LIMK2-1, une protéine impliquée dans la déficience intellectuelle
LIMK2-1 is a hominidae-specific isoform of LIMK2 expressed in central nervous system and associated with intellectual disability

Abstract

LIMK2 is involved in neuronal functions by regulating actin dynamics. Different isoforms of LIMK2 are described in databanks. LIMK2a and LIMK2b are the most characterized. A few pieces of evidence suggest that LIMK2 isoforms might not have overlapping functions. In this study, we focused our attention on a less studied human LIMK2 isoform, LIMK2-1. Compared to the other LIMK2 isoforms, LIMK2-1 contains a supplementary C-terminal phosphatase 1 inhibitory domain (PP1i). We found out that this isoform was hominidae-specific and showed that it was expressed in human fetal brain and faintly in adult brain. Its coding sequence was sequenced in 173 patients with sporadic non-syndromic intellectual disability (ID), and we observed an association of a rare missense variant in the PP1i domain (rs151191437, p.S668P) with ID. Our results also suggest an implication of LIMK2-1 in neurite
outgrowth and neurons arborization which appears to be affected by the p.S668P variation. Therefore our results suggest that LIMK2-1 plays a role in the developing brain, and that a rare variation of this isoform is a susceptibility factor in ID.

Keywords
Cytoskeleton remodeling; Hominidae-specific; Intellectual deficiency; LIM kinase; Neuron morphology; Patient mutation.

#LIMK #ID #intellectual deficiency #neuron #brain

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