SUPPLEMENTARY FILE

An inversion affecting the GCH1 gene as a novel finding in dopamine-responsive dystonia

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Supplementary Figure S1. Pedigree of the family. Arrow indicates proband who underwent genetic studies, filled symbol indicates affected, squares represent males and circles represent females. d. MVA = died in motor vehicle accident. Additional family members were not available for testing.



Supplementary Figure S2. Illumina short read whole genome sequencing data indicating a structural variant

(NC_000014.8:g.[55343254_55346605del;55346606_60822142inv;60822143_60823119del]) on chromosome 14 affecting *GCH1*. Chimaeric ("split") reads identified by the ClinSV tool are visualized in the IGV genome browser. To ease interpretation the alignments of segments of two representative reads are highlighted (A00488:195:HGJN7DSX2:3:2336:15573:12743 in red, and A00488:195:HGJN7DSX2:3:2160:31503:1219 in blue). Other tracks show the deletions which flank the inversion, and the exonic structure of *GCH1* transcript NM_000161.3. **(A)** The left-hand (centromere proximal) breakpoint region, associated with a 3.4 kb deletion. **(B)** The right-hand (centromere distal) breakpoint region, associated with a smaller (1.0 kb) deletion.



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Supplementary Figure S3. Oxford Nanopore long read sequencing (LRS) data supporting the proposed structural variant. Chimaeric nanopore sequences are visualized in the IGV genome browser. The sequence alignments confirm the breakpoints indicated by short read sequencing analysis, and extend wide enough for a high level of confidence in read locations. **(A, B)** Inversion breakpoint regions as in Fig. <u>S2</u>, but in a 50 kb window.

