

BIOBASE Product Release

Q3 2010

Summary of key release points:

- Addition of clinical trials data from clinicaltrials.gov, providing connections between diseases and drugs within the BKL
- Expansion of the Ontology Search tool to additionally support controlled vocabulary queries for diseases and drugs
- BIOBASE launches Genome Trax™, a new product that facilitates mapping of known genomic features to human genomic sequences for analysis of variations

Addition of clinical trials data:

- The data has been integrated from [clinicaltrials.gov](http://www.clinicaltrials.gov), a registry of clinical trials conducted in the US and other countries that is maintained by the US National Institutes of Health

<http://www.clinicaltrials.gov/>

- We have automatically processed >90,000 trial records, extracting those that map to a BKL disease entry and a BKL drug entry (remember that BKL drug entries are all taken from DrugBank)
- Key information regarding mapped trials is presented in a new Clinical Trials section on Disease Reports and Drug Reports

Disease Reports:

http://devel.incyte.com/cgi-bin/build_ghptywl/idb/1.0/pageview.cgi?view=DiseaseReport&mesh_id=D003882

Disease Report

Dermatomyositis [Synonyms : Dermatopolymyositis](#)

[Home](#)

Mousing over "i" information link open pop-up containing the trial summary.

Clinical Trials

Drugs under clinical trial investigation for treatment of
Provided by ClinicalTrials.gov

Drug	Phase				
Cyclosporine	Phase 3	Five-year Actively Controlled Clinical Trial in New Onset Juvenile Dermatomyositis	View study report	Recruiting	May 2006 - May 2011
Etanercept	Phase 2/Phase 3	Understanding the Pathogenesis and Treatment of Childhood Onset Dermatomyositis	View study report	Terminated	August 2002 - August 2002
Etanercept	N/A	A Pilot Study of Etanercept in Dermatomyositis	View study report	Active, not recruiting	January 2006 - May 2010
I-5	Phase 1	A Study to Evaluate Safety of Multi-Dose MEDI-545 in Adult Patients With Dermatomyositis or Polymyositis	View study report	Active, not recruiting	April 2007 - September 2010
Infliximab	Phase 2	Infliximab (Remicade(Registered Trademark)) to Treat Dermatomyositis and Polymyositis	View study report	Completed	April 2002 - May 2010

Juvenile dermatomyositis (JDMS) is one of the most serious of the childhood rheumatic diseases. The theory behind this trial is that early introduction of etanercept or methotrexate will prove to be effective in the treatment of JDMS. Pretreatment muscle biopsies, we believe there will be abnormalities in the blood vessels that will be correlated with worse physical strength and daily functional ability. The long-term goal is to improve the treatment of this serious childhood onset rheumatic disease and to better understand the pathogenic mechanism for the development of the vasculopathy (disorder of blood vessels) of JDMS. Identification of the specific mechanism of the vasculopathy may allow for the rational introduction of biologic treatments focused on vascular growth.

[\[more ...\]](#)

Link to Drug Reports

Clicking the "View study report" button opens the study report at the clinicaltrials.gov site.

ClinicalTrials.gov
A service of the U.S. National Institutes of Health

[Home](#) [Search](#) [Study Topics](#) [Glossary](#)

[Full Text View](#) [Tabular View](#) [No Study Results Posted](#) [Related Studies](#)

Understanding the Pathogenesis and Treatment of Childhood Onset Dermatomyositis

This study has been terminated.
(Incorporating the recommendations of the NIH-formed DSMB in the study procedures would make the project budget over the limit for this funding mechanism.)

First Received: May 7, 2002 Last Updated: April 15, 2009 [History of Changes](#)

Sponsor:	National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS)
Collaborator:	Immunex Corporation
Information provided by:	National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS)
ClinicalTrials.gov Identifier:	NCT00035958

Drug Reports:

http://devel.incyte.com/cgi-bin/build_ghptywl/idb/1.0/pageview.cgi?view=DrugReport&drug_acc=DR000003905

Drug Report
Home

Etanercept

Synonyms : etanercept, Tumor necrosis factor receptor 2, Tumor necrosis factor receptor superfamily member 1B precursor, CD120b, p75, Tumor necrosis factor receptor type II, TNF-R2, p80 TNF-alpha receptor

Brand name : Enbrel

Clinical Trials

Disease for which Etanercept is under clinical trial
Provided by ClinicalTrials.gov

Disease	Phase	Study Title	Status	Start Date	End Date
Uveitis	Phase 2	TNRF:Fc to Treat Eye Inflammation in Juvenile Rheumatoid Arthritis (i)	Completed	February 1999	March 2003
Arthritis	Phase 2	TNRF:Fc to Treat Eye Inflammation in Juvenile Rheumatoid Arthritis (i)	Completed	February 1999	March 2003
Vasculitis	Phase 2	Etanercept to Treat Wegener's Granulomatosis (i)	Completed	February 1999	March 2005
Granuloma	Phase 2	Etanercept to Treat Wegener's Granulomatosis			
Sjogren's Syndrome	Phase 2	Etanercept Therapy for Sjogren's Syndrome			

[\[more ...\]](#)

Home Search Study Topics Glossary

Full Text View
Tabular View
No Study Results Posted
Related Studies

TNRF:Fc to Treat Eye Inflammation in Juvenile Rheumatoid Arthritis

This study has been completed.

First Received: November 3, 1999 Last Updated: March 3, 2006 [History of Changes](#)

Sponsor:	National Eye Institute (NEI)
Information provided by:	National Institutes of Health Clinical Center (CC)
ClinicalTrials.gov Identifier:	NCT00001862

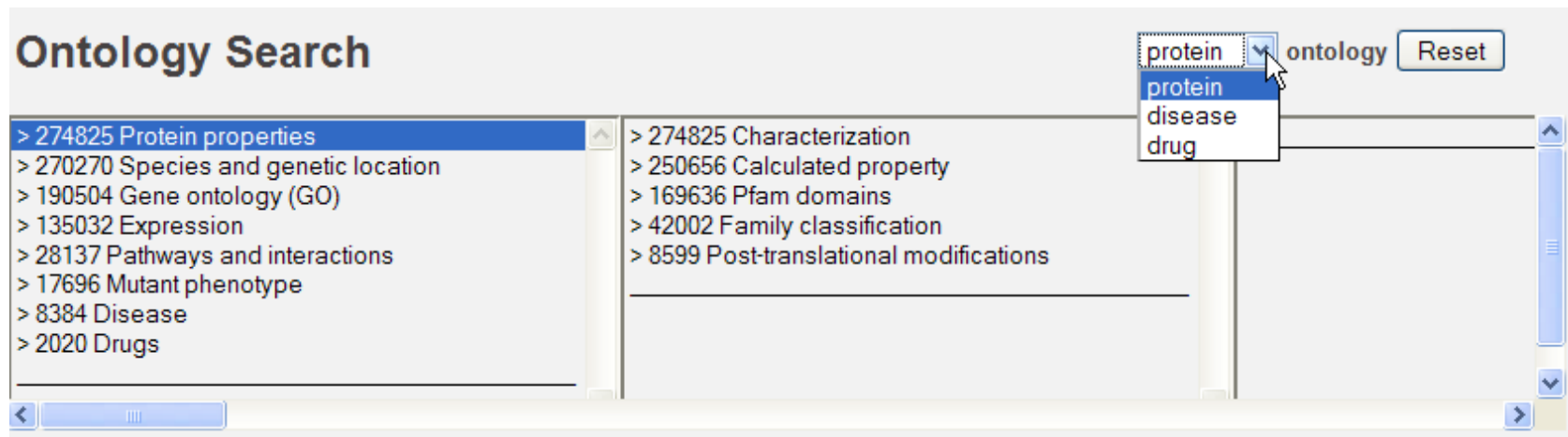
Link to Disease Reports

Clicking the "View study report" button opens the study report at the clinicaltrials.gov site.

Mousing over "i" information link open pop-up containing the trial summary.

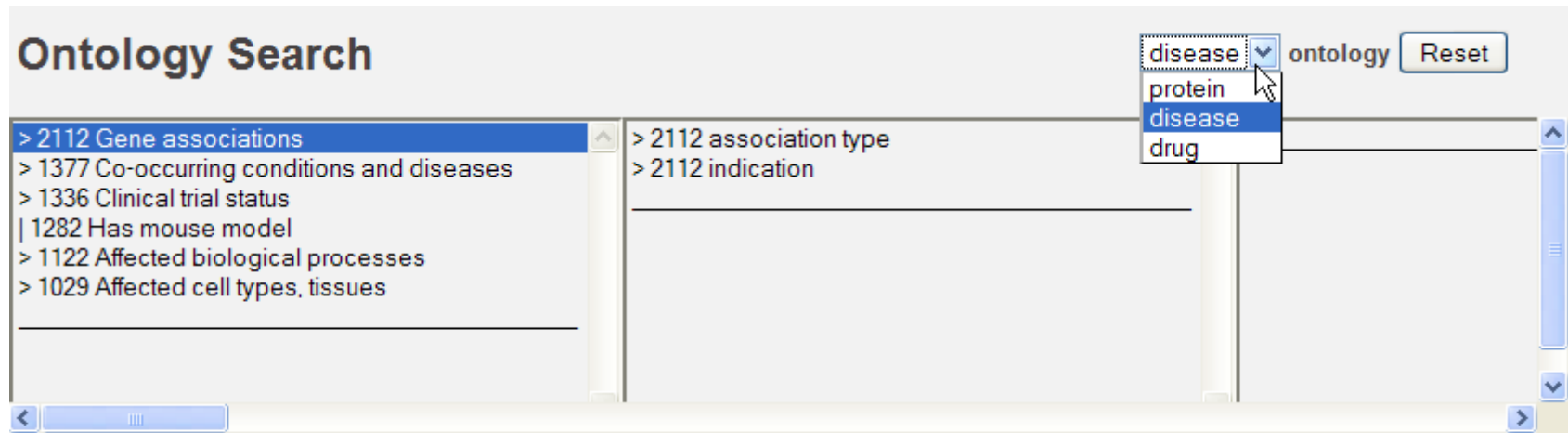
Expansion of the Ontology Search tool:

- Users can now use the Ontology Search tool to answer questions about diseases and drugs, in addition to proteins
- Users toggle between the search options using the new pull-down menu:



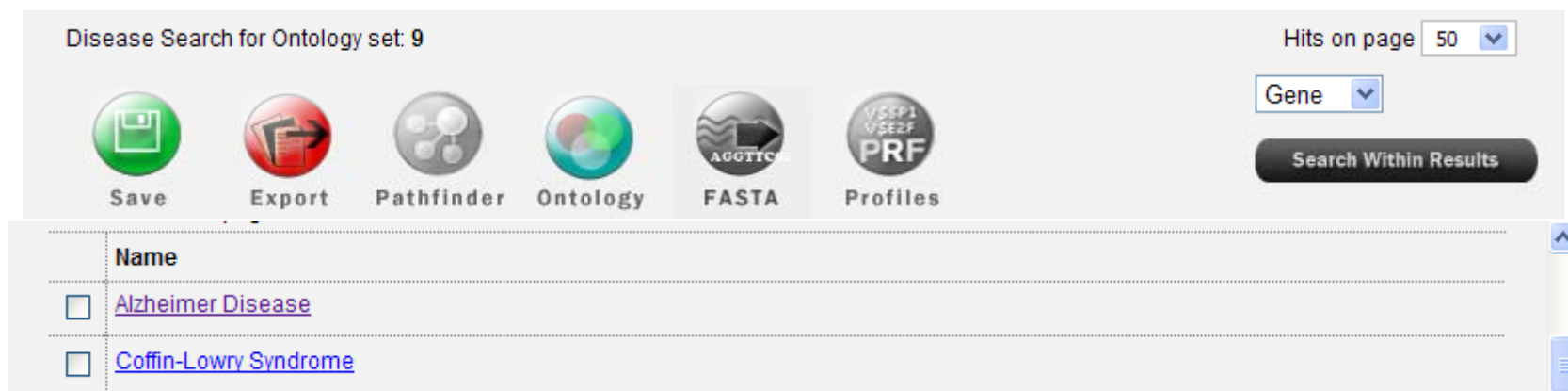
- As an aside, you'll notice that the regular protein hierarchies have been condensed into a more compact and user friendly presentation

- As a different search option is selected, the hierarchies are automatically reset:



The screenshot shows the 'Ontology Search' interface. At the top right, there is a dropdown menu currently set to 'disease'. A mouse cursor is hovering over the 'disease' option, which has opened a list of options: 'disease', 'protein', 'disease', and 'drug'. To the right of the dropdown is a 'Reset' button. Below the dropdown, the main content area is divided into two columns. The left column shows a list of categories with expandable arrows: '> 2112 Gene associations', '> 1377 Co-occurring conditions and diseases', '> 1336 Clinical trial status', '| 1282 Has mouse model', '> 1122 Affected biological processes', and '> 1029 Affected cell types, tissues'. The right column shows '> 2112 association type' and '> 2112 indication'. At the bottom of the interface, there are navigation arrows and a scrollbar.

- Searches work as normal, returning the relevant diseases or drugs as results:



The screenshot shows the 'Disease Search for Ontology set: 9' interface. At the top right, there is a 'Hits on page' dropdown set to '50'. Below this is a 'Gene' dropdown menu. A 'Search Within Results' button is located below the dropdowns. In the center, there are six icons representing different search options: 'Save', 'Export', 'Pathfinder', 'Ontology', 'FASTA', and 'Profiles'. Below the icons, there is a table of search results. The table has a header 'Name' and two rows of results, each with a checkbox and a link to the disease name: 'Alzheimer Disease' and 'Coffin-Lowry Syndrome'. At the bottom right of the table, there are navigation arrows and a scrollbar.

Example questions that can be answered include:

- Which diseases are associated with an altered immune response?
- Which of those are associated with a clinical trial in phase 3 or later?
- Which diseases are known to co-occur with hypertension?
- Which drugs are under investigation for treatment of diabetes?
- What pathways are those drugs associated with?
- Which pathways are associated with drugs that have been withdrawn from the market?

Genome Trax tool is launched:

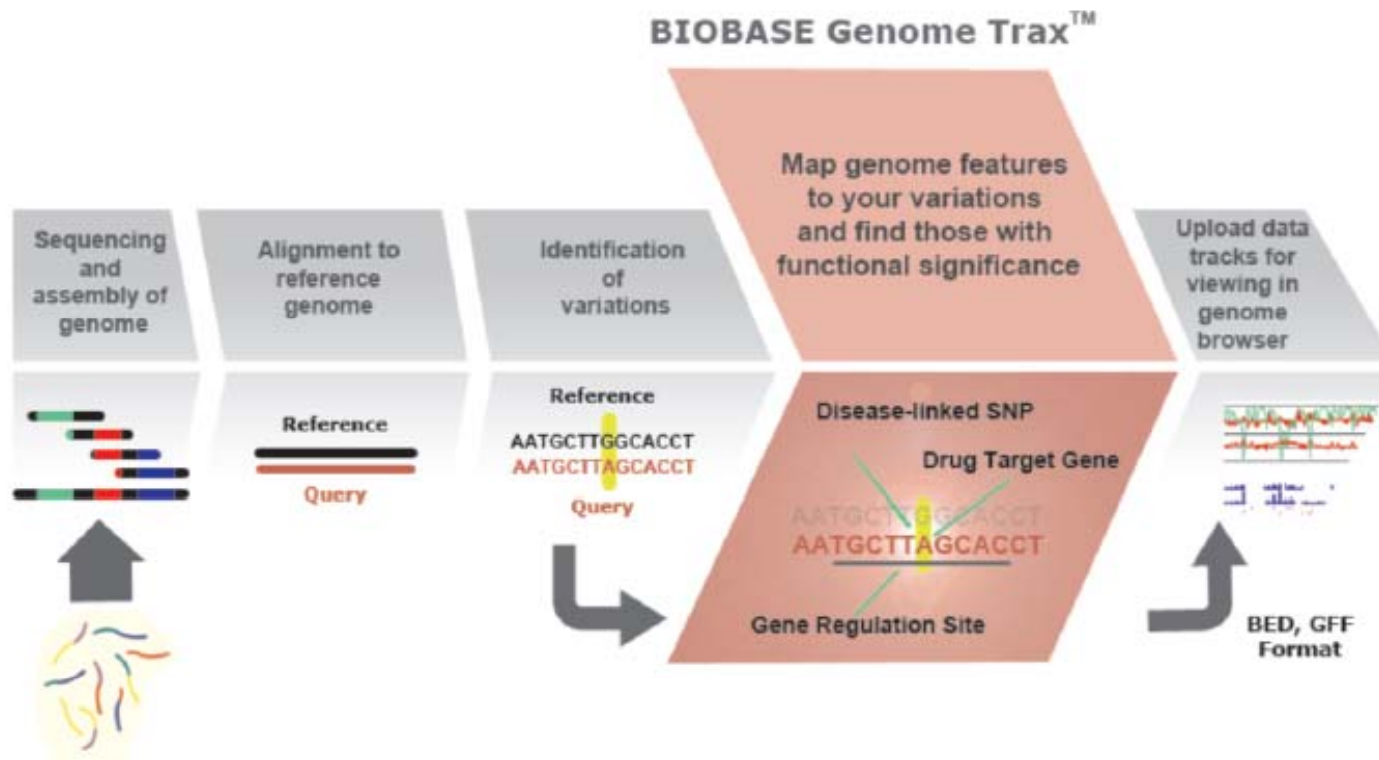
- All BKL subscribers will see a button for the new Genome Trax tool that is sold by separate subscription



- The button will be “on” (colored and clickable) for all online users. When clicked the user will be directed to the appropriate portal login screen and will only be able to gain access if they are a subscriber – in the same manner as the ExPlain link currently behaves.
- By default, the button will be “off” (gray and unclickable) for all installed customers, but it can be configured to be activated and point to our hosted online installation should the customer choose to upgrade their subscription. No local installations will be offered at this time.

Why would customers be interested in Genome Trax?

- It allows the user to identify which of the many variations found in their sequence are most likely to have functional significance, based on current knowledge of disease causing mutations and regulatory features



- The tool allows a user to upload a file of genomic coordinates specifying the location of sequence variations, which are then mapped to the genomic features that we have annotated in our database

Input Data

[Upload TXT Results](#)

1 Upload genomic coordinates of your variations: OR
(chr# <tab> start coordinate <tab> end coordinate)
[Don't see the browse button? Or having trouble uploading files?](#)

2 Select reference genome used in your analysis:


3 Annotate variations with the following features:

[Select/Deselect all](#)

Mutations and variations	Regulatory features
<input checked="" type="checkbox"/> Inherited disease mutations	<input checked="" type="checkbox"/> Experimentally verified TFBS
<input checked="" type="checkbox"/> dbSNP regulatory SNPs	<input checked="" type="checkbox"/> Predicted ChIP-Seq TFBS
<input checked="" type="checkbox"/> Ensembl regulatory SNPs	<input checked="" type="checkbox"/> Histone binding fragments
	<input checked="" type="checkbox"/> CpG islands
	<input checked="" type="checkbox"/> Microsatellites
	<input checked="" type="checkbox"/> Virtual Transcription Start Sites (TSSs)
	<input checked="" type="checkbox"/> Post translational modifications

4 Map variations

- The features are displayed in a tabulated form with links provided, when relevant, to BKL Site Reports and Fragment Reports, as well as HGMD mutation information, and dbSNP and Ensembl SNP reports

Search Results 

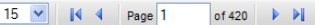

[New Search](#) [Upload TXT Results](#)

[View Feature Summary](#) [TXT Format](#) [BED Format](#) [GFF Format](#)

Filter Bar [Clear filters](#)

Type: Chromosome: [Go](#)

Type	Description	Chromosome	Feature Start	Feature End	Strand
CpG island	GC(%): 65 CpGs: 188	chr21	10989725	10991755	.
CpG island	GC(%): 64 CpGs: 38	chr21	15069582	15070142	.
CpG island	GC(%): 65 CpGs: 100	chr21	15076862	15078022	.
CpG island	GC(%): 64 CpGs: 68	chr21	15095390	15096320	.
CpG island	GC(%): 63 CpGs: 124	chr21	15435403	15437194	.
CpG island	GC(%): 61 CpGs: 80	chr21	15456221	15457462	.
CpG island	GC(%): 63 CpGs: 34	chr21	15646178	15646708	.
CpG island	GC(%): 62 CpGs: 31	chr21	15755178	15755692	.
CpG island	GC(%): 56 CpGs: 27	chr21	16299312	16299840	.
CpG island	GC(%): 55 CpGs: 25	chr21	17045836	17046336	.
CpG island	GC(%): 55 CpGs: 27	chr21	18748260	18748770	.
CpG island	GC(%): 65 CpGs: 145	chr21	18884621	18886271	.
CpG island	GC(%): 58 CpGs: 32	chr21	19093072	19093642	.
CpG island	GC(%): 55 CpGs: 28	chr21	19374952	19375522	.
CpG island	GC(%): 55 CpGs: 48	chr21	10884514	10885298	.

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- Users may export the data for viewing in formats supported by common genome browsers, or export the data for further analysis in other platforms

Benefits of using Genome Trax data compared to public data sets:

- Includes experimental transcription factor binding sites from TRANSFAC
- Includes the best predicted transcription factor binding site for experimental ChIP-Seq fragments, which provides a much more specific filter than the publicly available fragments
- Includes HGMD mutations
- Includes a subset of dbSNP and Ensembl SNPs that have been enriched due to co-occurrence with a known or predicted regulatory feature, which provides a much more specific filter than the full publicly available set of SNPs
- Includes histone binding fragments derived from the literature, not just ENCODE
- Includes a wealth of other computationally determined regulatory features

Availability of Genome Trax :

- An Online version is primarily targeted to bench scientists who want to analyze an individual genome sequence, or explore a particular area of a chromosome
- A Download version is primarily targeted to bioinformaticists and core facilities who want to parse the feature data for incorporation into their own sequence analysis pipeline or for use in their own specialized platforms. Download files are offered in two general formats: a BED format that has been optimized for use with the UCSC genome browser, and a GFF format that has been optimized for use with CLC Genomics Workbench.

Note that a freely accessible version of the tool is made available for education and evaluation purposes. This version is fully functional, but users are limited to the set of features associated with chromosome 21.

<http://custom.biobase-international.com/cgi-bin/ngs-free/index.cgi>