

Trichorhinophalangeal syndrome type II (TRPS2), also known as Langer-Giedion syndrome, is an extremely rare inherited multisystem disorder. TRPS2 is characterized by fine, thin hair; unusual facial features; progressive growth retardation resulting in short stature (dwarfism); abnormally short fingers and toes (brachydactyly); "cone-shaped" formation of the "growing ends" of certain bones (epiphyseal coning); and/or development of multiple bony growths (exostoses) projecting outward from the surfaces of various bones of the body. In addition, affected individuals may exhibit unusually flexible (hyperextensible) joints, diminished muscle tone (hypotonia), excess folds of skin (redundant skin), and/or discolored elevated spots on the skin (maculopapular nevi). Affected individuals may also exhibit mild to severe mental retardation, hearing loss (sensorineural deafness), and/or delayed speech development. The range and severity of symptoms varies greatly from case to case. TRPS2 is due to the absence of genetic material (chromosomal deletions) on chromosome 8. The size of the deletion varies from case to case.