PP-177

A case report: osteochondroma-associated with the Carney-complex?

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Introduction: The Carney Complex (CNC) is an autosomal dominant multiple neoplasia syndrome. Almost 60% of patients have a germline inactivating mutation in the Prkar1a gene. The various components of this complex include pigmented skin spots, myxomas (cardiac, cutaneous, ...), endocrine overactivity (the Cushing syndrome, acromegaly and sexual precocity), schwannomas and osteochondromyxomas. Osteochondroma is not known to be associated with the complex, so this is supposedly the first described case, where these two entities occur in one person.

Case Description: A 17-year-old Caucasian man (weighing 115 kg; 191 cm tall) was presented with osteochondroma of the humerus and several cutaneous myxomas on his chest. X-rays showed an exophytic chondro-osseus lesion at the proximal diaphysis. MRI, scintigraphy and immunohistological analysis after marginal ablation confirmed the diagnosis of osteochondroma. Due to the multiple cutaneous myxomas precise evaluation with relation to Carney-Complex followed. His Medical history revealed multiple angiomyxomas and calcifications of the testicular parenchyma. Furthermore the patient showed multiple narrow based papules on the chest, the right lower eyelid, the scrotum and the ventral surface of the tongue. The serum levels of IGF-I and of IGF-BP 3 were increased. In family history, his mother showed typical symptoms of CNC (cardiac myxomas). Although neither the mother, nor the presented man were positively tested for the two known associated loci of CNC.

Discussion: According to international classification criteria, the presented man can be diagnosed of CNC due to the appearance of multiple myxomas, a first-degree affected relative, calcification in testicular ultrasound and elevated serum levels of IGF. Prkar1a is a tumor suppressor gene with specific importance to cAMP responsive tissue and enhanced bone tumorgenesis. Myxomatous, cartilaginous and bony differentiation, like osteochondromyxomas are typical. In contrast, the appearance of osteochondroma is usually linked to tumor suppressor genes and differs in patient age and tissue composition. This presented case however displays the first manifestation of CNC and osteochondroma in one person. An association between these two entities cannot be excluded.