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DNA VARIANTS

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# An intragenic *Taq*I polymorphism in the faciogenital dysplasia (FGD1) locus, the gene responsible for Aarskog syndrome

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**Abstract** A *Taq*I polymorphism, located in intron 4 of the faciogenital dysplasia *(FGD1)* gene, the gene responsible for Aarskog syndrome, is described. *FGD1* encodes a putative Rho/Rac guanine nucleotide exchange factor involved in mammalian morphogenesis. The identification of an intragenic polymorphism will facilitate the accurate carrier detection of individuals at risk for Aarskog syndrome.

*Description.* The FGD1 probe is a 3.85-kb *Eco*RI cDNA insert, denoted as pFCF3.85, that encodes the complete open reading frame of the faciogenital dysplasia gene, the gene responsible for Aarskog syndrome (Pasteris et al. 1994).

*PCR primers.* E4-P9: 5′-TGAGGCCTCCCGCTGCCTGTTTC-3′. E5-P21: 5′-CTGATCCAGGAGATGGAGCCTGG-3′.

*Polymorphism.* Used as a probe to perform hybridization analyses of *Taq*I-digested genomic DNA, probe pFCF3.85 detected two alleles: A1, 6.0 kb and A2, 3.8 kb. The 6.0-kb fragment represented the 3.8-kb fragment (which included exon 4) and its adjacent 2.2-kb *Taq*I fragment (which included exon 5). To facilitate analysis, a PCR strategy was generated to investigate the presence or absence of the polymorphic *Taq*I site in intron 4. Genomic DNA was amplified using oligonucleotide primers directed against FGD1 exons 4 and 5. The polymorphism was identified by digesting the resulting 1.7-kb PCR product with *Taq*I; upon digestion, products containing the *Taq*I site yielded 1.2-kb and 0.5-kb fragments.

*Allelic frequency.* Estimated from 84 chromosomes of unrelated individuals (40 CEPH females and 2 random unrelated females). A1 = 0.13; A2 = 0.87.

*Chromosomal localization.* The FGD1 gene was localized to region Xp11.21 by somatic cell and radiation hybrid analyses (Pasteris et al. 1994).

*Mendelian inheritance.* Mendelian X-linked inheritance was observed in one family of five individuals.

*PCR conditions.* Amplifications were performed in 50-μl volumes containing 50 m*M* KCl, 10 m*M* Tris-HCl, pH 8.0, 10 mg/ml BSA, 1.5 m*M* $MgCl_2$, 200 μ*M* each of dATP, dGTP, dCTP, and dTTP, 100 ng of genomic DNA, 0.5 μ*M* primers, and 2.5 U Ampli*Taq* (Perkin Elmer/Cetus). Amplifications were performed in a DNA thermal cycler 480 (Perkin Elmer/Cetus); after an initial 10-min denaturation cycle (94° C), 34 cycles followed: 94° C denaturation 1 min, 65° C annealing 1 min, 72° C extension 2 min, with a final extension of 10 min at 72° C.

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## References

Pasteris NG, Cadle A, Logie LJ, Porteous MEM, Schwartz CE, Stevenson RE, Glover TW, Wilroy R, Gorski JL (1994) Isolation and characterization of the faciogenital dysplasia (Aarskog-Scott syndrome) gene: a putative Rho/Rac guanine nucleotide exchange factor. Cell 79:669–678