

# IMGT-ONTOLOGY and IMGT databases, tools and Web resources for immunoinformatics

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The international ImMunoGeneTics information system®  
Coordinator: M.-P. Lefranc, Montpellier, France <http://imgt.cines.fr>



# What is an ontology? and Why?

An **ontology** is the definition of the concepts and of their relations, necessary to share, to reuse and to represent the knowledge, in a domain.



The international ImMunoGeneTics information system®  
Coordinator: M.-P. Lefranc, Montpellier, France <http://imgt.cines.fr>



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# What is an ontology?

An **ontology** is the definition of the concepts...

Example: a gene

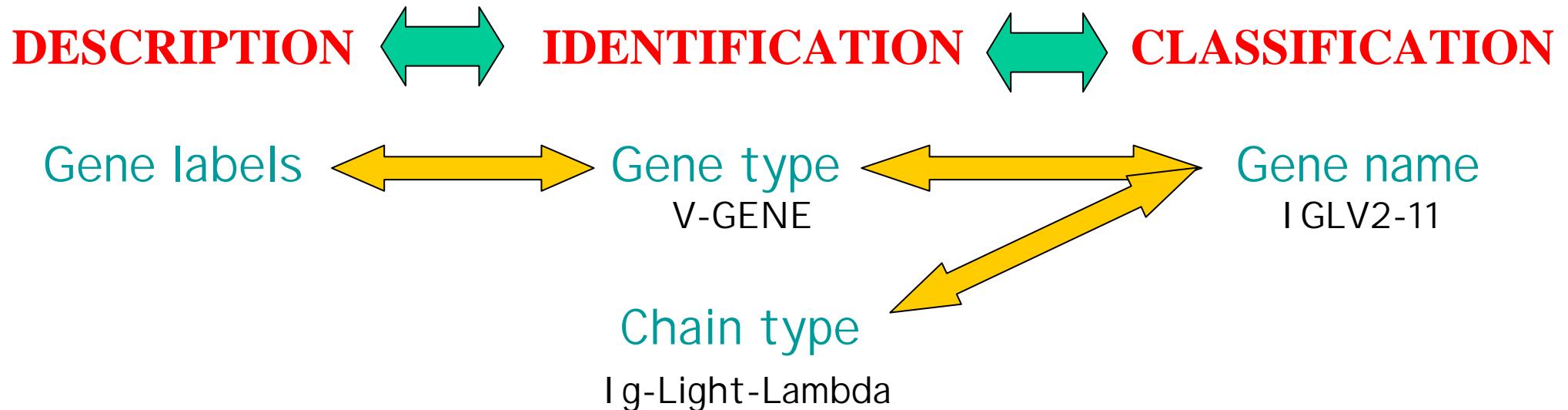
- gene type: concept of **IDENTIFICATION**
- gene name: concept of **CLASSIFICATION**
- gene labels: concept of **DESCRIPTION**



Controlled vocabulary

# What is an ontology?

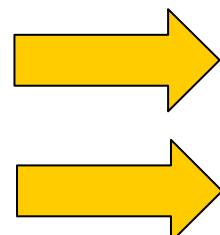
An **ontology** is the definition of the concepts and of their relations...



- In many ontologies, no distinction between « concepts » and « instances »
- In GO (GeneOntology), only 2 types of relations: « is a », « is part of »

# Why an ontology?

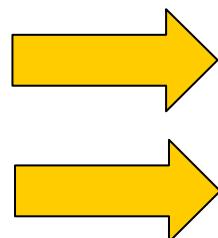
An **ontology** is the definition of the concepts and of their relations, necessary to share, to reuse and to represent the knowledge, in a domain.



Human beings  
Information systems

# Why an ontology?

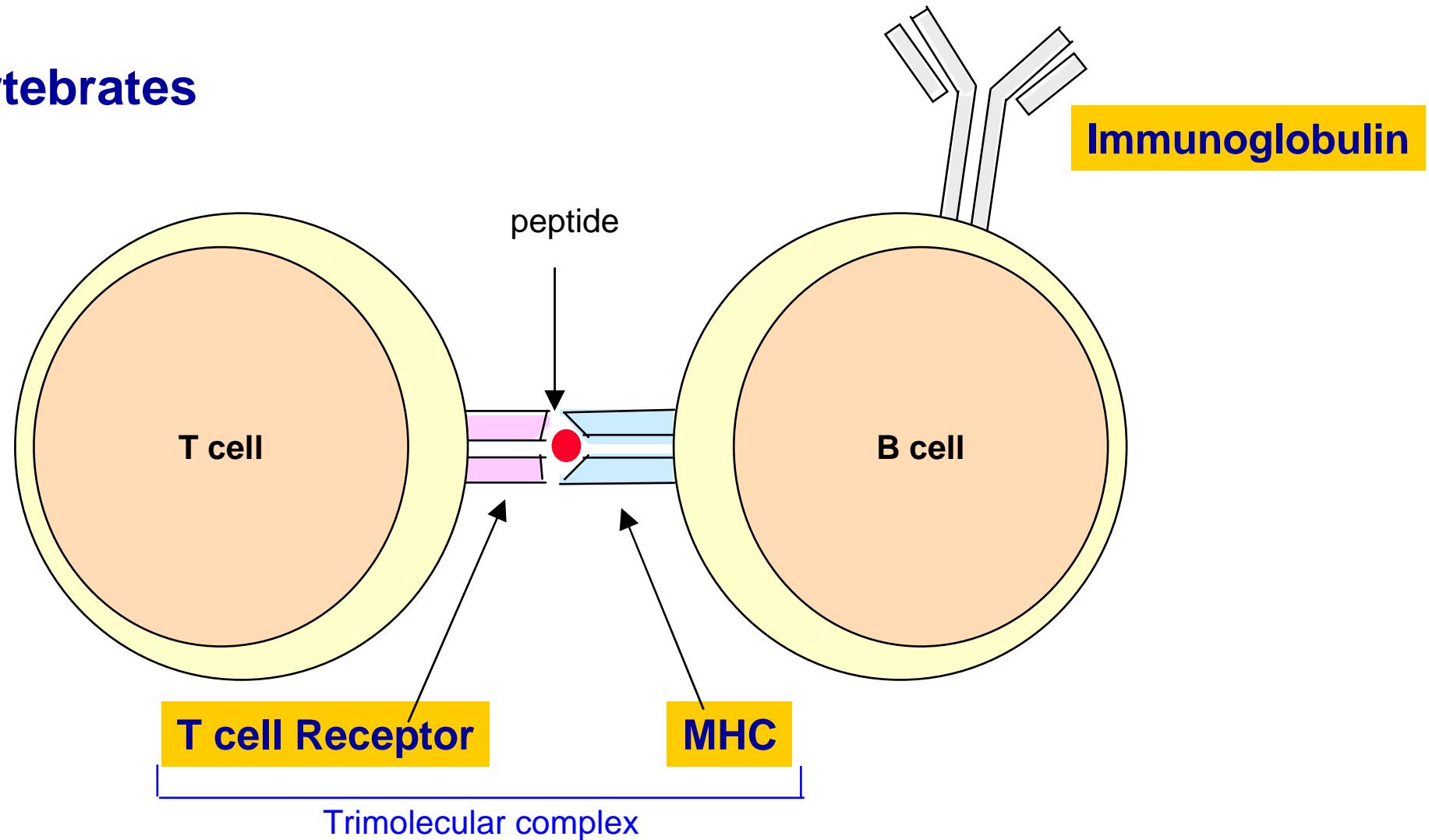
An **ontology** is the definition of the concepts and of their relations, necessary to share, to reuse and to represent the knowledge, in a domain.



Immunology  
Immunoinformatics

# IMGT domain of research: the adaptive immune system

## Vertebrates

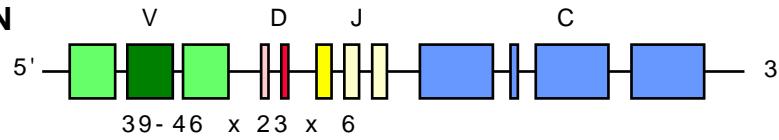


# Immunoglobulin (IG) and T cell receptor (TR) synthesis

150

## FUNCTIONAL IG GENES

HEAVY CHAIN



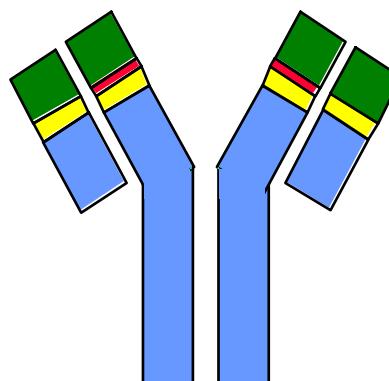
6300 POTENTIAL RECOMBINATIONS



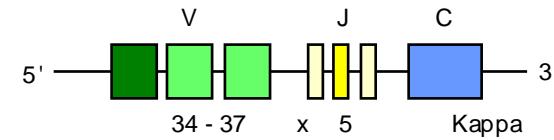
ABOUT  $6.3 \times 10^6$  POSSIBILITIES

$2 \times 10^{12}$

DIFFERENT ANTIBODIES



LIGHT CHAIN



185 + 165 POTENTIAL RECOMBINATIONS



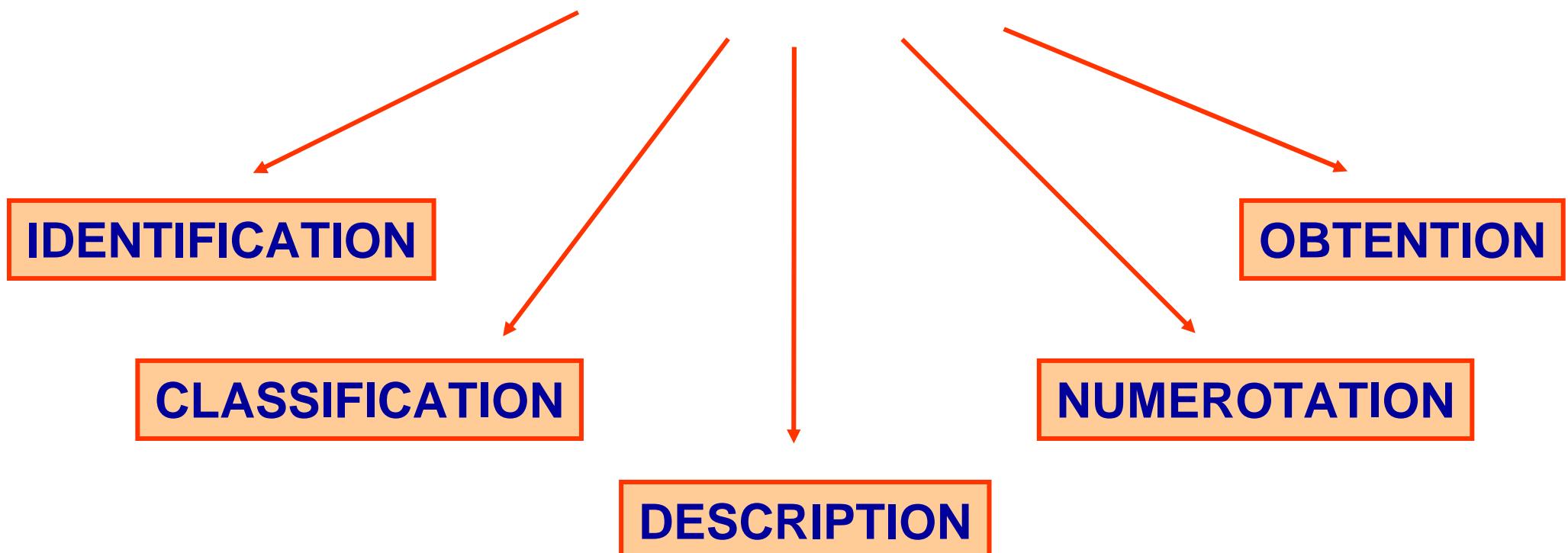
ABOUT  $3.5 \times 10^5$  POSSIBILITIES

$N\text{-DIVERSITY} \times 1000$

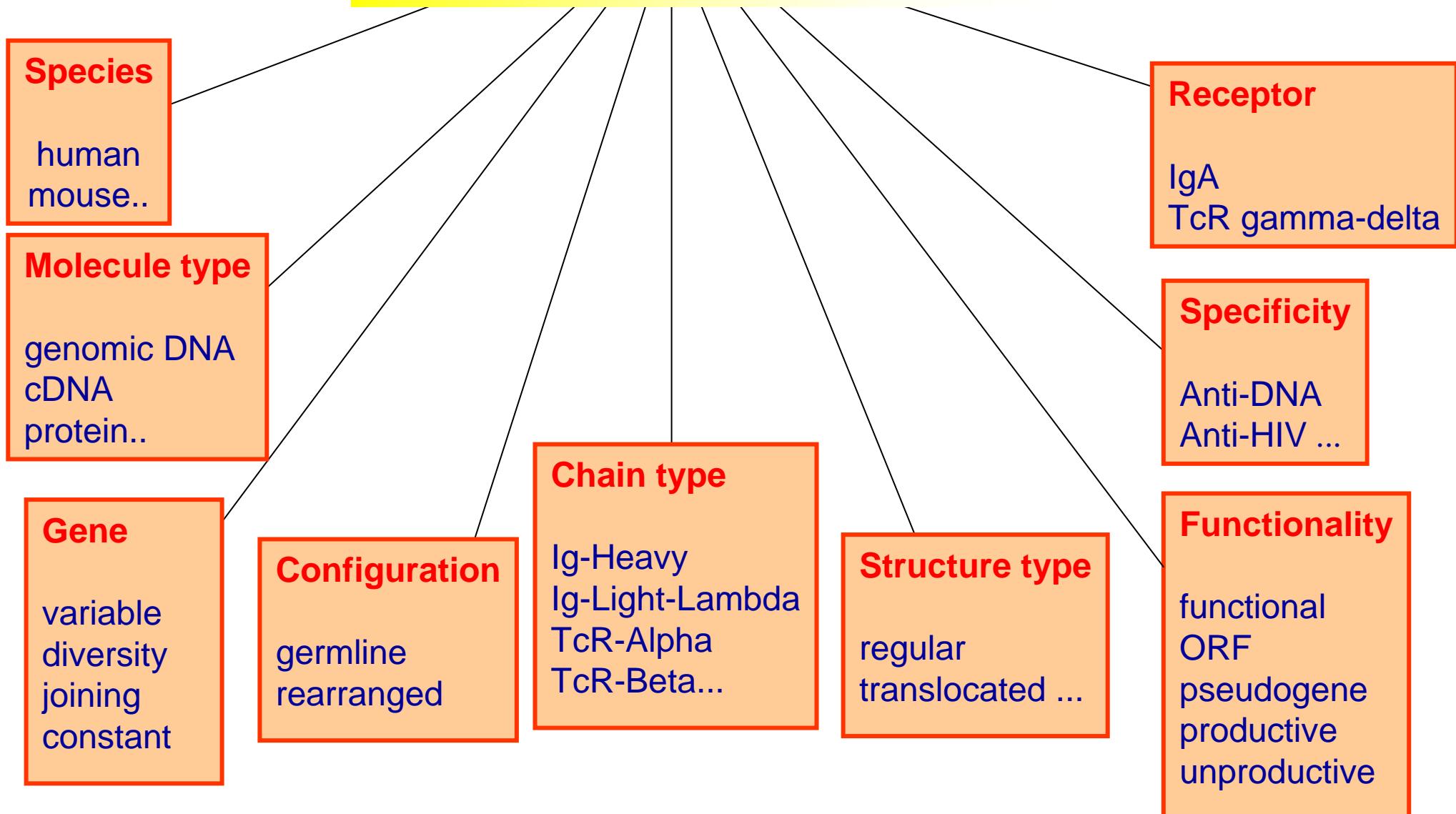
SOMATIC MUTATIONS

## IMGT-ONTOLOGY five main concepts

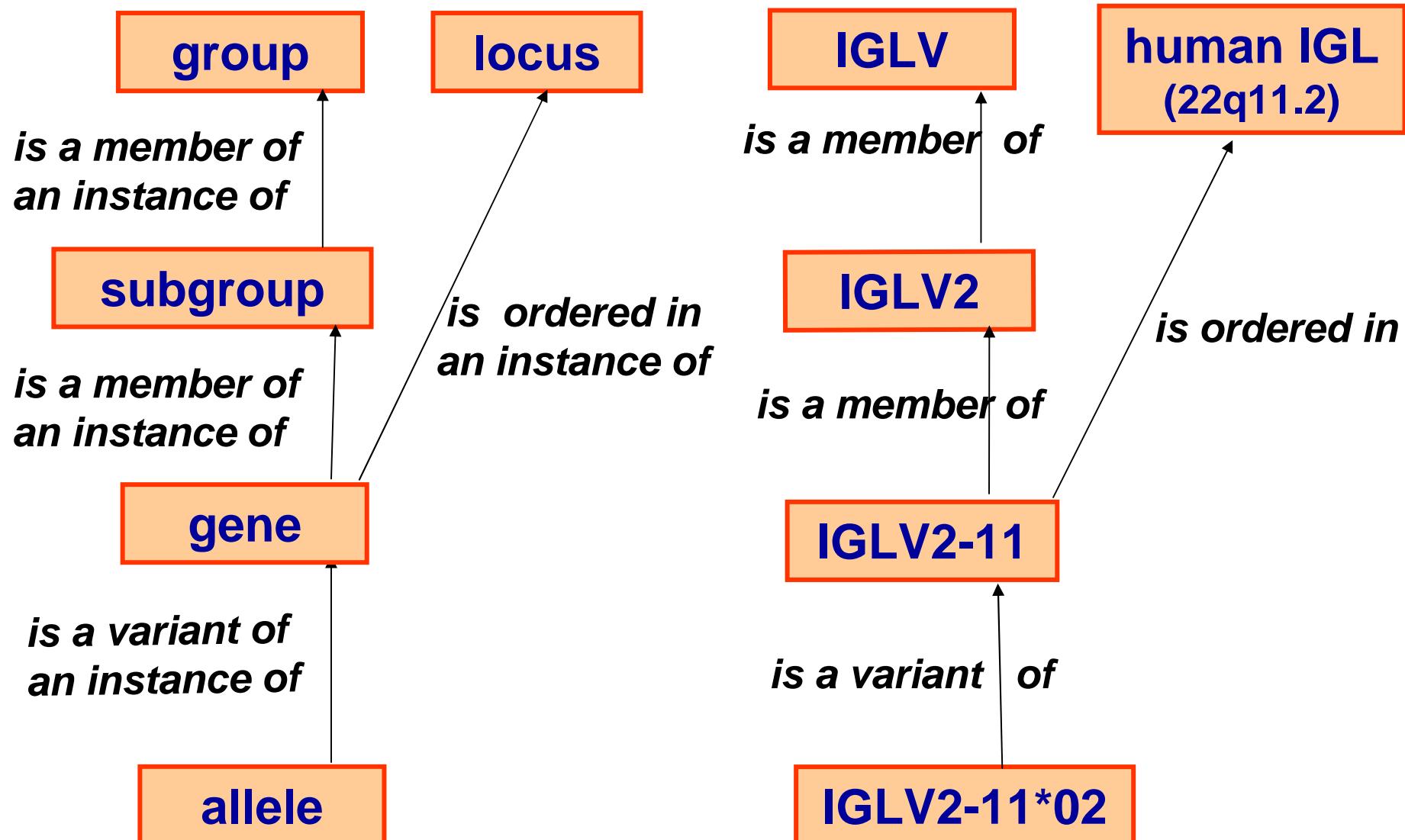
to share, reuse and represent knowledge  
in immunogenetics



# "IDENTIFICATION" concept



# "CLASSIFICATION" concept



« Concepts »

« Instances »



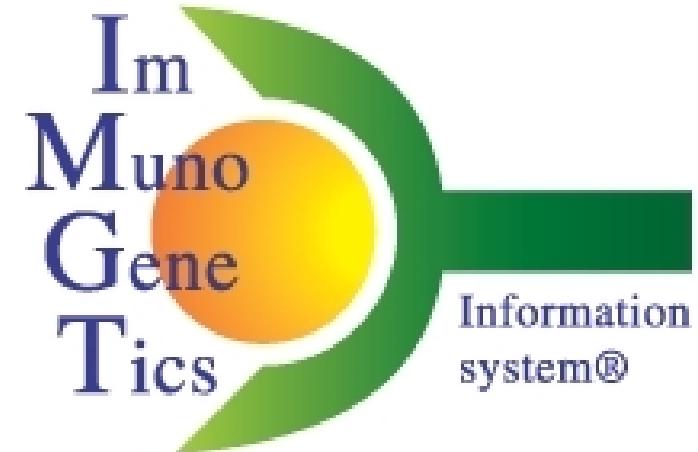
## Locus representation: Human IGL

Human IGL 2

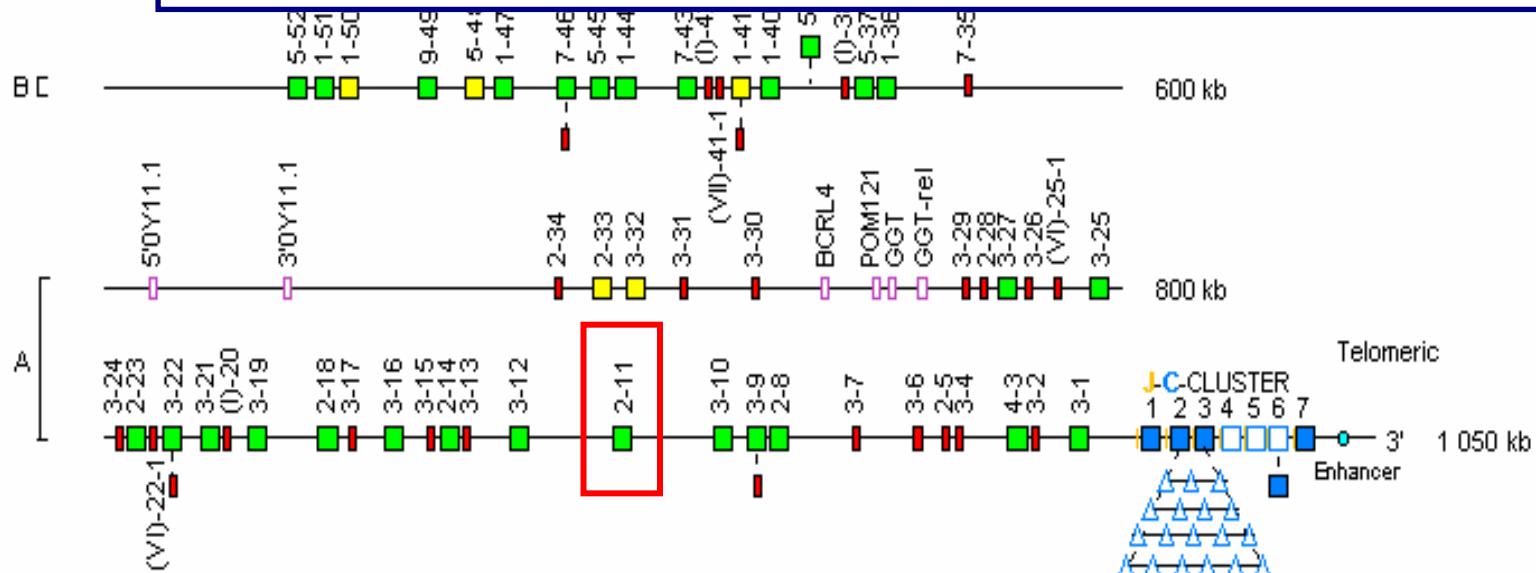
Centromere  
BCR  
5' →  
C  
0-63  
1-62  
+ -

**WELCOME !**  
**to IMGT/GENE-DB**

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INFORMATION SYSTEM®



<http://imgt.cines.fr>



Document : chargé



**LocusLink Report - Netscape**

Eichier Edition Afficher Aller Communicator Aide

**NCBI**

PubMed Entrez BLAST OMIM Map Viewer Taxonomy Structure

Search locusLink Display Best Organism: All

Query:

View ms IGLV2-11 One of 1 Loci Save All Loci

A B C D E F G H I J K L M N O P Q R S T U V W X Y Z

**PUB ACEVIEW MAP VAR GDB**

*Homo sapiens Official Gene Symbol and Name [HGNC]*

**IGLV2-11: immunoglobulin lambda variable 2-11**

**LocusID:** 28816

**Overview** [Submit GeneRIF](#) [?](#)

**Locus Type:** gene, segment

**Alternate Symbols:** V1-3, IGLV211

**Map Information** [?](#)

**Chromosome:** 22 [mv](#)

**Cytogenetic:** 22q11.2 [RefSeq](#)

**NCBI Reference Sequences (RefSeq)** [?](#)

**Category:** REVIEWED

**Genomic:** [NG\\_000002](#) [gb sv](#)

**Category:** NCBI Genome Annotation

**Genomic Contig:** [NT\\_011520](#) [gb sv mv ev mm](#)

**Related Sequences** [?](#)

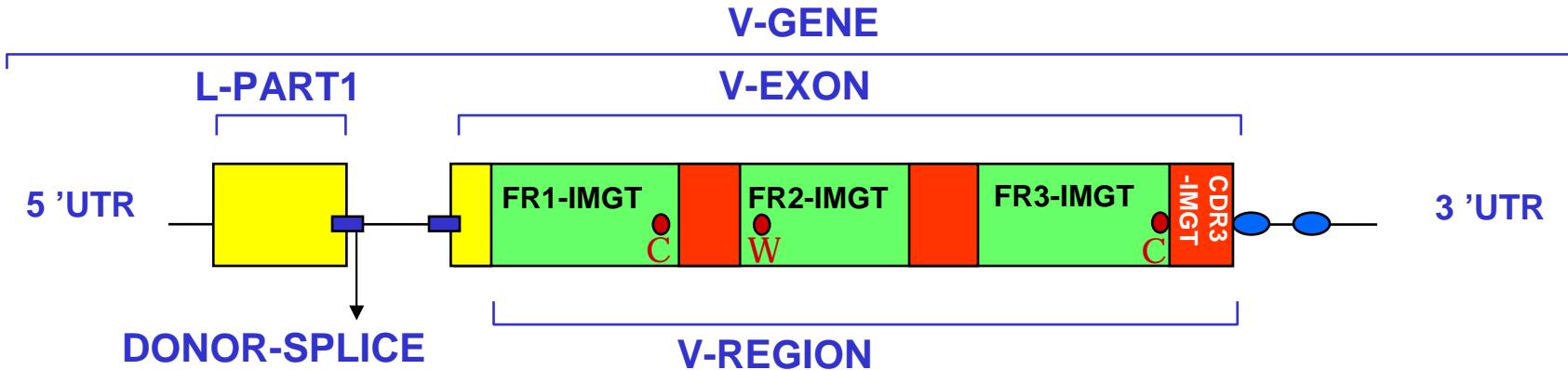
Nucleotide	Type	Protein
<a href="#">D86998</a>	g	<a href="#">BAA19994</a>
<a href="#">Z73657</a>	g	

**Additional Links** [?](#)

- [IMGT Repertoire for individual human immunoglobulin and T cell receptor genes](#)

Document : chargé

# "DESCRIPTION" concept



Label 1	Label 2	Relations entre Labels
V-GENE	V-EXON	[Red bracket spanning V-EXON]
FR3-IMGT	CDR3-IMGT	[Red bracket spanning CDR3-IMGT]
L-PART1	DONOR-SPLICE	[Red bracket spanning DONOR-SPLICE]
V-REGION	FR1-IMGT	[Red bracket spanning FR1-IMGT]
V-REGION	CDR3-IMGT	[Red bracket spanning CDR3-IMGT]

IMGT/LIGM-DB Consultation module v3 - Netscape

Fichier Edition Afficher Ailler Communicator Aide

Signets Adresse : <http://ligm.igh.cnrs.fr:8104/cgi-bin/IMGTlect.jv> Infos connexes

V-GENE <1..297> /partial

Help IMGT Home page IMGT Marie-Paule page New search

Created by Marie-Paule Lefranc (CNRS, Montpellier II University, France)  
<http://imgt.cines.fr>

IMGT/LIGM-DB ON LINE, HERE YOU ARE !

Five types of search are available : select one by clicking on the button

Catalogue accession number, mnemonic, definition, creation date, length, [annotation level](#)

FR1-IMGT 103..153  
/AA\_IMGT="39 to 55"  
/translation="VSWYQQHPGKAPKLMIY"

FR2-IMGT 109..111  
154..162  
/AA\_IMGT="56 to 58"  
/translation="DVS"

FR3-IMGT 163..270  
/AA\_IMGT="66 to 104, AA 73, 81, 82 missing"  
/translation="KRPSGVPDFSGSKSGNTASLTISGLQAEDEADYYC"

CONSERVED-TRP 268..270  
271..297  
/AA\_IMGT="105 to 113"  
/translation="CSYAGSYTF"

2nd-CYS

CDR1-IMGT

CDR2-IMGT

CDR3-IMGT

XX

SQ Sequence 297 BP; 60 A; 93 C; 71 G; 73 T; 0 other;  
cagtcgtcccc tggactcgacc tcggctcaatg tccgggtctc ctggacatgc agtcaccatc  
tccttacacta caaccacccaa tttatattttat ctttataact atatctccctt ctaccaacaa

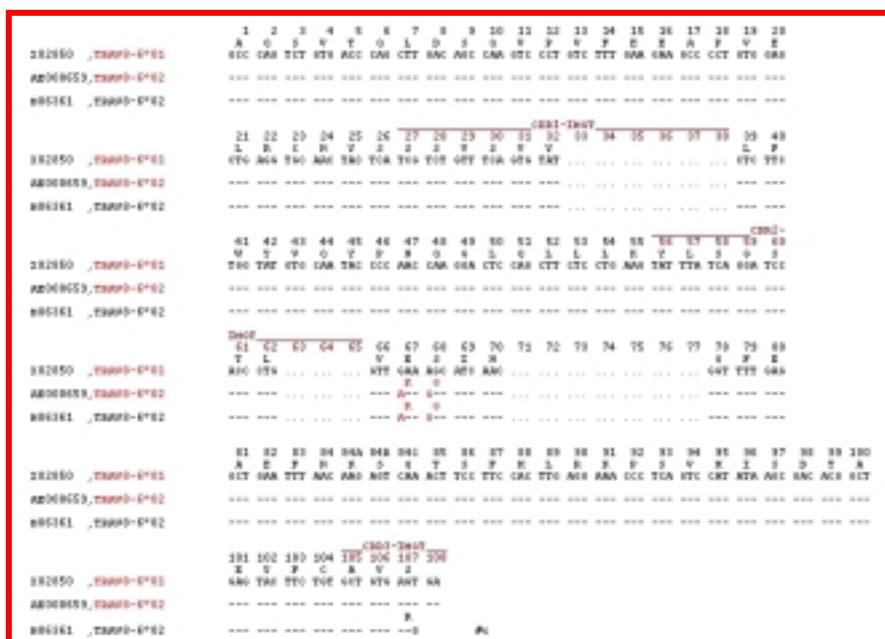
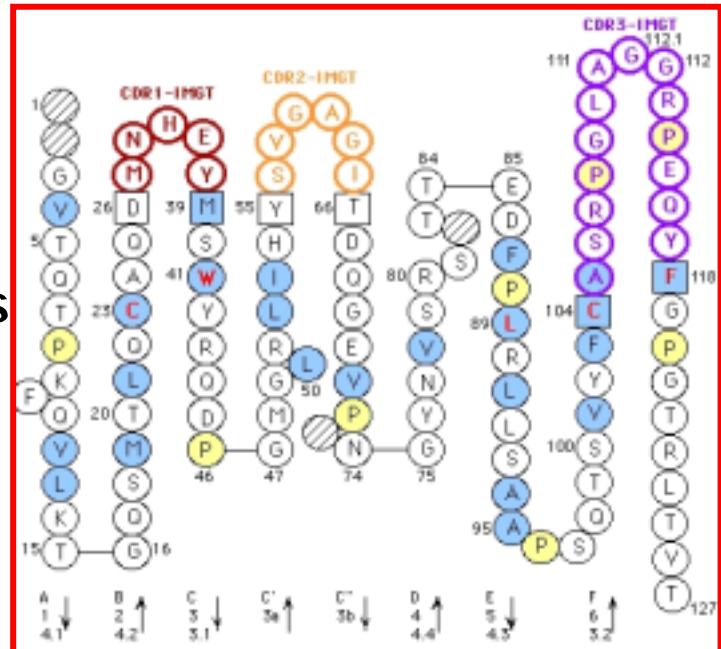
60 120

Information system®



# "NUMEROTATION" concept

Collier  
de Perles

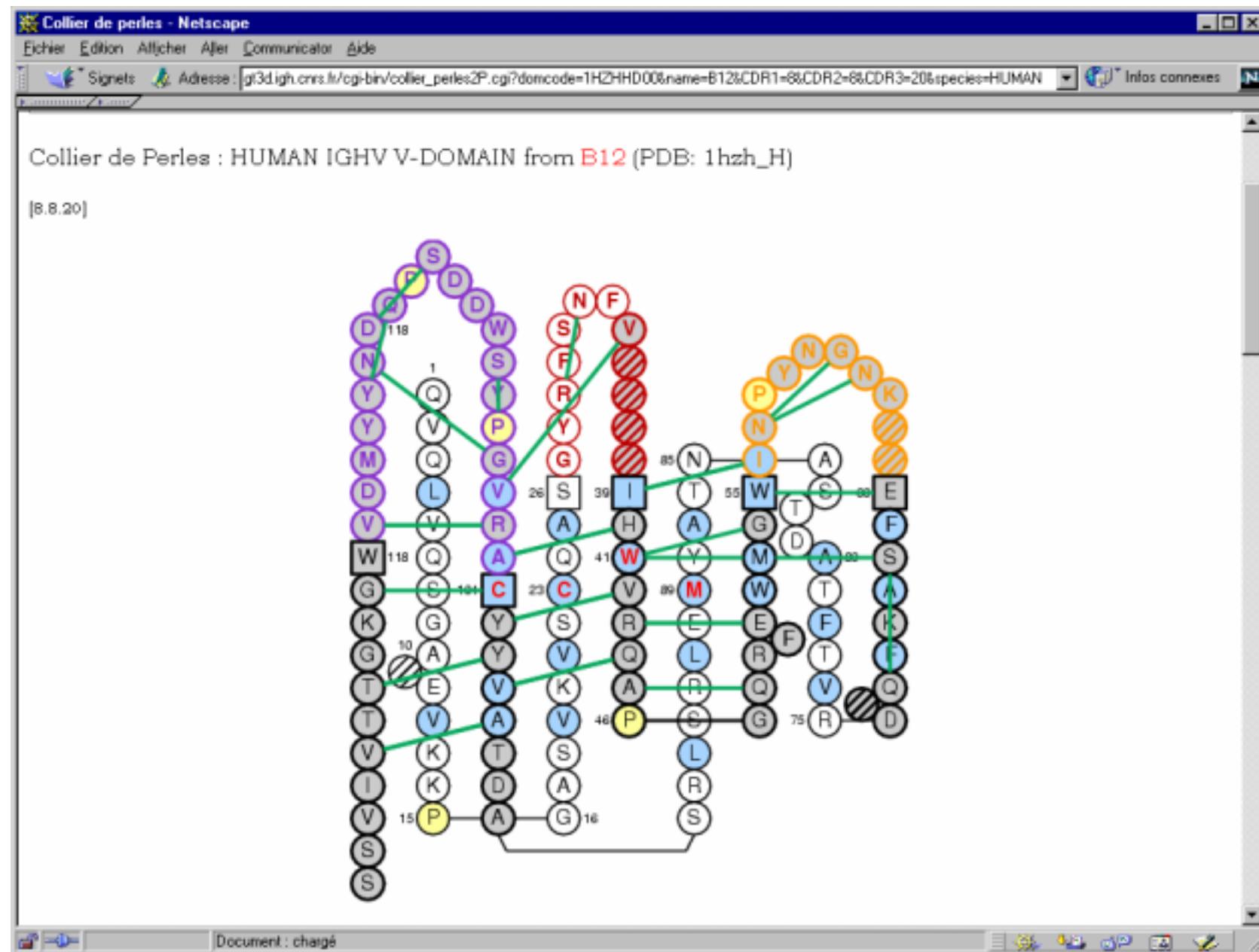


Alignment  
of alleles

Protein  
Display

TRAV gene	FR1-IMGT (1-26)	CDR1-IMGT (27-38)	FR2-IMGT (39-55)	CDR2-IMGT (56-65)	FR3-IMGT (66-104)	CDR3-IMGT (105-115)	
	1 10 20 30		40 50	60	70 80 84ABC 90	100 110	
	.....	.....	.....	.....	.....	.....	
AE000658, TRAV1-1	GQSLEQ. PSEVTAVEGAIVQINCTYQ	TSGFYG. ....	LSWYQQHDGGAPTFLSY	NALDG. ....	LEETG. ....	RFSSFLSRSDSYGYLLLQELQMKSASAYFC	AVR. ....
AE000658, TRAV1-2	GQNIDQ. PTETMATEGAIVQINCTYQ	TSGFNG. ....	LFWYQHAGEAPTFLSY	NVLDG. ....	LEEKG. ....	RFSSFLRSRKGSYSYLLKELQMKSASAYLC	AVR. ....
AE000658, TRAV2	KDQVFQ. PSTVASSEGAVVEIFCNHS	VSNAYN. ....	FFWYLHFPGCAPRLLVK	GSK. ....	PSQQG. ....	RYNMTIYER. FSSSLLILQVREADAAAVYYC	AVE. ....
AE000658, TRAV3	AQSVAQPEDQVNVAEGNPLTVKCTYS	VSGNPY. ....	LFWIVQYPNRLQFLLK	YITGDNL. ....	VKGSY. ....	GFEAEFNKSQTSFHLKKPSALVSDSALYFC	AVRD. ....
AE000658, TRAV4	LAKTTQ. PISMDSYEGQEVNITCSHN	NIATNDY. ....	ITWYQQPSQGPFRFIQ	GYKT. ....	KVTNE. ....	VASFNLPIADRKSSLTLSPRVLSDLTAVYYC	LVGD. ....
AE000659, TRAV5	GEDVEQS. LFLSVREGDSSVINTCYT	DSSSTY. ....	LYWYKQEPGAGLQLLKK	IIFSNMD. ....	MKQDQ. ....	RLTVLLNKKDKHLSLRIAQTGDSAIYLC	ALD. ....
AE000659, TRAV6	SQKIEQNSEALNIQEGKTATLTCNTY	NYSPAY. ....	LQWYRQDPGRGPVFLLL	IRENEK. ....	EKRKE. ....	RLKVTIFDTILKQSLFHITASQPADSATYLC	ALD. ....
AE000659, TRAV7	ENQVEHSPHFLGPQGDVASMCTYS	VSRFNN. ....	LQWYRQMTGMGPKHLLS	MYSAGY. ....	EKQKG. ....	RLNATLKK. NGSSLYITAVQPEDSATYFC	AVD. ....
AE000659, TRAV8-1	AQSVSQHNHHVILSEAASLELGONYS	YGGTVN. ....	LFWYVQYPGQHLQLLLK	YFGSDPL. ....	VKGIK. ....	GFEAEFKSFNLRKPSVQWSDTAEYFC	AVN. ....
AE000659, TRAV8-2	AQSVTQLDSHVSSEGTPVLLRCNYS	SSYSPS. ....	LFWYVQHPNKGLQLLLK	YTSAAVL. ....	VKGIN. ....	GFEAEFKKSETSFSFLTKPSAHMSDAAEYFC	VVS. ....
AE000659, TRAV8-3	AQSVTQPDIIHTVSEGASLELRCNYS	YGATPY. ....	LFWYVQSPGQQLQLLLK	YFGSDTL. ....	VQGIK. ....	GFEAEFKRSQSSFLRKPSVHWSDAAEYFC	AVG. ....
AE000659, TRAV8-4	AQSVTQLGSHVSVSEGALVLLRCNYS	SSVPPY. ....	LFWYVQYPNQGLQLLLK	YTSAAVL. ....	VKGIN. ....	GFEAEFKKSETSFSFLTKPSAHMSDAAEYFC	AVS. ....
X02850, TRAV8-6	AQSVTQLDSQVPVFEEAPVELRCNYS	SSVSVY. ....	LFWYVQYPNQGLQLLLK	YLSGSTL. ....	VESIN. ....	GFEAEFNKSQTSFHLRKPSVHISDTAEYFC	AVS. ....
AE000660, TRAV8-7	TQSVTQLDGHITVSEEAPLELKCNYS	YSGVPS. ....	LFWYVQYSSQSLQLLLK	DLTEATQ. ....	VKGIR. ....	GFEAEFKKSETSFSFLRKPSVHSDAAEYFC	AVGDR. ....
AF000659 TRAV0-1	GDWVWQTEGQWLPSEGDSLTYWCSVE	TTDVPS. ....	LFWYVQVPGEGPQDPLHLK	AMKAND. ....	KGRMK. ....	GFEAFMYRKFTTSFHLKFDNSWQESDLSAVYFC	ALS. ....

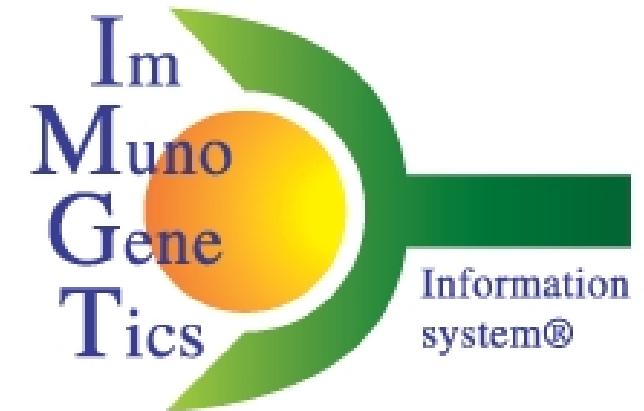
# IMGT Collier de Perles



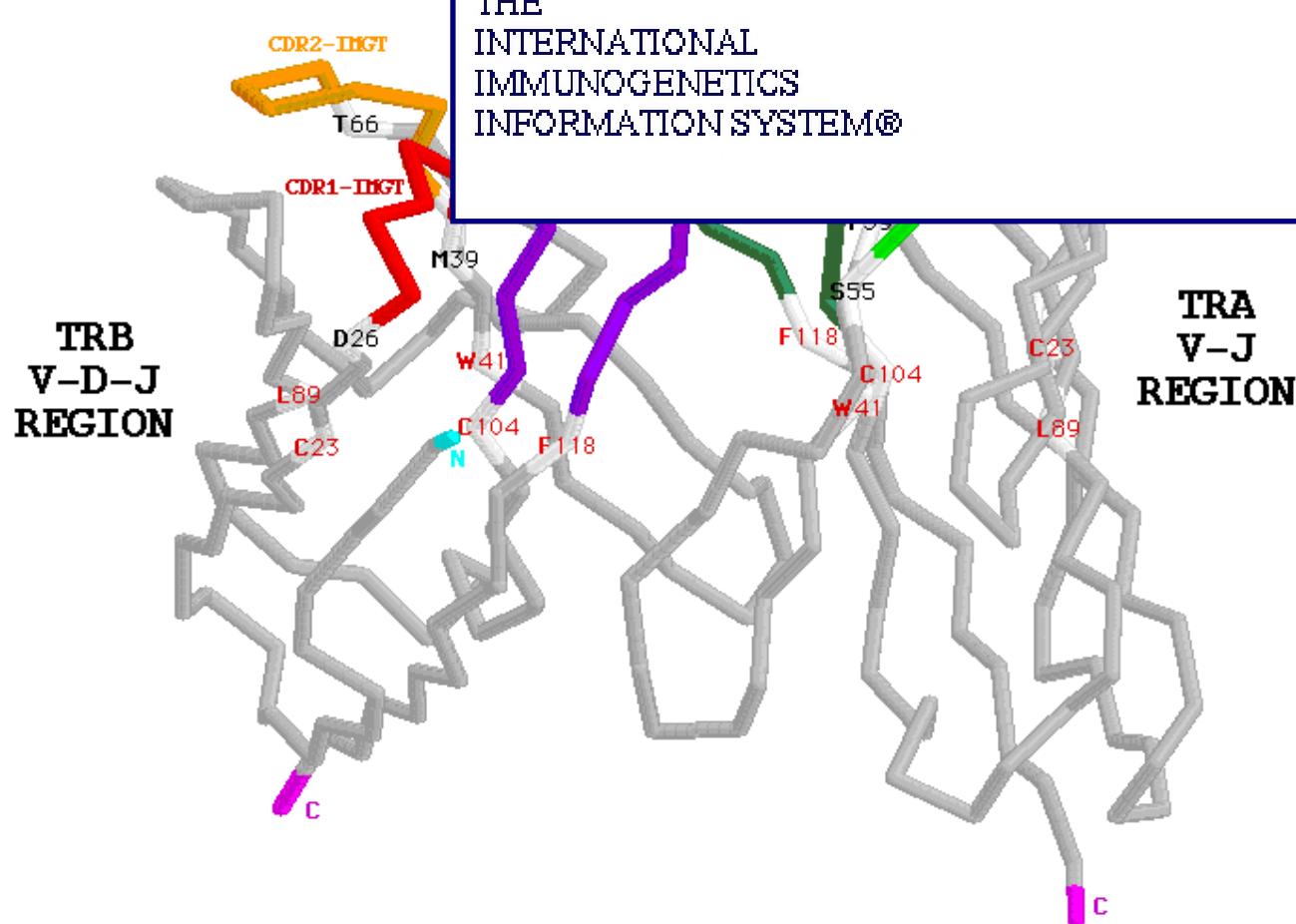
# V-DOMAIN 3D representation (TR A6, 1ao7)

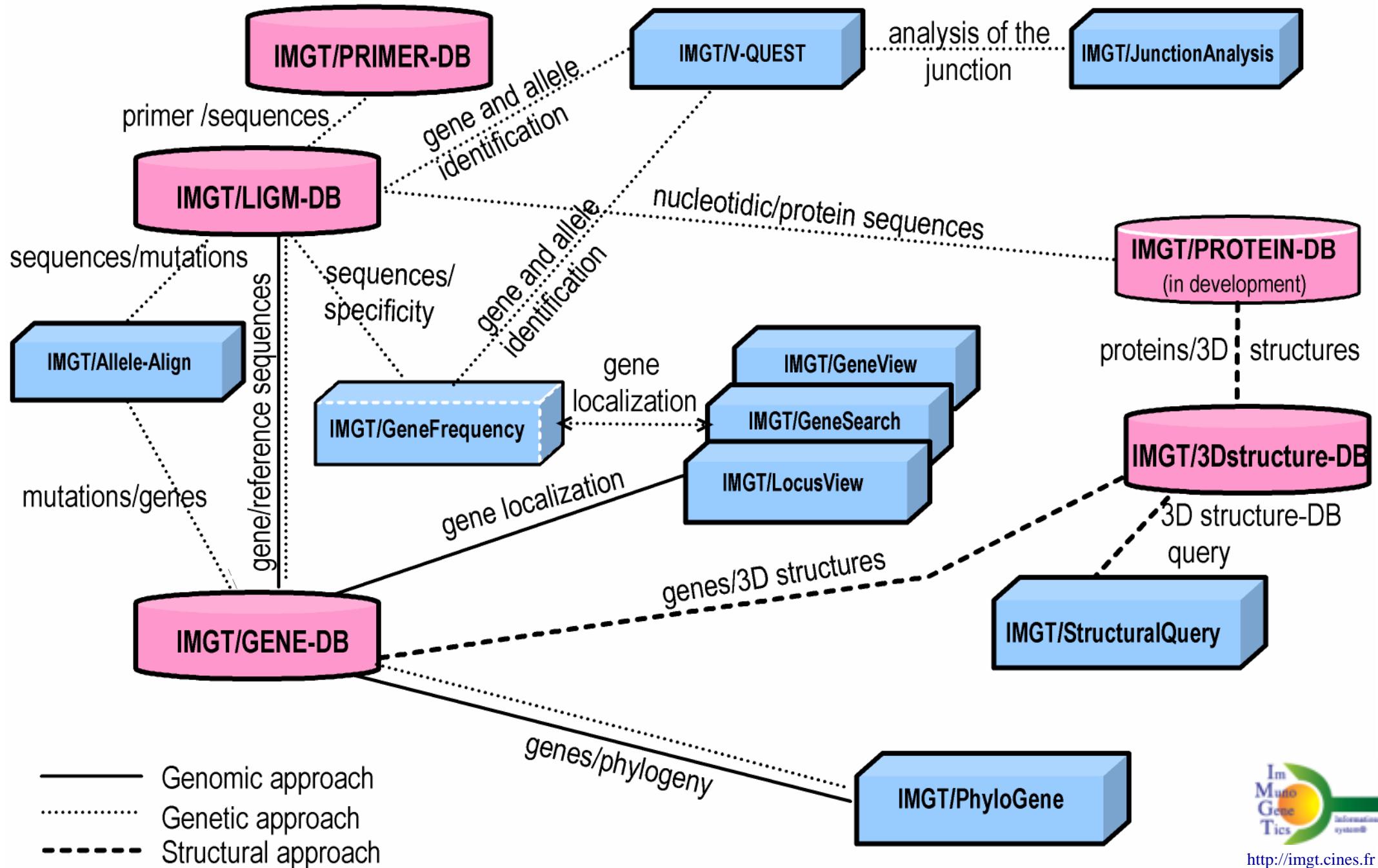


WELCOME !  
to IMGT/3Dstructure-DB



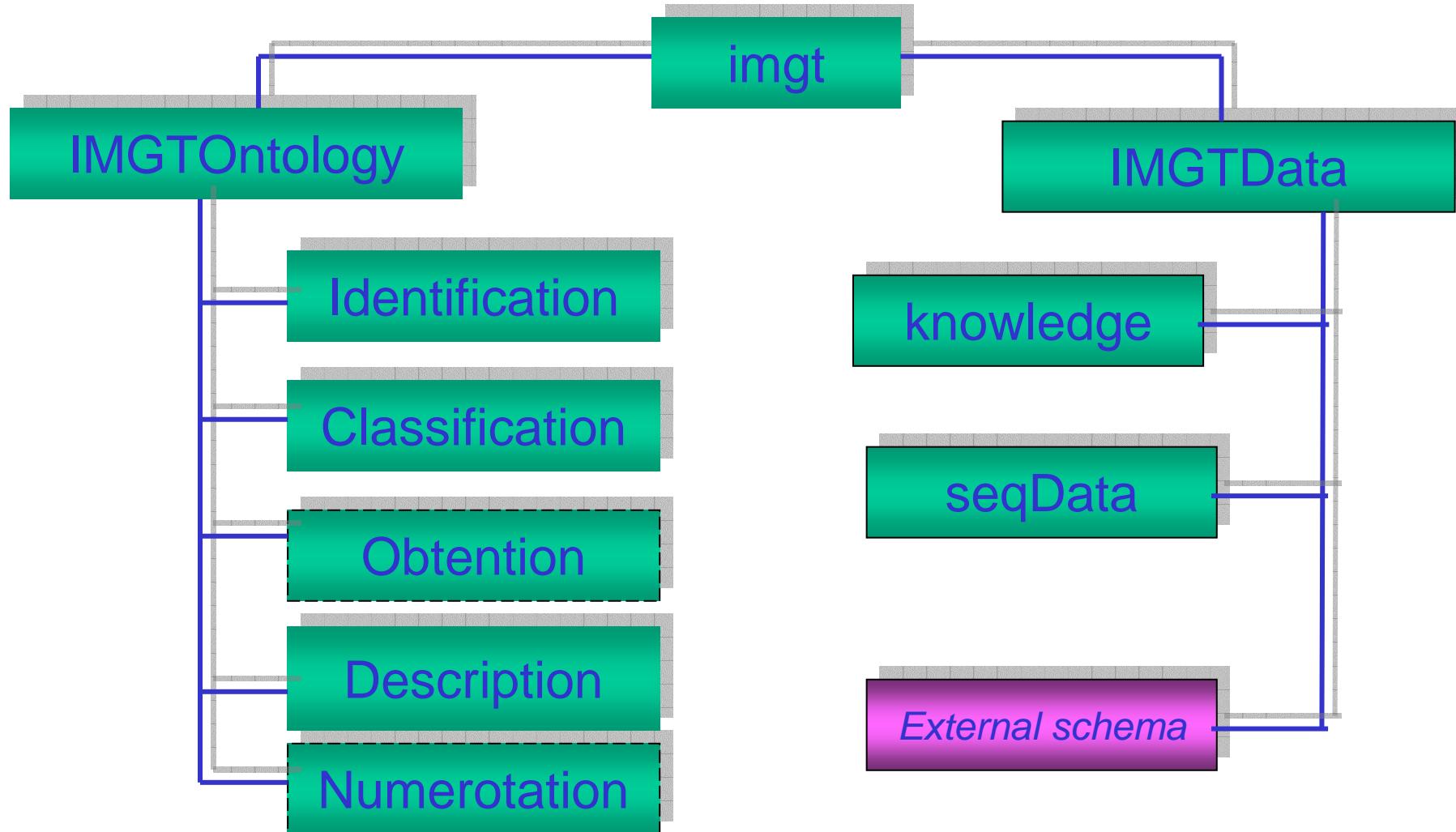
<http://imgt.cines.fr>



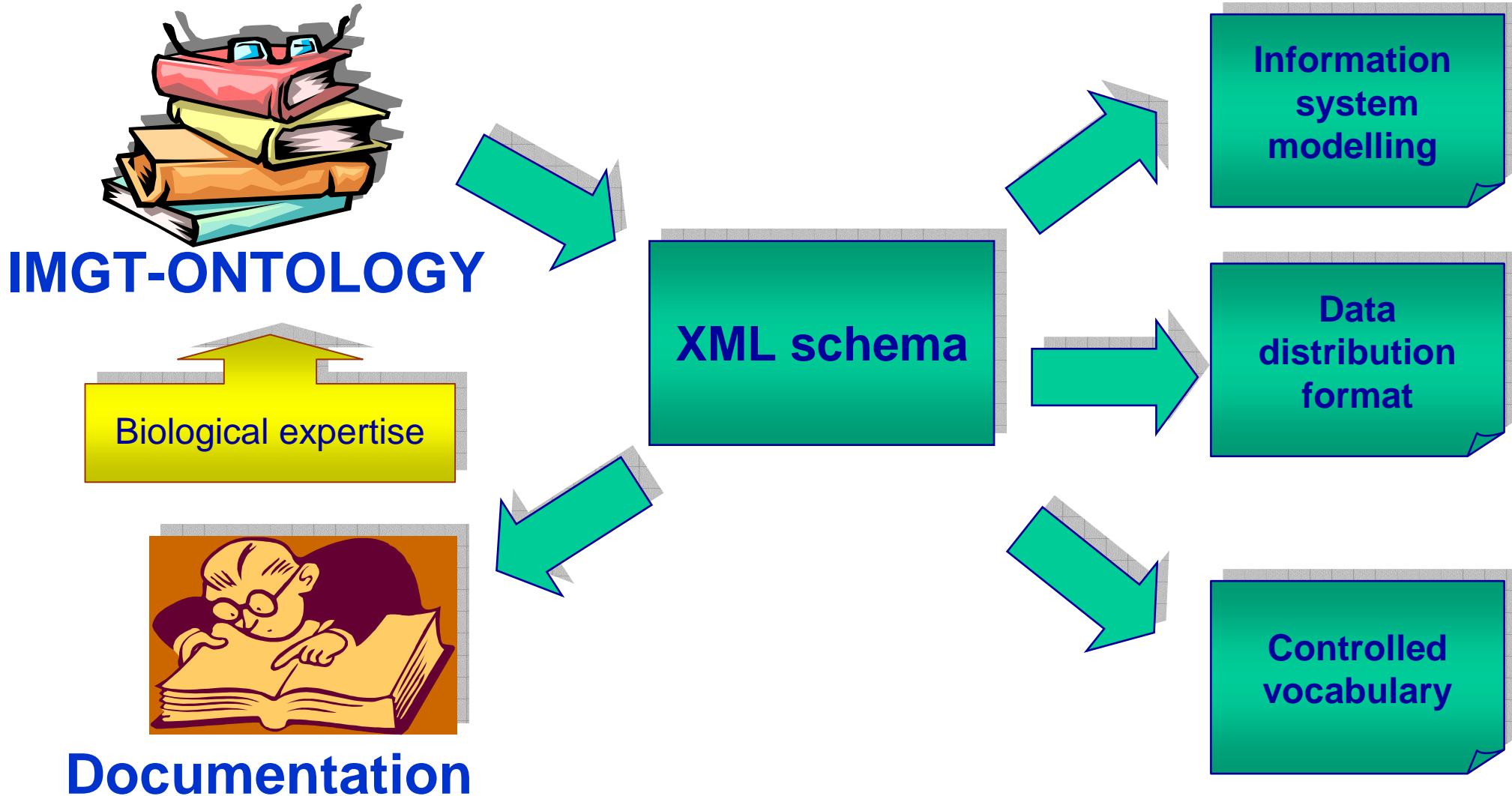


<http://imgt.cines.fr>

# IMGT-ML schema

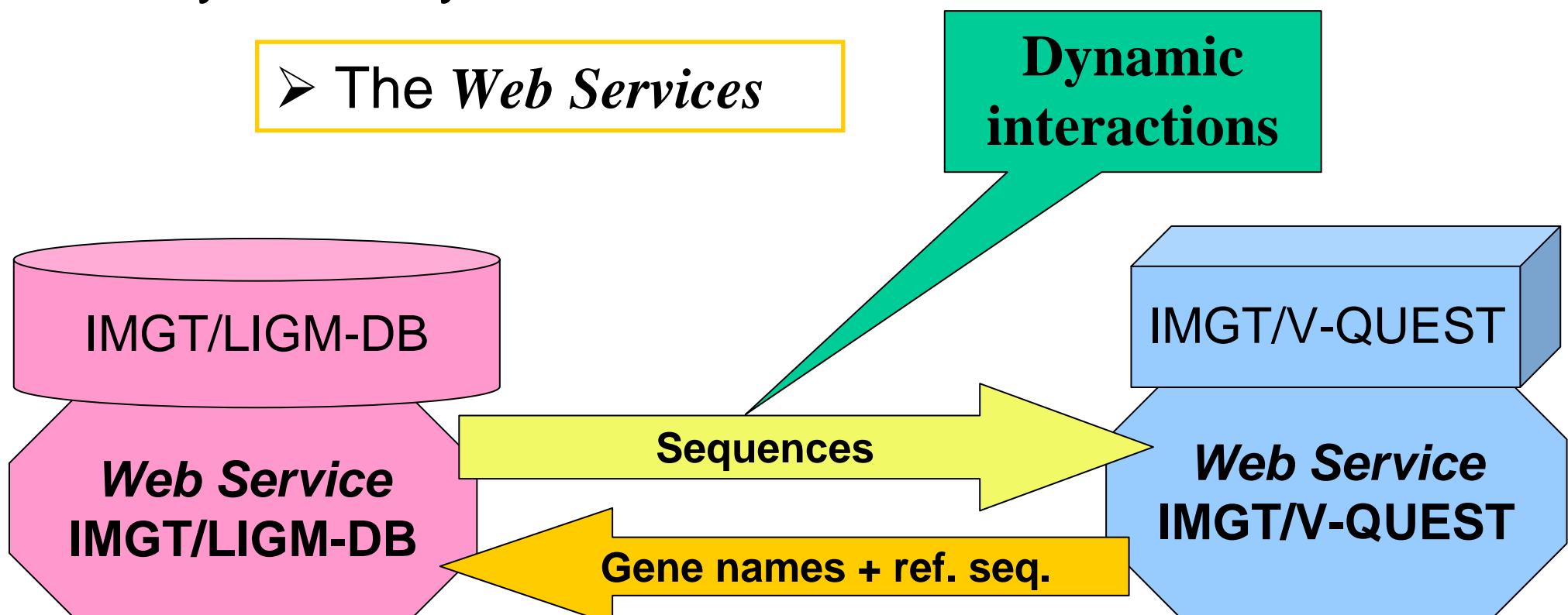


# IMGT-ML architecture



# Informatic answers to the biological problems

- Use IMGT-ONTOLOGY (and IMGT-ML)
- Allow IMGT components to dynamically interact



# Example of IMGT/V-QUEST results

## Alignment for V-GENE

AF402940  
X62109 IGHV1-3\*01  
X62107 IGHV1-3\*02  
M99637 IGHV1-8\*01  
L06612 IGHV1-46\*03  
X92343 IGHV1-46\*01

score GTGCAGCTGCTCGAGCAGTCTGGGGCT \_\_\_\_\_ GAGGTGAGCAAGCCTGGGGCTCAGTAAAGGTTCCCTGCA  
1146 CA.GTC.A...T.T.....AG.....G.....  
1110 CA.GTT.A...G.T.....AG.....G.....  
957 CA.GT..A...G.T.....AG.....G....C.....  
948 CA.GT..A...G.T.....AG.....G.....  
948 CA.GT..A...G.T.....AG.....G.....

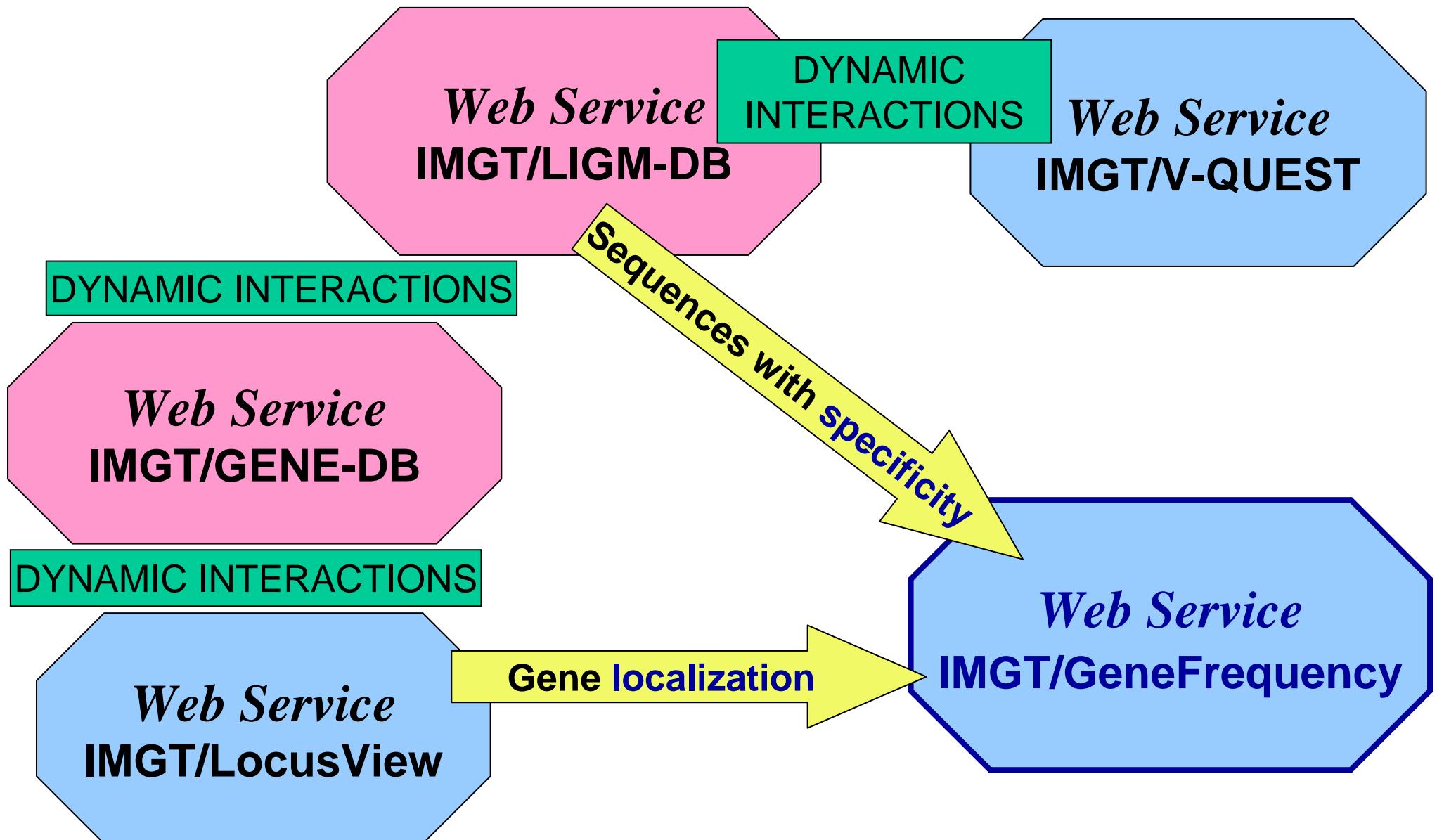
## Alignment for J-GENE

AF402940  
J00256 IGHJ3\*01  
X86355 IGHJ6\*02  
X86355 IGHJ3\*02

score CTTCACGGGGCGGGACGCTTGACGTCTGGGGCCAAGGGACCACGGTCACCGTCTCCTCA  
181 .....T....T..T.....A.T.....T...G  
179 T.A.TACTACTACT...G.A.....  
172 .....T....T..TA.....A.T.....T...G



# Diagram of collaboration: Analyse de repertoires

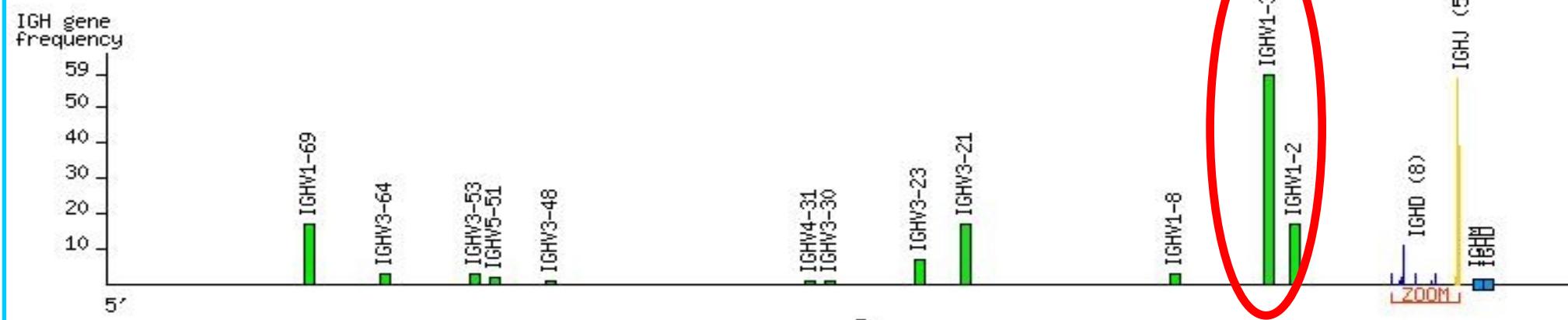


# Example of IMGT/GeneFrequency results

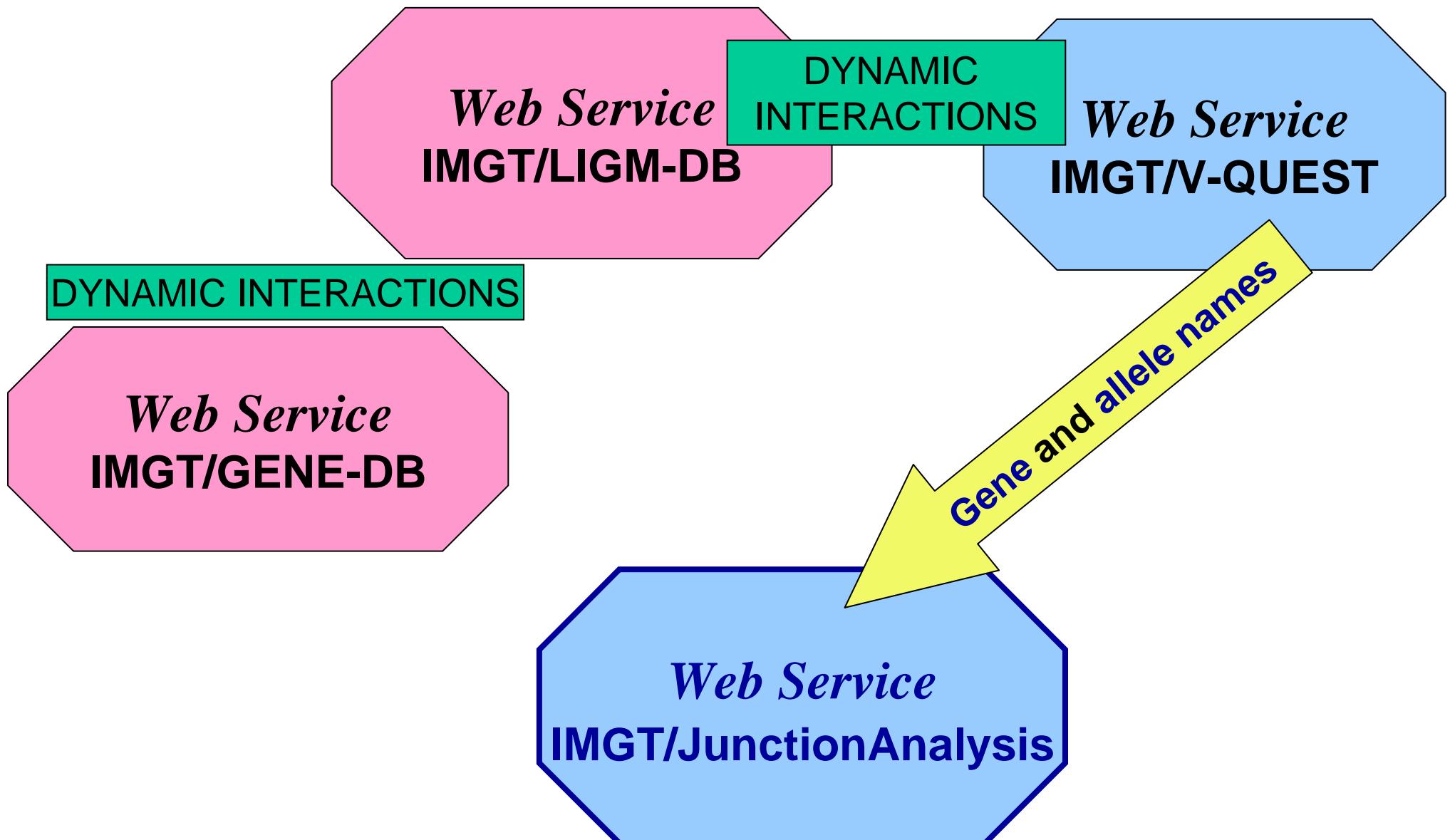
Your Selection :

## Human IGH, IGK and IGL Locus Specificity anti-thyroid peroxidase (TPO)

For the D and J genes, the number of genes is shown between parentheses when genes names could not be indicated for a click on the zoom for the D and J genes names.



# Diagram of collaboration: Analyse des jonctions



# Example of IMGT/JunctionAnalysis results

## Analysis of the JUNCTIONs

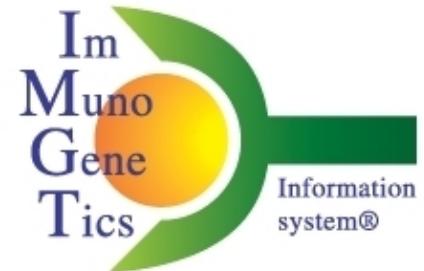
Input	V name	V-REGION	D-REGION	N2	J-REGION	
#1 AF402940	IGHV1-3*01	tgtgcgagag.	.....gcttcacgggg.....	cgggac	....gctttggacgtctgg	
Input	J name	D name	Vmut	Dmut	Jmut	Ngc
#1 AF402940	IGHJ3*01	IGHD3-10*01	0	4	2	5/6

## Translation of the JUNCTIONs

	105	107	109	112	114	116	118	CDR3-IMGT
	104	106	108	110	113	115	117	frame length
#1 AF402940	C	A	R	G	F	T	G	R D A L D V W
	tgt	gct	gca	ggc	ttc	acg	ggg	cgg gac gct ttg gac gtc tgg + 12

# THANK YOU for using IMGT/JunctionAnalysis

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<http://imgt.cines.fr>

## Analysis of the JUNCTIONS

Input	V name	V-REGION	N1	D-REGION	N2
#1 M62724	IGHV7-4-1*02	tatqcqaaqaga	aga	.taqcaatqqctacaa....	aata
#2 Z47269	IGHV1-69*06	tgtgcgagag.	ggggggctaagg	....tcgaattttggagtggtt.....	tcatgggt

Input	J-REGION	J name	D name	Vmut	Dmut	Jmut	Ngc
#1 M62724	....tttactactaa	IGHJ4*02	IGHD5-24*01	0	2	0	1/7
#2 Z47269	...actggttcgaccctgg	IGHJ5*02	IGHD3-3*02	0	2	0	13/20

## Translation of the JUNCTIONS

	105	106	107	108	109	110	111	111.1	111.2	112.3	112.4	112.1	112.2	113	114	115	116	117	118	CDR3-IMGT frame	length			
	C	A	R	E	D	S	N	G						Y	K	I	F	D	Y	W				
#1 M62724	tgt	gct	aga	gaa	gat	agc	aat	ggc						tac	aaa	ata	ttt	gac	tac	tgg	+	13		
	C	A	R	G	G	A	K	V	F	F	L	F	W	E	H	G	V	W	F	D	P	W		
#2 Z47269	tgt	gct	aga	ggg	ggg	gct	aag	gtc	gaa	ttt	ttg	gag	tgg	ttt	cat	ggg	tac	tgg	ttc	gac	ccc	tgg	+	20

[-> IMGT/JunctionAnalysis Search page](#)

[-> IMGT/JunctionAnalysis Documentation](#)

# IMGT-Choreography: Expressed IG and TR repertoires

THANK YOU  
for using [IMGT/JunctionAnalysis](#)

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IMMUNOGENETICS  
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<http://imgt.cines.fr>

## Analysis of the JUNCTIONS

Input	V name	V-REGION	N	J-REGION	J name	Vmut	Jmut	Ngc
#1 AF490920	IGKV1-33*01	tgtcaacactatgatgattccc...		attcactttc	IGKJ3*01	3	0	0/0
#2 AF490935	IGKV4-1*01	tgtcagcaatattatagtagtactcctc.		..tcactttc	IGKJ4*01	0	0	0/0
#3 AF490937	IGKV4-1*01	tgtcagcaatattatagtggtctcc		.gtacactttt	IGKJ2*01	2	0	0/0
#4 AF490932	IGKV3-15*01	tgtcagcactataataactggctcc	cc	tgtacactttt	IGKJ2*01	1	0	2/2

## Translation of the JUNCTIONS

	104	105	106	107	108	109	110	113	114	115	116	117	118	frame	CDR3-IMGT length
#1 AF490920	C	Q	H	Y	D	D		F	P	F	T	F			
	tgt	caa	cac	tat	gat	gat		ttc	cca	ttc	act	ttc		+	9
#2 AF490935	C	Q	Q	Y	Y	S		T	P	L	T	F			
	tgt	cag	caa	tat	tat	agt		act	cct	ctc	act	ttc		+	9
#3 AF490937	C	Q	Q	Y	Y	S		G	P	P	Y	T	F		
	tgt	cag	caa	tat	tat	agt		ggt	cct	ccg	tac	act	ttt		10
#4 AF490932	C	Q	H	Y	N	N	W	P	P	L	Y	T	F		
	tgt	cag	cac	tat	aat	aac	tgg	cct	ccc	ctg	tac	act	ttt	+	11



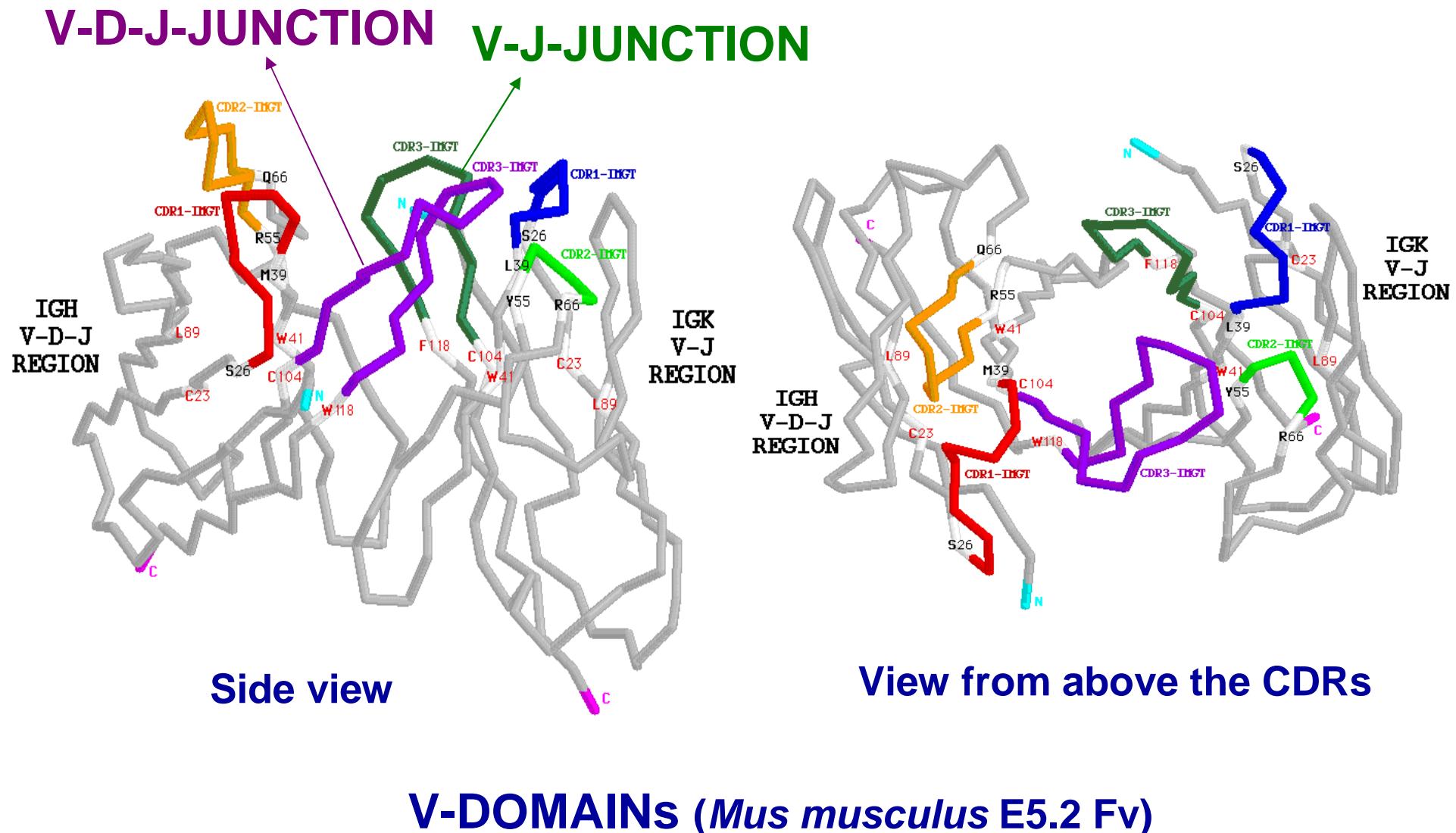
Transferring data from imgt.cines.fr...



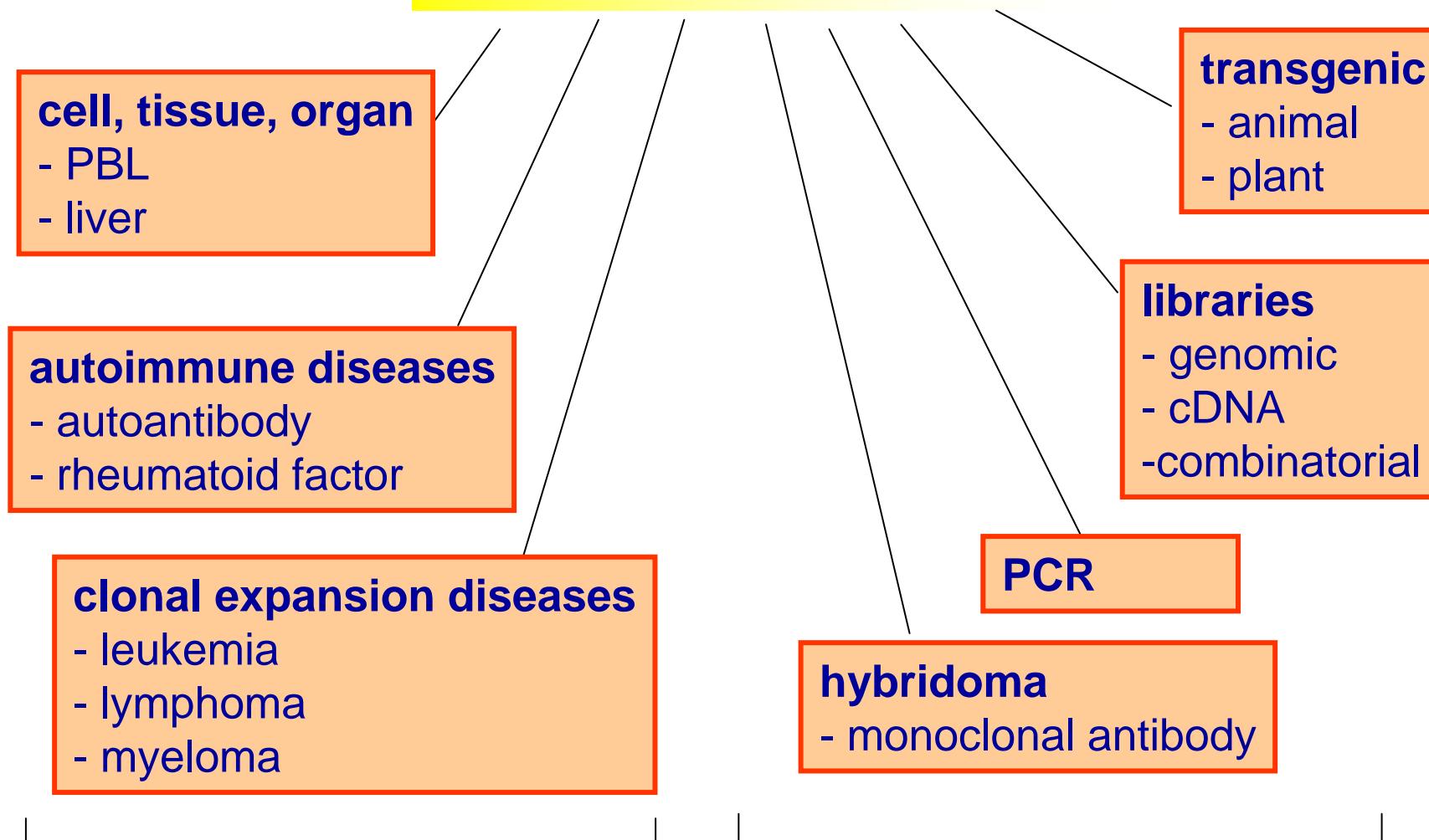
# IMGT-Choreography: 3D structures/specificities



<http://imgt.cines.fr>



## "OBTENTION" concept



origin

methodology

# Immunoinformatics

Data integration specific to Immunology

\*interactions host-pathogens

\*vaccinology

\*immunomodulation...

Gene

Transcript

Protein

Organelle

Bioinformatics,  
databases and tools

Microarrays

3D

Gene regulation  
Pathways  
Networks

Cell

Collection of  
clinical data

Population

Organism

Organ

Tissue

Mathematical and  
computational models

# Who is using IMGT?

## Medical research:

repertoire in autoimmune diseases, AIDS, leukemias, lymphomas, myelomas, translocations, detection of residual diseases

## Veterinary research:

IG and TR repertoire of domestic and farm species

## Genome diversity:

comparative and developmental immunology, evolution of the adaptive immune system

## Therapeutic approaches:

immunotherapy, grafts, immunomodulation, immunosuppression

## Biotechnology related to antibody engineering:

chimeric, humanized, human antibodies, scFv, combinatorial libraries, intrabodies

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