

# *The Database of Genomic Variants*

## User Tutorial

<http://dgv.tcag.ca/>

Updated, June 2013

# About the Database

- The objective of the Database of Genomic Variants is to provide a comprehensive summary of structural variation in the human genome. We define structural variation as genomic alterations that involve segments of DNA that are larger than 50bp. The content of the database only represents structural variation identified in healthy control samples.
- The Database of Genomic Variants provides a useful catalog of control data for studies aiming to correlate genomic variation with phenotypic data. The database is continuously updated with new data from peer reviewed research studies.

The specific aims of DGV include:

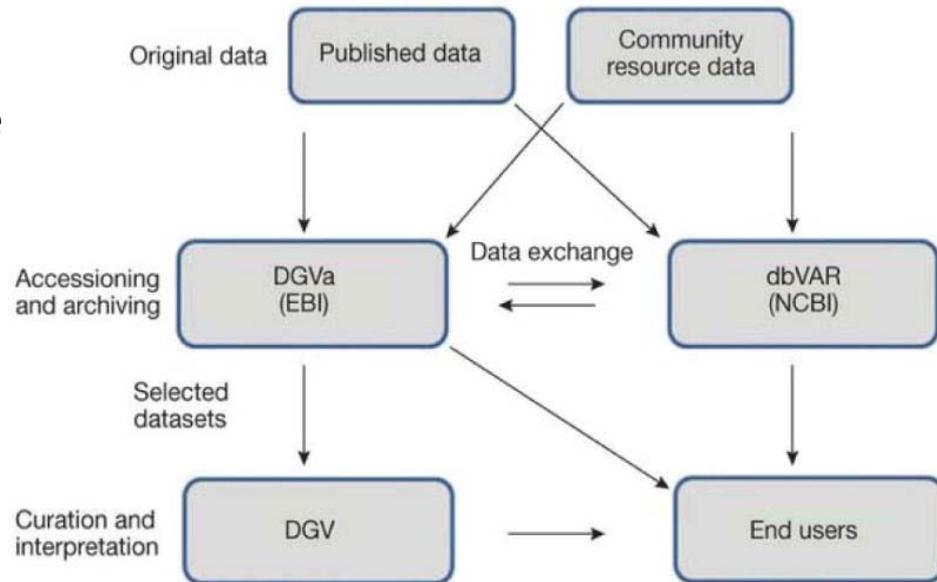
- To provide information about **genomic variants identified in control samples**
- To serve as a **resource** to both **clinical and research labs**
- To show **variation in genomic context** in a simple genome browser
- To transparently provide **access to underlying source data** (downloadable)
- To be **publicly available at no cost**

# The “New” DGV

<http://dgv.tcag.ca/>

- The Database of Genomic Variants (DGV) has been working in partnership with the new database archives at EBI (DGVa; <http://www.ebi.ac.uk/dgva/>) and NCBI (dbVar; <http://www.ncbi.nlm.nih.gov/dbvar>) to collect, organize and curate genome-wide information on copy number variation. The goal was to provide a fully integrated, standardized and accessioned set of variants.

- A schematic of how this data is generated and shared between the groups is highlighted here.



(Church DM et. al. Nat Genet. 2010 Oct;42(10):813-4 )

# Database Content

The database currently contains information from 53 different peer reviewed studies. Corresponding to ~ 265,000 CNV regions and over 1,100 inversions.

The majority of data sets in DGV are from four different types of studies:

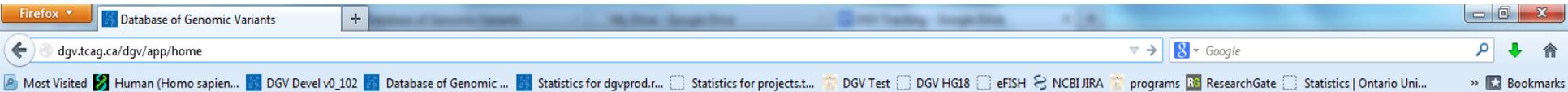
- Array based comparative genomic hybridization (CGH) and comparative intensity analysis (SNP/CNV arrays)
- Identification of deletions based on statistical analysis of SNP data
- Clone end sequence mapping
- Sequence trace mapping

What DGV is not...

- An uncurated repository of primary data
- A substitute for a well-designed control experiment
  - The database is still limited in content
  - About 6,500 individuals represented; not ethnically matched
- Due to biases in studies to date, the content is almost certainly not an accurate representation of structural variation on a population scale.

# DGV Home Page

http://dgv.tcag.ca/dgv/app/home



## Database of Genomic Variants

*A curated catalogue of human genomic structural variation*

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Keyword, Landmark or Region Search:   NCBI36/hg18 ▾

Examples: RP11-34P13; CFTR, 7q11.21; chr7:71890181-72690180

### Find DGV Variants

[by Study](#) [by Sample](#)  
[by Method](#) [by Variant](#)  
[by Platform](#) [by Chromosome](#)

### Summary Statistics

Stat	Merged-level	Sample-level
CNVs:	184148	2888526
Inversions:	238	3380

[Number of Studies:](#) 53

[News: May 2013 Update and Newsletter has been issued](#)

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Enter the Genome Browser

# Points of Entry into the Database

*D*atabase of *G*enomic *V*ariants  
A curated catalogue of human genomic structural variation

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[Genome Browser](#)   [Query Tool](#)   [Submissions](#)   [Contact Us](#)   [Training Resources](#)

**Keyword, Landmark or Region Search:**   NCBI36/hg18 ▾

**Examples:** RP11-34P13; CFTR, 7q11.21; chr7:71890181-72690180

### Find DGV Variants

[by Study](#)   [by Sample](#)  
[by Method](#)   [by Variant](#)  
[by Platform](#)   [by Chromosome](#)

Search by landmark or genomic feature

Search using the Query Tool

### Summary Statistics

Stat	Merged-level	Sample-level
CNVs:	184148	2888526
Inversions:	238	3380
<b><u>Number of Studies:</u> 53</b>		

[News: May 2013 Update and Newsletter has been issued](#)

# Genome Browser

- DGV uses the Generic Genome Browser (<http://gmod.org/wiki/GBrowseGMOD>) to provide a visual display of structural variation data.
- In addition to the structural variation data, we have provided numerous additional tracks of relevant annotations.
- The following slides will help to provide an introduction to the various features and content in our genome browser.

# Genome Browser

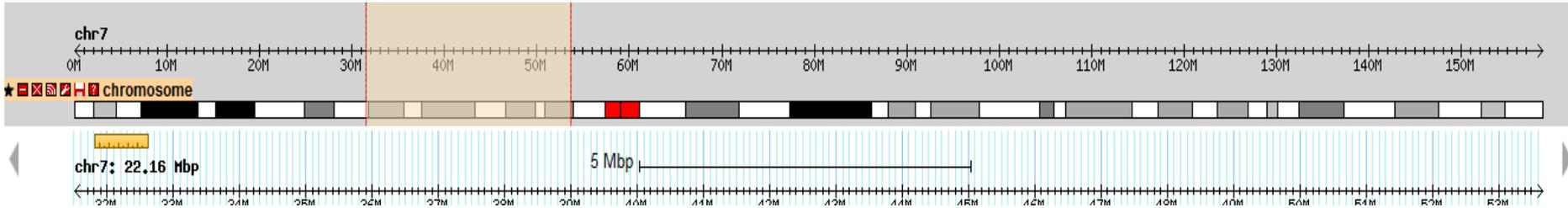
Many features of the genome browser are the same, with some functional differences and some new tools/options.

The screenshot shows the top section of the Database of Genomic Variants genome browser. At the top, the title "Database of Genomic Variants" is displayed in a stylized font, with the subtitle "A curated catalogue of human genomic structural variation" below it. A navigation menu includes "File" and "Help". The main content area shows the current view: "Genomic Variants in Human Genome (Build 36: Mar. 2006, hg18): 2 Mbp from chr5:148,499,447..150,499,447". Below this, there are tabs for "Browser", "Select Tracks", "Custom Tracks", and "Preferences". A "Search" section contains a "Landmark or Region:" input field with the value "chr5:148,499,447..150,499,447" and a "Search" button. Below the search field, there are "Examples:" and "Data Source:" sections. The "Data Source" dropdown is set to "Genomic Variants in Human Genome (Build 36: Mar. 2006, hg18)". To the right of the data source, there are "Scroll/Zoom:" controls with left and right arrow buttons, a "Show 2 Mbp" dropdown, and a "Flip" checkbox. At the bottom, there is a "Filter variants" section with a dropdown set to "study", an equals sign dropdown, and a text input field. Below the filter section are "Filter" and "Reset" buttons.

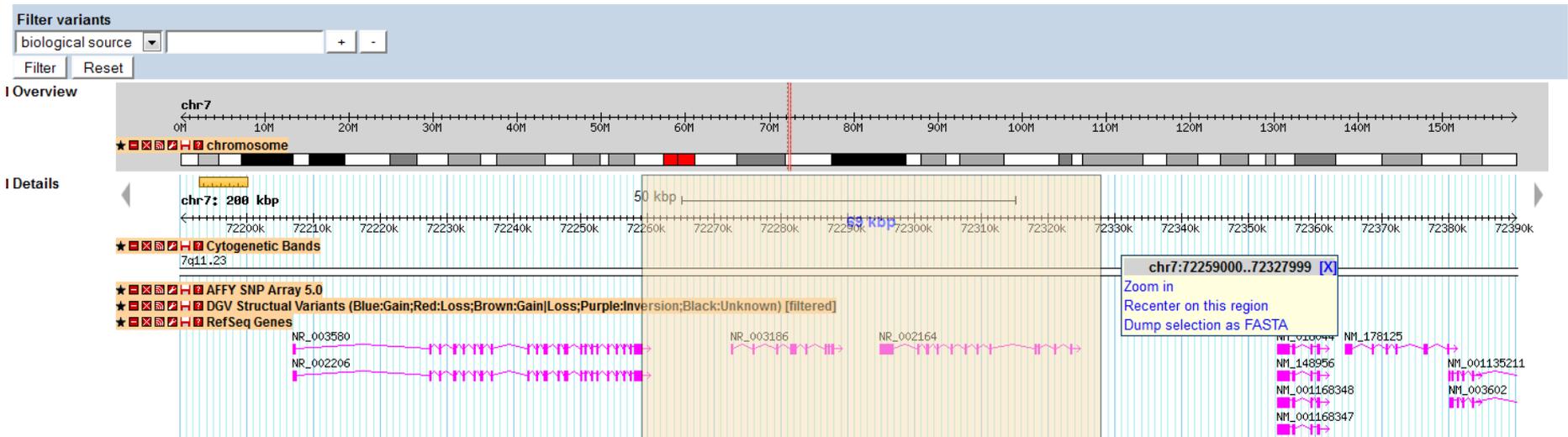
- At the top of the browser window, there are options modifying the display in the current browser
1. To turn on/off various tracks and to modify the display, chose the “Select Tracks” tab.
  2. To upload your own custom annotations, or to modify the display, chose the appropriate tabs.
  3. To filter the data displayed in the browser, to only show a subset of the variants, use the “Filter Variants” option, select the data type you want to filter by, and add a keyword to the box. To filter by greater than one data type, use the “+” key to add another row.

# Genome Browser Options

1. Click and drag on the chromosome to select a particular region



2. Click and drag on the genomic position bar to zoom in or centre on a region of interest.





# Genome Browser Options

Genomic Variants in Human Genome (Build 36: Mar. 2006, hg18): 1000 kbp from chr7:116,501,604..117,501,603

Browser Select Tracks Custom Tracks Preferences

- Show grid
- Image Width  
 600  760  980  1240
- Cache tracks
- Show tooltips

Highlight feature(s) (feature1 feature2...)  
CFTR [Clear highlighting](#)

Highlight regions (region1.start.end region2.start.end)  
chr7:116907253..117095954 [Clear highlighting](#)

Region Size (bp)  
200000

Update Appearance

In addition to adding a shaded area to the genome browser, the Preferences tab will allow you to modify the display on your computer by selecting the desired Image Width. You can Also turn the grid pattern on or off, and you may also highlight specific entries in the genome browser that you may be interested in. For example, if you input the term CFTR in the Highlight feature(s) box, you'll get the following display.



# Genome Browser Options

**Browser** **Select Tracks** Custom Tracks Preferences

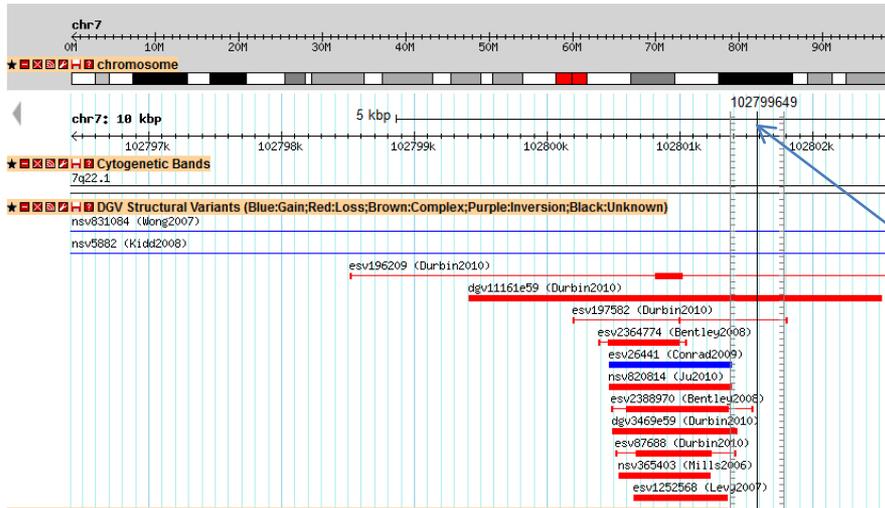
<< Back to Browser Show Favorites Only ★ Clear All Favorites ☆

Tracks

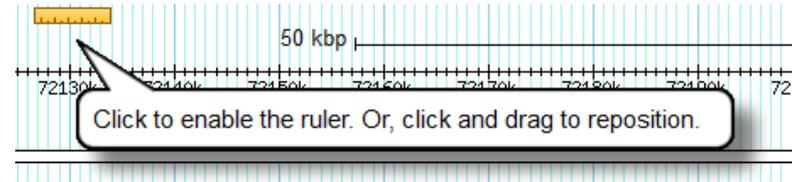
<input checked="" type="checkbox"/> Breakpoints <input type="checkbox"/> All on <input type="checkbox"/> All off	<input type="checkbox"/> Breakpoint annotations from Lam et al (Blue:Gain;Red:Loss;Indigo:Inversion;Brown:Complex) [?]	<input type="checkbox"/> Annotations from Conrad et al [?]	<input type="checkbox"/> Breakpoint annotations from Pang 2012 (Blue:Gain;Red:Loss;Indigo:Inversion;Brown:Complex) [?]
<input checked="" type="checkbox"/> CGH Arrays <input type="checkbox"/> All on <input type="checkbox"/> All off	<input type="checkbox"/> Agilent 244k [?]	<input type="checkbox"/> Clones on SMRT BAC Array [?]	
<input checked="" type="checkbox"/> CN Assays <input type="checkbox"/> All on <input type="checkbox"/> All off	<input type="checkbox"/> Applied Biosystems TaqMan Copy Number Assays [?]		
<input checked="" type="checkbox"/> Chromosome <input type="checkbox"/> All on <input type="checkbox"/> All off	<input checked="" type="checkbox"/> Cytogenetic Bands [?]	<input type="checkbox"/> Assembly [?]	<input type="checkbox"/> Gap [?]
<input checked="" type="checkbox"/> Clones <input type="checkbox"/> All on <input type="checkbox"/> All off	<input type="checkbox"/> BAC End Pairs [?]	<input type="checkbox"/> Fosmid End Pairs [?]	
<input checked="" type="checkbox"/> Disease <input type="checkbox"/> All on <input type="checkbox"/> All off	<input type="checkbox"/> ISCA Curated clinically relevant regions [?]	<input checked="" type="checkbox"/> Disease Genes (OMIM) [?]	<input type="checkbox"/> DECIPHER: Chromosomal Imbalance and Phenotype in Humans (Blue:Gain;Red:Loss;Brown:Complex;Black:NA) [?]
	<input type="checkbox"/> ISCA Clinical cytogenetic testing (Blue:Gain;Red:Loss;Brown:Complex;Black:NA) [?]	<input checked="" type="checkbox"/> DECIPHER Genomic Disorders [?]	
<input checked="" type="checkbox"/> Gene <input type="checkbox"/> All on <input type="checkbox"/> All off	<input checked="" type="checkbox"/> RefSeq Genes [?]	<input type="checkbox"/> mRNA [?]	<input type="checkbox"/> microRNA [?]
<input checked="" type="checkbox"/> General <input type="checkbox"/> All on <input type="checkbox"/> All off	<input type="checkbox"/> Personal Genome Variants (Blue:Gain;Red:Loss;Brown:Complex;Purple:Inversion;Black:Unknown) [?]	<input type="checkbox"/> SNPs [?]	
	<input type="checkbox"/> dbRIP [?]	<input type="checkbox"/> RepeatMasker [?]	
<input checked="" type="checkbox"/> SNP Arrays <input type="checkbox"/> All on <input type="checkbox"/> All off	<input type="checkbox"/> AFFY SNP Array 5.0 [?]	<input type="checkbox"/> ILMN HumanHap 300 [?]	<input type="checkbox"/> ILMN Human 660W [?]
	<input type="checkbox"/> AFFY SNP Array 6.0 [?]	<input type="checkbox"/> ILMN HumanHap 550 [?]	<input type="checkbox"/> ILMN HumanHap 1M [?]
	<input type="checkbox"/> AFFY CytoScan HD [?]	<input type="checkbox"/> ILMN HumanHap 650Y [?]	
<input checked="" type="checkbox"/> Segmental Duplications <input type="checkbox"/> All on <input type="checkbox"/> All off	<input checked="" type="checkbox"/> UCSC segmental duplications [?]		
<input checked="" type="checkbox"/> Study Variants <input type="checkbox"/> All on <input type="checkbox"/> All off	<input checked="" type="checkbox"/> DGV Structural Variants (Blue:Gain;Red:Loss;Brown:Complex;Purple:Inversion;Black:Unknown) [?]	<input checked="" type="checkbox"/> DGV Version 1 Structural Variants (Blue:Gain;Red:Loss;Brown:Complex;Purple:Inversion;Black:Unknown) [?]	<input type="checkbox"/> OPGP Affymetrix CytoHD Variants (blue=Gain; red=Loss; solidbox=AFFY_FILTER; solidline=MULTI_ALGO; dashedline=ONLY_CHAS) [?]
	<input type="checkbox"/> Supporting Variants (Blue:Gain;Red:Loss;Brown:Complex;Purple:Inversion;Black:Unknown) [?]	<input type="checkbox"/> Unannotated Studies (Blue:Gain;Red:Loss;Brown:Complex;Purple:Inversion;Black:Unknown) [?]	
<input checked="" type="checkbox"/> Overview <input type="checkbox"/> All on <input type="checkbox"/> All off	<input checked="" type="checkbox"/> Chromosome (overview)		

To select the tracks you want shown in the browser, navigate to the “Select Tracks” tab. Check the box beside the track name to turn the track on or off. To turn all the tracks on/off within a group (i.e. Disease), use the “All on” or “All off” options. The “?” beside the tracks will provide information regarding the content of the data displayed in this track.

# Genome Browser Options



Click on the ruler to add a vertical marker to align features.



File option allows users to export graphics for figures, and can access GFF tables.

The screenshot shows the 'Database of Genomic Variants' website. The header includes the title and subtitle: "A curated catalogue of human genomic structural variation". Below the header, there is a navigation bar with 'File' and 'Help' menus. A dropdown menu is open under 'File', showing options: 'Bookmark this', 'Share these tracks', 'Export as...', 'Get chrom sizes', 'Reset to defaults', and 'Examples: chr7:71890181..7269...'. The 'Export as...' option is selected, and a sub-menu is visible with options: '...low-res PNG image', '...editable SVG image', '...GFF annotation table', and '...FASTA sequence file'. At the bottom, there is a 'Data Source' dropdown set to 'Genomic Variants in Human Genome (Build GRCh37: Feb. 2009, hg19)' and a 'Scroll/Zoom' control with 'Show 10 kbp' and a 'Flip' button.

# Genome Browser Options

## Track Options

Minimize the track

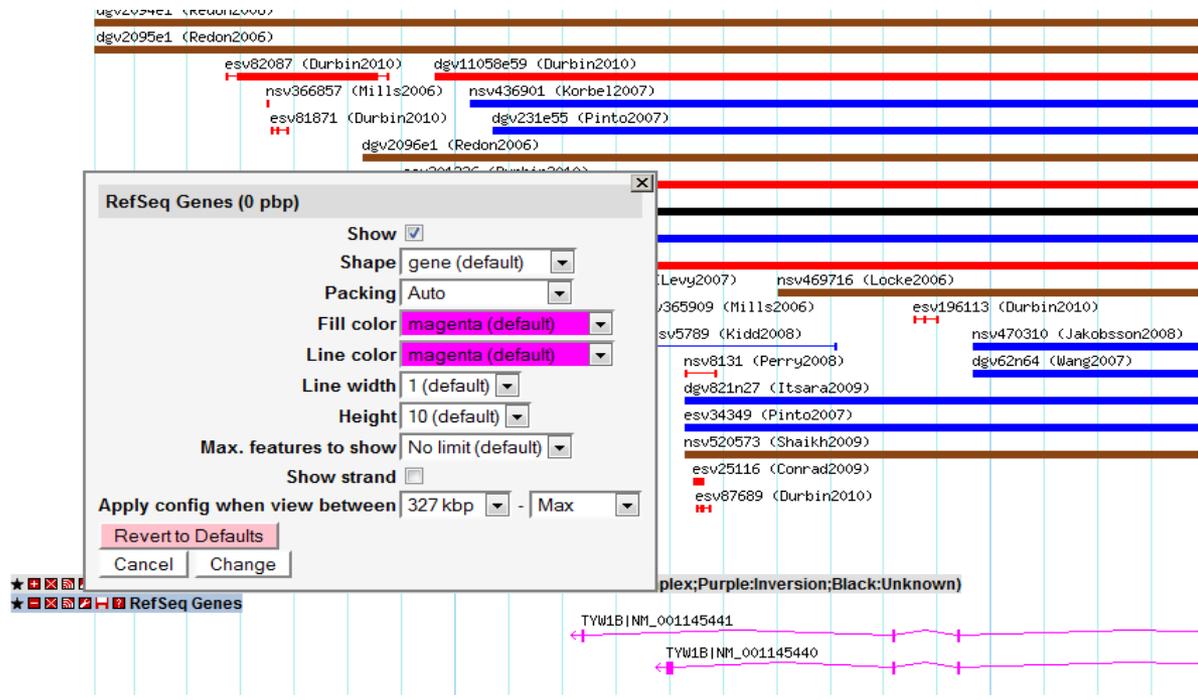
Displays information about the data  
In the track.



Close the track.

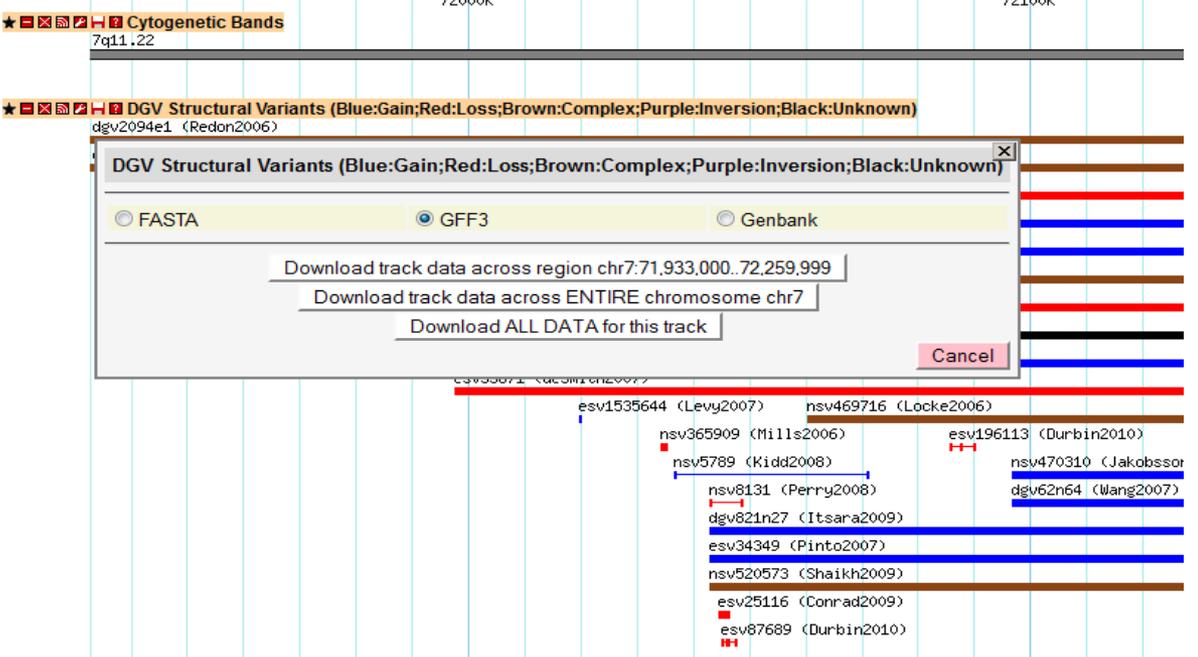
Save the data from this track  
This allows the user to download or save the underlying  
data as a GFF file.  
User may select the region displayed in the window,  
the whole chromosome or genome wide (see next slide).

Allows users to change how the data  
are displayed (see next slide).



Change Display  
(click on wrench icon)

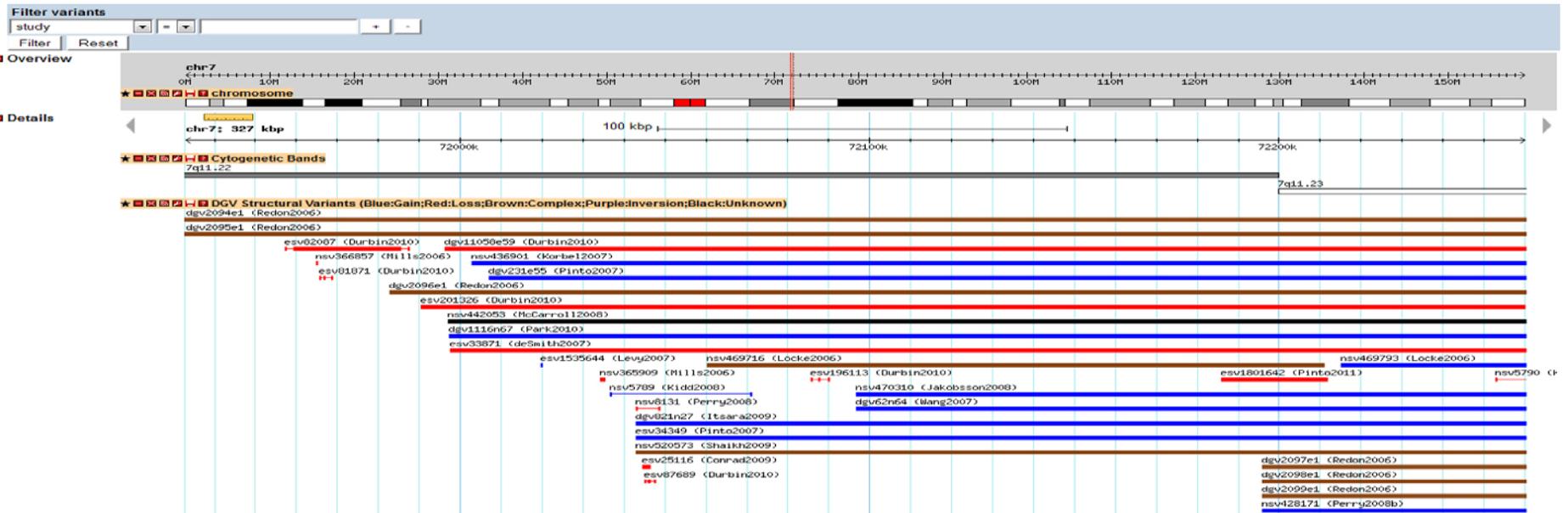
Example:  
-can force option to show strand/direction for genes regardless of the size of the window being used.



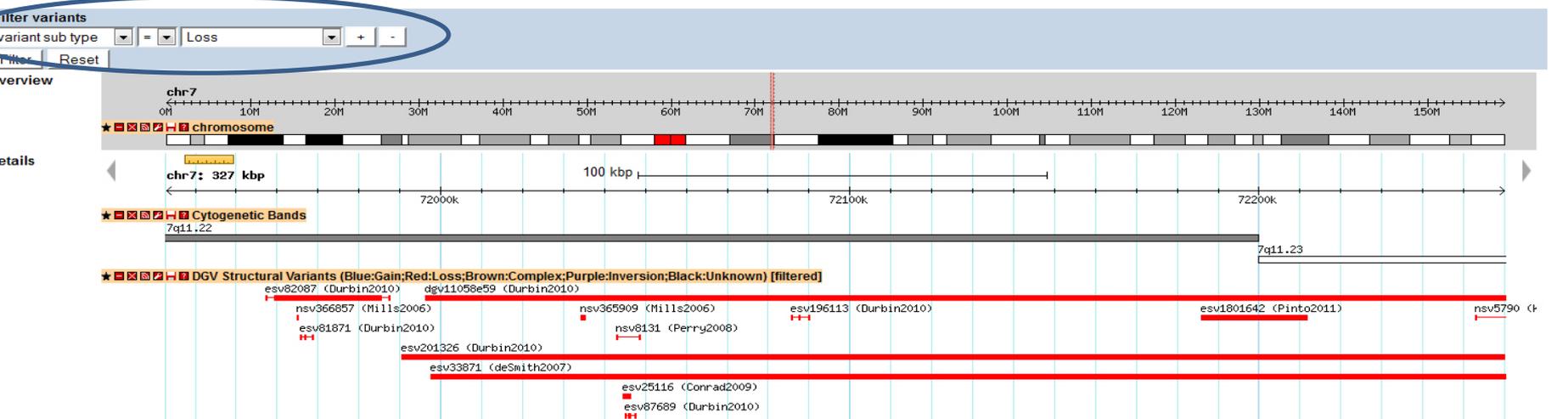
Save the Data  
(click on "save icon")

Example:  
-quick and easy method to generate a list of genes or other items that fall in your region of interest.

# Applying Filters to DGV Variants in Gbrowse



If you were only interested in comparing your data to deletions found in DGV, you can filter the data displayed in the browser by selecting the option in the “Filter Variants” box at the top.



# DGV Content

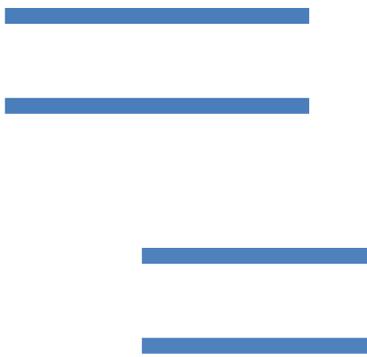
- With the release of the new DGV, there are a number of updates and changes to the content and representation of the data.
- Stable, long term accessions have been assigned and a controlled, and structured vocabulary of terms used to describe the data have been developed.
- The following slides will help introduce DGV users to some of these changes.

# New DGV Accessions

- Each study from DGV has been accessioned by one of the two groups; dbVAR have assigned nsv/nssv accessions, while DGVa has assigned esv/essv accessions. An nsv is an NCBI structural variant, and an nssv is an NCBI supporting structural variant. An esv is an EBI structural variant, and an essv is an EBI supporting structural variant.
- Supporting structural variants ("ssv") are typically sample level variants, where each ssv represents the variant called in a single sample/individual. In a few studies the ssv represents the variant called by a single algorithm. If multiple algorithms were used, overlapping ssv's from the same individual would be combined to generate a sample level sv. If there are many samples analysed in a study and if there are many samples which have the same variant, there will be multiple ssv's with the same start and end coordinates. These sample level variants are then merged and combined to form a representative variant that highlights the common variant found in that study. This is called a structural variant ("sv") record.
- DGV has always provided this type of summary/merged variant and we have continued to do so in cases where there are a number of overlapping variants that are almost identical, but may be slightly different due to the inherent variability between experiments. If there are clusters of variants within a single study that share at least 70% reciprocal overlap in size/location, we will merge these together and provide an accession record that has our internal "dgv" prefixed identifier. The dgv merged identifier has been updated to help improve the consistency and stability across updates of the database. The format of the dgv accession is now "dgv + variant number + study accession. As an example, the first merged variant from the Shaikh et al 2009 study (study accession=nstd21) would be dgv1n21. The second merged variant would be dgv2n21 and so forth.

# Examples of DGV Variant Types

## Example 1

DGV merged variant -merge of nsv/esv variant regions which overlap		dgv333n21 = nsv111+ nsv222 Samples:NA15510+NA10851+NA12291+NA18291
nsv/esv = variant regions -merge of nssv/essv variant calls which overlap		nsv111 = nssv123+ nssv321 Samples:NA15510+NA10851  nsv222 = nssv567+ nssv765 Samples:NA12291+NA18291
nssv/essv = variant calls -represent the supporting or sample level calls		nssv123 : Sample=NA15510  nssv321 : Sample=NA10851  nssv567 : Sample=NA12291  nssv765 : Sample=NA18291

# Examples of DGV Variant Types

## Example 2

DGV merged variant  
-merge of nsv/esv variant  
regions which overlap

No overlapping nsv/esv variants  
No DGV merged variant is created

nsv/esv = variant regions  
-merge of nssv/essv variant calls  
which overlap



nsv482 = nssv164+ nssv955  
Samples:NA15581+NA12761

nssv/essv = variant calls  
-represent the supporting  
or sample level calls



nssv164 : Sample=NA15581



nssv955 : Sample=NA12761

# Examples of DGV Variant Types

Example 3 (variant called in only 1 sample)

DGV merged variant  
-merge of nsv/esv variant  
regions which overlap

No overlapping nsv/esv variants  
No DGV merged variant is created

nsv/esv = variant regions  
-merge of nsv/esv variant calls  
which overlap.  
-there are no overlapping variants therefore  
the variant region is the same as the variant call.



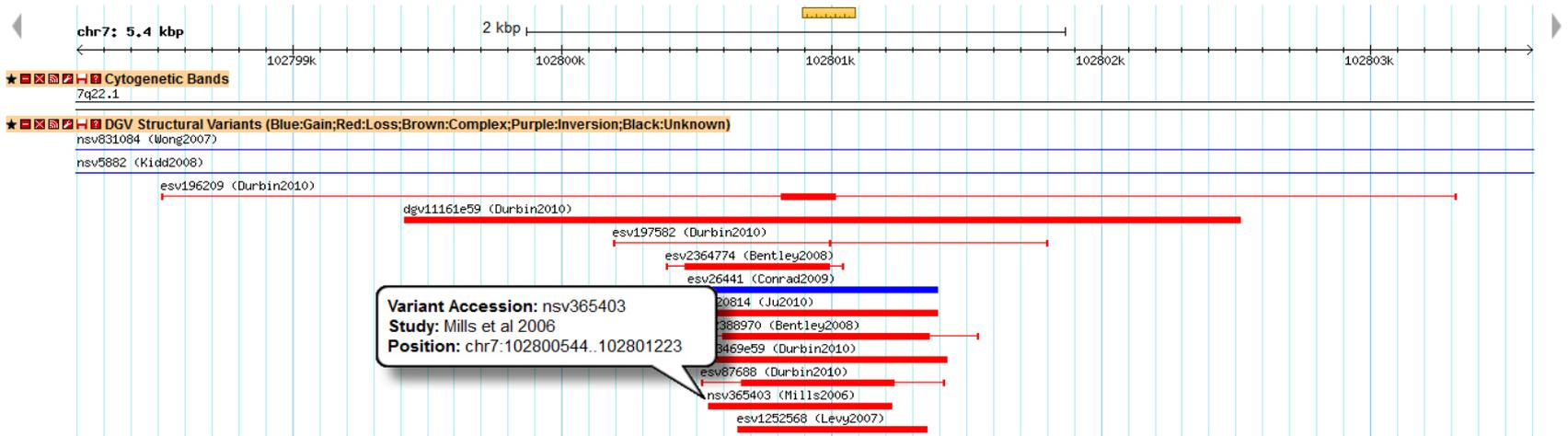
nsv372 = nsv862  
Samples:NA13781

nsv/esv = variant calls  
-represent the supporting  
or sample level calls.  
-in this case, the variant was  
only detected in one sample.



nsv862 : Sample=NA13781

# DGV data



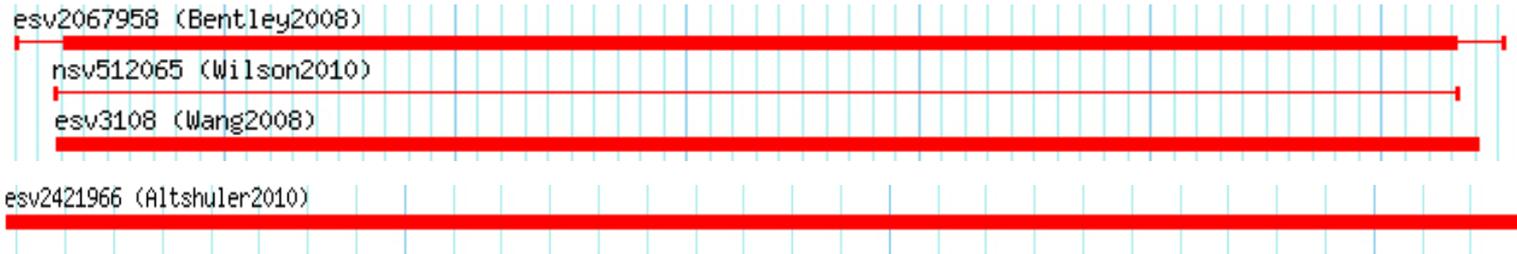
<b>Internal ID</b>	117790			
<b>Landmark</b>				
<b>Location Information</b>	<b>Type</b>	<b>Coordinates</b>	<b>Assembly</b>	<b>Other Links</b>
		<a href="#">chr12:109632315..109632415</a>	hg19	<a href="#">UCSC Ensembl</a>
		<a href="#">chr12:108116698..108116798</a>	hg18	<a href="#">UCSC Ensembl</a>
<b>Cytoband</b>	12q24.11			
<b>Allele length</b>	<b>Assembly</b>	<b>Allele length</b>		
	hg19	101		
	hg18	101		
<b>Variant Type</b>	CNV Deletion			
<b>Copy Number</b>				
<b>Allele State</b>	Homozygous			
<b>Allele Origin</b>	Not tested			
<b>Probe Count</b>				
<b>Merged Status</b>	S			
<b>Merged Variants</b>	<a href="#">esv1456600</a>			
<b>Supporting Variants</b>				
<b>Samples</b>	HuRef			
<b>Known Genes</b>	<a href="#">ACACB</a>			
<b>Method</b>	SNP_array			
<b>Analysis</b>	analysis_type=Sequence Alignment			
<b>Comments</b>				
<b>Reference</b>	Levy et al 2007			
<b>Pubmed ID</b>	<a href="#">17803354</a>			
<b>Accession Number(s)</b>	<a href="#">esv3652338</a>			
<b>Frequency</b>	<b>Sample Size</b>	2		
	<b>Observed Gain</b>	0		
	<b>Observed Loss</b>	1		
	<b>Observed Complex</b>	n/a		
	<b>Frequency</b>	n/a		

Users can mouse over the variations to obtain summary information, or can click on the image to go to the variants detail page (below).

Additional details such as the allele length, number of gains/losses and the allele state (heterozygous/homozygous) have been added (when available).

# DGV data

There are a few different types of images used to display structural variants in the genome browser. The different types are shown below with a corresponding description.

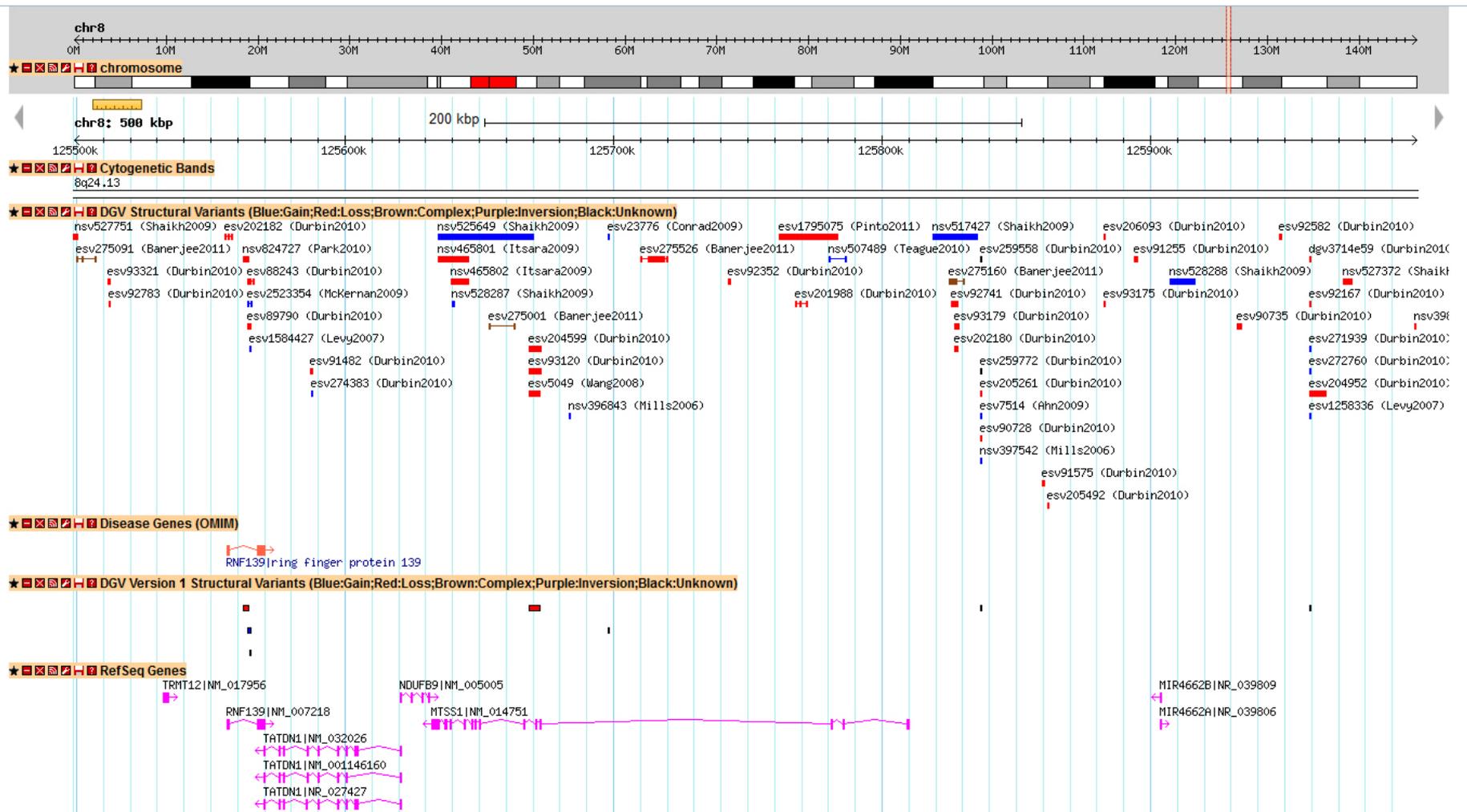


To accurately reflect the inherent differences in the resolution of different approaches, the assignment of boundaries for the structural variants has been updated and the display has been updated.

1. Variant boundaries may be assigned a start and stop position. This will be common for sequencing based studies where the actual breakpoints are known (example = esv3108; Wang2008).
2. An outer start and outer stop coordinate will be assigned for studies that use a mapping based strategy (paired-end, optical mapping) or BAC clone approach where the variant boundaries are likely overestimated, and the maximum or outer boundaries are known, but the actual variant likely resides somewhere within this region. (example = nsv512065; Wilson2010, Paired-end Mapping)
3. The inner start and inner stop coordinates are used for studies where the boundaries are likely underestimated and may include oligo (probe) based CGH experiments. The actual boundary of the variant would likely reside somewhere between the last positive probe on the array and the next neighbouring negative probe (example = esv2421966; Altshuler2010).
4. For some studies, a combination of outer start-outer stop and inner start-inner stop coordinates are described when information is available on the boundary regions (example = esv2067958, Bentley2008).

# DGV data

The original (DGV Version 1) and new DGV structural variation data are displayed for comparison.



# Query Tool

- The query tool is a set of inter-related tables containing all the data from the studies included in DGV. Options to search and filter the data across studies has been developed.
- Each table represents a specific category of data for all of the studies in the database.
  - The Study table contains general information about each study, including the number of samples and variants detected, with links to the published report in PubMed.
  - The Variant Table contains a list of all the variants reported in all the studies and their respective attributes.
  - The Sample Table contains information about the samples that were analysed in each study, including gender, ethnicity and source (where available).
  - The Method Table contains summary information on the approaches used, including details on the specific methods employed.
  - The Platform Table has specific information on the experimental approaches taken in each study used to identify and validate variants.
  - The Analysis Table has detailed information about the tools and algorithms used to analyse the data when generating the set of structural variants
- Options to select, filter and manage the data have been developed, providing the option to customize the output based on a number of terms and attributes.
- Searches and filters can be applied to view information from only a single study, or can be applied across studies to find all information related to a specific term. For example you could retrieve all variants from all studies identified in a single sample. Similarly, if you would like to obtain all the variants identified in a specific population (HapMap Yoruba), or derived from a single approach this would be an option as well.

# Query Tool

## Database of Genomic Variants

A curated catalogue of human genomic structural variation

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accession =  + -

[Study](#) [Variants](#) [Samples](#) [Methods](#) [Platforms](#) [Analyses](#)

Users can filter data based on the options presented in the tabs above.

## Database of Genomic Variants

A curated catalogue of human genomic structural variation

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Use ~ to perform a wildcard search

study ~  + -

[Study](#) [Variants](#) [Samples](#) [Methods](#) [Platforms](#) [Analyses](#)

Show 50 entries  
Showing 1 to 3 of 3 entries (filtered from 53 total entries) [Copy](#) [Print](#) [CSV](#) [Excel](#) [PDF](#) [FAQ](#)

accession	study	pubmed id	sample size	variant count
nstd2	Kidd et al 2008	18451855	9	18243
nstd35	Kidd et al 2010	20440878	9	39
nstd47	Kidd et al 2010b	21111241	9	1636

Showing 1 to 3 of 3 entries (filtered from 53 total entries) [First](#) [Previous](#) 1 [Next](#) [Last](#)

Example, where a user has selected to filter the database to obtain studies where the primary author is “Kidd”.

Use the “+” button to add additional terms  
Use the “-” button to remove search terms

# Query Tool

Users can save, copy or print the output using these options.

chromosome = Y + -  
assembly = NCBI36/hg18 + -  
Filter query Reset

Study Variants Samples Methods Platforms Analyses

Show 50 entries  
Showing 1 to 50 of 3,507 entries (filtered from 6,407,415 total entries)

chromosome	start	stop	study	variant accession	M=Merged, S=Sample Call	ethnicity	gender	assembly	variant type	variant subtype
Y	12101151	12209550	Durbin_et_al_2010	<a href="#">essv287618</a>	S	YRI	Male	NCBI36/hg18	CNV	Loss
Y	10519992	10654530	Durbin_et_al_2010	<a href="#">essv287075</a>	S	YRI	Female	NCBI36/hg18	CNV	Loss
Y	11926654	11943340	Ahn et al 2009	<a href="#">essv31307</a>	S	KOREAN	Male	NCBI36/hg18	CNV	Loss
Y	57393261	57399506	Durbin_et_al_2010	<a href="#">essv287096</a>	S	CEU	Female	NCBI36/hg18	CNV	Loss
Y	57229393	57236795	Durbin_et_al_2010	<a href="#">essv2301422</a>	S	CEU	Male	NCBI36/hg18	CNV	Loss
Y	2391693	2402131	Durbin_et_al_2010	<a href="#">essv287759</a>	S	CEU	Male	NCBI36/hg18	CNV	Loss
Y	22973854	23049013	Teague et al 2010	<a href="#">nsv508822</a>	S			NCBI36/hg18	CNV	Insertion
Y	5103369	5104505	Durbin_et_al_2010	<a href="#">essv287769</a>	S	CEU	Female	NCBI36/hg18	CNV	Loss
Y	12111044	12115461	Ahn et al 2009	<a href="#">essv28495</a>	S	KOREAN	Male	NCBI36/hg18	CNV	Loss
Y	10602703	10604281	Durbin_et_al_2010	<a href="#">essv252015</a>	M			NCBI36/hg18	CNV	Loss
Y	20837530	20837530	Levy et al 2007	<a href="#">essv4016907</a>	S	Caucasian	Male	NCBI36/hg18	CNV	Insertion
Y	57404391	57404705	Durbin_et_al_2010	<a href="#">essv287111</a>	S	CEU	Female	NCBI36/hg18	CNV	Loss
Y	21745263	21777102	Durbin_et_al_2010	<a href="#">essv151872</a>	S			NCBI36/hg18	CNV	Loss
Y	11921334	11944992	Ahn et al 2009	<a href="#">essv32152</a>	S	KOREAN	Male	NCBI36/hg18	CNV	Loss
Y	57264599	57265327	Durbin_et_al_2010	<a href="#">essv2301601</a>	S	CHB	Male	NCBI36/hg18	CNV	Loss
Y	57394520	57395721	Durbin_et_al_2010	<a href="#">dqv12705e59</a>	M			NCBI36/hg18	CNV	Loss
Y	57402930	57403514	Durbin_et_al_2010	<a href="#">essv287473</a>	S	CEU	Female	NCBI36/hg18	CNV	Loss
Y	57398842	57403535	Durbin_et_al_2010	<a href="#">essv152121</a>	S			NCBI36/hg18	CNV	Loss
Y	11924997	11942375	Ahn et al 2009	<a href="#">essv8763</a>	S	KOREAN	Male	NCBI36/hg18	CNV	Loss
Y	57263602	57264731	Durbin_et_al_2010	<a href="#">essv2301329</a>	S	YRI	Male	NCBI36/hg18	CNV	Loss
Y	10630453	10630629	Durbin_et_al_2010	<a href="#">essv287220</a>	S	CEU	Female	NCBI36/hg18	CNV	Loss
Y	20849751	20852550	Durbin_et_al_2010	<a href="#">essv151835</a>	S			NCBI36/hg18	CNV	Loss

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Another example where the user has selected variants on chromosome Y, mapped to assembly version hg18. To filter across all tables, select the “Filter query” button. If you then select the “Study” tab, only studies which have identified variants on the Y chromosome are included in the results.

# Query Tool

accession = estd59 + -  
Filter query Reset

Study Variants Samples Methods **Platforms** Analyses

Show 50 entries  
Showing 1 to 5 of 5 entries (filtered from 108 total entries)

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study	platform name	platform type	platform version	geo accession	arrayexpress accession
Durbin_et_al_2010	454 GS FLX				
Durbin_et_al_2010	ABI SOLiD System				
Durbin_et_al_2010	Illumina Genome Analyzer II				
Durbin_et_al_2010	Illumina Genome Analyzer II and 454 GS FLX				
Durbin_et_al_2010	Illumina Genome Analyzer II and ABI SOLiD System				

Showing 1 to 5 of 5 entries (filtered from 108 total entries) First Previous 1 Next Last

Additional details on the platforms used, and the specific types of analyses performed are available as shown for the 1,000 genomes pilot dataset (Durbin2010).

accession = estd59 + -  
Filter query Reset

Study Variants Samples Methods Platforms **Analyses**

Show 50 entries  
Showing 1 to 34 of 34 entries (filtered from 172 total entries)

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study	analysis type	analysis tool	analysis description	reference type	reference	reference description
Durbin_et_al_2010	Detection	NovelSeq_And_Cortex	Sequencing platform: Combination of Whole Genome Illumina and 454. Mapping algorithm: CORTEX on reference and NovelSeq. Type of computational approach: read pair mapping.	Ref_sequence	NCBI36/hg18	
Durbin_et_al_2010	Detection	BLAT	Sequencing platform 454. Mapping algorithm: BLAT. Type of computational approach: split read alignment.	Ref_sequence	NCBI36/hg18	
Durbin_et_al_2010	Detection	SPANNER	Sequencing platform: Whole Genome Illumina. Mapping algorithm: SPANNER. Type of computational approach: read pair mapping.	Ref_sequence	NCBI36/hg18	
Durbin_et_al_2010	Detection	MRFAS	Sequencing platform: Whole Genome Illumina. Mapping algorithm: mRFAS. Type of computational approach: read depth analysis.	Ref_sequence	NCBI36/hg18	

# Help and Support

<http://dgv.tcag.ca/dgv/app/contacts>

If you have any questions while using the DGV beta site, or if you notice any errors or bugs in the database, please contact the DGV Team at your earliest convenience.

Email: [dgv-contact@sickkids.ca](mailto:dgv-contact@sickkids.ca).

If you would like to receive updates and notifications about DGV, please sign up for our newsletter

Thank you for taking the time to test and use the database, your assistance and feedback is greatly appreciated.

Sincerely,  
The DGV Team

