Perry syndrome: a case of atypical parkinsonism with confirmed DCTN1 mutation: a response

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With great interest, we have read the article entitled “Perry syndrome: a case of atypical parkinsonism with confirmed DCTN1 mutation” by McManus et al published in 24 April edition of the New Zealand Medical Journal.1

We want to congratulate the authors for this important publication. Perry syndrome is a very rare disease with only handful of cases described around the world.2 Thus, it is interesting to know that this disease is also present in New Zealand, a country with only about five million inhabitants. It is very likely that the second case published by McManus et al1 is related to the first case published previously.3 They do share the same mutation, DCTN1 p.Y78C. It would be important to proceed with genealogical investigations locally and to perform molecular genetic experiments for haplotype sharing. These two approaches will help to determine if those two cases are indeed related.

The article by McManus et al1 also expands our knowledge on phenotypic presentations of Perry syndrome. Recurrent syncopes have not been previously associated with this condition. Due to postural hypotension seen in the Schellong test, it is possible that the patient's syncopal episodes originated from dysautonomia.1

The same mutation, DCTN1 p.Y78C, has also been identified in a Korean family. Interestingly, this family presented with an atypical phenotype consisting of PSP-like Parkinsonism in the absence of psychiatric symptoms, weight loss and hypoventilation.2,4 Thus, it is feasible that all three families albeit identified and described separately represent one extended kindred.

Competing interests: Nil.

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