Williams syndrome is a rare neurodevelopmental disorder characterized by: a distinctive facial appearance, along with a low nasal bridge; an unusually cheerful demeanor and ease with strangers; developmental delay coupled with strong language skills; and cardiovascular problems, such as supravalvular aortic stenosis and transient hypercalcemia.

It is caused by a deletion of about 26 genes from the long arm of chromosome 71. The syndrome has an estimated prevalence of 1 in 10,000 births2. Williams syndrome is caused by the spontaneous deletion of genetic material from the region q11.23 of chromosome 7. The deleted region includes more than 25 genes, and the loss of these several genes probably contributes to the characteristic features of this disorder. CLIP2, ELN, GTF2I, GTF2IRD1, and LIMK1 are among the genes that are typically deleted in people with Williams syndrome. Researchers have found that loss of the ELN gene, which codes for the protein elastin, is associated with the connective-tissue abnormalities and cardiovascular disease (specifically supravalvular aortic stenosis and supravalvular pulmonary stenosis) found in many people with this syndrome.

Despite their physical and cognitive deficits, individuals with Williams syndrome exhibit impressive social and verbal abilities. Williams patients can be highly verbal relative to their IQ. Some other strengths that have been associated with Williams syndrome are auditory short-term memory and facial recognition skills. The language used by individuals with Williams syndrome differs notably from unaffected populations, including individuals matched for IQ. One of deleted genes, GTF2I, has been associated with hypersociability3.

In this issue of the journal Chin-See-Chong et al describe a candidate gene study, in which they assessed a possible correlation between two polymorphisms of the GTF2I gene and positive emotionality in 3-year old children (n=862) , using data from the Generation R study4. Positive emotionality was measured by two observational tests (the Puppet Game and the Popping Bubbles). The authors found a trend to positive emotionality and one of the SNPs of the GTF2I gene. However, as the authors state, more research is warranted to draw conclusions about the possible genetic contribution to positive emotionality. They suggest that studying expression levels of GFT2I between cases and controls may be a promising approach.

References

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The dissection of Williams syndrome: searching for the gene coding for positive emotionality

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