

The exact cause of megalocornea intellectual disability syndrome is unknown. Megalocornea, when occurring as an isolated finding, is usually inherited as an X-linked recessive trait. Recessive genetic disorders occur when an individual inherits the same abnormal gene for the same trait from each parent. If an individual receives one normal gene and one gene for the disease, the person will be a carrier for the disease, but usually will not show symptoms. The risk for two carrier parents to both pass the defective gene and, therefore, have an affected child is 25 percent with each pregnancy. The risk to have a child who is a carrier like the parents is 50 percent with each pregnancy. The chance for a child to receive normal genes from both parents and be genetically normal for that particular trait is 25 percent. The risk is the same for males and females. Approximately 37 individuals with megalocornea intellectual disability syndrome have been reported in the medical literature. More males have been reported than females. The disorder is present at birth (congenital). The syndrome was first described in the medical literature in 1975.