HindIII/EcoRI polymorphism in the GAA gene

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Source/Description: Full length cDNA coding for human lysosomal glucosidase alpha gene was cloned in the eukaryotic expression vector pSG5 (1). The 3.6 kb EcoRI insert was used as a probe.

Polymorphism: In HindIII/EcoRI digested DNA the probe recognises a three-allele polymorphism with fragments of 5.2 (A1), 4.6 (A2), or 3.1 and 1.5 kb (A3). Constant fragments are 12 and 9 kb (see figure).

Frequency: In 17 unrelated Caucasians and 24 Asians:

	Caucasians	Asians
A1	–	0.29
A2	0.87	0.63
A3	0.13	0.08

Not Polymorphic For: PvuII, TaqI, RsaI, BclI, BamHI when tested in 18 individuals.

Chromosomal Localisation: 17q23.

Mendelian Inheritance: Co-dominant segregation of the A1 and A2 allele observed in three pedigrees.

Probe Availability: Contact A.J.J.Reuser.

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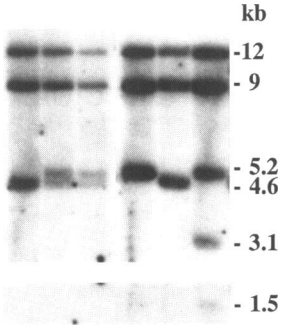
Reference: Hoefsloot,L.H. *et al.* (1988) *EMBO J.* **7**, 1697–1704.



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Dinucleotide repeat polymorphism at the D11S35 locus

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Source and Description: Phage 2–22 was from a human genomic library of CF52, a mouse X human somatic cell hybrid that retained a single translocated human chromosome, t(11;16)(q13;p11) on a rodent background (1). A Sau3A subclone of this phage that contained a $(GT)_{17}$ repeat was sequenced and sequences flanking the repeat (EMBL accession no. X52579) were used to design PCR primers.

PCR Primers:
(#780)-5′-ACAATTGGATTACTACTAGC-3′
(#781)-5′-TGTATTTGTATCGATTAACC-3′

Polymorphism: Allelic fragments were resolved on DNA sequencing gels. Lengths of allelic fragments (nt) were: A1=162, A2=160, A3=158, A4=156, A5=154, A6=152.

Frequencies: Allele frequencies in 17 unrelated European Caucasians: A1=.12, A2=.26, A3=.15, A4=.12, A5=.18, A6=.18. Heterozygosity=0.88; PIC=0.79.

Chromosomal Localization and Mendelian Inheritance: In situ hybridization and hybridization of phage 2–22 to genomic DNAs from a somatic cell hybrid panel indicated localization to 11q22 (1). Linkage analysis of the D11S35 MspI RFLP in CEPH families supports the order cen-D11S84-D11S35-D11S424-qter (2). Mendelian inheritance was observed in three informative families with a total of 23 children.

PCR Conditions: We carry out PCR in a total volume of 25 µl containing: 50 ng genomic DNA, 25 pmoles of each primer, 1.5 mM $MgCl_2$, 200 µM unlabeled dNTPs, 50 mM KCl, 10 mM Tris-Cl^-, pH 8.3, 0.6 units Taq polymerase (Perkin-Elmer/Cetus) and 0.01% gelatin. 1 µCi [^{32}P]dCTP is added to each sample. Amplification is for 30 cycles with denaturation at 93°C for 1 min, annealing at 40°C for 2 min and extension at 72°C for 2 min. Products are resolved on DNA sequencing gels and visualized by autoradiography.

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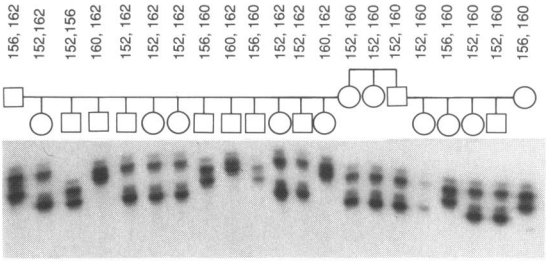
References: 1) Maslen,C.L., Jones,C., Glaser,T., Magenis,R.E., Sheehy,R., Kellogg,J. and Litt,M. (1988) *Genomics* **2**, 66–75. 2) Julier,C., Nakamura,Y., Lathrop,M., O'Connell,P., Leppert,M., Litt,M., Mohandas,T., Lalouel,J.-M. and White,R. (1990) *Genomics* **7**, 335–345.



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