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Welcome to Susan

We would like to introduce a new member of our team, Susan Tweedie, who joined the project as a nomenclature advisor in December. Susan studied Genetics at the University of Glasgow and completed a molecular biology PhD at University College London. Following postdoctoral research on a variety of topics including parasitology and eukaryotic DNA methylation, she left the lab and completed an MRes in Bioinformatics at the University of York. Most recently she has worked as a curator at the Drosophila model organism database FlyBase where she was responsible for annotating gene function using the Gene Ontology and curating fly models of human disease.

New improved Gene Families (beta release)

Here at the HGNC we have been working hard over the past few months to develop new improved gene family pages, which are now available as a beta release. We would very much like all of our newsletter readers to visit these new pages over the next six weeks and provide us with feedback. In the future the gene family pages will also be incorporated into our new Search, as described below.

Here is a summary of the improvements we have made:

Gene family index

Our gene family index is now ordered alphabetically according to family name, with root symbols shown in a separate column. Results are paginated and the user can choose to alter the number of results per page. Typing in either search box narrows down the results based on the input, as shown in the example below.

Fig. 1 The new gene family index
New gene family pages

Our gene family pages have also been redesigned to provide more information, conform to a more standardised format, and make it easier to browse between families. An example gene family page for *Cholinergic receptors, muscarinic* is shown below.

**Fig. 2 The new Gene Family page for “Cholinergic receptors, muscarinic”**

### Gene Family: Cholinergic receptors, muscarinic (CHRm)

*Also known as: “Muscarinic acetylcholine receptors”, “Muscarinic receptors”, “mAChRs”*

*A subset of: Airline receptors, Cholinergic receptors*

#### Gene family hierarchy map

*Muscarnic acetylcholine receptor:* Muscarinic receptors, or mAChRs, are acetylcholine receptors that form G protein-receptor complexes in the cell membranes of certain neurons and other cells. They play several roles, including acting as the main end-receptor stimulated by acetylcholine released from postganglionic fibers in the parasympathetic nervous system. Muscarinic receptors were named as such because they are more sensitive to muscarine than to nicotine. Their counterparts are nicotinic acetylcholine receptors (nAChRs), receptor ion channels that are also important in the autonomic nervous system. Many drugs and other substances (for example pilocarpine and scopolamine) manipulate these two distinct receptors by acting as selective agonists or antagonists. [Source: Wikipedia]

The mapped domains of **P11229**, encoded by the *CHRm1* gene, an example gene within the family. [Source: Pfam & UniProt]

### Genes contained within the family

<table>
<thead>
<tr>
<th>Approved Symbol</th>
<th>Approved Name</th>
<th>Previous Symbols</th>
<th>Synonyms</th>
<th>Chromosomes</th>
</tr>
</thead>
<tbody>
<tr>
<td>CHRm1</td>
<td>cholinergic receptor, muscarinic 1</td>
<td></td>
<td></td>
<td>15q12-q13</td>
</tr>
<tr>
<td>CHRm2</td>
<td>cholinergic receptor, muscarinic 2</td>
<td></td>
<td>7q35-q36</td>
<td></td>
</tr>
<tr>
<td>CHRm3</td>
<td>cholinergic receptor, muscarinic 3</td>
<td></td>
<td>1p23</td>
<td></td>
</tr>
<tr>
<td>CHRm4</td>
<td>cholinergic receptor, muscarinic 4</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>CHRm5</td>
<td>cholinergic receptor, muscarinic 5</td>
<td></td>
<td>11p12-11.2</td>
<td></td>
</tr>
<tr>
<td>CHRm6</td>
<td>cholinergic receptor, muscarinic 6</td>
<td></td>
<td>15p36</td>
<td></td>
</tr>
</tbody>
</table>

### Downloads

Current set only

### Gene family names, IDs and aliases

Each gene family now has a unique numerical ID that forms the last part of the gene family page URL to aid linking and downloading. For example the numerical ID for “Cholinergic receptors, muscarinic” is 180. Each family also has a unique gene family name and, where a family has a root symbol, this is shown in
parentheses next to the name. Please note that not all gene family pages equate to a particular set of genes with the same root symbol; in these cases only a
gene family name is displayed. Other commonly-used gene family names and abbreviations are listed following the text "Also known as:" e.g. "Cholinergic
receptors, muscarinic" are also known as "Muscarinic acetylcholine receptors", "Muscarinic receptors" and "mAChRs".

Gene family hierarchy map

The new gene family pages provide an improved display of curated hierarchical relationships between families and allow users to browse easily through each
hierarchy. As shown in Fig. 2 all gene families that fall into hierarchies include a "Gene family hierarchy map". Clicking on any gene family within the map opens a
pop-up that contains a link to that particular gene family page and highlights the current gene family and its direct relatives, and connects them with a red outline.
Users can move each gene family within the page by clicking and dragging the box. In addition to the map, text links to any related families are provided within the
page. For example, in Fig 2 there are direct links to the Amine receptors and Cholinergic receptors.

Gene family descriptions

Many of our gene family pages now contain a description of the family. These are often from Wikipedia (as shown in Fig 2) or UniProt (e.g. Integrins), in which
case the source is clearly marked with a link through to the original page; in some cases the descriptions have been written by HGNC curators and these can be
identified by [Source: HGNC] (e.g. IGH orphans); if they come from another source this will be clearly displayed within square brackets.

Example gene mapped domains graphic

Where gene family members share a particular protein domain we often show a graphical display of the protein domain structure for an example gene family
member, which is sourced from Pfam via UniProt ID. In Fig. 2 the domain structure is shown for the product of the CHRM1 gene. Clicking on a domain within the
graphic takes the user through to the Pfam description page for that domain.

Genes within the family

HGNC Symbol Reports for each gene within a family can be accessed by clicking on the Approved Symbol. By default the table of family members is sorted by
Approved Symbol, but where the family shares a root symbol the members can be sorted by that symbol even where it is a synonym or previous symbol, e.g. in the
DEAD box polypeptides (DDX) family INTS6 is sorted by its previous symbol of DDX26. Note that the symbol used for sorting is highlighted in green to make this
clear.

Above the table of family members there is a small pie chart symbol next to the text "Genes contained within the family". Clicking on the pie chart opens a pop-up
box containing statistics on the locus types of the genes included in the family. For example, the Tubulin gene family contains 22 protein-coding genes and four
pseudogenes.

Gene family downloads

We are now able to provide a much improved way of downloading gene families as data sets, allowing users to choose between downloading a single family or the
entire family hierarchy. For example, users can choose between downloading only the Cholinergic receptors, muscarinic genes shown in Fig 2 or they can browse
through to the G protein-coupled receptors and download all genes belonging to that hierarchy. Each gene family page has a download link at the bottom of the
page that generates a text file with all gene symbols and extra data fields such as "Approved Name" and "HGNC ID". Please note that some gene family pages do
not contain a list of genes because these are included to complete the hierarchical structure; these pages enable users to download all the genes from further
down the hierarchy, e.g. Amine receptors and Cholinergic receptors.

We look forward to receiving your comments on our new Gene Family pages.

Coming soon: an improved Search facility

In addition to our improved gene family pages, we are in the process of developing an expanded Search facility that will include the ability to search our gene
families. The new Search will have a similar look and feel to the current one, so this change should not disrupt the user experience but will make it easier for
people to find what they want.

Our current Search has a dropdown menu that provides a choice between "Search symbol reports" and "Search Site". As shown below, the dropdown menu for the
new Search will give the options "Search everything", "Symbol search", "Family search" and "Site Search".

When searching symbol reports the results filter on the left-hand side still provides the option to filter by locus group or locus type but the locus types are now
displayed under the relevant locus group to make the relationships between the groups and types clearer e.g. "RNA, long non-coding" is listed beneath "Non-coding RNA".

Fig. 3 Example results shown for our new Gene Search
This new Search will be released on genenames.org within the next two months, following testing and full release of the improved Gene Families.

Gene Symbols in the News

Several HGNC-approved symbols have appeared in the international media in recent months. A recent report gave new hope for the treatment of triple-negative breast cancer following the discovery that increased activity of the \textit{BCL11A} gene is associated with this disease. Another study has provided hope that a cholesterol-lowering drug may provide protection from heart disease – the drug ezetimibe inhibits the function of the \textit{NPC1L1} gene and the new study has found that individuals carrying an inactive copy of this gene are at a reduced risk of coronary heart disease. A study on the genetics of Finnish prisoners who have committed violent crimes found an association between violent behaviour and variants of the \textit{MAOA} and \textit{CDH13} genes, although it was noted that many individuals carry these variants without showing violent tendencies.

Meeting News

Beth and Elspeth attended the Livestock Genomics Meeting 2014 in Cambridge, UK from 18th-20th September where Beth gave a talk on her recent work on HCOP. Elspeth and Kris then attended Genome Informatics 2014 also in Cambridge, UK from 21st-24th September, where they presented a poster entitled "Using HCOP To Standardise Gene Nomenclature Across Vertebrate Genomes".

Ruth attended Computational RNA Biology in Hinxton, UK from 11th-13th November where she presented a poster on "The challenges of naming long non-coding RNA genes".

Publications


If you would like to be added to our HGNC Newsletter mailing list or if you have questions or comments on any human gene nomenclature issue, please email us at: hgnc@genenames.org

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