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Coffin-Lowry syndrome in a patient with osteosarcoma: cause or coincidence?

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Introduction: Osteosarcomas constitute a rare tumour entity, accounting for only 1% of all cancers diagnosed. The highest incidence can be found in children and around the 7th decade. Many hereditary syndromes are associated with development of osteosarcomas, as the Rothmund-Thomson syndrome, Bloom syndrome and Li-Fraumeni syndrome. Some hereditary syndromes follow the x-chromosomal pattern, from which the fragile X syndrome is most common, causing mental retardation.

Case Presentation: We herein present the case of a 22-year-old male patient with microcephaly, mental retardation and skeletal deformities. He was admitted to our department with severe knee pain caused by an osteosarcoma G3 of his right distal femur with involvement of the knee joint. Neoadjuvant chemotherapy was followed by overknee-amputation and adjuvant chemotherapy. One year later, there was no evidence of local or systemic recurrence. The patient's 8-year-old half brother presented with the same neurological impairments and growth delay. Moreover, the family history revealed multiple cancerous diseases as breast cancer, melanoma and laryngeal cancer. Therefore, the X-linked hereditary Coffin-Lowry syndrome (CLS) is supposed in both patients and a cancer syndrome may be present.

Discussion: The loss-of-function in the RSK2-gene is the cause for CLS, leading to mental retardation and growth disturbances. Affected females are rather asymptomatic, whereas males present with typical neurological deficits and bone deformities, as pseudoepiphyses and massively ossified fissures of the skull. As RKS2 plays a role in correct osteoblast function and is important for development of c-fos positive osteosarcoma, a relationship between our patient's disease and his mental retardation was supposed. However, this could not be approved, as deficient RSK2 seems to decrease the risk of developing an osteosarcoma.

Conclusion: To our knowledge, this is the very first case in literature of a patient with Coffin-Lowry syndrome developing an osteosarcoma. At the moment, genetic testing for p53 mutations is carried out in order to find out whether an additional hereditary cancer syndrome is present.