# NAA10-related disorder Arg83Cys

*https://pubmed.ncbi.nlm.nih.gov/31093388/*

The

NAA10

-related syndrome is a rare X-linked neurodevelopmental condition that was first described in 2011. The disorder is caused by pathogenic variants in the

NAA10

gene located on chromosome X at position Xq28. Clinical features typically include severe psychomotor developmental delay, cardiac disease, dysmorphic features, postnatal growth failure, and hypotonia, although there is significant variability in the severity of the phenotype among affected individuals. We describe a 5-year-old female with the syndrome; massively parallel exome sequencing and analysis revealed the c.247C>T (p.Arg83Cys) pathogenic variant that has been previously reported in ten affected individuals. Ocular manifestations of the

NAA10

-related syndrome are not uncommon, although they have not been well characterized in literature reports. From a systematic review of previously published cases to date, ocular abnormalities are present in more than half of patients with the syndrome. Common ocular findings reported include astigmatism, hyperopia, cortical vision impairment, microphthalmia/anophthalmia, and hypertelorism. Our patient presented with growth restriction, dysmorphic features, and hypotonia. Ocular manifestations identified in this child include downslanting palpebral fissures, myopic astigmatism, nystagmus, and exotropia. We speculate that the type and severity of ocular defects present in individuals with the

NAA10

-related syndrome are dependent on the specific

NAA10

pathogenic variant involved.