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A Novel Form of Human Polymorphism Involving the hDHFR-ψ1 Pseudogene Identifies Three RFLPs

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SOURCE AND DESCRIPTION OF CLONE: pCHB 235, a 0.42 kb XbaI-EcoRI fragment, subcloned into pUC12 from the 4.0 kb genomic insert of phage clone λ hDHFR-ψ_1 isolated from an EcoRI library, containing the 3' flanking region of hDHFR-ψ_1 pseudogene (Chen et al., 1982).

POLYMORPHISM: The hDHFR-ψ_1 pseudogene is present in DNA of some individuals and absent from DNA of others, representing a novel form of polymorphism (Anagnou et al., 1984).

FREQUENCY: The presence (+) or absence of the pseudogene (-) can be detected with digestion with at least three enzymes. PstI detects a two allele polymorphism with allelic fragments of 2.9 kb (-) and 2.5 kb (+). BglII detects a two allele polymorphism with allelic fragments of 6.7 kb (-) and 3.85 kb (+). Bam HI detects a two allele polymorphism with allelic fragments of 5.0 kb (-) and 33.0 (+). The allelic frequency of the pseudogene as defined by studying 180 chromosomes is: Mediterraneans 0.94, Asian Indians 0.77, Chinese 0.67, Southeast Asians 0.57 and American Blacks 0.33

CHROMOSOMAL LOCALIZATION: Using a panel of somatic cell hybrids, the hDHFR-ψ_1 pseudogene was localized to human chromosome 18 (Anagnou et al. 1987).

MENDELIAN INHERITANCE: Co-dominant segregation observed in at least 3 Caucasian families.

PROBE AVAILABILITY: Freely available for linkage studies.

OTHER COMMENTS: Low background at strigency of 0.1X SSC at 65°C.

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