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P2RY8 wt Allele

National Cancer Institute

Source

National Cancer Institute. <u>P2RY8 wt Allele</u>. NCI Thesaurus. Code C97684.

Human P2RY8 wild-type allele is located in the vicinity of Xp22.33 and Yp11.3 and is approximately 75 kb in length. This allele, which encodes P2Y purinoceptor 8 protein, is involved in G-protein coupled receptor signaling. A chromosomal deletion in the pseudoautosomal region 1 of either chromosome X or Y causing fusion of this gene with the CRLF2 gene is associated with B-cell acute lymphoblastic leukemia. Two pericentric inversions of the X chromosome inv(X)(q13;p22) and inv(X)(p22.3;q13.2) disrupt the gene and are associated with mental retardation.