

Aniridia, cerebellar ataxia, and mental deficiency, also known as Gillespie syndrome, is an extremely rare inherited disorder that is characterized by the absence, in whole (aniridia) or in part (partial aniridia), of the colored portion (iris) of the eye; impaired coordination of voluntary movements due to underdevelopment (hypoplasia) of the brain's cerebellum (cerebellar ataxia); and mental retardation. The condition usually affects both eyes (bilateral) but a few cases have been reported in which only one eye is affected. Some individuals with this syndrome also exhibit a delay in the acquisition of skills requiring coordination of muscular and mental activity (psychomotor retardation). ACAMD is thought to be inherited as an autosomal recessive genetic trait and is extremely rare, with only 20 to 30 cases reported in the medical literature. Aniridia, cerebellar ataxia and mental deficiency is an extremely rare inherited disorder that appears to affect females more than males, although the sample size is very low. Only about 20 or 30 cases have been reported in the medical literature. One report suggests that people with this syndrome make up about 2% of all patients with aniridia.