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NPHP1 wt Allele

National Cancer Institute

Source

National Cancer Institute. *NPHP1 wt Allele*. NCI Thesaurus. Code C75862.

Human NPHP1 wild-type allele is located in the vicinity of 2q13 and is approximately 83 kb in length. This allele, which encodes nephrocystin-1 protein, plays a role in the progression of adhesion-dependent signaling pathways. Mutations in the gene are associated with familial juvenile nephronophthisis type 1, Senior-Loken syndrome type 1, and Joubert syndrome type 4.