Noonan syndrome with multiple lentigines (LEOPARD)

Genetics

-PTPN11, RAF1, BRAF, MAP2K1

-AD

Clinical findings/Dysmorphic features

-Multiple lentigines

-Cardiac abnormalities (particularly hypertrophic cardiomyopathy)

-Poor linear growth/short stature; pectus deformity; SNHL

-Variable degree of cognitive deficits

-Café au lait macules (70%-80%)

-Facial features: hypertelorism, down slanting palpebral fissures, low set ears

Etiology

-Not known

Pathogenesis

-LoF mutations in PTPN11 ( vs. Noonan: GoF)

Genetic testing/diagnosis

-PTPN11 sequencing (90%), RAF1 (<5%), others rare