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CBS wt Allele

National Cancer Institute

Source

National Cancer Institute. *CBS wt Allele*. NCI Thesaurus. Code C129050.

Human CBS wild-type allele is located in the vicinity of 21q22.3 and is approximately 24 kb in length. This allele, which encodes cystathionine beta-synthase protein, is involved in the detoxification of homocysteine and the synthesis of cysteine. Mutation of the gene is associated with cystathionine beta-synthase deficiency (CBS), which can lead to homocystinuria and an increase risk of thrombosis.