

Gene Section

Mini Review

IGH (Immunoglobulin Heavy)

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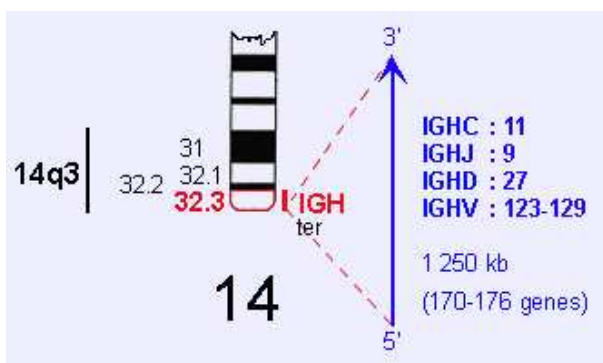
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Identity

HGNC (Hugo): IGH@

Location: 14q32.33



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

Note

The human IGH locus is located on the chromosome 14 at band 14q32.33, at the telomeric extremity of the long arm; the orientation of the locus has been determined by the analysis of translocations, involving the IGH locus, in leukemia and lymphoma.

DNA/RNA

Description

The human IGH locus at 14q32.33 spans 1250 kilobases (kb). It consists of 123 to 129 IGHV genes,

depending from the haplotypes, 27 IGHD segments belonging to 7 subgroups, 9 IGHJ segments, and 11 IGHC genes.

Eighty-two to 88 IGHV genes belong to 7 subgroups, whereas 41 pseudogenes, which are too divergent to be assigned to subgroups, have been assigned to 4 clans. Seven non-mapped IGHV genes have been described as insertion/deletion polymorphism but have not yet been precisely located.

The most 5' IGHV genes occupy a position very close to the chromosome 14q telomere whereas the IGHC genes are in a more centromeric position.

The potential genomic IGH repertoire is more limited since it comprises 38-46 functional IGHV genes belonging to 6 or 7 subgroups depending from the haplotypes 23 IGHD, 6 IGHJ, and 9 IGHC genes.

Thirty-five IGH genes have been found outside the main locus in other chromosomal localizations. These genes designated as orphans cannot contribute to the synthesis of the immunoglobulin chains, even if they have an Open Reading Frame (ORF). 9 IGHV orphans and 10 IGHD orphans have been described on chromosome 15 (15q11.2), and 16 IGHV orphans on chromosome 16 (16p11.2). In addition, one IGHC processed gene, IGHEP2 is localised on chromosome 9 (9p24.2-p24.1)

This is so far the only processed Ig gene described. The total number of human IGH genes per haploid genome is 170 to 176 (206 to 212 genes, if the orphans and the processed gene are included) of which 77 to 84 genes are functional.

each of the functional IGHG, IGHA and IGHE constant genes.

This recombination, accompanied by the deletion of the intermediary DNA, allows the IGHV-D-J initially transcribed with the IGHM, to be now transcribed with a IGHG, IGHA or IGHE gene, and translated into a gamma, alpha or epsilon chain.

Translation of the variable germline genes involved in the IGHV-D-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.

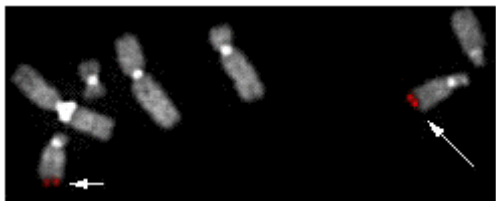
Mutations

Note

Mutations which correspond to allelic polymorphisms of the functional germline IGHV, IGHD, IGHI and IGHC genes are described in the IMGT database.

Implicated in

Translocations which frequently result from errors of the recombinase enzyme complex (RAG1, RAG2, etc.), responsible of the Immunoglobulin and T cell receptor V-J and V-D-J rearrangements, or from errors of the switch enzyme. IGHV, IGHD or IGHI recombination signals or isolated heptamer (first case) or switch sequences (second case) are observed at the breakpoints.



c-Immunoglobulin genes IgH at 14q32.33, in normal cells: PAC 998D24 - Courtesy Mariano Rocchi.

t(3;14)(q27;q32)

t(4;14)(p16;q32)

t(5;14)(q31;q32)

t(8;14)(q11;q32)

t(8;14)(q24;q32)

t(10;14)(q24;q32)

t(11;14)(q13;q32)

t(14;18)(q32;q21)

t(14;19)(q32;q13.1)

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