

Special Issue

New Insights into Neurocutaneous Syndromes in Children and in the Transitional Age

Message from the Guest Editor

Neurocutaneous syndromes are a heterogeneous group of diseases (mainly) affecting the skin, the nervous system and the eye. These conditions, however, may also affect many other organs or systems including the heart, vessels, lungs, kidneys and bones, thus configuring complex (malformation) syndromes. In the past few decades, the molecular genetic and cellular bases of an increasing number of neurocutaneous syndromes have been unravelled, shedding light on the interplays between intra- and extra-neuronal signalling pathways encompassing receptor-to-protein and protein-to-protein cascades involving the RAS, MAPK/MEK, ERK, mTOR, RHOA, PI3K/AKT, PTEN, GNAQ and GNA11 pathways, which also explain the phenotypic variability and overlapping. Newer imaging techniques helped clinicians to better diagnose and manage affected individuals. The general panorama of neurocutaneous syndromes is rapidly changing; new conditions are increasingly recognised and the respective frequencies of the various types have changed with recent studies.

Guest Editor

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