GENETICS & Treacher Collins Syndrome

Treacher Collins Syndrome (TCS) is Autosomal Dominant. Autosomal means that males and females are equally affected. In dominant inheritance the chance of passing on the disorder is 50%. If the gene is inherited it will result in an affected individual.

The features of TCS are, however, variable so that whilst some people are severely affected others are so mildly affected that it is very difficult to say whether they have the condition or not.

All human genetic information is carried on structures called chromosomes, of which there are 46 in most cells of the body. Of these 46 chromosomes, two are involved in determining the baby's sex whilst the remainder are made up of 22 pairs.

Each chromosome contains many genes, each one having the ability to produce a certain characteristic. Under normal circumstances each of a pair of chromosomes looks alike and at the same position on each chromosome are genes determining the same characteristic.

Usually these genes are identical although in some cases (for instance in TCS) one gene is different from its opposite number and cannot produce its normal characteristic - the genetic information is effectively broken.

If either parent has TCS there is a 50/50 chance that each of their children may be affected. If the TCS (broken) gene is passed from parent to child the child will be affected.

In approximately 60 per cent of cases, however, there is no previous family history of the condition - neither parent has it. In this case the child has developed TCS as a result of genetic mutation, not through hereditary causes.

Although the broken gene has now been found there is still a great deal of research to be done to gain a much better understanding of the mechanisms underlying TCS. Moreover, this may also lead to a better idea of the mechanisms responsible for other forms of conductive deafness.

Identification of the gene should also ultimately aid early diagnosis of TCS, which will be particularly useful for those people with very mild TCS and in whom clinical diagnosis is correspondingly difficult.

As a result of the earlier discovery of the chromosome upon which the gene responsible for causing TCS lies upon it is now possible to predict who is and who is not affected by TCS with a very high degree of accuracy (greater than 95%). Unfortunately a test such as this will not be applicable to all families until a later point in the ongoing research.

The families that would be potentially suitable for testing are those families in which there is a clearly defined history of TCS and not those where TCS has arisen without a previous family history.

At present there is no reliable prenatal testing for TCS. Specialist ultrasound can sometimes give an indication that a baby may be affected but cannot indicate the severity.

X-rays can sometimes help to determine whether a person has TCS.

Any genetic testing/consultation should be carried out by specially qualified geneticists who have experience of TCS.

For more information about the TCS research project contact:

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