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ABOUT US

Chromosome Disorder Outreach provides support and information to anyone diagnosed with a rare chromosome change, rearrangement or disorder. CDO actively promotes research and a positive community understanding of all chromosome disorders.

CDO is a 501c3 organization founded in 1992.

7q11.23
Williams Syndrome

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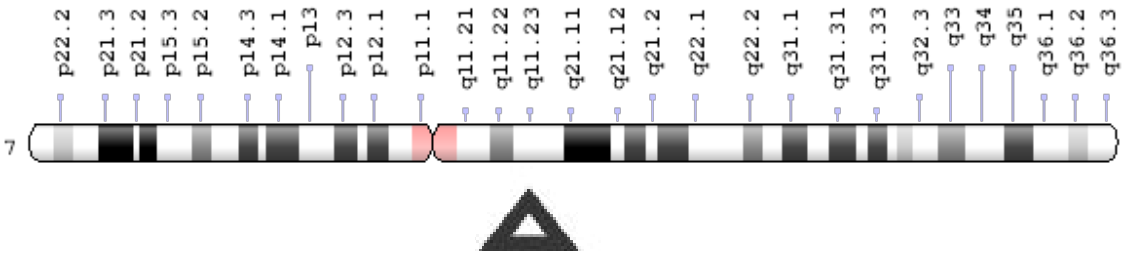
Williams Syndrome

The majority of syndromes caused by chromosomal deletions became known only after the discovery of chromosomal changes. Williams syndrome (WS) is a rare exception: it was described as an entity in 1961, although its chromosomal etiology was reported only in 1990s[1,2]. The main characteristics of WS are typical facial features, congenital heart defects, and a specific association of personality characteristics and intellectual disability. WS is a relatively frequent condition. It occurs approximately in 1 out of 10,000 newborns. Typical facial manifestations of WS include puffiness around the eyes, short upturned nose, full cheeks, wide mouth and elongated philtrum.



These characteristics allow recognition of typical WS patients in early childhood at a glance. Some authors have described these features as “elfin face”, and several reports on “elfin face” syndrome may be found in the literature even before 1961. Other very important traits are congenital heart defects found in ~90% of patients[3]. The most common abnormality is supravalvular aortic stenosis (narrowing of the aorta just above the aortic valve).

In many patients this heart defect has to be treated by surgery[3,4]. Stenosis of the pulmonary artery is the second most common type of heart defects in WS patients. Other forms of defects include abnormalities of coronary and other arteries. At the same time septal defects are relatively rate (~10% of all heart defects in WS patients).

The third main finding is intellectual disability which has very specific characteristics. The mean IQ for WS patients is ~70, but some may show normal IQ. The main problems include limited abstract reasoning and limited spatial orientation. As a result patients may have problems assembling puzzles. At the same time they usually are highly verbal (with an unusually wide lexicon) and overly sociable[2,5,6]. Many WS patients have absolute pitch and reveal love of music despite hyperacusis – an increased sensitivity to certain frequencies of sound.

In their younger age many WS patients have hypercalcemia[7], which causes colic-like symptoms in small babies. Usually hypercalcemia disappears without any specific treatment. Many patients may develop inguinal hernias. Severe morphological defects of the brain, kidney and gastro-intestinal tract are very rare. The main cause of death is cardiac insufficiency due to congenital heart defects. For patients without heart defects or for those who have had successful treatment of these defects, life expectancy is approximately normal.

The genetic basis of the syndrome is a deletion in the segment 7q11.23[2]. Although this deletion is relatively small (usually 1.5-1.7 Mb) it includes more than 25 different genes, including the elastin gene (ELN). The ELN gene is considered the main causal factor of defects in connective tissue and arterial abnormalities, common in WS patients. Other genes are involved in facial features and specific intellectual problems. Of course, deletions of this size may be recognized only by molecular testing or by using probes specific to the ELN gene. Patients with atypically large deletions, involving other genes may have some additional abnormalities.

Most deletions are sporadic, and in that case the risk of a recurrent birth of another WS child is close to zero. If WS patients have their own children the risk of the affected child will be 50%. There are several reports where mild manifestations of WS in one of the parents were noticed only after the birth of their child with typical WS manifestations. Therefore cytogenetic examination of parents is reasonable at least in families where WS in one parent may be suspected.There is no cure for WS. The patients with heart abnormalities may need surgical correction of the defect. Specific treatment for hypercalcemia may be also necessary, at least for some patients[7]. Older children and adult patients may need treatment by occupational therapists, psychologists and psychiatrists.