A severe form of epidermal nevus syndrome associated with brainstem and cerebellar malformations and neonatal medulloblastoma.


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Abstract

Here we report a boy with epidermal nevus syndrome associated with brainstem and cerebellar malformations and neonatal medulloblastoma. The patient had epidermal nevi and complicated brain malformations including macrocephaly with polymicrogyria, dysmorphic and enlarged midbrain tectum, enlarged cerebellar hemispheres with small and maloriented folia. The patient died after surgical resection of medulloblastoma which was newly recognized on MRI at 51 days of age. Postmortem pathological examinations showed very unique and bizarre malformation of the midbrain and hindbrain. The cerebellar cortex exhibited a coarse, irregular and bumpy surface, blurred border between the Purkinje cell layer and internal granule cell layer, and many foci of heterotopia in the cerebellar white matter. The brainstem showed multiple anomalies, including enlargement of superior colliculi, hypoplasia of pyramidal tracts and dysplasia of inferior olivary nuclei. The unusual constellation of brain malformations of our patient will widen the spectrum of epidermal nevus syndrome.

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