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Han Chinese patients with dopa-responsive dystonia exhibit a low frequency of exonic deletion in the *GCH1* gene

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ABSTRACT. We identified three novel mutations of the GTP cyclohydrolase 1 (*GCH1*) gene in patients with familial dopa-responsive dystonia (DRD), but were unable to identify meaningful sporadic mutations in patients with no obvious family DRD background. To investigate whether *GCH1* regional deletions account for the etiology of DRD, we screened for heterozygous exonic deletions in DRD families and in patients with sporadic DRD. Multiple ligation-dependent probe amplification analysis and quantitative real-time polymerase chain reaction amplification was performed in all members of our DRD cohort and in controls to detect exonic deletions in *GCH1*, tyrosine hydroxylase, and the epsilon-sarcoglycan-encoding (*SGCE*) genes. Using these techniques, we detected a *GCH1* exon 1 heterozygous deletion in 1 of 10 patients with sporadic DRD. Therefore, we concluded that exonic deletion in the *GCH1* gene only accounted for the etiology in a small percentage of patients with sporadic DRD in our Han Chinese cohort.

Key words: Dopa-responsive dystonia; GTP cyclohydrolase 1; Exonic deletion; Multiple ligation-dependent probe amplification analysis

