

Title: *PLP1*-Related Disorders *GeneReview*: Molecular Genetics, Normal allelic variants
Authors: Hobson GM, Kamholz J
Date: February 2013

Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

Normal Allelic Variants: Supplementary Data

Description of selected normal allelic variants reported in *PLP1*.

Changes that do not affect the coding sense of the gene have been reported:

- -445C>G (5' of transcription initiation site) in the promoter
- -220-210del (5' of transcription initiation site) [Hübner et al 2005]
- c.-102C>T [Hübner et al 2005]
- c.-31C>T [detected in an unaffected male, Hobson, unpublished observation]
- Dinucleotide (CA) repeat polymorphism in intron 1
- c.5-111T>C, Msp1 polymorphism in intron 1 [Hobson et al 2001]
- c.168A>G, an MvaI polymorphism in exon 2; detected in an unaffected male, Hobson, unpublished observation]
- c.243T>C in exon 3 [Hobson et al, unpublished observation]
- c.606T>C, an AhaI/BsaHI polymorphism in exon 4
- c.622+28C>G in intron 4 [Hübner et al 2005; Hobson & Sistermans, unpublished observations]
- c.*101C>T, reported as insT932 by Poduslo et al [1993] and Hobson et al [unpublished observations]

References

Hobson G, Stabley D, Funanage V, Marks H. A new polymorphism in the proteolipid protein (PLP1) gene and its use for carrier detection of PLP1 gene duplication in Pelizaeus-Merzbacher disease. *Hum Mutat.* 2001;17:152.

Hübner CA, Orth U, Senning A, Steglich C, Kohlschütter A, Korinthenberg R, Gal A. Seventeen novel PLP1 mutations in patients with Pelizaeus-Merzbacher disease. *Hum Mutat.* 2005;25:321-2.

Poduslo SE, Decker P, Astle H, Kurth J, LaBate M. Identification of a new polymorphism in the human proteolipid protein gene. *Neurochem Int.* 1993;23:413-7.