Trichorhinophalangeal syndrome type I (TRPS1) is an extremely rare inherited multisystem disorder. TRPS1 is characterized by thin, sparse scalp hair, unusual facial features, abnormalities of the fingers and/or toes, and multiple abnormalities of the "growing ends" (epiphyses) of the bones (skeletal dysplasia), especially in the hands and feet. Characteristic facial features may include a rounded (bulbous) "pear-shaped" nose, an abnormally small jaw (micrognathia), dental anomalies, and/or unusually large (prominent) ears. In most cases, the fingers and/or toes may be abnormally short (brachydactyly) and curved. In addition, affected individuals may exhibit short stature. The range and severity of symptoms may vary from case to case. In most cases, Trichorhinophalangeal syndrome type I has autosomal dominant inheritance. Trichorhinophalangeal syndrome type I is an extremely rare inherited disorder that affects males and females in equal numbers. In those individuals with mild symptoms, a diagnosis may be easily missed or go unreported. Therefore, it is difficult to determine the true frequency of this disorder in the general population. Many researchers suspect there may be a higher incidence of TRPS1 than is actually reported in the medical literature. The diagnosis of TRPS1 may be suspected upon identification of characteristic physical features (e.g., rounded [bulbous] nose; thin, sparse hair; etc.). The diagnosis may be confirmed by a thorough clinical evaluation, a detailed patient history, and X-ray studies of the skeleton that reveal distinctive abnormalities of the hands and feet (e.g., epiphyseal coning). Molecular genetic testing can reveal mutations of the TRPS1 gene.