



HGNC Newsletter Summer 2012

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Welcome to Kristian

We would like to introduce a new member of our team: Kristian Gray, who joined the project in May. Kris studied a B.Sc. in Microbiology at Imperial College London and a M.Sc. in Bioinformatics at the University of Manchester. In 2001 he joined the Wellcome Trust Sanger Institute as a laboratory scientist for the Cancer Genome Project before becoming a senior web developer/bioinformatician within the Core Software Services team at the same institute. Now within the HGNC Kris is the team's scientific programmer providing bioinformatical and IT support.

Gene Symbol Report changes

Is this a dagger which I see before me? quite possibly

We have added a dagger † next to certain gene symbols to indicate that the symbol used is a placeholder. If you have functional data about any of these genes or their product(s) please contact us and we can discuss changing the symbol to something better suited based on your information.

The i's have it

We have replaced + with to better reflect that the symbols are links to an information page. We found that the + symbols were confusing to some people in that they assumed that the plus expands a hidden field rather than an information link.

Hint, hint

Tooltips or hints have been added to the gene symbol report pages for certain links/symbols. In the past users used to have to click and leave the page on the orange **C** and **D** symbols to find out what they meant. Now our users can hover their pointer over the **C** and **D**'s and a little description will appear explaining that **C** stands for "Curated by the HGNC" and that **D** stands for "Downloaded from an external source" without leaving the page. Tooltips have also been added to the circular blue i's and the orange dagger symbol offering a quick description for these symbols.

Renaming genes with C\$orf# symbols

Renaming these placeholder symbols continues to be a priority for the HGNC. So far this year we have now renamed 187 C\$orf# symbols with more informative nomenclature.

Some of these genes have been renamed based on publications e.g. *C15orf2* has been reassigned as **NPAP1** based on "The imprinted NPAP1/C15orf2 gene in the Prader-Willi syndrome region encodes a nuclear pore complex associated protein" ([PMID: 22694955](#)), *C4orf49* has been renamed to **MGARP** based on "The expression of a mitochondria-localized glutamic acid-rich protein (MGARP/OSAP) is under the regulation of the HPG axis" ([PMID: 21447634](#)) and *C9orf46* has been renamed as **PLGRKT** based on "Regulation of macrophage migration by a novel plasminogen receptor Plg-R KT" ([PMID: 21940822](#)). Wherever possible, we advise that authors contact the HGNC prior to publication to discuss proposed symbol changes to ensure that the published symbol is unique and complies with our guidelines.

Other previous C\$orf# genes have been renamed based on homology to genes characterised in other species e.g. the gene formerly known as *C7orf44* has been named **COA1** and *C16orf42* has been named **TSR3**; in both cases the new symbols are based on orthologs in *S. cerevisiae*.

And some C\$orf# genes have been named based on domain composition e.g. **CPED1** (cadherin-like and PC-esterase domain containing 1) and **TRABD2A** (TraB domain containing 2A).

If you have information on any genes with a C\$orf# symbol that could be used as the basis for a rename, please either email us at hgnc@genenames.org or fill out our [gene symbol request form](#).

New Gene Family Resources

We have two new gene family pages:

[DNA polymerases](#)

and

[Glutathione S-transferases](#)

which is subdivided into

1. Soluble glutathione S-transferases
2. Microsomal glutathione S-transferases
3. Mitochondrial (kappa) glutathione S-transferase

We are currently working on the revision of our G protein-coupled receptor pages and hope to release this soon.

One gene family has been renamed:

The adrenergic receptors have been renamed to [adrenoceptors](#). This is reflected in the updated names of the associated members and represents common usage in the literature. The root symbol ADR# remains the same.

Gene Symbols in the News

We are happy to report that HGNC symbols continue to appear in the international media. Many of these news articles report the connection between specific genes and disease: a mutation in the [APP](#) gene has been found in 1% of the Icelandic population that [offers its carriers protection from Alzheimer disease](#); increased expression of the [FOXO1](#) gene has been found in [the brains of patients with Parkinson disease](#); a polymorphism in the [GNL3](#) gene has been [linked to osteoarthritis](#); mutations in the X-linked [AFF2](#) gene have been [associated with autism in boys](#); researchers have linked a mutation in the [PHF21A](#) gene [with Potocki-Shaffer syndrome](#); and women with high methylation levels of the [ATM](#) gene in white blood cells have been shown to be at [higher risk of developing breast cancer](#).

In drug function-related news, researchers have recently discovered why anti-TNF drugs, which have been successful in providing treatment for other immune-related disorders, have actually [exacerbated multiple sclerosis](#): a variant of the [TNFRSF1A](#) gene that has been previously linked to the disease encodes a short form of the TNFRSF1A protein. This shortened protein blocks TNF signalling, thereby having the same effect as the anti-TNF drugs. Finally, a recent study has provided hope for the development of future reversible male contraceptives following the discovery in mice that the [KATNAL1](#) ortholog is [needed for the final stages of sperm development](#).

Meeting News

The HGNC will be presenting its future plans for naming genes across vertebrate species at two conferences in Cambridge in this September: Elspeth and Ruth are attending [Genome Informatics 2012](#) at Robinson College from the 6-9th of the month; and Elspeth and Matt are attending the Livestock Genomics meeting from September 9th-11th.

Publications

Louise C Daugherty, Ruth L Seal, Mathew W Wright and Elspeth A Bruford. **Gene family matters: expanding the HGNC resource.** Human Genomics 2012, 6:4 (5 July 2012) [doi:10.1186/1479-7364-6-4](https://doi.org/10.1186/1479-7364-6-4)

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