Pediatric Neurology: Chapter 126. Opsoclonus-myoclonus syndrome (Handbook of Clinical Neurology)

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Opsoclonus–myoclonus syndrome is a very rare disorder with onset usually in the second year of life, and the clinical features of opsoclonus, myoclonus, ataxia, irritability, sleep disturbance, and, often but by no means invariably, an associated neuroblastoma. There is no diagnostic test; brain imaging is normal and other investigations produce nonspecific results; the diagnosis is clinical and the condition is not infrequently mistaken for acute cerebellar ataxia. The pathophysiology is thought to be immunological on the basis of the paraneoplasticity and the symptomatic (though often incomplete) response to immunomodulatory therapies; a number of autoantibodies have been identified to a variety of antigens and cerebrospinal fluid B-cell numbers found to be increased but no diagnostic immunological marker has yet been identified. Therapeutic benefit has been described with steroids, intravenous immunoglobulin, cyclophosphamide, azathioprine, and rituximab, but randomized trials are extremely difficult because of the rarity of the condition. Successful treatment of the tumor, when present, does not usually improve neurological outcome. Disease course may be monophasic or chronic relapsing and children are often left with long-term motor, behavioral, and cognitive sequelae.

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