

Gene Section

Mini Review

IGK@ (Immunoglobulin Kappa)

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Identity

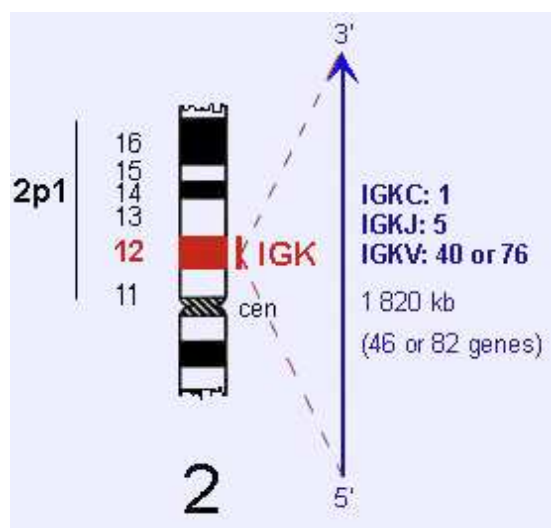
Other names: IGK (Immunoglobulin Kappa)

HGNC (Hugo): IGK@

Location: 2p12

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.



For complete Figure, see: chromosome 2, IMGT (The International ImMunoGeneTics information system ®) © Copyright 1995-2003 IMGT, IMGT is a CNRS trademark.

DNA/RNA

Description

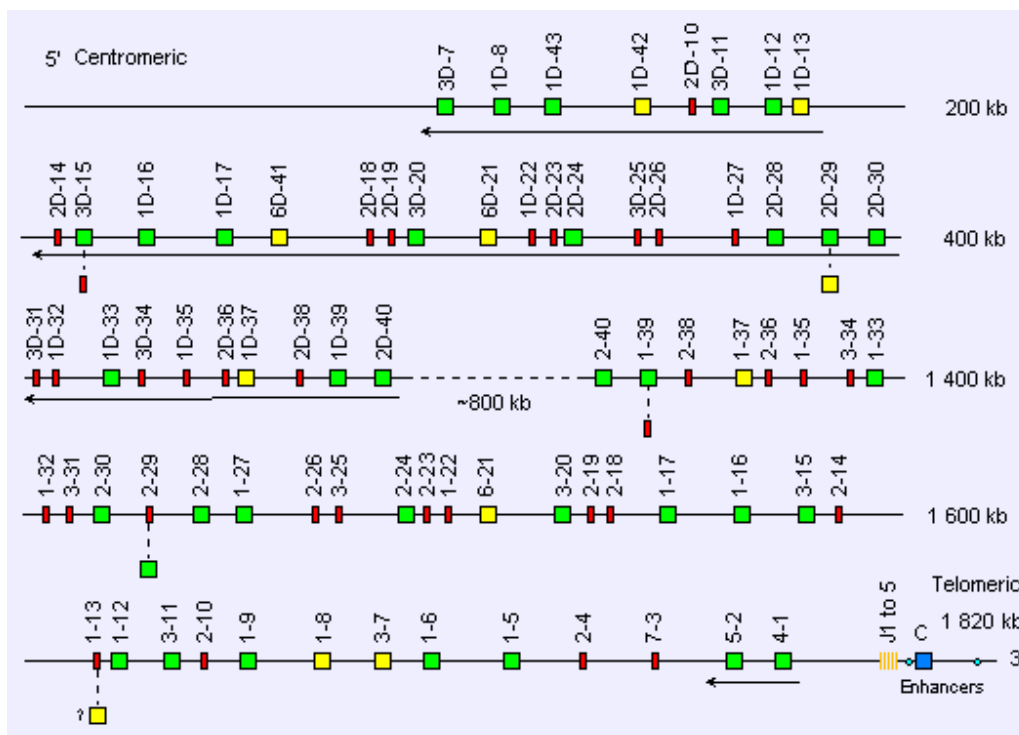
The human IGK locus at 2p12 spans 1820 kb. It consists of 76 IGKV genes belonging to 7 subgroups, 5 IGKJ segments, and a unique IGKC gene.

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.



IGK

V-GENE: Green box: Functional; Yellow box: Open reading frame; Red: Pseudogene.

J-GENE: Grey: Functional.

C-GENE: Blue: Functional.

For complete Figure, see: locus IGK, IMGT (The International ImMunoGeneTics information system ©) © Copyright 1995-2003 IMGT, IMGT is a CNRS trademark.

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.

List of the human IGK genes.

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 Protein displays. 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.

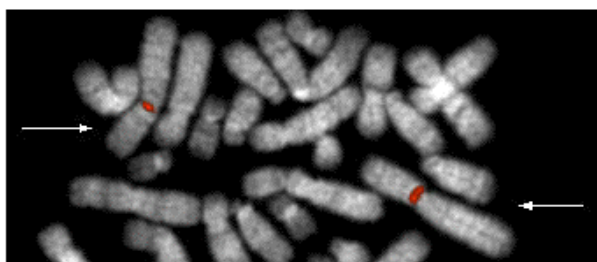
Mutations

Note

Mutations which correspond to allelic polymorphisms of the functional germline IGKV, IGKJ and IGKC genes are described in the IMGT database: (IMGT Repertoire> Alignments of alleles).

Implicated in

Translocations which frequently result from errors of the recombinase enzyme complex (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.



c-Immunoglobulin genes IgK at 2p12, in normal cells: PAC 1117G4 - Courtesy Mariano Rocchi, Resources for Molecular Cytogenetics.

t(2;3)(p12;q27); involve BCL6 in 3q27

Disease

B-cell non-Hodgkin lymphomas (NHL), mainly diffuse large cell lymphoma; adult aggressive lymphoma.

Prognosis

Controversial.

t(2;8)(p12;q24); involve C-MYC in 8q24

Disease

B-cell acute lymphoblastic leukemia (ALL3) and non-Hodgkin lymphomas (NHL), especially in the Burkitt lymphoma.

Prognosis

The prognosis has evolved with new treatments.

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