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Epidermal nevus syndrome and PIK3CA-Related Overgrowth Spectrum (PROS) in Neonates

Background: Epidermal nevus syndrome (ENS) is a spectrum within the broader category of neurocutaneous syndromes, as post-zygotic somatic mutations that affect the nervous system skin and other tissues. Mutation in the PIK signaling pathway, closely related to mTOR, causes alteration in cellular lineage, growth and morphogenesis. EN spectrum includes Proteus (PIK1) and CLOVES that clinically result in overgrowth of various tissues including the brain as hemimegaloencephaly (HME). HME also can occur as an isolated focal cerebral lesion, usually due to PIK3 mutation. The most frequent neurological manifestations of ENS are epilepsy, hemiparesis, cognitive/intellectual deficits and global developmental delay.

Objective: To correlate CNS dysmorphic overgrowth lesions with clinical presentation, especially in the neonatal period and infancy, in the context of neurocutaneous syndromes.

Methods: Synthesis of data on phenotype and clinical neurological presentation correlated with neuroimaging, EEG, neuropathological and genetic data, based upon personal experience with 20 ENS patients over years and extensive literature review.

Results: ENS exhibits a spectrum of phenotypes. Epilepsy is the earliest and most frequent neurological presentation, from the neonatal period or early infancy, as focal or multifocal seizures or infantile epilepsies including epileptic (infantile) spasms and Ohtahara syndrome. The most frequent underlying brain malformation is HME. Early diagnosis prenatally or in the neonate is important because prompt intervention with anti-seizure medications and mTOR inhibitors is feasible and justified for better outcome.

Conclusion: The presence since birth of nevi, cutaneous angiomata and overgrowth of extremities and visceral organs, sometimes more evident and progressive postnatally, is reason to perform brain MRI to exclude HME, and EEG to detect early subclinical epileptogenic foci. Early detection and anti-seizure treatment may delay or prevent the onset of severe infantile epilepsies. Detection of fetuses or neonates with HME by prenatal ultrasound or MRI may justify mTOR inhibitors (rapamycin) to arrest or attenuate progressive overgrowth.

Biography

Flores-Sarnat is professor of Paediatrics and Clinical Neurosciences at the University of Calgary and Alberta Children's Hospital, Canada. She is trained in both neonatalogy and paediatric neurology and is former Head of Paediatric Neurology at the Instituto Nacional de Pediatría, a large university children's teaching hospital in Mexico City. Her clinical and research interests are in the fields of fetal and neonatal neurology, brain malformations, neurocutaneous syndromes and early-onset epilepsy. She is the author or co-author of 60 research articles and many textbook chapters.

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