Brugada Syndrome

Genetics

-Most common SCN5A (3p21; 15-30%), also 22 other genes (<1%)

-AD, except KCNE5 (XLR)

Clinical findings/Dysmorphic features

-ST-segment abnormalities in leads V1-V3 on electrocardiogram

-Syncope (temporary loss of consciousness)

-Nocturnal agonal respiration

-High risk of ventricular arrhythmias and sudden death

-Mainly during adulthood (2 days to 85 yrs), mean age of sudden death: 40 yrs

-May present as SIDS or sudden unexpected nocturnal death syndrome (SUNDS)

Etiology

-Prevalence of the disease in endemic areas is on the order of 1:2,000

Pathogenesis

-SCN5A encodes the α-subunit of the cardiac sodium channel and is responsible for phase 0 of the cardiac action potential

-Pathogenic variants in SCN5A result in a decrease in Na+ current --> lack of expression of the mutated channel or accelerated inactivation of the channel

Genetic testing/diagnosis

-Serial single-gene testing or gene panel

Others

-In countries in Southeast Asia in which SUNDS is endemic, it is the second cause (following accidents) of death of men < 40 years