SHOX Deficiency

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WHAT IS SHOX DEFICIENCY

SHOX deficiency is a frequent cause of short stature. Human genes are made up of DNA and every person has two copies of each gene, one inherited from each parent. Genes act as instructions to make molecules called proteins and each protein has a specific function in the body. SHOX is a gene (short stature homeobox-containing gene), which plays an important role in bone growth and development. SHOX deficiency as a result of SHOX gene abnormalities causes poor growth in humans, usually in the first few years of life.

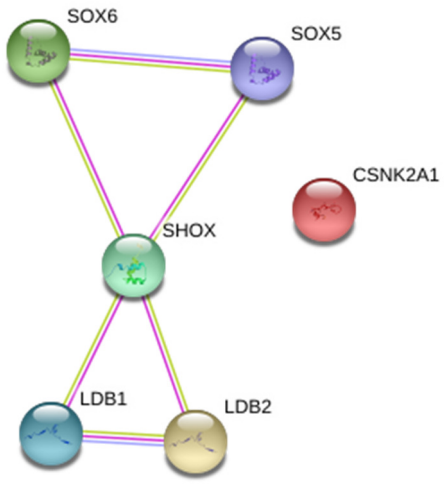
The SHOX genes are located on the sex chromosomes (the X and Y chromosomes) in humans. This gene was first found during a search for the cause of short stature in women with Turner syndrome, in which there is loss of genetic material from the X chromosome (classically loss of one entire X chromosome; 46XO). Since its discovery, the SHOX gene has been found to play a role in other forms of short stature, not related to Turner syndrome.

SHOX deficiency may be more severe and is more common in females.

Changes (mutations) or loss (deletions) of one of the SHOX genes causes growth failure. Mutations in SHOX have been identified in approximately 2-15% patients with unexplained short stature (idiopathic short stature or ISS). Changes in the SHOX gene

can also cause a type of skeletal dysplasia called Leri-Weill dyschondrosteosis. This a rare genetic condition which results in short stature and shortening of the bones of the arms and legs. The loss of both SHOX genes (complete lack of SHOX), is very rare and causes a very severe skeletal abnormality known as Langer syndrome. This results in extreme short stature and very short, underdeveloped bones.

In contrast, extra copies of SHOX due to abnormal numbers or structure of the sex chromosomes may cause tall stature.



DIAGNOSIS

A diagnosis of SHOX Deficiency may be suspected if a child has evidence of growth failure or short stature with no obvious cause. Some children with SHOX deficiency have shortening of their arms and legs but this may not be present in young children and may never be present in some people. X-rays e.g. of the hand and wrist might pick up particular bone changes associated with this condition and these may be requested as part of the initial investigations. Genetic analysis confirms the diagnosis.

TREATMENT

In the UK, SHOX deficiency is a licenced indication for synthetic growth hormone (GH), a man-made form of GH. GH is given by daily injection just under the skin (subcutaneous) and has been shown to improve the final height of SHOX deficient patients. SHOX patients are usually prescribed a similar GH dose to that given to patients with Turner syndrome and the effects of treatment are thought to be equivalent. In order to fulfil the criteria for GH treatment in the UK, children must have SHOX deficiency confirmed by DNA (genetic) analysis.

ACKNOWLEDGEMENTS

Many thanks to:

Dr Helen Storr

FURTHER INFORMATION

If you have any questions regarding the information contained in this sheet, then please contact:

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FUNDING

The Foundation funds research into many aspects of growth conditions such as the causes, effects, treatments and psychological impact. It also offers essential advice and experience to parents of children who have been diagnosed with growth problems. The annual convention provides a great forum for people to get together to discuss problems and solutions with others in a similar position. It also provides a chance to meet and learn from the doctors and professors dealing with child growth in the UK.

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