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HUGO Gene Nomenclature Committee

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Home | **Downloads** | **Gene Families** | **Tools** | **Useful links** | **About** | **Newsletters** | **Contact Us** | **Help** | **Request Symbol**

HGNC Newsletter Summer 2014

In this newsletter

New HCOP released

New features on our website

Gene Symbols in the News

Meeting News

Farewell to Matt

New HCOP released

A new and improved version of our HCOP tool has been released. This updated version of HCOP includes ortholog data from eggNOG, Ensembl Compara, HGNC, HomoloGene, Inparanoid, OMA, OrthoDB, OrthoMCL, Panther, PhylomeDB, TreeFam and ZFIN. The new HCOP also features a revamped, more user-friendly interface, and data for three new species: pig, xenopus and anole lizard. For more information about the new HCOP please visit the HCOP help page.

New features on our website

- We have recently changed our Symbol Report Search results to show the text that matches the actual query string within the matching field type, e.g. a search for "PARN" returns two records, with the top hit (**PARN)** showing the matching field "**Approved Symbol:** PARN" and the second hit (**PNLDC1**) showing "**Approved Name:**poly(A)-specific ribonuclease (PARN)-like domain containing 1". We hope that this will make the results clearer and will allow users to quickly identify their Symbol Report of interest.

- We would like to notify users of our REST service that we have changed the type associated with the hgnc_id field from int to string. The resulting hgnc_id will have the "HGNC:" prefix. We have made this change to make it clear that the HGNC ID is officially HGNC:# rather than just a number. The entrez_id field has also changed to a string due to backend code reasons. We hope that this has not caused too much disruption to anyone using our REST service.

- We have recently updated the HOMOLOGS section of our Symbol Reports to show multiple mouse and rat homologs for each human gene, where applicable. This also means that there will be multiple MGI or RGD IDs within the download files, which are comma space (ie ', ') 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).

- We have changed the name of "Symbol Checker" to "Multi-Symbol Checker" to make it clearer that this tool is intended to allow users to check the nomenclature of many genes at once.

Gene Symbols in the News

A news article recently reported that the gene variant of **EPAS1** which is thought to enable Tibetans to live at high altitudes is likely to have been inherited from Denisovan, not Homo sapien, ancestors. This discovery is somewhat surprising as the only Denisovan remains were found in a cave at an altitude of only 760m.

Also in the news was a study that linked a mutation in the **TBC1D4** gene with an increased risk of developing type 2 diabetes. The mutation is carried by 17% of the population of Greenland and leads to a ten fold increase in the likelihood of becoming diabetic. In other gene-to-disease news, a recent study found that a particular **PALB2** variant leads to a 33% risk of women developing breast cancer before the age of 70.

Meeting News

Kris attended OSCON (Open Source Convention) 2014 in Portland, Oregon, USA from 20-24 July 2014. OSCON is the main conference for the entire open source community, where delegates can find out about new and current open source resources, and discuss and learn from experts in the field. As our project is becoming evermore reliant on open source technologies such as the database, programming languages, and search we recently implemented, the conference enabled Kris to see how we can improve our use of the open source resources we are currently using, and to find out about other resources we could implement in future.

Farewell to Matt

After fourteen very fruitful years, Matt Wright has left the HGNC to join the RefSeq team at the NCBI. We are pleased that we will still be able to interact with Matt on a professional level and would like to wish him all the best for his new life in the USA.

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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