



HGNC Newsletter Summer 2010

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There are currently 29540 approved symbols

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In this newsletter: we present a new nomenclature scheme for the serine/arginine-rich proteins, a mitochondrial respiratory chain gene family page, a new locus type and news of several meetings attended by HGNC members.

SRSF family

We have recently revisited the nomenclature of serine/arginine-rich (SR) proteins following an effort led by Dr James Manley of Columbia University and Dr Adrian Krainer of Cold Spring Harbor. Dr Manley and Dr Krainer standardized the rules for which genes should be included in the genuine SR protein family based on the following protein sequence characteristics: one or two N-terminal RBDs [RNA recognition motifs], followed by a downstream RS domain of at least 50 amino acids with >40% RS content, characterized by consecutive RS or SR repeats." Following a discussion with the SR protein community and the HGNC, it was agreed that a suitable root symbol for these genes is SRSF# for "serine/arginine-rich splicing factor". The new symbols and a description of the family have been published in "[A rational nomenclature for serine/arginine-rich protein splicing factors \(SR proteins\)](#)" *Genes Dev.* 2010 Jun 1;24(11):1073-4. The publication includes a full list of scientists that are in support of the new nomenclature scheme.

New gene family page

We have created a gene family page for the human genes encoding components of the [mitochondrial respiratory chain](#). The page presents a full list of human genes that encode proteins for each of the five respiratory complexes to provide a quick reference for the nomenclature of the genes involved in this important biological process. Please contact us if you would like us to create a gene family page for genes that you are working on.

New locus type

We have recently added a new locus type: virus insertion site. This represents genomic target sequences that have been identified as viral DNA insertion sites in the human genome. For example, the adeno-associated virus (AAV) shows a preference for insertion at a particular target on 19q13, which has been named as [AAVS1](#) for "adeno-associated virus integration site 1".

Gene symbols in the news

Approved gene symbols continue to appear in the international media, proving that gene nomenclature aids communication beyond the scientific community. Here are some news stories featuring approved gene symbols from July 2010:

[SIRT1](#) was reported as [promoting memory in addition to longevity](#). A mutation in the [EPAS1](#) gene was reported as [allowing Tibetan people to thrive at high altitudes](#) and men carrying a mutated copy of the [BRCA2](#) gene were reported as [being at a greater risk of breast cancer](#).

Meeting news

Susan attended the [3rd Human Variome Project](#) meeting from 10-14 May in Paris. Her presentation entitled "A nomenclature for copy number variant genes?" can be viewed [here](#).

Elspeth and Ruth attended the [14th Human Genome Meeting](#) from 18-21 May in Montpellier, France. As ever, the HGNC pens proved very popular amongst delegates. The HGNC Nomenclature poster prize winners for using the most HGNC-approved gene symbols, each winning a year's subscription to the HUGO Journal and to one other Springer journal of their choice (with our thanks to Springer for their sponsorship!), were:

1st - Melissa Beaudoin from Montreal Heart Institute, Canada for poster no 38, "Identification of novel sequence variants using high-throughput re-sequencing of ulcerative colitis genes in French-Canadian patients"

2nd - Yasha Hasija from the Council of Scientific and Industrial Research in New Delhi, India for poster no 89, "SNPome: Human Polygenic Disease SNP Network"

This year for the first time the HGNC had a presence at the [International Society for Animal Genetics](#) meeting, held in Edinburgh (26-30 July). Elspeth presented a poster entitled "Speaking the Same Language: A Common Nomenclature for Vertebrate Genes" and had interesting discussions with delegates about plans for establishing gene nomenclature committees for groups of livestock species.

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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