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# OTX2 wt Allele

National Cancer Institute

## Source

National Cancer Institute. *OTX2 wt Allele*. NCI Thesaurus. Code C102952.

Human OTX2 wild-type allele is located in the vicinity of 14q22.3 and is approximately 10 kb in length. This allele, which encodes homeobox protein OTX2, plays a role in the positive regulation of gene expression during embryonic pattern formation. Mutation of the gene is associated with both microphthalmia syndromic type 5 and pituitary hormone deficiency combined type 6.