

TRICHORHINOPHALANGEAL SYNDROME (TRP)

TRP is a type of Ectodermal Dysplasia identified by problems with the hair, teeth and nails. There are several types of trichorhinophalangeal syndromes, TRP syndrome Type I is characterised primarily by abnormalities of the bones and hair. Some of the finger joints may be enlarged; the thumbs and big toes are usually short; other fingers and toes may also be short. Scalp hair is often fine, sparse and brittle, and is abnormal from birth. Scalp hair is especially scant in the front, simulating the male baldness pattern and some individuals may become completely bald.

Eyebrows are thick near the nose, but sparse on their lateral aspects. In addition, the tip of the nose is bulbous, the midportion of the upper lip is long and the upper lip is thin. Intraorally, extra teeth have been found. Nails may be thin. About one half of affected individuals are shorter than normal. Intelligence is normal as is the ability to perspire. In most families with the disorder, the inheritance pattern is autosomal dominant. However, a few families have been reported in which the condition may be inherited as an autosomal recessive. It is not possible to differentiate the dominant and recessive forms from one another on the basis of clinical or x-ray examination. So the family history and examination of both parents are important in order to determine the pattern of inheritance.

TRP syndrome Type II is also called the Langer-Giedion syndrome, named after the first two people who described the condition fully. Most patients with the condition do not have affected relatives. However, father to daughter transmission has been documented. The facial appearance and hair abnormalities are similar to those found in TRP syndrome Type I. Mild to moderate mental retardation has been present in most affected individuals, although some individuals may not be mentally retarded. Delayed onset of speech and, less frequently, hearing deficit have been reported, although the tones affected, age of onset and the degree of severity of this hearing loss have not been well documented. One of the prime features which differentiates this syndrome from TRP Type I is multiple bony bumps, called exostoses. They are usually present by the third or fourth year of life, although they may be found as early as the end of the first year. They are located primarily near the ends of the bones of the arms and legs, although other bones may also be affected.

Another feature which distinguishes between the TRP syndromes is the finding that in some patients with TRP syndrome Type II an abnormality of the chromosomes have been found; this abnormality involves chromosome 8. Specifically, a small portion of this chromosome is missing.

TRP syndrome Type III is like the others, but sufferers also have very short fingers and toes.

Supporting a normal lifestyle