

Trichorhinophalangeal syndrome type III (TRPS3), also known as Sugio-Kajii syndrome, is an extremely rare inherited multisystem disorder. TRPS3 is characterized by fine, thin light-colored 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 pear-shaped or rounded (bulbous) nose; an abnormally long prominent groove (philtrum) in the upper lip; and/or abnormalities such as delayed eruption of teeth. In addition, affected individuals also exhibit severe shortening of the fingers and toes (brachydactyly) due to improper development of bones in the hands and feet (metacarpophalangeal shortening). Additional features often include short stature (dwarfism) and/or additional skeletal abnormalities. The range and severity of symptoms may vary from case to case. TRPS3 is thought to have autosomal dominant inheritance. Trichorhinophalangeal syndrome type III is an extremely rare disorder that, in theory, affects males and females in equal numbers. However, of the reported cases, most affected individuals have been female. Approximately 15 cases have been reported in the medical literature. A diagnosis of trichorhinophalangeal syndrome type III may be suspected based upon identification of characteristic physical features (e.g., fine, sparse hair; facial abnormalities; etc.). The diagnosis may be confirmed by a thorough clinical evaluation, a detailed patient history, and X-ray studies of the skeleton that reveal severe shortening of bones in the hands and feet (i.e., metacarpals, metatarsals, and phalanges) and the abnormal development of the "growing ends" (epiphyses) of the phalangeal bones (epiphyseal coning).