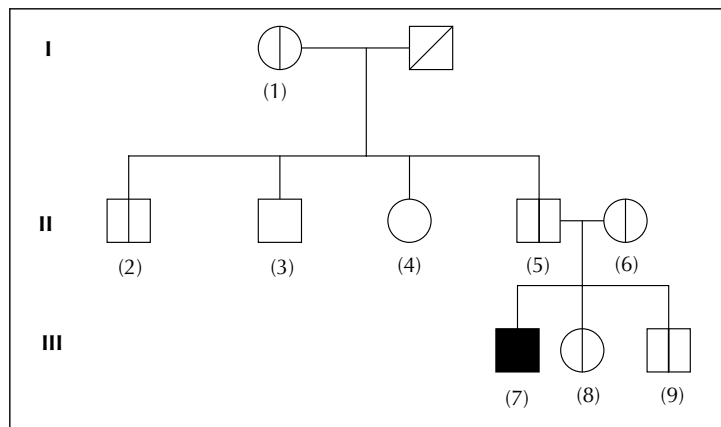


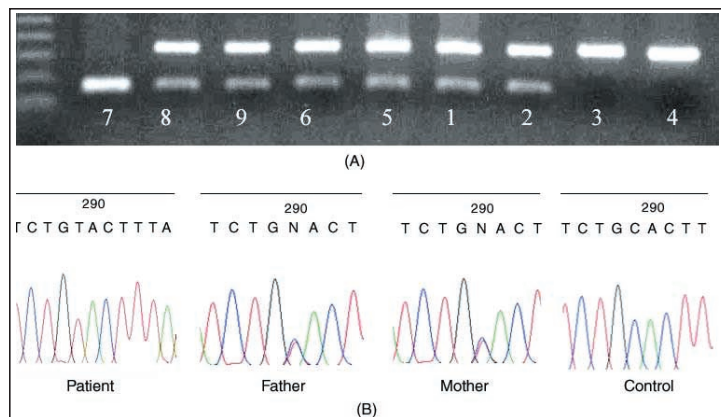
ucts were subjected to cycle sequencing using an ABI PRISM 377 (Applied Biosystems) DNA sequencer. A novel mutation was identified in exon 2 result-

ing in a change of histidine to tyrosine at amino acid position 173 (H173Y), which also creates an *RsaI* restriction site. To better visualize the restriction

products, exon 2 of the gene was re-amplified using custom-designed primers (5'-ACCTGACCTCCAATGTGG-3') and (5'-AGTGTGGA GACTCGAGAAG-3'). Amplified products (176 base pairs) were subjected to restriction endonuclease analysis by using *RsaI*; 0.5 units of enzyme were used for 10 mL of PCR reaction products. After a 3-hour incubation at 37°C, electrophoresis was carried out on a 2% agarose gel. Fig. 2 shows the restriction analysis for all the family members. The patient is homozygous for this mutation, whereas both the siblings and subjects 1, 2, 5 and 6 are heterozygous.



**Fig. 1: Pedigree of our patient with pantothenate kinase-associated neurodegeneration.** Circles represent females and squares, males. The square with an oblique line through it represents a deceased grandparent. The patient (homozygous for the mutation H173Y in the *PANK2* gene) is represented by a black box. Heterozygotes for the mutation are shown with half-cut boxes. Homozygotes for the wild-type genotype are shown with blank boxes. Numerical codes for all subjects are given in parentheses.



**Fig. 2: The mutation H173Y (C to T) creates an *RsaI* restriction site.** (A) A 2% agarose gel shows restriction analysis by the *RsaI* enzyme. The amplified product (176 base pairs) of exon 2 generated by custom-designed primers was digested by the enzyme at 37°C for 3 hours. The numbers below each lane represent numerical codes of subjects in Fig. 1. Subjects 3 and 4 carry the wild-type genotype and, thus, their amplified products remained undigested, showing only 1 band in the top panel. Both the siblings (8 and 9) and subjects 1, 2, 5 and 6 are heterozygous for this mutation, with 1 allele carrying the mutation and the other carrying the wild-type gene; thus, the top band shows the wild-type allele and the bottom one shows the mutant allele. Patient (7) is homozygous for the mutation, with both the alleles having the mutation, thus, displaying only 1 lower band. (B) Electrophoretogram of patient, father, mother and control. A transition from "C" in control to "T" in the patient can be seen, whereas the 2 parents are heterozygous.

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## Correction

A correction<sup>1</sup> that appeared in a recent issue of *CMAJ* should have also referred to the print version of the relevant article.<sup>2</sup>

## References

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