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Psychosomatic dysfunction in Rasopathies

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RASopathies are resulting from germ line mutations of the proto-oncogene HRAS. Many of these mutations affect SHP2, SOS1, RAS, RAF and MEK proteins. Dr White says. a group of related disorders including Costello syndrome, Noonan syndrome (NS), cardiofaciocutaneous (CFC) syndrome, and neurofibromatosis 1 (NF1), caused by abnormal functioning of the Ras□mitogen□activated protein kinase (RAS/MapK) pathway. Ras/MAPK pathway is an essential signalling pathway that controls the cell proliferation, differentiation, survival and its dysregulation causes clinically overlapping genetic disorders, called as 'Rasopathies'. In this pathway, Ras, a GTPase, transmits the extracellular signaling from the receptor tyrosine kinases to two serine/threonine kinases (Raf and MEK) and, finally, to the activation of MAPKs. She has led the implementation of exome sequencing (a genomic technique for sequencing all of the protein-coding regions of genes in a genome known as the exome) at The Royal Children's Hospital and the Murdoch Children's Research Institute (Melbourne, Australia). Aoki et al. discovered that these germline mutations altered the residues Gly12 and Gly13 in HRAS's P-loop and had been identified previously as somatic defects in various tumours. Rasopathies are developmental disorders characterised by postnatal growth inhibition with delayed skeletal maturation and psychomotor retardation. In 2009, gain-of-function missence mutation in SHOC2, C4a> G(Ps2g), identified in NS-like syndrome with loose anagen hair, severe intellectual disability, hyper nasal voice and skin abnormalities. HRAS consists of six exons Somatic mutation hotspots are bases encoding the glycines in Positions 12 and 13 and the glutamine in Position 61. Missense mutations at these positions lead to increased activity of the gene product. Germ line mutations affect similar codons, it can be inferred that they have a similar effect on the gene product. The splicing efficiency of activating HRAS mutations can determine the rasopathy phenotype and frequency in Cancer. This unravels a potential for the development of new anti-cancer therapies based on SSO-mediated HRAS exon 2 skipping. Pathway modulators or small molecule inhibitors such as statins causes significant improvement in verbal and nonverbal memory, visual attention & efficacy by inhibiting the posttranscriptional lipid modification of RAS. it is a potential targeted therapeutic drug to improve the stature of patients affected with disruption of the RAS/MEK/ERK pathway.

Biography

Ramachandran Muthiah, Consultant at Zion hospital, Azhagiamandapam and Morning Star hospital, Marthandam, Kanyakumari District, India. Completed primary education at Anaan vilai, keezhkulam and secondary education at Concordia Higher secondary school, Pootteti, MBBS in 1988 Worked as medical officer in Rural health services for 5 years (keezhachekkarakudi, Aryappapuram