SacI RFLPs at the D8S51 locus

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Source/Description: L48 is a 150 bp single copy genomic DNA fragment cloned into pUC13. The clone was isolated by microdissection and microcloning of the Langer-Giedion syndrome chromosome region (LGCR).

Polymorphisms: SacI detects a two-allele polymorphism:
A1 8.8 kb
A2 11.0 kb

Frequencies: Estimated from 36 chromosomes

A1	0.72
A2	0.28

Not Polymorphic For: AccI, AvaII, AspI, Asp700, BanI, BanII, BamHI, BclI, BglI, BglII, BstNI, BstXI, DraI, EcoRI, EcoRV, HindII, HindIII, HinfI, HgiAI, HpaI, KpnI, MspI, NciI, PstI, PvuII, RsaI, Sau96I, StuI, TaqI, XbaI in a screen of six unrelated Caucasian individuals.

Chromosomal Localization: Mapped to 8q23.2-q24.11 by quantitative Southern blot hybridization of DNA from patients with Langer-Giedion syndrome that have cytogenetically visible deletions.

Mendelian Inheritance: Autosomal codominant segregation was observed in two large Caucasian families.

Probe Availability: Available for collaboration: contact B.Horsthemke.

Other Comments: Insert can be released from the vector by EcoRI digestion.

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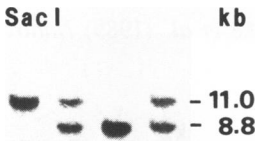
Reference: Lüdecke *et al.* (1989) *Nature* **338**, 348–350.



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Two polymorphisms in the non-coding regions of the BCHE gene

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Source/Description: Exon 1 of the butyrylcholinesterase (BCHE) gene contains a polymorphic site at nt-116 (TGC/TAC). Another polymorphism occurs at nt 1914 (A/G), 189 bases after the stop codon in exon 4. The rarer 1914 G creates an MaeII restriction site (1).

Frequencies: At nt −116, the frequency of G is 0.92 and that of A is 0.08. At nt 1914 the frequency of A is 0.74 and that of G is 0.26. Data are based on ds DNA sequencing of 33 unrelated, phenotypically usual, individuals. Numbers of heterozygous and homozygous individuals agree with the expected numbers from the Hardy-Weinberg equation.

Linkage: The above two polymorphisms appear to be in linkage disequilibrium: the less frequent A at nt −116 always showed the less frequent G at nt 1914, although only one pedigree was examined to establish this coupling. The quantitative K-variant BCHE mutation in exon 4, nt 1615 (GCA→ACA, Ala→Thr[539]), (frequencies of 0.87 and 0.13), is in linkage disequilibrium with the G at nt 1914: 12 informative individuals exhibited this haplotype, and 11 other individuals were heterozygous at both sites. At nt −116, the heterozygous G/A was always either heterozygous or homozygous for the K-variant. χ^2 tests indicate that the less frequent bases at these three sites are all in linkage disequilibrium. The point mutation causing the atypical BChE at Asp70 has been reported to be in linkage disequilibrium with the K-variant (88%) (2).

Chromosomal Localization: Localized to 3q26.2 (P.McAlpine, personal communication).

Mendelian Inheritance: Both polymorphisms were examined in nine key members of a 50-person pedigree. Several other BChE variants (atypical, fluoride-resistant, J-variant, K-variant) were present in this family so that linkage of the two polymorphisms with these established BCHE mutations could be examined.

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References: 1)Arpagaus (1990) *Biochem.* **29**, 124. 2) Bartels,C. *et al.* (1990) In *Proceedings of the 3rd International Meeting on Cholinesterases*. F.Bacou ed. ACS Books, Washington DC. In press.

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