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Risk of Chondrosarcoma Transformation from Multiple Hereditary Exostosis Under the Age of 18, and Suggestion for Screening Protocol. Literature Review.

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INTRODUCTION

Multiple hereditary exostosis (MHE) is a rare autosomal dominant condition with a prevalence of 1in 50,000. MHE usually occurs in long bones such as the femur, tibia, and humerus.

Malignant transformation into chondrosarcoma is rare and occurs in about 3-5%, however, usually at the 4th and the 7th decade of life.

AIMS / OBJECTIVES

This literature review aims to assess the actual need for screening for chondrosarcoma transformation in MHE patients underage of 18 by identifying the reported cases of transformation at pediatric age.

MATERIALS / METHODS

The research question and inclusion criteria were established in advance.
Inclusion Criteria:
1.All levels of evidence
2.MHE with documented malignant transformation
3.Patients aged ≤18 years
4.Both male and female patients
5.Studies involving human subjects

Online databases, PubMed, and Google Scholar, were used to identify literature on malignant transformation in children with MHE. No restrictions were applied regarding publication language or date.
An Excel spreadsheet was used to record the relevant data obtained from the included studies.

RESULTS

A total of 5 patients were identified. Four patients were male (80%), and the mean age was 11.4 years. The mean duration of symptoms experienced before consultation was 8 months.
The most common presenting symptoms were pain and swelling; and the common sites were the femur and fibula. In terms of chondrosarcoma grading, 4 patients had low-grade chondrosarcoma (80%).
All patients underwent tumor resection. Three patients had follow-up data available, with a mean follow-up duration of 4.5 years, during which no recurrences were reported.

CONCLUSION

Given the extremely low prevalence of malignant transformation in MHE at pediatric age and the limited data primarily derived from case reports, our study does not support the necessity of routine screening for malignant transformation in patients with MHE. We recommended spacing the surveillance protocol by skeletal survey at interval of 2-3years for asymptomatic children. However, further studies extracted from larger databases are warranted to support a strong recommendation.

REFERENCE

Bukara E, Buteera AM, Karakire R, Manirakiza F, Muhumuza S, Rudakemwa E, et al. Osteoblastic Osteosarcoma Arising beneath an Osteochondroma in an 11-Year-Old Male with Multiple Hereditary Exostoses. Case Reports in Orthopedics. 2018 Jul 12;2018:1–8.