

Central core disease (CCD) is a rare genetic neuromuscular disorder that is classified as a congenital myopathy, meaning that it is a muscle disorder (myopathy) that is present at birth (congenital). Affected infants have diminished muscle tone (hypotonia) resulting in abnormal "floppiness", muscle weakness, and a variety of skeletal abnormalities such as side-to-side curvature of the spine (scoliosis). Muscle weakness normally affects the proximal muscles, which are those muscles closest to the center of the body such as the muscles of the shoulder, pelvis and upper arms and legs. Affected infants may experience delays in acquiring motor milestones such as crawling or walking. Some individuals with CCD may be susceptible to developing malignant hyperthermia, a condition in which individuals develop adverse reactions to certain anesthetic drugs. CCD may be very mild or may cause serious complications. Most cases are inherited as autosomal dominant trait and associated with nonprogressive muscle disease and a favorable prognosis. Some cases are inherited as autosomal recessive traits and are more likely to be associated with severe complications.