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T [detected in an unaffected male, Hobson, unpublished observation]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G, an Mval polymorphism in exon 2; detected in an unaffected male, Hobson, unpublished observation]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C in exon 3 [Hobson et al, unpublished observation]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 AhaII/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G in intron 4 [Hübner et al 2005; Hobson & Sistermans, unpublished observations]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T, reported as insT932 by Poduslo et al [1993] and Hobson et al [unpublished observations]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

