
Print this Page for Your Records Close Window

**Control/Tracking Number:** 2019-A-405-ESHG
**Activity:** ESHG Abstract
**Current Date/Time:** 2/7/2019 10:39:02 AM

# Enhanced access to extensive phenotype and disease annotation of genes and genetic variation in Ensembl

**Author Block: I. M. Armean**, L. Gil, D. Lemos, A. Parton, H. Schuilenburg, A. Thormann, S. Hunt, F. Cunningham;
European Molecular Biology Laboratory, European Bioinformatics Institute, Hinxton, Cambridge, United Kingdom.

**Abstract:**
The accurate annotation and interpretation of genes and genetic variation is paramount in basic research and clinical diagnostics. Critical information such as population allele frequencies, the predicted molecular consequences of variants and observed associations with phenotype often lie in decentralized large-scale resources and are time consuming to integrate.
We import and harmonize phenotype and disease annotations from sources including ClinVar, DGVa/dbVar, the NHGRI-EBI GWAS Catalog, DDG2P, Orphanet, MIM morbid and the Cancer Gene Census into a standard structure and provide simple tools for data access.
To facilitate improved querying across conditions described differently in different studies and support aggregated views in the Ensembl genome browser, we map the phenotype descriptions used in these projects to ontology terms. We have recently updated the Ensembl Variant Effect Predictor (VEP) to provide enhanced phenotype and disease annotations alongside predicted molecular consequence and frequency data. We have also recently extended our REST API to support detailed querying of these data.
In Ensembl release 96 (April 2019) we have 54,859 human phenotype/disease annotations associated with genes and 5,910,851 associated with variants from 15 different sources. These can be extracted using a variety of interfaces and are available for variant annotation with VEP.
We import an extensive range of phenotype and disease annotations into one unified resource. Here we describe the data types we hold and the multiple access methods we have developed to simplify the use of these essential data within the community.
:

Author Disclosure Information:

**I.M. Armean:** None. **L. Gil:** None. **D. Lemos:** None. **A. Parton:** None. **H. Schuilenburg:** None. **A. Thormann:** None. **S. Hunt:** None. **F. Cunningham:** None.
**Topic (Complete)**: 16. Omics/Bioinformatics
**Keyword (Complete)**: vep ; phenotype ; ensembl
**Presentation Preference (Complete)**: Oral preferred
**Fellowships - Awards (Complete)**:
**Do you wish to apply for any of the Fellowships or Awards?**: No
**I am a: none of the above**
**I apply for the Conference Fellowship for European Countries: No**
**I apply for the Fellowship for Non-European Countries: No**
**I apply for the Fellowship of Excellence: No**
**I apply for the Young Investigator Award: No**

**Status:** Complete

ESHG Conference - Wiener Medizinische Akademie Kongress GmbH
Alser Strasse 4, A-1090 Vienna, Austria
Tel: (+43/1) 405 13 83-22
Fax (+43/1) 407 82 74

Feedback