HGNC Newsletter Spring 2012

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There are currently 33084 approved symbols

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In this newsletter

A new HGNC team member

We are excited to announce that we have appointed a new database developer, Kristian Gray, who starts with us on 1st May. We'd like to thank him for the help he has already provided prior to being an official team member! Look out for a full introduction to Kris in the next newsletter.

HGNC on twitter

For the most up-to-date news on the HGNC, you can now follow us on twitter @genenames. You can also link to our twitter account from the twitter icon in the 'Latest News' box on our homepage. In addition to news about the HGNC, we tweet news stories featuring our symbols and other interesting news from the world of genetics and genomics.

Update on renaming genes with C$orf# symbols

As reported in the previous newsletter, we are in the process of renaming genes with a *C$orf#* symbol wherever relevant new information is available (within *C$orf#* symbols $ represents the chromosome on which the gene is located and # is the next number in a numerical series). We renamed 155 *C$orf#*s with more informative gene symbols and names in 2011, and we have already renamed a further 70 so far in 2012. We aim to continue this process throughout this year; however, if a *C$orf#* symbol is consistently being used by the community, and there is no functional information for a rename, we will continue to support that symbol. For example, there are currently many publications on the ***C9orf72*** gene due to the discovery of a hexanucleotide repeat expansion in a noncoding region of this gene that causes neurodegenerative disease. Reflecting the utility of these symbols, all papers are currently using the approved ***C9orf72*** symbol, and so we would only consider a rename if the normal function of the gene was well characterised and the community was in support of such a change. If you do have information on a gene with a *C$orf#* symbol that you think could be used as the basis for a rename, please contact us via hgnc@genenames.org, or use our gene symbol request form.

New Gene Family Resources

New Ion Channels Resource

We now have a dedicated Ion Channel page which encapsulates the voltage-gated ion channels and ligand-gated ion channels, plus some "other" new ion channel pages. We reviewed our data in light of the latest edition of the Guide to Receptors and Channels (GRAC). Consequently, there have been a number of new gene families created, a number of new groups associated to current gene family pages and one gene family rename.

The new page has links to the following constituent groups: Voltage-gated ion channels, Ligand-gated ion channels and all other ion channels, and the latter two groups include some new gene families:

New Ligand-gated ion channel groups

1. 5-HT (serotonin) receptors, ionotropic
2. Acetylcholine receptors, nicotinic
3. GABA(A) receptors

4. Glutamate receptors, ionotropic
5. Glycine receptors
6. Purinergic receptors, ionotropic
7. Zinc-activated channels

New "other" ion channel groups
1. Pannexins
2. Sodium channels, nonvoltage
3. IP3 receptors
4. Ryanodine receptors
5. Sodium leak channels, non-selective
6. The Chloride channels page has the following NEW subgroups (in addition to the pre-exisiting Chloride intracelluar channels group)
   - Chloride channels, voltage-sensitive
   - Cystic fibrosis transmembrane conductance regulators
   - Chloride channels, calcium-activated

Re-naming of a gene family:

Amiloride-sensitive cation channels [ACCN] have been renamed to Acid-sensing (proton-gated) ion channels [ASIC]. All members of this family now have ASIC# symbols instead of ACCN# symbols. This is to reflect the adoption of this new nomenclature within the latest release of the GRAC and by IUPHAR, and to reflect the consistent usage of the ASIC# symbols in the literature. Members of the community have been notified, and are in agreement with the change.

Revised and updated Gene Families

We have improved the organization of the following gene family pages:

1. The Immunoglobins page has been reordered to group gene segments from the same loci together and to separate out the orphon gene segments. Groupings are as follows: IGH@ locus at 14q32.33, IGH orphons (not at the IGH@ locus), IGK@ locus at 2p11.2, IGK orphons (not at the IGK@ locus), IGL@ locus at 22q11.2, IGL orphons (not at the IGL@ locus), IGJ linker

2. The T cell receptors page has been reordered in a similar way, as follows: TRA@ locus at 14q11.2, TRB@ locus at 7q34, TRB orphons (not at the TRB@ locus), TRD@ locus at 4q11.2, TRG@ locus at 7p14

3. The Short chain dehydrogenase/reductase superfamily page has been organised into the following groups:
Classical SDR fold cluster 1
Classical SDR fold cluster 2
Classical SDR fold cluster 3
Extended SDR fold
Atypical members

4. The SP/KLF transcription factor page has been split to reflect the two separate gene families:
Specificity protein transcription factors
Kruppel-like transcription factors

New Gene Family pages

1. Cholinergic receptors
2. Paraneoplastic Ma antigens
3. Serine/threonine phosphatases The following subgroups are represented on this page:
   a) Protein phosphatase, catalytic and regulatory subunits:
      Protein phosphatase, catalytic subunits
      Protein phosphatase 1, regulatory subunits
      Protein phosphatase 2, regulatory subunits
      Protein phosphatase 3, regulatory subunits
      Protein phosphatase 4, regulatory subunits
      Protein phosphatase 6, regulatory subunits
   b) Protein phosphatases, Mg2+/Mn2+ dependent
   c) CTD aspartate-based protein phosphatases

Gene Symbols in the News

There have been a number of reports in the international media featuring approved symbols over the last few months. Many of these have been studies on how genetic variation affects our health and interaction with our surrounding environment: mothers carrying a ***PHLDA2*** genetic variant have been shown to be more likely to give birth to babies of a larger average weight; a variant of the ***HMGA2*** gene has been associated with larger brains and higher scores on IQ tests; variants of the ***TPH1*** and ***TPH2*** genes are associated with a higher risk of developing post-traumatic stress disorder; an ***IFITM3*** variant is associated with increased susceptibility to influenza virus; and mutation of the ***XRCC2*** gene has been linked to breast cancer. In further news, a mutation in the ***SHANK1*** gene has been associated with the development of autism but interestingly, only in male carriers even though the gene is on an autosomal chromosome.

A new study has implicated the ***CD44*** gene in insulin resistance and the formation of type 2 diabetes; experiments on *Cd44* knockout mice showed that lack of *Cd44* protected the mice from developing insulin resistance while the blood sugar levels of mice carrying functional *Cd44* copies were reduced after treatment with CD44 antibodies, providing hope for the early detection and treatment of insulin resistance in humans. Researchers have found that the activity of ***TLR9***, which has a role in pathogen recognition, is controlled by circadian rhythms; response to vaccination and survival rates following induced sepsis vary in mice depending on the level of *Tlr9* activity at particular times of day. Finally, there has been further success with ***RPE65*** gene therapy: patients that received gene therapy in one eye back in 2008 have now received treatment in their second eye and have reported improvements to their vision.

Meeting News

Matt attended HGM 2012 in Sydney, Australia from 11th-14th March, where he presented his first electronic poster on long non-coding RNA nomenclature. He enjoyed meeting the delegates at the conference and climbing the Sydney Harbour Bridge. Matt also attended the NC-IUPHAR Meeting of the International Union of Basic and Clinical Pharmacology Committee on Receptor Nomenclature and Drug Classification in Paris, France from 13th-15th April. The HGNC maintains an active collaboration with NC-IUPHAR in order to agree a parallel nomenclature for genes that encode receptors and channels, and to reach a consensus on when there is enough empirical evidence to rename orphan G-protein coupled receptors (GPRs) based on their cognate ligands.

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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