

Special Issue

New Molecular Insights into Neurocutaneous Syndromes

Message from the Guest Editor

Neurocutaneous syndrome is a category of genetic disease characterized by both skin and brain lesions with tumor formation, which comprises neurofibromatosis type 1 (NF1), tuberous sclerosis complex (TSC), Sturge–Weber syndrome (SWS), and various other disorders. In recent years, causative genes of these syndromes have been identified, and these molecules have been revealed to commonly regulate RAS/MAPK and PI3K/mTOR pathways, which are also shared with cancer. This has led to the emergence of a new concept based on which neurocutaneous syndromes have a common molecular basis as a framework, and clinical application of molecular targeted drugs such as mTOR inhibitors has been done. SWS (GNAQ) and some rare RAS mutation syndromes are caused by specific gain-of-function mutations as somatic mosaicism. In this Special Issue, we will focus on new molecular insight of neurocutaneous syndromes, welcoming all those studies on more sophisticated genetic diagnosis, analysis of molecular pathway dysfunctions and cross-talking abnormalities associated with gene mutations, and quest for possibilities of new molecular targeted drugs.

Guest Editor

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