Pediatric Neurology Part I: Chapter 41. Epidermal nevus syndrome (Handbook of Clinical Neurology)

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Epidermal nevus syndrome (ENS) is an inclusive term for a heterogeneous group of congenital disorders characterized by the presence of epidermal nevi associated with systemic involvement. These disorders, as are all primary neurocutaneous syndromes, are neurocristopathies. The epidermal nevi that follow the lines of Blaschko and most systemic anomalies in skeletal, ocular, cardiovascular, endocrine, and orodental tissues, as well as lipomas, are due to defective neural crest. The most important and frequent anomaly in the brain in all forms of epidermal nevus syndromes (ENSs) is hemimegalencephaly (HME). This malformation often is not recognized, despite being the principal cause of neurological manifestations in ENSs. They consist mainly of epilepsy and developmental delay or intellectual disability. The onset of epilepsy in ENS usually is in early infancy, often as infantile spasms. Several syndromic forms have been delineated. I propose the term “Heide's syndrome” for those distinctive cases with the typical triad of hemifacial epidermal nevus, ipsilateral facial lipoma, and hemimegalencephaly. Most ENSs are sporadic. The mechanism is thought to be genetic mosaicism with a lethal autosomal dominant gene. Specific genetic mutations (PTEN, FGFR3, PIK3CA, and AKT1) have been documented in some patients. The large number of contributors for over more than a century and a half to the description of these disorders precludes the use of new author eponyms.

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