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Role of SHANK3 gene in Phelan-McDermid syndrome (PMS)

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Phelan-McDermid syndrome (PMS) is a postnatal neurodegenerative disorder which is caused by the mutations in the gene called SHANK3 which is a member of the highly conserved SHANK family of synaptic scaffolding proteins. The function of *SHANK3* is to connect ion channels and receptors in the post-synaptic membrane to the cytoskeleton and to signal transduction pathways. The disruption of SHANK3 (Srchomology 3 (SH3) and multiple ankyrin repeat domains 3) /ProSAP2(Proline-rich synapse-associated protein 2) gene by balanced rearrangement and the alteration of *SHANK3* by a frame shift mutation lead to the language deficits and autistic characters of PMS and also *SHANK3* has increased actin polymerization and promoted spine formation. Haplo insufficiency of *SHANK3* gene is the cause of the major neurological features associated with 22q13 deletion. The PMS is a microdeletion syndrome resulting from loss of 22q13 by simple deletion, unbalanced translocation, ring chromosome formation, or other irregular structural change. PMS is characterized by developmental delay, delayed or absent speech, autistic-like behavior, normal to accelerated growth, and minor dysmorphic features including dolichocephaly, full brow and large or prominent ears. The genetic alterations that lead to the syndrome will mainly affect the gene *SHANK3* which codes for a scaffold protein localized in the postsynaptic densities of excitatory synapses, connecting membrane receptors to cytoskeleton. The present aim is to explain in depth of the clinical and genetic facets of the PMS, with more concentration on the domain architecture of *SHANK3* protein, by shedding light on the mutations involved in the protein families.

Biography

Sujitha Mariappan has completed BTech Biotechnology from Kalasalingam University 2011 and currently pursuing final year MTech Biotechnology from Karunya University, India. She has communicated one paper in Thomson Reuter's journal on title "Association of the SHANK3 gene mutations coupled with 22q13.3 deletion syndrome". She has also participated in "Stimulating Bio-Entrepreneurs Talk" conducted by ABLE Biotech.She has attended national level workshop on "Recent Techniques in Cell Culture Cytotoxity Assay" at Karunya University, Coimbatore.

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