RESOURCES FOR PARENTS


RESOURCES FOR PROFESSIONALS


RESOURCE MATERIALS

Gene Reviews: (www.genereviews.org). Gene Reviews is an online peer-reviewed database developed and maintained by the University of Washington. Gene Reviews currently lists 613 genetic disorders that have information authored by experts for a particular disorder. A wealth of information is presented including (1) major clinical features of the disorder, (2) what is known about the genetics and inheritance, and (3) suggestions about a differential diagnosis for the findings. Each entry has an extensive reference list and information about support groups and is updated on a regular basis. The site also includes educational materials about genetics, genetic disorders, and genetic testing including an illustrated glossary.

Genetic Alliance: (www.geneticalliance.org). The Genetic Alliance is a coalition of hundreds of genetic advocacy organizations, health professionals, clinics, hospitals, and companies. Type in a disorder and be transported to that support group’s website for information about the disorder and resources for people with the disorder.

Genetics Home Reference: (http://ghr.nlm.nih.gov). The Genetics Home Reference is the National Library of Medicine’s website for consumer information about genetic conditions and the genes or chromosomes responsible for those conditions. Summaries are provided about hundreds of genetic disorders. Each summary also contains information and Web links to other resource materials.

Genetic Testing Registry (http://www.ncbi.nlm.nih.gov/grtr). The Genetic Testing Registry is an online database maintained by the National Institutes of Health as a central repository of genetic tests. The registry lists more than 17,000 tests for 4400 disorders and 3000 genes. Information included for laboratories includes the test’s purpose, methodology, validity, evidence of the test’s usefulness, and laboratory contacts and credentials.

OMIM: (www.ncbi.nlm.nih.gov). Online Mendelian Inheritance of Man database is a catalog of genetic disorders and human genes. It is overseen by its originator, Dr. Victor A. McKusick, and maintained by the National Center for Biotechnology Information (NCBI). The database contains information about known inherited disorders, including information about the genes responsible for the disorder, a clinical synopsis, and references.