

Bioinformatics Chapter 6.

Genomic Mapping and Mapping Databases

&

E-mail: swchi@cbit.snu.ac.kr

Sung-Wook Chi

Center for Bioinformation Technology (CBIT)
Seoul National University

E-mail: btzhang@cse.snu.ac.kr

Byoung-Tak Zhang

Center for Bioinformation Technology (CBIT) &
Biointelligence Laboratory
School of Computer Science and Engineering
Seoul National University

This material is available at <http://bi.snu.ac.kr/> &
<http://cbit.snu.ac.kr/>

Outline

- ✍ Introduction
- ✍ Genomic Mapping
- ✍ Types of Maps
- ✍ Data Repositories
 - GDB, NCBI, MGI/MGD
- ✍ Mapping Projects and Associated Resources
- ✍ Practical Uses of Mapping Resources

Introduction

- ✍ “Map of Maps”
- ✍ The different types of markers and methods used for genomic mapping
- ✍ The inherent complexities in the construction and utilization of genome maps
- ✍ Several large community databases and method-specific mapping projects
- ✍ Practical examples of how these tools and resources can be used to aid in specific types of mapping studies

Genomic Mapping

Genetic Mapping

- Crossbreeding and pedigree
- Calculation of recombination frequency by linkage analysis

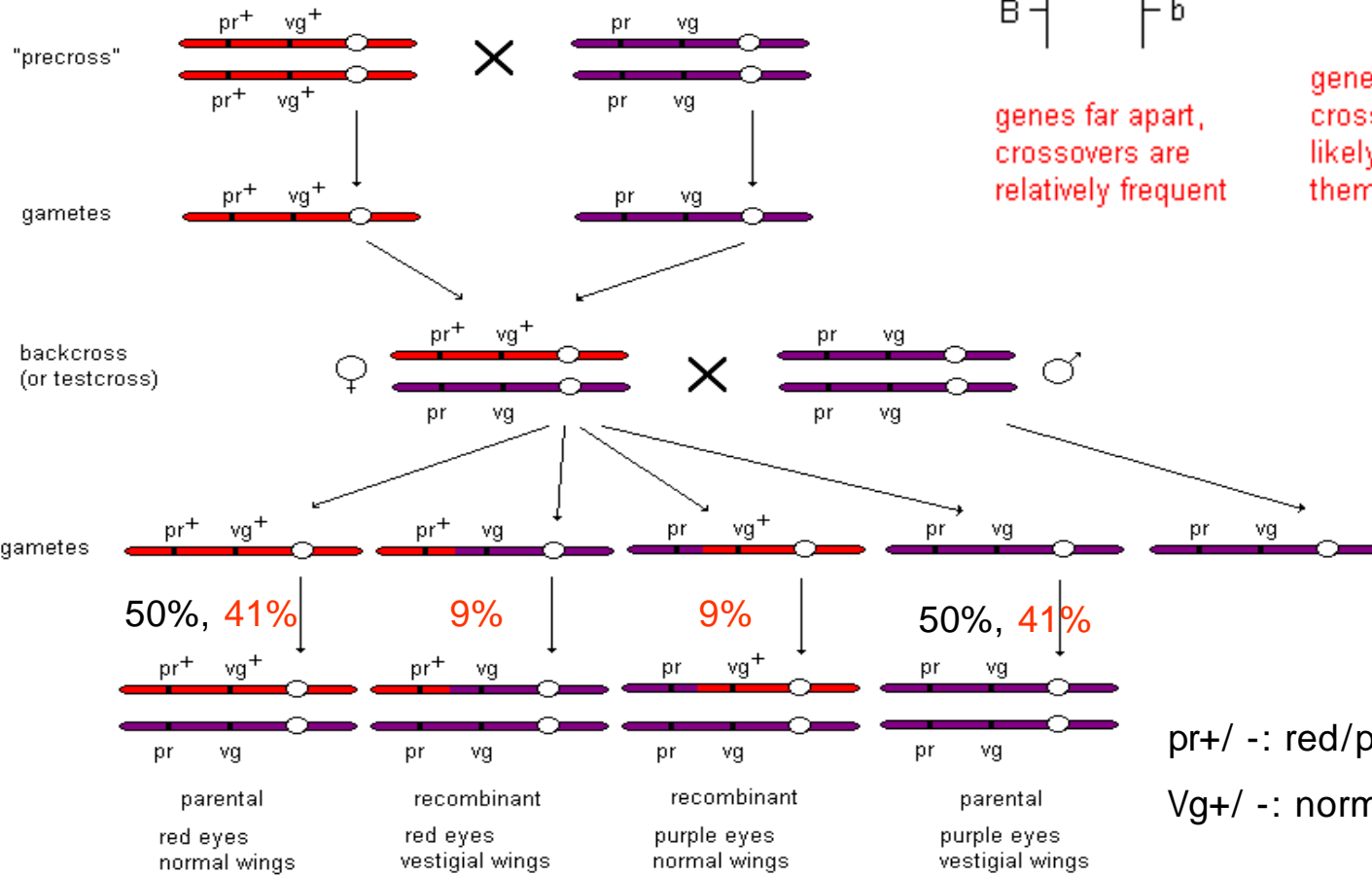
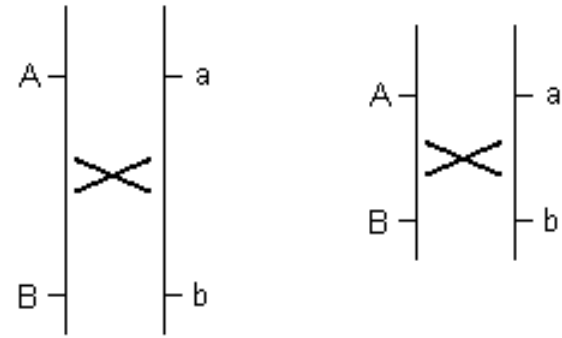
Cytogenetic Mapping

- FISH(Fluorescent In Situ Hybridization)

Physical Mapping

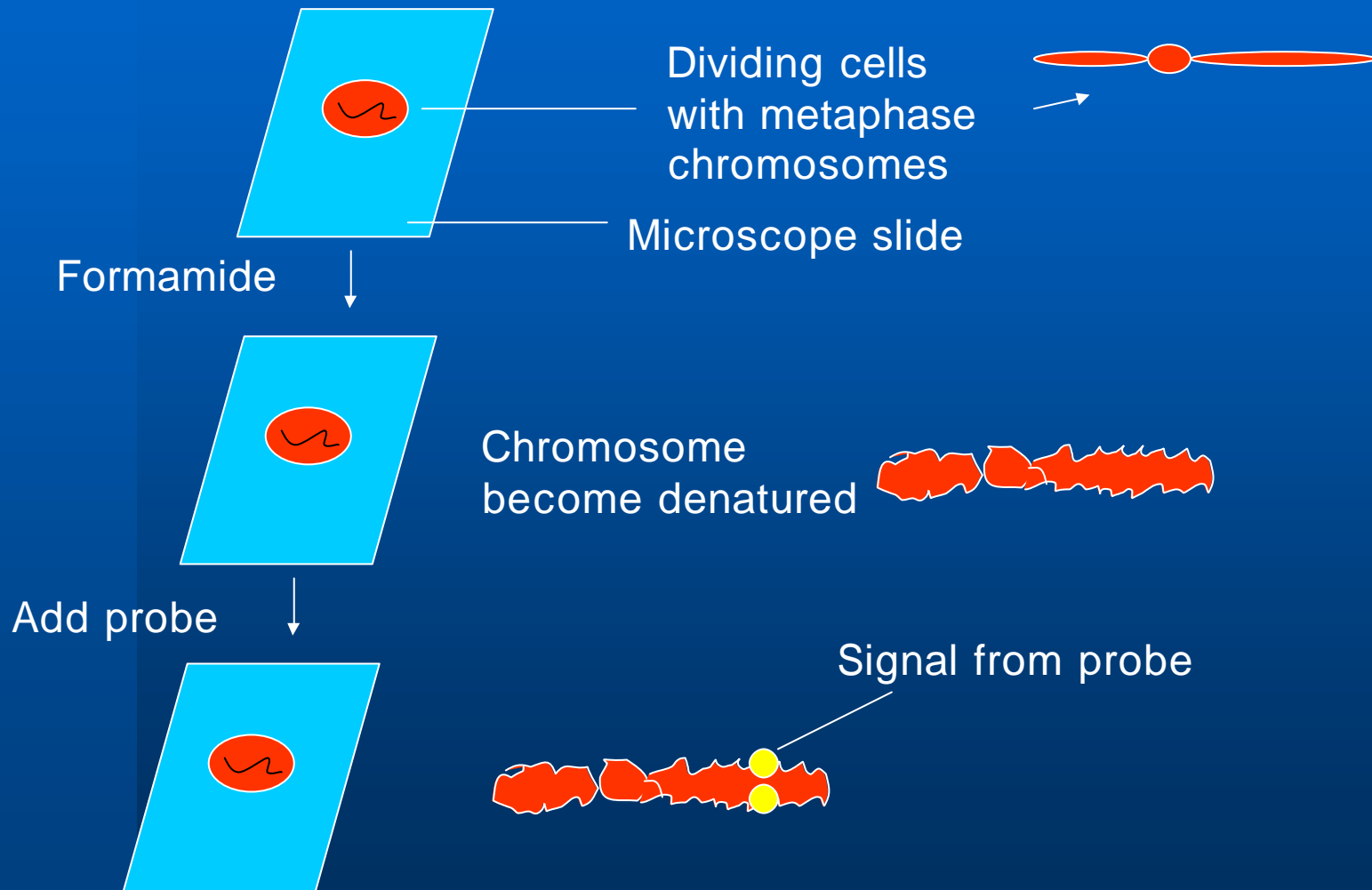
- Molecular biology technique (hybridization, PCR)
- Restriction Mapping
- STS(Sequence Tagged Site) Mapping
- Radiation-hybrid method, Clone library based method.

Linkage Analysis

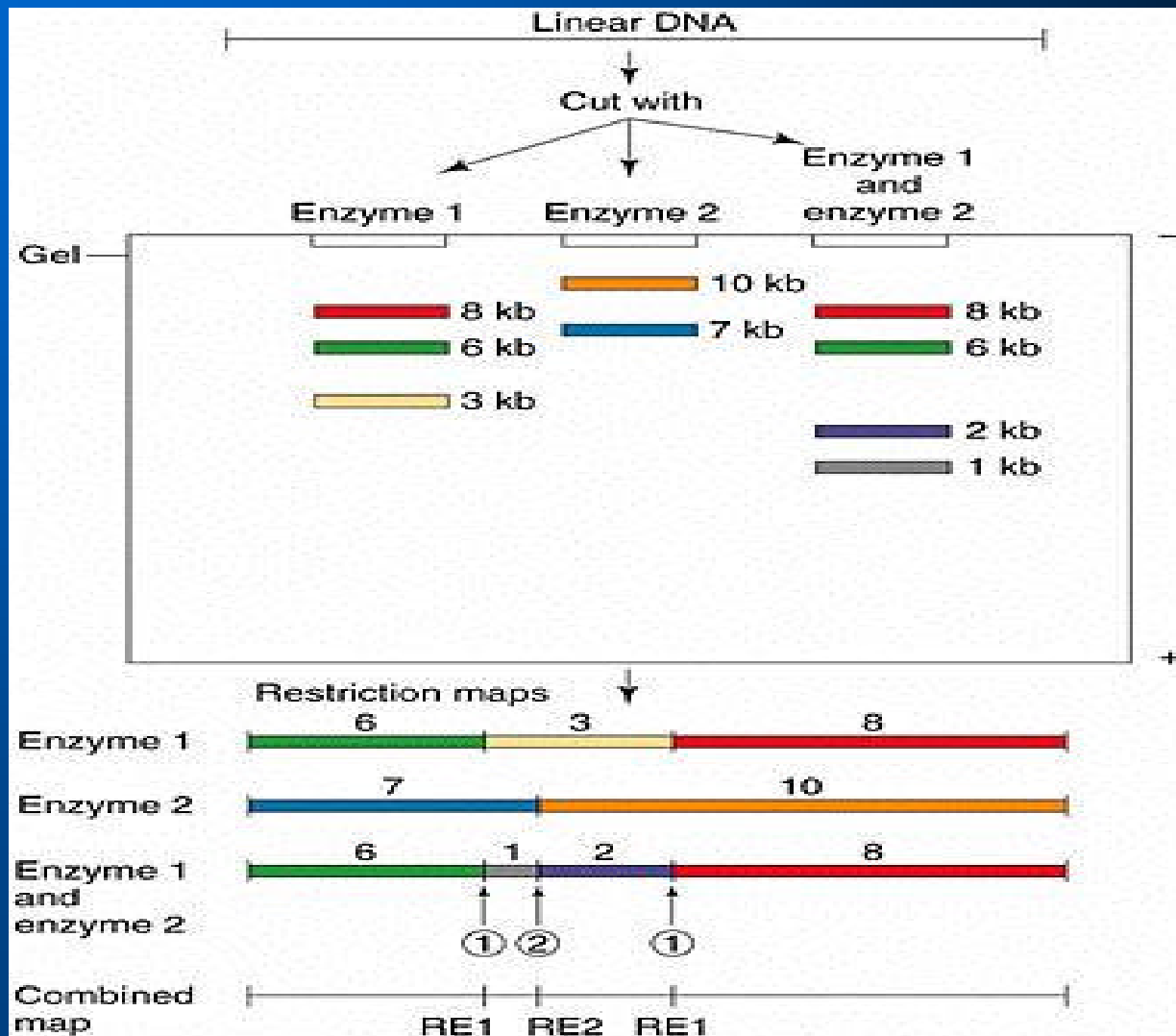


pr+/- : red/purple eyes
Vg+/- : normal/vestigial wings

FISH(Fluorescent In Situ Hybridization



Restriction Mapping



Genomic Map Elements

✍ DNA Markers

- ? A uniquely identifiable segment of DNA
- ? Usually ranging in size from one to 300-400 nucleotide bases in size
- ? Detection of markers may be either PCR based or hybridization based.

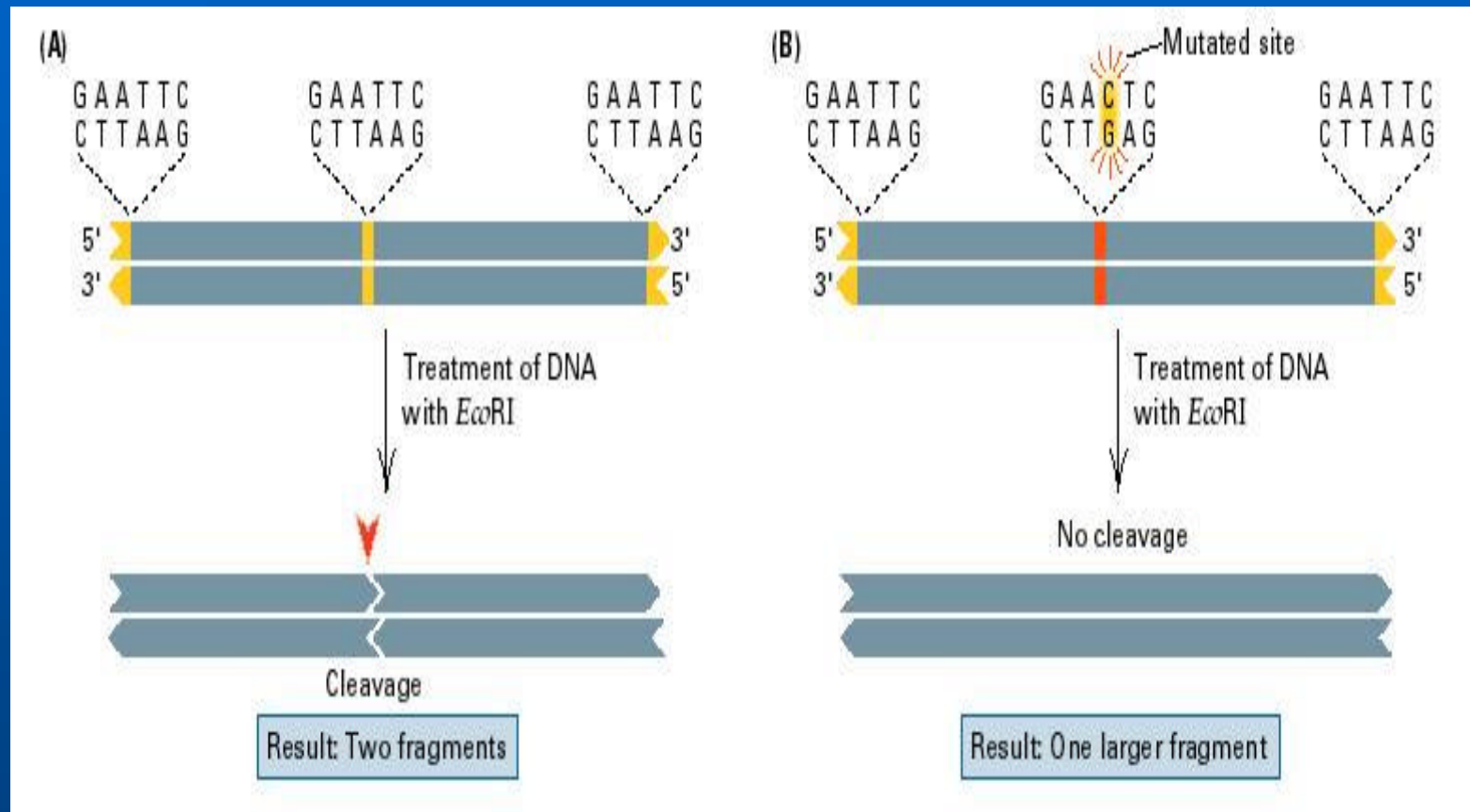
✍ Polymorphic Markers

- ? Show sequence variation among individuals
- ? To construct genetic linkage maps
- ? RFLP (Restriction fragment length polymorphism)

✍ DNA Clones

- ? Yeast artificial chromosome (YAC)
- ? Bacterial artificial chromosome (BACs)
- ? P1-artificial chromosome (PACs)

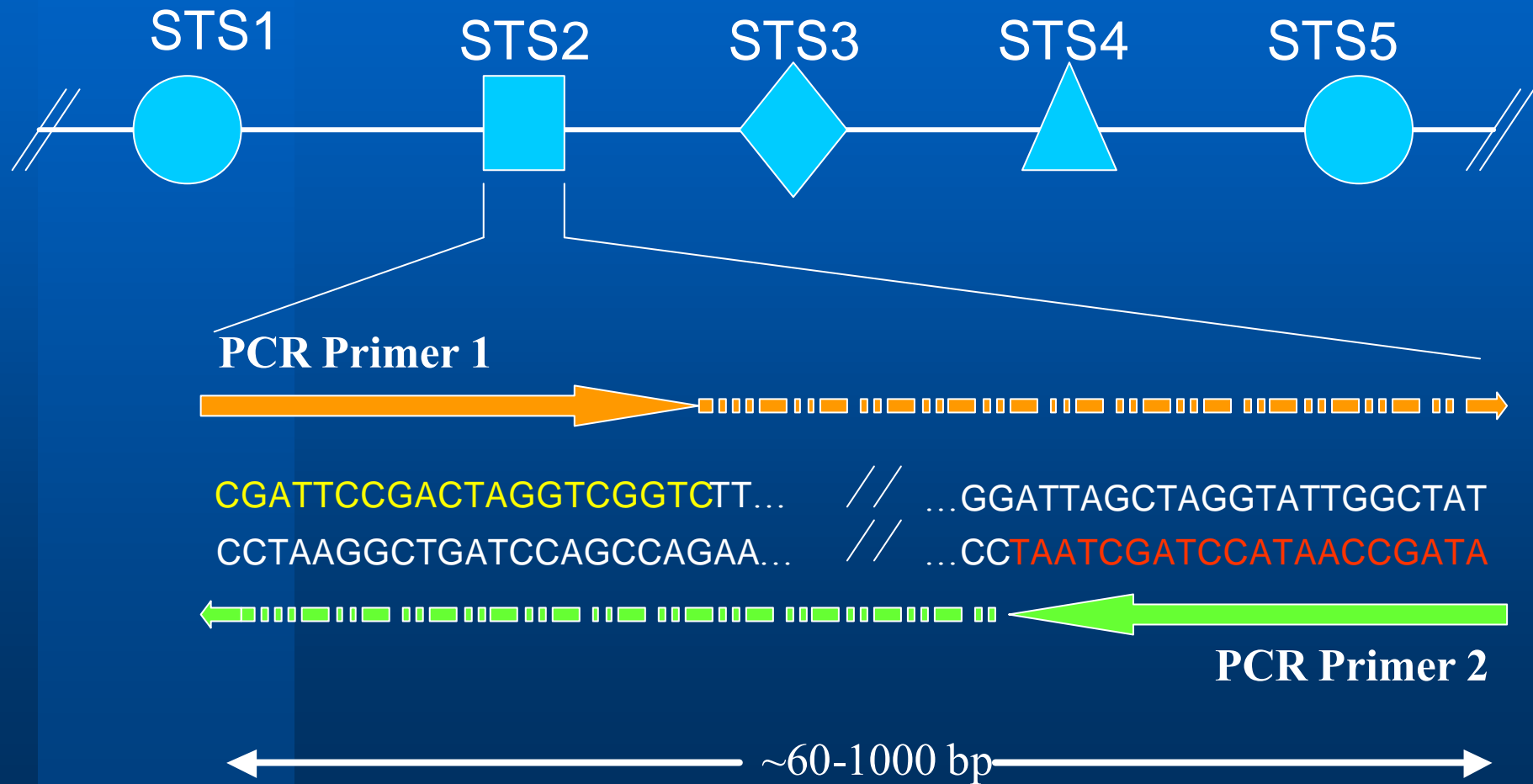
Basis for RFLP mapping



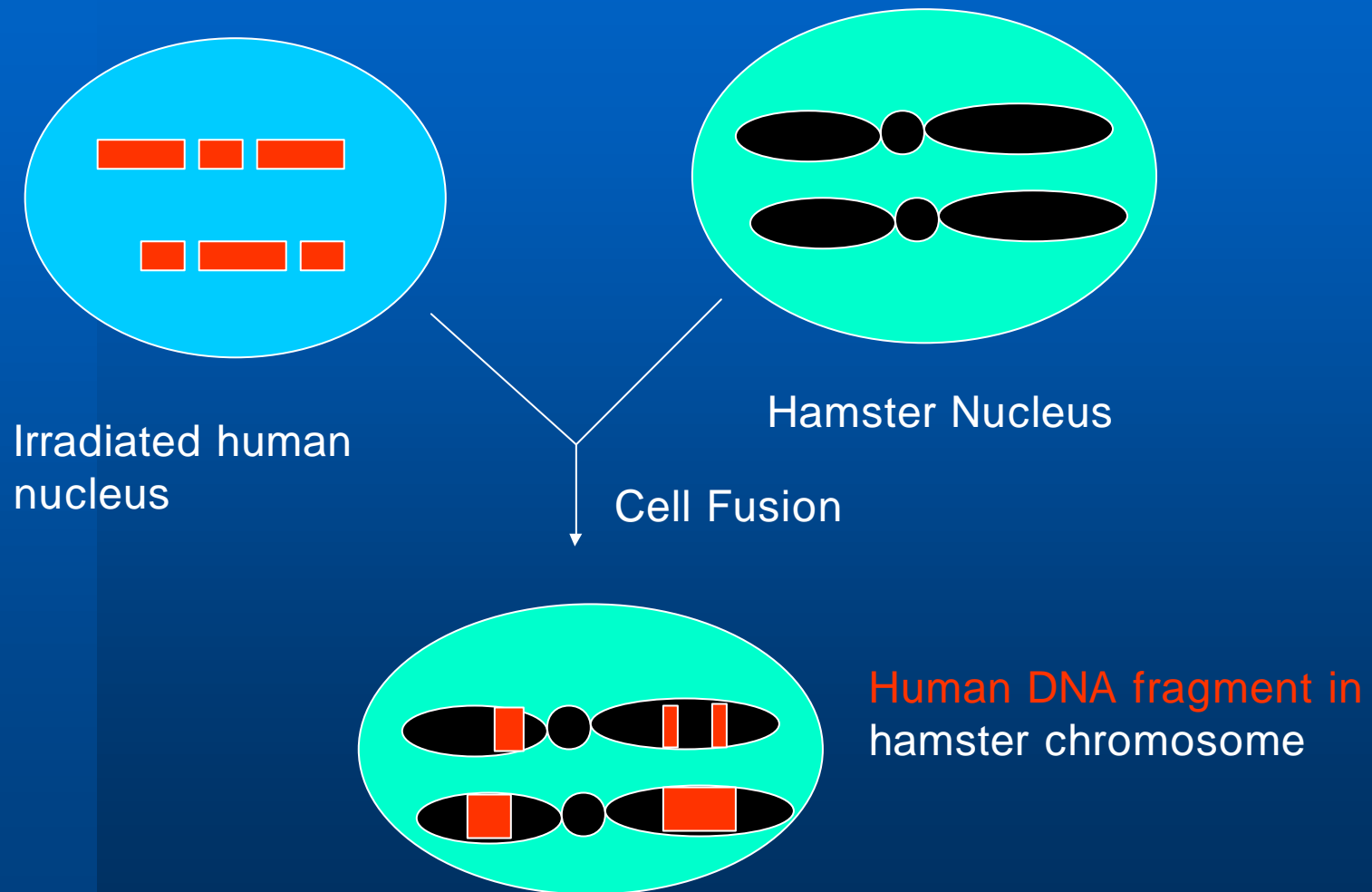
STS(Sequence Tagged Site)

- ✍ Simply a short sequence (100 ~ 500bp)
 - sequence must be known (PCR)
 - must have a unique location.
- ✍ Expressed sequence tags (ESTs)
 - comes from a unique gene
- ✍ SSLP (Simple sequence length polymorphism)
 - Minisatellites, VNTR(Variable number of tandem repeats)
 - Microsatellites, STRs(Simple tandem repeats)
- ✍ Random genomic sequences
 - obtained by sequence random pieces of cloned genomic DNA or from databases.

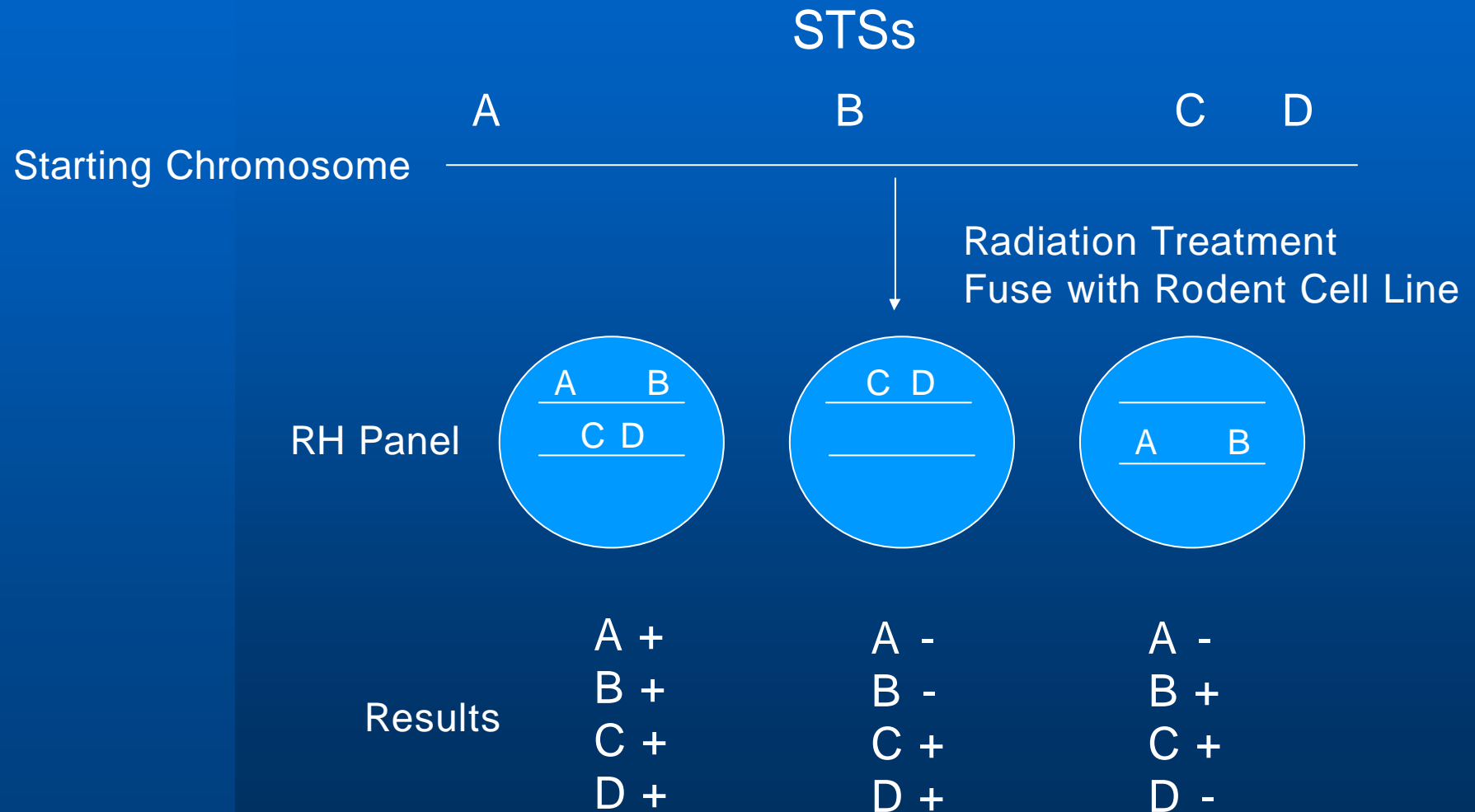
Sequence-Tagged Sites (STS) Mapping



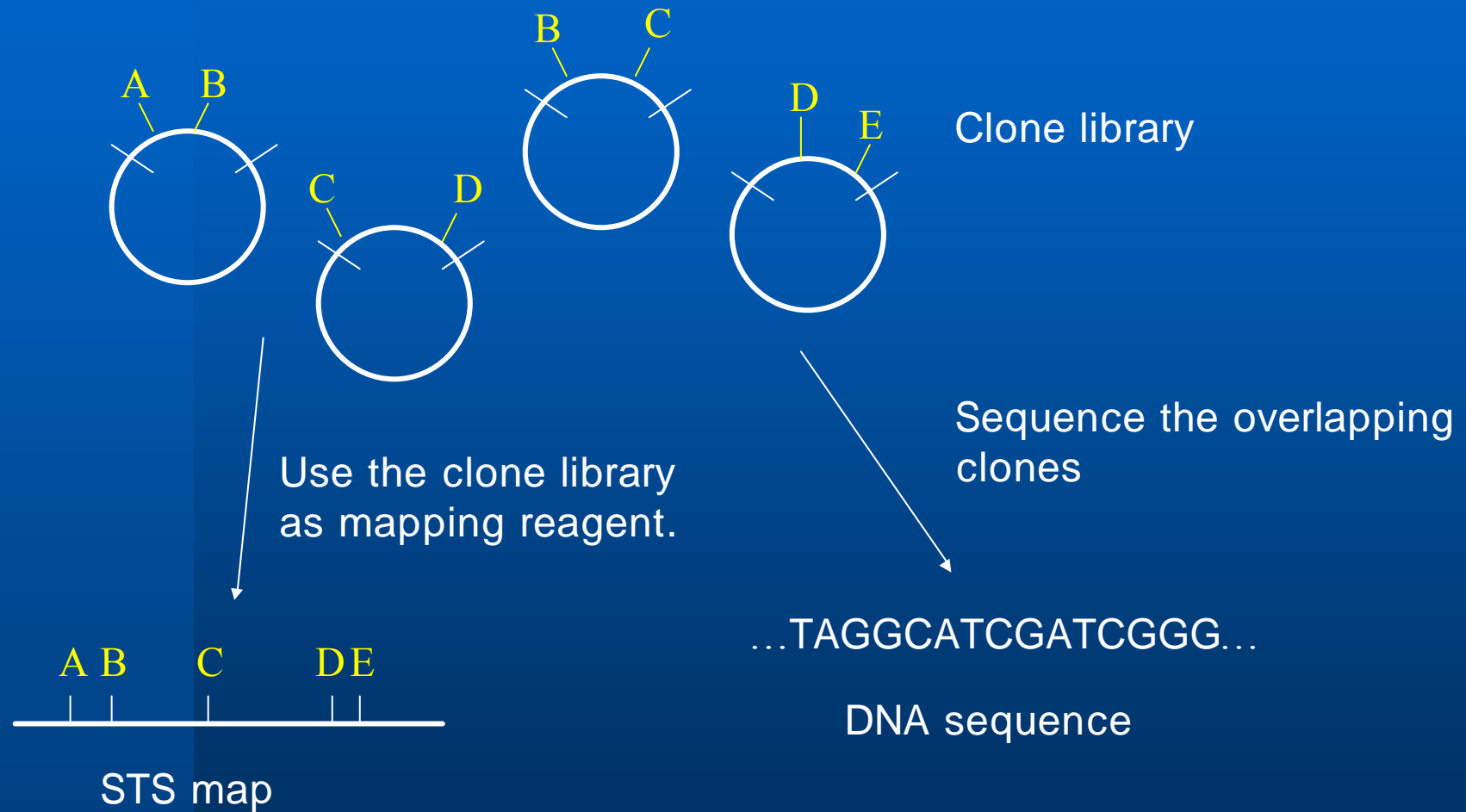
Radiation Hybrid (RH) Method



Radiation Hybrid (RH) Mapping



Clone library based Mapping

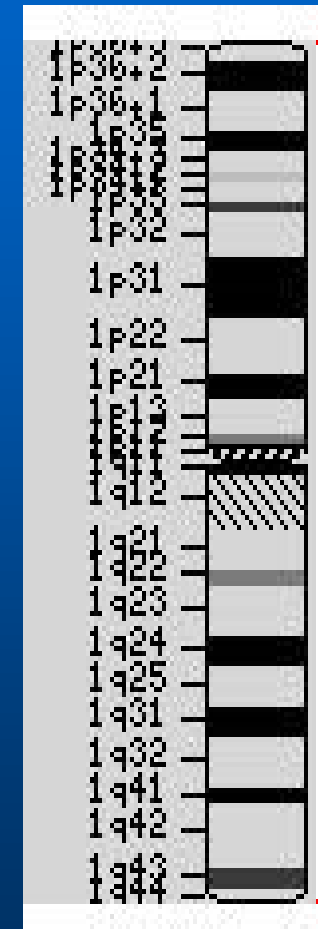


Type of Maps

- ✍ Cytogenetic maps
- ✍ Genetic Linkage maps
- ✍ Radiation hybrid maps
- ✍ Transcript maps
- ✍ Physical maps
- ✍ Comparative maps
- ✍ Integrated maps

Cytogenetic Maps

- ✍ Chromosome staining by Giemsa: G-bands
- ✍ Using FISH method
- ✍ Limited ordering range (<1-2mb)
- ✍ Not well-suited for high-throuput mapping.



Genetic Linkage Maps

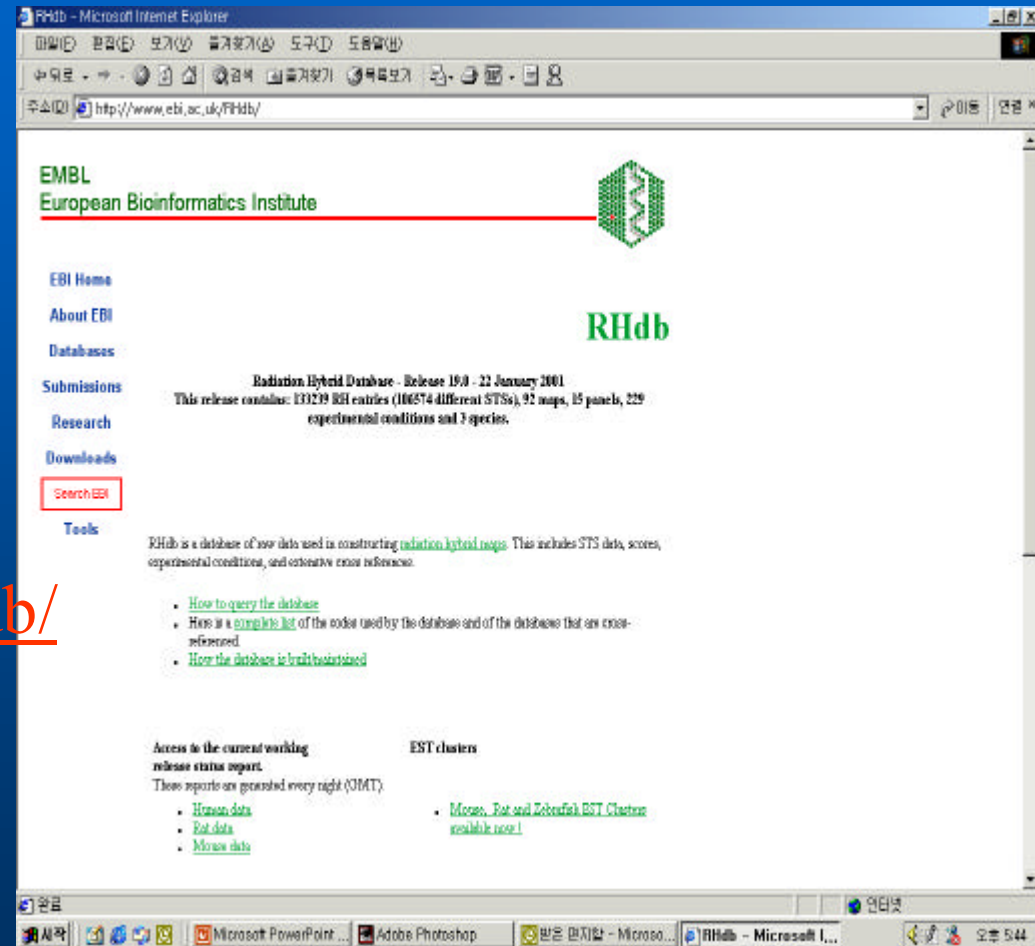
- ✍ 1 centiMorgans (cM) represents 1% probability of recombination.
- ✍ Not directly proportional to physical distance.
- ✍ MAP-O-MAT (map distance, static support for order)
 - <http://compgen.rutgers.edu/mapomat>
- ✍ CEPH(Centre d'Etude du Polymorphisme Humain)
 - Human DNAs from a set of reference pedigrees
 - highly polymorphic STR markers , SNP
 - <http://www.cephb.fr/cephdb/>

Radiation Hybrid Maps

✍ 1 Centirays (cR)
representing a 1%
probability of
chromosome breakage.

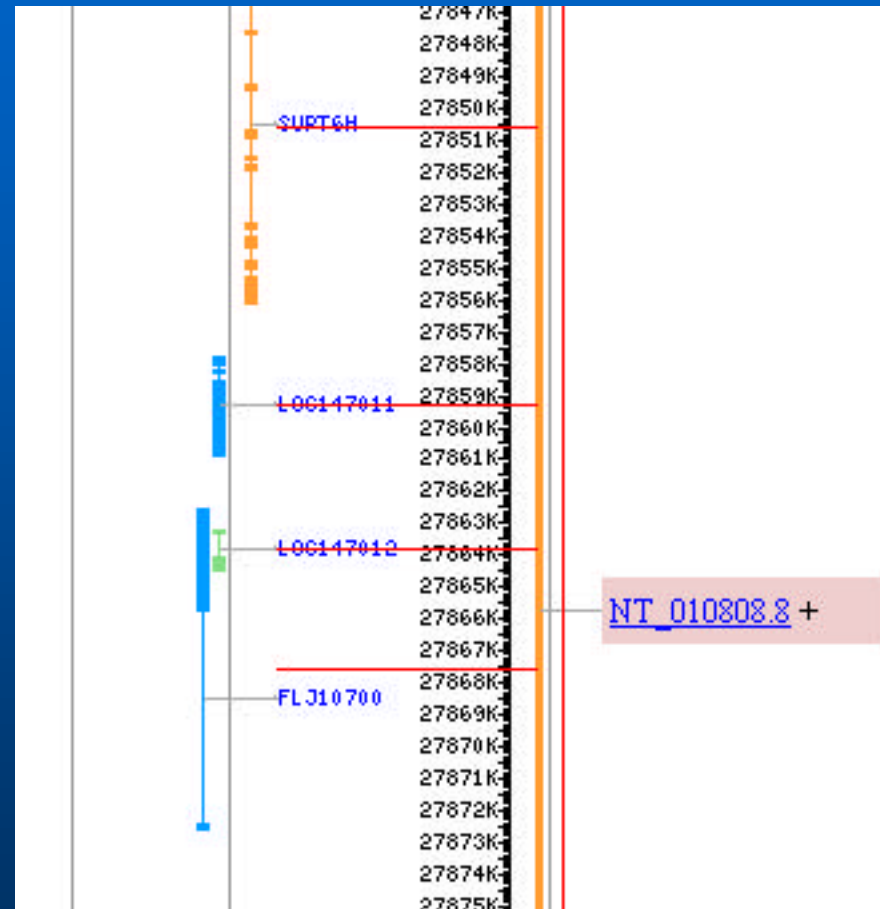
✍ Radiation Hybrid
Database (RHdb)

<http://www.ebi.ac.uk/RHdb/>



Transcript Maps

- ✍ Maps of transcribed sequences.
- ✍ Using Expressed Sequence Tag (EST) and known gene



Physical Maps

✍ STS content mapping (>1Mb)

- using PCR-based positional markers.
- Ordering of marker in clone library
- distance is measured by restriction mapping

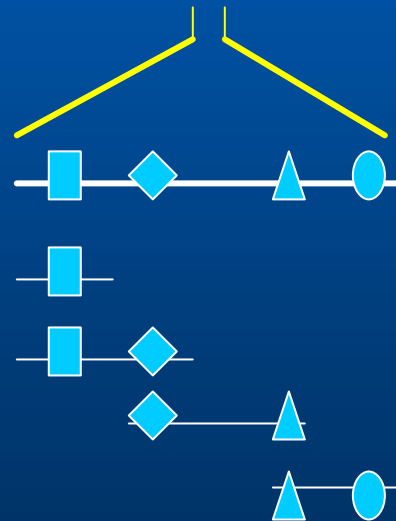
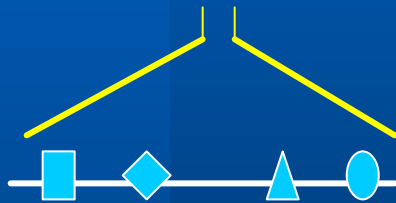
✍ CEPHYAC map

- used a combination of fingerprinting.

✍ Optical Mapping

Comparative Maps

- ✍ Process of identifying conserved chromosome segments across different species.
- ✍ Orthologous genes sharing an identical linear order within a chromosome region.
- ✍ Identify conserved segments and ancient chromosome breakpoints.



RH Map

Clone-Based Map

Sequence Map

Genome Database (GDB)

- ✍ The official central repository for genomic mapping data created by the Human Genome Project
- ✍ Currently, GDB comprises descriptions of three types of objects from humans
 - ? Genomic Segments (genes, clones, amplimers, breakpoints, cytogenetic markers, fragilesites, ESTs, syndromic regions, contigs, and repeats)
 - ? Maps (including cytogenetic, GL, RH, STS-content, and integrated)
 - ? Variations (primarily relating to polymorphisms)

Genome Database (GDB)

The screenshot displays two browser windows from the Genome Database (GDB) website.

The left window shows the main GDB homepage with the following content:

- Logo: **G D B**
- Hosted by: The Hospital for Sick Children, Toronto, Ontario CANADA and available at [other sites](#)
- What's New (8 May, 2001):
 - Server Upgrade, 8 May
 - P-PCR Tools and Database Updated, 20 April
- Normal Operations: [Editing re-enabled](#)
- Simple Search:
 - Search by:
 - Genomic Segments
 - All Biological Data (selected)
 - People
 - Citations
 - by:
 - Name/GDB ID
 - Keyword (selected)
 - DNA Sequence ID
 - Search term: BRCA
 - Buttons: Submit, Reset
- Note: When doing NameID searches, adding * to the end of your search text may improve your results. Also, precede a GDB Accession ID with either "GDB:", "REG:", or "CIT:".
- Other Search Options: [GDB Re-development Survey](#)
- Navigation: [Edit](#) [Help](#) [Site Map](#) [About GDB](#) [Reports](#) [Resources](#) [HGGO Chromosomes](#) [CP Mutations](#) [P...](#)

The right window shows the detailed view for the amplicon **Amplimer BRCA1-17/BRCA1-18** (Central Node). The URL is <http://www.gdb.org/gdb-bin/genera/accno?accessionNum=GDB:391238>.

View Maps of Region: View... Add... Edit...

Names:

Name	Name Status	Authority
BRCA1-17/BRCA1-18	Primary	Skolnick, Mark
BRCA1	Alias	Skolnick, Mark
BRCA-18	Alias	Skolnick, Mark
BRCA-17	Alias	Skolnick, Mark
BRCA17/BRCA18	Alias	Skolnick, Mark

Primers:

Primer Name	Primer Sequence
BRCA-17	GGAATTAATGAAGAGTATGAGC
BRCA-18	TGTGAGGGGACGCTCTTG

Cytogenetic Localization:

Band	Approval Status	Est MB	+/-
17q21-17q21	Unreviewed	47.7	5.6

Cyto. Loc. Evidence: Linkage Contig mapping

Localizations of Related Probes: (84 values) [\[Show All\]](#)

Related Genes:

Genes	Position	Relationship
BRCA1	Exon 11	Overlaps

GDB Mapview 2.4

View Maps in Region of BRCA1-17/BRCA1-18 - Microsoft Internet Explorer

View Maps in Region of BRCA1-17/BRCA1-18

Check maps to display: GDB Comprehensive Map | Annotator | Clones | Contigs | Genes | Others

Centig Maps

- 1 Mb region of TIGR surrounding BRCA1
- BRCA1 Region of Chromosome 17
- Whitehead Contig WCT1.3 July 1997
- Whitehead Contig WCT1.4 July 1997
- Whitehead Contig WCT1.5 July 1997
- Whitehead Contig WCT1.6 July 1997
- Whitehead Contig WCT1.7 July 1997

Cytogenetic Maps

- Chromosome 17 - Cytogenetic Map
- Chromosome 17 - Ideogram (No Internet)

Integrated Maps

- RH Consortium Gene Map 38 - Chromosome 17 (62 panel)
- RH Consortium Gene Map 38 - Chromosome 17 (684 panel)
- RH Consortium Transcript Map - Chromosome 17

Linkage Maps

17

File Edit View Maps Align Detail Help

Selected: Find...

Zoom: Repack Show only selected Show all

Genethon - Chr. 16 (March 1996) Whitehead Chr. 16 RH map (July 1997) GDB Comprehensive Map - Chr. 16

Markers... Markers... Markers... Gene

0 KcM pter 0 cR

D16S3521 D16S521
D16S3082 D16S2960
D16S3072 D16S2965
D16S3030 D16S506
D16S509 D16S502
D16S3092 D16S407
D16S3020 D16S497
D16S3064 D16S58
D16S414 NIB558
D16S3075 D16S499
D16S3060 D16S433E
D16S3079 AFMB354YF9
D16S501 D16S2974
D16S3036 D16S2626
D16S3045 D16S417
D16S417 D16S3181
D16S3113 D16S2984
D16S3116 TIGR-A002K05
D16S3145 D16S2982
D16S3081 D16S540
D16S411 D16S3183
D16S416 D16S2987
D16S3034 D16S2959
D16S3112 D16S408
D16S3140 D16S2941
D16S3057 D16S503
D16S3143 D16S2994
D16S508 D16S2609
D16S3043 WI-9392
D16S496 D16S2961
D16S3086 D16S496
D16S3025 D16S2991
D16S3018 D16S775
D16S3118 D16S516
D16S3142 AFMa109xe9
D16S504 D16S2625
D16S3119 D16S520
D16S402 D16S2621
D16S3077
D16S3023
D16S3121 qter

16p13.3
16p13.2
16p13.13
16p13.12
16p13.11
16p12.3
16p12.2
16p12.1
16p11.2
16p11.1
16q11.1
16q11.2
16q12.1
16q12.2
16q13
16q21
16q22.1
16q22.2
16q22.3
16q23.1
16q23.2
16q23.3
16q24.1
16q24.2
16q24.3

MPG, MEFV, TSC2, ABC3, CATM, ATP6
ZNF200, OR1F1, PKD1, HBQ1, DNASE1
HBZ, HBAP2, HBAP1, HBA2, HBA1, CC
CREBBP, DNASE1, HMOX2, IL9RP3, UE
GSPT1, ERCC4, IMPDHL1
SAH, CRYM
UMOD, PDE1B
ATP2A1, IL4R
STP1, CLN3
- SPN
RBL2, SLC6A2
BBS2
AMFR
DIA4
PSMD7, ZNF23
TAT, ALDOA
COX4, PLCG2
MPE16, PRSM1, FACA, CA5, PCOLN3
MC1R, CMAR, DPEP1, FA1

MYH11
CDR2
UQCRC2
SCNN1
SCNN1
ZNF48
ZNF44
CTF1
CYRN2, SALL1, TBS
CKBP1, PHKB
APOE1, CA7, LCA1
E2F4, HAS3, SLC9A
CBFB, PSKH1, CTRL
ZNF19
ATBF1

Jump Scroll: Horiz. Increment: 1 Vert: 1/2

NCBI

✍ Entrez provides integrated access to several different types of data for over 600 organisms

- ? Nucleotide sequences
- ? Protein structures and sequences
- ? PubMed/MEDLINE
- ? Genomic mapping information

✍ NCBI Human Genome Map Viewer

- ? Human genome sequence data as well as cytogenetic, genetic, physical, and radiation hybrid maps

Example: Homo sapiens Genome

View page

- ✍ A genome-wide search for the term CMT* returns 33 hits. representing the loci for forms of Charcot-Marie-Tooth neuropathy on eight different chromosomes.
- ✍ Selecting the Genes_seq link for the PMP2 gene (the gene symbol for CMT1A, on chromosome 17) returns the view of the sequence map for the region surrounding this gene.

주소(D) http://www.ncbi.nlm.nih.gov/cgi-bin/Entrez/maps.cgi?ORG=hum&MAP1=thon&R1=on&MAP2=sts& 이동 연결

Google Entrez Map PMP22 웹 검색 사이트 검색 PageRank 이미지 정보 위로 하이라이트

NCBI Entrez Genome

PubMed Entrez BLAST OMM Taxonomy Structure

Search Find in This View Find Advanced Search

Map Viewer Help
Human Maps Help
FTP
Chr. 17 Resource

Data As Table View
Maps & Options

Region Shown:
Go

out
zoom
in

17p13
17p12
17p11.2
17q11.1
17q11.2
17q12
17q21
17q22
17q23
17q24

***Homo sapiens* Map View build 28** [BLAST search the human genome](#)

Chromosome: [1](#) [2](#) [3](#) [4](#) [5](#) [6](#) [7](#) [8](#) [9](#) [10](#) [11](#) [12](#) [13](#) [14](#) [15](#) [16](#) [**17**] [18](#) [19](#) [20](#) [21](#) [22](#) [X](#) [Y](#)

Query: (pmp22)[SYM] [clear]

Master Map: Genes On Sequence **Maps & Options**

Total Genes On Chromosome: **2202** [43 not localized]

Region Displayed: **0-84M bp** [Download/View Sequence/Evidence](#)

Genes Labeled: **10** Total Genes in Region: **2159**

Genethon STS Genes_seq Genes_seq symbol

FLJ10979
OR1E1
NUDEL
PMP22
TRAF4
NEUROD2
AOC2
PRO1855
CACNG1
KIAA1453

The Human Genome - Microsoft Internet Explorer

주소(D) http://www.ncbi.nlm.nih.gov/genome/guide/human/

NCBI Home > Genomic Biology > Human

Search LocusLink for BRCA Go

Books
GenBank
GeneMap99
LocusLink
Maps
OMIM
Proteins
PubMed
UniGene
UniSTS

Web Resources
BLAST. Compare a DNA or protein sequence to the sequence to the database or its gene product.
Cytogenetics. A cyto-genetic resource of FISH-mapped, sequence-tagged clones.
dbSNP. Database of SNPs and other genetic variations.
e-PCR. Check your sequence for STSs and view in genomic context.
GEO. Gene Expression Omnibus, a public repository for expression data.
HomoloGene. Putative homologies among human, mouse, rat, and zebrafish.
Homology Map. Blocks of conserved synteny between mouse and human.

Building an information infrastructure
A major challenge facing researchers today is the ability to piece together and analyze the vast quantities of data currently being generated through the Human Genome Project. NCBI's Web site serves an integrated, one-stop, genomic information infrastructure for biomedical researchers from around the world so that they may use this data in their research efforts.
[More...](#)

Working Draft Analysis Published

- NLM Press Release
- NHGRI Press Release
- Interactive Tour of the Genome
- NCBI Genome Analysis Pipeline
- Nature (2/15/01) Human Genome Issue
- Science (2/16/01) Human Genome Issue

MapViewer tips and tricks
When browsing the genome using the new MapViewer, click on Display Settings to choose from several types of maps and . Below are three views of the BRCA2 locus.

Genes & Disease
G&D. Selected gene stories for students and the public.

Conservation
RB1. Complex of retinoblastoma (RB) protein with

주소(D) http://www.ncbi.nlm.nih.gov/LocusLink/list.cgi?Q=BRCA&ORG=Hs&V=0

NCBI LocusLink

PubMed Entrez BLAST OMIM Taxonomy Structure

Search: LocusLink Display: Brief Organism: Human

Query: BRCA Go Clear

View Loci Save Loci

LocusLink Home
Help
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

LocusID	Org	Symbol	Description	Position	Links
672	Hs	BRCA1	breast cancer 1, early onset	17q21	P O R G P H U V

Questions or Comments?
Write to the NCBI Service Desk
[Disclaimer](#) [Privacy statement](#)

Legend:
P PubMed
O OMIM
R RefSeq
G GenBank nucleotide
P Protein
H HomoloGene
U UniGene
V Variation

시작
MSN Messenger
Microsoft PowerPoint ...
The Human Geno...
인터넷
A 漢
오후 2:05

Entrez Map View - Microsoft Internet Explorer

주소(D) ih.gov/cgi-bin/Entrez/maps.cgi?ORG=hum&chr=17&ovr=ideogr&maps=scan-r,est,loc&VERBOSE=ON&cmd=focus&fill=40&size=40&query=BRCA1

Homo sapiens Map View build 28 BLAST search the human genome

Chromosome: [1](#) [2](#) [3](#) [4](#) [5](#) [6](#) [7](#) [8](#) [9](#) [10](#) [11](#) [12](#) [13](#) [14](#) [15](#) [16](#) **[17]** [18](#) [19](#) [20](#) [21](#) [22](#) [X](#) [Y](#)

Query: BRCA1 [\[clear\]](#)

Master Map: Genes On Sequence **Maps & Options**

Total Genes On Chromosome: 2202 [43 not localized]

Region Displayed: 43,284K-43,486K bp [Download/View Sequence/Evidence](#)

Genes Labeled: 13 Total Genes in Region: 13

Region	Gene Symbol	orient.	links	evidence	cyto.	full name
43290K	LOC146923	+	sv ev - seq mm	I	17q21.2	similar to Unknown (protein for IM,
43300K	RPL27	+	sv ev hm seq mm	?	17q21.1-q21.2	ribosomal protein L27
43310K	IFI35	+	sv ev - seq mm	C	17q21	interferon-induced protein 35
43320K	LOC146924	+	sv ev - seq mm	E	17q21.2	hypothetical gene LOC146924
43320K	VATI	+	sv ev hm seq mm	?	17q21	vesicle amine transport protein 1
43330K	LOC146925	+	sv ev - seq mm	E	17q21.2	similar to GTP-binding protein Rho7
43330K	RHO7	+	sv ev hm seq mm	C	17q21	GTP-binding protein Rho7
43360K	BRCA1	+	sv ev hm seq mm	?	17q21	breast cancer 1, early onset

Region Shown: 43,284K - 43,486K

Map View Options: ideogram, master

NCBI Sequence Viewer - Microsoft Internet Explorer

Reverse Complement Strand
View on plus strand

Protein coding genes

Search for gene: Find Refresh

CDS with gene and mRNA
 gene, tRNA, promoter...
 Other features
 Hide sequence

Located on Accession NT_010771
Located on Accession NT_010771

BRCA1

C *GDB:388557 C *GDB:389033 G T *D1781323 T T *G18044 *D178855
 NBR24 A C T G *GDB:388688 *GDB:454227 *GDB:391372 T C *D178932
 *D1781198 T C C A C T C *GDB:391358 C A T *SHGC-151111
 C T G G A T A C *GDB:454278 C G *GDB:391846 *GDB:391867
 A LLOC146927 A A T T A C C T G G T
 T G T A C *GDB:454349 C *GDB:391860 *GDB:391874
 C G C A C *GDB:454353 A *GDB:391881*
 T G T A *GDB:454372 C RH79896*
 A C *GDB:389847 G C C
 GDB:626905* G *GDB:454381 C T
 GDB:389054* *GDB:454417 C
 C T *GDB:391365
 C T LLOC146926
 GDB:454421* *GDB:454450 A
 GDB:454425* *GDB:454454 C
 GDB:454429* *GDB:454458 C
 G *G60034 C
 GDB:454433* *GDB:454462 C
 T *GDB:454466 C
 GDB:454437* T C
 GDB:454441* T
 T
 GDB:454446*

Legend:

- segment boundaries - CDS - RNR - gene
 - region - other feature
 - sequence fragment shown

Sequence:

1145890 GAACTCTGG GAGCGGGGCA GTTTGTAGCT CCGGAGGGA GCCCTAGGA TACGAGGGC

주소(D) http://www.ncbi.nlm.nih.gov/cgi-bin/Entrez/evv.cgi?contig=NT_010771.8&gene=BRCA1&lid=672

Evidence Viewer BRCA1

PubMed Nucleotide Protein OMIM Genome Taxonomy PopSet

Key for display of mRNAs aligning in this region:

Genomic sequence (C)
 model exons, single (M) mRNA exons, single (G, R)
 model exons, overlapping (M) mRNA exons, overlapping (G, R)
 C = contig, M = model mRNA, R = RefSeq mRNA, G = GenBank mRNA

EST density key (E):

1 EST 2-5 ESTs 6-20 ESTs
 21-99 ESTs >100 ESTs

Aligning 19 models, with an average of 17 exons each. Please be patient.

26 exons and 3 genes found in this genomic region spanning 81616 bp.
[View graphic only](#)

C NT_010771.8
 M XM_085638
 M XM_085640
 G AF005068
 G AK054630
 R NM_007294
 R NM_007295
 R NM_007296
 R NM_007297

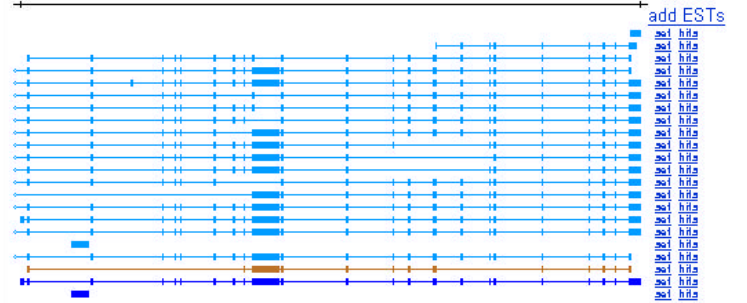
주소(D) http://www.ncbi.nlm.nih.gov/cgi-bin/Entrez/evv.cgi?contig=NT_010771.8&gene=BRCA1&lid=672 페이지 여는 중...

주소(D) http://www.ncbi.nlm.nih.gov/cgi-bin/Entrez/modelmaker.cgi?contig=NT_010771.8&gene=BRCA1&lid=672

Model Maker (Make Your Own Model by selecting an evidence exon "set" help legend and/or add/remove individual putative exons for inclusion in your model)

Evidence:

1145874<<< NT_010771.8 mv sv ev seq >>>1065059 change str...



Putative exons (graphic view):



Your model:

ORF Finder Save

Frame1, ORF= Frame2, ORF= Frame3, ORF=

Putative exons (table view):

1 66166C 1145874-1145497 CTG|GT => 2

주소(D) http://www.ncbi.nlm.nih.gov/Homology/view.cgi?map=ncbi_mgd&tax_id=9606&chr=17&symbol=BRCA1

NCBI Human-Mouse Homology Map

Map: ncbi vs. mgd Master: Human Chromosome: 17 Go

View as text

Human STS	Cytogen Pos	Human Symbol	Mouse chr	Mouse Symbol	cM Position	Mouse STS
17q24-17q25		ACOX1 *		Acox1		
17p13.3		DOC2B	11	Doc2b		
17p13.3		FLJ10979	11	310002B05Rik		
17p13.3		NXN	11	Nxn	45.2	
17p13.3		FLJ10581	11	4833420N02Rik		
17p13.3		LOC51031	11	1700082G03Rik		
17p13		TIMM22	11	Timm22		
17p13.3		ABR	11	Abr	45	
17p13		MYO1C	11	Myo1c	44.13	
17p13.3		PITPN	11	Pitpn	44.11	
17p13		SERPINF2	11	Serpinf2		
17p13.1		SERPINF1	11	Serpinf1		
17p13.3		RPA1	11	Rpa1	44	
17p13.3		DPH2L1	11	Dph2l1	47.7	
17p13.3		HIC1	11	Hic1	47.65	
17p13		SRR	11	Srr		
17p13.3		MNT	11	Mnt	44	
17pter-p13		ASPA	11	Aspa		
17q21.2		MGC10540	11	1110020N15Rik		
17q12-q21.1		RAMP2	11	Ramp2	61.5	
17q21		G6PC	11	G6pc		
17q21.2		MGC2744	11	1110069E20Rik		
17q21.1-q21.2		RPL27	11	Rpl27		
17q21		VAT1	11	Vat1		
17q21		RHO7	11	Arhn		
17q21		BRCA1	11	Brca1	60.5	
17q21.1		M17S2	11	Nbr1	60.5	
17q12-q21		ARF4L	11	Arf4l		
17q21.2		RAB5C *	11	Rab5c	60	
17q21.31		DDX8	11	Ddx8		
17q21		MEOX1	11	Meox1	58	
17q12-q21		SOST	11	Sost		
17q21		DUSP3	11	2210015O03Rik		
17q12-q21		MPP3	11	Mpp3	60	
17q21		PPY	11	Ppy		
17q21		HDAC5	11	Hdac5		

(1개 항목 남음) http://www.ncbi.nlm.nih.gov/Homology/view.cgi?map=ncbi_mgd&tax_id=9606&chr=17&symbol=BRCA1

MGI/MGD

- ✍ The Mouse Genome Initiative Database (MGI): the primary public mouse genomic catalogue resource.
- ✍ Located at The Jackson Laboratory, the MGI currently encompasses three cross-linked topic-specific database
 - ? The Mouse Genome Database (MGD)
 - ? The mouse Gene Expression Database (GXD)
 - ? The Mouse Genome Sequence project (MGS)

MGI

주소(D) http://www.informatics.jax.org/

Google - Entrez Map PMP22 - 열 검색 - 사이트 검색 - PageRank - 페이지 정보 - 위로 - 하이라이트

 The Jackson Laboratory

Query Forms

[What's New?](#) [MGI Home](#) [About MGD](#) [About GXD](#) [About MGS](#)

Searches, Data and Reports [HELP Using MGI Query Forms](#)

- [Genes, Alleles, and Phenotypes](#)
- [Molecular Probes and Segments](#)
- [Mammalian Homology and Comparative Maps](#)
- [Maps and Mapping Data](#)
- [Gene Expression](#)
- [Strains and Polymorphisms](#)
- [References](#)
- [Accession IDs](#)
- [Chromosome Committee Reports](#)
- [Database Reports](#)
- [Contributed Data Sets](#)

Quick Gene Search:

pmp22


Use % as wildcard

Prototypes

- [MouseBLAST](#)
- [Mouse Tumor Biology Database](#)
- [Rat Genome Data](#)

한글

주소(D) http://www.informatics.jax.org/mgihome/ 이동 연결

 MGI

Search the MGI Database

Services

- o [User Support](#)
- o [MGI E-mail List Service](#)
- o [Quick Guide to Nomenclature](#)
- o [Mouse Nomenclature Home Page](#)
- o [Submitting Mutant & Allele Data](#)
- o [Submitting Mouse Genome Data](#)

Resources

- o [Lee Silver's Mouse Genetics](#)
- o [MouseBLAST](#)
- o [Mouse Tumor Biology Database](#)
- o [Gene Family Reports](#)
- o [FTP Site](#)

Vocabularies

- o [Gene Ontology \(GO\) Browser](#)
- o [Mouse Anatomical Dictionary](#)
- o [Phenotype Classification Terms](#)

Related Information

- o [Publications](#)
- o [Collaborators](#)
- o [Acknowledgements](#)

What's New - March 21, 2002

- [New MGI Release Available, March 21, 2002](#)
- [Mouse Genome Monthly Newsletter - Issue #3, January, 2002](#)
- [UCSC Human Genome Browser Adds Links to MGI](#)
- [Scientific Curation Positions Available at MGI](#)
- [Listing of News and Announcements for 2001](#)
- [Listing of News and Announcements for 2000](#)

The [Mouse Genome Informatics Database](#) provides integrated access to data on the genetics, genomics and biology of the laboratory mouse. The projects contributing to this resource are:

Mouse Genome Database (MGD) Project
MGD includes data on gene characterization and nomenclature, mapping, gene homologies among mammals, sequence links, phenotypes, allelic variants and mutants, and strain data. (For details, [see About MGD.](#))

Gene Expression Database (GXD) Project
GXD integrates different types of gene expression information from the mouse and provides a searchable index of published experiments on endogenous gene expression during mouse development. (For details, [see About GXD.](#))

Mouse Genome Sequence (MGS) Project
The goal of the MGS project is to integrate emerging mouse genomic sequence data with the genetic and biological data available in MGD and GXD. MGS supports the MouseBLAST server as a sequence-level entry point into the MGI Database. (For details, [see About MGS.](#))

Gene Ontology (GO) Project
The Mouse Genome Informatics group is a founding member of the Gene Ontology Consortium (www.geneontology.org). MGI fully incorporates the GO in the database and provides a GO browser. (For details see [The Gene Ontology \(GO\) Project](#).)

A [brief history](#) of Mouse Genome Informatics is available.

인터넷

주소(D) http://www.informatics.jax.org/searches/quick_gene_report.cgi

Google Entrez Map PMP22 웹 검색 사이트 검색 PageRank 이미지 정보 위로 하이리이트

Mouse Genome Informatics The Jackson Laboratory

Main Menu | MGI Home | User Support | Help Documents | Submissions | Chr Comm
 Genes | Molecular | Homology | Mapping | Expression | Strain/Polymorphism | Refs | AccID

Query Forms

Genes, Markers and Phenotypes

Query Results -- Summary

2 matching items displayed

Chromosome	cM Position	Band	Symbol, Name
11	34.45		Pmp22 , peripheral myelin protein, 22 kDa
5	59.0		Pxmp2 , peroxisomal membrane protein 2

Citing These Resources
 Funding Information
 Warranty Disclaimer & Copyright Notice
 Send questions and comments to [User Support](#).

last database update 11 Mar 2002
 MGI 2.7

MGI 2.7 - Genes, Markers and Phenotypes Query Results (Details) - Microsoft Internet Explorer

주소(D) http://www.informatics.jax.org/searches/marker.cgi?12398

Google Entrez Map PMP22 웹 검색 사이트 검색 PageRank 이미지 정보 위로 하이리이트 Entrez

Mouse Genome Informatics The Jackson Laboratory

Main Menu | MGI Home | User Support | Help Documents | Submissions | Chr Comm
 Genes | Molecular | Homology | Mapping | Expression | Strain/Polymorphism | Refs | AccID

Query Forms

Genes, Markers and Phenotypes

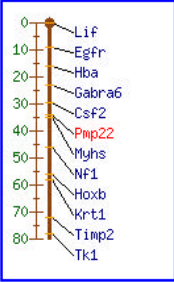
Query Results -- Details Your Input Welcome

Type: Gene
Symbol: Pmp22
Name: peripheral myelin protein, 22 kDa
Chromosome: 11
cM Position: 34.5
MGI Accession ID: MGI:97631

Synonyms: HNPP / Gas-3 / Tr

Additional Information:

- Mammalian Homology
- Marker Mapping Data (30)
- Phenotype (MLC)
- Phenotypic Alleles (6)
- RFLP/PCR Polymorphism (7)
- GXD Index Data (4)
- Molecular Probes and Segments (34)
- References(117)



Gene Classifications: (You can [browse the Gene Ontology \(GO\) Classifications](#))

Category	Classification Term	Evidence	Reference
Biological Process	cell cycle arrest	electronic annotation	J:60000
Biological Process	ionic insulation of neurons by glial cells	electronic annotation	J:60000
Cellular Component	integral membrane protein	electronic annotation	J:60000
Cellular Component	membrane	electronic annotation	J:72247

Result of an MGD
 Quick Gene Search
 for pmp22.

Mapping Projects and Associated Resources

- ✍ Cytogenetic resources
- ✍ Genetic linkage map resources
- ✍ Radiation hybrid map resources
- ✍ STS content maps and resources
- ✍ DNA sequence
- ✍ Integrated maps and genomic cataloguing
- ✍ Comparative resources
- ✍ Single-chromosome and regional map resources

Cytogenetic Resources

- ✍ The central repository for human cytogenetic information is GDB
 - ? Query for marker map and map information using cytogenetic coordinates
 - ? Cross-referencing cytogenetic positions with genes or regions of interest
- ✍ NCBI's LocusLink, UniGene, OMIM catalogues
 - ? Valuable repositories of cytogenetic positions
- ✍ NCI's Cancer Chromosome Aberration Project
- ✍ Southeastern Regional Genetic Group
- ✍ Coriell Cell Repositories

NCBI-UniGene - Microsoft Internet Explorer

UniGene

PubMed Entrez BLAST OMIM Taxonomy Structure

Search Human Go

NCBI UniGene Cluster Hs.194143 *Homo sapiens*

BRCA1 Breast cancer 1, early onset

SEE ALSO
 LocustLink: [672](#)
 OMIM: [113705](#)
 HomoloGene: [Hs.194143](#)

SELECTED MODEL ORGANISM PROTEIN SIMILARITIES
 organism, protein and percent identity and length of aligned region

Organism	Protein	Percent Identity	Length of Aligned Region
<i>H.sapiens</i>	pirA58881 - A58881 breast/ovarian cancer susceptibility protein BRCA1	100 %	1862 aa (see ProtEST)
<i>M.musculus</i>	pir2202221A - breast and ovarian cancer susceptibility protein	56 %	1857 aa (see ProtEST)
<i>A.thaliana</i>	pirT04938 - T04938 hypothetical protein F7J7.10 - Arabidopsis thaliana	26 %	196 aa (see ProtEST)
<i>C.elegans</i>	pirT19770 - T19770 hypothetical protein C36A4.8 - Caenorhabditis elegans	26 %	168 aa (see ProtEST)

MAPPING INFORMATION
 Chromosome: 17
 Genome View: [Chromosome 17](#)
 OMIM Gene Map: [17q21](#)
 UniSTS entries: [GDB:454417](#) Genomic Context: [Map View](#)
 UniSTS entries: [GDB:454433](#) Genomic Context: [Map View](#)
 UniSTS entries: [GDB:454381](#) Genomic Context: [Map View](#)

EXPRESSION INFORMATION
 cDNA sources: B-Cell fetal liver/spleen muscle breastHel a (cell line)

OMIM - BREAST CANCER, TYPE 1; BRCA1 - Microsoft Internet Explorer

OMIM

Online Mendelian Inheritance in Man

Johns Hopkins University

MIM *113705

Text
 Clinical Features
 Inheritance
 Clinical Management
 Population Genetics
 Mapping
 Molecular Genetics
 Gene Function
 Animal Model
 Allelic Variants

View List
 See Also
 References
 Contributors
 Creation Date
 Edit History

MINI-MIM
 Gene map

LocustLink
 Nomenclature
 Ref Seq
 GenBank
 Protein
 UniGene

LinkOut
 CCR
 HGMD

Search OMIM for Go Clear

Display Detailed Save Text Clip Add

[*113705](#) Nucleotide, Related Entries, Protein, PubMed, LinkOut

BREAST CANCER, TYPE 1; BRCA1

Alternative titles; symbols

BREAST CANCER 1, EARLY-ONSET
 BREAST-OVARIAN CANCER, INCLUDED

Gene map locus [17q21](#)

TEXT

For a general discussion of hereditary breast cancer, see [114480](#).

CLINICAL FEATURES

Familial Breast Cancer

Features characteristic of familial, versus sporadic, breast cancer are younger age at diagnosis, frequent bilateral disease, and frequent occurrence of disease among men [Hall et al, \(1990\)](#).

According to the conclusions of the [Breast Cancer Linkage Consortium \(1997\)](#), the histology of breast cancers in women predisposed by reason of carrying BRCA1 and BRCA2 mutations differs from that in sporadic cases, and there are differences between breast cancers in carriers of BRCA1 and BRCA2 mutations. The findings were interpreted as suggesting that breast cancer due to BRCA1 has a different natural history from BRCA2 or apparently sporadic disease, which may have implications for screening and management.

Proliferative Breast Disease (PBD)

http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=OMIM&dopt=Detailed&tpl=dispomimTemplate&li

Genetic Linkage Map Resources

- ✍ The starting point for many disease-gene mapping projects
- ✍ The backbone of many physical mapping efforts
- ✍ Genome-wide maps
 - ? The Cooperative Human Linkage Center
 - ✍ The CHLC has identified, genotyped, and/or mapped over 3,300 microsatellite repeat markers
 - ? The group at Généthon
 - ✍ Identified and genotyped over 7,800 dinucleotide repeat markers
 - ? The Center for Medical Genetics
 - ✍ Identified over 300 dinucleotide repeats
 - ✍ Constructed high-density maps using over 8,000 markers
- ✍ The ABI PRISM linkage mapping
 - ? Dinucleotide repeat markers derived from the Généthon linkage map
- ✍ The Map-O-MAT Web site
 - ? A marker-based linkage map server that provides several map-specific queries

Radiation Hybrid Map Resources

- ✍ Intermediate level of resolution between linkage and physical maps
- ✍ They are helpful for sequence alignment and will aid in completion of the human genome sequencing project
- ✍ The Radiation Hybrid Database (RHdb)
- ✍ The lowest-resolution human RH panel: the Genebridge4 (GB4)
- ✍ An intermediate level panel: The Stanford Generation 3 (G3)
- ✍ The highest resolution panel (“The Next Generation”, or TNG): Stanford

GM98: Chr.22 - Microsoft Internet Explorer

주소(D) http://www.ncbi.nlm.nih.gov/genemap/map.cgi?CHR=22

NCBI A NEW GENE MAP OF THE HUMAN GENOME GeneMap'99
The International RH Mapping Consortium

G??hon Sanger SHGC WICGR WTCHG EBI NCBI

Chromosomes: 1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 X

Search for:

Background
RH consortium
STS markers
RH mapping
Mapped genes
Gene distribution
Reference intervals
Error analysis
Disease genes

Using this site
Search using text
Marker view
Map view
Search by position
FAQs

Chromosome 22: pTEL-D22S420

RH Map Genetic Gene Ideogram
GB4 G3 Map Density

The interval shown is on the GB4 map
See also: equivalent interval on G3 map

Error Flags

- * Minor positional discrepancy
- ** Major positional discrepancy
- *** Chromosome assignment discrepancy

About This Interval

Top of interval: chr22_pTEL (0.0 cM)
Bottom of interval: D22S420 (0.0 cM)
Genetic size of bin: 0 cM
Physical size of bin: 15 cR₃₀₀₀

TELOMERE

0.00	P>3.00	T52917	EST
1.04	P0.19	stSG22362	ESTs
1.15	P0.16	A005T29	BCR breakpoint cluster region
1.50	P>3.00	R41599	COMT catechol-O-methyltransferase

STS Content Maps and Resources

The WICGR physical map

- ? STS content based and contains more than 10,000 markers for which YAC clones have been identified.
- ? Integrated with the Généthon GL and the WICGR RH maps

CEPH/Généthon YAC project

- ? Centered around screening of the CEPH MegaYAC library with a large set of STSs

DNA Sequence

- ✍ The existing human and forthcoming mouse draft genomic sequences are excellent sources for confirming mapping information, positioning and orienting localized markers, and bottom-up mapping of interesting genomic regions
- ✍ NCBI tools like BLAST can be very powerful in finding marker/sequence links.
- ✍ As the mammalian sequencing projects progress, a “sequence first” approach to mapping becomes more feasible

Integrated Maps and Genomic Cataloguing

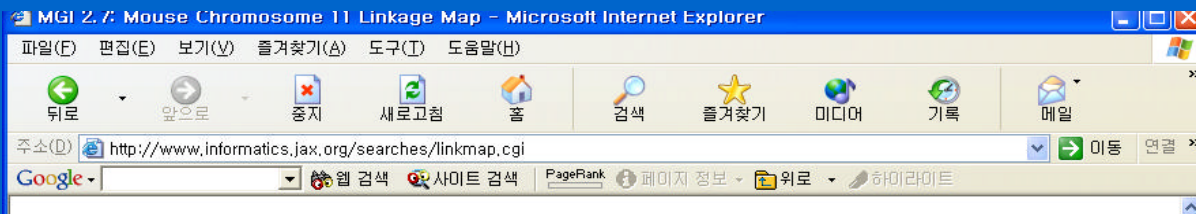
- ✍ GDB's Comprehensive Maps provide an estimated position of all genes, markers, and clones in GDB on a megabase scale.
- ✍ The estimated Mb position of the marker on each map:

Element	Chromosome	Map	Coordinate	Units	EST MB ⁻	+/-
D1S228	1	CeneMap '99	782.0000	cR	32.2	0.0

- ✍ LDB and UDB are two additional sites that infer physical position of a large, heterogeneous set of markers from existing maps using algorithm analogous to GDB's
- ✍ The eGenome project uses a slightly different approach for creating integrated maps of the human genome.
- ✍ Genomic catalogues help both to provide a single initial source containing most of the publicly available genomic information for a region and to make the task of monitoring new information easier.

Comparative Resources

- ✍ Studying the evolution and relatedness of genes between species and finding disease genes through position based orthology.
- ✍ NCBI's LocusLink database: Links to HomoloGene, a resource of curated and computed cross-species gene homologies
- ✍ The GDB does provide homology maps that simplify the reported literature for mouse, human, rat, and 17 other species.
- ✍ The Comparative Mapping by Annotation and Sequence Similarity (COMPASS) approach has been by researchers studying the cattle genome to construct cattle-human comparative maps with 638 identified human orthologs



Mouse Genome Info

Main Menu | [MGI Home](#) | [User Support](#) | [Help Documents](#)
[Genes](#) | [Molecular](#) | [Homology](#) | [Mapping](#) | [Expression](#)

Query Forms

MGD: Mouse Chromosome 11 Linkage Map with human support

Click on a marker symbol to retrieve a detailed marker record from the MGD (*Mouse Genome Database*).

- Page 0: 0.00-1.00 cM
- Page 1: 1.10-2.80 cM
- Page 2: 3.00-7.00 cM
- Page 3: 8.00-10.90 cM
- Page 4: 11.00-11.00 cM
- Page 5: 12.00-13.50 cM
- Page 6: 14.00-16.00 cM
- Page 7: 17.00-19.00 cM
- Page 8: 19.50-22.00 cM
- Page 9: 23.00-25.00 cM
- Page 10: 26.00-27.90 cM
- Page 11: 28.00-28.90 cM
- Page 12: 29.00-30.00 cM
- Page 13: 31.00-32.10 cM
- Page 14: 33.00-34.30 cM
- Page 15: 34.35-36.50 cM
- Page 16: 37.00-38.00 cM

MGI 2.7 - Build a Linkage Map - Microsoft Internet Explorer

주소(D) http://www.informatics.jax.org/searches/linkmap_form.shtml

Map Source:
MGD (stored value)

Chromosome:
11

Restrict map to a chromosomal region? (specify one of the following)

Between [] and [] (*Inclusive. Enter cM positions and/or marker symbols*)

Within [34.5] cM of [pmp22] (*cM position or marker symbol*)

Markers:

Include DNA segments? Yes No
Include syntenic markers? Yes No

Gene Classifications: (*You can [browse the Gene \(GO\) Ontology Classifications](#)*)

contains []

Molecular Function Biological Process Cellular Component

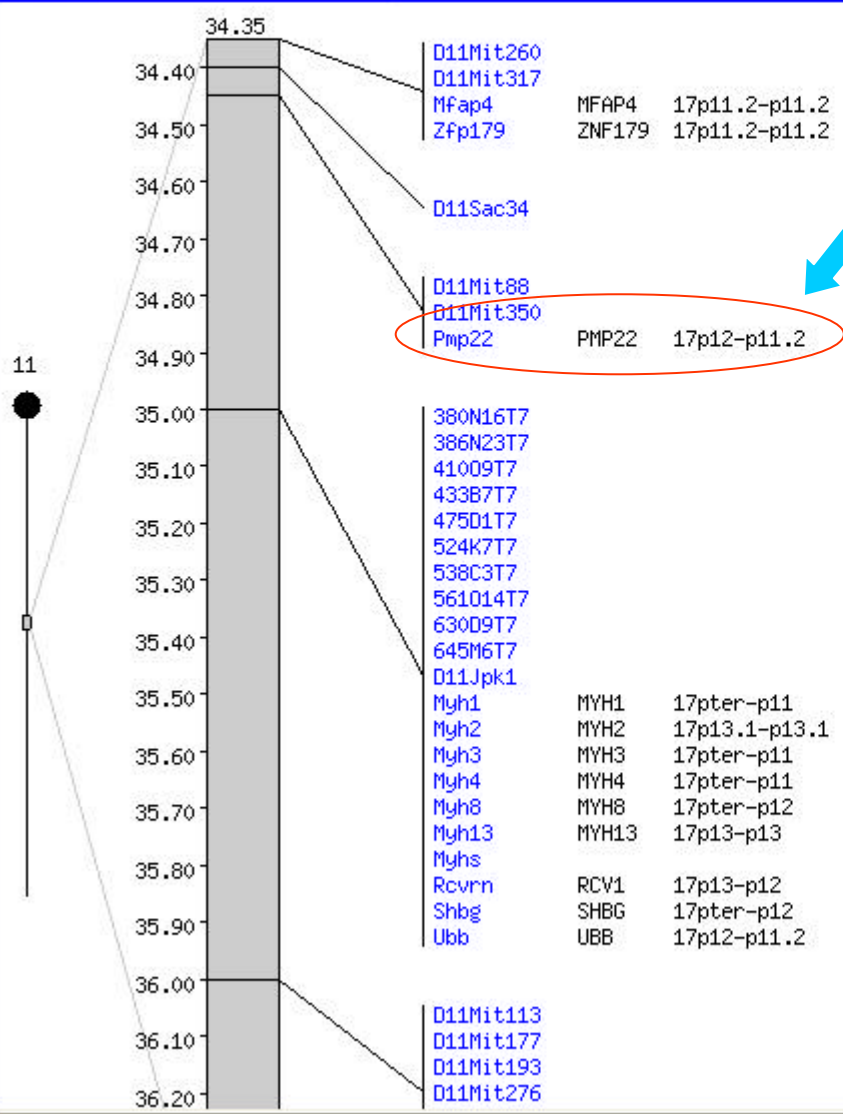
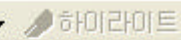
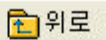
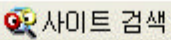
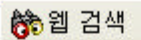
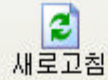
Marker Symbol/Name:
 NOT begins [] Search [current & withdrawn & synonyms]

Comparative Maps:

Show homologs from species:

- human (Homo sapiens)
- cat, domestic (Felis catus)
- cattle (Bos taurus)

URL: http://www.informatics.jax.org/searches/linkmap_form.shtml



Single-Chromosome and Regional Map Resources

- ✍ Data generated for only a single chromosome or a subchromosomal region are often important for fine mapping
- ✍ Most published human chromosome maps are listed and be viewed at GDB's Web site.
- ✍ Human Genome Organization (HUGO) has developed individual human chromosome Web pages.
- ✍ The Sanger Centre and the WUGSC have two of the most advanced collections of chromosome-specific genomic data, informatics tools, and resources.

Practical USES of Mapping Resources

- ✍ Defining a genomic region
- ✍ Determining and ordering the contents of a defined region
- ✍ Defining a map position from a clone or DNA sequence

Defining a Genomic Region

- ✍ A genomic region of interest is best defined by two flanking markers that are commonly used mapping purposes
 - ? Ploymorphic Généthon markers in humans
 - ? MIT microsatellites in mice
- ✍ Commonly used markers are often present on multiple, independently derived maps, so their “position” on the chromosome provides greater confidence for anchoring a regional endpoint
- ✍ In contrast, the exact location of less commonly used markers is often locally ambiguous.

Determining and Ordering the Contents of a Defined Region

- ✍ A good way to start is to identify a map that contains both flanking markers, either from a chromosome-wide or genome-wide map from the sources
- ✍ The map or maps containing the flanking markers can be used to create a consensus integrated map of the region.
- ✍ The most reliable tool for marker ordering is a DNA sequence or sequence contig.

Defining a Map Position from a Clone or DNA Sequence

- ✍ To determine whether the element of interest has already been localized
- ✍ If gene-specific or closely linked markers have been used previously for mapping, a position can usually be described in terms specific to the mapping method that was employed.
- ✍ If no previously localization exists for a genomic element, some experimental work must be undertaken.