Congenital Fiber-Type Disproportion

National Cancer Institute

Source

National Cancer Institute. Congenital Fiber-Type Disproportion. NCI Thesaurus. Code C120046.

A rare genetic disorder caused by mutations in the TPM3, ACTA1, RYR1 and SEPN1 genes. It is inherited in an autosomal dominant or recessive pattern and rarely in an X-linked pattern. It manifests with myopathy throughout the body, particularly in the muscles of the shoulders, upper arms, hips, and thighs. Affected individuals may have contractures, lordosis, or scoliosis. In a minority of cases mild to severe breathing problems may occur.