

Welcome

Written by Administrator

Friday, 03 July 2009 11:59 - Last Updated Friday, 03 July 2009 14:20

Recently a class of developmental syndromes, the 'RASopathies', is caused by germline mutations in genes that encode protein components of the critical signal transduction cascade, the Ras/mitogen activated protein kinase (MAPK) pathway. At present, these syndromes include:-

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1) Noonan syndrome

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2) LEOPARD syndrome

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3) Hereditary Gingival fibromatosis type 1

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4) Capillary malformation-AV malformation syndrome

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5) Neurofibromatosis type 1

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6) Legius syndrome

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7) Costello syndrom

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8) Cardio-facio-cutaneous syndrome

- 9) Autoimmune lymphoproliferative syndrome

The Ras/MAPK pathway is essential in the regulation of the cell cycle, differentiation, growth and cell senescence, all of which are critical to normal development. Therefore, it is not surprising that its dysregulation has profound effects on development. Each syndrome in this class of developmental syndromes, exhibits unique phenotypic features, however, since they all cause dysregulation of the Ras/MAPK pathway, there are numerous overlapping phenotypic features among the syndromes, including characteristic facial features, cardiac defects, cutaneous abnormalities, neurocognitive delay and a predisposition to malignancies.

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