Peripheral Nerve Disorders: Chapter 49. Early onset (childhood) monogenic neuropathies (Handbook of Clinical Neurology)

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Hereditary neuropathies (HN) with onset in childhood are categorized according to clinical presentation, pathogenic mechanism based on electrophysiology, genetic transmission and, in selected cases, pathological findings. Especially relevant to pediatrics are the items “secondary” versus “primary” neuropathy, “syndromic versus nonsyndromic,” and “period of life.” Different combinations of these parameters frequently point toward specific monogenic disorders. Ruling out a neuropathy secondary to a generalized metabolic disorder remains the first concern in pediatrics. As a rule, metabolic diseases include additional, orienting symptoms or signs, and their biochemical diagnosis is based on logical algorithms. Primary, motor sensory are the most frequent HN and are dominated by demyelinating autosomal dominant (AD) forms (CMT1). Other forms include demyelinating autosomal recessive (AR) forms, axonal AD/AR forms, and forms with “intermediate” electrophysiological phenotype. Peripheral motor neuron disorders are dominated by AR SMN-linked spinal muscular atrophies. (Distal) hereditary motor neuropathies represent <10% of HN but exhibit large clinical and genetic heterogeneity. Sensory/dysautonomic HN involves five classic subtypes, each one related to specific genes. However, genetic heterogeneity is larger than initially suspected. Syndromic HN distinguish “purely neurological syndromes”, which are multisystemic, such as spinocerebellar atrophies +, spastic paraplegias +, etc. Peripheral neuropathy is possibly the presenting feature, including in childhood. Autosomal recessive forms, on average, start more frequently in childhood. “Multiorgan syndromes”, on the other hand, are more specific to Pediatrics. AR forms, which are clearly degenerative, prompt the investigation of a large set of pleiotropic genes. Other syndromes expressed in the perinatal period are mainly developmental disorders, and can sometimes be related to specific transcription factors. Systematic malformative workup and ethical considerations are necessary. Altogether, >40 genes with various biological functions have been found to be responsible for primary HN. Many are responsible for various phenotypes, including some without the polyneuropathic trait, and some for various types of transmission.

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