ATP7B gene

ATP7B	
13q14.3-q21.1	
Associated diseases	Wilson's disease
Function	encodes the product of the same name
OMIM	606882
HGNC	870

The ATP7B gene (13q14.3-q21.1; OMIM: 606882) encodes the product of the same name. The full name is: ATPase, Cu 2+ transporter, beta polypeptide.

Mutations in this gene cause Wilson's disease . Because ATP7B ATPase is a large transmembrane protein (8 transmembrane regions, copper binding domain and ATP binding domain), there are many mutations that cause this protein to malfunction.

So far, about 300 such mutations are known to occur in different regions of the protein. Nevertheless, about 40% (value varies geographically) of Wilson's disease is caused by a single mutation (H1069Q). This affects the ATP binding domain and prevents the cleavage of an already bound ATP molecule.

Links

External links

- Oregon Health&Science University (https://www.ohsu.edu/xd/)

References

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