



# GENETests Overview

**GeneTests** promotes the appropriate use of genetic services in patient care by providing current, authoritative information on inherited disorders to physicians, other healthcare providers, and researchers.

**GeneTests** comprises:



## GeneReviews

Over 500 expert-authored, peer-reviewed disease descriptions that apply genetic testing to the diagnosis, management, and genetic counseling of patients and families with specific inherited conditions.

## Laboratory Directory

Voluntary international listing of clinical and research laboratories offering in-house genetic and biochemical tests.

Linked to *GeneReviews* articles to provide information on availability of genetic tests.

## Clinic Directory

Voluntary international listing of genetics clinics offering genetic evaluation and genetic counseling.

## Educational Materials

Illustrated glossary, information on genetic services, and annotated Internet resources.



# GENETests

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# GENETests

[www.genetests.org](http://www.genetests.org)

Free Online  
Medical Genetics  
Information Resource

**Funding Support:**  
National Institutes  
of Health (NIH)

**Sponsoring Institution:**  
University of Washington  
Seattle, Washington

PMID: 20301295

- A valued international resource for physicians and clinicians
- Each entry indexed in PubMed
- Searchable on GeneTests Web site by multiple options, including:
  - Disease Name
  - Gene Symbol
  - Protein Name
  - Author
- Written by practicing physicians, clinicians, and genetics professionals
- Updated approximately every three years

- Provides links to genomic databases, consumer resources, policy statements and guidelines for genetic testing

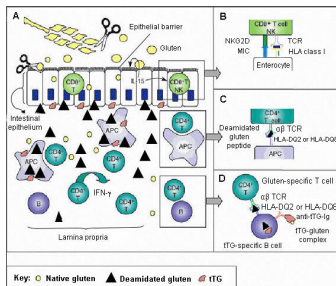


## Testing

- Links to the GeneTests Laboratory Directory to locate current testing

## Research

- Figures and illustrations included at author's discretion



- US and international clinical and research laboratories
- Laboratory contact information and links to laboratory website (when available)
- Focused on in-house molecular genetic testing, specialized cytogenetic testing, and biochemical testing for inherited disorders
- Searchable by:
  - Disease Name
  - Laboratory Name
  - Gene Symbol
  - Laboratory Director Name
  - Protein Name
  - Location (State or Country)
  - Miscellaneous Testing and Banking
- Updated annually
- Links to *GeneReviews* (when available) and other resources

## Educational Materials

- **Illustrated Glossary**
  - 225+ terms defined
  - 100+ terms illustrated
  - Links to *Case Examples* and *Learn More* illustrations
- **About Genetic Services**

Basic information about genetic counseling and testing concepts
- **Other Internet Resources**

Links to resources for health professionals and consumers

- US and international genetic clinics providing genetic evaluation and genetic counseling
- Lists services provided:
  - Adult genetics
  - Pediatric genetics
  - Preimplantation genetic diagnosis
  - Cancer genetic counseling/risk assessment
  - Telemedicine
- Searchable by:
  - Service
  - US Directory (Zip Code, State)
  - International Directory (Country)
- Updated annually
- Link to Directories of Genetic Professionals
- Links to clinic Web sites (when available)

**mitochondrial inheritance**

Mitochondria, cytoplasmic organelles that produce the energy source ATP for most chemical reactions in the body, contain their own distinct genome; mutations in mitochondrial genes are responsible for several recognized syndromes and are always maternally inherited since ova contain mitochondria, whereas sperm do not.

**Pedigree Illustrating Maternal Inheritance of a Mitochondrial Disorder**

**Key**

- Female: unaffected
- Female: affected
- Male: unaffected
- Male: affected

**Note:** Affected females transmit the disease to all their children.

**Note:** Affected males do not transmit the disease to their children.

**Case example (variable expressivity):**

**Tuberous sclerosis**

**Posted:** 10-1-02

**Related Terms:** heteroplasm; mode of inheritance; penetrance;

**Case Example** | **Full Glossary** | **Instructions**

Brain has tuberous sclerosis, an **autosomal dominant** disorder. His only clinical manifestations are typical depigmented skin lesions detectable on examination with a Woods lamp. Brian and his wife, Sai, have two affected children, Brianna and Kate. Kate's only manifestations are skin lesions. Brianna is more severely affected. In addition to typical skin findings, she has epilepsy, moderate developmental delay, and cerebral hamartomas detected by MRI. All three affected family members have the same genetic mutation, but their clinical presentations vary.

**Brian** - "Ash leaf spots" on Woods lamp examination of the skin

**Kate** - "Ash leaf spots" on Woods lamp examination of the skin

**Brianna** - "Ash leaf spots" - Intra-cerebral lesions - Epilepsy - Moderate developmental delay

**Some Clinical Implications**

- For a family in which a single individual is affected with an **autosomal dominant** disorder, accurate recurrence risk counseling may be based on clinical examination of the parents; careful examination is imperative to detect mild manifestations of the disorder.
- For **autosomal dominant** conditions with **variable expressivity**, an affected individual has a 50% risk of transmitting the mutation to his or her offspring, but the phenotype in the offspring may be mild to severe.