Jervell and Lange-Nielsen Syndrome

Genetics

-KCNQ1 (K+ channel protein; 11p15.5) and KCNE1 (K+ voltage-gated channel; 21q22.1)

-AR (Heterozygotes at risk for AD long QT a.k.a. Romano Ward syndrome)

Clinical findings/Dysmorphic features

-Congenital severe-profound bilateral SNHL

-Prolonged QT interval --> at risk for arrhythmia, syncope, and sudden death

Etiology

-high (1:200,000) in North Europe --> founder variants

Pathogenesis

-In cardiac cells: abnormal repolarization of the ventricular action potential

-In cochlear cells: abnormal depolarization of the auditory nerve

Genetic testing/diagnosis

-LoF variants in: KCNQ1 sequencing (90%), KCNE1 (10%)

Others

-Cochlear implants for HL, beta blockers, cardiac pacemakers, and/or implantable defibrillators

-Avoid QT prolonging drugs