Isolation and mapping of a polymorphic DNA sequence (pEFZ38) on chromosome 2 [D2S60]


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SOURCE/DESCRIPTION: A 4.0 kb Rsal fragment from cosmid EFZ38 was subcloned into the HincII site of pUC18.

POLYMORPHISM: Rsal identifies a 2 allele polymorphism with a band at 4.0 kb (R1) or 3.4 kb (R2) without a constant band. TaqI also identifies a 2 allele polymorphism with a band at 2.0 kb or 1.7 kb, but this is not yet examined by three generation families.

FREQUENCY: Estimated from 94 unrelated Caucasians.

<table>
<thead>
<tr>
<th>Allele</th>
<th>Frequency</th>
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<tbody>
<tr>
<td>R1</td>
<td>0.72</td>
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<tr>
<td>R2</td>
<td>0.28</td>
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NOT POLYMORPHIC FOR: MspI

CHROMOSOMAL LOCALIZATION: pEFZ38 has been assigned to chromosome 2 by multipoint linkage analysis(1) with loci (ACP, D2S1) known to span this region(2).

MENDELIAN INHERITANCE: Co-dominant segregation was observed in 47 three generation families.

PROBE AVAILABILITY: Freely available (contact Y.N.) or will be available from ATCC.

OTHER COMMENTS: RFLPs were observed after competitive hybridization with total human DNA.

REFERENCES: