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# HSPG2 wt Allele

National Cancer Institute

## Source

National Cancer Institute. *HSPG2 wt Allele*. NCI Thesaurus. Code C95447.

Human HSPG2 wild-type allele is located within 1p36.1-p34 and is approximately 115 kb in length. This allele, which encodes basement membrane-specific heparan sulfate proteoglycan core protein, plays a role in both the structure of and cell interaction with the basement membrane. Mutation of the gene is associated with both Schwartz-Jampel syndrome and Silverman-Handmaker type dyssegmental dysplasia.