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A highly polymorphic locus in 5p15.2-5p15.3 (213-274EC) revealed by an anonymous single copy DNA fragment

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Source Description: 213-274EC is a single copy 1.7 Kb EcoRI fragment cloned into pUC8. The fragment was subcloned from a phage isolated from an MboI partial digest library (in EMBL-4) prepared from a somatic cell hybrid (HHW 213) that contains an intact short arm and small portion of the long arm of human chromosome 5 as its only human DNA (Carlock and Wasmuth, 1985).

Polymorphisms: In a sample of 12 unrelated individuals, polymorphisms were identified with BglI, EcoRI, HindII, HindIII, PstI, PvuII, SphI, SstI, TaqI, XbaI and XmnI. SphI alone reveals at least 8 different alleles (See Figure for SphI polymorphisms revealed in 6 individuals). The PIC value of the marker using just SphI is approximately 0.84.

Chromosomal Location: Localized to 5p15.2-5p15.3 by Southern blot analysis of a panel of somatic cell hybrids (Overhauser, Beaudet and Wasmuth, Am. J. Hum. Genet.; in press).

Mendelian Inheritance: Codominant inheritance according to Mendelian expectations shown for two families.

Probe Availability: Available for studies other than on cri du chat syndrome.

Other Comments: A combination of SphI, HindII and XmnI appears to maximize the informativeness of the marker. Low background at a washing stringency of 1xSSC.

Reference: Carlock and Wasmuth. "Molecular Approach to Analyzing the Human 5p Deletion Syndrome, Cri du Chat" Som. Cell Mol. Genet. 11, 267-276 (1985).

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