HGNC Newsletter Spring 2014

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There are currently 38,863 approved symbols

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In this newsletter

The HGNC join CCDS

We have recently become members of the Consensus CDS (CCDS) project, which aims to develop a set of high quality gene annotations for human and mouse protein-coding genes. This project is a collaboration between Ensembl, HAVANA, NCBI RefSeq and now MGI and the HGNC. NCBI provide automatic and manually curated RefSeq annotations and Ensembl provide an annotation set that includes automated Ensembl annotations and HAVANA manual annotations. Annotations with matching genomic coordinates that pass a set of stringent quality control tests are assigned a unique CCDS ID. CCDS IDs flagged for update or withdrawal undergo a voting process that includes annotators from RefSeq and HAVANA, and curators from the HGNC for human and from MGI for mouse. For more infomation on the CCDS project, see their latest publication "Current status and new features of the Consensus Coding Sequence database" Nucleic Acids Res. 2014 Jan 1;42(1):D865-72 PMID: 22434842.

We look forward to many productive discussions with our CCDS co-members.

Introducing new improved HCOP

For the last few months we have been working on a new and improved version of our HCOP tool for displaying consensus orthologs to human genes. This new version is now very close to release and you can take a sneak preview of what we've been working on by going to the beta HCOP page on our site (http://www.genenames.org/cgi-bin/hcop (now live)). The new HCOP sees the addition of lots of new data, with ortholog assertions from EggNog, OrthoDB, OrthoMCL, Panther, and PhylomeDB being added in addition to the ortholog resources we already had. We've also added three new species: pig, anole lizard and western clawed frog. You'll notice changes to the way HCOP looks too, as we've tried to make it more user friendly. Please let us know what you think of the changes we've made and if you would like us to include additional ortholog data sources just ask and we'll see if they can be incorporated. Although the release is imminent please bear in mind that we will be tweaking the way the pages look, and functions and some data for some of the species may not yet be available.

New features on our website

We have improved the REFERENCES section within the Symbol Report pages to display the title, authors and citation of the publications as well as providing links out to Europe PMC and Pubmed. Next to the links you will find a '+' icon and clicking on this icon will additionally reveal the abstract and full author list for the publication.

We are now able to display multiple OMIM cross references in our Symbol Reports (e.g. **IGH**) when in the past we only displayed one. This change has also affected the "Complete HGNC Dataset" download file (available on our Statistics & Downloads page) in that the omim_id column in the tab separated file contains multiple OMIM IDs separated by a comma and space (i.e ', '). This is also applicable to the data downloaded from our Custom Downloads application when returning OMIM IDs.

We have also recently updated the HOMOLOGS section of our Symbol Reports to show multiple mouse and rat homologs per human gene where applicable e.g. see the **BCL2A1** report. This also means that there will be multiple MGI or RGD IDs within the download files, which are comma space (i.e. ', ') separated within the tab separated output. The REST API output has also changed for RGD and MGD data as they are now within an array named mgd_id and rgd_id (see REST web-service help).

Genes Symbols in the News

There have been two high profile gene-to-disease associations reported in the media recently. First, the **S1PR2** gene has been linked to gender bias in multiple sclerosis; levels of **S1PR2** are higher in the brains of women with the disease than in men. Second, an Australian study has found a mutation in the **POT1** gene that confers a high risk of developing melanoma. In other news, there has been a study linking genetic variation to pain perception. Individuals suffering with chronic pain are more likely to report lower pain levels if they carry a particular **DRD1** variant, those reporting moderate pain are more likely to carry specific **COMT** or **OPRK1** variants, while those suffering from severe pain are likely to carry a variant of the **DRD2** gene.

Meeting News

Ruth attended HGM 2014 in Geneva, Switzerland from 27th-30th April where she gave a talk entitled "Naming variant genes on the reference human genome" as part of the session on Phenotypes-Genotypes and Therapy.

Kris recently attended the jQuery UK 2014 conference for developers in Oxford, UK on 16th May; the purpose of jQuery is to make it much easier to utilise JavaScript within webpages.

Publications

Wright MW. **A short guide to long non-coding RNA gene nomenclature.** Hum Genomics. 2014 Apr 9;8(1):7.PMID:24716852 PMCID:PMC4021045

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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