Welcome to Release 3.0 of the DBD Genes Database

This release of the DBD Genes Database introduces a refreshed look and new features, expanding the amount of information available, and facilitating the ability to analyze it. This document will describe the new features in this release, including:

- A new Site Search Bar for easier searching of genes within the database
- The inclusion of classifications from ClinGen, GenCC, SFARI, and DDG2P for comparison
- More powerful searching and filter capabilities
- Additional data export formats

GENE SEARCH

This release introduces the Site Search Bar, located in the top site banner as shown below:

To use the Site Search Bar, simply begin typing a gene symbol or HGNC ID. For every character typed, the top matches are displayed. The more characters typed, the further refined the list will be. Once a desired gene is displayed, simply select from the list to view the LoF or Missense detail page for that gene.
The returned search matches will only return the top matches for genes within the DBD Genes Database. If a gene is not present in the database, it will not be displayed, even if the entire symbol name has been entered.

**TABLE CHANGES**

A major component of this release is a new data management system and table display designed to accommodate more content and provide more functionality in an intuitive manner. Compared to previous versions of the DBD Genes Database, nearly all aspects of the displayed table have changed without any loss of previous functionality. This section will review all changes.

**GENE INFORMATION**

As in previous releases, the DBD Genes Database maintains separate gene listings for those with LoF and Missense variants. However, in this release, it is now easier to determine which gene on the LoF list also has additional Missense variant data, and vice versa. Genes with both LoF and Missense variants will have an information indicator (ตรา Vaults) next to their names in the list, as shown below:
As always, clicking on the gene symbol name will take you to the respective gene detail page. However, as a shortcut, clicking on the information indicator icon for a LoF gene will take you to the Missense gene detail page. Similarly, clicking on the information indicator icon for a Missense gene will take you to the LoF gene detail page.

Any row can be expanded for more gene level information by simply click on any white area in that row. To contract the row, simply click again.
While previous releases of the DBD Genes Database displayed disorder data in a relative bar chart format which proved hard to navigate, especially on touch devices, this new release presents disorder data in a more traditional sortable and searchable column format.

The new format allows you to resort on a particular disorder using standard column sort controls. For example, if you are primarily interested in genes associated with Epilepsy, you can sort on the Epilepsy (EP) column by decreasing number of cases:

This new release of the DBD Genes Database also allows you to view only genes associated with disorders of interest. Next to the “Disorders” title in the table header is an icon shaped as a funnel. Clicking this will bring up a selector list as shown below:
Using this menu, you may deselect and select any disorder simply by mousing over the menu item and clicking on it. Once you are done, you can hide the menu again by clicking on the funnel icon again, or anywhere outside the menu. In the example below, we moused over the attention deficit hyperactivity disorder (ADHD) to hide from the table any genes only associated with that disease.

Note that genes will remain displayed if they are associated with any disorders still selected. For example, unselecting Intellectual Disability (ID) will not remove ACACA from the display, as the gene is also associated with autism spectrum disorder (ASD). However, unselecting both Intellectual disability (ID) and autism spectrum disorder (ASD) will remove ACACA from the display.
Filtering by disorders is not the only options available in the DBD Genes Database. As in previous releases, you could also filter based on Tier classifications.

**TIERS**

DBD Genes Database Tiers classifications are an important analytical tool, and as such are much more prominently displayed within the display table, both in summary and in row form. With this new release, hiding and showing genes associated with one or more specific Tiers is now as easy as clicking on the Tier buttons:

For example, to remove all Tier 4 and AR genes from the display, simply click on the Tier 4 and AR buttons. Notice that the Tier selection is fully integrated with Disorder Selection. In our example, the display now lists only those Tier 1, Tier 2, and Tier 3 genes associated with disorders other than ID and ASD. You can confirm this not only visually by the genes listed in the table, but also by the Active Filers monitor.
To restore Tier 4 or AR genes, simply click on the associated Tier button again. Similarly, to restore ID and ADSD disorders, simply open the Disorders Filter menu (▼) and reselect those disorders.

**Important:** Note that refreshing the browser will clear all active filters. A planned future release of the DBD Genes Database will introduce features allowing you to save and restore settings.

**VIEWING CLASSIFICATIONS**

This release of the DBD Genes Database also allows you to display current ClinGen, GenCC, SFARI, and DDG2P classifications for comparison purposes. By default, the table loads in the “View Disorders” mode, as we have previously seen. To switch to the “View Classifications” mode and view these classifications, click on the “View Classifications” button.

To further refine the display to your choosing, you can resort the listing based on the classification strength using the standard column sort capabilities. Each of the classification strings are links to the source’s gene page, thus to understand more about why gene ABCC9 is classified as Limited by ClinGen, simply click on the Limited string in the
ClinGen column for that gene. To return to the “View Disorders” mode, simply click on the “View Disorders” button.

Sometimes, having two predefined table views is simply not accommodating enough to satisfy the needs of desired tasks. In those cases, you can also mix and match columns from either view for a total custom display, as discussed in the next section.

**HIDING AND SHOWING COLUMNS**

At times, you want to customize the display in ways not covered by the standard view options. Previous releases of the DBD Genes Database allowed you to select which columns to show and hide, and this release continues the tradition. To bring up the column selector menu, click on the “Show/Hide More Columns” button on the table toolbar shown below.

There are no limits to which columns can be displayed or hidden, however, there must always be at least one column displayed. You can mix columns from the “View Disorders” mode and the “View Classifications” mode as desired. In the example below, we’ve eliminated a few columns from the standard “View Classifications” mode, including the ID disorder, then added the classifications for GenCC and SFARI:
Note that while there are no limits to which column you can show, and you can easily show all columns, the physical size of your display and web browser may limit available space. In such cases, the table will automatically add a horizontal scroll ability. You can elect to utilize the scroll function, or if possible, widen your browser window to accommodate the additional data.

COLUMN SEARCHING

A powerful new feature for the DBD Genes Database is the ability to limit a string search to just a specific column. This feature is called “Column Search” and is activated from the table toolbar by clicking on the “Column Search” button.

To search any one column, for example the gene ACACA, you can simply enter “ACACA” in the search box below the Gene column title. However, the full power in column searching lies in the ability to combine multiple columns. For example, to show genes that are Tier 2 and have 1 case of Epilepsy, you can enter “2” in the Tier column search box and “1” in the EP search box as shown below. Note that each box is a text search, thus “1” will also match the “10” of CNKSR2. However, it is a way to quickly refine the table display, and you can always add additional column search values to further eliminated unwanted genes.
Column Search is completely integrated Disorder and Tier Filters discussed previously. In the above example, while clicking on a Tier button would be redundant, you can use the Disorder Filter to further refine the display and show only those Tier 2 genes with 1 case of Epilepsy (EP) and any number reported cases of autism spectrum disorder (ASD).

**TABLE EXPANSION**

Another new feature of the DBD Genes Database is the ability to expand the view of the table display to the full size of the browser. This is especially useful for presentations and documentation. To expand the table size at any time, simply click on the “Fullscreen” button in the table toolbar.
To restore the table back to its original size, simply click on the “Fullscreen” button again.
DATA EXPORT

While previous releases allowed a limited selection of data export formats, this new release of the DBD Genes Database more than double the options, bringing the complete set of choices to:

- JSON
- XML
- CSV (Comma Separated Values)
- TXT (Text)
- SQL
- Excel
- PDF

To choose the desired format and export the visible contents of the data table, click on the “Export data” button in the table toolbar:
As in previous releases, the export will only include those rows and columns visible within the table. In the case of multiple pages, it will only export the current page. If the intent is to export all selected data from the table, even if it is on a later page, then simply change the “Rows per Page” selector to “All”. If you only need a sample of data, such as for demonstration or presentation purposes, the setting the “Rows per Page selector to the desired number may be more applicable. The “Rows per Page” selector is located along the right, immediately above the table, as shown below.
While previous releases of the DBD Genes Database has evolved the display of a single gene, this release further refines the display for a more structured and easier to view. For the main Summary page, no additional content is added, however the view is more structured as show below:

Again, the Site Search Bar has been added, making it easier to navigate from gene to gene without continuously revisiting the listing pages.

Perhaps the most significant enhancement has been made to the “Cases” tab, where same table management system has been implemented, including powerful filter and search capabilities. In particular, notice the same Filter icon (🔍) buttons located within the Variant Type, Inheritance, and Method column titles.
Each of these filters behaves exactly like the Disorders Filter we saw earlier, with each selection menu content relevant to its column. And like we saw earlier; each filter is fully integrated with other filters on the page and with the Column Search feature.

Using the more expansive content provided by the ADNP gene, the below example illustrates how to view only cases that have a Variant Type of “Nonsense”, a Disorder which includes ID, and utilized the Whole Exome Sequencing Method.