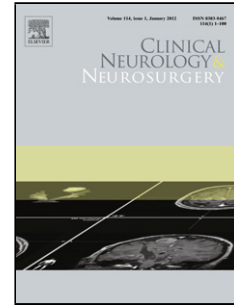


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**Novel homozygous *GBA2* mutation in a patient with complicated spastic paraplegia**

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**Highlights**

1. Identification of a novel homozygous biallelic c.452-1G>C mutation in *GBA2* gene causative for spastic paraplegia type 46
3. The impact of Next Generation Sequencing technology on human genetics
4. To collect information and experience on hereditary spastic paraplegias cases

**Abstract:** Hereditary spastic paraplegias (HSPs) are a heterogeneous group of neurological disorders characterized primarily by a pyramidal syndrome with lower limb spasticity, which can manifest as pure HSP or associated with a number of neurological or non-neurological signs (i.e., complicated HSPs). The clinical variability of HSPs is associated with a wide genetic heterogeneity, with more than eighty causative genes known. Recently, next generation sequencing (NGS) has allowed increasing genetic definition in such a heterogeneous group of

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