Open Peer Review on Qeios

Noonan Syndrome

National Cancer Institute

Source

National Cancer Institute. Noonan Syndrome. NCI Thesaurus. Code C34854.

A genetic syndrome caused by mutations in the PTPN11 gene (over 50% of the cases) or less frequently mutations in the SOS1, RAF1, or KRAS genes. It is characterized by short stature, webbed neck, hypertelorism, low-set ears, deafness, and thrombocytopenia or abnormal platelet function.