Novel functions of proto-oncogenes in human development

Yoko Aoki MD PhD

(Tohoku University, Graduate School of Medicine, Assistant Professor)

【Outline of survey】

Costello syndrome, cardio-facio-cutaneous (CFC) syndrome and Noonan syndrome are congenital anomaly syndromes characterized by heart defects, facial dysmorphism, ectodermal abnormalities and mental retardation. Recently we discovered *HRAS* mutations in Costello syndrome and *KRAS* and *BRAF* mutations in CFC syndrome, suggesting the important role of the RAS/RAF/MEK/ERK pathway in human development. The aim of this project is to identify new genetic causes of patients with Noonan syndrome and related disorders. We will analyze the pathogenetic mechanisms of these disorders to explore novel functions of proto-oncogenes in human development.

【Expected results】

- Novel genetic causes for Noonan syndrome and related disorders will be identified.
- The novel functions in proto-oncogenes in human development will be explored.

【References】

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【Term of project】 FY2007－2011	【Budget allocation】 32,000,000 yen (2007 direct cost)

【Homepage address】 http://www.medgen.med.tohoku.ac.jp/