

Open Peer Review on Qeios

EYA1 wt Allele

National Cancer Institute

Source

National Cancer Institute. <u>EYA1 wt Allele</u>. NCI Thesaurus. Code C75391.

Human EYA1 wild-type allele is located in the vicinity of 8q13.3 and is approximately 165 kb in length. This allele, which encodes eyes absent homolog 1 protein, may be involved in the development of the eyes, ears, branchial arches and kidney. Mutations of this gene are associated with branchiootorenal dysplasia syndrome, branchiootic syndrome, congenital cataracts and ocular anterior segment anomalies.

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