



Pediatric Neurology Part I: Chapter 36. Genetics of neural crest and neurocutaneous syndromes (Handbook of Clinical Neurology)

Harvey B. Sarnat, Laura Flores-Sarnat

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Neural crest progenitor cells are identified at the lateral margins of the neural placode at the time of gastrulation. With folding of the placode, these precursors are brought to the dorsal midline of the neural tube at the site of closure, become committed to neural crest lineage and almost immediately migrate peripherally to various predetermined sites in the body and then differentiate as a variety of cellular types in all three of the traditional “germ layers.” All of these processes of migration and differentiation of neural crest are precisely genetically programmed, temporally and spatially, by a variety of genes. Primary neurocutaneous syndromes are all very different diseases with different genetic mutations, but the unifying factor amongst them is that all are neurocristopathies and can be explained as such, including the tumor-suppressor function of several of these genes, especially those of neurofibromatosis 1 and 2 and tuberous sclerosis. This chapter reviews the principal genes that program neural crest development and also are documented, implicated, or suspected in the pathogenesis of neurocutaneous syndromes. Recent genetic discoveries are noted in epidermal nevus syndrome, including Proteus syndrome and their association with hemimegalencephaly and congenital infiltrating lipomatosis of the face.

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