Williams Syndrome

National Cancer Institute

Source

National Cancer Institute. Williams Syndrome. NCI Thesaurus. Code C85232.

A rare syndrome caused by multiple gene deletions from a region of chromosome 7, including the deletion of CLIP2, ELN, GTF2I, GTF2IRD1 and LIMK1 genes. It is characterized by distinctive facial appearance (elfin facies), mild-to-moderate mental retardation, cheerfulness, cardiovascular abnormalities and infantile hypercalcemia.