Ollier disease

Ollier disease is a rare disorder characterised by the development of enchondromatosis in long bones.

Ollier disease and Maffucci syndrome are rare syndromes in which there is deforming dysplasia of cartilage, primarily but not exclusively involving the metaphyses and diaphyses of long bones. In a minority of patients, dysplasia can lead to sarcomatous degeneration, producing chondrosarcomas. There also appears to be an association with other neoplasms. Little has been written about the association between Ollier disease and intracranial tumors.

Some of these patients also develop other unrelated tumors, such as gliomas, that harbor IDH mutations, suggesting that an IDH mutation is a common genetic event in the tumorigenesis in this group of patients. Tan et al., illustrated an interesting case of multifocal IDH-mutant astrocytomas in an OD patient with 8 years of follow-up. They first demonstrated identical IDH mutations in the brain tumor samples from various locations in this patient, but different 1p,19q results by fluorescent in-situ hybridization, different whole genome copy number profiles by OncoScan analysis, and a discrepant IDH2M131I mutation unique to one tumor, supporting a multifocal disease process in the setting of somatic IDH mosaicism [1)]

Case reports

A 35-year-old woman who was diagnosed with OD at age 24 underwent resection surgery for multifocal tumors located at the right and left frontal lobes that were discovered incidentally. No apparent spatial connection was observed on preoperative magnetic resonance imaging. Pathological examinations revealed tumor cells with a perinuclear halo in the left frontal lobe tumor, whereas astrocytic tumor cells were observed in the right frontal lobe tumor. Based on positive IDH1 R132H immunostaining and the result of 1p/19q fluorescent in situ hybridization, pathological diagnoses were IDH mutant and 1p/19q-codeleted oligodendroglioma in the right frontal lobe tumor and IDH mutant astrocytoma in the left frontal lobe tumor, respectively. The DNA sequencing revealed IDH1 R132H mutation in the peripheral blood sample and frontal lobe tumors. This case suggested that in patients with OD, astrocytoma and oligodendroglioma can co-occur within the same individual simultaneously, and IDH1 R132H mutation was associated with supratentorial development of gliomas [2)]

2012

Pearce et al. present a 19-year-old man with Ollier disease who also developed three synchronous brain tumours. Craniotomy, biopsy and debulking was performed for one lesion followed by a period of observation, and 9 months later he underwent a second craniotomy and debulking for symptomatic progression. Histopathological examination revealed a diagnosis of multifocal diffuse glioma (World Health Organization grade II). This report highlights the increased incidence of primary brain tumours in patients with Ollier disease and identifies the importance of screening patients with Ollier disease for primary neoplasms [3)].

2011

Mitchell et al. describe an unusual case of oligodendroglial GC diagnosed in a 16-year-old boy with Ollier disease. This is the first case of GC reported in a child with Ollier disease [4].

2009

Ranger et al. present the case of a 6-year-old girl with left arm osseous changes consistent with Ollier disease and a biopsy-proven thalamic glioblastoma multiforme. They then examine the co-occurrence of brain tumors in conjunction with a dyschondroplasia syndrome in children and adolescents to assess the presentation, treatment offered, and disease course of similar cases. Eight other such cases were identified, 6 in patients with Ollier disease (ranging in age from 7 to 18 years), and 2 with Maffucci syndrome (both in late adolescence). Including our own patient, 7 of the 9 cases of comorbid dyschondroplasia and intracranial malignancy occurred in girls. Some patients presented soon after the acute onset of symptoms, and others had a more subtle, protracted course over as many as 2 years. Some tumors were deemed resectable and others not. In only 1 instance was follow-up beyond 1 year reported [5].

1)
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2)
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5)
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