Tricho-rhino-phalangeal syndrome

TRPS1, TRPS2, TRPS3, Langer-Giedion syndrome, LGS

Background

Tricho-rhino-phalangeal syndrome is a rare genetic condition affecting the hair (tricho), nose (rhino), and fingers (phalangeal). The term is often abbreviated to TRPS. There are two main types: Type 1 (or TRPS1) and Type 2 (or TRPS2, sometimes also called Langer-Giedion syndrome, named after the two radiologists who described the first patients). Both conditions cause slow growth with a short stature as an adult, sparse and slow-growing hair, an unusually formed nose of which the tip is more prominent than usual, and short fingers and toes. Type 3 is a term used by some people to describe a more severe form of TRPS Type 1.

What are the causes?

TRPS is a genetic condition, meaning it is caused by an alteration to a person's genetic makeup. This person may be the first in the family with the disease, or may have inherited it from a parent. Sometimes the syndrome can be traced back through several generations. In some people, the genetic alteration may be a change in the genetic code of a gene called TRPS1 which is found on chromosome 8. In others, the whole gene may be missing. In people with TRPS type 2 (Langer-Giedion syndrome), a small piece of chromosome 8 is usually missing, which means that not only is the TRPS1 gene missing, but also another genes, and this can cause additional problems. Commonly, a gene called EXT1 is missing, which causes benign bony growths called exostoses to develop.

What are the symptoms?

The most common problems are short stature, sparse hair, and restricted joint movements. There are often other problems with the bones and joints, which may appear as congenital hip problems, Perthes disease of the hip, and early onset osteo-arthritis of other joints. Almost any joint can be affected. People with TRPS type 2 can also develop multiple exostoses, which are benign bony tumours (or lumps) that grow on the bones. Although not painful themselves, these can press on surrounding muscles or ligaments, causing limitations of movement, discomfort or pain. Some people with TRPS also suffer from learning difficulties and this is more common in TRPS type 2. People with TRPS often comment that they look similar to other people with TRPS that they have met. Orthodontic problems, deafness and susceptibility to infections can also occur.

How is it diagnosed?

The diagnosis is usually made on the findings at clinical examination, and the shape of the bones (cone-shaped epiphyses) when seen on a hand X-ray. The diagnosis can in some cases be confirmed by checking the patient's chromosomes for a deletion on chromosome 8q, or by molecular genetic analysis of the TRPS1 gene.

How is it treated?

There is currently no treatment that will cure the whole syndrome. Individual problems are managed as needed. In childhood, growth and development should be monitored.

Simple analgesia can relieve joint pains. A reduction in mobility should be investigated as Perthes disease of the hip can frequently occur. Multiple exostoses can develop in TRPS2, typically from early-mid childhood. Adults frequently require joint replacement surgery in the fifth and sixth decades due to degenerative changes in the large joints. Sparse hair and premature hair loss can result in reduced self-esteem. Hair extensions may help some, but the exceptionally brittle hair seen in some patients may not support them.

Inheritance patterns and prenatal diagnosis

Inheritance patterns

Autosomal dominant with many cases occurring as a sporadic new mutation.

Prenatal diagnosis

Chorionic villus sampling (CVS) or amniocentesis may be available for a family if the diagnosis has been confirmed on chromosome or DNA analysis. Referral to a regional genetics centre for genetic counselling is indicated.

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Support

TRPS Support Group UK

c/o Contact a Family, 209-211 City Road, London, EC1V 1JN
Tel: 0808 808 3555 (group number available for those without internet access)
e-mail: info@trpsuk.org
Web: http://www.trpsuk.org
TRPS Support Group UK formed in 2009 and has been developing its services over the last couple of years. It currently offers an opportunity for those affected by this condition in the UK to meet others to share experiences and knowledge and has been instrumental in the development of a UK clinic for this condition. Contact the group for further details of the clinics. The group has close links with an international forum which is based in American.

Trichorhinophalangeal Syndrome Association

The US based support group, Trichorhinophalangeal Syndrome Association, offers an e-mail group at http://health.groups.yahoo.com/group/TRPSA (information about the use of online groups is available in our 'Medical Information on the Internet: Seeking Quality' article). This service is moderated. The Association is also in the process of developing and extending the support which they provide to include an annual family meeting, newsletter and website. If you do not have internet access please telephone our free phone helpline, Tel: 0808 808 3555.
Group details last updated February 2009.

