

Ruvalcaba syndrome is a rare inherited disorder characterized by short stature, abnormalities affecting the head and facial (craniofacial) area, mental retardation, skeletal malformations, and/or underdeveloped (hypoplastic) genitalia. Characteristic craniofacial features include an abnormally small head (microcephaly); an abnormally small, narrow nose; and down-slanting eyelid folds (palpebral fissures). Skeletal malformations may include fifth fingers that are permanently fixed in a bent position (clinodactyly) and/or abnormally short bones between the wrists and the fingers (metacarpals) and the ankles and toes (metatarsals), resulting in unusually small hands and feet. In addition, affected children may have abnormal side-to-side curvature of the spine (scoliosis) and/or unusual prominence of the breastbone (pectus carinatum). Ruvalcaba syndrome is inherited as an autosomal dominant genetic trait.

Ruvalcaba syndrome is a rare disorder that, in theory, affects males and females in equal numbers. Probably fewer than a dozen confirmed cases have been reported in the medical literature. Several cases of what were initially diagnosed as Ruvalcaba syndrome proved to be cases of two related disorders — trichorhinophalangeal syndrome type III and Hunter-McAlpine craniosynostosis syndrome. (For more information on these disorders, see the Related Disorders section below.) It is highly likely that some cases of Ruvalcaba syndrome may go unrecognized and remain undiagnosed, making it difficult to determine its true frequency in the general population. The diagnosis of Ruvalcaba syndrome may be suspected based upon identification of characteristic physical features (e.g., microcephaly, characteristic facial abnormalities, skeletal malformations, etc.). The diagnosis may be confirmed by a thorough clinical evaluation, a detailed patient history, and x-ray studies of the skeleton that reveal shortening of bones in the hands and feet (i.e., metacarpals, metatarsals, and phalanges).