KpnI RFLP for the human CSF-1 gene

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Source and Description of Clone: The plasmid p3ACSFRI contains a 2.0 kb EcoRI-XhoI fragment of human CSF-1 cDNA subcloned into pXM (1)

Polymorphism: KpnI digestion gives one invariant band of 3.8 kb and a two allele polymorphism with bands of 16 kb (K1) and 8.0 kb and 7.6 kb (K2)

Frequency: (among 15 unrelated Caucasians)

K1 (16 kb): 0.53

K2 (8.0 + 7.6 kb): 0.47

Not Polymorphic For: (among 10 unrelated individuals) Taq I, Pst I, XbaI, EcoRV, BamHI, Hind III, PvuII, EcoRI, MspI, StyI.

Chromosomal Location: The CSF-1 gene has been mapped to human chromosome 5q33.1 (2).

Mendelian Inheritance: Segregation was demonstrated in one family of 18 individuals.

Probe Availability: Available from Dr. S. Clark, Genetics Institute, Inc., 87 Cambridge Park Drive, Cambridge, MA 02410.

References: 1) Wong, G.G. et al. (1987) Science 235:1504-1508. 2) Pettenati, M.J. et al. (1987) Proc. Natl. Acad. Sci. USA 84:2970-2974.

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