Pediatric Neurology Part III: Chapter 194. Progressive dystonia (Handbook of Clinical Neurology)

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Progressive dystonias are a clinically and genetically heterogeneous group of movement disorders. In the primary forms, dystonia is the only sign of the disease, and the cause is either unknown or genetic. In the secondary forms, dystonia is usually only one of several disease manifestations and the cause may be genetic or due to other insults. Monogenic defects have been found to underlie many forms of dystonia syndromes, which are designated DYT1-20. Dystonias with known genes include DYT1 and DYT6 dystonia, presenting as isolated torsion dystonia, as well as DYT5 (dopa-responsive dystonia), DYT11 (myoclonus-dystonia), and DYT12 (rapid-onset dystonia-parkinsonism), where dystonia occurs in conjunction with other types of movement disorders. All of these conditions follow an autosomal dominant mode of inheritance, usually develop in childhood or early adolescence, and show an initially progressive course with stabilization in early adulthood. In secondary dystonias, there are often atypical features and additional neurological signs, such as prominent tongue and perioral involvement, pyramidal signs, ataxia, oculomotor abnormalities, or cognitive disturbances. Acquired brain lesions typically affect the putamen, thalamus, or globus pallidus and cause contralateral hemidystonia. Dystonia can be part of the clinical syndrome in many heredodegenerative disorders, or may be drug-induced or psychogenic.

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