Reviewer's report

Title: Transient, Recurrent, White Matter Lesions in X-linked Charcot-Marie-Tooth Disease with Novel Mutation of gap junction protein beta 1 gene in China: a case report

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Reviewer: Byung-Ok Choi

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The authors report the transient and recurrent white matter lesions in X-linked Charcot-Marie-Tooth Disease patient with a novel mutation of GJB1 gene. They said that the novel T278G mutation of Gap junction protein beta I maybe could result in X-linked Charcot-Marie-Tooth disease type 1 with predominant leucoencephalopathy, and the white matter changes in MRI of X-linked CMTX1 patient are reversible. However, those are well known phenotype in CMTX1, because Cx32 is expressed in the myelinating Schwann cells of the peripheral nerves, and the protein is also widely expressed in the oligodendrocyte of the central nervous system.

The authors said that the male patient with novel T278G mutation of GJB1 had homozygous, but his mother had heterozygous mutation. His father had normal sequence. The CMTX1 has been known to be an X-linked dominant inheritance pattern. So, I wonder the reason why this genotype appeared, and the authors must show the chromogram of this patient.

In addition, muscle weakness was developed in his both lower limbs, and disappeared. Also, the patient showed weakness of both upper limbs twice. It is very difficult to understand, because brain lesion of MRI show left corticospinal tract involvement.