A novel mutation in the transmembrane 6 domain of GABBR2 leads to a Rett-like phenotype

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Résumé en anglais	We read with great interest the recent article published by Yooet al1reporting 4 additional Rett-like (RTT) patients with therecurring A567TGABBR2mutation.2More interestingly, theyshowed, with in vitro and in vivo functional studies, that theseverity of the phenotype caused byGABBR2mutations wasdirectly linked to their impact onc-aminobutyric acid (GABA)signaling activity, this latter being more reduced with the 2 mis-sense mutations, S695I and I705N, associated with epilepticencephalopathy (EE).1,3They hypothesized that the position ofvariants in different transmembrane (TM) domains ofGABBR2,TM6 for S695I and I705N, and TM3 for A567T, could deter-mine the phenotypic expression. This hypothesis was recentlyreinforced with the report of a novelGABBR2mutation also inTM6 and associated with infantile epileptic spasms.
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Liens
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