Emanuel syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Emanuel syndrome. ORPHA:96170

Emanuel syndrome is a constitutional genomic disorder due to the presence of a supernumerary derivative 22 chromosome and characterized by severe intellectual disability, characteristic facial dysmorphism (micrognathia, hooded eyelids, upslanting downslanting parebral fissures, deep set eyes, low hanging columnella and long philtrum), congenital heart defects and kidney abnormalities.