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# NR0B1 wt Allele

National Cancer Institute

## Source

National Cancer Institute. *NR0B1 wt Allele*. NCI Thesaurus. Code C75865.

Human NR0B1 wild-type allele is located within Xp21.3-p21.2 and is approximately 5 kb in length. This allele, which encodes nuclear receptor subfamily 0 group B member 1 protein, is involved in the regulation of both transcription and receptor-mediated signal transduction. Mutations in the gene are associated with both X-linked congenital adrenal hypoplasia and hypogonadotropic hypogonadism. A duplication part of the short arm of the X chromosome, which contains this gene, in XY individuals is linked to dosage-sensitive sex reversal.