CORRESPONDENCE

Familial transthyretin-related amyloid polyneuropathy in a Malaysian patient of ethnic Chinese descent

We read with interest the article by Goh et al. in the 2008 December issue. They report a Malaysian patient of ethnic Chinese descent with familial amyloid polyneuropathy (FAP) and a TTR mutation. Because the first 20 codons of TTR encode a signal peptide, which is cleaved to form a mature transthyretin protein, the numbering system for transthyretin mutations does not include the 20-codon signal peptide (see The Human Gene Mutation Database at the Institute of Medical Genetics in Cardiff, http://www.hgmd.cf.ac.uk; or Data Base on Transthyretin Mutations, http://www.ibmc.up.pt/mjsaraiva/trrmut.html). Therefore, the transthyretin mutation Goh et al. report should be Ala97Ser, but not Ala117Ser. Although wide spectrum of TTR mutations of FAP have been described in Caucasians, there is still limited information in Chinese. Seven unrelated Taiwanese patients of Chinese in origin with FAP had been reported to carry TTR Ala97Ser.2-4

This report by Goh et al. further stresses the possibility that TTR Ala97Ser is a mutation hotspot among Chinese FAP patients.

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REFERENCES


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