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Osteochondroma slips into the silent period of puberty ------ 20 years follow-up without any intervention

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Background Majority of the bone tumors are progressive and damage motor function in children. Osteochondroma is a kind of skeletal dysplasia, which can form bone ridges of different sizes on the bone. It is an autosomal dominant hereditary disease. Most patients have a family history of inheritance.

Case presentation We report a family with osteochondroma, including the proband and her mother with similar manifestations of multiple bony masses throughout the body and plagued by it nearly 20 years from the childhood of the mother to the time of definite diagnosis of proband. Fortunately, the osteochondroma of mother entered a quiescent period during puberty and did not damage motor function and quality of survival. With meaningless prenatal intervention, the pathogenic gene was passed on to the proband which cause similar disease again. Further genetic examination revealed mutations in the EXT1 gene (c.913C>T) and eventually confirmed the diagnosis of hereditary multiple exostoses(HME).

Conclusion The unique clinical follow-up covering two generations provides the real-world study of a non-intervention osteochondroma which expands our understanding of the development of the HME in adolescence and adulthood, and provides basis for prenatal diagnosis and intervention.

Biography

Dr. Li Jinrong's main research fields are growth and development, management of high-risk children, and rare diseases related to growth disorders. She has excellent clinical experience in rare disease case management, and shared the diagnosis , treatment methods and follow-up of these rare patients with multi-level hospitals and medical universities.

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