Enchondromatosis

Enchondromatosis is a rare skeletal disorder characterized by the development of multiple enchondromas, which can also manifest non-cartilage tumors including gliomas.

Ollier disease and Maffucci syndrome are non-hereditary skeletal disorders characterized by enchondromatosis (Ollier disease) combined with spindle cell hemangiomas (Maffucci syndrome).

The characteristics of gliomas in patients with enchondromatosis suggest that these tumors, as cartilaginous tumors, result from somatic IDH mosaicism and that the timing of IDH mutation acquisition might affect the location and molecular characteristics of gliomas. Early acquisition of IDH mutations could shift gliomagenesis towards the brainstem thereby mimicking the regional preference of histone mutated gliomas [1].

OD and Maffucci syndrome differ with respect to the distribution of intracranial malignancies by histology, and geographical and age distribution of cases, with OD patients younger by approximately a decade, and Maffucci syndrome patients more likely to live in Asia or South America [2].

Forty-six patients with 47 intra-cranial malignancies were identified, with nine of the patients being 18 years old or less and categorized as youths. The incidence of intracranial chondrosarcomas peaked in the fourth decade of life, in parallel with the peak number of MS cases; conversely, both non-sarcomas and OD peaked in the third decade of life. Six of nine youths (67%) versus 17 of 36 adults with gender data (47%) were female (P = 0.30). There was no difference in geographic distribution by continent (P = 0.82). Four youths (44%) versus 16 adults (43%) had a chondrosarcoma (P = 0.95), and there was no statistically significant difference by tumour site (P = 0.42). However, seven (77%) of the youths had Ollier's disease as their underlying enchondromatosis syndrome, versus just 17 (46%) of the adults, a difference that approached statistical significance (P = 0.086). The association between enchondromatosis and intracranial malignancy seems to be roughly the same in youths versus adults, though Ollier's disease cases appear to predominate among youths [3].

Case reports

2017

Achiha et al. describe a genetic analysis of a low-grade glioma that developed in an enchondromatosis case. A 32-year-old man with a long history of enchondromatosis developed a left frontal tumor. The histopathological findings of his surgical specimen revealed characteristics of a low-grade glioma with an IDH1 c.395G>A (R132H) mutation and 1p/19q codeletion, which led to a definitive diagnosis of oligodendroglioma. A common point mutation in IDH1 (R132H) was detected in the patient's enchondroma and glioma-matched pair specimens. To the best of our knowledge, this is the first case of molecularly confirmed oligodendroglioma associated with enchondromatosis. Furthermore, identification of a common IDH1 mutation in enchondroma and oligodendroglioma-matched pair specimens supports the hypothesis that IDH1/2 mosaicism initiates tumorigenesis [4].

1)

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