House of Lords Inquiry on Genomic Medicine Visit
Lawton Chiles International House
National Institutes of Health
Bethesda, MD
June 5, 2008

Bioinformatics Infrastructure:

Education, Public Databases, Standards

Betsy L. Humphreys, MLS
National Library of Medicine, National Institutes of Health
U.S. Department of Health and Human Services
http://www.nlm.nih.gov

National Library of Medicine (NLM)

- Library Operations and Services 1836-
- Grant Programs 1965-
- Toxicology/Environmental Health Program 1966-
- Lister Hill National Center for Biomedical Communications 1968-
- National Center for Biotechnology Information (NCBI) 1988-
- National Information Center on Health Services Research & Health Care Technology 1993-

NLM Resources

- Current Budget:
  - $329 million
    - $262 million – Intramural
      - Includes: PubMed/MEDLINE, PubMed Central, GenBank, PubChem, dbGaP, consumer health information services, environmental/toxicology databases, NLM collection, computer centers, intramural research, health IT standards development, etc.
    - $67 million – Extramural
      - Includes support for: Informatics research, research training ($15M), National Network of Libraries of Medicine($12M)
- Staff: 1,330 employees & contractors

NLM Long Range Plan Goals for 2006-16

- **Seamless, Uninterrupted Access** to expanding collections of biomedical data, medical knowledge, and health information
- **Trusted Information Services** that promote health literacy, improve health outcomes, and reduce health disparities wordwide
- **Integrated Biomedical, Clinical, and Public Health Information Systems** that promote scientific discovery and speed transition of research into practice
- **A Strong and Diverse Workforce** for biomedical informatics research, systems development, and innovative service delivery

[some of] NLM's UK Connections

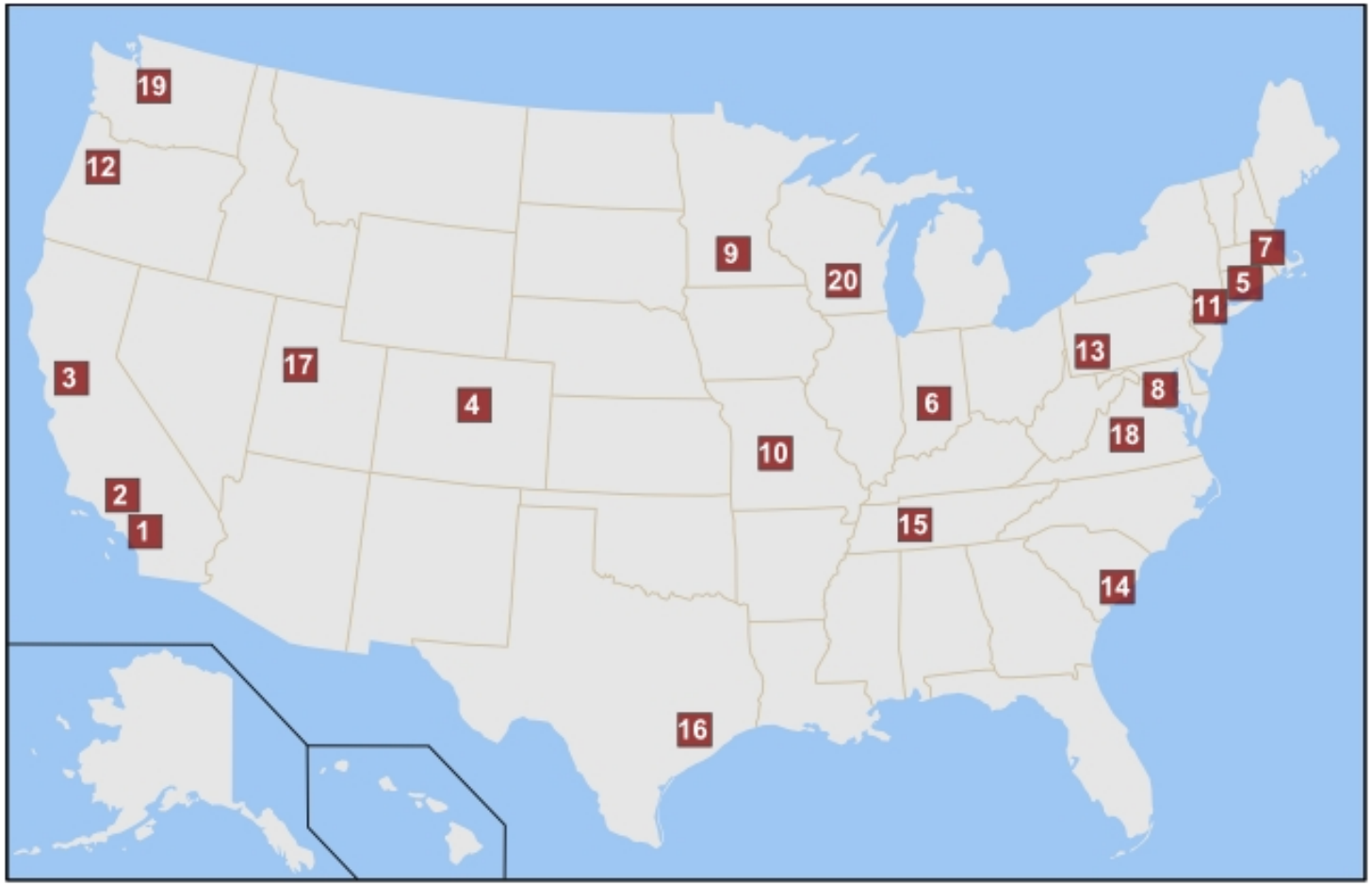
- **Research** – NCBI intramural scientists
  - Cambridge, University College, Royal Veterinary College, Universities of: Bangor, Liverpool, Portsmouth, York
- **Scientific data** – GenBank, PubChem …
  - European Bioinformatics Institute, EMBL
  - Structural Genomics Consortium, Oxford
- **Clinical trials registration** – ClinicalTrials.gov
  - International Standard Randomised Controlled Trial Register
- **Standard terminologies** – UMLS, SNOMED CT
  - NHS Connecting for Health
- **Biomedical literature** – MEDLINE/PubMed/Pubmed Central
  - British Library (1965-)
  - Wellcome Trust, JISC, Other UK research funders – UKPMC
- **Health information –** MedlinePlus, Genetic Home Reference, Bookshelf…
  - NHS Direct
  - National Library of Health

NLM Bioinformatics Activities

- Intramural Research
- Extramural Support
  - Research, Infrastructure
  - Education/training
- Direct Infrastructure Development/Access
  - Linked databases, Standards, Software Tools, Inter-organizational Networks

NLM's University-based Biomedical Informatics Research Training Programs

Listed in alphabetical order by state



Areas of Emphasis for NLM's University-based Biomedical Informatics Research Training Programs					
Training Program	Health care informatics	Bioinformatics or computational biology	Clinical research translational informatics	Public health informatics	Short-term training
1. University of California Irvine Irvine, CA Program contact: Pierre Baldi		✔	✔		
2. University of California Los Angeles Los Angeles, CA Program contact: Alex Bui			✔		✔
3. Stanford University Stanford, CA Program contact: Russ Altman	✔	✔	✔		
4. University of Colorado Denver/HSC Aurora Aurora, CO Program contact: Larry Hunter		✔			✔
5. Yale University New Haven, CT Program contact: Sandra Frawley	✔	✔	✔	✔	✔
6. Regenstrief/Indiana University Indianapolis, IN	✔		✔	✔	

Focus Issue

FOCUS ISSUE INTRODUCTION: Building the role of medical libraries in bioinformatics
Renata C. Geer and Diane C. Rein
J Med Libr Assoc. 2006 July; 94(3): **284–285.**
| Full Text | PDF–43K |

Broad issues to consider for library involvement in bioinformatics
Renata C. Geer
J Med Libr Assoc. 2006 July; 94(3): **286-298, E152-E155.**
| Abstract | Full Text | PDF–445K |

A Web-based assessment of bioinformatics end-user support services at US universities
Donna J. Messersmith, Dennis A. Benson, and Renata C. Geer
J Med Libr Assoc. 2006 July; 94(3): **299-305, E156-E187.**
| Abstract | Full Text | PDF–866K |

Vignettes: diverse library staff offering diverse bioinformatics services
David L. Osterbur, Kristine Alpi, Catharine Canevari, Pamela M. Corley, Medha Devare, Nicola Gaedeke, Donna K. Jacobs, Peter Kirlew, Janet A. Ohles, K.T.L. Vaughan, Lili Wang, Yongchun Wu, and Renata C. Geer
J Med Libr Assoc. 2006 July; 94(3): **306, E188-E191.**
| Abstract | Full Text | PDF–76K |

Design and implementation of a library-based information service in molecular biology and genetics at the University of Pittsburgh
Ansuman Chattopadhyay, Nancy Hrinya Tannery, Deborah A. L. Silverman, Phillip Bergen, and Barbara A. Epstein

J Med Libr Assoc

NLM Individual Fellowship for Informationist Training (F37) (PAR 06-509)
http://grants.nih.gov/grants/guide/pa-files/PAR-06-509.html

Correction

- Correction to Stipend and Allowances, PAR-06-509, NLM Individual Fellowship for Informationist Training (F37) (11/30/07)

Purpose

The National Library of Medicine (NLM) believes that clinical care, biomedical research and education, and public health administration can be improved by the inclusion of in-context information specialists (informationists) into work and decision settings. Informationists are information specialists who have received graduate training and practical experience that provides them with disciplinary background both in medical or biological sciences and in information sciences/informatics. This program expires December 9, 2009 unless reissued.

NLM Contact

Dr. Valerie Florance, florancev@mail.nih.gov

Deadlines

Deadlines for New or Revised Applications: April 8, August 8, December 8 each year
Full listing of deadlines for competing applications: http://www.nlm.nih.gov/ep/Deadlines.html

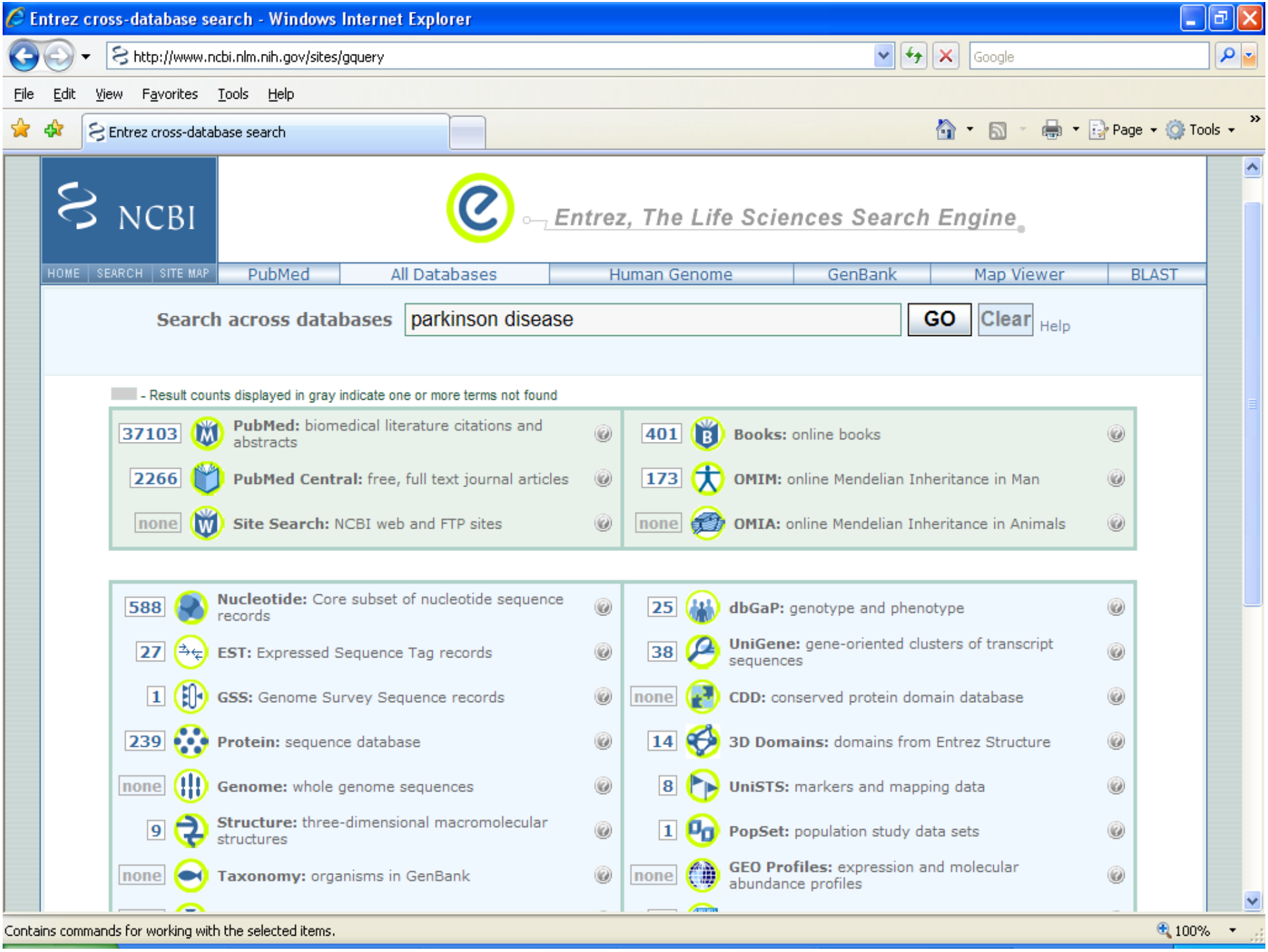
See http://era.nih.gov/ElectronicReceipt/ for details and timetable for electronic submission of F37 grants applications.

Helper Links for Fellowships
Recent NLM Awards
NIH Research Training Opportunities

Grant Program Overviews
Research Support
Resource Support
Career Support
Training Support
Small Business R & D Support

Public Databases and Standards

- A huge international success story
  - Significant impact of joint action by research funders, journal editors
- Amount of data, use/utility, maintenance costs rising together
  - But economies of scale
    - International Nucleotide Sequence Database Collaboration – an excellent model
- Direct deposit/use fees highly counterproductive
  - For both Databases *and* Standards
- Integration of basic, clinical research, healthcare, consumer data adds value
  - Full employment for many R & D groups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NCBI
Entrez, The Life Sciences Search Engine
HOME SEARCH SITE MAP
PubMed
All Databases
Human Genome
GenBank
Map Viewer
BLAST
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GO
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- Result counts displayed in gray indicate one or more terms not found
37103 PubMed: biomedical literature citations and abstracts
2266 PubMed Central: free, full text journal articles
none Site Search: NCBI web and FTP sites
401 Books: online books
173 OMIM: online Mendelian Inheritance in Man
none OMIA: online Mendelian Inheritance in Animals
588 Nucleotide: Core subset of nucleotide sequence records
27 EST: Expressed Sequence Tag records
1 GSS: Genome Survey Sequence records
239 Protein: sequence database
none Genome: whole genome sequences
9 Structure: three-dimensional macromolecular structures
none Taxonomy: organisms in GenBank
25 dbGaP: genotype and phenotype
38 UniGene: gene-oriented clusters of transcript sequences
none CDD: conserved protein domain database
14 3D Domains: domains from Entrez Structure
8 UniSTS: markers and mapping data
1 PopSet: population study data sets
none GEO Profiles: expression and molecular abundance profiles
Contains commands for working with the selected items.
100%

Parkinson's Disease

Also called: Paralysis agitans, Shaking palsy

Parkinson's disease is a disorder that affects nerve cells, or neurons, in a part of the brain that controls muscle movement. In Parkinson's, neurons that make a chemical called dopamine die or do not work properly. Dopamine normally sends signals that help coordinate your movements. No one knows what damages these cells. Symptoms of Parkinson's disease may include

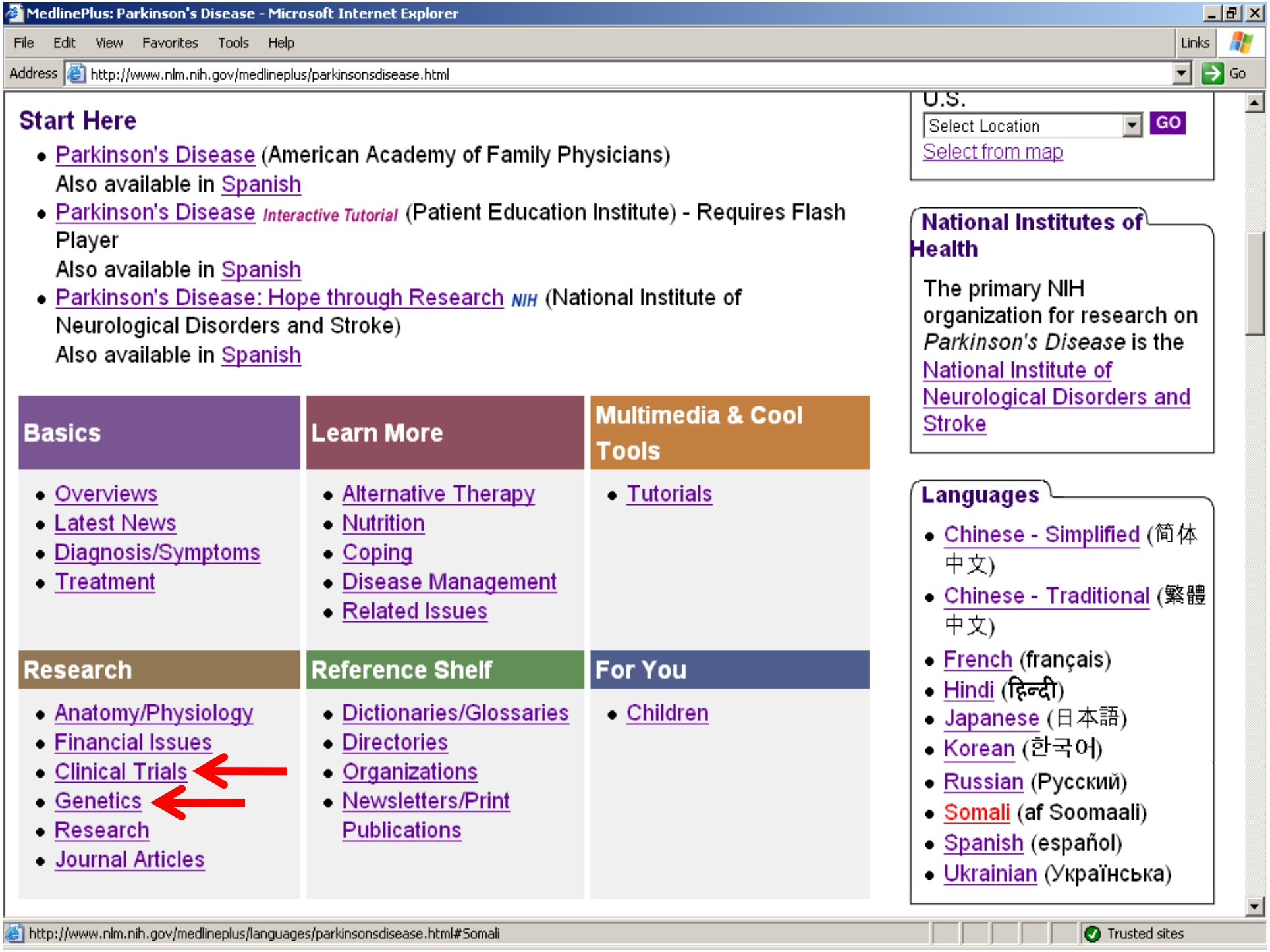
- Trembling of hands, arms, legs, jaw and face
- Stiffness of the arms, legs and trunk
- Slowness of movement
- Poor balance and coordination

As symptoms get worse, people with the disease may have trouble walking, talking or doing simple tasks. They may also have problems such as depression, sleep problems or trouble chewing, swallowing or speaking.

*ADAM.

Related Topics

- Movement Disorders
- Brain and Nerves
- Seniors


MedlinePlus: Parkinson's Disease - Microsoft Internet Explorer
File Edit View Favorites Tools Help
Links
Address http://www.nlm.nih.gov/medlineplus/parkinsonsdisease.html
Go
Start Here
Parkinson's Disease (American Academy of Family Physicians)
Also available in Spanish
Parkinson's Disease Interactive Tutorial (Patient Education Institute) - Requires Flash Player
Also available in Spanish
Parkinson's Disease: Hope through Research NIH (National Institute of Neurological Disorders and Stroke)
Also available in Spanish
Basics
Overviews
Latest News
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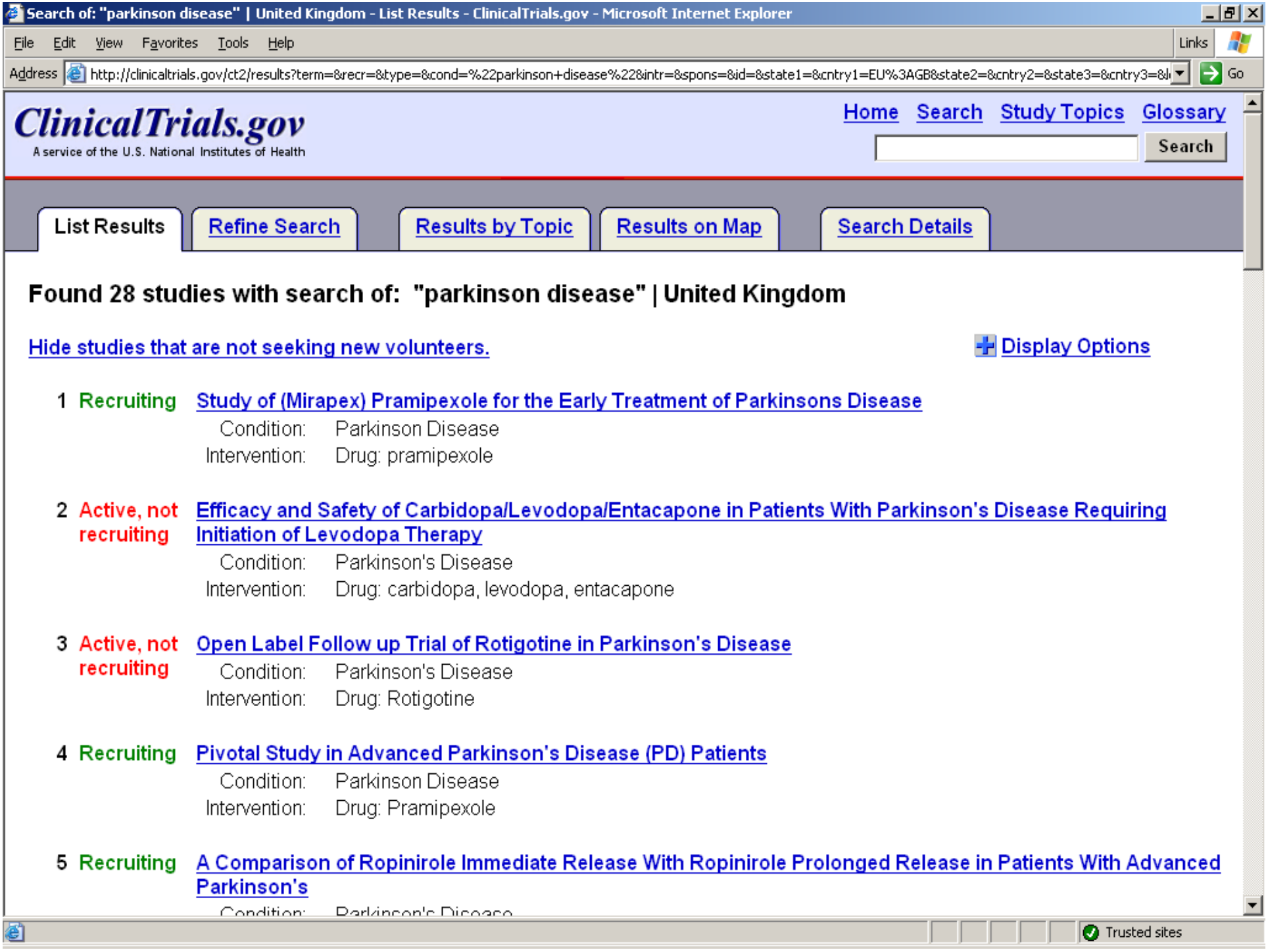
National Institutes of Health
The primary NIH organization for research on Parkinson's Disease is the National Institute of Neurological Disorders and Stroke
Languages
Chinese - Simplified (简体中文)
Chinese - Traditional (繁體中文)
French (français)
Hindi (हिन्दी)
Japanese (日本語)
Korean (한국어)
Russian (Русский)
Somali (af Soomaali)
Spanish (español)
Ukrainian (Українська)
http://www.nlm.nih.gov/medlineplus/languages/parkinsonsdisease.html#Somali
Trusted sites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Refine Search
Results by Topic
Results on Map
Search Details
Found 28 studies with search of: "parkinson disease" | United Kingdom
Hide studies that are not seeking new volunteers.
Display Options
1 Recruiting Study of (Mirapex) Pramipexole for the Early Treatment of Parkinsons Disease
Condition: Parkinson Disease
Intervention: Drug: pramipexole
2 Active, not recruiting Efficacy and Safety of Carbidopa/Levodopa/Entacapone in Patients With Parkinson's Disease Requiring Initiation of Levodopa Therapy
Condition: Parkinson's Disease
Intervention: Drug: carbidopa, levodopa, entacapone
3 Active, not recruiting Open Label Follow up Trial of Rotigotine in Parkinson's Disease
Condition: Parkinson's Disease
Intervention: Drug: Rotigotine
4 Recruiting Pivotal Study in Advanced Parkinson's Disease (PD) Patients
Condition: Parkinson Disease
Intervention: Drug: Pramipexole
5 Recruiting A Comparison of Ropinirole Immediate Release With Ropinirole Prolonged Release in Patients With Advanced Parkinson's
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Gene Tests
DNA test labs
ClinicalTrials.gov
Research studies
PubMed
Recent literature
OMIM
Genetic disorder catalog

Parkinson disease

On this page: Description Genetic changes Inheritance Treatment Additional information Other names Glossary definitions

Reviewed July 2007

What is Parkinson disease?

Parkinson disease is a progressive disorder of the nervous system. The disorder affects several regions of the brain, including an area called the substantia nigra that controls balance and movement. Parkinson disease may also affect regions of the brain that regulate involuntary functions such as blood pressure and heart activity.

Often the first symptom of Parkinson disease is trembling or shaking (tremor) of a limb, especially when the body is at rest. Typically, the tremor begins on one side of the body, usually in one hand. Tremors can also affect the arms, legs, feet, and face. Other characteristic symptoms include rigidity or stiffness of the limbs and trunk, slow movement (bradykinesia) or the inability to move (akinesia), and impaired balance and coordination (postural instability).

Many Parkinson disease symptoms occur when nerve cells (neurons) in the substantia nigra die or become impaired. Normally, these cells produce a chemical messenger called dopamine, which transmits signals within the brain to produce smooth physical movements. When these dopamine-producing neurons die or become impaired, communication between the brain and muscles weakens, and eventually, the brain is unable to control muscle movement. In most cases of Parkinson disease, protein deposits called Lewy bodies appear in dead or dying dopamine-producing neurons. (Cases without Lewy bodies are sometimes referred to as parkinsonism instead of Parkinson disease.) It is unclear whether Lewy bodies play a role in killing nerve cells, or if they are part of a protective process.

Generally, Parkinson disease that begins after age 50 years is called late onset disease. It is known as early onset disease if signs and symptoms begin before age 50. Cases that begin before age 20 are sometimes referred to as juvenile onset Parkinson disease.

LRRK2-Related Parkinson Disease

[*PARK 8, LRRK2-Associated Parkinson Disease*]

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Owen A Ross, PhD

Department of Neuroscience
Mayo Clinic
Jacksonville, FL

All GeneReviews
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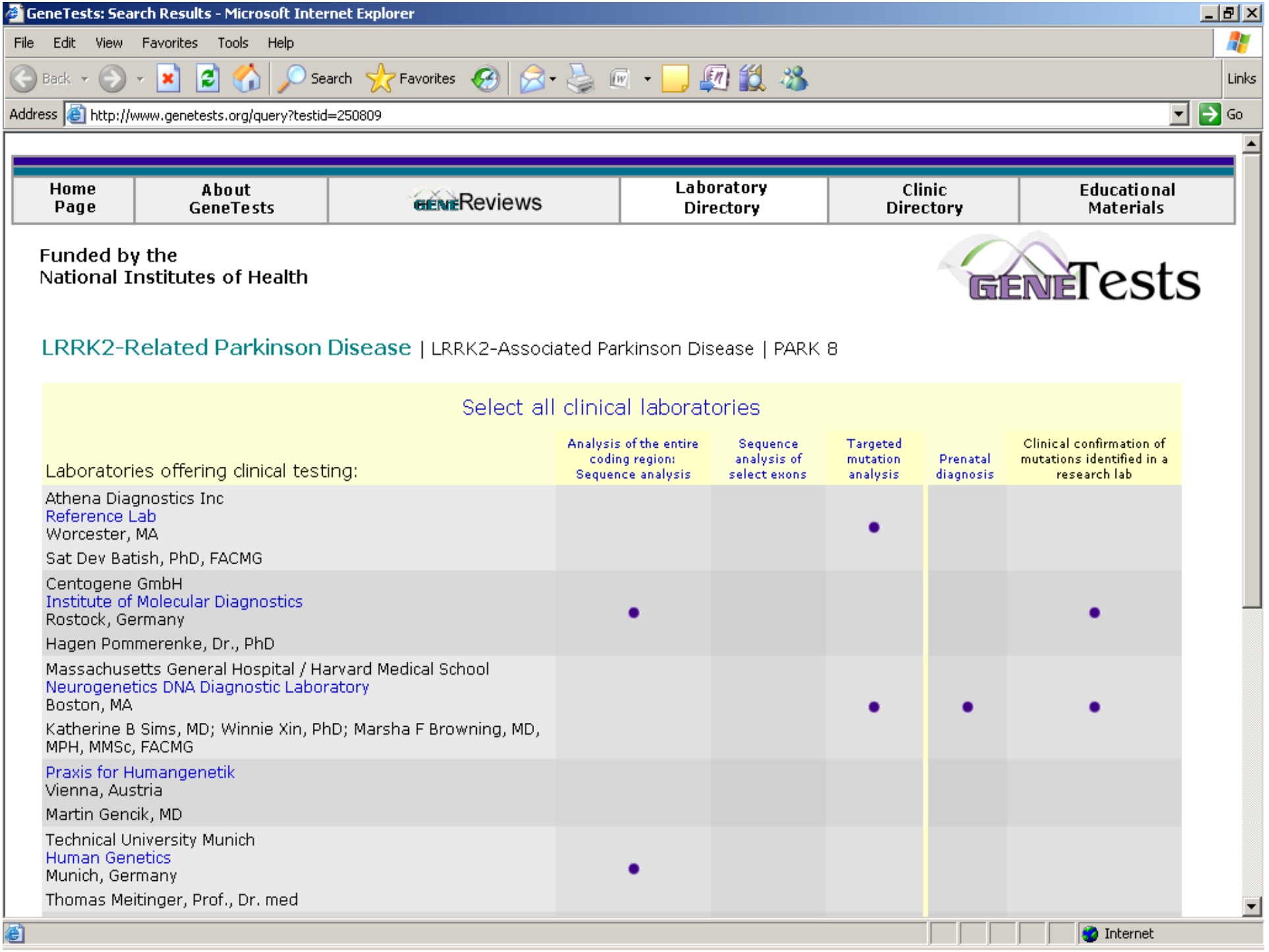
In this page

Summary
Diagnosis
Clinical Description
Differential Diagnosis
Management
Genetic Counseling
Molecular Genetics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Laboratories offering clinical testing:
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Sat Dev Batish, PhD, FACMG
Centogene GmbH
Institute of Molecular Diagnostics
Rostock, Germany
Hagen Pommerenke, Dr., PhD
Massachusetts General Hospital / Harvard Medical School
Neurogenetics DNA Diagnostic Laboratory
Boston, MA
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Human Genetics
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Thomas Meitinger, Prof., Dr. med
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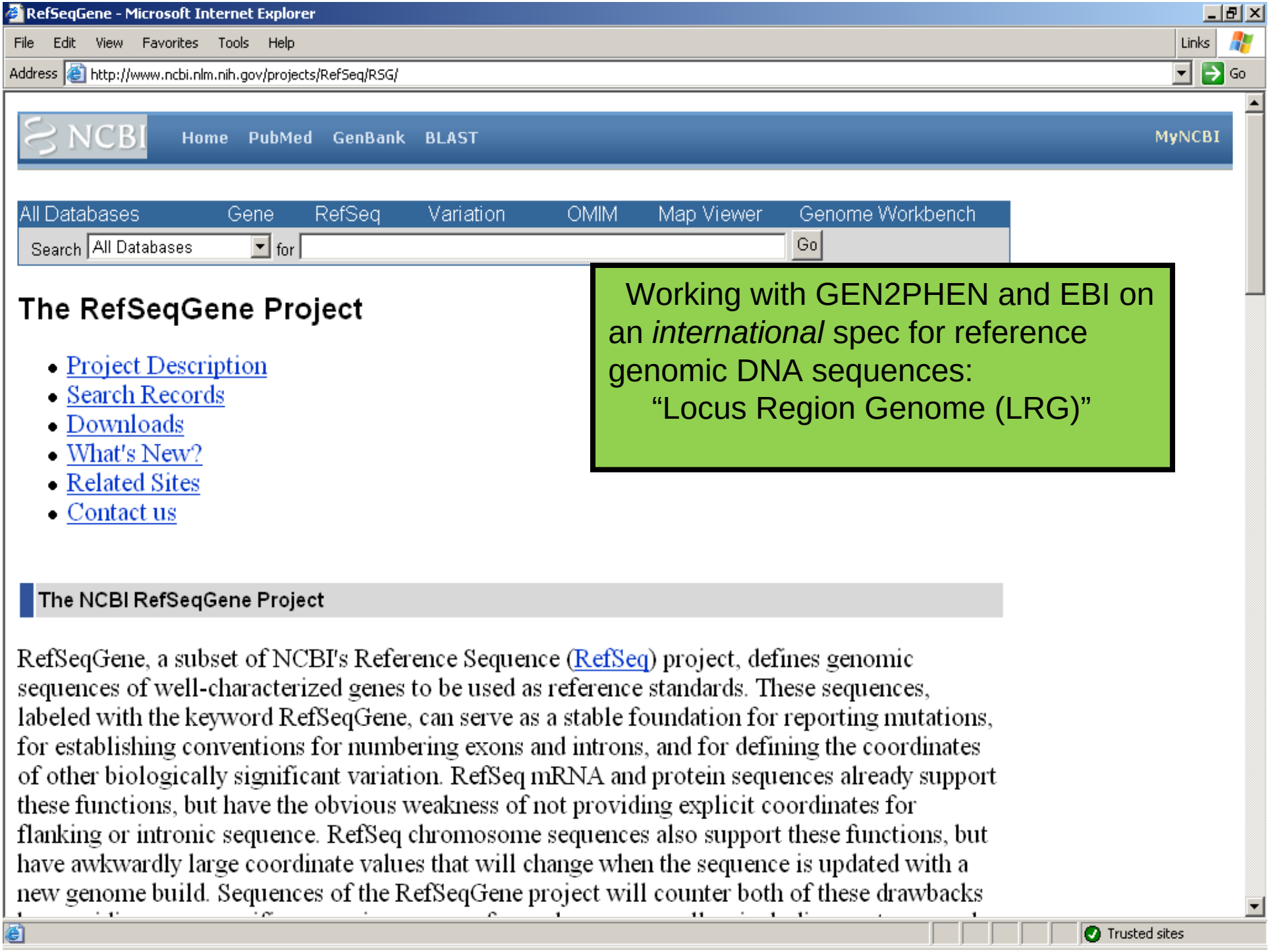

Working with GEN2PHEN and EBI on an *international* spec for reference genomic DNA sequences:

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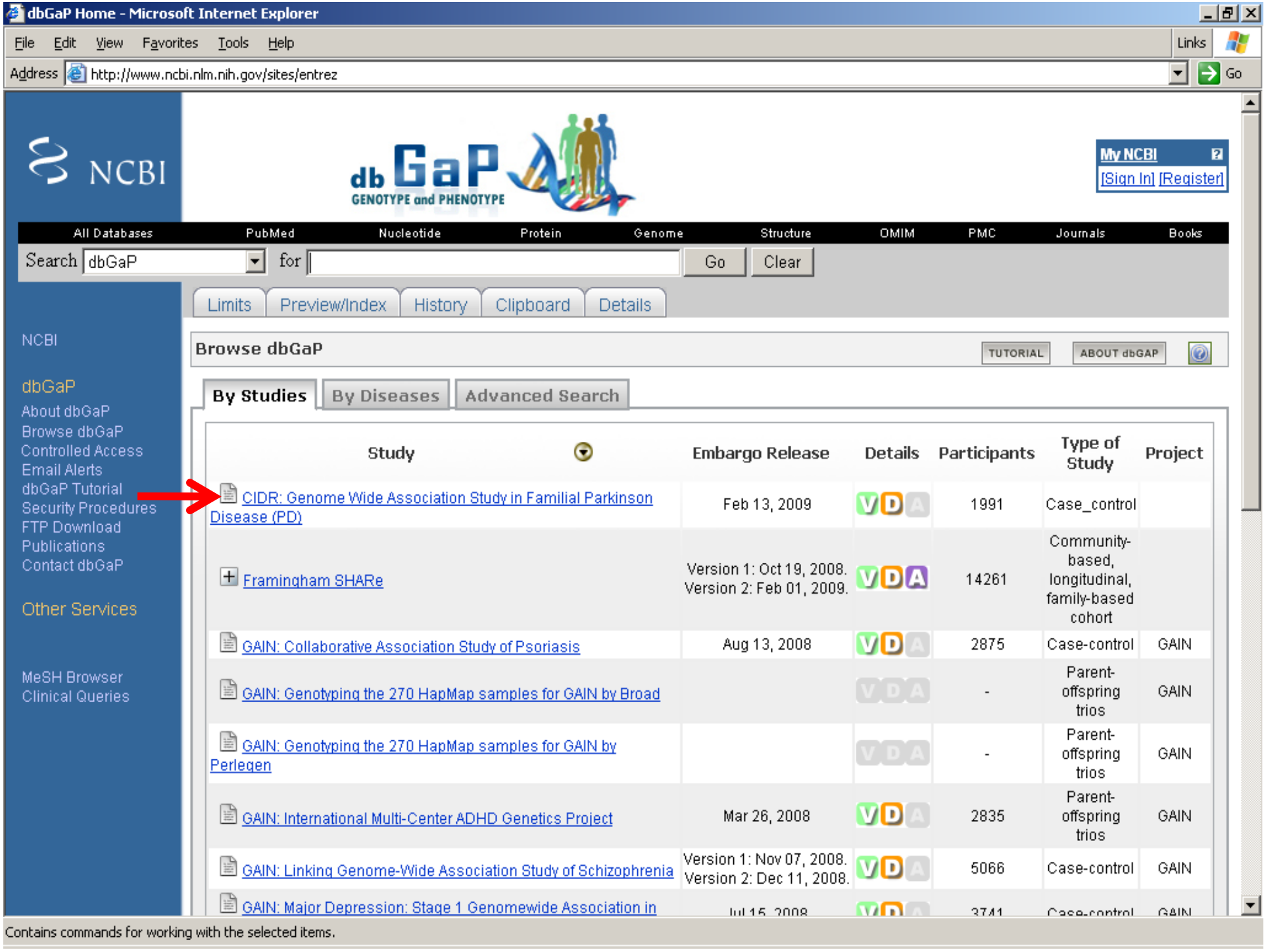
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CIDR: Genome Wide Association Study in Familial Parkinson Disease (PD)
Feb 13, 2009
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Version 2: Feb 01, 2009.
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GAIN: Collaborative Association Study of Psoriasis
Aug 13, 2008
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GAIN: Genotyping the 270 HapMap samples for GAIN by Broad
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GAIN: Genotyping the 270 HapMap samples for GAIN by Perlegen
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GAIN: International Multi-Center ADHD Genetics Project
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2835
Parent-offspring trios
GAIN
GAIN: Linking Genome-Wide Association Study of Schizophrenia
Version 1: Nov 07, 2008.
Version 2: Dec 11, 2008.
5066
Case-control
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GAIN: Major Depression: Stage 1 Genomewide Association in
Contains commands for working with the selected items.

study-logo

CIDR: Genome Wide Association Study in Familial Parkinson Disease (PD)

Study Accession: phs000126.v1.p1

Study | Variables | Documents | Analyses

Document Name and Accession

Name: IUPUI and Clarian Informed Consent Statement for Parkinson's Research: The Organized Genetics Initative (PROGENI)
Accession: phd000920.1

Document

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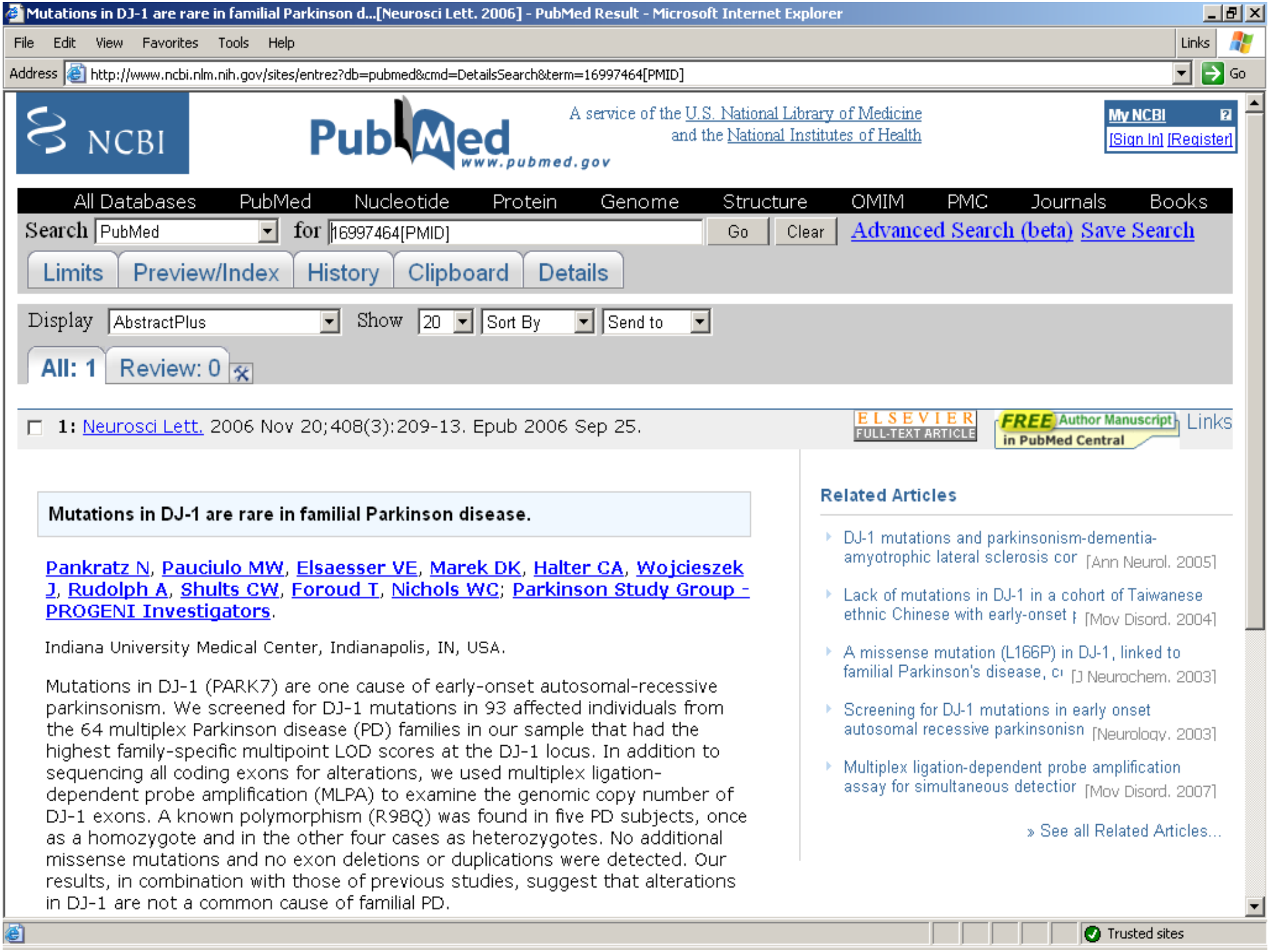
IUPUI and Clarian Informed Consent Statement for Parkinson's Research: The Organized Genetics Initative (PROGENI)

Associated Documents

- CIDR: Genome Wide Association Study in Familial Parkinson Disease (PD)
  - Parkinson Disease
    - Protocols and Instruction

- IUPUI and Clarian Informed Consent Statement for P arkinson's Research: The O rganized Gen etics I nitative (PROGENI)
- Protocol for Parkinson Disease Collaborative Study of Genetic Linkage A.K.A. P arkinson's R esearch: The O rganized Gen etics I nitiative [PROGENI]
- Institutional Review Board


Mutations in DJ-1 are rare in familial Parkinson d...[Neurosci Lett. 2006] - PubMed Result - Microsoft Internet Explorer
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1: Neurosci Lett. 2006 Nov 20;408(3):209-13. Epub 2006 Sep 25.
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Mutations in DJ-1 are rare in familial Parkinson disease.
Pankratz N, Pauciulo MW, Elsaesser VE, Marek DK, Halter CA, Wojcieszek J, Rudolph A, Shults CW, Foroud T, Nichols WC; Parkinson Study Group - PROGENI Investigators.
Indiana University Medical Center, Indianapolis, IN, USA.
Mutations in DJ-1 (PARK7) are one cause of early-onset autosomal-recessive parkinsonism. We screened for DJ-1 mutations in 93 affected individuals from the 64 multiplex Parkinson disease (PD) families in our sample that had the highest family-specific multipoint LOD scores at the DJ-1 locus. In addition to sequencing all coding exons for alterations, we used multiplex ligation-dependent probe amplification (MLPA) to examine the genomic copy number of DJ-1 exons. A known polymorphism (R98Q) was found in five PD subjects, once as a homozygote and in the other four cases as heterozygotes. No additional missense mutations and no exon deletions or duplications were detected. Our results, in combination with those of previous studies, suggest that alterations in DJ-1 are not a common cause of familial PD.
Related Articles
DJ-1 mutations and parkinsonism-dementia-amyotrophic lateral sclerosis cor [Ann Neurol. 2005]
Lack of mutations in DJ-1 in a cohort of Taiwanese ethnic Chinese with early-onset I [Mov Disord. 2004]
A missense mutation (L166P) in DJ-1, linked to familial Parkinson's disease, c [J Neurochem. 2003]
Screening for DJ-1 mutations in early onset autosomal recessive parkinsonism [Neurology. 2003]
Multiplex ligation-dependent probe amplification assay for simultaneous detection [Mov Disord. 2007]
» See all Related Articles...
Trusted sites

Neurosci Lett. Author manuscript; available in PMC 2006 December 19.
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PMCID: PMC1706076
NIHMSID: NIHMS13438

Mutations in *DJ-1* are rare in familial Parkinson disease

Nathan Pankratz, Ph.D.,[1] Michael W. Pauciulo, M.B.A.,[2] Veronika E. Elsaesser, B.S.,[2] Diane K. Marek, B.S.,[2] Cheryl A. Halter, M.S.,[1] Joanne Wojcieszek, M.D.,[1] Alice Rudolph, Ph.D.,[3] Clifford W. Shults, M.D.,[4] Tatiana Foroud, Ph.D.,[1] William C. Nichols Ph.D.,[2,5] and the Parkinson Study Group – PROGENI Investigators

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[2] Cincinnati Children's Hospital Medical Center, Cincinnati, OH;

[3] University of Rochester, Rochester, NY;

[4] University of California, La Jolla, CA; VA San Diego Healthcare System, San Diego, CA;

[5] University of Cincinnati College of Medicine, Cincinnati, OH

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