

Symptoms of Wieacker syndrome include stiffening of the muscles and joints of the feet (contracture), slowly progressive atrophy of certain muscles of the legs and arms, and mild intellectual disability. Other symptoms are impairment or inability to move the eyes despite the wish to do so, and impairment in the use of face and tongue muscles. In some cases, affected individuals may have droopy eyelids (ptosis), crossed eyes (strabismus), and farsightedness (hyperopia). Abnormal curvature of the spine may also occur (kyphoscoliosis). The prevalence of Wieacker syndrome is not known. Wieacker syndrome usually affects males, but some carrier females show mild manifestations of the disorder. As of 2015, the syndrome has been reported in 5 families. ZC4H2 gene mutations have also been identified in 3 sporadic patients who presented more or less severe intellectual disability and congenital contractures of multiple joints, at least equinovarus of the feet, associated with neurological symptoms such as muscle weakness, spasticity, seizures and ptosis. In one of the families, those affected had neonatal respiratory distress responsible for early death. Wieacker syndrome is therefore now considered to be part of a wider phenotype consisting of arthrogryposis multiplex congenita (congenital multiple joint contractures) and intellectual disability, caused by ZC4H2 gene mutations. In some instances in the history of the family in which the syndrome was first described, the syndrome was present at birth.