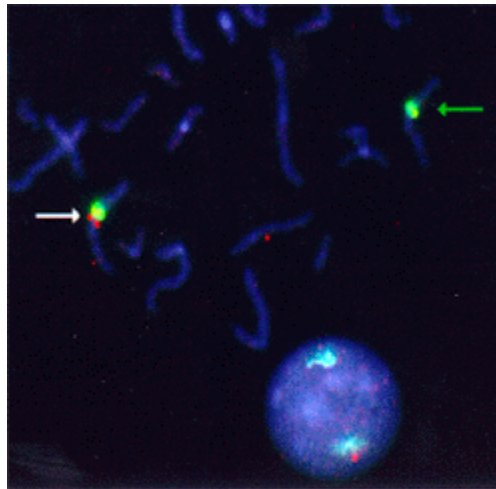




Williams syndrome



Williams syndrome is caused by a deletion of part of chromosome 7 that includes the LIM kinase and elastin coding sequences. Above, this sequence (stained red) can no longer be seen in the chromosome with the deletion (green arrow). [Photograph kindly provided by L. G. Shaffer, Baylor College of Medicine.]

Williams syndrome is a rare congenital disorder characterized by physical and development problems. Common features include characteristic "elfin-like" facial features, heart and blood vessel problems, irritability during infancy, dental and kidney abnormalities, hyperacusis (sensitive hearing) and musculoskeletal problems. Although individuals with Williams syndrome may show competence in areas such as language, music and interpersonal relations, their IQs are usually low.

In Williams syndrome individuals, both the gene for elastin and an enzyme called LIM kinase are deleted. Both genes map to the same small area on chromosome 7. In normal cells, elastin is a key component of connective tissue, conferring its elastic properties. Mutation or deletion of elastin lead to the vascular disease observed in Williams syndrome. On the other hand, LIM kinase is strongly expressed in the brain, and deletion of LIM kinase is thought to account for the impaired visuospatial constructive cognition in Williams syndrome.

Williams syndrome is a contiguous disease, meaning that the deletion of this section of chromosome 7 may involve several more genes. Further study will be required to round up all the genes deleted in this disease. The remarkable musical and verbal abilities of individuals with Williams syndrome, and their tendency to be very sociable, has led to the suggestion that children with Williams syndrome were an inspiration for folktales and legends, as the 'wee, magical people' were often musicians and storytellers.

Related diseases

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