

Trichorhinophalangeal Syndrome Type I

Subjects: Genetics & Heredity

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Trichorhinophalangeal syndrome type I (TRPS I) is a condition that causes bone and joint malformations; distinctive facial features; and abnormalities of the skin, hair, teeth, sweat glands, and nails.

Keywords: genetic conditions

1. Introduction

Trichorhinophalangeal syndrome type I (TRPS I) is a condition that causes bone and joint malformations; distinctive facial features; and abnormalities of the skin, hair, teeth, sweat glands, and nails. The name of the condition describes some of the areas of the body that are commonly affected: hair (tricho-), nose (rhino-), and fingers and toes (phalangeal).

In people with TRPS I, the ends (epiphyses) of one or more bones in the fingers or toes are abnormally cone-shaped. Additionally, the fingernails and toenails are typically thin and abnormally formed. Affected individuals often have short feet.

Individuals with TRPS I may have a misalignment of the hip joints (hip dysplasia), which often develops in early adulthood but can occur in infancy or childhood. Children with TRPS I often have an unusually large range of movement (hypermobility) in many of their joints. Over time, however, the joints may break down (degenerate), leading to joint pain and a limited range of joint movement.

The characteristic appearance of individuals with TRPS I involves thick eyebrows; a broad nose with a rounded tip; large ears, a long, smooth area between the nose and the upper lip (philtrum); a thin upper lip; and small teeth that are either decreased (oligodontia) or increased (supernumerary) in number. Almost all affected individuals have sparse scalp hair. Males are particularly affected by hair loss with many being nearly or completely bald soon after puberty. Some children with this condition have loose skin, but the skin becomes tighter over time. Individuals with TRPS I may experience excessive sweating (hyperhidrosis).

2. Frequency

TRPS I is a rare condition; its prevalence is unknown. In the Netherlands, at least 35 people have TRPS I.

3. Causes

TRPS I is caused by mutations in the *TRPS1* gene. This gene provides instructions for making a protein that is found within the cell nucleus where it interacts with specific regions of DNA to turn off (repress) the activity of certain genes. Research suggests that the TRPS1 protein plays a role in regulating genes that control the growth of bone and other skeletal tissues.

TRPS1 gene mutations lead to the production of an altered TRPS1 protein. The altered protein has a reduced ability to control the activity of genes that regulate the growth of bone and other tissues, leading to abnormal bones in the fingers and toes, joint abnormalities, distinctive facial features, and other signs and symptoms of TRPS I.

A condition similar to TRPS I is caused by the loss of the *TRPS1* gene and its neighboring genes. This condition, called trichorhinophalangeal syndrome type II (TRPS II), has many of the same signs and symptoms of TRPS I, as well as multiple benign (noncancerous) bone tumors called osteochondromas and intellectual disability. These additional features are associated with the loss of genes near *TRPS1*.

3.1 The gene associated with Trichorhinophalangeal syndrome type I

- TRPS1
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4. Inheritance

TRPS I is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

In most cases, an affected person inherits the mutation from one affected parent. Some cases result from new mutations in the gene and occur in people with no history of the disorder in their family.

5. Other Names for This Condition

- trichorhinophalangeal dysplasia type I
- TRP syndrome
- TRPS I
- TRPS1

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