Pediatric Neurology Part III: Chapter 173. Neuronal ceroid lipofuscinoses (Handbook of Clinical Neurology)

Brigitte Chabrol, Catherine Caillaud, Berge Minassian

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Neuronal ceroid lipofuscinoses (NCL) represent a group of autosomal recessive neurodegenerative disorders, presenting with myoclonic epilepsy, psychomotor delay, progressive loss of vision, and early death. Four main clinical forms have been delineated (infantile, late infantile, juvenile, and adult), but many other variants have also been described. At least 14 genetically distinct NCL, designated CLN1 to CLN14, are presently known.The identification of the deficient protein and/or the genetic defect is required for a specific diagnosis, which is necessary for a reliable genetic counseling in at-risk families.

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