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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, [sobrien1@nova.edu](mailto:sobrien1@nova.edu)*

Arthur W. Nienhuis

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### A novel form of human polymorphism involving the hDHFR- $\psi_1$ pseudogene identifies three RFLPs

N.P.Anagnou\*, S.E.Antonarakis<sup>1</sup>, S.J.O'Brien<sup>2</sup> and A.W.Nienhuis

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

**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  $\lambda$ hDHFR- $\psi_1$  isolated from an EcoRI library, containing the 3' flanking region of hDHFR- $\psi_1$  pseudogene (Chen et al., 1982).

**POLYMORPHISM:** The hDHFR- $\psi_1$  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- $\psi_1$  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 stringency of 0.1X SSC at 65°C.

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

**REFERENCES:** Chen MJ et al. Proc. Natl. Acad. Sci. USA 79:7435-7439, 1982  
Anagnou NP et al. Proc. Natl. Acad. Sci. USA 81:5170-5174, 1984  
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