The Genetic Landscape of Rasopathies in Hong Kong

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Abstract

Rasopathies are a genetically heterogeneous group of genetic diseases characterized by disturbance of the RAS-MAPK signaling pathway. With an incidence of 1/1,000-2,500, the most common condition among the rasopathies is Noonan syndrome, a well-known dysmorphic syndrome and a common reason for referral to the Clinical Genetic Service. Since the identification of the *PTPN11* gene in 2001, there are more than 10 genes implicated in Noonan syndrome. Diagnosing Noonan syndrome has moved from a "genetic" approach to a "genomic" one. This presentation summarized the molecular defects found in over 180 local patients with Noonan syndrome and two other phenotypically overlapping disorders known as Cardiofaciocutaneous syndrome and Costello syndrome. The distinguishing molecular and clinical features were highlighted.