2008-08-07

User manual version 1.0

Contents

1. System of VarySysDB

Figure 1-1 Home

Table 1. Subsystems and Pages included in VarySysDB

Subsystem	Page	Function
VaryGene 2	Polymorphism Search	Retrieving and displaying genetic polymorphism.
	Polymorphism Table	Displaying detailed information on polymorphisms.
	Transcript Search	Retrieving and displaying transcript information.
	Transcript Table	Displaying detailed information on transcripts.
	Sequence View	Displaying cDNA sequence with information on polymorphism and functional domains.
	STR/SAR Search	Retrieving and displaying short tandem repeats (STRs) and simple amino acid repeats (SARs).
	CNV Search	Retrieving and displaying Copy Number Variations (CNVs).
	CNV Table	Displaying detailed information on CNVs.
	Keyword Search	Retrieving and displaying by ID, gene name, or Definition.
	System Information	Displaying summary table showing total numbers of transcripts and polymorphisms in VaryGene2.
LD-Search	-	Retrieving and displaying LD-bins within the specified region.
GBrowse	-	Displaying genomic region specified with HITs, HIXs, and polymorphisms.

The results of searches can be downloaded and easily displayed on the computer screen.

1.1. VaryGene2

A menu bar of Varygene2 is designed to select search pages from “Polymorphisms” (i.e., search by feature of SNPs and DIPs), “Transcripts” (i.e., by feature of HITs), “STRs/SARs” (i.e., by feature of STRs and SARs), and “CNVs” (i.e., by feature of CNVs) (Figure 1-1).

1.1.1. Polymorphism Search

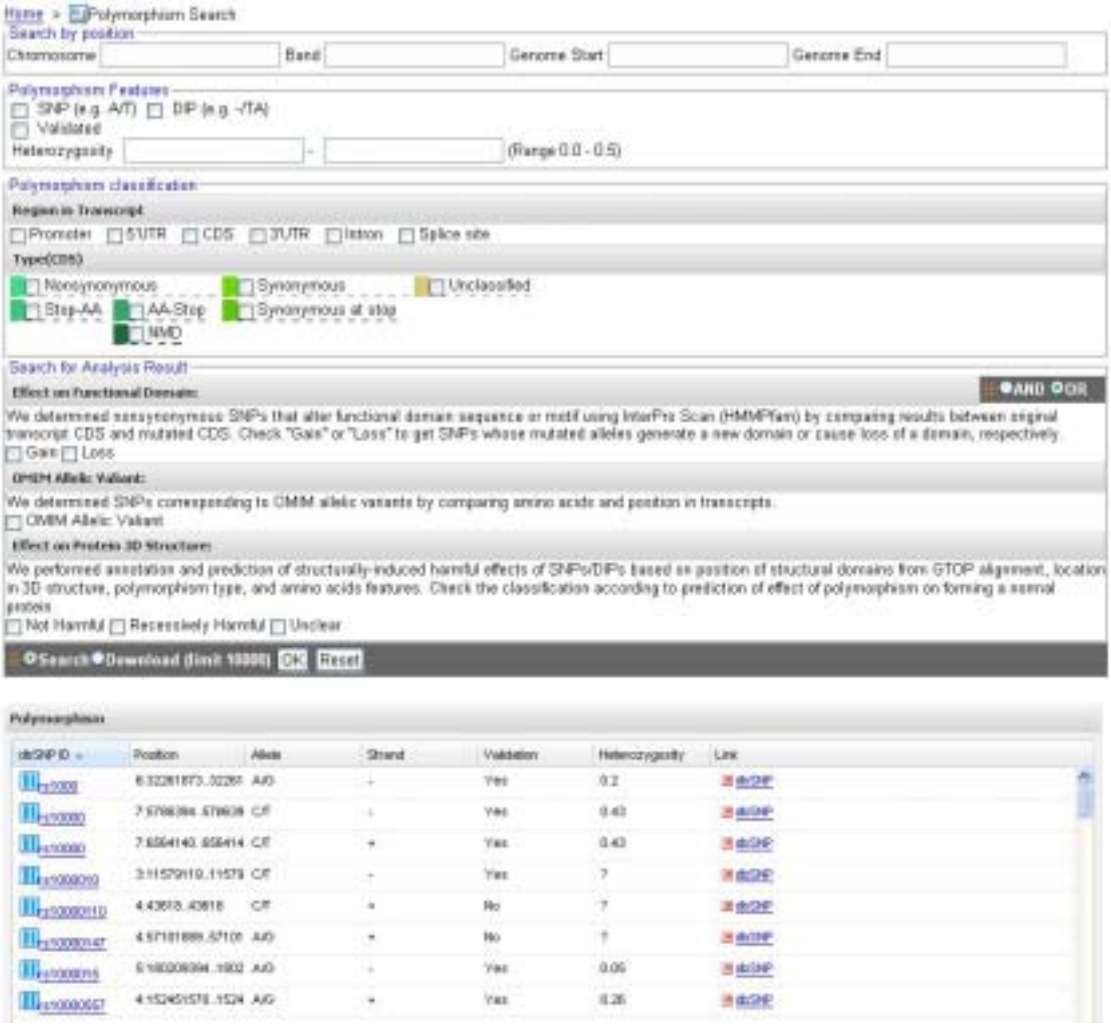

Figure 1-2 Polymorphism Search

In Polymorphism Search page, users can search polymorphism by features, classification, and our analysis results such as effects on functional domains and protein 3D structures. Search criteria "between" boxes are searched by "AND" search, which offers polymorphisms that include all criteria. Search criteria "within" box are searched by "OR" search, except the "Polymorphism Classification" box and "Search for Analysis Result" box, in which user can chose search conditions either "AND" or "OR" searches. Clicking "db SNP" of the retrieved results in "Polymorphism" table below leads users to "Polymorphism Table" page. User can download the retrieved data by clicking Download button.

1.1.2. Polymorphism Table

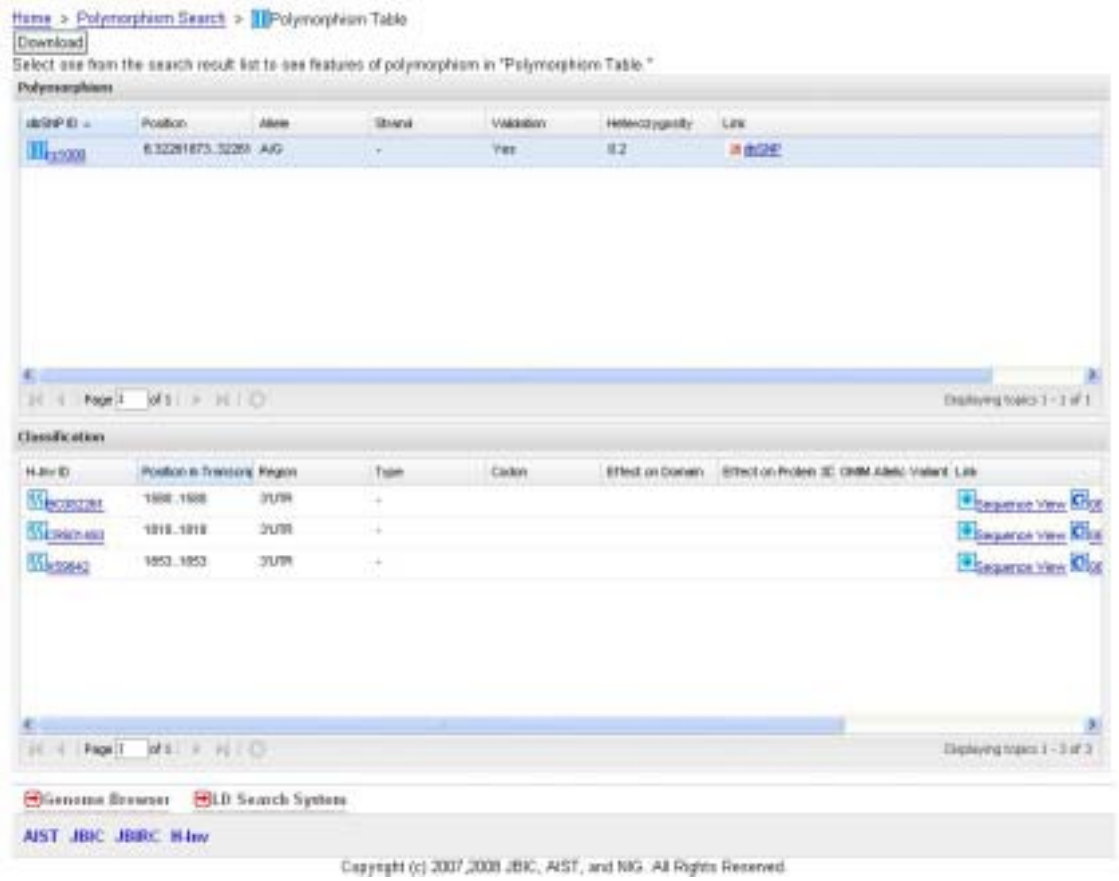

Figure1-3 Polymorphism Table

Click somewhere on a low in “Polymorphism Table” and user can browse classification and analysis results of the selected polymorphism, such as Position in H-Inv Transcript and effects on domain. “Classification” table contains links to the "Transcript Table" , “Sequence View”, "GBrowse", “Transcript View” and "Locus View." User can download the retrieved data by clicking Download button.

1.1.3. Transcript Search

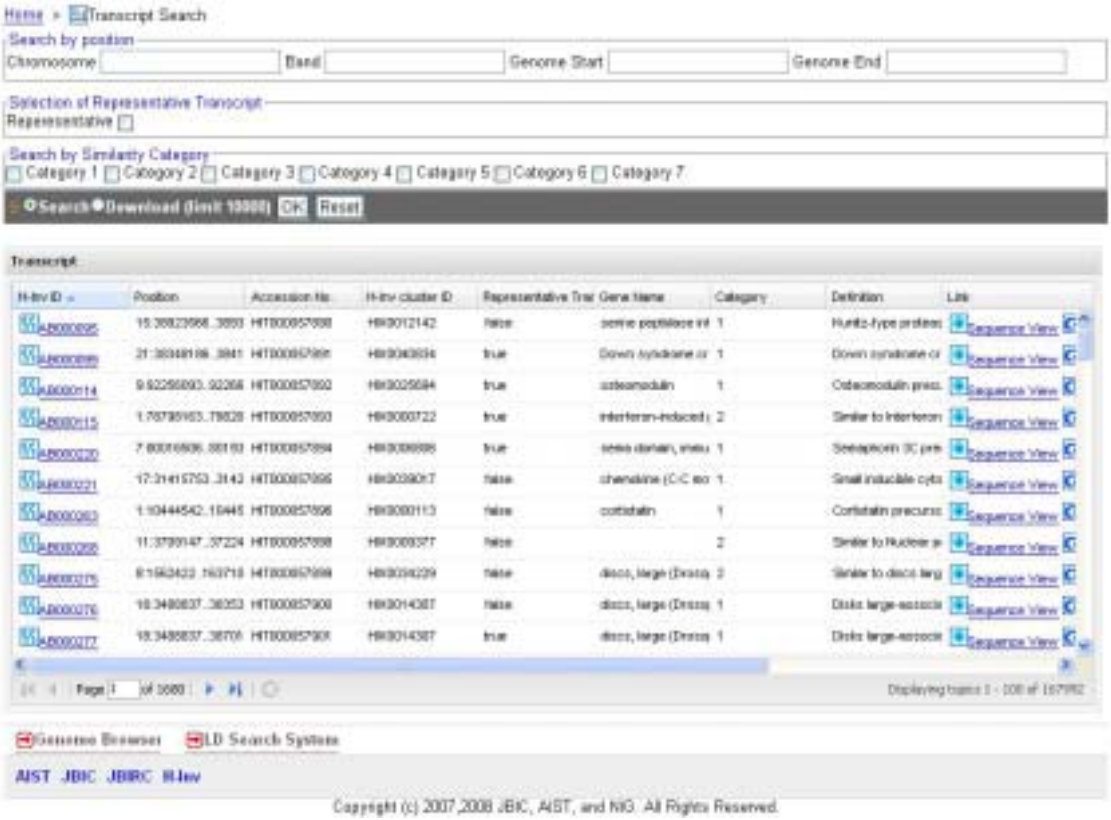

Figure1-4 Transcript Search

In Transcript Search page, users can search transcript by features. Search condition concerning between and within box is the same as Polymorphism search page. Clicking “H-Inv ID” of the retrieved results in “Transcript” table below leads users to “Transcript Table” page. “Transcript” table showing retrieved results contains links to the “Sequence View” , "GBrowse", “Transcript View”, and “Locus View.” User can download the retrieved data by clicking Download button.

1.1.4. Transcript Table

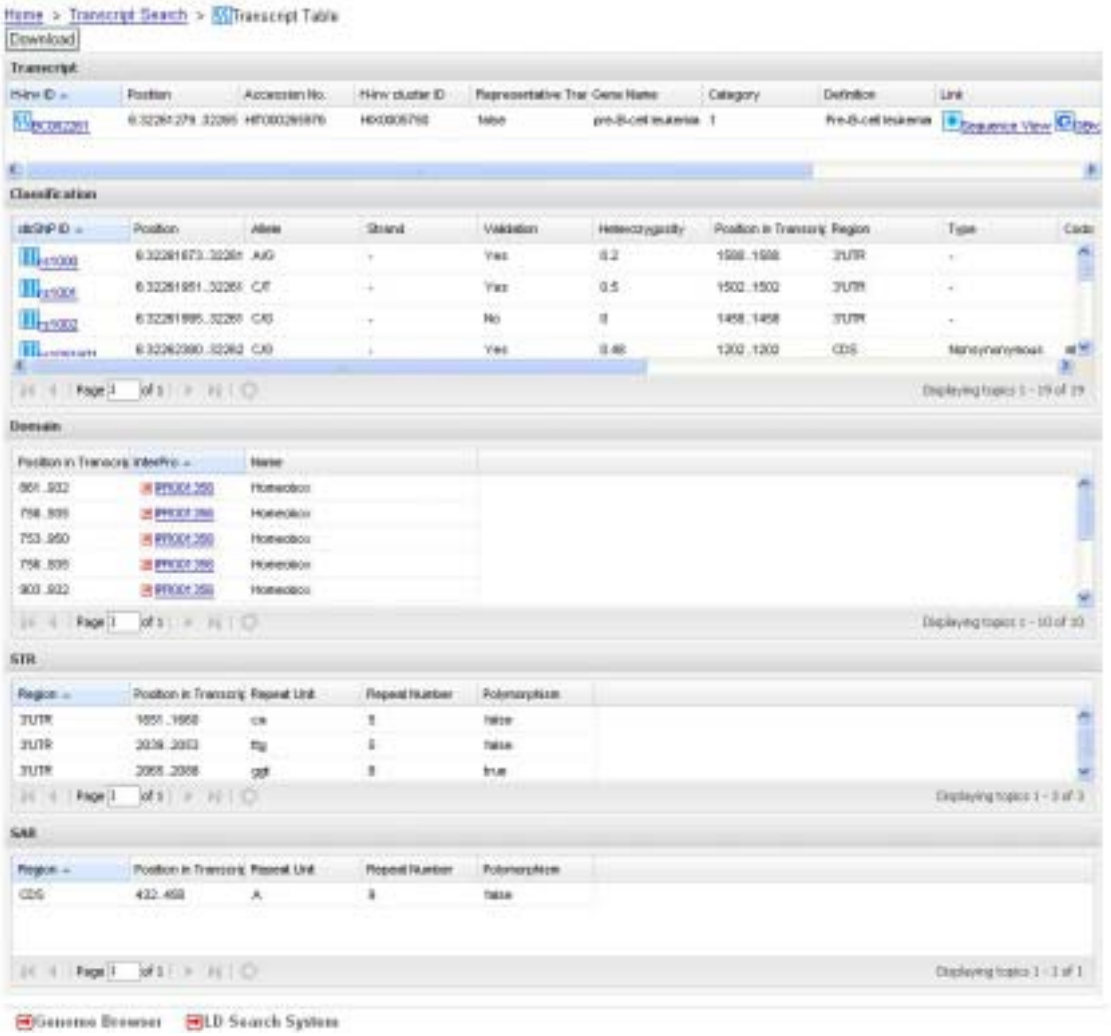

Figure1-5 Transcript Table

Transcript Table page displays information of transcript, polymorphisms, functional domains, STRs, and SARs. “Transcript” table above contains links to the “Sequence View”, "GBrowse", “Transcript View”, and “Locus View” of H- InvDB. User can download the retrieved data by clicking Download button.

1.1.5. Sequence View

Figure1-6 Sequence View

Sequence View page displays transcript sequence with polymorphism classification and location of CDS and functional domain. Click on colored area and users can see detailed information in a separate window. User can download the displayed data by clicking Download button.

1.1.6. STR/SAR Search

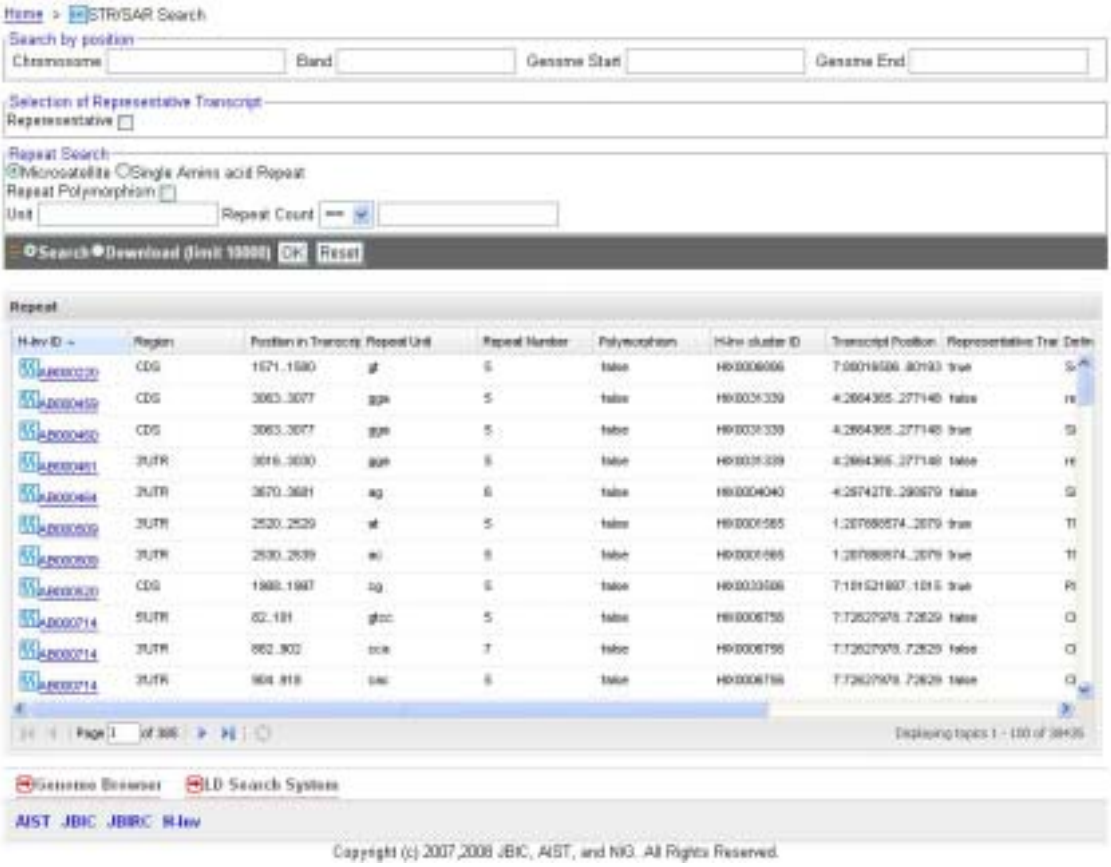

Figure1-7 STR/SAR Search

In STR/SAR Search page, user can search STRs and SARs with transcript information. Clicking “H-Inv ID” of the retrieved results in “Repeat” table below leads users to “Transcript Table” page. “Repeat” table contains links to the “Transcript View”, and “Locus View” of H- InvDB. User can download the retrieved data by clicking Download button.

1.1.7. CNV Search

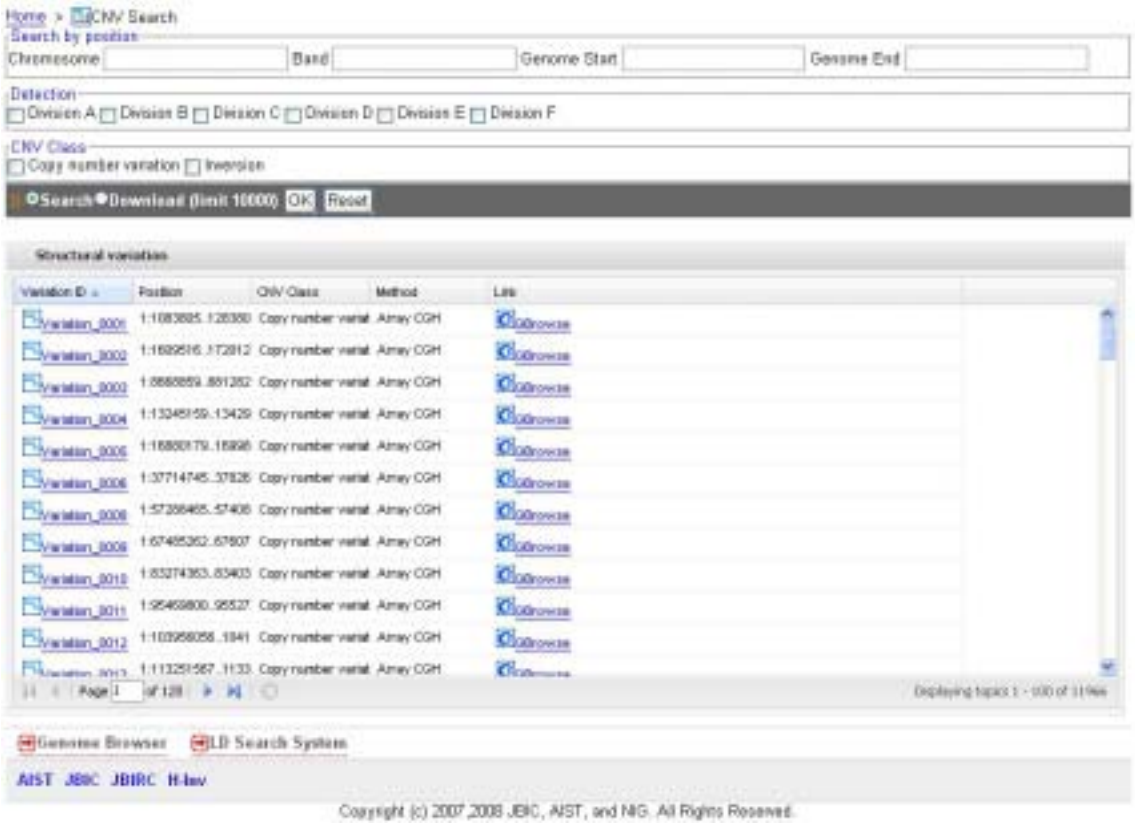

Figure1-8 CNV Search

In CNV Search page, user can search CNVs. Clicking “Variation ID” of the retrieved results in “Structural variation” table below leads users to “CNV Table.” User can download the retrieved data by clicking Download button.

1.1.8. CNV Table

Figure1-9 CNV Table

CNV Table page displays information of CNVs. User can download the retrieved data by clicking Download button.

1.1.9. Keyword Search

Figure1-10 Keyword Search

In the search box above in each page, user can search IDs, genes, and definitions of H-Inv transcript by keyword. “Keyword Search” table contains links to suitable page according to retrieved keyword types. For example, when “dbSNP ID” is selected for keyword search, link to “Polymorphism” table will be appeared.

1.1.10. System Information

Figure1-11 System Information

System Information page displays summary table showing total numbers of transcripts and polymorphisms in VaryGene2.

1.2. LD Search System

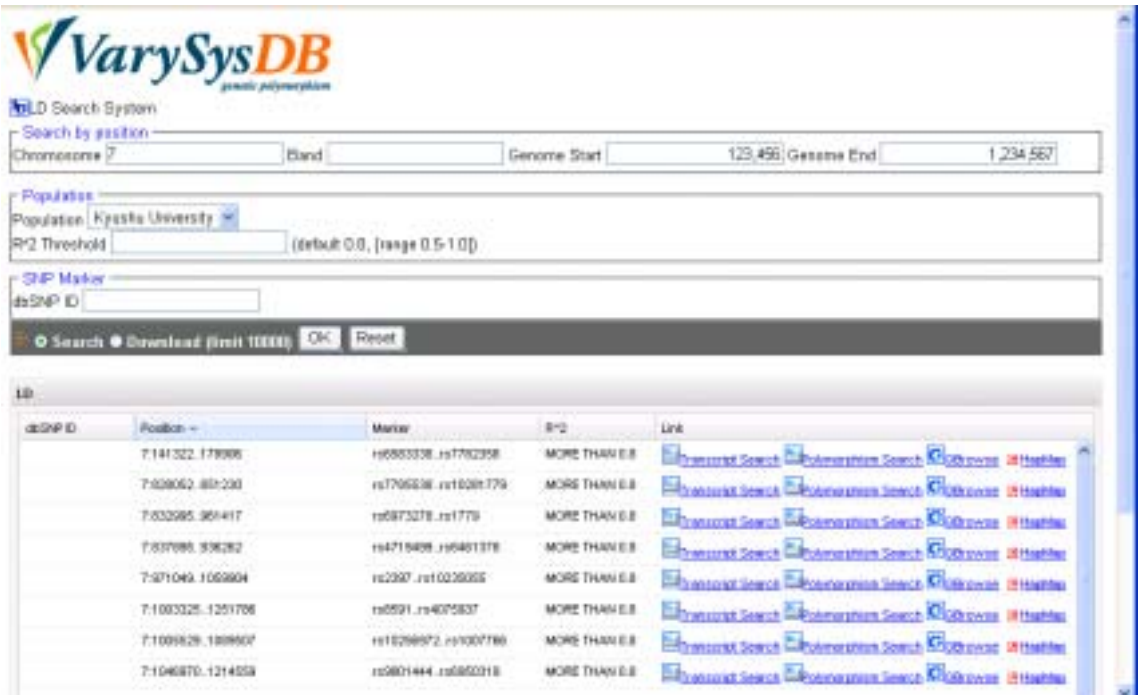

Figure2-1 LD Search System

LD Search System is a subsystem to retrieve LD-bin distributed within a specified region of the chromosome. Data of the LD-bins presented here are definitive haplotypes that originate from a single sperm, indicating that they are free from errors, which is typically caused by the inference from diploid genotypes. The data of LD-bin are provided by Prof. Kenshi Hayashi and Dr. Koichiro Higasa at Kyusyu University (D-HaploDB Phase II on Build 36). For detail of data, please see web page of D-HaploDB (http://finch.gen.kyushu-u.ac.jp/) (1).

1.3. GBrowse

Figure3-1 GBrowse

GBrowse in VarySysDB can be used to navigate positional relationships among HITs, HIXs, and polymorphisms. Since GBrowse is an open source architecture with various functions, users can conveniently download information from the retrieved region and upload their own data to make comparisons with the information in VarySysDB.

URL or GBrowse:

http://www.gmod.org/wiki/index.php/Gbrowse

2. Acknowledgements and Reference

Data of LD-bin (Definitive haplotype):
Koichiro Higasa and Kenshi Hayashi
(Research Center for Genetic Information, Medical Institute of Bioregulation, Kyushu University)

Advice and Discussion:
Shinsei Minoshima
(Photon Medical Research Center, Hamamatsu University School of Medicine)
Satoshi Fukuchi
(Center for Information Biology and DNA Data Bank of Japan, National Institute of Genetics, Shizuoka,)

1. **Higasa, K., Miyatake, K., Kukita, Y., Tahira, T. and Hayashi, K. (2007) D-HaploDB: a database of definitive haplotypes determined by genotyping complete hydatidiform mole samples. *Nucl. Acids Res.*, 35, D685-689.**