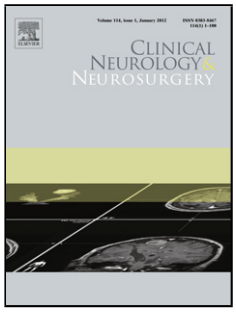

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Novel homozygous *GBA2* mutation in a patient with complicated spastic paraplegia

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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