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# GJA1 wt Allele

National Cancer Institute

## Source

National Cancer Institute. *GJA1 wt Allele*. NCI Thesaurus. Code C115171.

Human GJA1 wild-type allele is located in the vicinity of 6q22.31 and is approximately 14 kb in length. This allele, which encodes gap junction alpha-1 protein, plays a role in the modulation of the activity of gap junctions. Mutation of the gene is associated with atrioventricular septal defect 3, autosomal recessive craniometaphyseal dysplasia, hypoplastic left heart syndrome 1, oculodentodigital dysplasia and syndactyly, type III.