

Cleidocranial dysplasia is a rare skeletal dysplasia characterized by short stature, distinctive facial features and narrow, sloping shoulders caused by defective or absent collarbones (clavicles). Major symptoms may include premature closing of the soft spot on the head (coronal), delayed closure of the space between the bones of the skull (fontanel), narrow and abnormally shaped pelvic and pubic bones and deformations in the chest (thoracic region). Delayed eruption of teeth, moderately short stature, a high arched palate, a wide pelvic joint, failure of the lower jaw joints to unite, and fingers that are irregular in length may also be present. Cleidocranial dysplasia is inherited as an autosomal dominant genetic trait. Cleidocranial dysplasia is a very rare disorder that is apparent at birth and affects males and females in equal numbers. Approximately 1,000 cases of this disorder have been reported in the medical literature. The birth prevalence is approximately 1 in 1 million.