

Aetiology

Aetiology

There is often no obvious aetiology; most abnormalities appear to be multifactorial with both genetic and environmental influences. There are well-recognised associations. Jacqueline Anne Noonan , 1928–2020, pediatric cardiologist, the University of Kentucky College of Medicine, Lexington, KY , USA, described this condition in 1963. Mary Clayton Holt , 1924–1993, cardiologist, The London Hospital for Women and Children, London, UK. Samuel Oram , 1913–1991, cardiologist, King’s College Hospital, London, UK. Holt and Oram described this syndrome in a joint paper in 1960. John Langdon Haydon Down , 1828–1896, physician, The London Hospital, London, UK. John Hilton Edwards , 1928–2007, Professor of Genetics, University of Oxford, Oxford, UK. Klaus Patau , 1908–1975, German-born American geneticist, University of Wisconsin–Madison, Madison, WI, USA. Henry Hubert Turner , 1892–1970, Professor of Medicine, The University of Oklahoma, Oklahoma City , OK, USA. Harry Klinefelter , 1912–1990, American rheumatologist and endocrinologist, first described the syndrome in 1942. Angelo M DiGeorge , 1921–2009, Professor of Pediatrics, Temple University , Philadelphia, PA, USA. John CP Williams , b. 1922, New Zealand born cardiologist, described the condition in 1961. Recognised associations with congenital heart disease /uni25CF /uni25CF /uni25CF /uni25CF /uni25CF

Maternal (environmental) factors Infection: rubella Disease: systemic lupus erythematosus, diabetes mellitus, maternal phenylketonuria Drugs/medications: alcohol abuse, warfarin, phenytoin, lithium, thalidomide Genetic factors Single gene defects: Marfan, Noonan and Holt-Oram syndromes; numerous single-gene disorders Chromosomal defects: trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome), trisomy 13 (Patau syndrome), Turner syndrome, Klinefelter syndrome Deletions: DiGeorge and Williams syndromes

Revision #1

Created 2025-12-31 15:22:04 UTC by Omar Ayman

Updated 2025-12-31 15:22:04 UTC by Omar Ayman