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Recent developments in neurofibromatosis type 1 and RASopathies: Novel therapeutic targets

Neurofibromatosis type 1 (NF1) is a common autosomal dominantly inherited tumor predisposition syndrome affecting 1/3000-4000 individuals worldwide. Neurofibromin, the protein encoded by *NF1* gene down-regulates Ras. The Ras/MAPK pathway plays an essential role in regulating the cell cycle and cellular growth, differentiation and senescence, all of which are critical to normal development. In NF1 patients, benign plexiform neurofibromas can transform into aggressive malignant tumors called Malignant Peripheral Nerve Sheath Tumors (MPNSTs), currently, there are no effective treatments for MPNSTs. The RASopathies are a clinically defined group of medical genetic syndromes caused by germline mutations in genes that encode components or regulators of the Ras/MAPK pathway. These disorders include neurofibromatosis type 1, Legius syndrome, Noonan syndrome, Noonan syndrome with multiple lentigines (LEOPARD), capillary arteriovenous malformation syndrome, Costello syndrome cardiofaciocutaneous syndrome and SYNGAP1 autism. Because of the common underlying Ras/MAPK pathway dysregulation, they have overlapping phenotypic features, including cancer, facial dysmorphia, neurocognitive impairment, pain and cardiovascular, musculoskeletal, gastrointestinal and cutaneous abnormalities. NF1 was the first Rasopathy syndrome reported in the RAS pathway. Several ongoing clinical trials exist for RASopathies including NF1 and effective treatments for certain clinical features are on horizon. NF1 also has great importance and significance in a number of sporadic cancers and functions as a central tumor suppressor gene in these cancers. With the recent advances in sequencing technologies, high-throughput drug discovery platforms, increasing availability of more sophisticated animal models and application of the state-of-art tumor imaging techniques, diagnosis and treatment of patients with RASopathies is improving.

Biography

Professor Meena Upadhyaya OBE, is a distinguished professor in the Division of Cancer and Genetics, Cardiff university, UK. She obtained her PhD in Medical Genetics from Cardiff University and gained FRCPath from Royal College of Pathologists, London. She has made substantial contributions to the molecular understanding of a number of genetic conditions including neurofibromatosis type 1, facioscapulohumeral muscular dystrophy, Legius syndrome, Charcot Marie Tooth disease, Duchenne Muscular Dystrophy, Sotos syndrome, Hunters syndrome inter alia and also supervised numerous PhD, MD, MSc and undergraduate students. Prof. Upadhyaya has published well over 200 papers in peer reviewed journals and co-edited three books and sits on a number of Committees. She is also passionate about women's equality in all spheres of life.

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