# HGNC Newsletter Summer 2013

*<Previous Issue*

There are currently 37,656 approved symbols

*Next Issue>*

## In this newsletter

NHGRI funding for HGNC

Gene symbol changes...

...and gene name updates

Antisense or Bidirectional RNAs?

Gene Symbols in the News

Publications

New link within 'Gene Symbol Reports'

## NHGRI funding for HGNC

We are very pleased to confirm the renewal of our funding from the NHGRI for four years (U41HG003345). This key funding for the HGNC, in concert with our funding from the Wellcome Trust, will enable us to continue our assignment of names for novel protein-coding, RNA genes and pseudogenes, as well as the updating of uninformative identifiers based on novel functional data. Further, it will also fund us to begin our work on naming genes across other vertebrate species that currently do not have a dedicated gene nomenclature committee. And crucially, this grant also funds subcontracts for our specialist advisors for naming vertebrate genes within two complex gene families: the cytochrome P450 family, with David Nelson and Jed Goldstone, and the olfactory receptor family with Doron Lancet and Tsivya Olender. Please keep reading this newsletter for updates on our progress with these aims.

## Gene symbol changes...

We have recently received a number of requests from individuals asking if they could be notified of updates in gene symbols for specific genes they are interested in. As a small team it is not feasible for us to contact interested individuals personally, though we do make considerable efforts to notify groups who have published on a specific gene if we are planning to update the gene symbol. However, we could investigate the possibility of setting up an automatic notification system. If you think this would be of interest to you, please let us know via our feedback form, and tell us approximately how many genes you would be interested in receiving notifications for.

Please also note, we strive to avoid changes to established and well-used gene symbols wherever possible. The main exceptions are made for the following specific cases:

- updating a previously uninformative symbol (e.g. a C#orf symbol) based on novel data
- reclassifying a gene into a specific gene family
- correcting a name that has since been found to be misleading

Our new role expanding human gene nomenclature to other species may also involve some changes in gene nomenclature away from designations based on human phenotypes, as these often make little or no sense in other species. For example - and as mentioned above - if a gene is a member of a known gene family but is currently named for causing a human phenotype, we would look into reclassifying it as a gene family member. We appreciate that such symbol changes may prove unpopular among some sectors in the short-term, but we are certain these updates will be beneficial to everyone in the long-term. We feel sure our users appreciate that human gene names, more than those in any other species, are used throughout many different disciplines, and also by the media and general public, and in many cases it is not possible to assign a nomenclature that satisfies all of these groups with their different areas of interest.

## ...and name updates

In collaboration with our colleagues in the mouse and rat gene nomenclature committees we are also looking to remove references to species from gene names, and where possible replace them with descriptive functional information; this will also aid gene name transferral to orthologous genes in other species, by simplifying the gene names. Note that this will usually only affect the gene name, and will not involve a change in gene symbol. A few recent examples of this are:

SYMBOL: *APH1A*
PREVIOUS NAME: anterior pharynx defective 1 homolog A (C. elegans)
NEW NAME: APH1A gamma secretase subunit

SYMBOL: *BUB1*
PREVIOUS NAME: budding uninhibited by benzimidazoles 1 homolog (yeast)
NEW NAME: BUB1 mitotic checkpoint serine/threonine kinase

SYMBOL: *CNPY1*
PREVIOUS NAME: canopy 1 homolog (zebrafish)
NEW NAME: canopy FGF signaling regulator 1

If you spot any gene names that currently include a species in brackets at the end of the gene name that you think could be removed - and perhaps replaced with a brief functional desciption - please let us know.

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## 'Antisense' or 'Bidirectional' RNAs?

Matt attended RNA2013, the Eighteenth Annual Meeting of the RNA Society in Davos, Switzerland from June 11th-16th. During discussions at our poster, "A short guide to human lncRNA gene nomenclature", some researchers proposed an update to our nomenclature for antisense long non-coding RNA (lncRNA) genes. LncRNA genes that overlap an antisense protein-coding gene are named using the approved HGNC symbol for the protein-coding gene with the suffix '-AS' for 'antisense', e.g. the lncRNA gene on the opposite strand to the *BACE1* gene is '*BACE1-AS*' for 'BACE1 antisense RNA'. LncRNAs are also named as antisense if they are found in a head to head arrangement with, and less than 1kb away from, a protein-coding gene, e.g. '*SPATA8-AS1*' does not overlap the *SPATA8* gene but the 5' ends of each gene are separated by less than 100 base pairs. Such non-coding transcripts potentially share a bidirectional promoter with the protein-coding gene and so are now commonly referred to as bidirectional RNAs; however although they may share a promoter they might have entirely divergent expression. Should we continue to name bidirectional RNAs like all other antisense transcripts with an -AS suffix, or should we label these genes with a specific suffix to show they are putatively bidirectional, such as -BD? We'd appreciate your feedback on this issue.

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## Gene Symbols in the News

A study has found that mutations in the *AQP5* gene are the cause of autosomal-dominant diffuse non-epidermolytic palmoplantar keratoderma, a condition characterised by the white, spongy appearance of affected areas of skin upon exposure to water. This research may help to explain how skin maintains its waterproof barrier.

Many behavioural disorders feature hyperactivity in individuals with severe inner ear dysfunction but no genetic link has been proven. Recently, mutations of *the Slc12a2* mouse gene (human ortholog: *SLC12A2*) have intriguingly revealed that an inner ear dysfunction can induce specific molecular changes in a mouse brain that cause maladaptive behaviours, such as hyperactivity.

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## Publications

Dhanoa BS, Cogliati T, Satish AG, Bruford EA, Friedman JS. Update on the Kelch-like (KLHL) gene family. Hum Genomics. 2013 May 15;7(1):13. PMID:23676014 PMCID:PMC3658946

Please also see the KLHL gene family page.

Hediger MA, Clémençon B, Burrier RE, Bruford EA. The ABCs of membrane transporters in health and disease (SLC series): introduction. Mol Aspects Med. 2013 Apr-Jun;34(2-3):95-107. PMID:23506860

Please also see the SLC gene family page. In addition to this published update on the solute carriers, we have now added direct links to the BioParadigms website from each SLC gene symbol report, under the "specialist database" links section.

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## New link within 'Gene Symbol Reports'

We have added a link to BioGPS on our "Gene Symbol Reports" under the heading "OTHER DATABASE LINKS". The link will take you into the BioGPS application for the gene in question, showing a gene expression and activity chart as well as other gene related information. If you would like to know more about BioGPS please look up their about us page.

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