

Coffin-Lowry syndrome is a rare genetic disorder characterized by mental retardation; abnormalities of the head and facial (craniofacial) area; large, soft hands with short, thin (tapered) fingers; short stature; and/or various skeletal abnormalities. Characteristic facial features may include an underdeveloped upper jawbone (maxillary hypoplasia), an abnormally prominent brow, downslanting eyelid folds (palpebral fissures), widely spaced eyes (hypertelorism), large ears, and/or unusually thick eyebrows. Skeletal abnormalities may include abnormal front-to-back and side-to-side curvature of the spine (kyphoscoliosis) and unusual prominence of the breastbone (sternum) (pectus carinatum). Coffin-Lowry syndrome is caused by mutations in the RSK2 gene and is inherited as an X-linked dominant genetic trait. Males are usually more severely affected than females. Coffin-Lowry syndrome affects as many males as females. However, symptoms may be more severe in males.