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CRYAB wt Allele

National Cancer Institute

Source

National Cancer Institute. *CRYAB wt Allele*. NCI Thesaurus. Code C115114.

Human CRYAB wild-type allele is located within 11q22.3-q23.1 and is approximately 15 kb in length. This allele, which encodes alpha-crystallin B chain protein, plays a role in maintaining the structure of the lens. Mutation of the gene is associated with myofibrillar myopathy type 2, dilated cardiomyopathy type 11i; cataract 16 and alpha-B crystallin-related, fatal infantile hypertrophy myofibrillar myopathy.