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Nicholas P. Anagnou National Heart, Lung, and Blood Institute

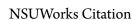
S. E. Antonarakis *Johns Hopkins University*

Stephen J. O'Brien
National Cancer Institute at Frederick, sobrien 1@nova.edu

Arthur W. Nienhuis

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A novel form of human polymorphism involving the hDHFR- ψ_1 pseudogene identifies three RFLPs

N.P.Anagnou*, S.E.Antonarakis1, S.J.O'Brien2 and A.W.Nienhuis

Clinical Hematology Branch, NHLBI, NIH, Bethesda, MD 20205, ¹Department of Pediatrics, Johns Hopkins University, Baltimore, MD 21205 and ²Laboratory of Viral Carcinogenesis, NCI, NIH, Frederick, MD 21701, IISA

SOURCE AND DESCRIPTION OF CLONE: pCHB 235, a 0.42 kb XbaI-EcoRI fragment, subcloned into pUCl2 from the 4.0 kb genomic insert of phage clone $\lambda\,hDHFR-\,\psi_1$ 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 0.1% SSC at 65°C.

*Present address: Department of Medicine, RG-25, University of Washington, Seattle, WA 98195, USA

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