

Congenital fiber type disproportion (CFTD) is a rare genetic muscle disease that is usually apparent at birth (congenital myopathy). It belongs to a group of muscle conditions called the congenital myopathies that tend to affect people in a similar pattern. Major symptoms may include loss of muscle tone (hypotonia) and generalized muscle weakness. Delays in motor development are common and people with more marked muscle weakness also have abnormal side-to-side curvature of the spine (scoliosis), dislocated hips, and the permanent fixation of certain joints in a flexed position (contractures), particularly at the ankle. CFTD affects males and females in equal numbers. The incidence of the disorder in the general population is unknown but it is uncommon. The disorder is usually present at birth (congenital) but may not be recognized for many months. Case reports describing children with the features of CFTD first appeared in the medical literature in the 1960s and 70s. The term congenital fiber type disproportion was first used in 1973.