GPHN wt Allele

National Cancer Institute

Source

National Cancer Institute. GPHN wt Allele. NCI Thesaurus. Code C97526.

Human GPHN wild-type allele is located in the vicinity of 14q23.3 and is approximately 674 kb in length. This allele, which encodes gephyrin protein, plays a role in both protein-cytoskeleton interactions and biosynthesis of the molybdenum cofactor. Mutation of the gene is associated with both molybdenum cofactor deficiency type C and startle disease. A chromosomal translocation t(11;14)(q23;q23) of this gene and the MLL gene is involved in acute myeloid leukemia.