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Disease Portals: A Platform for Genetic and Genomic Research



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Short Abstract: The Disease Portals at the Rat Genome Database provide a comprehensive platform for physiological genomics discovery through the integration of heterogeneous datasets into the context of the genome using multiple ontologies and sophisticated data mining and visualization tools.

Long Abstract:

The Disease Portals at the Rat Genome Database provide a comprehensive platform for physiological genomics discovery through the integration of heterogeneous datasets into the context of the genome using multiple ontologies and sophisticated data mining and visualization tools. RGD serves a disparate community of users often defined by specific disease research areas and the Disease Portals provide both the novice and experienced user with an easy access to a comprehensive, integrated knowledgebase that can be tailored to the particular interests of the user. In addition, these initiatives define the focus and scope for data acquisition and curation projects. Current and planned components of the Disease Portals include: 1) comprehensive rat, human and mouse gene sets associated with diseases, related phenotypes, pathways and biological processes; 2) all rat QTLs related to the disease area as well as associated mouse and human QTLs; 3) rat strains used as models in studying these diseases; 4) rat phenotype data including values and experimental conditions for model strains; 5) gene expression data; 6) related references; 7) genome-wide view of disease genes and QTLs via GViewer; 8) comparative maps of disease related regions, 10) customization of datasets and download options; 11) analysis and visualization of function and cellular localization makeup of gene sets. These portals are designed to highlight genetic and genomic data generated from rat research in diseases related to the cardiovascular, nervous, musculoskeletal, digestive, endocrine and immune systems as well

as metabolic diseases and cancer.

The disease portals have been built using a combination of next-generation web technologies including Ajax-enabled navigation components, custom-developed Flash components, and 3rd-party graphing components utilizing Flash and XML. As the user drills down into specific disease categories, back-end components produce lists of annotated genes, QTLs and strains, categorized using multiple ontologies and presented in tabular format, along with a graphical view using the GViewer component (annotated objects placed on a visual representation of the genome). Using the GOSlim ontology (rolled-up GO terms), the disease portal also presents dynamic pie charts showing representation of the disease associated genes across biological processes (BP), molecular functions (MF), cellular components (CC), and pathways (PW).

The goal of the disease portals is to provide researchers with a singular view of genetic and genomic data related to a particular disease both within a species and across species. Researchers can then use this portal as a starting point for linking out to relevant sources to further explore the disease. RGD expects the disease portals to evolve as more people use them and discover better ways to visualize the data and integrate it with other relevant sources of information.