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Psychomotor Dysfunction in Rasopathies

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ABOUT THE STUDY

Rasopathies are developmental disorders characterized by postnatal growth retardation with delayed skeletal maturation, psychomotor dysfunction, cutis laxa, 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 (RAS/MapK) signalling pathway that controls cell proliferation, differentiation and survival. In this pathway, Ras, a GTPase, transmits extracellular signaling 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 Transferase (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 and efficacy by inhibiting the posttranscriptional lipid modification of RAS. RAF-1 inhibition by C-type Natriuretic Peptide (CNP) improved bone growth in preclinical animal models and it is a potential targeted therapeutic drug to improve the stature of patients. Gene correction of the germ line 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 ascorbate with ribose). Causes a reduction in oxidative stress biomarkers in parallel with improvement of clinical features [1-5]. 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 tumor development. It is low cost, no side effects, orally administered and useful for all genetic syndromes (**Fig. 1**).

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Fig. 1. 9 years old boy with radiant smile.

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