Gene Section
Mini Review

IGK (Immunoglobulin Kappa)

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Identity

HGNC (Hugo): IGK@

Location: 2p12

For complete Figure, see the international ImMunoGeneTics information system; Copyright 1995-2003 IMGT.

Note

The human IGK locus is located on chromosome 2 on the short arm, at band 2p12. The orientation of the locus has been determined by the analysis of translocations, involving the IGK locus, in leukemia and lymphoma.

DNA/RNA

Description

The human IGK locus at 2p12 spans 1820 kb. It consists of 76 IGKV genes belonging to 7 subgroups, 5 IGKJ segments, and a unique IGKC gene.

The 76 IGKV genes are organized in two clusters separated by 800 kb. The IGKV distal cluster (the most 5' from IGKC and in the most centromeric position) spans 400 kb and comprises 36 genes. The IGKV proximal cluster (in 3' of the locus, closer to IGKC, and in the most telomeric position) spans 600 kb and comprises 40 genes.

The potential genomic IGK repertoire comprises 31 to 35 functional IGKV genes belonging to 5 subgroups, the 5 IGKJ segments, and the unique IGKC gene.

One rare IGKV haplotype has been described which contains only the proximal cluster. This haplotype comprises the 40 proximal IGKV genes belonging to 7 subgroups, of which 17 to 19 are functional and belong to 5 subgroups.

Twenty-eight IGKV orphans have been identified and sequenced: 3 on the short arm of chromosome 2 but outside of the main IGK locus, 13 on the long arm of chromosome 2, 6 on chromosome 22, one on chromosome 1, one on chromosome 15, and 4 outside of chromosome 2.

If both the proximal and distal IGKV clusters are present, the total number of human IGK genes per haploid genome is 82 (110 genes, if the orphans are included) of which 37-41 are functional. If only the proximal IGKV cluster is present, the total number of genes per haploid genome is 46 (74 genes, if the orphans are included) of which 23-25 genes are functional.
**Protein**

**Description**

Proteins encoded by the IGK locus are the immunoglobulin kappa chains. They result from the recombination (or rearrangement), at the DNA level, of two genes: IGKV and IGKJ, with deletion of the intermediary DNA to create a rearranged IGKV-J gene. The rearranged IGKV-J gene is transcribed with the IGKC gene and translated into an immunoglobulin kappa chain.

Translation of the variable germline genes involved in the IGKV-J rearrangements are available at IMGT Repertoire. Compared to the germline genes, the rearranged variable genes will acquire somatic mutations during the B cell differentiation in the lymph nodes, which will considerably increase their diversity. These somatic mutations can be analysed using IMGT/V-QUEST tool.

**Implicated in**

Translocations which frequently result from errors of the recombinase enzyme complexe (RAG1, RAG2, etc.), which is responsible of the Immunoglobulin and T cell receptor V-J and V-D-J rearrangements. IGKV or IGKJ recombination signals or isolated heptamer are observed at the breakpoints.

<table>
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<tr>
<th>Translocation</th>
<th>Note</th>
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<tr>
<td>t(2;3)(p12;q27)</td>
<td>c-Immunoglobulin genes IgK at 2p12, in normal cells: PAC 1117G4 - Courtesy Mariano Rocchi.</td>
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<tr>
<td>t(2;8)(p12;q24)</td>
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**References**

Hieter PA, Max EE, Seidman JG, Maizel JV Jr, Leder P. Cloned human and mouse kappa immunoglobulin constant and J region genes conserve homology in functional segments. Cell. 1980 Nov;22(1 Pt 1):197-207


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