Two additional RFLPs at the D4S10 locus, useful for Huntington's disease (HD)-family studies

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SOURCE AND DESCRIPTION OF THE CLONES:
The original phage clone G8 (Gusella et al. 1983) was used to screen a total human cosmid library and a single 42 kb G8 positive cosmid (C5.5) isolated H5.52 and F5.53 are the two end-clones of this cosmid. H5.52 contains two unique inserts, a 2.4 kb EcoRI and a 1.2 kb EcoRI/BglII, in the cosmid vector c2RB (Bates and Swift 1983). F5.53 contains an 2.8 kb EcoRI/HindIII insert in the same vector.

POLYMORPHISMS:
H5.52 reveals a two allele MspI RFLP with allele sizes of 2.8 and 3.6 kb. F5.53 reveals a two allele SinI RFLP with allele sizes of 3.7 and 3.0 kb, also detects two constant bands of 1.5 kb and 1.0 kb.

FREQUENCY:
Studied in 60 unrelated chromosomes from Dutch individuals.
H5.52 2.8 kb allele (A1) 0.60 3.5 kb allele (A2) 0.40
F5.53 3.7 kb allele (A1) 0.65 3.0 kb allele (A2) 0.35

CHROMOSOMAL LOCALIZATION: The total cosmid DNA was used to localize the D4S10 locus at 4p16.3 by in situ hybridisation (Landegent et al. 1986).

MENDELIAN INHERITANCE:
Both RFLPs segregate with the known G8 (D4S10) RFLPs in normal as well as in HD-families.

PROBE AVAILABILITY:
Available for collaborators.

OTHER COMMENTS:
Thusfar, we have found no evidence for linkage disequilibrium with the earlier described RFLPs at the D4S10 locus (Gusella et al. 1983). The overall heterozygosity at this locus is now increased to over 95%.

REFERENCES:

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