



RESEARCH & REVIEW

Discussion of "Novel ATP7B mutations in Vietnamese patients with Wilson disease" by Tue Nguyen *et al.*

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L902P is not novel. It was reported in a Vietnamese paper in 2013 along with the other novel mutations including T850I, P868P-fs (del C), L1015R, D1027H, IVS 15-2:A>G and IVS20+3:A> G. The 11-year-old girl with homozygote of L902P was diagnosed with H2 phenotype (Ferenci *et al.*, 2003), Kayser Fleischer ring, low ceruloplasmin (10.4 mg%) and high 24h urinary copper (396 mcg/24h).

The papers in *Nhi Khoa* (the local Vietnamese journal) are cited as:

Hoàng Lê Phúc, Krsin Zinober, Claudia Willheim, Peter Ferenci (2013). Phân tích đột biến phân tử gen ATP7B ở bệnh nhân Wilson Việt Nam. *Nhi khoa* 6 (4): 21-28.

Hoàng Lê Phúc, Phạm Lê An, Trần Diệp Tuấn (2015). Xác định bước đầu mối liên quan kiểu gen-kiểu hình của bệnh Wilson ở trẻ em Việt Nam. *Nhi khoa* 8 (6): 67-76.

References

Ferenci P, Caca K, Loudianos G, Mieli-Vergani G, Tanner S, Sternlieb I, *et al.* (2003). Diagnosis and phenotypic classification of Wilson disease. *Liver Int*, 23, 139-142

Closure to "Novel ATP7B mutations in Vietnamese patients with Wilson disease" by Tue Nguyen *et al.*

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The papers "Phân tích đột biến phân tử gen ATP7B ở bệnh nhân Wilson Việt Nam" and "Xác định bước đầu mối liên quan kiểu gen-kiểu hình của bệnh Wilson ở trẻ em Việt Nam" by Hoàng *et al.* were published in a Vietnamese journal that does not have an online version. These papers, even their abstracts, are not available in the Internet. Therefore, we did not know their existence when writing our manuscript; consequently, we have not referenced them in our paper. We confirm that the L902P mutation was detected independently, and the overlap of the result is entirely random. As L902P was reported by both Hoàng *et al.* and our group, it appears that this novel mutation might be popular in Vietnamese Wilson patients.