Project title:

Diverse rare diseases caused by functional dysregulation of Rho GTPase-related proteins

Project description:

RAS GTPase signaling alterations contribute to several developmental diseases. In particular, diverse rare genetic disorders are caused by mutations in the subfamily of Rho GTPase genes, such as CDC42 and RAC1.

The aim of this project is to identify novel disease-causing mutations occurring in Rho GTPases-related genes and to characterize their functional impact in vitro.

Researcher(s):

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