

## Resources

For further information on genetic testing, the resources provided below will be helpful:

**American Society of Human Genetics**

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

**Centers for Disease Control and Prevention**

[www.cdc.com](http://www.cdc.com)

**National Institutes of Health**

[www.health.nih.gov](http://www.health.nih.gov)

**National Center for Biotechnology Information**

[www.ncbi.nlm.nih.gov](http://www.ncbi.nlm.nih.gov)

**National Human Genome Research  
Institute**

[www.genome.gov](http://www.genome.gov)

**Genetic Alliance**

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

**National Society of Genetic Counselors**

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



## Examples of Genetic Syndromes

### Angelman Syndrome

*This is a condition of severe intellectual disability with absent speech, seizures, facial findings of prominent jaw with protruding tongue and wide mouth with widely spaced teeth, hypopigmentation, inappropriate laughter with a happy disposition, and characteristic gait with stiff legs and arms held flexed and perpendicular to the body.*

### Fragile X Syndrome

*Fragile X syndrome (FXS) is the most common cause of inherited mental impairment. This impairment can range from learning disabilities to more severe cognitive or intellectual disabilities. FXS is the most common known cause of autism or "autistic-like" behaviors. Symptoms also can include characteristic physical and behavioral features and delays in speech and language development.*

### Prader-Willi Syndrome

*Prader-Willi syndrome is a rare disorder present at birth that results in a number of physical, mental and behavioral problems. A key feature of Prader-Willi syndrome is a constant sense of hunger that usually begins after the first year of life. People with Prader-Willi syndrome want to eat constantly and usually have trouble controlling their weight. Many complications of Prader-Willi syndrome are due to obesity.*

**Empowerment through  
education, support and advocacy**



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For more information on this medical topic please call  
PCHC at 215-546-0300 or visit us at [www.PCHC.org](http://www.PCHC.org)

# Genetic Testing



## What is Genetic Evaluation and Testing?

Genetic evaluation and testing involves examining DNA, the chemical database that carries instructions for the body's functions. A genetic test result can help determine if there are genetic components to a disease or medical condition.

## What is the Purpose of Genetic Evaluation and Testing?

This evaluation and testing can reveal changes or alterations in genes that may cause illness or disease. Through genetic testing, health care practitioners can determine developing conditions, or medical and behavioral issues related to a specific genetic condition. In many instances, knowing the underlying cause can be very helpful in supporting the person not only currently but also as they age. In the field of intellectual and developmental disabilities, there are hundreds of genetic conditions that are known to affect behavior as well as mental and physical health.

## How Do You Obtain Genetic Evaluation and Testing?

- Discuss your concerns and your desire to have a genetic consultation/evaluation by a genetic professional (geneticist, counselor) with your primary care practitioner or specialty physician. Determine together if a consultation with a genetic professional is recommended.
- Contact your insurance company to verify that your insurance plan covers genetic consultations/evaluations/testing. Determine what is necessary for coverage of this service; many insurance providers require the primary care or specialty physician to provide documentation proving that the test is “medically necessary” for pre-authorization.
- Once approved, obtain a list of providers within the insurance plan network and make your appointment with the genetic professional. At the time of making the appointment, discuss what information will be needed for the visit. This may include insurance information, health history of the person to be tested (both physical and mental), developmental history, and extended family health history (both physical and mental).
- Meet with the genetic professional and provide any information requested; this specialist will determine what specific testing should be ordered and what is involved (e.g., blood test, mouth swab).
- Have the genetic tests completed.
- Patiently wait for the test results as the waiting period can vary by facility.
- Return to the genetic professional for your test results, counseling and planning.
- Share the test and consultation results with your primary care or specialty physician.

