Two polymorphisms for the human G\textsubscript{M2} activator protein gene

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Source/Description: A 300 bp HincII/EcoRI fragment of the human G\textsubscript{M2} activation protein cDNA (1) was subcloned into Bluescript vector and used as a probe.

Polymorphism: Mspl identifies a two allele polymorphism with bands at either 3.4 kb (A1) or 2.9 kb (A2) and invariant bands at 1.0 kb, 1.8 kb and 4.0 kb.

Rsal identifies a two allele polymorphism with a band at either 4.1 kb (B1) or 3.4 kb (B2) and one invariant band at 2.5 kb.

Frequency: Mspl—studied in 27 unrelated healthy Caucasians (North America)
A1 = 0.77 and A2 = 0.23
Rsal—studied in 23 unrelated healthy Caucasians (North American)
B1 = 0.18 and B2 = 0.82

Not Polymorphic For: EcoRI, BglII, BamHI, HindIII, Asp700, TaqI and PstI.

Chromosomal Localization: The G\textsubscript{M2} activator protein gene has been localized to chromosome 5 by enzyme linked immunosorbent assay (ELISA) on human mouse somatic cell hybrids (2).

Mendelian Inheritance: Co-dominant inheritance shown in 10 Caucasian families.

Probe Availability: Contact D.Mahuran.


A rare insertion/deletion polymorphism at the HEXA locus

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Source/Description: A cDNA clone pHexA49 (1), encoding the α-subunit of β-hexosaminidase. The entire cDNA (excised with Xhol) was used as a probe.

Polymorphism: Xbal identifies a two allele polymorphism with bands at either 5.0 kb (A1) or 5.2 kb (A2). Two invariant bands of 9.6 kb and 12 kb are also detected. Both SstI and TaqI identify the same polymorphism indicating the presence of an insertion/deletion type of polymorphism.

Frequency: Studied in 45 unrelated Caucasians.
A1 = 0.07 and A2 = 0.93
Also studied in 41 unrelated Ashkenazi Jews
A1 = 0.04 and A2 = 0.96.

Not Polymorphic For: EcoRI, BglII, HincII, BamHI, HindIII and PstI.

Chromosomal Localization: The cDNA encoding for the HEXA gene has been localized to 15q23–24. (2).

Mendelian Inheritance: Co-dominant inheritance demonstrated in 3 families.

Probe Availability: pHexA49 is available from the American Type Culture Collection.

Other Comments: The 5.0 kb allele was not present in several Tay-Sach's cell lines and carriers.