

Myotonia congenita is a rare genetic disorder in which an abnormality of voluntary (skeletal) muscle fiber membranes causes an unusually exaggerated response to stimulation (hyperexcitability). As a result, affected individuals have difficulty relaxing certain muscles after contracting them (myotonia), muscle stiffness (rigidity), and associated symptoms. Such symptoms tend to occur when attempting to move certain muscles after rest. In many cases, individuals with myotonia congenita also have abnormal enlargement of the muscles (hypertrophy), resulting in a "herculean" or "body-builder like" appearance.