The butyrylcholinesterase gene (BCHE) at 3q26.2 shows two RFLPs

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Source and Description: Exon 1 probe has a 0.8 kb PstI-HindIII insert and exon 3 has a 1.7 kb EcoRI-XbaI insert (Arpagaus *et al.*, 1990).

Polymorphism: The exon 1 probe identifies two alleles on PstI digests at 23.1 kb (C1) and 4.4 kb (C2). The exon 3 probe identifies two alleles on MspI digests at 10.5 kb (D1) and 5.4 kb (D2). To date complete correspondence in phenotypes with both enzyme-probe combinations found.

Frequency:

MspI:	C1: 0.87
	C2: 0.13
PstI:	D1: 0.87
	D2: 0.13

Studied in 19 Caucasian Canadians, 11 females, 8 males.

Not Polymorphic For: Not polymorphic with EcoRI, PvuII, TaqI.

Mendelian Inheritance: Codominant inheritance demonstrated in several two and three generation families.

Probe Availability: From ATCC.

Other Comments: No problems with RFLP analysis under normal stringency.

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Reference: Arpagaus, M. *et al.* (1990) *Biochem.* **29**, 124.

VNTR polymorphism in the hepatic lipase gene (LIPC)

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Source/Description: A CT repeat is located in intron 8 of the human hepatic lipase gene at the 3' end of an Alu sequence (1). Two oligonucleotides flanking the CT repeat and including the poly-T tail of the Alu sequence, HLIP1 and HLIP2, were used to selectively amplify the sequence from genomic DNA by the polymerase chain reaction (PCR).

Primer Sequences:

HLIP1 = ATGTGATGTCAGTGCTGCCAGTCCA

HLIP2 = ACTGACATTGAAAGATACGACCAC

Frequency: This was estimated in 38 unrelated Caucasian English individuals:

Allele (nt)	Frequency
174	0.013
171	0.184
169	0.013
167	0.645
165	0.145

The heterozygosity index was 0.53 and polymorphic information content 0.42.

Chromosomal Localization: Hepatic lipase gene has been assigned to chromosome 15q21-q23 (2).

Mendelian Inheritance: Codominant segregation was observed in 5 families with 28 meioses.

Other Comments: The PCR reaction was performed on genomic DNA (1) using end labelled oligo HLIP1 and unlabelled HLIP2. DNA was denatured at 94°C for 5 minutes, followed by 30 one minute cycles of denaturing at 94°C, annealing at 48°C, and extension at 72°C, with a final extension step of 9 minutes. The PCR buffer was made up to 14 mmol MgCl₂, and 0.25 µl of PerfectMatch™ (Stratagene) was added to the 25 µl reaction volume. The PCR products were sized on a 6% denaturing polyacrylamide gel by simultaneously running the dideoxy chain termination reaction products of phage M13mp18 (Sequenase 2.0). The odd size of allele 1 may be due to polymorphism in the poly-T region of the Alu-sequence.

References: 1) Ameis, D. *et al.* (1990) *JBC* **265**, 6552-6555.
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