The RefSeqGene Project

A collection of sequences as foundation for gene-based coordinates

https://www.ncbi.nlm.nih.gov/refseq/rsg/

National Center for Biotechnology Information • National Library of Medicine • National Institutes of Health • Department of Health and Human Services

Scope of the RefSeqGene project

RefSeqGene, a subset of NCBI's Reference Sequence (RefSeq) project, defines genomic sequences to be used as reference standards for well-characterized genes. These sequences provide a stable foundation for reporting mutations, establishing conventions for numbering exons and introns, and defining the coordinates of variations such as single nucleotide (SNV), multiple nucleotide (MNV), insertions, and deletions. Sequences from this project solve the drawbacks and limitations present in mRNA- or whole chromosome-based systems by providing gene-specific genomic sequences for each gene with its upstream, intronic, and downstream flanking regions. Modifications made to RefSeqGene sequences are versioned and a tool is provided to facilitate conversion of coordinates among versions: www.ncbi.nlm.nih.gov/genome/tools/remap/#tab=rsg

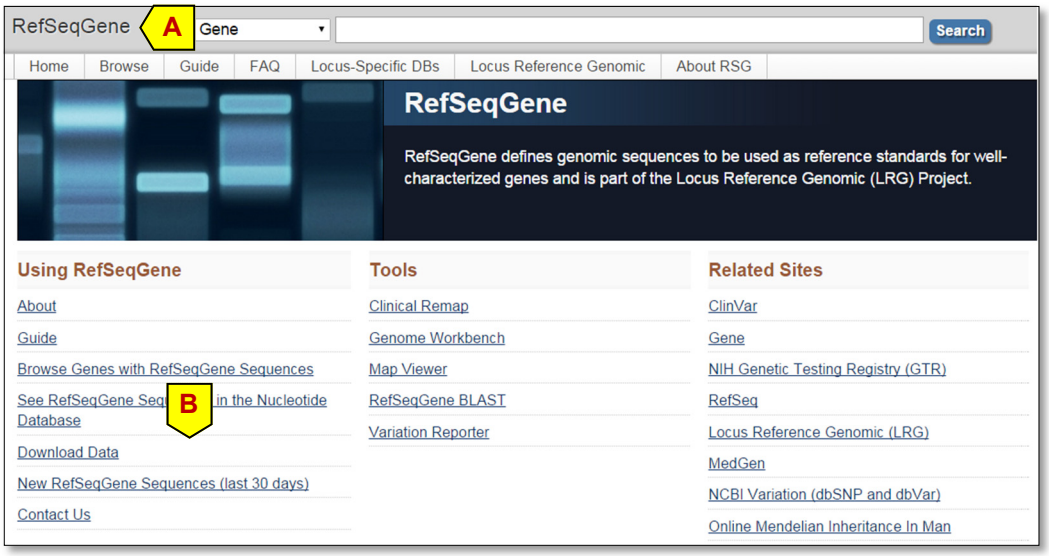
The RefSeqGene project is an active member of the Locus Reference Genomic (LRG) collaboration. Input and leadership from Dr. M. L. Gulley and the Molecular Pathology Resource Committee of the College of American Pathologists has greatly facilitated its implementation [1].

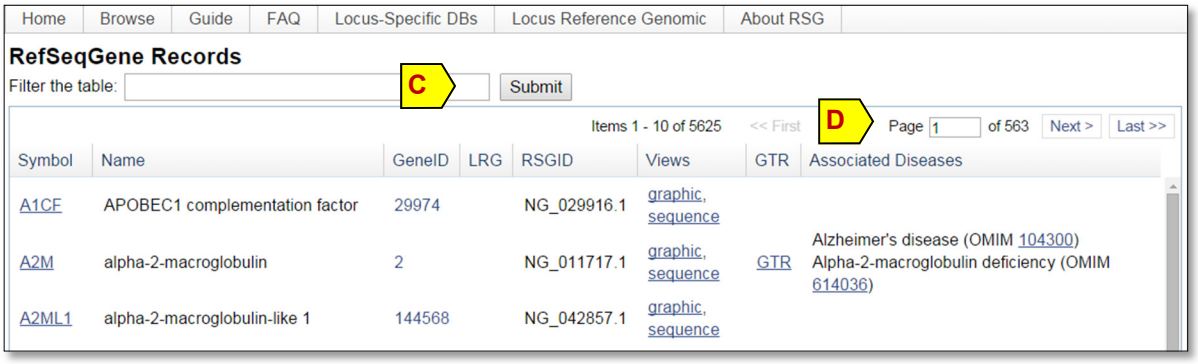
Sequence selection

Sequences in the RefSeqGene set represent well-supported, naturally occurring haplotypes, and prevalent alleles. The RefSeqGene group collaborates with multiple Locus Specific Databases (LSDBs) and the LRG project of GEN2PHEN [2] to establish and maintain these standard sequences. The RefSeqGene-annotated genes with identified LSDB counterparts can be retrieved from NCBI Gene (www.ncbi.nlm.nih.gov/gene) using the term *refseqgene*. Sequences of RefSeqGene entries can be retrieved using fielded term *refseqgene[keyword]* from the NCBI Nucleotide database (www.ncbi.nlm.nih.gov/nuccore).

Data access

Over 5,300 RefSeqGene records are available to serve as the foundation for gene-based coordinates. The RefSeqGene homepage (**A**) provides access to browse and search for RefSeqGene entries (**B**). It lists the available entries in a summary table and provides official symbols, gene name, GeneID, and other information. The list can be filtered by terms entered in the text box (**C**) or browsed by paging (**D**). RefSeqGene sequences are also accessible through the BLAST homepage.



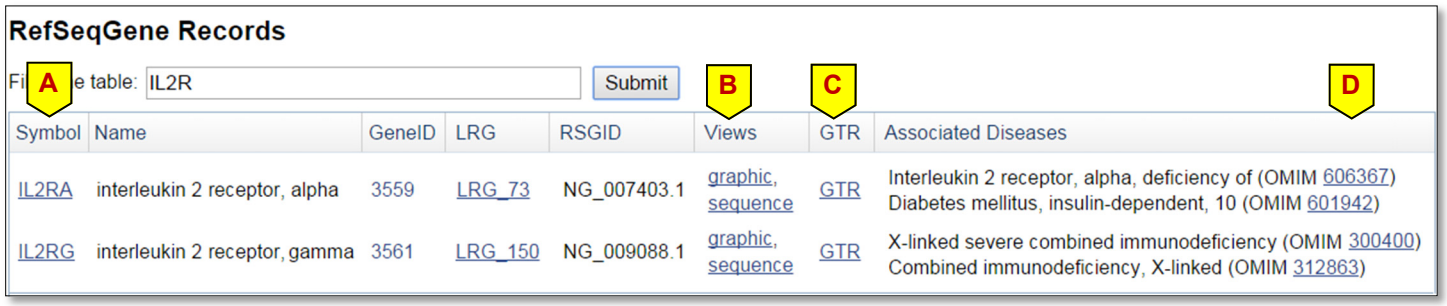


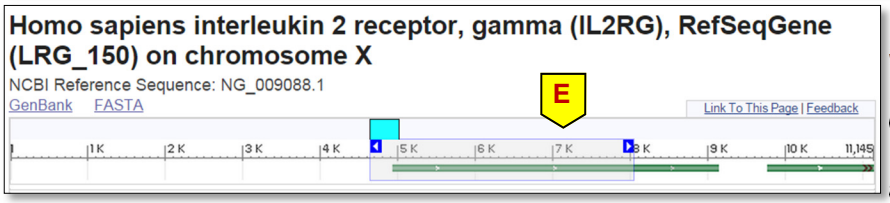
1. Gulley et al. Clinical laboratory reports in molecular pathology. 2007. Arch Pathol Lab Med. 131(6): 852 -63.

2. Dalgleish et. al. Locus Reference Genomic sequences: an improved basis for describing human DNA variants. 2010.

Contents of a gene record

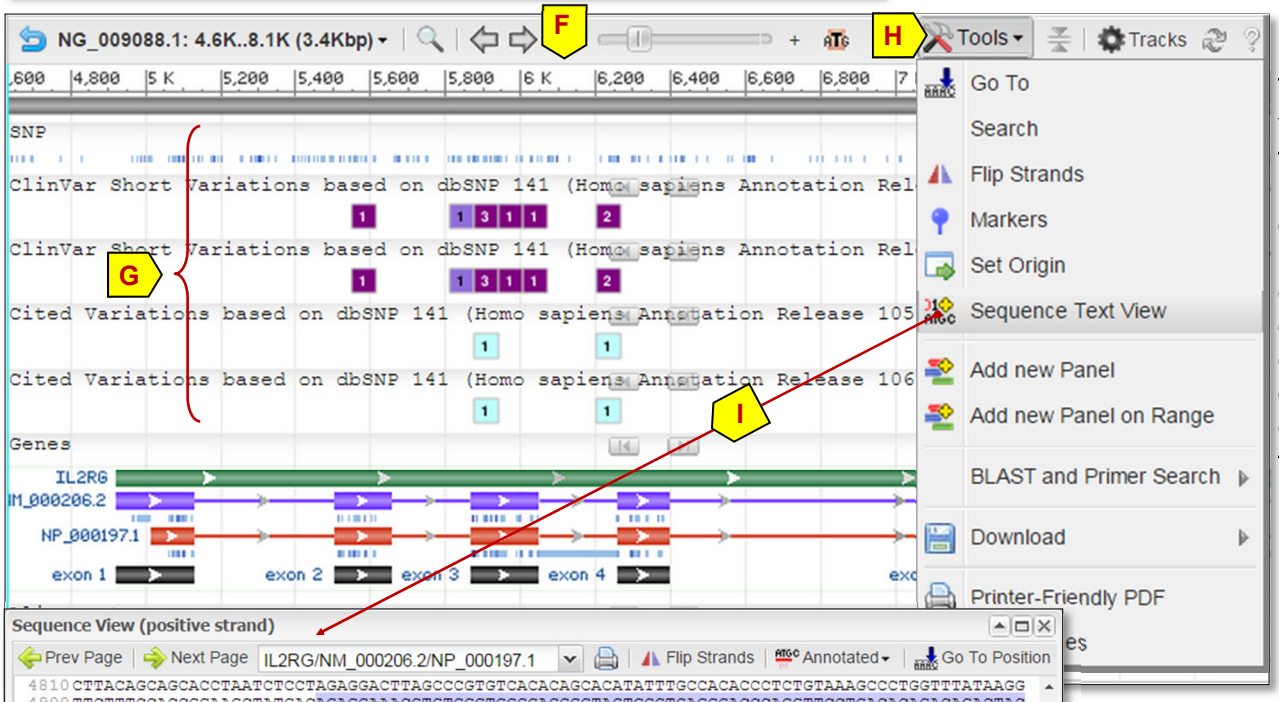
In the RefSeqGene Browse list (shown below), summary information and links to records in other databases are provided for each entry. Here, official gene symbols (**A**) in the "Symbol" column link to Gene records. The "graphic" (**B**) in the "Views" column displays sequences in the NCBI Sequence Viewer. The "GTR" column (**C**) provides links to the NIH Genetic Testing Registry (GTR). Additionally, IDs (**D**) in the "Associated Diseases" columns link to OMIM records with disease-specific information.





Sequence Viewer display

In the Sequence Viewer display (left), the complete gene locus, represented by the RefSeqGene, is depicted at the top (**E**) and details for the Gene are shown in the panel below (**F**). The set of variation tracks (**G**) provides an comprehensive overview of existing data in the context of gene features.



Clicking the "Tools" icon (**H**) opens a popup menu allowing the selection of "Sequence Text View" option to display the annotated sequences in a floating text window (**I**) within the Sequence viewer display.