Pediatric Neurology Part III: Chapter 193. Joubert syndrome and related disorders (Handbook of Clinical Neurology)

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Joubert syndrome (JS) is a rare autosomal recessive condition characterized by a peculiar midbrain-hindbrain malformation, known as the molar tooth sign (MTS). The neurological presentation of JS includes hypotonia that evolves into ataxia, developmental delay, abnormal eye movements, and neonatal breathing abnormalities. This picture is often associated with variable multiorgan involvement, mainly of the retina, kidneys, and liver, defining a group of conditions termed Joubert syndrome and related disorders (JSRDs), that share the MTS. To date, 16 causative genes have been identified, all encoding for proteins expressed in the primary cilium or its apparatus. Indeed, JSRD present clinical and genetic overlap with a growing field of disorders due to mutations in ciliary proteins, that are collectively known as “ciliopathies.” These include isolated nephronophthisis, Senior–Løken syndrome, Bardet–Biedl syndrome and, in particular, Meckel syndrome, which is allelic at JSRD at seven distinct loci. Significant genotype-phenotype correlates are emerging between specific clinical presentations and mutations in JSRD genes, with relevant implications in terms of molecular diagnosis, clinical follow-up, and management of mutated patients. Moreover, the identification of mutations allows early prenatal diagnosis in couples at risk, while fetal neuroimaging may remain uninformative until the late second trimester of pregnancy.

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