(E)-Figure 1 legend - Ohtake et al. Identification of SNCB alterations in DLB.

A. Detection of the V70M amino acid substitution. Here we partial sequence from exon 4 of the *SNCB* gene showing a heterozygous alteration at amino acid position #70 that changes a GTG codon (Valine) to a ATG codon (Methionine).

B. Detection of the P123H amino acid substitution. Here we see partial sequence from exon 5 of the *SNCB* gene showing a heterozygous alteration at amino acid position #123 that changes a CCC codon (Proline) to a CAC codon (Histidine).

C. Screening for the P123H alteration by *Dra* III restriction endonuclease digestion. DNA samples from 416 individuals of the same population group as the index case were PCR-amplified using the standard B5F primer and a modified B5R primer containing a *Dra* III recognition site to serve as a positive control for adequate restriction endonucelase digestion. As shown schematically, uncut PCR product should be 295 bp, while *Dra* III cut product will be 263 bp if homozygous normal. Samples heterozygous for the P123H alteration should yield a 263 bp fragment, a 141 bp fragment, and a 122 bp fragment after *Dra* III restriction digestion. In the accompanying ethidium-stained agarose gel, the results of a representative P123H SNCB allele screen are shown. Uncut product migrates at 295 bp (lane 1), while *Dra* III cut product from three normal controls (lane 2, 3 and 9) migrates at 263 bp, indicating that they lack the P123H SNCB allele. DNAs isolated from peripheral blood lymphocytes (lanes 4 and 6), skin (lane 5), and brain (lane 7) all yield PCR products that upon *Dra* III restriction digestion migrate at 263 bp, 141 bp, and 122 bp. Thus, while patient material consistently revealed the heterozygous pattern in each run, normal control samples always showed just the 263 bp product. Note that lane 8 is a negative PCR control and lacks DNA template.

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