

Genetics of Williams Syndrome

Williams syndrome is caused by the deletion of 1.5-1.8 megabases (measurement of length) of genetic material at chromosome 7q11.23. Although there are several genes included in the deleted region of chromosome 7, no single gene has been identified as causing Williams syndrome.

Genes that are typically deleted include; Elastin, LIMK1, CLIP2, GTF2IRD1 AND GTF2I.

Elastin is missing in 95-99% of individuals with Williams syndrome and therefore is the main gene used to genetically diagnose Williams syndrome. Elastin is a protein found in connective tissue which is important for connecting organs and tissues, and for storing nutrients. The loss of elastin is associated with connective tissue abnormalities and cardiovascular (heart) disease.