Gen ATP7B

This article has been translated from WikiSkripta; the **formatting** needs to be checked.
This article has been translated from WikiSkripta; ready for the **editor's review**.

Template:Infobox - gen The **ATP7B gene** (13q14.3-q21.1; OMIM: 606882 (https://omim.org/entry/606882)) encodes the eponymous product. The full name is: ATPase, Cu^{2+} transporting, beta polypeptide.

Mutations of this gene cause Wilson's disease. As ATP7B ATPase is a large transmembrane protein (8 transmembrane segments, a copper-binding domain and an ATP-binding domain), there are many mutations causing the non-functionality of this protein.

File:Atp7b.jpg

So far, about 300 such mutations are known, manifesting in different sections of the protein. However, about 40 % (the value varies geographically) of Wilson's disease is caused by a single mutation (H1069Q). This affects the ATP-binding domain and makes it impossible to split the already bound ATP molecule.

Links

Source

- ws:Gen ATP7B

External links

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

References

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