Developing an Online Encyclopedia on Genetic Variation and Human Health

Food for Thought!

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CDC National Office of Public Health Genomics

What is a Field Synopsis?

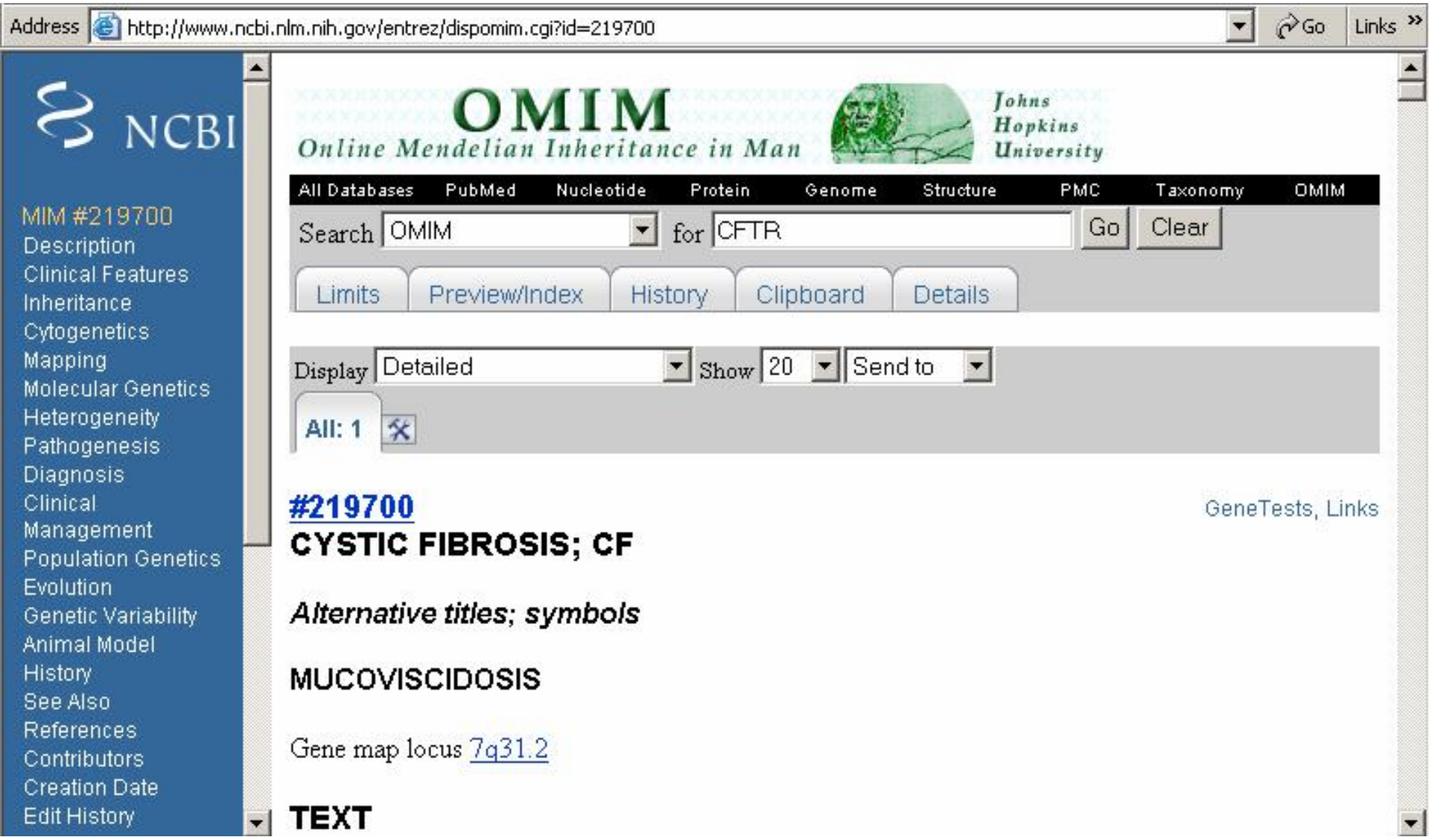
- A regularly updated snapshot of "what we know and what we don't know" about the association of a particular field (disease or group of conditions) with human genetic variation

- Some desirable features of field synopses
  - Systematic (published/unpublished)
  - Accessible online
  - Updated
  - Authoritative
  - Cumulative quantitative assessment
  - Grading of evidence
  - Linkage with other data/information

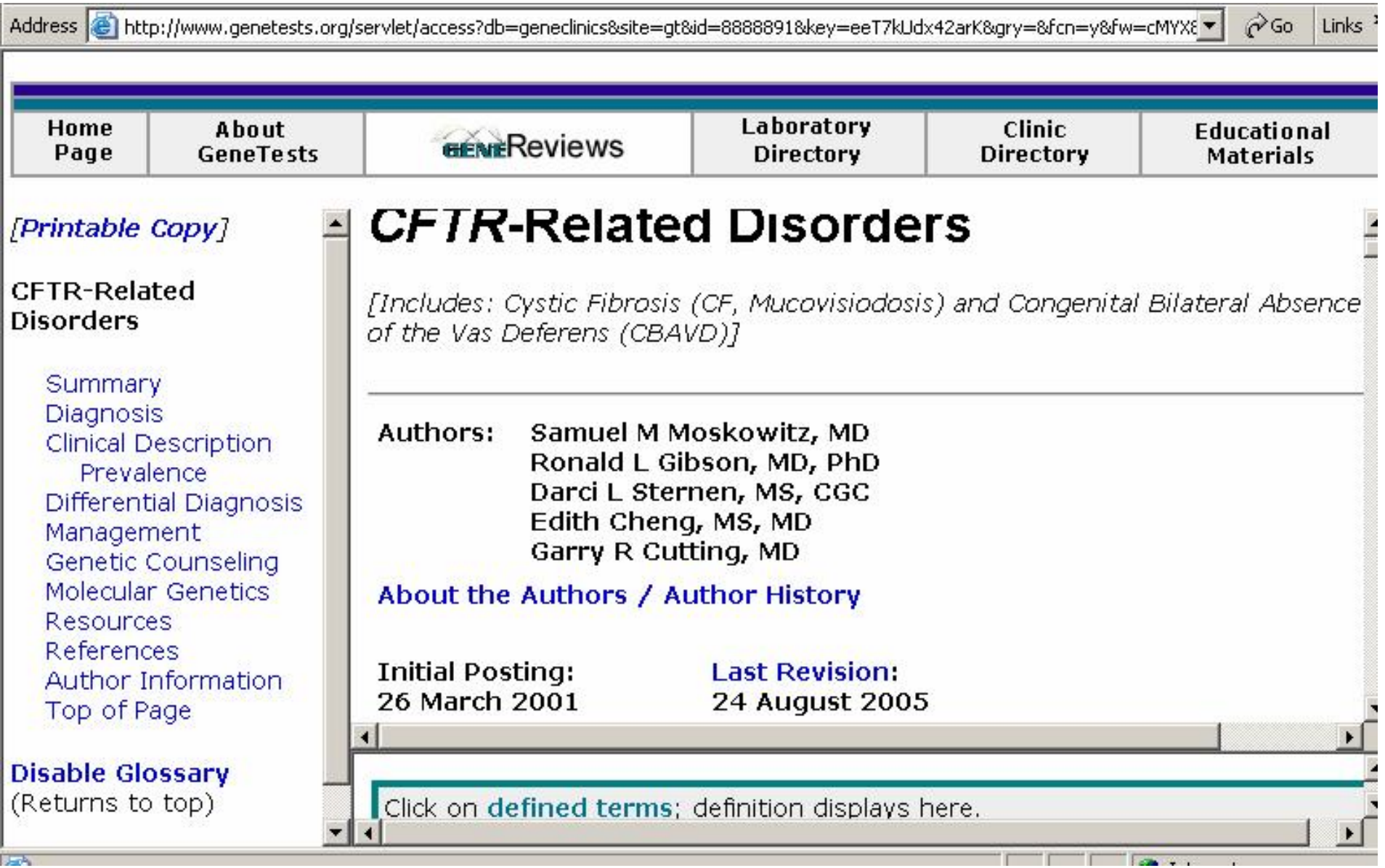
What is an Online Encyclopedia?

- Collection of field synopses but different models exist
- Is it impossible to do in the field of genomics given rapidly changing information?
- A proposal for a collaborative approach

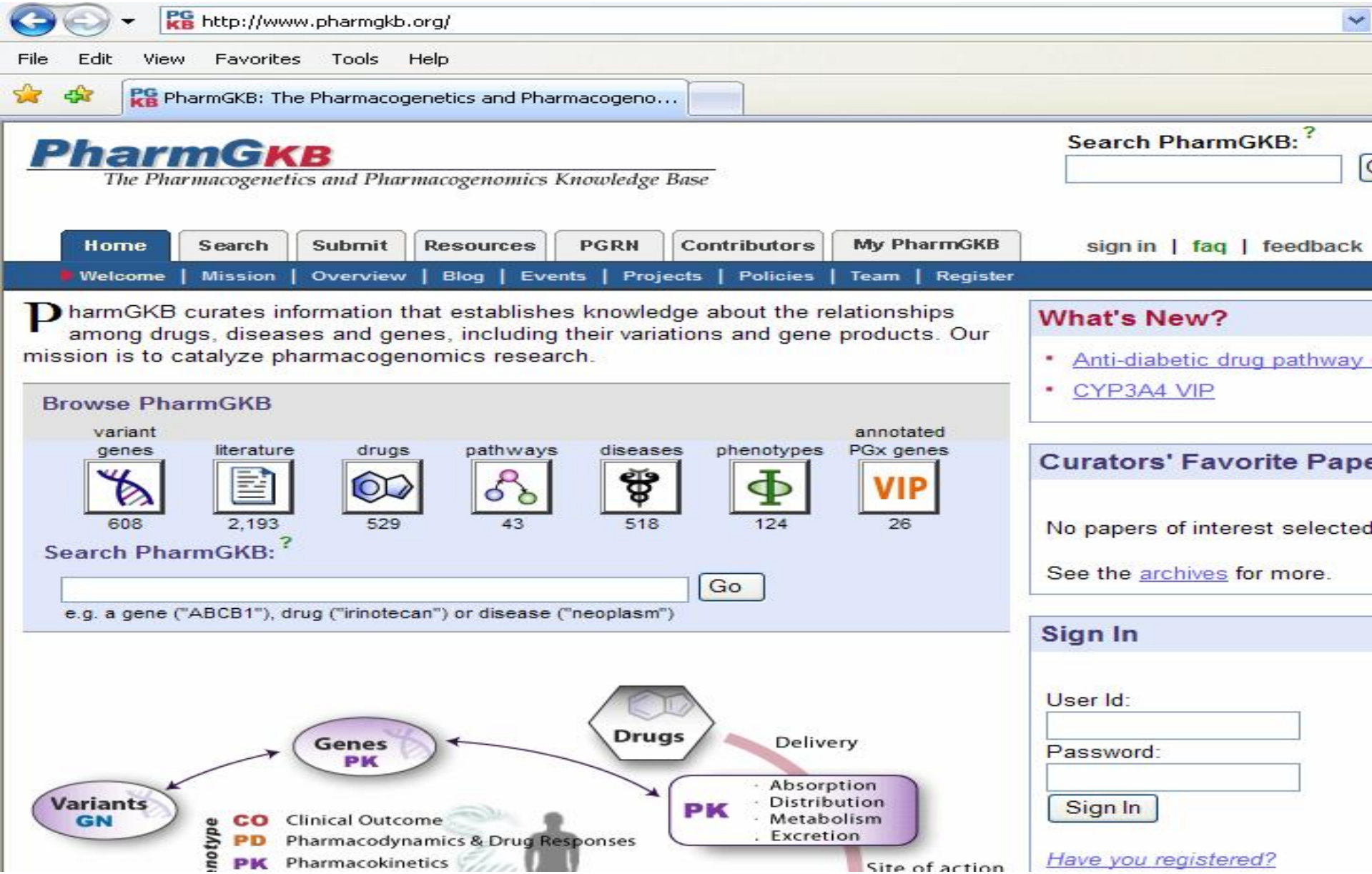
Models for Online Encyclopedia 1



Models for Online Encyclopedia 2



Models for Online Encyclopedia 3



Models for Online Encyclopedia 4

Models for Online Encyclopedia 5

Address http://en.wikipedia.org/wiki/Main_Page Go Links

Sign in / create account

main page | discussion | view source | history

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The Free Encyclopedia

navigation

- Main Page
- Community Portal
- Featured articles
- Current events
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search

Go Search

Welcome to Wikipedia,

the free encyclopedia that anyone can edit.

1,461,789 articles in English

- Arts
- History
- Society
- Biography
- Mathematics
- Technology
- Geography
- Science
- **All portals**

Overview · Searching · Editing · Questions · Help

Categories · Featured content · A–Z index

Today's featured article

Kochi is a city in the Indian state of Kerala, and one of the principal seaports of the country. Kochi is located in the district of Ernakulam, about 220 km north of the state capital Thiruvananthapuram. The city has an estimated population of 650,000, with an extended metropolitan population of over 1.6 million, making it the largest urban agglomeration and the second largest city in

In the news

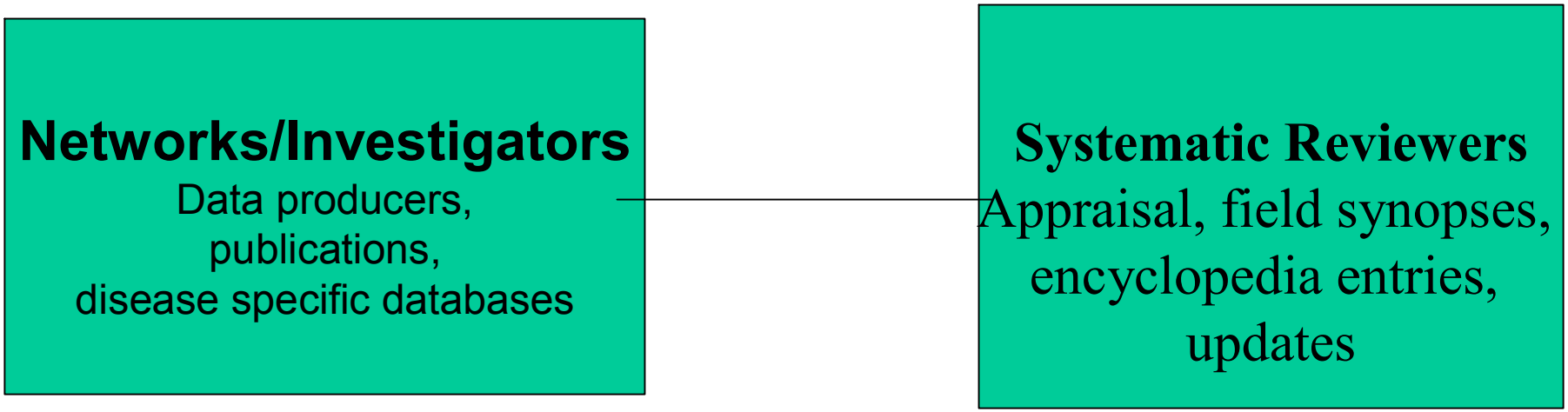
- The World Confederation of Labour and the International Confederation of Free Trade Unions merge to form the **International Trade Union Confederation**, representing 166 million members.
- Hassan Nasrallah, the Secretary General of Hezbollah, says negotiations

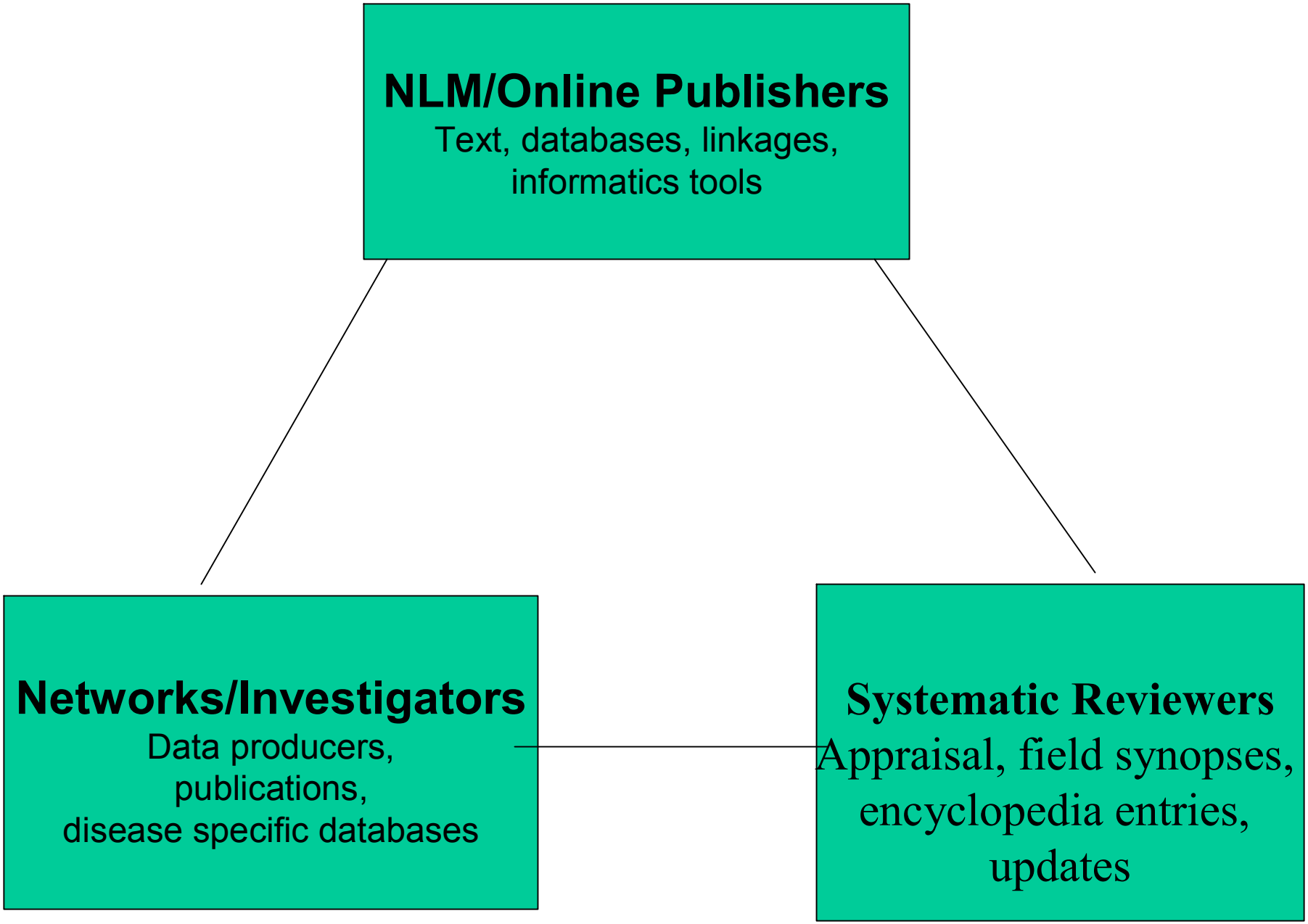
Collaborative Approach Needed

- **Investigators/**networks/consortia/GWAS/ biobanks
- **Systematic reviewers:** conduct field synopses and write knowledge base summaries
- **Methods developers:** tools and resources
- **Publishers**: journal editors, NCBI-connecting text and databases

Networks/Investigators

Data producers,
publications,
disease specific databases


Networks/Investigators
Data producers,
publications,
disease specific databases
Systematic Reviewers
Appraisal, field synopses,
encyclopedia entries,
updates


NLM/Online Publishers
Text, databases, linkages,
informatics tools
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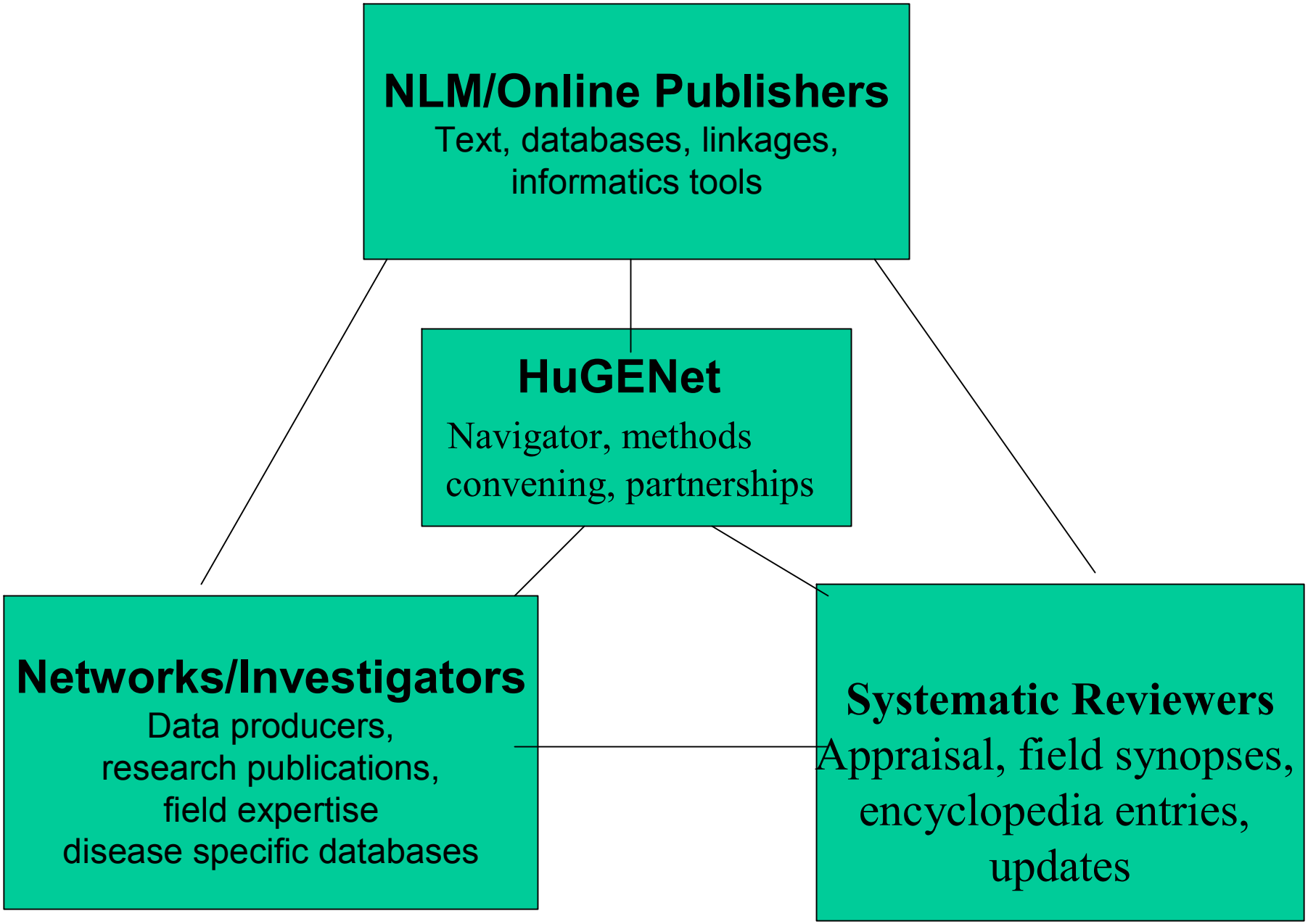
NLM/Online Publishers
Text, databases, linkages,
informatics tools
HuGENet
Navigator, methods
convening, partnerships
Networks/Investigators
Data producers,
research publications,
field expertise
disease specific databases
Systematic Reviewers
Appraisal, field synopses,
encyclopedia entries,
updates

HuGE Navigator (version 1.0)

An integrated, searchable knowledge base of genetic associations and human genome epidemiology.

HuGEpedia

- Select options, Disease or Gene.
- Enter one disease term or gene information (gene symbol, alias names etc) into the text box.
- Click All to see A-Z list of diseases or genes.
- Use the Search dropdown list to switch to other HuGE Navigator applications.

HuGEPedia is an online Human Genome Epidemiology (HuGE) encyclopedia that allows users search integrated information on genetic associations and other HuGE information including the published HuGE literature, HuGE investigators, candidate genes, association data extracted from meta-analysis publications and HuGE field synopses generated by domain experts, as well as analysis of temporal and geographic trends.

How to cite this site:

HuGE Navigator (version 1.0)

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HuGEpedia

Alzheimer Disease

Summary

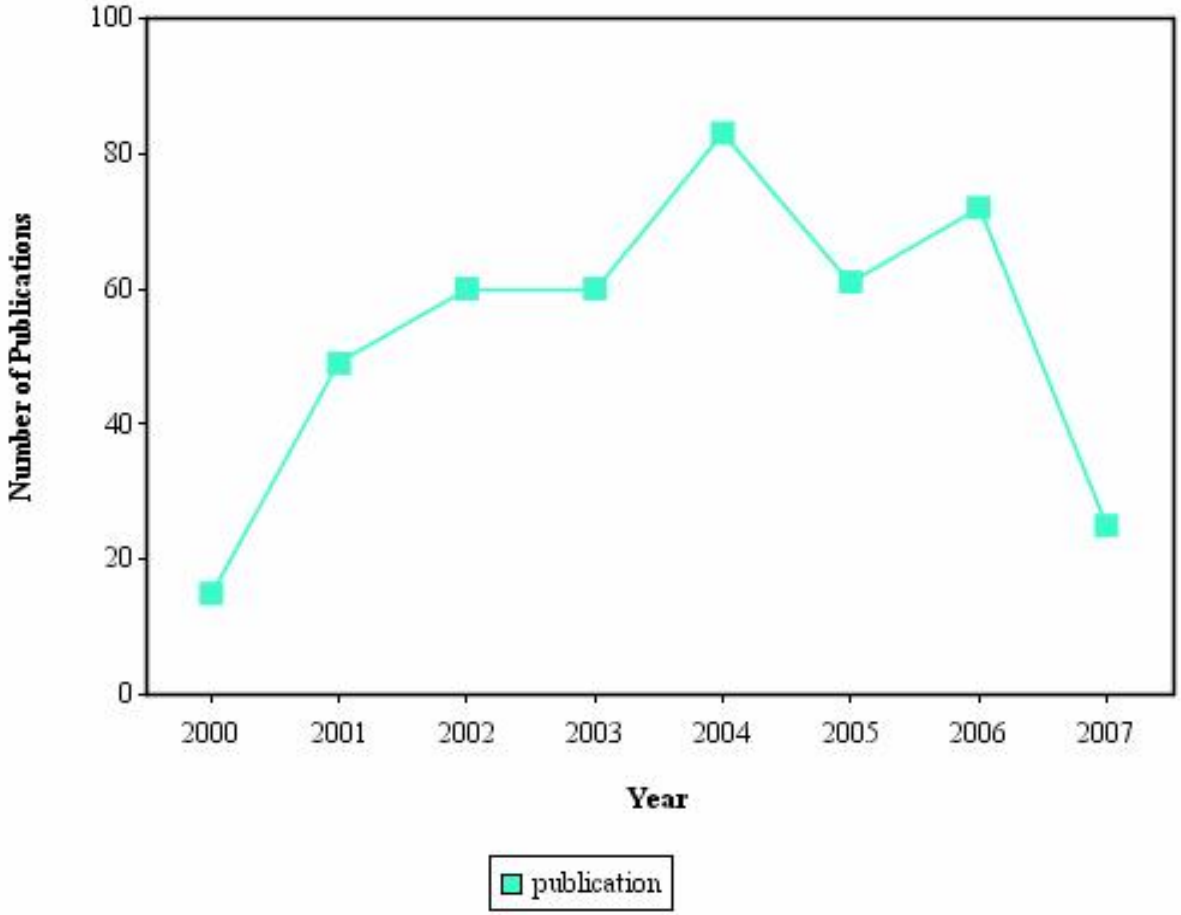
- Total Publications 1157
- Meta-Analyses 43
- MA Summary
- Genes 299
- GWAS Publications 6
- Investigators 1356(F/L) 4661(All)
- Trend/Pattern

Last Update: 27 Dec 2007

299 genes have been studied in association with Alzheimer Disease

[Click to re-sort the table]

Gene	Total Pub	MA Pub (Summary)	Trend/Pattern
APOE	589	20	
ACE	34	5	
PSEN1	32	3	
A2M	29	1	
IL1A	29	1	
IL1B	23	0	
BDNF	23	0	
MTHFR	23	1	
BCHE	22	1	
CYP46A1	21	1	
MAPT	19	1	
SLC6A4	18	0	
TNF	18	1	



Note: The chart illustrates the temporal trend based on Medline/PubMed records indexed with the disease MeSH term (Alzheimer's Disease). Un-MeSHed PubMed records are not included.

Trend/Pattern Information - Windows Internet Explorer provided by ITSO

http://hugenavigator.net/HuGENavigator/getWatch.do?type=disease_gene&geneSymbol=APOE&cuiID=C0002395

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Done

Internet

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Field Synopsis
by Bertram et al.

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Alzheimer

GWAS Database
NA

Disease Site
AlzGene

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SLC6A4	18	0	
TNF	18	**1**	
CTSD	18	**1**	
IL6	17	0	
LRP1	15	**3**	
PON1	14	0	
IDE	14	**1**	
CST3	14	**1**	
ESR1	14	**2**	
APP	13	0	
PRNP	13	**5**	
PSEN2	13	0	
IL10	12	0	
SERPINA3	12	**1**	
LDLR	11	**1**	
HFE	11	0	
NOS3	11	**1**	
CHAT	10	0	
ABCA1	10	0	
BACE1	10	**2**	
COMT	9	0	
HTR2A	9	0	
TF	9	**1**	
TGFB1	8	0	
MPO	8	**2**	
NCSTN	8	**1**	
APOC1	7	0	
IL1RN	7	0	
UBQLN1	7	0	

1: Nat Genet. 2007 Jan;39(1):17-23. Related Articles, Links

nature genetics

Systematic meta-analyses of Alzheimer disease genetic association studies: the AlzGene database.

Bertram L, McQueen MB, Mullin K, Blacker D, Tanzi RE.

Genetics and Aging Research Unit, MassGeneral Institute for Neurodegenerative Disease (MIND), Department of Neurology, Massachusetts General Hospital, Charlestown, Massachusetts 02129, USA. bertram@helix.mgh.harvard.edu

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ALZGENE - PUBLISHED AD CANDIDATE GENES

AlzGene

Updated 31 August 2007

Chromosome: 1 2 3 4 5 6 7 8 9 10 11 12 13
14 15 16 17 18 19 20 21 22 X Y

Gene: -- Select -- Go

Protein: -- Select -- Go

Polymorphism: -- Select -- Go

Study: -- Select -- Go

Keyword: Go

AlzGene Recent Updates

PSEN2 Go

Top AlzGene Results

View Top Results Methods

1. APOE (ε2/3/4)
2. CHRNB2
3. CH25H
4. PGBD1

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Meta Table for Alzheimer Disease and PRNP

Variant: codon 129
Gene(s): (PRNP)

Phenotype	Reference	Analysis Type*	#Studies	#Cases	#Controls	Contrast	Effect size*	Het*
Alzheimer's disease	Qian HR, 2006	A	4	1095	940	(MM + VV) vs. MV	1.100 (0.890-1.350)	none found
Alzheimer's disease	Qian HR, 2006	A	Not Reported	Not Reported	Not Reported	V* vs MM	0.800 (0.650-0.980)	none found
Alzheimer's disease	Qian HR, 2006	A	Not Reported	Not Reported	Not Reported	M* vs VV	1.380 (1.010-1.890)	none found

A: Meta analysis of group level data (and/or HuGE review)
B: Consortium-based analysis with individual level data
Effect Size: OR unless noted otherwise
Het: Heterogeneity

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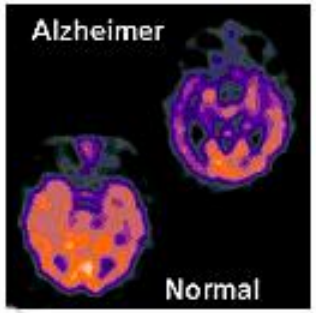
Genes and Disease

NCBI

Alzheimer disease

Alzheimer disease (AD) is the fourth leading cause of death in adults. The incidence of the disease rises steeply with age. AD is twice



Brain scans of a healthy elderly person and a patient with Alzheimer's disease. [Image supplied Keith Johnson, Bringham

Trend/Pattern

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NCBI

dbGaP GENOTYPE and PHENOTYPE

Linking Genome-Wide Association Study of Schizophrenia

Accession: phs000021.v1.p1

Description

The goal of the study is to find susceptibility genes for schizophrenia.

Note: The data for this study have not been completely submitted and processed into the dbGaP database yet. Public summaries of phenotype variables as well as encoded study documents are coming soon. Thank you for your patience.

GAIN The Genetic Association Information Network

- Participants: 5188
- Type: Case-control

Individual-Level Data

- Request to Download Individual-Level Data from dbGaP Authorized Access
- Data Use Certification Requirements (DUC)
- Release Date for Individual-Level Data: December 25, 2007
- Embargo Release Date: August 25, 2008

- **Use Restrictions**

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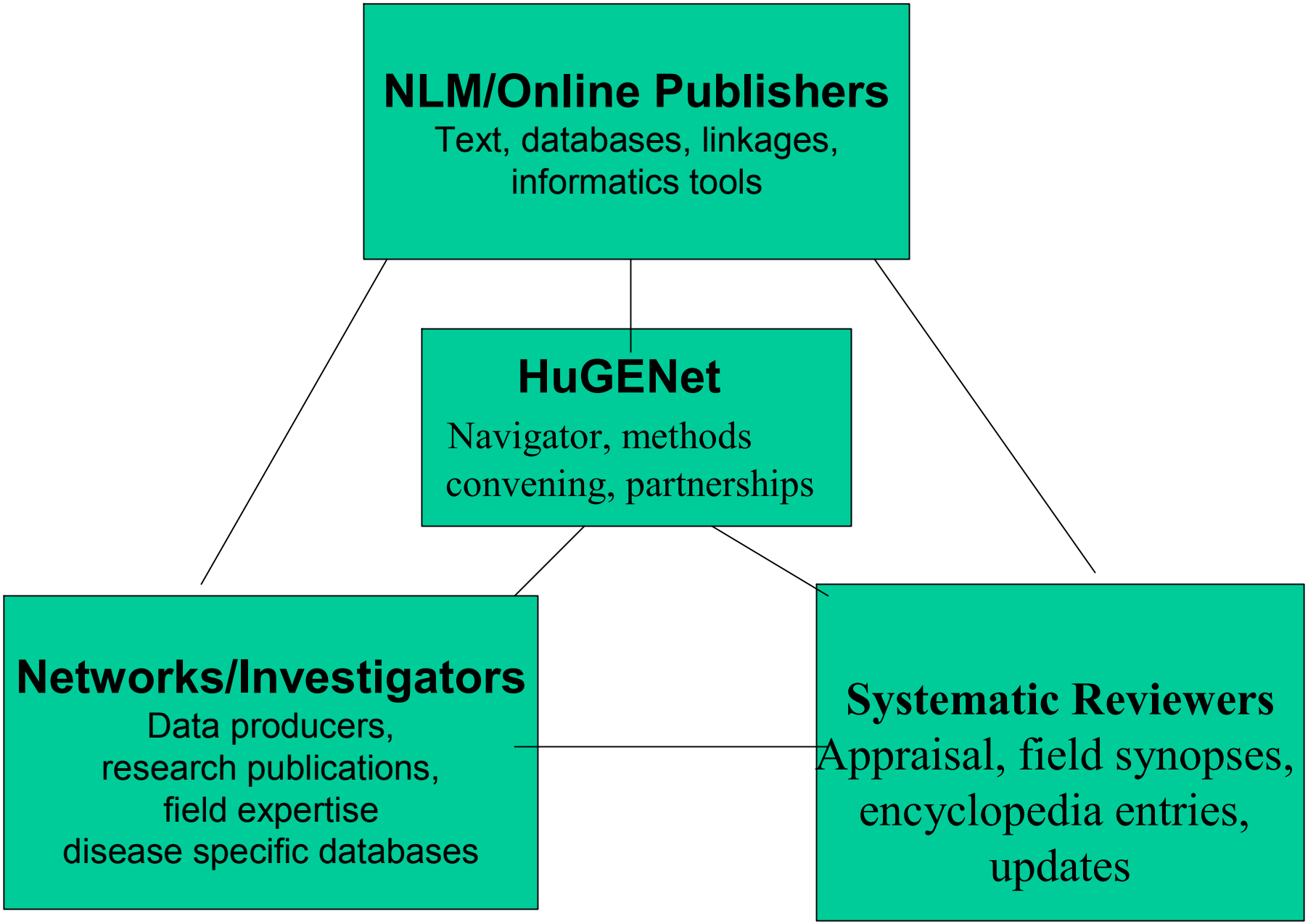
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