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World Conference on ADDICTION PSYCHIATRY

July 06, 2022 | Webinar

Psychomotor dysfunction in Rasopathies

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Rasopathies are developmental disorders characterised by postnatal growth retardation with delayed skeletal maturation, psychomotor dysfunction, cutis lax, and acanthosis Nigerians and 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 Ras mitogen activated protein kinase signalling pathway that controls cell proliferation, differentiation and survival. In this pathway, Ras, a GTPase, transmits extracellular signalling from receptor tyrosine kinases to two serine/threonine kinases (Raf and MEK) and to the activation of MAPKs. Psychosomatic dysfunction is a common feature of rasopathies. Isoprenylation involves the enzyme farnesyl transferees (FTase) transferring a farnesyl group from farnesyl pyrophosphate (FPP) to the pre-Ras protein. 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. RAF-1 inhibition by C-type Natriuretic of the gremlin mutations to restore normal protein functions is anticipated as a new therapeutic option. Oxidative stress and free radicals- determine non-neoplastic clinical features such as elastin anomalies, alteration of skin and appendages, developmental retardation and cardiac defects. PAR therapy (potassium acerbate with ribose). Causes a reduction in oxidative stress biomarkers in parallel with improvement of clinical features. It combines the antioxidant action of vitamin C with stabilizing intracellular effects of potassium and causes improvement of skin and appendage lesions, better evolution of psychomotor development, no Progression of heart hypertrophy, nor tumour development. It is low cost, no side-effects, orally administered and useful

Biography

Ramachandran Muthiah, Consultant Physician & Cardiologist, Zion hospital, Azhagiamandapam and Morning star hospital, India. He was born on 10/5/1966. He completed his School education from Annan villain primary school and Concordia higher secondary school, Potted and graduated MBBS from Tirunelveli medical college under Madurai Kama raj university in 1989 and MD General medicine in 1996 (Tirunelveli medical college), DM cardiology in 2003 (Madurai medical college) under MGR medical university, Chennai. Worked as medical officer in rural health services for 5 years (keelachekkarakudi, arryapapuram PHCs, and ESI hospital at Singanallur) and teaching faculty at Coimbatore, Madras, Kanyakumari and Thoothukudi Gov. Medical colleges. Marital status troubled at Coimbatore in 2004 and thereafter further family living get prevented. Published many papers in cardio source, American College of Cardiology Foundation, Case Reports in Clinical Medicine (SCIRP) and Journal of Saudi Heart Association.