D₃ as well as urinalysis and complete blood count, were within the normal limits. ECG, EEG, cardiac, abdominal and retroperitoneal ultrasonography, fundal examination, chest radiographs and electromyography were normal. Cerebral MRI revealed only slight nonspecific ventricular asymmetry. Skeletal radiography showed diffuse nonhomogeneous soft tissue calcification (Fig. 2), with shallow and irregular margins in the left temporal region, right acromion and coracoid process (Fig. 3), right elbow, right and left forearm, right and left wrist, right thigh (Fig. 4), plantar and dorsal regions of the left foot (Fig. 5), left heel and left ankle. In addition there was a thin, but not short, fourth metacarpal, and a periosteal reaction.
Fig. 1. Photograph showing patches of curly, unruly hair alternating with the patient's normal straight hair.

Fig. 2. Drawing of the main soft tissue calcification nodules as revealed on physical and radiographic examination.

Fig. 3. Radiograph of the shoulder joint showing soft tissue calcification in the acromion and coracoid process.

Fig. 4. Radiograph of the thigh showing soft tissue calcification in the central region.

Fig. 5. Radiograph of the foot showing soft tissue calcifications in the plantar and dorsal regions, and at the heel.

Fig. 6a, b, c. Photomicrographs of the biopsy sample showing well-formed spicules of bone in the dermis and subcutaneous tissues. The bone appears lamellated with haversian canals containing blood vessels and connective tissue. (H & E × 100) d Photomicrograph showing osteoblasts and osteocytes within the lesion and fibroblast-like appearance of the cells around the bony lesion (H & E × 250).

Discussion

This patient showed the typical features of primary osteoma cutis. She presented with multiple subcutaneous slow-growing nodules, mostly localized at the extremities but detected also in other areas, and with radiographic evidence of diffuse soft tissue calcification. Histological examination revealed these areas to be ectopic bone formation. Cutaneous and soft tissue ossification is traditionally divided into primary and secondary...