USERS GUIDE TO THE DECEMBER 2017 VERSION OF OMIA

Thanks to some excellent work by software engineer Josh Stretton, whose time and skills were generously provided by the University of Sydney through the university's Sydney Informatics Hub and Centre for Translational Data Science, a new version of OMIA was launched in December 2017.

There are two new features in this new version.

1. **The first is a new URL, namely http://omia.org.**

This was purchased (for the princely sum of US$425!) to highlight OMIA as a global resource and to emphasise OMIA's complementarity with OMIM (http://omim.org), the human catalogue on which it is based and to which it is reciprocally hyperlinked. The previous URL http://omia.angis.org.au continues to function, and anyone who uses this URL will be directed automatically to the new one.

When writing papers on particular traits/disorder, authors are strongly encouraged to use the relevant full OMIA ID to identify a trait/disorder. Authors are also strongly encouraged to ensure that all mentions of an OMIA ID in a paper include the relevant hyperlink, which is a simple function of the OMIA ID. For example, the OMIA ID for Haemophilia B in cats is OMIA 000438-9685 and its URL is http://omia.org/OMIA000438/9685/. Hyperlinks to OMIA IDs can be included in papers in a very tidy fashion (without displaying the URL) by associating the URL with the ID, e.g. OMIA 000438-9685

2. **The second new feature is the inclusion of tabulated information on likely causal variants using standard nomenclature.**

In the summary table on the home page, you will see there is an extra (Likely causal variants) row. This generates a sortable table of all variants (last column) or all those in a particular species (other columns). The table includes hyperlinks to the relevant OMIA page, to the relevant paper in PubMed, and (if the variant has been submitted to a variant database) to the relevant entry in Ensembl, with a link to dbSNP. At the bottom of each table is a tab for CSV download and another for summary stats. If you wish to download a table so as to retain the hyperlinks, simply copy and paste the table straight into Excel.

There is also a table of variants for each phene-species in which at least one likely causal variant has been reported. If you look, for example, at http://omia.org/OMIA000683/9913/, you will see a table of the variants for muscular hypertrophy (double muscling) in cattle. In this example, three of the variants have rs IDs, which are hyperlinked to the Ensembl entry for that variant.

Another way to access variants is to use any of the searching tools available from the Search tab on the home page. For any search, it is now possible to return a list of phenes or a list of genes or a list of variants.

As soon as is feasible, OMIA will store only a variant's rs ID, which will be used to populate the OMIA variant tables with always-up-to-date information directly from the European Variant Archive (EVA). Obviously, this can be done only for variants that have been submitted to a variant database! Given that fewer than 10% of variants in the OMIA tables have an rs ID at present, there is a serious need for authors to submit all newly-discovered likely causal variants, and for a big effort to do the same for the backlog. FN is working with Gary Saunders and other EVA personnel to facilitate the submission process. If you wish to be part of the team developing a suitable submission process, please contact FN.

The information in the variant tables has been compiled manually since 2013 by FN, incorporating information from other acknowledged sources. Ensembl's Variant Effect Predictor (VEP) has been an invaluable tool in verifying particular variants and in providing genomic locations. There are many variants with only a verbal description or with incomplete information and, no doubt, there are many errors. There are also many variants with absent or incomplete breed and other information. **FN urges anyone who has variant information that should be included to contact him. Thanks to Josh's efforts, we now have a new variant curation page that will enable curators to fix anything that anyone spots.**