Single base polymorphism at -511 in the human interleukin-1β gene (IL1β)

Section of Molecular Medicine, Department of Medicine and Pharmacology, University of Sheffield, Royal Hallamshire Hospital, Sheffield S10 2JF, UK

Source/Description: We have identified, by direct sequencing of the human IL-1β 5'-region (—702 to +603), a single-base variation at position —511. PCR Primers:
5'-TGGCATTGATCTGGTTCATC-3'
5'-GTCTTAGGAATCTTCCACTT-3'
Polymorphism: Transition 'C to T', the 'C allele completing an Aval site.
Frequency/Alleles: Estimated from 39 unrelated Caucasians. Allele A1: 0.59. Allele A2: 0.41. Observed frequency of heterozygotes is 0.51.
Chromosomal Location: The human IL-1β gene (1, 2) has been assigned to 2q13—21 (3).
Mendelian Inheritance: Co-dominant segregation has been demonstrated in 4 families of 42 individuals.
Comments: Population screening was performed by PCR amplification of —702 to —398 followed by Aval digestion and silver-staining of PAGE gels. Aval restriction of genomic DNA is not informative.
Acknowledgements: We are grateful to R.Mountford and H.N.Hughes for their kind gift of genomic DNA from families.


Dinucleotide repeat polymorphism at the RBP3 locus in chromosome band 10q11.2

L.Papi1,2*, A.Tunnacliffe1 and B.A.J.Ponder1
1CRC Human Cancer Genetics Research Group, Department of Pathology, University of Cambridge, UK and 2Department of Clinical Physiopathology, Section of Medical Genetics, University of Florence, Italy

Source and Description: A polymorphic CA repeat was isolated and characterised from cosmid cTBIRBP9. From the same cosmid a TaqI RFLP was previously characterised (1).
Primer Sequences:
Primer LLO: 5'TAATCTTTTCTCAAAAGGTG-3'
Primer FO: 5'-TAGATGCTGACAGTGGGAAG-3'
Polymorphism and Frequency: Five alleles were detected in 56 chromosomes of unrelated individuals. Heterozygosity 0.79.

<table>
<thead>
<tr>
<th>Allele</th>
<th>Allele size</th>
<th>Allele frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>A1</td>
<td>387</td>
<td>0.12</td>
</tr>
<tr>
<td>A2</td>
<td>381</td>
<td>0.23</td>
</tr>
<tr>
<td>A3</td>
<td>375</td>
<td>0.25</td>
</tr>
<tr>
<td>A4</td>
<td>361</td>
<td>0.18</td>
</tr>
<tr>
<td>A5</td>
<td>355</td>
<td>0.21</td>
</tr>
</tbody>
</table>
Chromosomal Localisation: RBP3 has been assigned to chromosome 10q11.2 (2).
Mendelian Inheritance: Co-dominant segregation was observed in 6 Multiple Endocrine Neoplasia type 2 (MEN 2) pedigrees.
PCR Conditions: PCR amplification was performed in a total volume of 30 µl containing 20—200 ng of genomic DNA, 30 pmol of each primer, 10 mM Tris-HCl (pH 8.3), 1.5 mM MgCl2, 50 mM HCl, 100 µg/ml gelatin, 250 µM dNTPs. Thirty-five cycles of denaturation at 95°C for 1 min, annealing at 58°C for 45 s and elongation at 72°C for 1 min, with a final extension step at 72°C for 5 min, were performed. PCR products were analysed on a 5% acrylamide denaturing gel.
Comments: Clinical relevance; presymptomatic screening of MEN 2 syndrome.

Acknowledgements: This work was supported by grants from the Cancer Research Campaign and the EEC (L.P.).


*To whom correspondence should be addressed at present address: Dip. Fisiopatologia Clinica, Sezione di Genetica Medica, Viale Pieraccini 6, Firenze, Italy