Pediatric Neurology Part I: Chapter 30. Cognitive and medical features of chromosomal aneuploidy (Handbook of Clinical Neurology)

Christa Hutaff-Lee, Lisa Cordeiro, Nicole Tartaglia

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This chapter describes the physical characteristics, medical complications, and cognitive and psychological profiles that are associated with chromosomal aneuploidy conditions, a group of conditions in which individuals are born with one or more additional chromosome. Overall, chromosomal aneuploidy conditions occur in approximately 1 in 250 children. Information regarding autosomal disorders including trisomy 21 (Down syndrome), trisomy 13 (Patau syndrome), and trisomy 18 (Edward syndrome) are presented. Sex chromosome aneuploidy conditions such as Klinefelter syndrome (47,XXY), XYY, trisomy X, and Turner syndrome (45,X), in addition to less frequently occurring tetrasomy and pentasomy conditions are also covered. Treatment recommendations and suggestions for future research directions are discussed.

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