HGNC Newsletter Summer 2011

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There are currently 31546 approved symbols

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New Gene Family resources

We have continued to add many new gene family/grouping pages to our public website. Please remember that there are now links from the gene symbol reports to relevant gene families. The following families have been added over the last quarter:

A-G

Actin related protein 2/3 complex subunits
Adenylate kinases
Anaphase promoting complex subunits
Anoctamins
Chemokine (C-X-C motif) receptors
DDB1 and CUL4 associated factors
Enhanced RNAi three prime mRNA exonuclease family
Ephrins
Fermitin family
Fibulins
Formyl peptide receptor family
Gamma-glutamyltransferase family
GPN-loop GTPase family
GTPase, IMAP family

H-K

Hydroxy-carboxylic acid receptors
IKAROS family zinc fingers
INO80 complex subunits
Keratin associated proteins
KN motif and ankyrin repeat domain family

L-P

Lysophospholipid receptors
Myocyte enhancer factors
Na+/K+ transporting ATPase interacting family
N-terminal EF-hand calcium binding protein family
NOL1/NOP2/Sun domain family
Parkinson disease
Patatin-like phospholipase domain containing
Perilipins
Poly (ADP-ribose) polymerase family
POTE ankyrin domain family
Prenyltransferase alpha subunit repeat containing family
Prostate and testis expressed family

R-W

Rho GTPase activating protein family
SAM and SH3 domain containing family;
Shisa homologs
Speedy homologs
Tectonic family
Ubiquitin protein ligase E3 component n-recognin
UBX domain containing
WW, C2 and coiled-coil domain containing

There are instances where large gene families/groups have been subdivided into smaller subgroupings. For example the Chromatin-modifying enzyme group has been divided into three subgroups: K-demethylases (KDM), K-acetyltransferases (KAT) and K-methyltransferases (KMT). These subgroups all have their own separate common root (or stem) which allows easy identification of the members.

Another large new gene family is the Homeobox gene family, this has also has been divided into twelve subgroups as detailed in Holland PW, Booth HA, Bruford EA. **Classification and nomenclature of all human homeobox genes.** BMC Biol. 2007 Oct 26;5:47. PMID:17963489

Gene Family downloads

We have recently developed new and additional ways to download our gene family data. There are now four different options for downloading these data:

1) From www.genenames.org select the Downloads tab. Here you will find a direct link to the "complete HGNC dataset" which includes our gene family data fields, Gene Family Tag and Gene Family Description.

2) Alternatively, if you only want information relating to the gene family data select "complete HGNC Gene Family dataset". This will give you a file with the following fields: URL /Gene Family Tag/Gene Family Description/Symbol/HGNC ID

3) Our Custom Downloads page lets you specify what exact fields you require. Select the Gene Family Description and/or Gene Family Tag fields to include gene family data in your output.

4) Finally, if you just want the data for the gene family associated to your gene of interest, click the link "Download gene family data" beneath the gene family table on the relevant gene family page.

Gene Symbols in the news

Approved gene symbols continue to be used by the international media. There have been several reports on genetic variation affecting the appearance and behaviour of individuals. Two different genes have been associated with fat levels: the *KLF14* gene product has been shown to control a number of genes found in fat tissue. A variant of the *IRS1* gene is associated with reduced fat under the skin but not around the internal organs, meaning that carriers may appear slim whilst still carrying dangerous levels of internal fat. A variant of the *DRD4* gene has been reported to be associated with "high-risk" behaviours, including promiscuity.

There have also been reports on new gene functions. A recent study has found that the human *CRY2* encodes a protein that can function as a magnetosensor when expressed in flies, although whether humans can sense magnetic fields is still under debate. The *CXCL5* gene product has previously been associated with the pain mechansim and has recently been shown to be produced in skin burnt by UV rays, meaning this could be a target for painkillers in the future.

Additionally, there have been several reports on the association of genes with disorders. An exome sequencing study found four genes associated with autism; while three of these genes have been identified with this condition before (*FOXP1*, *GRIN2B* and *SCN1A*), the association between the *LAMC3* gene and autism is a new finding. Research has shown that there may be a genetic link for migraines; a study on female sufferers found an association with the *PRDM16*, *TRPM8* and *LRP1* genes. And a study has shown that men carrying a particular variant of the *DEFB126* gene have low rates of fertility; the *DEFB126* gene product has been associated with movement of sperm through the female reproductive tract.

Finally, there has been recent hope for the treatment of malignant melanoma. The drug Vemurafenib inhibits *BRAF* activity and has been shown to extend the life of melanoma patients. Previous studies have shown that this gene is frequently mutated in melanoma, showing how gene-association disease studies can have direct impact on treatments.

Meeting News

Louise and Ruth attended the European Human Genetics Conference (ESHG 2011) in Amsterdam, Netherlands from May 28th-31st. They presented a poster entitled "Using HGNC to improve and expand Gene Family resources" and enjoyed meeting the other delegates at the conference.

Matt attended the RNA 2011, the Sixteenth Annual Meeting of the RNA Society in Kyoto, Japan from June 14th-18th. He presented a poster detailing his recent work on naming non-protein coding RNA genes. For more details on this work, please read our publication: Wright MW, Bruford EA. **Naming 'junk': Human non-protein coding RNA (ncRNA) gene nomenclature**. Hum Genomics. 2011 Jan 1;5(2):90-8. PMID:21296742

Michael will be attending the "YAPC::Europe" conference 2011 in Riga, Latvia from 15th-17th August.

Supporting parallel gene nomenclature across vertebrate species

In May Elspeth & Matt travelled to the Jackson Lab in Bar Harbor, Maine to meet with our colleagues from both the Mouse Genome Database, and from the Rat Genome Database in Milwaukee, Wisconsin. Discussions ranged from nomenclature usage in journals to naming pseudogenes, readthrough transcripts, long non-coding RNAs and transposable elements, and enabled us to ensure that we continue naming genes in human, mouse and rat in a concordant way. We would also like to take this opportunity to thank Lois Maltais, who has recently retired from her role as MGI Nomenclature Coordinator, for all the help and support she has given the HGNC team over the years.

Elspeth also attended the second Quest for Orthologs meeting at Hinxton in June, which brought together researchers in the field of orthology with the aims of evaluating different approaches to ortholog identification, establishing benchmarking, increasing interoperability between resources, and improving orthology-based protein function prediction. This work is very relevant for both our HCOP resource (new release coming soon!) and for gene naming across vertebrates.

Publications

Mayer J, Blomberg J, Seal RL. **A revised nomenclature for transcribed human endogenous retroviral loci.** Mob DNA. 2011 May 4;2(1):7. PMID:21542922

This paper describes our work with the ERV community to devise a nomenclature system for transcriptionally active ERVs. The new nomenclature only applies to ERV loci that are represented by mRNA sequence in a public database corresponding to at least one viral gene. Each symbol is of the format ERV + group symbol + unique number *e.g.* ERVK-1. Group symbols are based on a mixture of established Repbase designations and well-supported symbols used in the literature. All ERV genes have the locus type "endogenous retrovirus" and can be grouped using this data field. If you have any questions, please contact us at hgnc@genenames.org.

Farewell to Susan

Susan Gordon has recently left the HGNC and returned to her native Canada. We wish her all the best for the future, and would particularly like to thank her for all of the essential work she did on creating our lovely new website.

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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