198. Juvenile Huntington's Disease: Unusual Presentation in Three Children

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Huntington's disease (HD) is a genetically determined degenerative condition characterized by progressive dementia, personality change, and chorea. Only 5% of HD patients present before the age of 14 years, and onset before 6 years is exceptionally rare. We report on 3 children with unique features: very early onset (12–14 mo in one) and severe, intractable seizures. Our methods involved retrospective analysis of 3 children, including detailed electroencephalograms, in a brother, sister, and half sister.

Results are seen in the Table:

Due to unique presentation, extensive neurological workup was undertaken. Clinical diagnosis emerged when the father developed adult-onset HD. Subsequently, gene analysis revealed the mutation in Patient III. Juvenile HD deserves consideration in familial intractable epilepsy with loss of developmental milestones. Prognosis seems worst with earliest age of onset.

Reference

1. Farrel LA, Conneally PM. A genetic model for age at onset in Huntington disease. Am) Hum Genet 1985;37:350–357

Patient	I	II	III
Onset of motor delay (mo)	12–14	36	72
Onset of cognitive delay (mo)	14–16	36	72
Seizure onset (mo)	24	36	9 yr
Initial seizure type	Generalized tonic	Generalized tonic	Complex partial seizure
Death (yr)	6	7 (dying)	16 (alive)