



Nome News Issue 29

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New Team Member

We are pleased to announce the appointment of a new member to the HGNC team. Dr Kate Sneddon (no relation to Dr Tam Sneddon!) has joined us as a new nomenclature advisor and will be based at the London office

Entrez Gene

This month saw LocusLink superceded by Entrez Gene. Although we are sad to lose LocusLink we would like to thank the NCBI for their prominent positioning of the HGNC approved 'Official Symbol and Name' fields and the direct link to our webpage.

New Logo

The HGNC logo is currently being redesigned and will replace the present logo on our website later this month...see if you can identify which chromosome we used!

Hot Topic

'Does it matter if a disease phenotype and its causative cloned gene have the SAME symbol?' Many inherited diseases have a symbol for the mapped locus whose existence is inferred from; pedigree analysis. When the gene which is mutated in the disease is identified it may be realised that this is a known and well characterised gene which already has an approved name and symbol. If so, the phenotype-based symbol is merged into the already approved gene name and symbol. If the gene is novel but recognised as a member of a gene family, or if there is direct functional information about the gene product, an appropriate new gene name and symbol can be approved and the phenotype symbol merged into this. However, problems arise when the only useful information known about a gene is that when it mutates the disease occurs.

There are then two choices:-

- 1) Introduce a new symbol or add some extra letter to the phenotype symbol, in each case as a temporary solution.
- 2) Keep the original phenotype-derived name and symbol until more is known about the normal function of the gene.

Why does this matter?

Sometimes the following situations cause problems, for example:

- It is difficult to write a clear discussion of gene and phenotype correlations and interactions when the gene and phenotype have the same symbol, e.g. SPG4 , spastic paraplegia 4.
- A gene may be found to cause other disorders than the one from which the gene symbol was originally derived, e.g. the OPA3 gene is named after "optic atrophy 3", but mutations in OPA3 also cause methylglutaconic aciduria.
- Other genes may be found that are related by sequence to the disease causing gene, but have nothing to do with the disease, e.g. VMD2 (vitelliform macular dystrophy 2) has related genes called VMD2L3 , VMD2L2 , and VMD2L1 which are not involved in vitelliform macular dystrophy.

We would like to know what you think.

Please email hgnc@genenames.org to vote for option 1 or option 2

Meetings

Tam recently attended the International Workshop on Encoding Information in DNA Sequences in Okinawa, Japan, thanks to funding from the Japanese Government. With 19 renowned lecturers and 39 invited participants, this provided stimulating and valuable insights into future directions for genomic analysis.

The Workshop was a precursory event for the [Okinawa Institute of Science and Technology \(OIST\)](#), a graduate university established by the Government of Japan.

Publication

Ross MT, Grafham DV, Coffey AJ *et al.* The DNA sequence of the human X chromosome. *Nature* **434**: 325-337 ([PMID:15772651](#)).

If you would like to be added to our Nome News mailing list or if you have questions or comments on any human gene nomenclature issue, please email us at: hgnc@genenames.org