

# Genes in Diseases

*Computational Biology camp for high school students*  
*January 29, 2012*



**MIRcore**

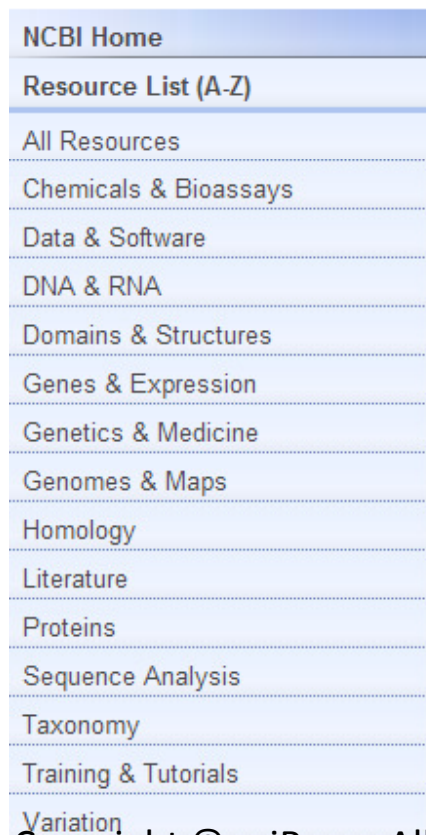
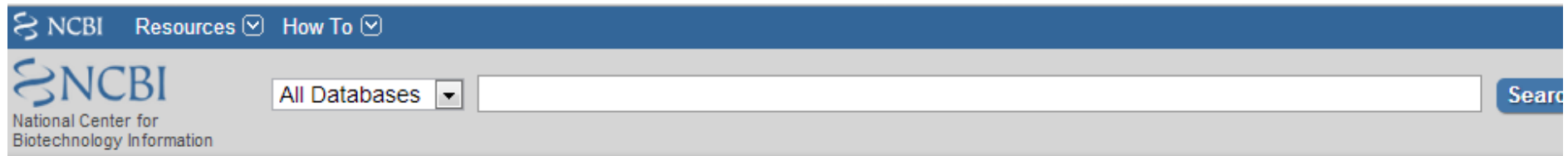
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# Choose a disease

# NCBI

(The National Center for Biotechnology Information)

<http://www.ncbi.nlm.nih.gov/>



## Welcome to NCBI

The National Center for Biotechnology Information advances science and health by providing access to biomedical and genomic information.

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## Get Started

- [Tools](#): Analyze data using NCBI software
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- [Submissions](#): Submit data to GenBank or other NCBI databases

## Genetic Testing Registry

A portal to clinical genetics resources with detailed information about genetic tests and laboratories.

GO

## Popular Resources

[PubMed](#)

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[SNP](#)

[Gene](#)

[Protein](#)

[PubChem](#)

## NCBI Announcements

Come to the NCBI Disco on February 4&5!

Spaces are still available  
**MIRCOE**

# Pubmed Health

<http://www.ncbi.nlm.nih.gov/pubmedhealth/>



# Search your disease

A.D.A.M. Medical Encyclopedia.

Click one under the Medical Encyclopedia

[illegible]



# Alzheimer's disease

<http://www.ncbi.nlm.nih.gov/pubmedhealth/PMH0001767/>

Review Date: 9/26/2011.

A.D.A.M. Medical Encyclopedia: For professional overview

Home > Diseases and Conditions > Alzheimer's disease

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## ★ Recommended reading: [Alzheimer's disease: Do medications containing memantine help?](#)

Medications containing the drug memantine are supposed to help people who have Alzheimer's disease remember things and better manage their daily tasks. Trials show that memantine can somewhat delay the deterioration of mental abilities. Other abilities important to daily life may also last longer.

A.D.A.M. Medical Encyclopedia.

## Alzheimer's disease

Senile dementia - Alzheimer's type (SDAT); SDAT

Last reviewed: September 26, 2011.

[Dementia](#) is a loss of brain function that occurs with certain diseases. Alzheimer's disease (AD), is one form of dementia that gradually gets worse over time. It affects memory, thinking, and behavior.

## Causes, incidence, and risk factors

You are more likely to get Alzheimer's disease (AD) if you:

- Are older. However, developing AD is not a part of normal aging.
- Have a close blood relative, such as a brother, sister, or parent with AD.
- Have certain genes linked to AD, such as APOE epsilon4 allele

The following may also increase your risk, although this is not well proven:

- Being female

## What works?



Dementia in Alzheimer's disease: Can Ginkgo help?

Ginkgo biloba could help some people with Alzheimer's disease to perform daily tasks again. Adverse effects are not very common, but interactions with other medications cannot be ruled out.

[See all \(63\) ..](#)

## Figures



## Learn about...



The science behind screening tests

[Screening tests can lead to](#)

# CDC (Centers for Disease Control and Prevention)

<http://www.cdc.gov/diseasesconditions/>

CDC Home



Centers for Disease Control and Prevention

CDC 24/7: Saving Lives. Protecting People.™

autism

SEARCH

A-Z Index for All CDC Topics

## Diseases & Conditions

### Chickenpox Vaccine

Chickenpox Can Be Serious: Protect Your Child

Learn More »



### A-Z Index for Diseases and Conditions

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### Topics

- [ADHD](#)
- [Arthritis](#)
- [Asthma](#)
- [Autism](#)
- [Avian Influenza](#)
- [Heart Disease](#)
- [Hepatitis](#)
- [HIV/AIDS](#)
- [HPV \(Human papillomavirus\)](#)
- [Measles](#)
- [Mononucleosis](#)
- [Norovirus](#)
- [Pertussis](#)
- [Rabies](#)
- [Scarlet Fever](#)
- [Shingles](#)
- [Strep Throat](#)
- [Tuberculosis](#)
- [Whooping Cough](#)
- [Zika Virus](#)

### Campaigns & Programs

[Colorectal Cancer Control Program \(CRCCP\)](#)

[Inside Knowledge: Get the Facts About Gynecologic Cancer campaign](#)

[Learn the Signs. Act Early.](#)

[Million Hearts](#)

[National Breast and Cervical Cancer Early Detection Program](#)

[One Test. Two Lives](#)

[Pre-teen Vaccine Campaign](#)

[Screen for Life: National Colorectal Cancer Action Campaign](#)

[Tobacco Control Program](#)

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Protecting People.™

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### News & Events

September is  
National Prostate  
Cancer Awareness  
Month

[Press Room »](#)

<http://www.cdc.gov/ncbddd/autism/index.html>

CDC Home

**CDC** Centers for Disease Control and Prevention  
CDC 24/7: Saving Lives. Protecting People.™

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## Autism Spectrum Disorders (ASDs)

[National Center Homepage](#)


Autism spectrum disorders (ASDs) are a group of developmental disabilities that can cause significant social, communication and behavioral challenges. CDC is committed to continuing to provide essential data on ASDs, search for risk factors and causes, and develop resources that help identify children with ASDs as early as possible.

### Mary Elizabeth's Story

One mom's journey with two children with autism.

[Learn More»](#)

< 1 2 3 4 5 >





## Autism Spectrum Disorder Topics



### Basics

Facts, signs, causes, and what to do if you're concerned.



### Screening and Diagnosis

Information for families and health professionals.



### Treatments

Types of treatment services and early intervention.



### Data & Statistics

Data and statistics highlights.



### Research and Tracking

What we've learned.



### Articles

Scientific articles.



### Free Materials

Print or order free materials.



### About Us

Overview of CDC's work.

# Limited Scientific Information for Most Genetic Tests

- Despite the many scientific advances in genetics, researchers have only identified a small fraction of the genetic component of most diseases. Therefore, genetic tests for many diseases are developed on the basis of limited scientific information and may not yet provide valid or useful results to individuals who are tested. However, many genetic tests are being marketed prematurely to the public through the Internet, TV, and other media. This may lead to the misuse of these tests and the potential for physical or psychological harms to the public. At the same time, valid and useful tests, such as those for hereditary breast and ovarian cancer or for Lynch syndrome, a form of hereditary colorectal cancer, are not widely used, in part because of limited research on how to get useful tests implemented into practice across U.S. communities. Individuals can learn more about specific genetic tests by visiting the Web sites listed below or by talking with their doctor.

<http://www.cdc.gov/genomics/gtesting/index.htm>

# OMIM<sup>®</sup> - Online Mendelian Inheritance in Man<sup>®</sup> <http://omim.org/>

**OMIM<sup>®</sup>**

Online Mendelian Inheritance in Man<sup>®</sup>

An Online Catalog of Human Genes and Genetic Disorders

Updated 25 January 2013

[Search](#)

[Sample Searches](#)

[Advanced Search: OMIM, Clinical Synopses, OMIM Gene Map , Search History](#)



# Caution

- NOTE: OMIM is intended for use primarily by physicians and other professionals concerned with genetic disorders, by genetics researchers, and by advanced students in science and medicine. While the OMIM database is open to the public, users seeking information about a personal medical or genetic condition are urged to consult with a qualified physician for diagnosis and for answers to personal questions. (from OMIM site)
- Genes associated with diseases are mainly in DNA level.



# Search your disease

- 1 : # 104300. ALZHEIMER DISEASE; AD  
ALZHEIMER DISEASE, FAMILIAL, 1, INCLUDED  
Cytogenetic locations: 4p14-p13 , 6p22.2 , 7q36 , 7q36.1 , 7q36.2 , 10q22.2 , 10q24 , 11q24.1 , 12p13.31 , 12p11.23-q13.12 , 17q11.2 , 17q22 , 17q23.3 , 19p13.2 , 20p , 21q21.3  
Matching terms: alzheimer, disease
- 2 : \* 104760. AMYLOID BETA A4 PRECURSOR PROTEIN; APP  
Cytogenetic location: 21q21.3 , Genomic coordinates (GRCh37): 21:27,252,860 - 27,543,445  
Matching terms: alzheimer, disease
- 3 : + 107741. APOLIPOPROTEIN E; APOE  
APOLIPOPROTEIN E, DEFICIENCY OR DEFECT OF, INCLUDED  
Cytogenetic location: 19q13.32 , Genomic coordinates (GRCh37): 19:45,409,038 - 45,412,649  
Matching terms: alzheimer, disease
- 4 : \* 104311. PRESENILIN 1; PSEN1  
Cytogenetic location: 14q24.2 , Genomic coordinates (GRCh37): 14:73,603,142 - 73,690,398  
Matching terms: alzheimer, disease
- 5 : # 607822. ALZHEIMER DISEASE 3  
ALZHEIMER DISEASE, FAMILIAL, 3, WITH SPASTIC PARAPARESIS AND UNUSUAL PLAQUES, INCLUDED  
Cytogenetic locations: 14q24.2  
Matching terms: alzheimer, disease
- 6 : \* 157140. MICROTUBULE-ASSOCIATED PROTEIN TAU; MAPT  
Cytogenetic location: 17q21.31 , Genomic coordinates (GRCh37): 17:43,971,747 - 44,105,699  
Matching terms: alzheimer, disease
- #: disease centric information  
%: disease where the cause is unknown  
+: gene with known sequence and phenotype  
\*: gene with known sequence

# Google “autism gene”

## Recent studies

### [Genetic test predicts risk for autism spectrum disorder](http://www.sciencedaily.com/releases/2012/09/120912093827.htm)

[www.sciencedaily.com/releases/2012/09/120912093827.htm](http://www.sciencedaily.com/releases/2012/09/120912093827.htm)

Sep 11, 2012 – A team of Australian researchers has developed a **genetic** test that is able to predict the risk of developing **autism** spectrum disorder (ASD).

### [Scientists Link Gene Mutation to Autism Risk - New York Times](http://www.nytimes.com/.../scientists-link-rare-gene-mutations-to-heightene...)

[www.nytimes.com/.../scientists-link-rare-gene-mutations-to-heightene...](http://www.nytimes.com/.../scientists-link-rare-gene-mutations-to-heightene...)

Apr 4, 2012 – Scientists have for the first time identified several **gene** mutations that they say sharply increase the chances of **autism**, and have found that the ...

### [Genetics: DNA modification marks autism genes —](http://sfari.org/News%20Opinion/In%20Brief/2012)

[sfari.org › News & Opinion › In Brief › 2012](http://sfari.org/News%20Opinion/In%20Brief/2012)

by I Profiles

Dec 12, 2012 – Studies in the past few years have found that DNA methylation tends to concentrate around **autism**-linked **genes** and that it changes in ...

### [New Gene Variants Linked to Autism | TIME.com](http://healthland.time.com/2013/01/15/new-gene-variants-linked-to-autism/)

[healthland.time.com/2013/01/15/new-gene-variants-linked-to-autism/](http://healthland.time.com/2013/01/15/new-gene-variants-linked-to-autism/)

Jan 15, 2013 – In one of the largest-ever studies of genetics and **autism**, researchers have identified 24 new **gene** variants associated with **autism** spectrum ...

### [Researchers Identify 24 More “High Impact” Autism Gene Changes ...](http://www.autismspeaks.org/.../researchers-identify-24-more-high-impact...)

[www.autismspeaks.org/.../researchers-identify-24-more-high-impact...](http://www.autismspeaks.org/.../researchers-identify-24-more-high-impact...)

Jan 14, 2013 – Researchers report their identification of 24 **genetic** changes that individually more than double the risk of **autism**.

### ['Autism gene discovered' by researchers - Health News - NHS Choices](http://www.nhs.uk/news/.../Autism-gene-discovered-by-researchers.aspx)

[www.nhs.uk/news/.../Autism-gene-discovered-by-researchers.aspx](http://www.nhs.uk/news/.../Autism-gene-discovered-by-researchers.aspx)

by NHS Choices - 2012

Nov 8, 2012 – There are more than half a million people with **autism** in the UK. “**Genetic** mutation discovered in people with **autism**,” The Daily Telegraph ...

Need to check  
the scientific source

# Wikipedia: Heritability of autism

[http://en.wikipedia.org/wiki/Heritability\\_of\\_autism](http://en.wikipedia.org/wiki/Heritability_of_autism)

Gene	OMIM/#	Locus	Description
CDH9, CDH10		5p14.1	A 2009 pair of genome-wide association studies found an association between autism and six single-nucleotide polymorphisms in an intergenic region between CDH10 (cadherin 10) and CDH9 (cadherin 9). These genes encode neuronal cell-adhesion molecules, implicating these molecules in the mechanism of autism. <sup>[57]</sup>
MAPK3		16p11.2	A 2008 study observed a de novo deletion of 593 kb on this chromosome in about 1% of persons with autism, and similarly for the reciprocal duplication of the region. <sup>[58]</sup> Another 2008 study also found duplications and deletions associated with ASD at this locus. <sup>[59]</sup> This gene encodes ERK1, one of the extracellular signal regulated kinase subfamily of mitogen-activated protein kinases which are central elements of an intracellular signaling pathways that transmits signals from cell surfaces to interiors. 1% of autistic children have been found to have either a loss or duplication in a region of chromosome 16 that encompasses the gene for ERK1. A similar disturbance in this pathway is also found in neuro-cardio-facial-cutaneous syndromes (NCFC), which are characterized by cranio-facial development disturbances that also can be found in some cases of autism. <sup>[60]</sup>
SERT (SLC6A4)		17q11.2	This gene locus has been associated with rigid-compulsive behaviors. Notably, it has also been associated with depression but only as a result of social adversity, although other studies have found no link. <sup>[61]</sup> Significant linkage in families with only affected males has been shown. <sup>[62][63]</sup> Researchers have also suggested that the gene contributes to hyperserotonemia. <sup>[64]</sup> However, a 2008 meta-analysis of family- and population-based studies found no significant overall association between autism and either the promoter insertion/deletion (5-HTTLPR) or the intron 2 VNTR (STin2 VNTR) polymorphisms. <sup>[65]</sup>
CACNA1G		17q21.33	Markers within an interval containing this gene are associated with ASD at a locally significant level. The region likely harbors a combination of multiple rare and common alleles that contribute to genetic risk for ASD. <sup>[66]</sup>
GABRB3, GABRA4		multiple	GABA is the primary inhibitory neurotransmitter of the human brain. Ma <i>et al.</i> (2005) concluded that GABRA4 is involved in the etiology of autism, and that it potentially increases autism risk through interaction with GABRB1. <sup>[67]</sup> The GABRB3 gene has been associated with savant skills. <sup>[68]</sup> The GABRB3 gene deficient mouse has been proposed as a model of ASD. <sup>[69]</sup>
Engrailed 2 (EN2)		7q36.2	Engrailed 2 is believed to be associated with cerebellar development. Benayed <i>et al.</i> (2005) estimate that this gene contributes to as many as 40% of ASD cases, about twice the prevalence of the general population. <sup>[70]</sup> But at least one study has found no association. <sup>[71]</sup>

# Choose a gene



# Task example 1

- Identify a gene related to the increased risk of developing late-onset Alzheimer's with specific DNA sequences. This gene is on chromosome 19 and one of the four forms of this gene accounts for about 40 percent of all cases of late-onset Alzheimer's. Write its general symbol, not differentiating the four forms.