Pediatric Neurology Part I: Chapter 57. Chromosome disorders associated with epilepsy (Handbook of Clinical Neurology)

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Epilepsy is a feature of several hundred chromosome abnormalities. However, there are relatively few conditions in which epilepsy is a consistent feature and even fewer in which the electroclinical phenotype is recognizable. Advances in cytogenetics and molecular genetics are leading to the detection of more complex and smaller chromosomal re-arrangements, duplications, and deletions using techniques such as comparative genome hybridization (CGH). This will provide new challenges for the epilepsy specialist who, in partnership with the geneticist, will have to judge the clinical relevance of these abnormalities. Most chromosome anomalies associated with epilepsy are individually rare therefore clinicians must continue to collaborate to describe novel electroclinical phenotypes. Cytogenetic studies should be requested in all individuals with refractory epilepsy and no clear underlying cause even in cases with no dysmorphic features, no learning disability, and an EEG suggestive of genetic generalized epilepsy. In syndromes where epilepsy is a consistent feature the seizure semiology and EEG features can suggest a specific diagnosis and guide the clinician to the appropriate cytogenetic investigation. An early correct diagnosis can save unnecessary investigations and guide prognosis. Children with chromosomal disorders frequently have learning disability, which can be further compromised by an epileptic encephalopathy. Medications should be targeted to specific seizure types.

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