Autosomal dominant transmission of complicated hereditary spastic paraplegia due to a dominant negative mutation of *KIF1A*, SPG30 gene

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¹Department of Pediatrics, Pusan National University Children's Hospital, Yangsan, Korea. ²Research Institute for Convergence of Biomedical Science and Technology, Pusan National University Yangsan Hospital, Yangsan, Korea. ³Rare Disease Research Center, Korea Research Institute of Bioscience and Biotechnology, Daejeon, Korea. ⁴Center for Synaptic Brain Dysfunctions, Institute for Basic Science (IBS), Daejeon, Korea. ⁵Department of Biological Sciences, Korea Advanced Institute of Science and Technology, Daejeon, Korea. Figure S1. Expression of KIF1A-MD mutants is comparable to WT KIF1A-MD. (A) WT KIF1A-MD and KIF1A-MD mutants were transfected in HEK293 cells using the calcium phosphate method and the expression of the KIF1A-MD proteins was assessed in the total cell lysates by Western blotting using an anti-EGFP antibody after 2 days of transfection. The levels of expressions of KIF1A-MD mutants were comparable to WT KIF1A-MD. (B) The expression of WT KIF1A-MD and KIF1A-MD mutants were confirmed by immunocytochemistry using EGFP antibody in transfected COS1 cells as described in the methods. KIF1A-MD mutants as well as WT KIF1A-MD were evenly distributed throughout the whole cell body.

