HGNC Newsletter Spring 2013

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There are currently 36,852 approved symbols

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In this newsletter

Welcome to Beth...

We would like to introduce a new member of our team: Beth Yates, who joined the project in April. Beth completed an M.Sc in Bioinformatics at the University of Manchester. She subsequently worked as a research assistant providing bioinformatics support to the *Aspergillus fumigatus* and *Aspergillus flavus* genome projects at the University of Manchester and North Carolina State University respectively. In late 2006 Beth joined the Ensembl project at the Wellcome Trust Sanger Institute where she worked as a web developer before moving to the HGNC.

... and to another new arrival

Congratulations to our team member Ruth and her husband Jon on the safe arrival of their son Evan on 11th April.

New journal support for HGNC nomenclature

Support from journals is absolutely essential for the work of the HGNC and we encourage all journals to ask that submitting authors use HGNC-approved symbols in their papers. Therefore, we are delighted to report that all BioMed Central (BMC journals) now advise that their authors use approved nomenclature e.g. see the instructions listed for the BioMed Biology journal. It is equally good news that all American Association for Cancer Research (AACR) journals now require authors to include approved nomenclature when each gene is mentioned; see their instructions for authors.

New Gene Family Resources

We continue in our goal of increasing our Gene Family resources. Here is a list of the new gene family pages from the last few months:

1-acylglycerol-3-phosphate O-acyltransferases
Adenylate cyclases
Argonaute/PIWI family
Armadillo repeat containing
Arylsulfatases
Ataxins
AT rich interactive domain containing
Beta 3-glycosyltransferases
Beta 4-glycosyltransferases
Beta-1,3-glucuronyltransferases
Bone morphogenetic proteins
Endogenous ligands
Exostosin glycosyltransferase family
Fatty acid binding protein family
Fibrinogen C domain containing
Fibronectin type III domain containing
Laminins

Low density lipoprotein receptors
Netrins
Phosphatidylinositol glycan anchor biosynthesis
RNA binding motif (RRM) containing
RNA pseudouridylate synthase domain containing
Secretory carrier membrane proteins
SH2 domain containing
Tetraspanins
tRNA-splicing endonuclease subunits
Tubulin tyrosine ligase-like family
Ubiquilins
Vesicle-associated membrane proteins
Vanins

New links to Europe PMC

Regular users of our website may have noticed that our Symbol Reports no longer include links to CiteXplore in the Publications field, but instead include links to Europe PubMed Central (PMC). Europe PMC is a collaboration between the EBI, Manchester University and the British library and contains many full text articles in addition to abstracts. It uses similar text mining technologies to its predecessor CiteXplore so that users can highlight and browse keywords for genes/proteins, organisms, GO terms, diseases, accession numbers and chemicals. It also allows sorting of results by publication date, search relevance or the number of times each article has been cited. We are continuing to provide links to PubMed for all publications in addition to links to Europe PMC.

Gene Symbols in the News

There have been several appearances of approved gene symbols in the international media over the past few months, most of which have reported connections between particular genes and diseases. While **BRCA2** is well-known for its association with breast and ovarian cancer, a study has found that prostate cancer patients with a BRCA2 variant suffer from a more aggressive form of the disease. Another "famous" gene, **FTO**, which is well-known for its association with body weight has been linked to a greater risk of melanoma; the mutation linked to skin cancer is found within intron 8 while the mutation linked to body mass is found within intron 1. A mutation in the **TAP1** gene has been associated with susceptibility to leprosy, and a variant form of the **DEPDC5** has been linked to focal epilepsy.

A mouse model has revealed an intriguing link between the **SNX27** gene on chromosome 1 and Down Syndrome, also known as trisomy 21. The study found that the extra copy of chromosome 21 leads to loss of the **SNX27** protein in humans and restoring expression of Snx27 in the Down Syndrome mouse model improves cognitive function and behaviour, leading to hope that this gene could be the cause of learning and memory problems in the human condition. Finally, a new study has shed light on the association between the **LRRC6** gene and the disease primary ciliary dyskinesia. The cilia from patients carrying a mutant form of the gene showed irregular formation and reduced motility. The **LRRC6** protein is now thought to have a central role in the construction of cilia.

Meeting News

Elspeth and Matt attended the 6th International Biocuration Conference in Cambridge, UK from 7th-10th April. Matt presented a poster "Let's talk about sets" which discussed our dedicated webpages for displaying curated gene sets. Gene sets are generally grouped together by shared homology or function. Please email us if you think we need to update a gene set or you have a group of genes that could be made into a set.

Immediately following the Biocuration conference they then both flew to Singapore to attend the HGM 2013 and 21st International Congress of Genetics, which ran from 13th-18th April. Elspeth presented her poster, and gave a brief talk, about our plans to extend the utility of approved gene nomenclature across vertebrate species and Matt presented a poster on our progress to date on naming non-coding RNA genes.

Publications

Seal RL, Wright MW, Gray KA, Bruford EA. **Vive la difference: naming structural variants in the human reference genome.** Hum Genomics. 2013 May 1;7(1):12. [Epub ahead of print] PMID: 23634723

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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