Announcing the Musculoskeletal Knowledge Portal

We are pleased to announce the Musculoskeletal Knowledge Portal (MSK-KP), an open-access resource that aggregates, integrates, analyzes, and displays 'omic results relevant to musculoskeletal traits and diseases. The MSK-KP will facilitate and accelerate musculoskeletal disease research by comprehensively amassing and distilling "big data" to make them accessible to all researchers.

Motivation

Many barriers exist for researchers who would like to take advantage of large-scale 'omic data to design new therapeutic targets or to better understand the regulatory networks and pathways underlying disease: the datasets are often only accessible to members of large consortia, and can only be handled and interpreted by scientists with computational biology expertise. The MSK-KP breaks down those barriers by harmonizing and analyzing datasets, and summarizing the most significant results for each gene, genomic region, or genetic variant. It also provides interactive tools that allow researchers to interact with datasets and perform custom queries while protecting data privacy.

"The rapid advances in human genetics of complex diseases have, to some extent, left the musculoskeletal field behind," says the Chair of the IFMRS MSK-KP Working Group, Dr. Douglas Kiel, Senior Scientist at the Hinda and Arthur Marcus Institute for Aging Research, Professor of Medicine at Harvard Medical School, and Associate Member at the Broad Institute. "We are excited to be able to come together as a scientific community to collect and curate 'omic data on diseases like osteoporosis, osteoarthritis, and sarcopenia on a state-of-the-art knowledge portal. This resource will provide a useful platform for discovery and efficient use of the unique data that we all have to contribute."
Exploring the MSK-KP

There are three major entry points to the results in the MSK-KP:

• enter a gene name, variant ID, or genomic coordinates into the home page search box to view a page with details on genetic associations and epigenomic annotations in that region
• select a phenotype to view a genome-wide plot and table of the top associations for that phenotype
• use the Variant Finder tool to retrieve a list of variants meeting custom criteria

Watch this video for a brief tutorial on navigating the MSK-KP. More written and video documentation will be available on the MSK-KP Resources page.

Currently, the MSK-KP includes genetic association datasets relevant to osteoporosis from the musculoskeletal research community as well as representative datasets for anthropometric, cardiovascular, and metabolic traits. Genetic association datasets are listed and described on the Datasets page. Osteoblast DNAse hypersensitive regions and ATAC-seq peaks, both indicative of open chromatin, are displayed with regional LocusZoom plots. Results from computational methods that integrate multiple data types to predict associations between genes, variants, tissues, and phenotypes are also available. In the future, diverse "big data" types will be added: not only genetic association studies, but also epigenomics, chromatin conformation, transcriptomics, functional results, and more. While the focus now has been on osteoporosis, the next area will be osteoarthritis. Other musculoskeletal disease focus areas will be determined in the future.

About the project

The MSK-KP project is a collaboration between the International Federation of Musculoskeletal Research Societies (IFMRS) and scientists and software engineers at the Broad Institute. The IFMRS was conceived in 2012 to provide scientific opportunities for its member societies with regards to education, collaboration and funding; the IFMRS Big Data Working group is charged with developing a means to share musculoskeletal databases globally. "The IFMRS is excited to work with the Broad to fulfill the goal of providing musculoskeletal data not only to investigators in countries rich in resources, but also sharing with those who may not have access to this information," says Dr. Lynda Bonewald, Executive Director of the Indiana Center for Musculoskeletal Health and Professor of Anatomy, Cell Biology, Physiology and Orthopaedic Surgery at Indiana University School of Medicine.

Along with the other portals of the Knowledge Portal Network, the MSK-KP is powered by the Human Genetics Amplifier (HuGeAMP) software platform, developed at the Broad Institute with support from the Accelerating Medicines Partnership in Type 2 Diabetes.

As we develop the MSK-KP into a comprehensive resource facilitating the translation of big data into actionable knowledge for human health, we welcome all suggestions, comments, questions, and submissions of relevant datasets; please contact us, and sign up to receive email updates on the project.