Identification of a HindIII and a TaqI RFLP at the acid alpha glucosidase (GAA) locus

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Source/Description: A 1.9 kb cDNA EcoRI segment subcloned into pUC 19 (GAA67; '2 kb') (1).

Polymorphisms: HindIII detects a two allele polymorphism 5.6 kb (A1) and 5.3 kb (A2). TaqI detects a two allele polymorphism 10 kb (A1) and 9 kb (A2).

Frequency: HindIII: studied in 22 unrelated Caucasians, gene frequency of 5.6 kb allele = 0.05 and of 5.3 kb allele = 0.95, heterozygosity = 0.09. TaqI: studied in 25 unrelated Caucasians, gene frequency of 10 kb allele = 0.94 and of 9 kb allele = 0.06, heterozygosity = 0.12.

Chromosomal Localization: 17q23.

Mendelian Inheritance: HindIII: Six matings studied were all monomorphic. However, the frequencies of heterozygotes and homozygotes among independent non-family members fit Hardy-Weinberg equilibrium. TaqI: Co-dominant inheritance in three matings with three offspring; gene frequencies fit Hardy-Weinberg equilibrium.

Probe Availability: Contact F. Martiniuk, NYU Medical Center.

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Reference: 1) Martiniuk *et al.* (1986) *PNAS* **83**, 9641-9644.

Further characterization of PstI RFLPs at the acid alpha glucosidase (GAA) locus

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Source/Description: A 1.9 kb cDNA EcoRI segment subcloned into pUC 19 (GAA-67; '2 kb') (1) and subfragments indicated below.

Polymorphisms: PstI detects three polymorphisms; A: a two allele polymorphism 4.9 kb (A1) and 4.4 kb (A2), also detected with a 0.65 kb SacI/NcoI subfragment, bp 1857 to 2520 (2); B: a three allele polymorphism 3.8 kb (B1), 3.6 kb (B2) and 3.3 kb (B3), also detected with a 0.7 kb NcoI/SacI subfragment, bp 2520 to 3236 (2); C: a two allele polymorphism 1.55 kb (C1) and 1.35 kb (C2), also detected with a 0.4 kb EcoRI/SacI subfragment, bp 1506 to 1857 (2).

Frequency: PstI studied in 60 unrelated Caucasians: 12 randomly ascertained individuals; 12 from Utah kindreds and 1 from Family 2 from NIGMS Human Genetic Mutant Cell Repository at the Coriell Institute, Camden, NJ; 18 from a large Venezuelan kindred (3); 17 from von Recklinghausen neurofibromatosis kindreds (4).

A1 = .07	B1 = .26	C1 = .74
A2 = .93	B2 = .73	C2 = .26
	B3 = .01	

Three haplotypes in the Venezuelan kindreds were

A: A2 B2 C1, .63 B: A2 B1 C2, .28 C: A1 B2 C1, .09

Chromosomal Localization: 17q23.

Mendelian Inheritance: Co-dominant segregation in 14 matings.

Probe Availability: Contact F. Martiniuk, NYU Medical Center.

Other Comments: There is apparent linkage disequilibrium between PstI B2 and PstI C1 seen in 58 individuals. Additionally there is apparent linkage disequilibrium between PstI B2, PstI C1 and RsaI A2 (5) seen in 18 of 19 individuals as well as between SacI A2 (6) seen in 15 of 18 individuals.

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References: 1) Martiniuk *et al.* (1986) *PNAS* **83**, 9641-9644. 2) Martiniuk *et al.* (1990) *DNA Cell Biol.* **9**, 85-94. 3) Tanzi *et al.* (1988) *Genomics* **3**, 127-136. 4) Seizinger *et al.* (1987) *Cell* **49**, 589-594. 5) Tzall *et al.* (1990) *Nucl. Acids Res.* **18**, 1661. 6) Tzall *et al.* (1990) *Nucl. Acids Res.* **18**, 1930.