

## CA repeat polymorphism at the ASS locus

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**Introduction:** We report a polymorphism at a CA repeat in the human argininosuccinate synthetase gene (ASS) which has been mapped to 9q34 (Carritt and Povey, 1979; Su *et al.*, 1984).

**Source/Description:** Using universal sequencing primer, ASSG1, a EcoRI-PvuII fragment from the 14th intron of ASS (Northrup *et al.* 1989) in pUC8, was found to contain a dinucleotide repeat: (CA)<sub>16</sub>. Flanking primers were designed giving a 132 bp PCR product.

**TG primer:** 5'GGGAGCTATAAAAATGACCA3'

**CA primer:** 5'TTAGGTCCGAAAACACAAAG3'

**Polymorphism:** Ten alleles were detected, designated A1 (121 bp), A2 (131 bp), A3 (133 bp), A4 (135 bp), A5 (137 bp), A6 (139 bp), A7 (141 bp), A8 (143 bp), A9 (145 bp) and A10 (147 bp).

**Frequency:** From 62 unrelated individuals: A1 = 0.01; A2 = 0.15; A3 = 0.26; A4 = 0.07; A5 = 0.14; A6 = 0.02; A7 = 0.13; A8 = 0.12; A9 = 0.08; A10 = 0.02. Of these individuals 64% were heterozygous.

**Mendelian Inheritance:** Autosomal co-dominant segregation was observed in 23 families with 203 individuals.

**Other Comments:** PCR was carried out using a Perkin-Elmer Cetus (Norwalk, CT) DNA Thermal Cycler. Each 10 µl reaction contained 200 ng genomic DNA, 30 ng of each primer, 0.6 U Taq DNA polymerase, 100 µM of each of dATP, dGTP and TTP, 0.4 µM dCTP, 25 nCi [alpha 32P]dCTP at 3000 Ci/mmol. Amplification conditions were 98°C/10 mins, then 35 cycles of 94°C/15 secs; 51°C/30 secs; 72°C/30 secs with a final elongation step of 72°C/10 mins. A 1.5 µl aliquot was electrophoresed on a 6% polyacrylamide sequencing gel which was fixed, dried and exposed to Fuji RX film. The EMBL Data Library has assigned accession number X53535 to the sequences referred to herein.

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## A TaqI RFLP in the human *ret* proto-oncogene

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**Source/Description:** A cDNA clone of *ret* proto-oncogene (pN6) was isolated from a neuroblastoma cell line, Nagai (1). A 562 bp Sall-BamHI fragment subcloned into Bluescript pKS-M13+ (pN6SB) was used as a probe.

**Polymorphism:** TaqI identifies a two allele polymorphism with bands at 2.8 + 1.7 kb (A1) and 2.6 + 1.8 kb (A2). Invariant bands at 0.8 kb and 0.7 kb are also present (Fig. 1).

**Frequency:** Studied in 46 unrelated Japanese individuals:

A1: 0.43

A2: 0.57

**Not Polymorphic For:** EcoRI, BamHI.

**Chromosomal Localization:** Human *ret* proto-oncogene has been mapped to chromosome 10q11.2 (2).

**Mendelian Inheritance:** Co-dominant segregation shown in one informative family with 5 individuals (Fig. 2).

**Probe Availability:** Contact M.Nagao.

**Other Comments:** This RFLP was observed under normal stringencies.

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**References:** 1) Tahira,T. *et al.* (1990) *Oncogene* **5**, 97-102. 2) Ishizaka,Y. *et al.* (1989) *Oncogene* **4**, 1519-1521.

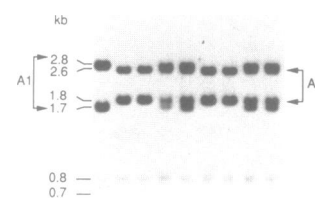


Fig. 1

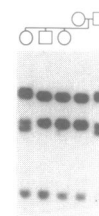


Fig. 2

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