Pontine tegmental cap dysplasia

A hindbrain malformation caused by defective neuronal migration

Figure Imaging features of pontine tegmental cap dysplasia

Fast spin echo/T2-weighted MRI showing flat ventral pons and a dorsal vault (arrow) projecting from the pontine tegmentum into the fourth ventricle (A); very thin (B) or not visible (C) middle cerebellar peduncles (arrowheads); abnormal shape and course of superior cerebellar peduncles (arrowheads) (D, E); very narrow internal auditory canals (arrows) (F).

A 13-month-old girl presented with developmental delay, generalized hypotonia, vertical pendular nystagmus, and cranial nerves IV, V, and VII paresis. MRI (figure) revealed pontine hypoplasia with ectopic dorsal transverse pontine fibers (tegmental cap). Concomitant profound sensorineural deafness and a butterfly Th_2 vertebra led to the syndromic diagnosis of pontine tegmental cap dysplasia (PTCD).

PTCD has been described to date in several patients.^{1,2} It is suggested that many cases of pontocerebellar hypoplasia or Moebius syndrome should be revised for the features of PTCD.² Pathomechanism of the condition involves defect in migration or navigation of axons of rhombencephalic neurons.¹

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