



Pediatric Neurology Part I: Chapter 12. Neuroembryology and brain malformations: an overview (Handbook of Clinical Neurology)

Harvey B. Sarnat, Laura Flores-Sarnat

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Modern neuroembryology integrates descriptive morphogenesis with more recent insight into molecular genetic programming and data enabled by cell-specific tissue markers that further define histogenesis. Maturation of individual neurons involves the development of energy pumps to maintain membrane excitability, ion channels, and membrane receptors. Most malformations of the nervous system are best understood in the context of aberrations of normal developmental processes that result in abnormal structure and function. Early malformations usually are disorders of genetic expression along gradients of the three axes of the neural tube, defective segmentation, or mixed lineages of individual cells. Later disorders mainly involve cellular migrations, axonal pathfinding, synaptogenesis, and myelination. Advances in neuroimaging now enable the diagnosis of many malformations in utero, at birth, or in early infancy in the living patient by abnormal macroscopic form of the brain. These images are complimented by modern neuropathological methods that disclose microscopic, immunocytochemical, and subcellular details beyond the resolution of MRI. Correlations may be made of both normal and abnormal ontogenesis with clinical neurological and EEG maturation in the preterm or term neonate for a better understanding of perinatal neurological disease. Precision in terminology is a key to scientific communication.

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