CGH array of the proband revealed an interstitial dele- tion of at least 179.4 kb on chromosome 15q21.1 (genomic coordinates chr15:44,960,975–45,140,413; GRCh37/hg19) (Figure 3a). The maximum possible size of the dele- tion was 193.6 kb, spanning from chr15:44,957,934 to chr15:45,151,560. The deleted region encompassed three genes: PATL2, B2M, and TRIM69. According to ClinGen and published literature, none of them has been associ- ated with human disease in case of haploinsufficiency. PATL2 and B2M are OMIM morbid genes, but they are associated with recessive disorders with no apparent rela- tion to the patient's phenotype (Oocyte maturation defect 4, and Immunodeficiency 43, respectively); and TRIM69 may play a role in apoptosis. The deletion does not overlap with polymorphic CNVs reported in Database of Genomic Variants or gnomAD, and comparable variants have nei- ther been reported in ClinGen, DECIPHER nor published anywhere to the best of our knowledge. The deletion, however, was very close to the 5′ end of the SPG11 gene. According to current guidelines, the genetic loss was re- ported and classified as a variant of uncertain clinical sig- nificance. No additional rare CNVs were detected

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