



## HGNC Newsletter Summer 2015

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### Brand new BioMart released

Since 2008 we have provided a BioMart service to allow our users to query and download the data that they want easily and quickly. Due to infrastructure and data changes it has become necessary to close this service, but fear not, as the old BioMart service has been replaced by a new BioMart server using the latest BioMart (0.9.0) code. Despite looking very different the new service retains an easy to use interface, includes our gene family data in addition to the gene symbol report dataset, and allows our users to access and query a lot more of the HGNC data than was previously possible. Bioinformaticians and web developers can also interact with our BioMart server via REST/SOAP, SPARQL and a Java BioMart API available from the [BioMart project](#) website.

You can find the new Biomart at [biomart.genenames.org](http://biomart.genenames.org) or via the link under our Tools menu. Our tool actually includes two separate HGNC Marts, Gene and Family, which have different interfaces and allow users to choose to search all gene data or gene family data. The BioMart tool supports bulk uploads of data and allows users to filter sets of genes that have particluar data associated with them, e.g. you can search for all genes with a CCDS ID or all genes with an associated LSDB link. You select exactly which attributes you want to see in your results. Check out the [BioMart help](#) page for a full description of what the new BioMart offers.

### Changes to genes named after homologs

We are in the process of removing the names of other species from genes that are named after homologs. These species names have sometimes caused confusion, and in many cases the same symbol/name is used across many species already, negating the relevance of citing a single source species in the name. Where possible we are adding extra information into the names of these genes. For example, we have updated the name of the **MON2** gene from "MON2 homolog (S. cerevisiae)" to "MON2 homolog, regulator of endosome-to-Golgi trafficking", we have updated the name of **ATOH1** from "atonal homolog 1 (Drosophila)" to "atonal bHLH transcription factor 1" and we have updated the name of **TOM1** from "target of myb1 (chicken)" to "target of myb1 membrane trafficking protein". All original gene names are displayed in the "Previous symbols and names" section of our Symbol Reports and are fully searchable on [genenames.org](http://genenames.org).

### New Gene Families

We hope that you have all been finding our updated Gene Families useful. We are continually updating this resource - within the last few months we have added over 70 new families. Where relevant we organise our gene families into related hierarchies. For example, we have recently created the [Cystatin superfamily](#) page, which has the subfamilies [Cystatins, type 1](#), [Cystatins, type 2](#), [Cystatins, type 3](#) and [Cystatins, type 4](#). Another new family hierarchy is the [Histone deacetylase superfamily](#) which is grouped into [Histone deacetylases, class I](#), [Histone deacetylases, class IIA](#), [Histone deacetylases, class IIB](#), [Sirtuins](#) (also called Histone deacetylases, class III) and [Histone deacetylases, class V](#); while the new [Peptidylprolyl isomerases](#) page has the subfamilies [Parvulins](#) and [Immunophilins](#), which is further split into [cyclophilin peptidylprolyl isomerases](#) and [FKBP prolyl isomerases](#). If you are working on a gene family that is missing from our resource please let us know.

### HGNC is tweeting...

We would like to remind our users that we have a twitter account. Please follow [@genenames](https://twitter.com/genenames) for all our latest news, including important new gene family releases.

## Gene Symbols in the News

In gene therapy news, patients suffering from Leber's congenital amaurosis-2, a sight condition, caused by mutation of the **RPE65** gene, [showed improvement following targeted treatment to their eyes with an active copy of the gene](#). However, this improvement peaked at 6-12 months following treatment. Researchers say a better gene therapy vector is needed.

Recently, a team has found one genetic explanation as to [why some children suffer from more middle ear infections than others](#) - children carrying a variant of the **A2ML1** gene appear to be more prone to these types of infection. Also in the news, [scientists have found a possible target for reducing age-related cognitive decline](#) after noticing that there is an increase in levels of **B2M** protein in the blood of older humans and mice. They found that injecting young mice with B2M resulted in poorer performance in memory-related tasks.

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## Meeting News

Beth attended the [Quest for Orthologs 4](#) from May 25th to 27th in Barcelona, Spain where she presented a talk entitled "HGNC efforts to standardise gene nomenclature across vertebrate species". Ruth attended [ESHG 2015](#) in Glasgow, UK from 6th to 9th June where she presented a poster on our new gene families.

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## Publications

Smedley D, Haider S, Durinck S, Pandini L, Provero P, ... Gray K, ... Kasprzyk A. **The BioMart community portal: an innovative alternative to large, centralized data repositories**. Nucleic Acids Res. 2015 Apr 20. pii: gkv350. [Epub ahead of print] PubMed PMID: [25897122](#).

Kubagawa H, Carroll MC, Jacob CO, Lang KS, Lee KH, Mak T, McAndrews M ... Seal R, Wang JY, Wiestner A, Coligan JE. **Nomenclature of Toso, Fas Apoptosis Inhibitory Molecule 3, and IgM FcR**. J Immunol. 2015 May 1;194(9):4055-7. PMID: [25888699](#)

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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](mailto:hgnc@genenames.org)

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