genetics resources: a directory

Connecticut Department of Public Health

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Connecticut Department of Public Health
Office of Planning, Communication, and Workforce Development
410 Capitol Avenue, MS#13PCW
Hartford, Connecticut 06134-0308

Telephone: (860) 509-7122
Fax: (860) 509-7160
E-mail: webmaster.dph@po.state.ct.us

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STATE OF CONNECTICUT

DEPARTMENT OF PUBLIC HEALTH

J. Robert Galvin, M.D., M.P.H.
Commissioner

M. Jodi Rell
Governor

October 1, 2004

Dear Reader:

I am pleased to present the Connecticut Department of Public Health's new publication, **Genetics Resources: A Directory.**

Advances in genetics hold the promise of great benefits to the health of Connecticut residents. Yet there is a growing need for enhanced genetics knowledge among physicians, other health care providers, and the general public. To help meet this need, the department has developed a genetics resources directory - a timely source for accessing local, regional, and national genetics information. This directory was developed to help Connecticut residents and their medical caregivers access information about the significant role that genes play in human health and disease, and locate services such as genetic testing, treatment, and follow-up.

The Directory begins with an overview of basic genetics, and provides sources for additional information. It also displays clusters of various genetic conditions and lists specialized resources and supports for each condition whenever possible.

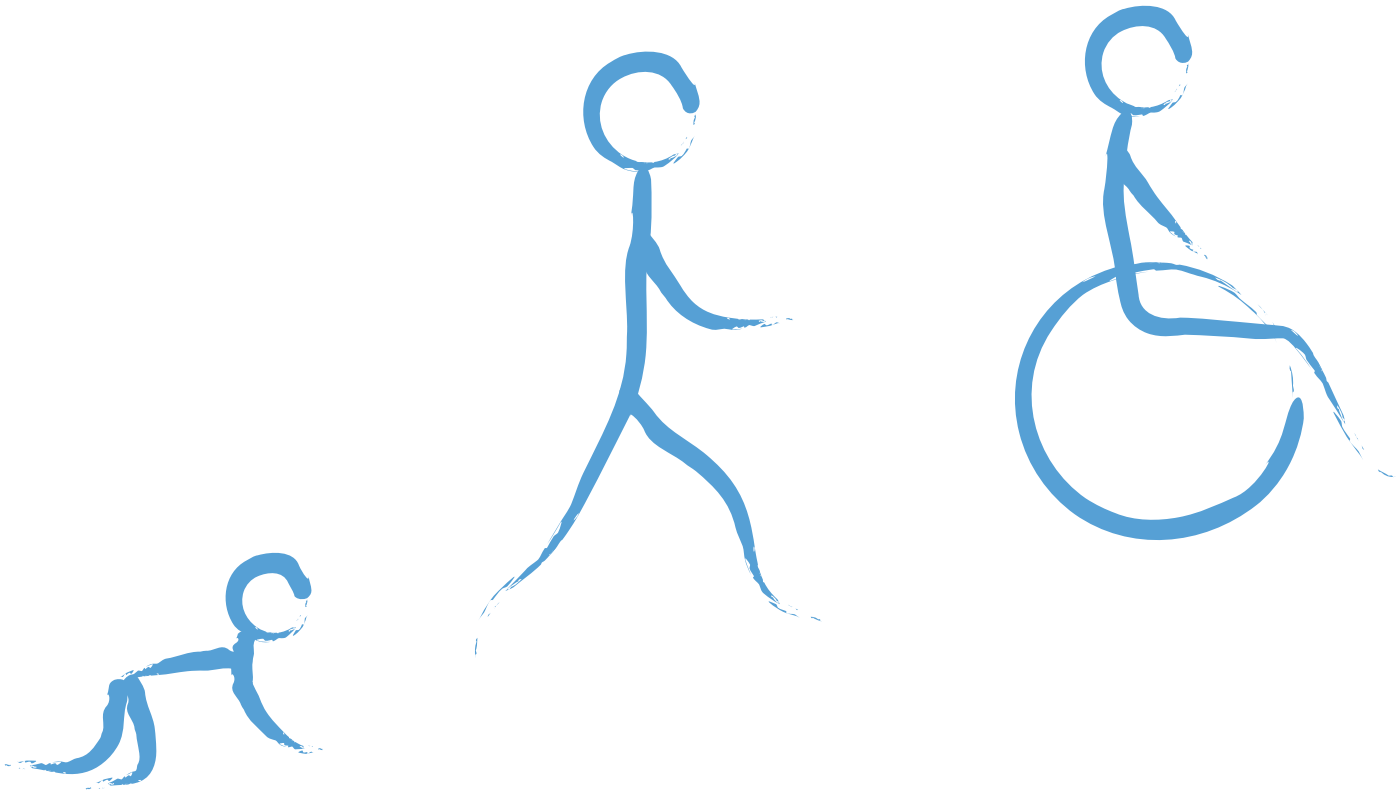
We hope you find this Directory a useful tool in your search for genetics information. We welcome your feedback on its utilization and your suggestions regarding future public education efforts in genetics.

Sincerely,

J. Robert Galvin, M.D., M.P.H.
Commissioner

PHONE: (860) 509-7101 FAX: (860) 509-7111
410 CAPITOL AVENUE - MS#13COM, P.O. BOX 3340308, HARTFORD, CONNECTICUT 06134-0308

Genetics Resources: A Directory



Connecticut Department of Public Health

ACKNOWLEDGEMENTS

Contributors:

CT Department of Public Health

Genetics Planning Team:

Meg A. Hooper, M.P.A., Project Director
Lloyd Mueller, Ph.D., Supervising Epidemiologist
Beverly Burke, M.S.W., Lead Planner
Carol Stone, Ph.D., M.S., M.A.S., Epidemiologist
Joan Foland,M.H.S.,M.Phil., Epidemiologist

William Gerrish, M.B.A., Director
Office of Planning, Communication & Workforce Development

Prepared by:
Leslie Ciarleglio, C.G.C., N.E.R.G.G., Inc.

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PREFACE

Rapid developments are causing genetics to become an increasingly integral part of medical care. The optimal translation of genetic developments into medical practice is just emerging. Additionally, the communication of genetic information between medical providers and patients, patients and their medical providers, and genetic services and the public will require special caution and counseling because genetics – and genetic information- affects people in unique ways. Such information may predict events that will occur years in the future, or not at all. It may predict the future of other family members, and has the potential impact to challenge, and even stigmatize both individuals and families.[1]

This publication, **Genetics Resources: A Directory**, is a product of a multi-year genetics planning initiative at the Connecticut Department of Public Health. The development of the directory was prompted by concern that a lack of general, current genetics knowledge among the public and medical professionals could pose the potential for missed health promotion opportunities. The development of this up-to-date resource for genetics information is an important part of a strategy aimed at increasing genetics 'literacy' for all.

In that light, this directory is intended to assist the general public, affected families, and the medical community to further their knowledge of genetics, genetic conditions, services, and supports – and to make it easier for those searching for such sources to locate and access them. Readers will note that the information and resources included in the Directory extend beyond those relating to the traditional public health genetics efforts in newborn screening to encompass the impact of genetics across the lifespan. You will also note that wherever possible, accompanying the listing relating to a particular diagnosis or genetic condition, are additional sources of information in print and further readings.

It should be noted that this first printing of Connecticut's **Genetics Resources: A Directory**, will require updating and revision as the "genomics revolution" continues its rapid expansion, poses new challenges and the creation of new resources in response to them.

As our planning and preparation for the impacts of genetic developments on the public's health in Connecticut continue, the need for sharing timely, accurate and useful information will grow. Public health will continue to play a critical role in addressing the challenges posed by genetics in the future.

1. N.E.R.G.G., The Genetic Resource, Volume 10, No.2

Table of Contents

I. GENETICS THROUGHOUT THE LIFESPAN

Human genetics. Understanding ourselves and our make-up from the most basic of perspectives can be overwhelming. The study of genetics at one time was considered obscure. Even today, with the completion of the Human Genome Project and the related media exposure, the general public's perception about the risk for birth defects or genetic disease is often stated as a "one in-a-million" possibility. Many people believe that this science is remote from their day-to-day lives. The reality, however, is that genomic medicine is here to stay and genetic testing and information will be encountered by all people at some time in their lives.

Genetics is the transfer of information from one generation to the next. The foundation of genetics is genes, which are made up of molecules of DNA (deoxyribonucleic acid). Genes come in pairs, and are packaged in structures called chromosomes, which collectively form an organism's genome.

Every cell in an individual contains every gene in its' entire genome. It was originally believed that humans had approximately 100,000 genes; however, it is now known that the human genome is made up of only about 35,000 genes. Humans have a total of 46 chromosomes in each cell, which like genes, also come in pairs, 23 pairs altogether. One chromosome in each pair comes from an egg from the individual's mother, and the other member of the pair comes from sperm from the individual's father. The joining of egg and sperm at conception results in a fertilized egg with 46 chromosomes, 23 pairs. Of the 23 pairs of chromosomes, 22 pairs are called the autosomes. The 23rd pair are the sex chromosomes, with females having two "X" chromosomes, and males having one "X" chromosome and one "Y" chromosome.

Everything about us, including our health, at least in part, is determined by our genes. Our genetic blueprint not only determines whether or not we or our offspring have a serious genetic disorder, but also determines our chance to develop a chronic disease, or our susceptibility to an infectious disease.

Traditionally, genetic testing and counseling has centered on prenatal and pediatric genetic diagnoses. This is changing and rapidly expanding into other medical fields. However, there are key times throughout the lifespan that genetic issues are likely to be encountered. To begin, patients and families involved in reproductive decision-making face genetic concerns. For example, prior to pregnancy, parental carrier testing is routinely available for a variety of genetic diseases more common in specific ethnic groups. Gamete donors are often asked to submit to a variety of genetic tests. Pre-implantation genetic diagnosis affords couples known to be at risk for genetic disease in the fetus the option of making that diagnosis in fertilized eggs *in-vitro* prior to transfer to the uterus.

Historically, women 35 years of age or older at the time of expected delivery were the typical prenatal genetics referral. Now, with the growing list of prenatal testing options, all pregnant women and women planning a pregnancy are potential candidates for genetic counseling.

Each year, state newborn screening programs test millions of newborns for disorders that require early detection and medical treatment to prevent serious illness or death. The Connecticut Department of Public Health currently tests for nine disorders through its Newborn Screening Program. Determining which disorders should be included in newborn screening is controversial. The challenge is maintaining the ability to manage test results effectively and provide quality care to patients, while keeping pace with advancing technology and the ability to test for additional conditions.

Genetic evaluation and testing is nothing new for pediatricians and other health care professionals who work with children with birth defects. However, a new challenge is the controversial issue of testing minors for adult-onset conditions.

The identification of susceptibility genes for common adult-onset genetic disease is moving the field of genetic testing and counseling in a new and demanding direction. Diabetes, certain cancers, and Alzheimer's disease are some examples of conditions for

which a genetic component has been identified. Understanding and uncovering the genetic basis of “common” diseases is bringing genetics into primary care and into adult medicine, a relatively new area. Additionally, genomic medicine confronts the way we have historically defined health and illness. Pre-symptomatic and susceptibility testing have defined an entirely new concept – people described as “genetically unwell” – healthy individuals who are expected to develop or have an increased risk to develop a disorder. The psychosocial implications of this information are still being explored and represent a continuing challenge to the genetics profession.

Geneticists and the individuals and families they serve, have valid concerns about genetic testing and genomic medicine. They worry about the possible misuse of information about genetic traits or predisposition as a basis for denial of health coverage or employment.

The issue of gene patents is another area of controversy. There are legal and ethical issues posed by storage of DNA samples. Also, genetic information is familial; the test results of one person have implications for other family members. Finally, many genetic conditions remain difficult to treat or prevent, and genetic testing may be limited to providing information only, rather than affecting the clinical course of disease.

Given these concerns, detailed counseling, informed consent, and confidentiality should be key aspects of the genetic testing process. Exciting and stimulating, the study of genetics and genomics continues to bring hope to many individuals and families.

II. GENETIC SERVICES IN CONNECTICUT

Bridgeport Hospital

267 Grant Street
Bridgeport, CT 06610
www.bridgeporthospital.com

Maternal Fetal Medicine
Phone: 203-384-3049
Pediatric
Phone: 203-384-3574
An outreach site of Yale University
School of Medicine

Connecticut Department of Public Health

Newborn Screening Program
410 Capitol Avenue
P.O. Box 340308
Hartford, CT 06134-0308
Phone: 860-509-8081
Fax: 860-509-7720
www.dph.state.ct.us

Commissioner: J. Robert Galvin, MD, MPH
Deputy Commissioner: Norma Gyle, RN, PhD

Connecticut State Law mandates that all newborns delivered in Connecticut be screened for selected genetic and metabolic disorders. The Newborn Screening Program consists of three components: testing,tracking, and treatment. The aim of this program is to screen all babies prior to hospital discharge or within the first 4 days of life. The goal is early identification of infants at increased risk for disorders so that medical treatment can be promptly initiated to avert complications and prevent irreversible problems or death.

Greenwich Hospital

5 Perryridge Road
Greenwich, CT 06830
203-863-3000

Genetic Consultation Service
Phone: 203-863-3552
An outreach site of Yale University

Perinatal Genetic Counseling
Phone: 203-863-3917
Fax: 203-863-3467
Email: jodir@greenhosp.org
www.greenhosp.org

Norwalk Hospital

Maple Street
Norwalk, CT 06856
www.norwalkhosp.org

Perinatology
Phone: 203-855-3588
Fax: 203-852-3610

Stamford Hospital

Shelburne Avenue at West Broad Street
Stamford, CT 06904
Phone: 203-325-7000
www.stamhealth.org

Maternal Fetal Medicine
Phone: 203-325-7060
Fax: 203-325-7908

University of Connecticut Health Center

Division of Human Genetics
UConn Health Partners
65 Kane Street, MC 7120
West Hartford, CT 06119
Phone: 860-523-6464
Fax: 860-523-6465
www.uchc.edu/genetics

General Genetics Consultation Service
Phone: 860-523-6464
Fax: 860-523-6465
Provides up-to-date information about the heritability and cause of genetic disorders, such as birth defects, mental retardation and late onset adult diseases, provide evaluation, diagnosis and management of persons with genetic disorders; provides information about the recurrence risk to families with a genetic disorder; and providessupportive counseling to them and their families.

Hereditary Cancer Program
Phone: 860-523-6464
Fax: 860-523-6465
A referral/consultation service, which evaluates families with multiple members with cancer, to assess the likelihood for hereditary cancer. Genetic testing is offered to families with appropriate histories.

Patients also seen at:

Middlesex Hospital
28 Crescent Street
Middletown, CT 06457
www.midhosp.org

Gaucher Disease Center

Phone: 860-523-6464
Fax: 860-523-6465
Established through the National Gaucher Foundation (www.gaucherdisease.org), to work with patients and doctors in diagnosis, management and treatment of Gaucher Disease.

Genetic Inborn Errors of Metabolism Service

Phone: 860-523-6464
Fax: 860-523-6465
Provides diagnostic evaluation of metabolic disorders that are detected by the Connecticut State Newborn Screening Program or by other evaluations.

Other Collaborative Programs

Phone: 860-523-6464
Fax: 860-523-6465
Hemophilia
Huntington Disease
Neurogenetics Program
Neuromuscular Disorders
Newborn Cystic Fibrosis Screening Program
Education
Phone: 860-523-6464
Fax: 860-523-6465
Certified residency in medical genetics, clinical electives/rotations for medical students, residents, genetic counseling students, and graduate fellows, undergraduate, graduate and continuing education.

Prenatal Genetics Program

Phone: 860-523-6464
Fax: 860-523-6465
Provides up-to-date information about the inheritance of genetic conditions, about the cause for various birth defects, and the tests that are available to detect whether an individual or couple has a chance to have a child with such a condition.

Offers tests to determine whether a pregnancy may be affected by a birth defect or genetic condition.

Patients also seen at:

Middlesex Hospital
28 Crescent Street
Middletown, CT 06457
www.midhosp.org

Hartford Hospital
Maternal-Fetal Medicine
80 Seymour Street
Hartford, CT 06102
www.harthosp.org

Connecticut Pregnancy Exposure Information Service

Phone: 800-325-5391/860-523-6419
Fax: 860-523-6465
www.docdb.uchc.edu/genetics/PregnancyExposure.htm

As a state-funded program that provides up-to-date information on all types of exposures during pregnancy for Connecticut residents or women with Connecticut physicians. This service is a member of The Organization of Teratology Information Services.
Located in West Hartford.

Patients with workplace exposures also seen in Occupational and Environmental Reproductive Hazards Clinic
UConn Health Center
Dowling North Building, 3rd floor
Farmington, CT 06032
Phone: 800-325-5391/860-523-6419
www.oehc.uchc.edu/clinser/reprohaz.htm

Laboratory Services

Human Genetics Laboratories
UConn Health Center
263 Farmington Avenue
Butler Building #5
Farmington, CT 06030-6140
Phone: 860-679-2469
Fax: 860-679-3616

Yale University School of Medicine

Department of Genetics
333 Cedar Street
New Haven, CT 06520
www.info.med.yale.edu/genetics

Genetic Consultation Service
Phone: 203-785-2660
Fax: 203-785-3404
Diagnosis, management and genetic counseling for a broad range of hereditary and genetic-influenced conditions. Ongoing treatment including nutritional treatment, bone marrow and stem cell transplant, pharmacological therapy, and multidisciplinary management.

Patients also seen at:

Bridgeport Hospital
267 Grant Street
Bridgeport, CT 06610
Phone: 203-384-3049
www.bridgeporthospital.com

Greenwich Hospital
5 Perryridge Road
Greenwich, CT 06830
Phone: 203-863-3552
www.greenhosp.org

Danbury Hospital
24 Hospital Avenue
Danbury, CT 06810
Phone: 203-797-7124
www.danhosp.org

Genetic Inborn Errors of Metabolism Service
Phone: 203-785-2660
Fax: 203-785-3404

Statewide Genetics Services Program
Phone: 203-785-2660
Fax: 203-785-3404
Part of the State Department of Public Health program to community based medical genetic services with public health planning.

Other Collaborative Programs:

Craniofacial Genetics
Phone: 203-737-2049
Fax: 203-785-5714

Huntington Disease
Phone: 203-785-2661
Fax: 203-785-7673

Neurofibromatosis Program
Phone: 203-785-2660
Fax: 203-785-3404
Neurogenetics Clinic
Phone: 203-785-4085
Fax: 203-785-5694

Cancer Genetic Counseling Program
Yale Cancer Center
55 Church Street
Suite 800B
New Haven, CT 06510
Phone: 203-764-8400
Fax: 203-764-8401
www.info.med.yale.edu/ycc

Outreach sites:

Praxair Cancer Center
Danbury Hospital
24 Hospital Avenue
Danbury, CT 06810
www.danhosp.org

Bendheim Cancer Center
Greenwich Hospital
77 Lafayette Place
Greenwich, CT 06830
www.greenhosp.org

Whittingham Cancer Center
Norwalk Hospital
Maple Street
Norwalk, CT 06856
www.norwalkhosp.org

Education:

Phone: 203-785-2660
Fax: 203-785-3404
Certified residency in medical genetics, clinical electives/rotations for medical students, residents, genetic counseling students, and graduate fellows, undergraduate, graduate and continuing education. Laboratory training in clinical molecular genetics, clinical biochemical genetics, and clinical cytogenetics

PRENATAL SERVICE
Department of Genetics
333 Cedar Street
New Haven, CT 06520
Phone: 203-785-2661
Fax: 203-785-7673

Prenatal Outreach Sites:

Lawrence and Memorial Hospital
365 Montauk Avenue
New London, CT 06320
www.lmhospital.org

Waterbury Hospital
64 Robbins Street
Waterbury, CT06721
www.waterburyhospital.org

LABORATORY SERVICES
Biochemical Disease Detection Laboratory
Yale University School of Medicine
Department of Genetics
333 Cedar Street
WWW 313
P.O. Box 208005
New Haven, CT 06520
Phone: 203-785-2662
FAX: 203-785-3535
Email: biochemical.lab@yale.edu

Studies include amino acid analysis (HPLC), total homocysteine (HPLC), organic acid analysis (GC/MS), carnitine and acylcarnitine profiles (electrospray tandem MS), screening for fatty acid oxidation disorders (urine acylglycine profile).

CYTOGENETICS
(including molecular cytogenetics)
Yale University School of Medicine
Department of Genetics
333 Cedar Street
WWW 333
New Haven, CT 06520

Laboratory Director: 203-785-6317
Administrative Assistant: 203-785-2146
Prenatal laboratory: 203-785-5140
Bone Marrow & Blood laboratory:
203-785-2656
Research Cytogenetics: 203-785-6523

DNA Diagnostics Laboratory
Yale University School of Medicine
Department of Genetics
333 Cedar Street
New Haven, CT 06520
Phone: 203-785-5745
Fax: 203-785-7227

Studies include fragile X testing, adult polycystic kidney disease, Duchenne and Becker muscular dystrophy, Gorlin syndrome, Hemophilia A, medium chain acyldehdrogenase deficiency, multiple endocrine neoplasia, and ornithine transcarbamylase deficiency.

Did You Know...?

Goldfish have more.....
chromosomes than humans.

III. UMBRELLA ORGANIZATIONS & REFERENCES

A. Consumer Organizations:

Genetic Alliance

4301 Connecticut Avenue, NW
Suite 404
Washington, DC 20008
Phone: 800-336-4363/202-966-5557
Fax: 202-966-8553
Email: info@geneticalliance.org
www.geneticalliance.org

The Genetic Alliance is an international coalition comprised of millions of individuals with genetic conditions and over 600 advocacy, research and health care organizations. The staff and board work together to empower individuals and families living with genetic conditions.

March of Dimes

1275 Mamaroneck Avenue
White Plains, NY 10605
Phone: 888-663-4637/914-428-7100
Fax: 914-997-4763
Email: askus@marchofdimes.com
www.modimes.org
The mission of the foundation is to improve the health of babies by preventing birth defects and infant mortality. The March of Dimes funds programs of research, community services, education, and advocacy.

In Connecticut:
255 Pitkin Street
East Hartford, Ct 06108
Phone: 860-290-5440
Fax: 860-290-5433
Email: CT322@marchofdimes.com
www.marchofdimes.com/connecticut

South Division
11 Belden Avenue
Norwalk, CT 06850
Phone: 203-849-9800
Fax: 203-847-8060
Email: CT322@marchofdimes.com
www.marchofdimes.com/connecticut

National Organization for Rare Disorders (NORD)

55 Kenosia Avenue
P.O. Box 1968
Danbury CT 06813
Phone: 800-999-6673/203-744-0100
Fax: 203-798-2291
Email: orphan@rarediseases.org
www.rarediseases.org

NORD is a unique federation of voluntary health organizations dedicated to helping people with rare "orphan" disease and assisting the organizations that serve them. NORD is committed to the identification, treatment, and cure of rare disorders, through programs of education, advocacy, research, and service.

National Healthy Mothers, Healthy Babies Coalition

121 North Washington Street
Suite 300
Alexandria, VA 22314
Phone: 703-836-6110
Fax: 703-836-3470
Email: info@hmhb.org
www.hmhb.org

Focuses attention on raising public awareness of the basic components of prenatal care – early care, good nutrition, avoidance of drugs (including tobacco and alcohol), and promotion of breastfeeding.

B. On-Line Resources

On-line Mendelian Inheritance in Man (OMIM)

www.ncbi.nlm.nih.gov/entrez/query/fcgi?db=OMIM
This database is a catalog of human genes and genetic disorders authored and edited by Dr. Victor A. McKusick and his colleagues.

GeneTests

www.genetests.org
Information for health professionals about hundreds of genetic tests. Also includes links to disease information and geographic listings of genetic services.

U.S. National Screening Status Report

www.genes-r-us.uthscsa.edu/resources/newborn/screenstatus.htm
Lists status of newborn screening in the United States, state by state, including links to additional conditions states are screening for using tandem mass spectrometry. Part of National Newborn Screening and Genetics Resource Center, a cooperative agreement between the Maternal and Child Health Bureau Genetic Services Branch and the University of Texas Health Science Center at San Antonio Department of Pediatrics.

National Institutes of Health

www.nih.gov
U.S. Department of Health and Human Services, A-Z index of NIH health resources, clinical trials, health hotlines, MEDLINE plus, drug information.

National Library of Medicine

www.ncb.nlm.nih.gov
Part of the NIH, the world's largest medical library. Search on health topics, drug information, medical encyclopedia and dictionary, news, directories and other resources. Includes a link to "Genetics Home Reference", a website for consumer information about genetic conditions, disease summaries, gene and chromosome summaries, and a glossary of genetic and medical terms.

National Human Genome Research Institute

www.genome.gov
An on-line listing of information available on the Web for researchers, consumers, support and advocacy groups, health professionals, policy makers and more. Includes links to clinical genetics resources, search engines and family history tools, consumer resources, support groups, legal and ethical issues in genetics, and educational and genetic resources for health professionals such as primary care providers and nurses.

Information for Genetic Professionals

www.kumc.edu/gec/geneinfo.html
Comprehensive on-line service based at University of Kansas Medical Center, provides information for genetic professionals, with clinical, research, and educational resources for genetic counselors, clinical geneticists and medical geneticists. Includes Human Genome Project resources, centers, news and other online sites, educational resources including curricula, lesson plans and activities, courses, books and other publications, videotapes, and computer programs. Also includes listings of upcoming professional meetings in genetics, and information on careers in human genetics

C. Professional Genetics Society

American College of Medical Genetics (ACMG)

9650 Rockville Pike
Bethesda, MD 20814
Phone: 301-634-7127
Fax: 301-634-7275
Email: acmg@acmg.net
www.acmg.net

ACMG is an organization composed of biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors and other health care professionals committed to the practice of medical genetics. The ACMB provides education, resources and a voice for the medical genetics profession. To make genetic services available to and improve the health of the public, the ACMG promotes the development and implementation of methods to diagnose, treat and prevent genetic disease.

Did You Know...?

If typed, the genetic code of a human would fill about 134 complete sets of the Encyclopedia Brittanica.

American Society for Human Genetics

www.ashg.org

The American Society for Human Genetics is a professional organization composed of researchers, academicians, clinicians, laboratory professionals, genetic counselors, and nurses with the principal objectives of sharing research, informing health professionals, legislators, health policy makers and the general public about human genetics, and facilitation interactions amongst all of these groups.

National Society of Genetic Counselors (NSGC)

Executive Office
233 Canterbury Drive
Wallingford, PA 19086
Phone: 610-872-7608
Fax: call for faxing information
Email: FYI@nsgc.org
www.nsgc.org

The NSGC is the leading voice, authority and advocate for the genetic counseling profession, with the aim of promoting the genetic counseling profession as a recognized and integral part of health care delivery, education, research and public policy. In addition, the NSGC promotes the professional interests of genetic counselors and provides a network for professional communications. Local and national continuing education opportunities and the discussion of all issues relevant to human genetics and the genetic counseling profession are an integral part of belonging to the NSGC.

National Coalition for Health Professional Education in Genetics (NCHPEG)

2360 West Joppa Road
Suite 320
Lutherville, MD 21093
Phone: 410-583-0600
Fax: 410-583-0520
Email: geneticsinfo@nchpeg.org
www.nchpeg.org

A national effort to promote health professional education and access to information about advances in human genetics. NCHPEG members are an interdisciplinary group of leaders from health professional organizations, consumer and voluntary groups, government agencies, private industry, managed care organizations, and genetics professional societies seeking to capitalize on the collective expertise and experience of members and to reduce duplication of effort.

D. Reference Texts:

Emery and Rimoin's Principles and Practices of Medical Genetics

by Connor, J. M., Pyeritz, R., Korf, B., and Rimoin, D.
Churchhill Livingstone, 4th Edition, 2001
Extensive reference on the basic science and clinical applications of medical genetics. For primary care physicians, specialists, and students.

Management of Genetic Syndromes

by Cassidy, S. B., and Allanson, J. E.
Wiley-Liss, 2001
Covering 30 common genetic syndromes. Information on incidence, etiology and pathogenesis, natural history, diagnostic criteria, spectrum of variation, recurrence risk in siblings and offspring, and availability of prenatal diagnosis and diagnostic testing.

Smith's Recognizable Patterns of Human Malformation

by Jones, K. L.
W. B. Saunders Company, 5th Edition, 1996
Clinical atlas on malformations. Discusses morphogenesis, genetics, genetic counseling and clinical diagnosis.

The Metabolic and Molecular Bases of Inherited Disease

by Scriver, C. R., Sly W. S., Childs, B., Beaudet, A. L., Valle, D., Kinzler, K. W., and Vogelstein, B.
McGraw-Hill, 8th Edition, 2000
The undisputed authority on genetic inheritance, you can explore what is currently known about every inherited disease known to exist.

THE PRACTICAL GUIDE TO THE GENETIC FAMILY HISTORY
by Bennett, R. L.
Wiley-Liss, 1999
Excellent and thorough reference, well-organized with complete index and references, especially useful for the primary care physician.

PRACTICAL GENETIC COUNSELING
by Harper, P. S.
Arnold Publishers, 5th Edition, 1998
Provides clinicians and other staff members with an updated guide through the profusion of new information and the emerging psychosocial and ethical concerns connected with genetic counseling.

A GUIDE TO GENETIC COUNSELING
by Baker, D. L., Schuette, J. L., and Uhlmann, W. R.
John Wiley & Sons, 1998
Defines theory, goals, and core competencies associated with the practice of genetic counseling.

STRUCTURAL FETAL ABNORMALITIES: THE TOTAL PICTURE
by Sanders, R..
C. V. Mosby, 2002
Wonderful reference text of ultrasound diagnosed fetal anomalies.

ULTRASOUND OF FETAL SYNDROMES
by Benacerraf, B. R.
Churchhill Livingstone, 1998
Thorough quick reference volume of ultrasound anomalies and possible syndromic etiologies, juxtaposed with ultrasound findings in specific syndromes.

IV. CONSUMER SUPPORT ORGANIZATIONS AND RESOURCES
(alphabetically by condition)

~ ACHONDROPLASIA ~

Little People of America
5289 NE Elam Young Parkway
Suite F-700
Hillsboro, OR 97124
Phone: 888-LPA-2001/503-846-1562
Fax: 503-846-1590
Email: info@lpaonline.org
www.lpaonline.org

In Connecticut:
District 1 (CT, ME, MA, NH, RI, VT)
Phone: 203-641-9760
Fax: none
Email: chayim76@yahoo.com
www.lpad1.org

Additional web resources:

International Skeletal Dysplasia Registry at Cedars-Sinai Health System
www.csmc.edu/medgenetics/3086.asp
A referral center for research into the diagnosis, management, and etiology of skeletal dysplasias.

Additional literature:

Dwarfism: The Family and Professional Guide
by Scott, C.J., Mayeaux, N, Crandall, R, and Weiss, J.
Short Stature Foundation, 1994
Non-fiction

Little People: Learning to See the World Through My Daughter's Eyes
by Kennedy, D.
Rodale Books, 2003
Non-fiction

Stones from the River
by Hegi, U.
Simon & Schuster, 1994
Fictional story of a woman with a skeletal dysplasia, coming of age just as Hitler's pronouncements begin to threaten the Jewish residents of her town, and all of Germany.

Mendel's Dwarf
by Mawer, S
Penguin Books, 1999
This novel tells the story of Benedict Lambert, a distant relative of Gregor Mendel. Like Mendel, Lambert is a brilliant geneticist. Unlike Mendel, Lambert is a dwarf. The story is a mix of humor and philosophy.

~ ADRENAL DISORDERS ~

National Adrenal Diseases Foundation
505 Northern Boulevard
Great Neck, NY 11021
Phone: 516-487-4992
Fax: 516-829-5710
Email: nadfmail@aol.com
www.medhelp.org/www/nadf.htm
No Connecticut chapter

Additional web resources:

Your Adrenal Glands
www.endocrineweb.com/adrenal.htm
Information on the anatomy and physiology of the adrenal glands.

~ ALAGILLE SYNDROME ~

Alagille Syndrome Alliance
10630 SW Garden Park Place
Tigard, OR 97223
Phone: 503-639-6217
Fax: none available
Email: alagille@earthlink.net
www.alagille.org
No Connecticut chapter.

Additional web resources:

Cincinnati Children's Hospital Medical Center page on Alagille syndrome
www.cincinnatichildrens.org/svc/prog/liver/diseases/alagille-syndrome.htm
Lists explanation, genetics, signs and symptoms, diagnosis, treatment, and prognosis for Alagille syndrome.

~ ALBINISM AND HYPOPIGMENTATION ~

National Organization for Albinism and Hypopigmentation (NOAH)

P.O. Box 959
East Hampstead, NH 03826
Phone: 800-473-2310/603-887-2310
Fax: 603-887-6049
Email: info@albinism.org
www.albinism.org
For Connecticut information, call national office.

Hermansky-Pudlak Syndrome Network, Inc.

1 South Road
Oyster Bay, NY 11771
Phone: 800-789-9477/516-922-3440
Fax: 516-922-4022
Email: hpsn@juno.com
www.medhelp.org/web/hpsn.htm
A special interest group of NOAH.

Additional web resources:

International Albinism Center at the University of Minnesota

www.sbs.umn.edu/iac
Web page of a multidisciplinary group of researchers with interests in clinical genetics, molecular biology, ophthalmology, dermatology, and biochemistry, all with a central theme of understanding the cause and effect of albinism and other forms of pigment loss in humans.

Additional literature:

Living with Albinism

by Landau, E.
Franklin Watts, 1999
Children's book, defines disease and discusses cause and associated problems, lifestyle and well-being. A positive book.

Too White to be Black and Too Black to be White

by Edwards, L.G.
Authorhouse, 2001
Expresses true emotion and life experiences of author, an African American with albinism.

~ ALZHEIMER'S DISEASE ~

Alzheimer's Association

225 North Michigan Avenue
Suite 1700
Chicago, IL 60601
Phone: 800-272-3900/312-335-8700
Fax: 312-335-1110
Email: info@alz.org
www.alz.org

In Connecticut:
96 Oak Street
Hartford, CT 06106
Phone numbers:
866-363-6679/800-356-5502/860-956-9560
Fax: 860-956-9590
Email: see website for multiple board members' email addresses
www.alzct.org (with links to 98 support groups)

Alzheimer's Foundation of America

322 8th Avenue, 6th floor
New York, NY 10001
Phone: 866-232-8484
Fax: 646-638-1546
Email: info@alzfdn.org
www.alzfdn.org
No Connecticut chapter

Additional web resources:

Alzheimer's Disease Education and Referral Center (ADEAR)

www.alzheimers.org
A service of the National Institute on Aging, the ADEAR center is a site that can be used to find current, comprehensive Alzheimer's Disease and resources.

Additional literature:

Alzheimer's Early Stages: First Steps for Family, Friends, and Caregivers
by Kuhn, D. and Bennett, D.A.
Hunter House, 2nd Edition, 2003
Latest information on risk factors, treatment, prevention, medications, financial aspects, and reflections by family members.

Learning to Speak Alzheimer's: A Groundbreaking Approach for Everyone Dealing with the Disease
by Coste, J.K.
Houghton Miiflin, 2003
Provides hundreds of practical tips to ease life for everyone involved with Alzheimer's disease.

Did You Know...?

Sometimes genes "jump" around on a chromosome.

~ AMYOTROPHIC LATERAL SCLEROSIS (ALS) ~

ALS Association

27001 Agoura Road
Suite 150
Calabasas Hills, CA 91301
Phone: 800-782-4747, 818-880-9007
Fax: 818-880-9006
Email: alsinfo@alsa-national.org
www.alsa.org

In Connecticut:
4 Oxford Road
Unit D1
Milford, CT 06460
Phone: 877-257-2281/203-874-5050
Fax: 203-874-7070
Email: als.assoc@snet.net
www.alsact.org

Additional web resources:

ALS Survival Guide
www.lougehrigsdisease.net
Informational and inspirational site with links to What is ALS, causes, news treatments, and an "Ask the Expert" link, support groups and other useful links.

Additional literature:

Amyotrophic Lateral Sclerosis: A Guide for Patients and Families
by Mitsumoto, H. and Munsat, T.L.
Demos Medical Publishing, 2nd Edition, 2001
Medical aspects, rehabilitative management, day-to-day living, end of life issues, resources for families, legal, ethical, and spiritual issues.

~ ASTHMA ~

Allergy and Asthma Network – Mothers of Asthmatics, Inc.

2751 Prosperity Avenue
Suite 150
Fairfax, VA 22031
Phone: 800-878-4403/703-641-9595
Fax: 703-573-7794
Email: aanma@aol.com
www.aanma.org
No Connecticut chapter.

American Lung Association

61 Broadway, 6th floor
New York, NY 10006
Phone: 800-586-4872/212-315-8700
Fax: 212-265-5642
Email: no main email address
www.lungusa.org

In Connecticut:
American Lung Association of Connecticut
45 Ash Street
East Hartford, CT 06108
Phone: 800-922-2263/860-289-5401
FAX: 860-289-5405
Email: alaofct@alact.org
www.alact.org

American Academy of Allergy Asthma and Immunology

611 East Wells Street
Milwaukee, WI 53202
Phone: 800-822-2762/414-272-6071
Fax: 414-276-3349
Email: info@aaaai.org
www.aaaai.org
No Connecticut chapter.

Asthma and Allergy Foundation of America, Inc.

1233 20th Street NW
Suite 402
Washington, DC 20036
Phone: 800-727-8462/202-466-7643
Fax: 202-466-8940
Email: info@aafa.org
www.aafa.org
No Connecticut chapter.

New England Chapter:

220 Boylston Street
Chestnut Hill, MA 02467
Phone: 877-2-ASTHMA/617-965-7771
Fax: 617-965-8886
Email: aafane@aol.com
www.asthmaandallergies.org

Additional web resources:

Global Initiative for Asthma (GINA)

www.ginasthma.com
Objectives of GINA include: increase awareness of asthma and its public health consequences, reduce morbidity and mortality, improve management, and increase the availability and accessibility of effective asthma therapy.

Additional literature:

Allergies and Asthma for Dummies

by Berger, W.E.
John Wiley & Sons, 2000
Prevention, treatment, recognition and management of triggers, medications for short and long term relief. Easy to understand.

The Harvard Medical School Guide to Taking Control of Asthma: A Comprehensive Prevention and Treatment Plan for You and Your Family

by Fanta, C.H., Cristiano L.M., and Haver, K.
Simon & Schuster, 2003
Discussed diagnosis, symptoms individual management plans, current therapies, pros and cons of medications.

~ ATAXIA TELANGIECTASIA ~

Ataxia Telangiectasia Children's Project

6685 Military Trail
Deerfield Beach, FL 33442
Phone: 800-543-5728/954-481-6611
Fax: 954-725-1153
Email: info@atcp.org
www.atcp.org
For local resources, call national office.

A-T Medical Research Foundation

5241 Round Meadow Road
Hidden Hills, CA 91301
Phone: 818-704-8146
Fax: 818-703-8310
Email: gsmith@gspartners.com
www.gspartners.com/at
No Connecticut chapter.

Additional web resources:

Ataxia Telangiectasia Mutation Database

www.vmresearch.org/bri_investigators/atm.htm
Scientific site with listings of AT mutations associated with cancer, AT polymorphisms, and variants, and nucleotide sequence of AT gene. Includes information on AT.

~ AUTISM ~

Autism Research Institute

4182 Adams Avenue
San Diego, CA 92116
Phone: 619-281-7165
Fax: 619-563-6840
Email: none
www.autismresearchinstitute.com
No Connecticut chapter.

Autism Society of America

7910 Woodmont Avenue
Suite 300
Bethesda, MD 20814
Phone: 800-3AUTISM/301-657-0881
Fax: 301-657-0869
Email: see website for specific departments
www.autism-society.org

In Connecticut:
Autism Society of Connecticut
207 Metacomet Drive
Meriden, CT 06450
Phone: 203-235-7659
Fax: none
Email: asconn@sbcglobal.net
www.autismsocietyofct.org

National Alliance for Autism Research

Research Park
99 Wall Street
Princeton, NJ 08540
Phone: 888-777-6227/609-430-9160
Fax: 609-430-9163
Email: naar@naar.org
www.naar.org
No Connecticut chapter.

NAAR – New England Office

124 Watertown Street
Suite 3B, Box 6
Watertown, MA 02472
Phone: 617-924-3300
Fax: 617-924-3311
Email: newengland@naar.org

Additional web resources:

Exploring Autism: A Look at the Genetics of Autism

www.exploringautism.org
Website dedicated to helping families who are living with challenges of autism. A collaborative effort of Autism Genetics Cooperative, a group of researchers and clinicians working to find the genetic causes of autism.

Additional literature:

Children with Autism: A Parent's Guide

by Powers, M.D. and Grandin, T.
Woodbine House, 2nd Edition, 2000
Includes chapters on diagnosis and treatment, adjusting to life with an autistic child, finding good education programs, legal rights of the autistic child, becoming an advocate for a child, special problems and needs of the adult with autism. Provides list of resources for autistic individuals in the United States.

Facing Autism: Giving Parents Reasons for Hope and Guidance for Help

By Hamilton, L.M.
Waterbrook Press, 2000
A treasury of detailed, helpful information from a mother who carefull investigated all promising treatment approaches.

Family Pictures: A Novel

by Miller, S.
Harper, 1999
A fictional story told through the eyes of four family members, the complexity of family relationships is illustrated through their reactions to a family member with autism.

Nobody Nowhere: The Extraordinary Autobiography of an Autistic

by Williams, D.
Perennial Currents, 1994
The author details what it is like to grow up autistic and the price one pays for being "high-functioning".

Somebody Somewhere:
Breaking Free from the World of Autism
by Williams, D.
Three Rivers Press, 1995
Sequel to **Nobody Nowhere,** the author describes her life as a published author and as a graduate student.

~ BATTEN DISEASE ~

Batten Disease Support and Research Association

120 Humphries Drive
Suite 2
Reynoldsburg, OH 43068
Phone: 800-448-4570/740-927-4298
Fax: 614-445-4191
Email: bdsra1@bdsra.com
www.bdsra.org
No Connecticut chapter.

New England chapter:

Diane Burl
22 Carver Street
Granby, MA 01033
Phone: 413-467-2294
Fax: none
Email: none
Web: none

Additional web resources:

The Natalie Fund

www.nataliefund.org
Striving to find a treatment and cure for Batten's Disease. Goals include raising funds for research, enhancing community awareness, and providing information and support for families with affected children.

~ BECKWITH-WIEDEMANN SYNDROME ~

Beckwith-Wiedemann Support Network

2711 Colony Road
Ann Arbor, MI 48104
Phone: 800-837-2976/734-973-9721
Fax: 734-973-9721
Email: bwsn@beckwith-wiedemann.org
www.beckwith-wiedemann.org

No Connecticut chapter.

~ CANAVAN DISEASE ~

Canavan Foundation

450 West End Avenue #10C
New York, NY 10024
Phone: 877-4-CANAVAN/212-873-4640
Fax: 212-873-7892
Email: info@canavanfoundation.org
www.canavanfoundation.org
No Connecticut chapter.

National Tay-Sachs and Allied Diseases Association

2001 Beacon Street
Suite 204
Brighton, MA 02135
Phone: 800-906-8723
Fax: 617-277-0134
Email: info@ntsad.org
www.ntsad.org

In Connecticut:
(Serving the tri-state area)
1202 Lexington Avenue #288
New York, NY 10028
Phone: 888-354-7788/212-431-0431
Fax: 888-354-4884
Email: info@ntsad-ny.org
www.ntsad-ny.org

Additional web resources:

Center for Jewish Diseases
Mt. Sinai School of Medicine
www.mssm.edu/jewish_genetics/diseases/canavan.shtml
Includes disease description, natural history, genetics and testing information.

Medical College of
Wisconsin HealthLink:
The Facts about Canavan Disease
www.healthlink.mcw.edu/article/921391101.htm
Provides disease description, including screening and diagnosis, signs and symptoms, and research, and links to other informative sites.

~ CANCER ~

American Cancer Society
1599 Clifton Road, NE
Atlanta, GA 30329
Phone: 800-227-2345/404-320-3333
Fax: none
Email: through website only
www.cancer.org

In Connecticut:
372 Danbury Road
Wilton, CT 06897
Phone: 203-563-0740
Fax: 203-563-0738

238 West Town Street
Norwich, CT 06360
Phone: 860-887-2547
Fax: 860-885-0820

538 Preston Avenue
Meriden, CT 06450
Phone: 203-379-4700
Fax: 203-379-5060

Candlelighters Childhood Cancer Foundation
3910 Warner Street
P.O. Box 498
Kensington, MD 20895
Phone: 800-366-2223/301-962-3520
Fax: 301-962-3521
Email: info@candlelighters.org
www.candlelighters.org
No Connecticut chapter.

Additional web resources:

National Cancer Institute's CancerNet
www.cancer.gov/cancerinfo/prevention-genetics-causes
Authoritative information about cancer genetics.

Harvard Center for Cancer Prevention
www.yourcancerrisk.harvard.edu
Personalized estimation of cancer risk and tips for prevention.

Guide to Internet Resources for Cancer
www.cancerindex.org
Nonprofit guide contains over 100 pages and over 4000 links to cancer related information. Site is regularly updated.

Additional literature:

A Practical Guide to
Human Cancer Genetics
by Hodgson, S.V. and Maher, E.R.
Cambridge University Press, 2nd Edition, 1999
Essential overview of the latest developments in human cancer genetics.

Counseling About Cancer:
Strategies for Genetic Counseling
by Schneider, K.
Wiley-Liss, 2nd Edition, 2001
Comprehensive resource including medical and psychological issues, cancer risk counseling, predisposition testing, essential references.

Healing Lessons

by Winawer, S.J. and Taylor, N.
Rutledge, 1999
Story of Dr. Sidney Winawer, chief of gastroenterology at Memorial Sloan-Kettering Cancer Center, and his wife, Andrea, dealing with her diagnosis of metastatic stomach cancer.

It's Not About the Bike; My Journey Back to Life

by Armstrong, L and Jenkins, S.
Berkley Publishing Group, 2001
The story of Lance Armstrong - a world-class athlete nearly struck down by cancer, only to recover and win the grueling and intense Tour de France.

Staying Alive – A Family Memoir

by Reibstein, J.
Bloomsbury Publishing PLC, 2002
Follows a successful, ethnically mixed family typical in America today over fifty years, atypical only in the diagnosis of breast cancer that hangs like a dark cloud over all the women in the family.

The Truth About Breast Cancer Risk Assessment

by Hollingsworth, A.B.
National Writers Press, 2000
Learn how risk factors are assembled into a personal profile.

~ CARBOHYDRATE-DEFICIENT GLYCOPROTEIN SYNDROME ~

CDG Family Network Foundation

P.O. Box 149
Anamorse, ND 58710
Phone: 800-250-5273
Fax: 972-633-8088
Email: cdgaware@aol.com

~ CEREBRAL PALSY ~

United Cerebral Palsy Association

1660 L Street NW
Suite 700
Washington, DC 20036
Phone: 800-872-5827/202-776-0406
Fax: 202-776-0414
Email: national@ucp.org
www.ucp.org

UCP of Eastern Connecticut

Shaw's Cove 6, Suite 101
New London, CT 06320
Phone: 860-447-3889
Fax: 860-447-3789
Email: united.cerebra.palsy@snet.net
www.neighborhoodlink.com/org/ucpect

UCP of Greater Hartford

80 Whitney Street
Hartford, CT 06105
Phone: 860-236-6201
Fax: 860-236-6205
Email: srne@mindspring.com

UCP of Southern Connecticut

94-96 South Turnpike Road
Wallingford, CT 06492
Phone: 203-269-3511
Fax: 203-269-7411
Email: ucpasouthernct@yahoo.com

Additional web resources:

National Disability Sports Alliance

www.ndsaonline.org
Servicing all individuals with physical disabilities in the areas of sports, fitness, and recreation. Also known as the United States Cerebral Palsy Athletic Association.

The Cerebral Palsy Network

www.geocities.com/Heartland/Plains/8950
A resource for information, sharing, and support.

ADDITIONAL LITERATURE:

CEREBRAL PALSY:
A COMPLETE GUIDE FOR CAREGIVING
by Miller, F. and Bachrach, S.J.
Johns Hopkins University Press, 1998
Overview of CP, explains medical procedures, medical and psychological implications, discusses advocacy, list of resources and recommended reading.

MY PERFECT SON HAS CEREBRAL PALSY:
A MOTHER'S GUIDE OF HELPFUL HINTS
by Kennedy, M.
AuthorHouse, 2001
Practical advice based on personal account.

~ CHARCOT-MARIE-TOOTH ~

CHARCOT-MARIE-TOOTH ASSOCIATION
2700 Chestnut Street
Chester, PA 19013
Phone: 800-606-2682
Fax: 610-499-9567
Email: CMTAssoc@aol.com
www.charcot-marie-tooth.org

In Connecticut
(serving Fairfield, CT and
Westchester County, NY):
Contact Beverly Wurzel
Phone: 845-783-2815
Fax: none
Email: cranomat@frontiernet.net

ADDITIONAL WEB RESOURCES:

CMTNET
www.users.rcn.com/smith.ma.ultrranet/CMTneto.html
Repository of information for research and treatment of CMT intended to provide information for the medical and non-medical community.

~ CHROMOSOME ABNORMALITIES ~

(See also Down syndrome, Klinefelter syndrome, Turner syndrome, Fragile X syndrome)

CHROMOSOME DELETION OUTREACH
P.O. Box 724
Boca Raton, FL 33429
Phone: 888-CDO-6880/561-395-4252
Fax: 561-395-4252 call first
Email: cdo@worldnet.att.net
www.chromodisorder.com
No Connecticut chapter.

4P- SUPPORT GROUP
P.O. Box 1676
Gresham, OR 97030
Phone: 503-661-7546
Fax: none
Email: fourthchromosome@aol.com
www.4p-supportgroup.org
No Connecticut chapter.

5P- SOCIETY
P.O. Box 268
Lakewood, CA 90714
Phone: 888-970-0777/562-804-4506
Fax: 562-920-5240
Email: director@fivepminus.org
www.fivepminus.org or www.criduchat.org
No Connecticut chapter.

Northeastern Regional
Family Support Coordinator:
Contact Ruthie Liberman
Lexington, MA
Phone: 781-861-3962

8P DUPLICATION SUPPORT GROUP
The Genetics Center
1 Children's Plaza
Dayton, OH 45404
Phone: 937-641-3800/937-641-5645
Fax: 937-463-5325
Email: callif-daleyF@childrensdayton.org
www.cmc-dayton.org
No Connecticut chapter.

Chromosome 9p- Network
393 North Grass Valley Road
Pine Valley, UT 84781
Phone: 435-574-1121
Fax: 435-574-2000
Email: Beverly.udell@9pminus.org
www.9pminus.org
No Connecticut chapter.

11q Research and Resource Group
54 Elmridge Road
Pawtucket, CT 06379
Phone: 860-599-4015
Fax: 860-441-6159
Email: david_m_george@groton.pfizer.com
www.web.ukonline.co.uk/c.jones/11q/contents.htm

Trisomy 12p Parent Support Organization
175 Lawndale Road
Mansfield, MA 02048
Phone: 508-339-1680
Fax: 508-339-0504
Email: maguirecb@comcast.net
No Connecticut chapter.

IDEAS (Isodicentric 15 Exchange, Advocacy, and Support)
c/o Paul Rivard
P.O. Box 4616
Manchester, NH 03108
Phone:, not available
Fax: not available
Email: info@idic15.org
www.idic15.org
No Connecticut chapter.

Disorders of Chromosome 16 Foundation
331 Haddon Circle
Vernon Hills, IL 60061
Phone: 847-816-0627 check number
Fax: 847-367-4031
Email: kblange1@aol.com
www.trisomy16.org/foundation.html
No Connecticut chapter.

Support Organization for Trisomy 18, 13 and Related Disorders (SOFT)
2982 South Union Street
Rochester, NY 14624
Phone: 800-716-7638/716-594-4621
Fax: 716-594-4621
Email: barbsoft@rochester.rr.com
www.trisomy.org

Connecticut chapter:
1473 Forbes Street
East Hartford, CT 06118
Phone: 860-568-0171
Fax: none
Email: norbertonhanel@aol.com

Trisomy 18 Support Foundation, Inc.
4301 Connecticut Avenue, NW
Suite 404
Washington, DC 20008
Phone: 703-878-2369
Fax: 703-878-2369
Email: info@trisomy18support.org
www.trisomy18support.org
No Connecticut chapter.

Chromosome 18 Registry and Research Society
6302 Fox Head
San Antonio, TX 78247
Phone: 210-657-4968
Fax: 210-657-4968
Email: cody@chromosome18.org
www.chromosome18.org

New England Regional Coordinator:
Deb Ammann
Phone: 631-223-3039
Fax: none
Email: debpaul94@optonline.net

22q and You Center
The Department of Clinical Genetics
The Children's Hospital of Philadelphia
34th Street and Civic Center Boulevard
Philadelphia, PA 19104
Phone: 215-590-2920
Fax: 215-590-3298
Email: lunny@email.chop.edu
www.chop.edu/consumer/jsp/division/generic.jsp?id=74631
No Connecticut chapter.

Ring Chromosome 22 Email Discussion List
14 Westwood Acres
Morris, MN 56267
Phone: 320-589-1050
Fax: none
Email: r22@maelstrom.stjohns.edu
maelstrom.stjohns.edu/archives/r22.htm
No Connecticut chapter.

Additional literature:

Chromosome Abnormalities and Genetic Counseling
by Gardner, RT.J.M. and Sutherland, G.R.
Oxford University Press, 2nd Edition, 2003
Second edition of a textbook/reference for genetic counselors and cytogenetic laboratory workers. Presents the theoretical basis of clinical cytogenetics.

~Cleft Lip/Palate and Craniofacial Disorders ~

AboutFace USA
P.O. Box 969
Batavia, IL 60510
Phone: 888-486-1209
Fax: 630-761-2985
Email: info@aboutfaceusa.org
www.aboutfaceusa.org
Call main office for Connecticut contacts.

American Cleft Palate – Craniofacial Association
104 South Estes Drive
Suite 204
Chapel Hill, NC 27514
Phone: 919-933-9044
Fax: 919-933-9604
Email: info@acpa-cpf.org
www.cleftpalate-craniofacial.org
No Connecticut chapter.

Children's Craniofacial Association
13140 Coit Road
Suite 307
Dallas, TX 75240
Phone: 800-535-3643
Fax: 214-570-8811
Email: csmith@ccakids.com
www.ccakids.com
No Connecticut chapter.

Cleft Palate Foundation
1504 East Franklin Street
Suite 102
Chapel Hill, NC 27514
Phone: 800-242-5338
Fax: 919-933-9044
Email: info@cleftline.org
www.cleftline.org

F.A.C.E. (Families Advancing Craniofacial Excellence)
P.O. Box 185
Unionville, CT 06085
Phone: 860-673-1829
Fax: 860-673-1829
Email: smilesforchildren@sbcglobal.net
www.smilesforchildren.org

FACES: The National Craniofacial Association
P.O. Box 11082
Chattanooga, TN 37401
Phone: 800-332-2373
Fax: 423-267-3124
Email: faces@faces-cranio.org
www.faces-cranio.org
No Connecticut chapter.

Let's Face It
P.O. Box 29972
Bellingham, WA 98228
Phone: 360-676-7325
Fax: contact office for faxing information
Email: letsfaceit@faceit.org
www.faceit.org
No Connecticut chapter.

ADDITIONAL WEB RESOURCES:

SMILES
www.cleft.org
A group of dedicated families who have developed a first-hand understanding of the needs of children with cleft lip, cleft palate and craniofacial abnormalities.

WIDE SMILES
www.widesmiles.org
Offers support, inspiration, information and networking for families who may be dealing with the challenges associated with clefting.

ADDITIONAL LITERATURE:

A PARENT'S GUIDE TO CLEFT LIP AND PALATE
by Moller, K.T., Starr, C.D. and Johnson, S.A.
University of Minnesota Press, 1990
One parent has said that this book should be required reading for every new parent of a child with a cleft lip or palate.

BABYFACE: A STORY OF HEART AND BONES
by McDermott, J.
Penquin Books, 2002
Story of a child with Apert syndrome.

CLEFT LIP AND PALATE: FROM ORIGIN TO TREATMENT
by Wyszynski, D.F.
Oxford University Press, 2002
Major advances in the diagnosis and treatment of oral clefts have been made in the past 50 years, and recent genetics and epidemiological studies have led to new theories about the causes of cleft lip and palate. Addressing issues that are relevant to clinicians, researchers and family members, this book is a comprehensive well-illustrated and up-to-date account of the many facets of this common disorder.

~ COFFIN-LOWRY SYNDROME ~

COFFIN-LOWRY SYNDROME FOUNDATION
3045 255th Avenue SE
Sammamish, WA 98075
Phone: 425-427-0939
Fax: none
Email: cclsfoundation@yahoo.com
www.clsf.info
No Connecticut chapter.

~ CONGENITAL ADRENAL HYPERPLASIA ~

CARES FOUNDATION, INC. (CONGENITAL ADRENAL HYPERPLASIA, RESEARCH, EDUCATION AND SUPPORT)
189 Main Street
2nd floor
Millburn, MJ 07041
Phone: 866-227-3737/973-912-3895
Fax: 973-912-3894
Email: Kelly@caresfoundation.org
www.caresfoundation.org

Connecticut contact:
Lynn Torony
Phone: 203-264-6898
Fax: 203-264-0529
Email: ltorony@earthlink.net

ADDITIONAL WEB RESOURCES:

CAH EDUCATION AND SUPPORT NETWORK
www.congenitaladrenalhyperplasia.org
Includes "What Is CAH?", message boards, medical sites, glossary, and Frequently Asked Questions.

~ CONGENITAL HYPOTHYROIDISM ~

AMERICAN FOUNDATION OF THYROID PATIENTS
4322 Douglas Avenue
Midland, TX 79703
Phone: not available
Fax: not available
Email: thyroid@flash.net
www.thyroidfoundation.org
No Connecticut chapter.

Thyroid Foundation of America, Inc.
1 Longfellow Place
Suite 1518
Boston, MA 02114
Phone: 800-832-8321
Fax: 617-534-1515
Email: info@allthyroid.org
www.allthyroid.org
No Connecticut chapter.

Additional web resources:

Educating Parents of Extra-Special Children
www.epeconline.com/Congenital Hypothyroidism.html
A resource of information for adults with special needs and parents of special needs children. Overview of congenital hypothyroidism with links to American Academy of Pediatrics.

~ CORNELIA DE LANGE SYNDROME ~

Cornelia de Lange Syndrome Foundation
302 West Main Street
Suite 100
Avon, CT 06001
Phone: 800-753-2357/800-223-8355
Fax: 860-676-8337
Email: info@cdlsusa.org
www.cdlsusa.org

Additional web resources:

CDLS Online Support Group
www.cdls-support.org
Free online group focuses on parents, caregivers and families, friends, teachers and other professionals. Includes email group for siblings, and Spanish speaking members.

~ CROHN'S AND COLITIS ~

Crohn's and Colitis Foundation of America
386 Park Avenue South
17th floor
New York, NY 10016
Phone: 800-932-2423/212-685-3440
Fax: 212-779-4098
Email: info@ccfa.org
www.ccfa.org

In Connecticut:
Central Connecticut Chapter:
P.O. Box 185431
Hamden, CT 06518
Phone: 203-248-4625
Fax: 203-248-7832
Email: ctccff@aol.com

Fairfield/Westchester Chapter Address:
200 Bloomingdale Road
White Plains, NY 10605
Phone: 914-328-2874
Fax: 914-328-2946
Email: Westfield@ccfa.org

Northern Connecticut Affiliate
P.O. Box 370614
West Hartford, CT 06137

Additional web resources:

Crohn's Disease Resource Center
www.healingwell.com/ibd
Links to books on Amazon.com as well as resource directory, message boards, chat rooms, products and services, and other organizations.

Additional literature:

The First Year – Crohn's Disease and Ulcerative Colitis: An Essential Guide for the Newly Diagnosed
by Sklar, J. and Sklar, M.
Marlowe & Company, 2002
Covers strategies for necessary lifestyle changes, guidelines and tips for modifying diet, choosing a medical team, discussing the condition with family, current medical research, support, and more.

The Crohn's Disease and Ulcerative Colitis Fact Book
by the Crohn's and Colitis Foundation
Banks, P.A., Present, D.H. and Steiner, P.
John Wiley & Sons, 1983
Older text but good general introduction to disease, vast amount of background information and general overview on the nature of the disease, treating the disease, and living with the disease.

~ CYSTIC FIBROSIS ~

The Cystic Fibrosis Foundation
6931 Arlington Road
Suite 200
Bethesda, MD 20814
Phone: 800-344-4823/301-951-4422
Fax: 301-951-6378
Email: info@cff.org
www.cff.org

In Connecticut:
185 Silas Deane Highway
Wethersfield, CT 06109
Phone: 800-841-2828/860-257-6907
Fax: 860-257-6903
Email: conn@cff.org
www.cff.org/connecticut.htm

Additional web resources:

Cystic Fibrosis Mutational Database
www.genet.sickkids.on.ca/cftr
Collection of mutations in CFTR gene to provide CF researchers and other related professionals with up-to-date information about individual mutations.

Cystic Fibrosis.com
www.cysticfibrosis.com/info/info.htm
CF information, frequently asked questions, "Just for Kids" section, search for CF associations, clinical trials, and other links.

Cystic Fibrosis Foundation Genotyping Center
www.hopkinsmedicine.org/cfgenotyping
Their mission is to detect CFTR mutations in patients with unusual forms of CF, coordinate collection of genotype/phenotype information, to increase and facilitate communication between CF researchers and clinicians working with patients with non-classical CF.

The CF Pharmacy
www.cfpharmacy.com
Dedicated to research and development of new cost-effective pharmacological approaches to care and treatment of CF.

Additional literature:

Alex: The Life of a Child
by Deford, F.
Rutledge Hill Press, 1997
Sportswriter Deford's story of his courageous daughter who died at age 8 due to complications of cystic fibrosis.

Alive at 25: How I'm Beating Cystic Fibrosis
by Lipman, A.
Longstreet Press, 2002
Candid, honest, inspiring personal account.

Cystic Fibrosis:
A Guide for Patient and Family
by Orenstein, D.M.
Lippincott, Williams and Wilkins,
2nd Edition, 1997
Clear advice on day-to-day management, school, travel, exercise, nutrition, medications, psychological effects, treatment, complications, long-term issues, and prospects for a cure.

~ CYSTINOSIS ~

Cystinosis Foundation, Inc.
604 Vernon Street
Oakland, CA 94610
Phone: 800-392-8458
Fax: 559-222-7997
Email: check website for individual emails
www.cystinosisfoundation.org
No Connecticut chapter.

Cystinosis Research Network
10 Pine Avenue
Burlington, MA 01803
Phone: 866-276-3669/781-229-6182
Fax: 781-229-6030
Email: CRN@cystinosis.org
www.cystinosis.org
No Connecticut chapter.

Additional web resources:

Cystinosis Central
www.medicine.ucsd.edu/cystinosis/INDEX.htm
Up-to-date site including general information, links to other resources, physician information, and research.

~ DEAFNESS/HEARING IMPAIRMENT ~

Alexander Graham Bell Association for the Deaf and Hard of Hearing, Inc.
3417 Volta Place, NW
Washington, DC 20007
Phone: 800-432-7543/202-337-5220
Fax: 202-337-8314
Email: agbell2@aol.com
www.agbell.org

In Connecticut:
P.O. Box 565
Bristol, CT 06011
Phone: not available
Fax: not available
Email: biernat@comcast.net
www.agbellct.org

American Hearing Research Foundation
8 South Michigan Avenue
Suite 814
Chicago, IL 60603
Phone: 312-726-9670
Fax: 312-726-9695
Email: blederer@american-hearing.org
www.american-hearing.org
No Connecticut chapter.

National Association of the Deaf
814 Thayer Avenue
Suite 250
Silver Spring, MD 20910
Phone: 301-587-1788
Fax: 301-587-1791
Email: NADinfo@nad.org
www.nad.org

In Connecticut:
Connecticut Association of the Deaf
Contact Mary Beers
Email: catbeers@aol.com

Additional web resources:

Finding Genes for Non-syndromic Deafness
www.people.vcu.edu/~nance/index
Provided by Gallaudet University and the Medical college of Virginia, includes information on causes of deafness, patterns of inheritance, syndromic deafness gene mapping and molecular characterization of genetic deafness, treatment, and research.

ADDITIONAL LITERATURE:

GENETICS AND AUDITORY DISORDERS
by Keats, B.J.B., Popper, A.N. and Fay, R.R.
SPRINGER-VERLAG, 2001
Informs reader on many clinical forms of genetic hearing loss, the mutations responsible and the function of proteins encoded by mutant genes.

HEREDITARY HEARING LOSS AND ITS SYNDROMES
by Gorlin, R.J., Toriello, H.V. and Reardon, W.
Oxford University Press, 2nd Edition, 2004
Comprehensive and organized resource.

I'M DEAF AND IT'S OKAY
by Aseltine, L., Mueller, E. and Tait, M.
Albert Whitman & Company, 1986
For children ages 4-8

MY SENSE OF SILENCE: MEMOIRS OF A CHILDHOOD WITH DEAFNESS
by Davis, L.J.
University of Illinois Press, 2000
Growing up as a hearing child of deaf parents, a candid, affecting, and often funny memoir.

~ DIABETES ~

AMERICAN DIABETES ASSOCIATION
1701 North Beauregard Street
Alexandria, VA 22311
Phone: 800-DIABETES,
Fax: call for faxing information
Email: ASKADA@diabetes.org
www.diabetes.org

In Connecticut:
306 Industrial Park Road
Middletown, CT 06457
Phone: 888-DIABETES/203-639-0385
Fax: 860-632-5098
Email: not available

JUVENILE DIABETES RESEARCH FOUNDATION
120 Wall Street
19th floor
New York, NY 10005
Phone: 800-533-2873
Email: info@jdrf.org
www.jdrf.org

In Connecticut:
NORTH CENTRAL CONNECTICUT AND WESTERN MASSACHUSETTS CHAPTER
18 North Main Street
3rd floor
West Hartford, CT 06107
Phone: 860-561-1153
Fax: 860-561-3440
Email: northcentralct@jdrf.org
www.jdrf.org/CT/North-Central-CT-Western-MA

FAIRFIELD COUNTY CHAPTER
200 Connecticut Avenue
Suite 5H
Norwalk, CT 06854
Phone: 203-854-0658
Fax: 203-854-0798
Email: fairfield@jdrf.org
www.jdrf.org/chapters/ct/fairfield-county

GREATER NEW HAVEN CHAPTER
2969 Whitney Avenue
Hamden, CT 06518
Phone: 203-248-1880
Fax: 203-248-1820
Email: newhaven@jdrf.org
www.jdrf.org/chapters/ct/Greater-New-Haven

ADDITIONAL WEB RESOURCES:

CHILDREN WITH DIABETES
www.childrenwithdiabetes.com
Online community for kids, families and adults with diabetes.

JOSLIN DIABETES CENTER
www.joslin.org
Harvard Medical School affiliate, an internationally recognized treatment, research, and education institution headquartered in Boston, Massachusetts.

Did You Know...?

Humans are 99.9% identical – only 0.1% of our genetic makeup differs.

American Association of Diabetes Educators
www.aadenet.org
A multidisciplinary professional membership organization dedicated to advancing the practice of diabetes self-management, training, and care.

Additional literature:

American Diabetes Association Complete Guide to Diabetes
by the American Diabetes Association,
McGraw-Hill/Contemporary Distributed Products, 3rd Edition, 2002
One volume comprehensive home reference on the best self-care techniques, latest medical breakthroughs, and all the information needed to live an active, healthy life with diabetes.

Diabetes for Dummies
by Rubin, A.L.
John Wiley & Sons, 1999
Addresses Type 1 and Type 2 diabetes, discusses causes, symptoms, treatment, and the importance of diet and exercise, finding the right practitioner, building a support team, and how to find help online.

Living with Juvenile diabetes: A Practical Guide for Parents and Caregivers
by Puerrung, V.
Hatherleigh Press, 2001
Written by a mother of two children with diabetes, provides the latest facts and treatments, tips on exercise and nutrition, recipes, supplies, research trends, and more.

~ DOWN SYNDROME ~

National Down Syndrome Congress
1370 Center Drive
Suite 102
Atlanta, GA 30338
Phone: 800-232-6372/770-604-9500
Fax: 212-979-2873/770-604-9898
Email: info@ndsccenter.org
www.ndsccenter.org

In Connecticut:
Connecticut Down Syndrome Congress
P.O. Box 340385
Hartford, CT 06134
Phone: 888-486-8537, 860-257-8882
Fax: call for faxing information
Email: admin@ctdownsyndrome.org
www.ctdownsyndrome.org

National Down Syndrome Society
666 Broadway
Suite 810
New York, NY 10012
Phone: 800-221-4602
Fax: 212-979-2873
Email: info@ndss.org
www.ndss.org
No Connecticut chapter.

Association for Children with Down Syndrome
4 Fern Place
Plainview, NY 11803
Phone: 516-933-4700
Fax: 516-933-9524
Email: information@acds.org
www.acds.org
No Connecticut chapter.

Additional web resources:

Growth Charts for Children with Down Syndrome
www.growthcharts.com

Down Syndrome: Health Issues
www.ds-health.com
News and information for parents and professionals, provided by a pediatrician who is the father of a child with Down syndrome.

Recommended Down Syndrome Sites on the Internet
www.ds-health.com/ds_sites.htm
Probably the most useful site on Down syndrome.

Additional literature:

Babies with Down Syndrome: a New Parent's Guide
by Stray-Gundersen, K.
Woodbine House, 2nd Edition, 1995
A book designed to answer most questions new parents have.

A Parent's Guide to Down Syndrome: Toward a Brighter Future
by Pueschel, S.M.
Paul H. Brooks Publishing Co., 2nd Edition, 2000
Reference book written by a physician parent of a child with Down syndrome.

Keys to Parenting a Child with Down Syndrome
by Brill, M.T.
Barron's Educational Series, 1993

Teaching the Infant with Down Syndrome: A Guide for Parents and Professionals
by Hanson, M.J.
Pro-Ed, 2nd Edition, 1986

Expecting Adam: A True Story of Birth, Rebirth, and Everyday Magic
by Beck, M.
Berkley Publishing Group, 2000
Autobiographical tale of academically oriented Harvard couple who discover prenatally that their baby has Down syndrome.

Choosing Naia: A Family's Journey
by Zuckoff, M.
Beacon Press, 2003
Follows the story of a couple struggling with grief and confusion and the decisions they face when they are confronted by abnormal prenatal diagnosis test results.

Understanding Down Syndrome: An Introduction for Parents
by Cunningham, C.
Brookline Books, 1996
An excellent overview for new parents and professionals.

~ DYSTONIA ~

Dystonia Medical Research Foundation
1 East Wacker Drive
Suite 2430
Chicago, IL 60601
Phone: 800-377-3978/312-755-0198
Fax: 312-803-0138
Email: dystonia@dystonia-foundation.org
www.dystonia-foundation.org

In Connecticut:

Central Connecticut Support Group
Contact Larry Stahl
11 Hammick Road
West Hartford, CT 06107
Phone: 860-565-2564
Fax: 860-565-1629
Email: artscifi2@aol.com

Stratford Support Group
Contact Barbara Benowitz
Phone: 203-386-1982

Additional web resources:

International Dystonia On-line Support Group
www.dystonia-support.org
Created by affected individuals, includes links to information about their group, medical descriptions, personal accounts, chat rooms, parents information, and children's email club.

Additional literature:

Holding the Hope: a Parent's Guide to Living with Dystonia
by Ross, K.K.
The Dystonia Foundation, 1996
A guide book for families coping with dystonia. Addresses the impact a child's chronic condition may have on the entire family.

The Official Patient's Sourcebook on Dystonia disorders: A Revised and Updated Directory for the Internet Age
by Icon Health Publications
Icon Health, 2002
Created for patients doing their own education and research. A reference book organized into 3 main parts, basic research techniques to find general information such as guidelines for diagnosis, treatment and prognosis, how to research on specific topics in dystonia, and guide to the latest scientific research.

~ EHLERS-DANLOS SYNDROME ~

Ehlers-Danlos National Foundation
6399 Wilshire Boulevard
Suite 200
Los Angeles, CA 90048
Phone: 800-956-2902/323-651-3038
Fax: 323-651-1366
Email: staff@ednf.org
www.ednf.org
No Connecticut chapter.

EDS Today
P.O. Box 88814
Seattle, WA 98138
Phone: 253-835-1735
Fax: 253-835-1735
Email: info@edstoday.org
www.edstoday.org
No Connecticut chapter.

Additional web resources:

Ehlers-Danlos Syndrome
www.orthop.washington.edu/arthritis/types/ehlersdanlos/01
Website of the University of Washington Orthhopedics and Sportsmedicine Group. Article reviews incidence, risk factors, causes, symptoms, diagnosis, management, treatment, and coping with Ehlers-Danlos syndrome.

~ EPILEPSY ~

Epilepsy Foundation of America
4351 Garden City Drive
Landover, MD 20785
Phone: 800-332-1000/301-459-3700
Fax: 301-577-2684
Email: postmaster@efa.org
Wwww.epilepsyfoundation.org

In Connecticut:
Epilepsy Foundation CT, Inc.
386 Main Street
Middletown, CT 06457
Phone: 800-899-3745/860-346-1924
Fax: 860-346-1928
Email: efct@aol.com
www.epilepsyfoundation.org/connecticut

Additional web resources:

American Epilepsy Society
www.aesnet.org
Neurological professional organization seeking to promote interdisciplinary communication, scientific investigation and exchange of clinical information.

Epilepsy.com
www.epilepsy.com
Information on diagnosis, treatment, living with epilepsy, news and other resources.

Additional literature:

Growing Up with Epilepsy: A Practical Guide For Parents
by Blackburn, L.B.
Demos Medical Publishing, 2003
Provides advice on discipline, social development, education, medications and side effects, psychological concerns, and other issues.

Handbook of Epilepsy
by Brown, T.R. and Holmes, G.L.
Lippincott, Williams and Wilkins,
3rd Edition, 2003
Pocket-sized reference book contains concise up-to-date, clinically oriented information on diagnosis and treatment. Includes guidelines and classification of epilepsy syndromes from the American Academy of Neurology and the American Epilepsy Society.

~ FAMILIAL DYSAUTONOMIA ~

Dysautonomia Foundation, Inc.
633 3rd Avenue
12th floor
New York, NY 10017
Phone: 212-949-6644
Fax: 212-682-7625
Email: fdinfo@videobureau.com
www.familialdysautonomia.org
No Connecticut chapter.

National Dysautonomia Research Foundation
1407 West 4th Street
Suite 160
Red Wing, MN 55066
Phone: 651-267-0525
Fax: 651-267-0524
Email: ndrf@ndrf.org
www.ndrf.org
No Connecticut chapter.

Additional web resources:

Center for Jewish Diseases
Mt. Sinai School of Medicine
www.mssm.edu/jewish_genetics
Follow links to familial dysautonomia. Includes disease description, natural history, genetics and testing information.

~ FANCONI ANEMIA ~

Fanconi Anemia Research Fund, Inc.
1801 Willamette Street
Suite 200
Eugene, OR 97401
Phone: 541-687-4658
Fax: 541-687-0548
Email: info@fanconi.org
No Connecticut chapter.

Additional web resources:

Fanconi Anemia Mutation Database
www.rockefeller.edu/fanconi/mutate/
Established as a cooperative effort to accelerate the availability of information. Divided into a public section listing mutations that have already been reported in the literature, and a private section with unpublished data.

FA: Hemotology 101
www.dceg.cancer.gov/clinicalhematology101.html
Powerpoint slide show on Fanconi Anemia by Dr. Blanche Alter of the National Cancer Institute.

Additional literature:

Molecular Mechanisms of Fanconi Anemia
by Ahmad, S.
Landes Bioscience, 2004
Medical text book.

~ FATTY OXIDATION DISORDER ~

FOD Family Support Group
805 Montrose Drive
Greensboro, NC 27410
Phone: 336-547-8682
Fax: call for faxing information
Email: deb@fodsupport.org
www.foodsupport.org
No Connecticut chapter.

~ FIBRODYSPLASIA OSSIFICANS PROGRESSIVA ~

International FOP Association
P.O. Box 196217
Winter Springs, FL 32719
Phone: 407-365-4194
Fax: 407-365-3213
Email: together@ifopa.org
www.ifopa.org
No Connecticut chapter.

~ FRAGILE X ~

National Fragile X Foundation
P.O. Box 190488
San Francisco, CA 94119
Phone: 800-688-8765/925-938-9300
Fax: 925-938-9315
Email: natlfx@fragileX.org
www.fragileX.org

In Connecticut:
Fragile X Society of Connecticut
20 Mohegan Drive
West Hartford, CT 06117
Phone: 860-233-1904
Fax: not available
Email: xfamily@aol.com

FRAXA Research Foundation, Inc.
45 Pleasant Street
Newburyport, MA 01950
Phone: 978-462-1866
Fax: 978-463-9985
Email: info@fraxa.org
www.fraxa.org
No Connecticut chapter.

Additional web resources:

Genetic Counseling for Fragile X Syndrome: Recommendations of the National Society of Genetic Counselors
www.guideline.gov/summary/summary.aspx?doc_id=2546&nbr=1772

Additional literature:

Children with Fragile X Syndrome: A Parents' Guide
by Weber, J.D.
Woodbine House, 2000
A comprehensive book for parents. Topics include diagnosis, parents' emotions, daily care, family life, education, and how to seek further help and expertise.

Fragile X Syndrome: Diagnosis, Treatment, and Research
by Hagerman, R.J. and Hagerman, P.J.
Johns Hopkins University Press,
3rd edition, 2002
Textbook discusses clinical approaches to diagnosis, epidemiology, molecular genetics, neuropsychology, treatment, genetic counseling, pharmacotherapy, intervention and gene therapy.

~ GALACTOSEMIA ~

Parents of Galactosemic Children, Inc.
885 Del Sol Street
Sparks, NV 89436
Phone: 775-626-0885
Fax: not available
Email: mesameadow@aol.com
www.galactosemia.org
No Connecticut chapter.

Additional web resources:

Galactosemia Resources and Information
www.galactosemia.com
Includes general information on galactosemia, galactose content in foods, contact information for other families.

~ GAUCHER DISEASE ~

National Gaucher Foundation, Inc.
5410 Edson Lane
Suite 260
Rockville, MD 20852
Phone: 800-925-8885/301-816-1515
Fax: 301-816-1516
Email: ngf@gaucherdisease.org
www.gaucherdisease.org
No Connecticut chapter.

Children's Gaucher Research Fund
P.O. Box 2123
Granite Bay, CA 95746
Phone: 916-797-3700
Fax: 916-797-3707
Email: research@childrensgaucher.org
www.childrensgaucher.org
No Connecticut chapter.

Other web resources:

Gaucher Disease Summary
www.gaucher.mgh.harvard.edu/summary.html
A guide for patients, parents, relatives, and friends provided by the Neurology Service at Massachusetts General Hospital. Includes links to diagnosis, evaluation and treatment summaries, section on living with Gaucher disease, and questions and answers about therapies.

Center for Jewish Diseases
Mt. Sinai School of Medicine
www.mssm.edu/jewish_genetics
Follow links to Gaucher disease. Includes disease description, natural history, genetics and testing information.

Additional literature:

The Official Parent's Sourcebook on Gaucher's Disease: A Revised and Updated Directory for the Internet Age
by Parker, P.M. and Parker, J.N. (eds.)
Icon Health, 2002
Informs parents where and how to look for information, also useful for doctors, caregivers, and other health professionals.

~ GLYCOGEN STORAGE DISEASE ~

Association for Glycogen Storage Disease
P.O. Box 896
Durant, IA 52747
Phone: 563-785-6038
Fax: 563-785-6038
Email: maryc@agsdus.org
www.agsdus.org
No Connecticut chapter.

The Children's Fund for Glycogen Storage Disease Research
917 Bethany Mountain Road
Cheshire, CT 06416
Phone: 203-272-CURE, 203-272-7744
Email: info@curegsd.org
www.curegsd.org

American Liver Foundation
75 Maiden Lane
Suite 603
New York, NY 10038
Phone: 800-465-4837/212-688-1000
Fax: 212-483-8179
Email: info@liverfoundation.org
www.liverfoundation.org

In Connecticut:
127 Washington Avenue
North Haven, CT 06473
Phone: 203-234-2022
Fax: 203-234-1386
Email: ctalf@liverfoundation.org
www.ctalf.org

~ GROWTH DISORDERS ~

Human Growth Foundation
997 Glen Cove Avenue
Glen Head, NY 11545
Phone: 800-451-6434/516-671-4041
Fax: 516-671-4055
Email: hgf1@hgfound.org
www.hgfound.org
No Connecticut chapter.

MAGIC Foundation for Children's Growth
6645 West North Avenue
Oak Park, IL 60302
Phone: 800-362-4432/708-383-0808
Fax: 708-383-0899
Email: mary@magicfoundation.org
www.magicfoundation.org
No Connecticut chapter.

~ HEART DISORDERS ~

American Heart Association
7272 Greenville Avenue
Dallas, TX 75231
Phone: 800-242-8721/214-373-6300
Fax: 214-373-0268
Email: inquire@heart.org
www.americanheart.org

In Connecticut:
5 Brookside Drive
P.O. Box 5022
Wallingford, CT 06492
Phone: 203-294-0088
Fax: 203-294-3577

Congenital Heart Anomalies Support, Education, and Research
2112 North Wilkins Road
Swanton, OH 43558
Phone: 419-825-5575
Fax: 419-825-2880
www.chsu.edu/~hfmth006/chaser/
No Connecticut chapter.

Mended Hearts Inc.
7272 Greenville Avenue
Dallas, TX 75231
Phone: 888-432-7899/214-706-1442
Fax: 214-706-5245
Email: info@mendedhearts.org
www.mendedhearts.org

Connecticut Chapter
Marvin Keyser, President
Hartford, CT
Phone: 860-582-0299
Email: DACBULDOG3@aol.com
www.mendedheartsct.org

Children's Heart Information Network
1561 Clark Drive
Yardley, PA 19067
Phone: 215-493-3068
Fax: 215-493-3068
Email: mb@tchin.org
www.tchin.org
No Connecticut chapter.

Little Hearts, Inc.
P.O. Box 171
Cromwell, CT 06416
Phone: 860-635-0006
Fax: 860-635-0006
Email: info@littlehearts.net
www.littlehearts.net

Additional web resources:

Texas Heart Institute
www.tmc.edu/thi/congenit.html

Congenital Heart Disease Center
www.heartcenteronline.com
Provides patient guides on many congenital heart anomalies, includes animated videos of how the heart works.

Did You Know...?

Scientists have been able to study the DNA of 30-milliion-year-old termites and a Neanderthal.

ADDITIONAL LITERATURE:

CARDIAC KIDS: A BOOK FOR FAMILIES WHO HAVE A CHILD WITH HEART DISEASE
by Elder, V. and King, A.
Tenderhearts Publishing Company, 1984
For parents and children to read together, book explains many of the medical tests a child will experience after being diagnosed with heart disease. It also touches on some of the stress siblings may feel.

HEART DEFECTS IN CHILDREN: WHAT EVERY PARENT SHOULD KNOW
by Wild, C.J.
Wiley, 1998
Designed for parents of newly diagnosed children, also a useful resource for families of prenatally diagnosed patients as well.

THE HEART OF A CHILD: WHAT FAMILIES NEED TO KNOW ABOUT HEART DISORDERS IN CHILDREN
by Clark, E.B., Clark, C. and Neill, C.A.
Johns Hopkins University Press,
2nd Edition, 2001
Designed for parents of newly diagnosed children, also a useful resource for families of prenatally diagnosed patients as well.

THE PARENT'S GUIDE TO CHILDREN'S CONGENITAL HEART DEFECTS: WHAT THEY ARE, HOW TO TREAT THEM, HOW TO COPE WITH THEM
by Kramer, G.F. and Maurer, S.
Three Rivers Press, 2001
Designed for parents of newly diagnosed children, also a useful resource for families of prenatally diagnosed patients as well.

FETAL ECHOCARDIOGRAPHY
by Drose, J.A.
W. B. Saunders Company, 1998
An essential reference for anyone involved in fetal echocardiography.

~ HEMOCHROMATOSIS ~

HEMOCHROMATOSIS FOUNDATION, INC.
P.O. Box 8569
Albany, NY 12208
Phone: 518-489-0972
Fax: 518-489-0227
Email: not available
www.hemochromatosis.org
No Connecticut chapter.

AMERICAN HEMOCHROMATOSIS SOCIETY
4044 West Lake Mary Boulevard
Suite 104, PMB 416
Lake Mary, FL 32746
Phone: 888-655-4766/407-829-4488
Fax: 407-333-1284
Email: mail@americanhs.org
www.americanhs.org
No Connecticut chapter.

AMERICAN LIVER FOUNDATION
75 Maiden Lane
Suite 603
New York, NY 10038
Phone: 800-465-4837/212-688-1000
Fax: 212-483-8179
Email: info@liverfoundation.org
www.liverfoundation.org

In Connecticut:
127 Washington Avenue
North Haven, CT 06473
Phone: 203-234-6304
Fax: 203-234-1386
Email: ctalf@liverfoundation.org
www.ctalf.org

ADDITIONAL WEB RESOURCES:

HEMOCHROMATOSIS INFORMATION SOCIETY
www.hemoinfo.org
Online resource with information on condition, treatment, how to get an at-home test kit, links for further information.

Additional literature:

Living with Hemochromatosis
by Everson, G. and Weinberg, H.
Hatherleigh Press, 2003
Patient guide on signs, symptoms, diagnosis and treatment options, and new areas of research. Also physical, emotional, nutritional and financial issues.

The Iron Disorders Institute Guide to Hemochromatosis
by Garrison, C.D., Phatak, D.D., Weinber, E.D. and Burke, W.
Cumberland House Publishing, 2001
Valuable information on diagnosis, treatment, lifestyle, nutrition and genetics.

The Official Patient's Sourcebook on Hemochromatosis
by Parker, J.N. and Parker, P.M.
Icon Health, 2002
Tells patients, doctors, caregivers and other health professionals how to look for information on hemochromatosis.

~ HEMOPHILIA ~

National Hemophilia Foundation
116 West 32nd Street
11th floor
New York, NY 10001
Phone: 800-424-2634/212-328-3700
Fax: 212-328-3777
Email: info@hemophilia.org
www.hemophilia.org

New England Hemophilia Foundation
347 Washington Street
Suite 402
Dedham, MA 02026
Phone: 781-326-7645
Fax: 781-329-5122
Email: neha@theworld.com
www.newenglandhemophilia.org

Additional web resources:

BloodLine
www.bloodline.net
An online resource for hematology education and news.

~ HUNTINGTON'S DISEASE ~

Huntington's Disease Society of America
158 West 29th Street
7th floor
New York, NY 10001
Phone: 800-345-4372/212-242-1968
Fax: 212-239-3430
Email: hdsainfo@hdsa.org
www.hdsa.org

Connecticut Chapter:
Phone: 860-679-4441

Hereditary Disease Foundation
1303 Pico Boulevard
Santa Monica, CA 90405
Phone: 310-450-9913
Fax: 310-450-9532
Email: cures@hdfoundation.org
www.hdfoundation.org
No Connecticut chapter.

Additional web resources:

The Huntington's Disease Association Online
www.hda.org.uk
Offers news and information about Huntington's disease for people affected, their families, friends, and health care professionals.

Huntington's Disease Advocacy Center
www.hdac.org
Chat rooms, research updates, information on living with Huntington's disease, managing symptoms, caregiving, finances, and nursing homes.

ADDITIONAL LITERATURE:

HUNTINGTON'S DISEASE: THE FACTS
by Quarrell, O.
Oxford University Press, 1999
Pocket guide that offers practical advice, discusses medical facts, genetic aspects and counseling, neuropathology, and support organizations.

FACES OF HUNTINGTON'S
by Leal-Pock, C.
Living Hope Inc., 1998
A unique collection of writings which illuminates the many facets of Huntington's Disease.

MAPPING FATE: A MEMOIR OF FAMILY, RISK, AND GENETIC RESEARCH
by Wexler, A.
University of California Press, 1996
Personal account of a family dealing with Huntington's Disease.

~ HYDROCEPHALUS ~

NATIONAL HYDROCEPHALUS FOUNDATION
12413 Centralia Road
Lakewood, CA 90715
Phone: 888-857-3434/562-402-3523
Fax: 562-924-6666
Email: hydrobrat@earthlink.net
www.nhfonline.org
No Connecticut chapter.

HYDROCEPHALUS ASSOCIATION
870 Market Street
Suite 705
San Francisco, CA 94102
Phone: 888-598-3789/415-732-7040
Fax: 415-732-7044
Email: info@hydroassoc.org
www.hydroassoc.org
No Connecticut chapter.

GUARDIANS OF HYDROCEPHALUS RESEARCH FOUNDATION
2618 Avenue Z
Brooklyn, NY 11235
Phone: 800-458-8655/718-743-4473
Fax: 718-743-1171
Email: GHRF2618@aol.com
www.ghrf.homestead.com/ghrf.html
No Connecticut chapter.

HYDROCEPHALUS FOUNDATION
910 Rear Broadway
Route 1
Saugus, MA 01906
Phone: 781-942-1161
Fax: not available
Email: HyfII@netscape.net
www.hydrocephalus.org
No Connecticut chapter.

ADDITIONAL WEB RESOURCES:

THE HYDROCEPHALUS CENTER
www.patientcenters.com/hydrocephalus

NIH HYDROCEPHALUS LINKS
www.nlm.nih.gov/medlineplus/hydrocephalus.html

ADDITIONAL LITERATURE:

HYDROCEPHALUS: A GUIDE FOR PATIENTS, FAMILIES AND FRIENDS
by Toporek, C. and Robinson, K., Lamb, L.
O'Reilly Media, Inc., 1999
Addresses selecting a skilled neurosurgeon, treatments, lifestyles, and where to turn for support.

PEDIATRIC HYDROCEPHALUS
by Cinalli, G., Maixner, W.J. and Sainte-Rose, C.
Springer-Verlag, 2004
Medical reference text for health care professionals, includes classification, etiology, pathophysiology, genetics, and recent progresses in the field.

~ KLINEFELTER SYNDROME ~

American Association for Klinefelter Syndrome Information and Support
2945 West Farwell Avenue
Chicago, IL 60645
Phone: 888-466-5747/773-761-5298
Fax: 773-761-5298
Email: xxyinfo@aaksis.org
www.aaksis.org
No Connecticut chapter.

Klinefelter Syndrome and Associates
11 Keats Court
Coto De Caza, CA 92679
Phone: 888-999-9428/916-773-1449
Fax: 916-773-1449
Email: help@genetic.org
www.genetic.org
No Connecticut chapter.

Additional web resources:

Klinefelter Syndrome Support Group Home Page
www.klinefeltersyndrome.org
Includes links to various and prenatal diagnosis information, support group information, research studies, medical conferences, and links to other useful websites.

Additional literature:

Klinefelter's Syndrome: A Medical Dictionary, Bibliography, and Annotated Research Guide to Internet References
by Icon Health Publications
Icon Health, 2004
Complete medical dictionary, lists of bibliographic citations, information on Internet resources. Designed for physicians, medical students, medical researchers, and patients.

~ KLIPPEL-TRENNAUNAY-WEBER ~

Klippel-Trenaunay Support Group
5404 Dundee Road
Edina, MN 55436
Phone: 952-925-2596
Fax: 952-925-4708
Email: ktnewmembers@yahoo.com
Web: www.k-t.org
No Connecticut chapter.

~ LESCH-NYHAN DISEASE ~

Lesch-Nyhan Syndrome Registry
New York University School of Medicine
Department of Psychiatry
New Bellevue 18 18E13
Bellevue Hospital
New York, NY 10012
Phone: 212-263-6458
Fax: 212-629-9523
Email: lta1@nyu.edu
www.lndinfo.org
No Connecticut chapter.

Lesch-Nyhan Syndrome Children's Research Foundation
210 South Green Bay Road
Lake Forest, IL 60045
Phone: 847-234-3154
Fax: 847-234-3136
No Connecticut chapter.

Additional web resources:

National Institute of Neurological Disorders and Stroke; Lesch-Nyhan Syndrome Information Page
www.ninds.nih.gov/health_and_medical/disorders/lesch_doc.htm

~ LEUKODYSTROPHY ~

United Leukodystrophy Foundation
2304 Highland Drive
Sycamore, IL 60178
Phone: 800-728-5483/815-895-3211
Fax: 815-895-2432
Email: ulf@tbc.com
www.ulf.org
No Connecticut chapter.

Additional literature:

21st Century Complete Medical Guide to Leukodystrophies: Authoritative Government Documents, Clinical References, and Practical Information for Patients and Physicians
by PM Medical Health News
Progressive Management, 2004
Electronic, thoroughly-researched collection on CD-ROM. For patients, practical information is provided in clearly written education documents. For medical professionals, doctor reference tools and texts had detailed technical information and clinical background material.

~ LUPUS ~

American Autoimmune Related Diseases Association, Inc.
22100 Gratiot Avenue
Eastpointe, MI 48021
Phone: 586-776-3900
Fax: 586-776-3903
Email: aarda@aarda.org
www.aarda.org
No Connecticut chapter.

SLE Foundation, Inc.
149 Madison Avenue
Suite 205
New York, NY 10016
Phone: 212-685-4118
Fax: 212-545-1843
Email: lupus@lupusny.org
www.lupusny.org
No Connecticut chapter.

Lupus Foundation of America, Inc.
2000 L Street, NW
Washington, DC 20036
Phone: 800-558-0121/202-349-1155
Fax: 202-349-1156
Email: info@lupus.org
www.lupus.org

Connecticut chapter:

97 South Street
Suite 110
West Hartford, CT 06110
Phone: 860-953-0387
Fax: 860-953-0483
Email: CTLFA@aol.com
www.lupusct.org

Additional web resources:

Lupus Suite 101
www.suite101.com/welcome.cfm/lupus
Written by a nurse who is also a lupus patient. Provides information, support and help.

The Lupus Site
www.uklupus.co.uk
Information on lupus including symptoms, diagnosis, tests, medications, email list, and surveys.

Additional literature:

Coping with Lupus: A Guide to Living With Lupus for You and Your Family
by Phillips, R.H.
Avery Publishing Group, 2nd Edition, 1990
Updated, includes new research, treatments, and information.

The Lupus Handbook for Women
by Dibner, R.
Fireside, 1994
"Must read" book of common sense, advice, basics, tips for daily living, entire chapter on pregnancy.

~ MARFAN SYNDROME ~

National Marfan Foundation
22 Manhassett Avenue
Port Washington, NY 11050
Phone: 800-862-7326/516-883-8712
Fax: 516-883-8040
Email: staff@marfan.org/support@marfan.org
www.marfan.org
No Connecticut chapter.

Additional web resources:

National Institute of Arthritis and Musculoskeletal and Skin diseases
www.niams.nih.gov/hi/topics/marfan/marfan.htm

Additional literature:

Connective Tissue and Its Heritable Disorders: Molecular, Genetic and Medical Aspects
by Royce, P.M. and Steinmann, B.
Wiley-Liss, 1993
Reference text which provides up to date clinical and scientific information for medical specialists treating affected individuals.

~ MENTAL RETARDATION ~

The ARC (formerly known as The Association for Retarded Citizens)
1010 Wayne Avenue
Silver Spring, MD 20910
Phone: 800-433-5255/301-565-3842
Fax: 301-565-5342
Email: info@thearc.org
www.thearc.org

In Connecticut:
(22 chapters listed alphabetically by town)

Sarah Seneca Residential Services
11 Business Park Drive
Suite 1
Branford, CT 06405
Phone: 203-315-3770
Email: sarahsenecares@snet.net

Farmington Valley ARC, Inc.
225 Commerce Drive
P.O. Box 1099
Canton, CT 06019
Phone: 860-693-6662
Email: favarh@favarh.org
www.favarh.org

Tri-County ARC, CT
65 Route 66 East
Columbia, CT 06237
Phone: 860-228-2070 X103

WeCAHR
211 Main Street
Danbury, CT 06810
Phone: 203-792-3540
www.wecahr.org

The ARC of Quinebaug Valley
687 Cook Hill Road
Danielson, CT 06239
Phone: 860-774-2827

The ARC of Greater Enfield
75 Hazard Avenue
Unit E
Enfield, Ct 06082
Phone: 860-763-5411

Sarah, Inc.
246 Goose Lane
Suite 101
Guilford, CT 06437
Phone: 203-458-4040
www.sarah-inc.org

Sarah Tuxis Residential Services, Inc.
45 Boston Street
Guilford, CT 06437
Phone: 203-458-8532
Email: tuxis@cshore.com
www.sarah-tuxis.org

MARC, Inc. of Manchester
376R West Middle Turnpike
Manchester, CT 06040
Phone: 860-646-5718
Email: info@marcct.org
www.marcct.org

The ARC of Meriden-Wallingford, Inc.
224-226 Cook Avenue
Meriden, CT 06451
Phone: 203-237-9975
Email: mwshinc@hotmail.com
www.mwsinc.org

Directions, Inc.
62 Washington Street
Middletown, CT 06457
Phone: 860-347-5099
Email: PDFutures@aol.com

Options Unlimited, Inc.
584 West Hill Road
New Hartford, CT 06057
Phone: 860-738-1410
Email: optionsunltd@myexcel.com

Friends of New Milford, Inc.
238 Chesnutland Road
New Milford, CT 06776
Phone: 860-355-5343

STAR, Inc., Lighting the Way
P.O. Box 470
Norwalk, CT 06851
Phone: 203-846-9581
Web: www.starinc-lightingtheway.org

New London County ARC
125 Sachem Street
Norwich, CT 06360
Phone: 860-889-4435
Email: nlarc@snet.net

The ARC of Plainville
367 New Britain Avenue
Plainville, CT 06062
Phone: 860-747-1560

The ARC of Southington, Inc.
201 West Main Street
Plantsville, CT 06479
Phone: 860-628-9220
Email: advocacy@arcsouthington.org
www.arcsouthington.org

Marc Community Resources, Ltd.
12 Fairview Street
P.O. box 126
Portland, CT 06480
Phone: 860-342-0700
Email: marcinfo@ucoonect.net
www.marcweb.org

Litchfield County ARC
84-R Main Street
Torrington, CT 06790
Phone: 860-482-9364
Email: larc@litchfieldarc.org
www.litchfieldarc.org

Waterbury ARC
1929 East Main Street
Waterbury, CT 06705
Phone: 203-575-0707
Email: warc@snet.net

Family Options
51 Depot Road
Watertown, CT 06795
Phone: 860-274-0757

The ARC of Connecticut
1030 New Britain Avenue
Suite 102
West Hartford, CT 06110
Phone: 860-953-8335
Email: arcct@aol.com
www.arcct.com

American Association on Mental Retardation
444 North Capitol Street NW
Suite 846
Washington, DC 20001
Phone: 800-424-3688, 202-387-1968
Fax: 202-387-2193
Email: dcroser@aamr.org
www.aamr.org

Connecticut chapter:
Region X
Contact James T. Morrison
Email: marcjamesm@hotmail.com

~ MITOCHONDRIAL DISORDERS ~

United Mitochondrial Disease Foundation
8085 Saltsburg Road
Suite 201
Pittsburgh, PA 15239
Phone: 412-793-8077
Fax: 412-793-6477
Email: info@umdf.org
www.umdf.org
No Connecticut chapter.

New England Chapter of UMDF
39 Bay Farm Drive
Plymouth, MA 02360
Phone: 413-593-5920
Fax: none
Email: NEngChapter@umdf.org

Mitochondria Research Society
P.O. Box 1952
Buffalo, NY 14221
Phone: see website for individual contacts
Fax: not available
Email: mitoresearch@mitoresearch.org
www.mitoresearch.org
No Connecticut chapter.

Additional web resources:

Mitochondrial Disease in Perspective: Symptoms, Diagnosis and Hope for the Future
www.mitoresearch.org/Quest_6_5.htm
Online lecture covering diagnosis, management and symptoms new research and promising treatments.

Additional literature:

Mitochondrial Disease: Models and Methods
by Lestienne, P.
Springer-Verlag, 1999
Covers molecular aspects of mitochondrial disorders, diagnosis and mutations; discusses role of mitochondria in apoptosis and aging.

~ MOEBIUS SYNDROME ~

Moebius Syndrome Foundation
P.O. Box 147
Pilot Grove, MO 65276
Phone: 660-834-3406
Fax: 660-834-3407
Email: vmccarrell@mid-mo.net or
Moebius@ciaccess.com
www.moebiussyndrome.com
No Connecticut chapter.

Additional web resources:

Moebius Syndrome
www.moebius1.org
Site to promote international awareness, offer information resource links and provide interactive support format. A personal approach.

Additional literature:

My Face
by Abbott, M. and Abbott, B.
Forward Face, 1998
Children's book written by mother of affected child.

~ MUCOPOLYSACCHARIDOSIS ~

National MPS Society, Inc.
P. O. Box 736
Bangor, ME 04402
Phone: 207-947-1445
Fax: 207-990-3074
Email: info@mpssociety.org
www.mpssociety.org

~ MULTIPLE SCLEROSIS ~

National Multiple Sclerosis Society
733 3rd Avenue
New York, NY 10017
Phone: 800-344-4867/212-986-3240
Fax: 212-986-7981
Email: generalmailbox@nmss.org
www.nationalmssociety.org

In Connecticut:
Greater Connecticut Chapter
705 North Mountain Road
Newington, CT 06111
Phone: 860-953-0601
Email: lgerrol@ctnmss.org
www.ctnmss.org

Western Connecticut Chapter
1 Selleck Street
Suite 500
Norwalk, CT 06855
Phone: 203-838-1033
Email: lparizeau@msswct.org
www.msswct.org

Multiple Sclerosis Association of America
706 Haddonfield Road
Cherry Hill, NJ 08002
Phone: 800-532-7667
Fax: 856-661-9797
Email: msaa@msaa.com
www.msa.com

Connecticut contacts:

Marva Llwellyn
Stamford, CT
Phone: 203-921-1984

Jeannie Beresford
Suffield, CT
Phone: 860-668-2792

Additional web resources:

All About Multiple Sclerosis
www.mult-sclerosis.org
Aims to provide accurate and comprehensive medical information about multiple sclerosis written in plain English by people living with the disease. Includes encyclopedia archives of news stories and personal accounts.

Additional literature:

Multiple Sclerosis Q & A: Reassuring Answers to Frequently Asked Questions
by Hill, B.A. and Wojcieszek, J.
Avery Publishing Group, 2003
Insightful, informative, and empathic resource, discusses traditional and complementary therapies, explains medical terminology and diagnosis, addresses lifestyle issues.

Managing the Symptoms of Multiple Sclerosis
by Schapiro, R.T.
Demos Medical Publishing, 4th Edition, 2003
Symptom by symptom discussion, overview of the disease and relation to patient's total health.

~ MUSCULAR DYSTROPHY ~

Muscular Dystrophy Association
3300 East Sunrise Drive
Tucson, AZ 85718
Phone: 800-572-1717/520-529-2000
Fax: 520-529-5300
Email: mda@mdausa.org
www.mdausa.org

Connecticut contact:
Health Services Coordinator
Phone: 860-633-4466

Muscular Dystrophy Family Foundation
2330 North Meridian Street
Indianapolis, IN 46208
Phone: 800-544-1213/317-923-6333
Fax: 317-923-6334
Email: info@mdff.org
www.mdff.org
No Connecticut chapter.

Additional web resources:

Parent Project Muscular Dystrophy
www.parentprojectmd.org
Founded by parents of kids with Duchenne and Becker muscular dystrophies, they identify, fund and disseminate information on research, support an international conference, and provide a newsletter. Includes links to information and resources, breaking news, treatments, and emotional issues.

Additional literature:

Muscular Dystrophy in Children: A Guide for Families
by Siegel, I.M.
Demos Medical Publishing, 1999
Text for consumers, includes information on signs and symptoms, medical treatments, psychosocial issues, probable course of disease, and therapies.

Muscular Dystrophy: The Facts
by Emery, A.E.H.
Oxford University Press, 2nd Edition, 2000
Easy to understand language explaining the complexities of muscular dystrophy, including daily life issues.

Moonrise: One Family, Genetic Identity, and Muscular Dystrophy
by Wolfson, P.
St. Martin's Press, 2003
Written by a mother of a son with Duchenne muscular dystrophy. Personal account through diagnosis, prognosis, and natural history. Explores special education, prenatal diagnosis, and genetics.

~ NEUROFIBROMATOSIS ~

National Neurofibromatosis Foundation, Inc.
95 Pine Street 16th floor
New York, NY 10005
Phone: 800-323-7938/212-344-6633
Fax: 212-747-0004
Email: nnff@nf.org
www.nf.org
No Connecticut chapter.

NNFF Northern New England Chapter
75 McNeil Way
Suite 201
Dedham, MA 02026
Phone: 888-585-5316/508-879-5638
Fax: 781-326-4940
Email: nfne.ed@verizon.net

Neurofibromatosis, Inc.
9320 Annapolis Road Suite 300
Lanham, MD 20706
Phone: 800-942-6825/301-918-4600
Fax: 301-918-0009
Email: info@nfinc.org
www.nfinc.org
No Connecticut chapter.

NF Inc. New England
9 Bedford Street
Burlington, MA 01803
Phone: 781-272-9936
Email: info@nfincne.org
www.nfincne.org

Additional web resources:

Neurofibromatosis Resources
www.neurosurgery.mgh.harvard.edu/NFR
Online listing of neurofibromatosis information on the web, including general information, genetics, support, news groups, homepages, and information for clidren and adolescents.

Additional literature:

Living with Genetic Disorder: The Impact of Neurofibromatosis I.
by Ablon, J.
Auburn House, 1999
A chronicle of the life experiences of adults with Neurofibromatosis I.

Neurofibromatosis Type I: From Genotype to Phenotype
by Upadhyaya, M. and Cooper, D.N.
Academic Press, 1998
Medical reference text written for students through professionals. Includes clinical aspects, gene structure, expression, and mutation, animal models, disease treatment and prevention.

~ NIEMANN-PICK ~

National Niemann-Pick Foundation, Inc.
P.O. Box 49
415 Madison Avenue
Fort Atkinson, WI 53538
Phone: 877-287-3672/920-563-0930
Fax: 920-563-0931
Email: nnpdf@idcnet.com
www.nnpdf.org
No Connecticut chapter.

Additional web resources:

Center for Jewish Diseases
Mt. Sinai School of Medicine
www.mssm.edu/jewish_genetics
Includes disease description, natural history, genetics and testing information.

Additional literature:

The Official Parent's Sourcebook on Niemann-Pick Disease: A Revised and Updated Directory for the Internet Age
by Parker, J.N. & parker, P.M., (eds)
Icon Health, 2002
Guide to how to look for information online, how to find a doctor, how to locate the atest research.

~ ORGANIC ACIDEMIA ~

Organic Acidemia Association
13210 35th Avenue North
Plymouth, MN 55441
Phone: 763-559-1797
Fax: 763-694-0017
Email: oaanews@aol.com
www.oaanews.org
No Connecticut chapter

~ OSTEOGENESIS IMPERFECTA ~

Osteogenesis Imperfecta Foundation, Inc.
804 West Diamond Avenue Suite 210
Gaithersburg, MD 20878
Phone: 800-981-2663/301-947-0083
Fax: 301-947-0456
Email: bonelink@oif.org
www.oif.org
No Connecticut chapter.

Children's Brittle Bone Foundation
7701 95th Street
Pleasant Prairie, WI 53158
Phone: 866-694-2223
Fax: 262-947-0724
Email: info@cbbf.org
www.cbbf.org
No Connecticut chapter.

Additonal web resources:

The Bones Page
www.geocities.com/CapeCanaveral/Lab/3608
Tips for everyday life, basic information, and links to other resources.

~ PARKINSON'S DISEASE ~

American Parkinson Disease Association
1250 Hylan Boulevard
Suite 4B
Staten Island, NY 10305
Phone: 800-223-2732
Fax: 718-981-4399
Email: adpa@apdaparkinson.org
ww.apdaparkinson.com or org

In Connecticut:
APDA Connecticut Chapter
27 Allendale Drive
North Haven, CT 06473
Phone: 203-288-0546
Fax: 203-288-0546
Email: gladkt@hotmail.com
www.ctapda.com
See Connecticut website for 20 local contacts.

Parkinson's Disease Foundation, Inc.
710 West 168th Street
New York, NY 10032
Phone: 800-457-6676/212-923-4700
Fax: 212-923-4778
Email: info@ppdf.org
www.pdf.org

No Connecticut chapter

Additional web resources:

Parkinson's Information
www.parkinsonsinfo.com
Information about Parkinson's disease, a directory or resources, and frequently asked questions.

Michael J. Fox Foundation
www.michaeljfox.org
Dedicated to ensuring the development of a cure for Parkinson's disease within this decade.

Additional literature:

300 Tips for Making Life with Parkinson's Disease Better
by Schwarz, S.P.
Demos Medical Publishing, 2002
Tips, techniques, and shortcuts learned from personal experience, arranged in categories of daily activities.

Parkinson's Disease: A Guide for Patient and Family
by Duvoisin, R.C. and Sage, J.
Raven Press, LTD., 3rd Edition, 1990
In depth information on diagnosis, medications, management, and genetics, and hopes for the future.

Did You Know...?

2003 marked the 50th anniversary of the discovery of the structure of DNA.

~ PHENYLKETONURIA ~

Children's PKU Network
3970 Via de la Valle
Suite 120 E
Del Mar, CA 92014
Phone: 858-509-0767
Fax: 858-509-0768
Email: pkunetwork@aol.com
www.pkunetwork.org
No Connecticut chapter.

Additional web resources:

National PKU News
www.pkunews.org
News and information about PKU including personal stories, information for students writing papers, support information, and information on legislation and policies relevant to PKU.

Low Protein Recipes
www.lowprotein.com
"Successful and sumptuous" low protein recipes put together by a mother of a child with homocystinuria.

National Coalition for PKU and Allied Disorders
www.pku-allieddisorders.org
Organization comprised of individuals, metabolic support groups and professionals seeking to improve the identification, treatment, and management of PKU and allied disorders.

Additional literature:

Low Protein Cookery for Phenylketonuria
by Schuett, V.E.
University of Wisconsin Press, 1997
Provides recipes for American style cooking, including instructions for calculating nutrient content, and special circumstances such as kid's parties, etc.

~ PORPHYRIA ~

American Porphyria Foundation
P.O. Box 22712
Houston, TX 77227
Phone: 713-266-9617
Fax: 713-871-1788
Email: porphyrus@aol.com
www.porphyriafoundation.com
No Connecticut chapter.

~ PRADER-WILLI SYNDROME ~

Prader Willi Syndrome Association
5700 Midnight Pass Road Suite 6
Sarasota, FL 34242
Phone: 800-926-4797/941-312-0400
Fax: 941-312-0142
Email: national@pwsausa.org
www.pwsausa.org

In Connecticut:
Prader-Willi Syndrome Association
Connecticut Chapter, Inc.
35 Ansonia Drive
North Haven, CT 06473
Phone: 203-239-9902
Fax: none
Email: pwsactchapter@yahoo.com

Additional web resources:

Uniparental Disomy: Prader-Willi Syndrome, Angelman Syndrome
www.lpch.org/DiseaseHealthInfo/HealthLibrary/genetics/uniparen.html
Brief overview provided by the Lucile Packard Children's Hospital at Stanford.

Did You Know...?

12,000 letters of DNA were decoded by the Human Genome Project every second.

Additional literature:

Prader-Willi Syndrome: Development and Manifestations
by Whittington, J. and Holland, T.
Cambridge University Press, 2004
Based on the largest cohort of studies on Prader Willi syndrome, provides information on the conditions, and management issues in medical, nutritional, psychological, educational, social, and therapeutic needs.

~ PRUNE BELLY SYNDROME ~

Prune Belly Syndrome Network, Inc.
P.O. Box 2092
Evansville, IN 47728
Phone: 310-825-6865
Fax: 310-794-9962
Email: rmoehlmann@prunebelly.org
www.prunebelly.org
No Connecticut chapter.

~ PSEUDOXANTHOMA ELASTICUM ~

National Association for Pseudoxanthoma Elasticum, Inc.
8764 Manchester Road
Suite 200
St. Louis, MO 63144
Phone: 314-962-0100
Fax: 314-962-0100
Email: pxenapee@napxe.org
www.napxe.org
No Connecticut chapter.

PXE, International
4301 Connecticut Avenue NW, Suite 404
Washington, DC 20008
Phone: 203-362-9599
Fax: 202-966-8553
Email: pxe@pxe.org
www.pxe.org
No Connecticut chapter.

PXE New England Regional Office
Gordon & Wendy Wood Hubbard
73 Marnoch Drive
Seekonk, MA 02771
Phone: 508-336-7461
Email: PXENewEng@aol.com

~ RETINITIS PIGMENTOSA ~

Retinitis Pigmentosa International
P.O. Box 900
Woodland Hills, CA 91365
Phone: 818-992-0500
Fax: 818-992-3265
Email: info@rpinternational.org
www.rpinternational.org
No Connecticut chapter.

Foundation Fighting Blindness
11435 Cronhill Drive
Owings Mills, MD 21117
Phone: 888-394-3937/410-568-0150
Fax: 410-363-2393
Email: info@blindness.org
www.blindness.org

In Connecticut:
Contact Leonard Roberto
Phone: 203-268-4031
Email: robbylenn@aol.com

American Council of the Blind, Inc.
1155 15th Street, NW, Suite 1004
Washington, DC 20005
Phone: 800-424-8666/202-467-5081
Fax: 202-467-5085
Email: info@acb.org
www.acb.org

In Connecticut:
Connecticut Council of the Blind
Alice Jackson
191 Centrebrook Road
Hamden, CT 06518
Email: ajackson212@comcast.net

American Foundation for the Blind
11 Penn Plaza, Suite 300
New York, NY 10001
Phone: 800-232-5463/212-502-7600
Fax: 212-502-7777
Email: afbinfo@afb.net
www.afb.org
No Connecticut chapter.

Additional web resources:

Visionchannel
www.visionchannel.net/retinitis
Includes overview of retinitis pigmentosa, signs, symptoms, risk factors, treatment, and diagnosis.

Additional literature:

Ordinary Daylight: Portrait of an Artist Going Blind
by Potok, A.
Bantam, 2003
Story of a gifted painter with retinitis pigmentosa who loses his sight. Depressed and angry, he ultimately discovers it is not the end of the world, it is the beginning.

Amazing Grace: Autobiography of a Survivor
by Halloran, G.
North Star Publications, 1993
Personal account of blind mother and her son, her unconventional therapies, challenges and triumphs.

~ RETT SYNDROME ~

International Rett Syndrome Association
9121 Piscataway road, 2B
Clinton, MD 20735
Phone: 800-818-7388/301-856-3334
Fax: 301-856-3336
Email: irsa@rettsyndrome.org
www.rettsyndrome.org
No Connecticut chapter.

Rett Syndrome Research Foundation
4600 Devitt Drive
Cincinnati, OH 45246
Phone: 513-874-3020
Fax: 513-874-2520
Email: mgriffin@rsrf.org
www.rsrf.org

Connecticut contact:
Monica Coenraads
67 Under Cliff Road
Trumbull, CT 06611
Phone: 203-445-9233
Email: monica@rsrf.org

Additional web resources:

The DRM WebWatcher: Rett syndrome
www.disabilityresources.org/RETT.html
Links to sites for information on Rett syndrome.

Additional literature:

Grief Dancers:
A Journey into the Depths of the Soul
by Zimmerman, S.
Nemo Press, 1996
Mother's story of raising a daughter with Rett Syndrome.

~ SCLERODERMA ~

Scleroderma Foundation
12 Kent Way, Suite 101
Byfield, MA 01922
Phone: 800-722-4673/978-463-5843
Fax: 978-463-5809
Email: sfinfo@scleroderma.org
www.scleroderma.org

Connecticut chapter:

Tri-State Chapter (Connecticut, Northern New Jersey, South Central and Eastern New York)
62 Front Street
Binghamton, NY 13905
Phone: 800-867-0885/607-723-2239
FAX: 607-723-2039
Email: sdtrristate@aol.com
www.scleroderma/org/chapter/tristate

Margaret Whitehead Van Why Connecticut Support Group
Contact Mary Mannillo
Phone: 860-521-3024

Scleroderma Research Foundation
2320 Bath Street
Suite 315
Santa Barbara, CA 93105
Phone: 800-441-2873/805-563-9133
Fax: 805-563-2402
Email: srfcure@srfcure.org
www.sclerodermausa.org
No Connecticut chapter.

Additional web resources:

Scleroderma From A to Z
www.sclero.org
Resource available in multiple languages with links to medical information, support groups, personal accounts, books, message boards, and news.

Additional literature:

The Scleroderma Book:
A Guide for Patients and Families
by Mayes, M.D.
Oxford University Press, 1999
Up-to-date practical information provided by a leading expert in the field, a reliable resource.

Scleroderma:
A New Role for Patients and Families
by Brown, M.
Scleroderma Press, 2002
Written from the perspective of a patient and family, useful resource on both medical and non-medical issues.

~ SICKLE CELL ANEMIA ~

Sickle Cell Disease Association of America
200 Corporate Point
Suite 495
Culver City, CA 90230
Phone: 800-421-8453/310-216-6363
Fax: 310-215-3722
Email: scdaa@sicklecelldisease.org
www.sicklecelldisease.org

Connecticut chapter:

Southern Regional Sickle Cell Association, Inc.
177 State Street, 3rd floor
Bridgeport, CT 06604
Phone: 888-745-2327
Email: srsca@sicklecelldisease.org

Additional web resources:

Sickle Cell Information Center, Atlanta, GA
www.scinfo.org
Provides both the patient and the professional information on news, research updates, and world wide sickle cell resources.

Information Center for Thalassemia and Sickle Cell Disease
www.sickle.bwh.harvard.edu
Source of current information on sickle cell, thalassemia, and disorders of iron metabolism. Includes overviews of basic and clinical research, management and new developments in the field.

Additional literature:

Sickle Cell Disease: Basic Principles and Clinical Practice
by Embury, S.H., Hebbel, R.P. and Mohandas, N.
Lippincott, Williams & Wilkins, 1994
A synopsis of what is currently know about sickle cell disease, summarizing the core of basic principles and clinical practice. Sickle Cell Disease in Children and Adolescents: Diagnosis, Guidelines for Comprehensive Care, and Care Paths and Protocols for Management of Acute and Chronic Complications by the Sickle Cell Disease Care Consortium.

Did You Know...?

Scientists estimate that humans have only 1/3 more genes than the simple roundworm!

~ SJOGREN'S SYNDROME ~

Sjogren's Syndrome Foundation
8120 Woodmont Avenue
Suite 530
Bethesda, MD 20814
Phone: 800-475-6473/301-718-0300
Fax: 301-718-0322
Email: cwilt@sjogrens.org
www.sjogrens.org

In Connecticut:
State support leader; Isabel Lopez
Norwalk, CT
Phone: 203-846-1031

Additional web resources:

SjS World
www.sjsworld.org
Online community that offers patients and families a place to meet and share experiences, books, chat rooms, medical info, and email groups.

Additional literature:

The New Sjogren Syndrome Handbook
by Wallace, D.J., Bromet, E.J. and the Sjogren Syndrome Foundation
Oxford University Press, 1998
Updated version of original book to be released soon.

A Body Out of Balance: Understanding and Treating Sjogren Syndrome
by Fremes, R., Carteron, N. and Grayzel, A.
Avery Publishing Group, 2003
A comprehensive guide that covers all aspects of living with Sjogren syndrome.

The Sjogren Syndrome Survival Guide
by Rumpf, T.P. and Hammitt, K.M.
New Harbinger Publications, 2003
Detailed information on how to cope with this condition.

~ SMITH-LEMLI-OPITZ SYNDROME~

Smith-Lemli-Opitz/RSH Syndrome Advocacy and Exchange
2650 Valley Forge Drive
Boothwyn, PA 19061
Phone: 610-485-9663
Fax: not available
Email: bhook@erols.com
www.members.aol.com/slo97/index.html
No Connecticut chapter.

~ SOTOS SYNDROME ~

Sotos Syndrome Support Association
P.O. Box 4626
Wheaton, IL 60189
Phone: 888-246-7772
Fax: not available
Email: sssa@well.com
www.well.com/user/sssa
No Connecticut chapter.

Additional literature:

Sotos Syndrome: A Handbook for Families
by Anderson, R.R. and Buehler, B.A.
Sotos Syndrome Support Association.
Includes description of the condition, medical and developmental evaluations, and glossary. Order through the Sotos Syndrome Support Association.

~ SPINA BIFIDA ~

Spina Bifida Association of America
4950 MacArthur Boulevard NW, Suite 250
Washington, DC 20007
Phone: 800-621-3141/202-944-3285
Fax: 202-944-3295
Email: sbaa@sbaa.org
www.sbaa.org

In Connecticut:
Spina Bifida Association of Connecticut
P.O. Box 2545
Hartford, CT 06146
Phone: 800-574-6274
Fax: 860-345-2600
Email: sbac@sbac.org
www.sbac.org

Additional web resources:

Children with Spina Bifida: A Resource Page for Parents
www.waisman.wisc.edu/~rowley/sbkids/index.html
Links to spina bifida organizations, articles, websites, online discussion groups, related diagnoses, learning issues, family support, tests, surgeries and treatments, prenatal diagnosis and fetal surgery, and genetics.

Additional literature:

Views from Our Shoes: Growing Up with a Brother or Sister with Special Needs
by Meyer, D.J. and Pillo, C.
Woodbine House, 1997
About using a wheelchair. Recommended by a girl with spina bifida.

Children With Spina Bifida: A Parent's Guide
by Lutkenhoff, M.
Woodbine House, 1999

Spinabilities: A Young Person's Guide to Spina Bifida
by Lutkenhoff, M. and Oppenheimer, S.G.
Woodbine House, 1997

Did You Know...?

Identical twins do not have identical fingerprints.

~ TAY SACHS ~

National Tay-Sachs and Allied Diseases Association
2001 Beacon Street, Suite 204
Brighton, MA 02135
Phone: 800-906-8723
Fax: 617-277-0134
Email: info@ntsad.org
www.ntsad.org

In Connecticut:
(Serving the tri-state area)
1202 Lexington Avenue
#288
New York, NY 10028
Phone: 888-354-7788/212-431-0431
Email: info@ntsad-ny.org
www.ntsad-ny.org

Additional web resources:

Center for Jewish Diseases
Mt. Sinai School of Medicine
www.mssm.edu/jewish_genetics
Follow links to Tay-Sachs Disease. Includes disease description, natural history, genetics and testing information.

Additional literature:

Tay-Sachs Disease
by Desnick, R.J. and Kaback, M.M.
Academic Press, 2001
Highly recommended medical reference text written and edited by recognized leaders in the field.

~ THALASSEMIA ~

Cooley's Anemia Foundation, Inc.
129-09 26th Avenue
Suite 203
Flushing, NY 11354
Phone: 800-522-7222/718-321-2873
Fax: 718-321-3340
Email: info@cooleysanemia.org
www.cooleysanemia.org

In Connecticut:
Contact Peter Chieco
Phone: 914-232-1808
Email: anemia@optonline.net

Additional web resources:

Information Center for Thalassemia and Sickle Cell Disease
www.sickle.bwh.harvard.edu
Source of current information on sickle cell, thalassemia, and disorders of iron metabolism. Includes overviews of basic and clinical research, management and new developments in the field.

Northern California Comprehensive Thalassemia Center
www.thalassemia.com
Information for patients, families, health professionals and interested community members to improve the quality of life and survival of thalassemia patients.

Additional literature:

The Thalassemia Syndromes
by Weatherall, D.J. and Clegg, J,B,
Blackwell Publishers, 4th Edition, 2001
New edition that summarizes the evidence suggesting that the clinical picture of this disorder may result from several different inherited defects of globin synthesis. Includes historical background on the field.

~ TOURETTE SYNDROME ~

Tourette Syndrome Association, Inc.
42-40 Bell Boulevard
Bayside, NY 11361
Phone: 800-237-0717/718-224-2999
Fax: 718-279-9596
Email: ts@ts-usa.org
www.tsa-usa.org

In Connecticut:
Connecticut Chapter
Phone: 203-912-7310
Email: ts@tsact.org

Additional web resources:

Tourette-Syndrome.com
www.tourette-syndrome.com
Comprehensive online community devoted to children and adults with Tourette syndrome.

Additional literature:

Tourette's Syndrome: Finding Answers and Getting Help
by Waltz, M.
Patient Center Guides, 2001
A consumer guide offering help to families living and dealing with Tourette's.

Icy Sparks
by Rubio, G.H.
Penguin Books, 2001
This book is about a young girl who has Tourette's, but doesn't know it. She grows up in the back hills of Kentucky and no one knows what to make of her.

~ TREACHER COLLINS SYNDROME ~

Treacher Collins Foundation
P.O. Box 683
Norwich, VT 05055
Phone: 800-823-2055/802-649-3050
Fax: not available
Email: hopecharkins@hotmail.com
www.treachercollinsfnd.org
No Connecticut chapter.

Additional web resources:

Reflections on Treacher Collins Syndrome
www.treachercollins.org
Website written by a pediatric resident with Treacher Collins syndrome. Includes many resource links.

~ TUBEROUS SCLEROSIS ~

Tuberous Sclerosis Alliance
801 Roeder Road
Suite 750
Silver Spring, MD 20910
Phone: 800-225-6872/301-562-9890
Fax: 301-562-9870
Email: info@tsalliance.org
www.tsalliance.org
No Connecticut chapter.

Additional web resources:

The Cardiff-Rotterdam Tuberous Sclerosis Mutation Database
www.archive.uwcm.ac.uk/uwcm/mg/tsc_db
Contains published mutations and polymorphisms in the TSC2 gene.

Additional literature:

Tuberous Sclerosis Complex: From Basic Science to Clinical Phenotypes
by Curatolo, P., editor
Cambridge University Press, 2003
Correlation between new genetic and basic science data and clinical presentation.

~ TURNER SYNDROME ~

Turner Syndrome Society of the United States
14450 TC Jester
Suite 260
Houston, TX 77014
Phone: 800-365-9944/832-249-9988
Fax: 832-249-9987
Email: manager@turner-syndrome-us.org
www.turner-syndrome-us.org

In Connecticut:
Connecticut Turner Syndrome Society
Contact Sandra Gittleman
57 Cianci Drive
Southington, CT 06489
Phone: 860-628-8729
Email: barry1157@aol.com

~ UREA CYCLE DISORDERS ~

National Urea Cycle Disorders Foundation
4841 Hill Street
La Canada, CA 91011
Phone: 800-386-8233/818-790-2460
Fax: 818-952-2184
Email: info@nucdf.org
www.nucdf.org
No Connecticut chapter.

Additional web resources:

Urea Cycle Disorders
www.meadjohnson.com/metabolics/ureacycle.html
Detailed overview of the conditions, therapies, and common disorders.

~ VELO-CARDIO-FACIAL SYNDROME ~

Velo-Cardio-Facial Syndrome Educational Foundation, Inc.
708 Jacobsen Hall
Upstate Medical University
Syracuse, NY 13210
Phone: 315-464-6590
Fax: 315-464-6593
Email: info@vcfsef.org
www.vcfsef.org
No Connecticut chapter.

Northeast VCFS Support Group
Contact Maureen Anderson
2 Lansing Drive
Salem, NH 03079
Phone: 603-898-6332
Fax: not available
Email: mladja@aol.com

Did You Know...?

Most leading causes of death have a genetic component.

~ VON HIPPEL-LINDAU SYNDROME ~

VHL Family Alliance
171 Clinton Road
Brookline, MA 02445
Phone: 800-767-4845/617-277-5667
Fax: 858-712-8712
Email: info@vhl.org
Web: www.vhl.org

In Connecticut:
Contact Northeast Chapter
Email: northeast@vhl.org

~ WILLIAMS SYNDROME ~

Williams Syndrome Association
P.O. Box 297
Clawson, MI 48017
Phone: 800-806-1871/248-244-2229
Fax: 248-244-2230
Email: info@willims-syndrome.org
www.williams-syndrome.org

Connecticut contact:
Holly Weston
495 Route 87
Columbia, CT 06237
Phone: 2860-228-1020
Email: hweston@williams-syndrome.org

Additional web resources:

The Williams Syndrome Comprehensive Web Site
www.wsf.org
Includes medical and general information, behavioral issues, research, and information for families.

ADDITIONAL LITERATURE:

UNDERSTANDING WILLIAMS SYNDROME: A GUIDE TO BEHAVIORAL PATTERNS AND INTERVENTIONS
By Semel, E. and Rosner, S.R.
Lawrence Erlbaum Associates, Inc., 2003
Includes basic information, neurogenetic implications, challenges and interventions.

JOURNEY FROM COGNITION TO BRAIN TO GENE: PERSPECTIVES FROM WILLIAMS SYNDROME
by Bellugi, U. and St. George, M.I.
MIT Press, 2001
Presents the work of a team of scientists using a multidisciplinary integrated approach to link genes with human behavior.

~ WILSON'S DISEASE ~

WILSON'S DISEASE ASSOCIATION INTERNATIONAL
1802 Brookside Drive
Wooster, OH 44691
Phone: 800-399-0266/330-264-1450
Fax: 509-757-6418
Email: wda@sssnet.com
www.wilsonsdisease.org

In Connecticut:
Lenore and Russell Sillery
152 Cheese Spring Road
Wilton, CT 06897
Phone: 203-762-2372/203-961-9993
Fax: 203-961-9993
Email: lsillery@sillery.com
www.sillery.com

ADDITIONAL WEB RESOURCES:

WILSON'S DISEASE RESOURCES AND INFORMATION
www.acsu.buffalo.edu/~drstall/wilsons.html
Website by physician who has Wilson's disease.

LOW COPPER DIET FOR WILSON'S DISEASE
www.gicare.com/pated/edtgs17.htm
Gastroenterologist's website with detailed nutritional information for Wilson's disease patients.

ADDITIONAL LITERATURE:

WILSON'S DISEASE: A CLINICIAN'S GUIDE TO RECOGNITION, DIAGNOSIS, AND MANAGEMENT
by Brewer, G.J.
Kluwer Academic Publishers, 2001.

WILSON'S DISEASE FOR THE PATIENT AND FAMILY: A PATIENT'S GUIDE TO WILSON'S DISEASE AND FREQUENTLY ASKED QUESTIONS ABOUT COPPER
by Brewer, G.J.
George J. Brewer, MD through Xlibris Publishing Services Provider, 2001

COMMON GENETIC TERMS

Allele: Variant forms of the same gene. Different alleles produce variations in inherited characteristics such as eye color or blood type.

Allelic heterogeneity: A single disorder, trait, or pattern of traits caused by different mutations within a gene.

Allelic variant: An alteration in the normal sequence of a gene, the significance of which may be unclear. Complete gene sequencing often identifies numerous allelic variants (sometimes hundreds) for a given gene.

Amino acid: Any of a class of 20 molecules that are combined to form proteins in living things. The sequence of amino acids in a protein, and therefore protein function, are determined by the genetic code.

Aneuploidy: The occurrence of one or more extra or missing chromosomes leading to an unbalanced chromosome complement.

Anticipation: The tendency in certain genetic disorders for individuals in successive generations to present at an earlier age and/or with more severe manifestations; often observed in disorders resulting from the expression of a trinucleotide repeat mutation that tends to increase in size and have a more significant effect when passed from one generation to the next.

Autosomal: Any of the chromosomes other than the sex-determining chromosomes (X and Y) or the genes on these chromosomes.

Autosomal dominant: A gene on one of the non-sex chromosomes that is always expressed, even if only one copy is present.

Autosomal recessive: Describes a trait or disorder requiring the presence of two copies of a gene mutation at a particular locus in order to express observable phenotype; specifically refers to genes on one of the 22 pairs of non-sex chromosomes.

Background risk: The proportion of individuals in the general population who are affected with a particular disorder or who carry a certain gene; often discussed in the genetic counseling process as a comparison to the patient's personal risk given his or her family history or other circumstances.

Band level: Terminology used in reference to cytognetic analysis. Refers to the total number of stripes, or bands, elicited on each chromosome with staining techniques. Band level is the total number of bands estimated to be present in a haplotype set (23) of chromosomes. When analysis is performed at an early stage of mitosis (prometaphase), chromosomes appear longer, with approximately 700-1200 bands. At a later stage of mitosis (metaphase), chromosomes are more condensed, with approximately 300-600 bands. At higher band levels, the greater resolution increases the ability to identify more subtle chromosomal abnormalities and their breakpoints.

Base pair: The two complementary, nitrogen-rich molecules held together by weak chemical bonds. Two strands of DNA are held together in the shape of a double helix by the bonds between their base pairs. Carrier: An individual who has a recessive, disease-causing allele at a particular locus on one chromosome of a pair and a normal allele at that locus on the other chromosome.

Carrier detection or carrier testing: Testing used to identify usually asymptomatic individuals who have a gene mutation for an autosomal recessive or X-linked recessive disorder.

Carrier frequency: The proportion of individuals in a population who have inherited a single copy of a specific recessive gene mutation.

Cell cycle: The complete series of events from one cell division to the next.

Centromere: The constricted portion of the chromosome at which the chromatids are joined and to which the spindle attaches during mitosis and meiosis.

Chromosome: Structure found in the nucleus of a cell, which contains the genes. Chromosomes come in pairs, and a normal human cell contains 46 chromosomes (23 pairs).

Coding region: Sequence of DNA consisting of a series of nucleotide bases (code) giving rise to the mature messenger RNA that will be translated into the specific amino acids of the protein product.

Codominance: Situation in which two different alleles for a genetic trait are both expressed.

Codon: In DNA or RNA, a sequence of three nucleotides that codes for a certain amino acid or signals the termination of translation (stop or termination codon).

Compound heterozygote: An individual who has two different abnormal alleles at a particular locus, one on each chromosome of a pair; usually refers to individuals affected with an autosomal recessive disorder.

Congenital: Present from birth, not necessarily genetic.

Consultand: The individual (not necessarily affected) who presents for genetic counseling and through whom a family with an inherited disorder comes to medical attention.

Cross over (meiosis): A specialized cell division in which a single diploid cell undergoes two nuclear divisions following a single round of DNA replication in order to produce four daughter cells, each with half the number of chromosomes as the original diploid cell. Meiosis occurs during the formation of gametes from diploid organisms and at the beginning of haplophase in those organisms that alternate between diploid and haploid generations.

Crossing over: The exchange of a segment of DNA between two homologous chromosomes during meiosis leading to a novel combination of genetic material in the offspring.

Cytogenetics: The study of the structure, function, and abnormalities of human chromosomes.

De novo gene mutation: An alteration in a gene that is present for the first time in one family member as a result of a mutation in the egg or sperm cell that led to that person's conception.

Deletion: Absence of a segment of DNA; may be as small as a single base or large enough to encompass one or more entire genes. Large deletions involving a whole segment of a chromosome may be detected by routine examination of the chromosomes; intermediate deletions involving a few genes may be detected by using fluorescent in situ hybridization (FISH); smaller deletions involving a portion of a gene may only be detected by analyzing the DNA.

Deoxyribonucleic acid: DNA. The molecules inside cells that make up the genes, that carry genetic information from one generation to the next.

Derivative chromosome: Term used to denote an abnormal chromosome consisting of segments from two or more chromosomes joined together as the result of a translocation, insertion, or other rearrangement.

Diploid: The normal number of chromosomes in a somatic cell; in humans, 46 chromosomes (22 pairs of autosomes plus two sex chromosomes).

Direct DNA analysis: Molecular genetic testing used to detect a genetic alteration associated with a specific disorder; direct DNA analysis is possible only when the gene, genes, or genomic region associated with a disorder is known.

Disease causing mutation: A gene alteration that causes or predisposes an individual to a specific disease.

DNA banking: The process through which DNA is extracted from any of a number of possible cell sources and stored indefinitely by freezing or refrigerating for future testing; done when a specific test is not presently available or when the decision to have testing has not been made.

Double heterozygote: An individual who has two different gene mutations at two separate genetic loci.

Embryo: An animal in the early prenatal stages of growth and differentiation, specifically refers to the developing human during the first trimester of pregnancy.

Euploid: Any chromosome number that is a multiple of the haploid number.

Exon: The protein-coding DNA sequence of a gene. Each exon codes for a specific portion of the complete protein. In some species (including humans), a gene's exons are separated by long regions of DNA (called introns) that have no apparent function.

False paternity: The situation in which the alleged father of a particular individual is not the biological father.

False positive: A test result which indicates that an individual is affected and/or has a certain gene mutation when he or she is actually unaffected and/or does not have the mutation; i.e., a positive test result in a truly unaffected individual.

Familial: Describes a trait that is observed with higher frequency within the same family, whether the etiology is genetic or environmental, or a combination of the two.

Family history: The genetic relationships and medical history of a family; when represented in diagram form using standardized symbols and terminology, usually referred to as a pedigree. A record of a person's current and past illnesses, and those of his or her parents, brothers, sisters, children, and other family members. A family history shows the pattern of certain diseases in a family, and helps to determine risk factors for those and other diseases.

Fertility: The capacity to conceive or to induce conception. It may refer to either the male or female.

Fetus: An animal in the early prenatal stages of growth and differentiation, specifically refers to the developing human during the second and third trimesters of pregnancy.

Fibroblast: A connective tissue cell that makes and secretes collagen proteins.

First-degree relative: Parent, sibling, or offspring of an individual. A relative that shares one half of the persons genes.

Flanking marker: An identifiable, polymorphic region of DNA located to the side of a gene, but not within the gene itself. Used in linkage analysis to track the coinheritance of the gene in question.

Fluorescent in situ hybridization: (FISH) A cytogenetic technique used to identify the presence of specific chromosomes or chromosomal regions.

Gametogenesis: The meiotic process by which mature eggs and sperm are formed. Oogenesis refers specifically to the production of ova (eggs) and spermatogenesis to the production of sperm.

Gene: The functional and physical unit of heredity passed from parent to offspring. Genes are pieces of DNA, and most genes contain the information for making a specific protein.

Gene expression: The detectable effect of a gene.

Gene product: Genes are transcribed into segments of RNA (ribonucleic acid), which are translated into proteins. Both RNA and proteins are products of the expression of the gene.

Gene therapy: Treatment of a genetic disorder by the insertion of usually genetically altered genes into cells especially to replace defective genes or to provide a specialized disease fighting function (as the destruction of tumor cells).

Genetic predisposition: Increased susceptibility to a particular disease due to the presence of one or more gene mutations, and/or a combination of alleles (haplotype), not necessarily abnormal, that is associated with an increased risk for the disease. Also refers to a family history that suggests an increased risk for the disease.

Genetic testing: Analyzing DNA to look for a genetic alteration that may indicate an increased risk for developing a specific disease or disorder.

Genomics: The study of the complete DNA sequence, containing all coding and non-coding genetic information and supporting proteins, in the chromosomes of an individual or species. The study of the sequence, structure, and function of the genome. The comprehensive analysis of all the genes, and their interactions, of an organism.

Genotype: The genetic constitution of an organism or cell; also refers to the specific set of alleles inherited at a locus.

Genotype-phenotype correlation: The association between the presence of a certain mutation or mutations (genotype) and the resulting physical trait, abnormality, or pattern of abnormalities (phenotype).

Germline: The cell line from which egg or sperm cells (gametes) are derived.

Germline mosaicism: Two or more genetic or cytogenetic cell lines confined to the precursor cells of the egg or sperm; also called gonadal mosaicism.

Germline mutation: The presence of an altered gene within the egg and sperm (germ cell) such that the altered gene can be passed to subsequent generations.

Haploid: Half the diploid or normal number of chromosomes in a somatic cell; the number of chromosomes in a gamete (egg or sperm) cell, which in humans is 23 chromosomes, one chromosome from each chromosome pair.

Heterozygote: With respect to a particular trait or condition, an individual who has inherited two different alleles, usually one normal and the other abnormal, at a particular locus.

Homologous chromosomes: A pair of particular chromosomes, normally one inherited from the mother and one from the father, containing the same genetic loci in the same order.

Homozygote: With respect to a particular trait or condition, an individual who has inherited identical alleles at a particular locus.

Imprinting: A phenomenon in which the disease phenotype depends on which parent passed on the disease gene. This is determined by the process by which maternally and paternally derived chromosomes are uniquely chemically modified leading to different expression of a certain gene or genes on those chromosomes depending on their parental origin.

Infertility: Incapable of or unsuccessful in achieving pregnancy over a considerable period of time (as a year) in spite of determined attempts by heterosexual intercourse without contraception.

Inheritance pattern: The manner in which a particular genetic trait or disorder is passed from one generation to the next. Autosomal dominant, autosomal recessive, X-linked dominant, X-linked recessive, multifactorial, and mitochondrial inheritance are examples.

Insertion: A chromosome abnormality in which material from one chromosome is inserted into another chromosome; or a mutation in which a segment of DNA is inserted into a gene or other segment of DNA, potentially disrupting the coding sequence.

Intrafamilial variability: Variation in clinical presentation of a particular disorder among affected individuals within the same immediate or extended family.

Intron: Non-coding sequence of DNA removed from mature messenger RNA prior to translation. DNA initially transcribed to messenger RNA consists of coding sequences (exons) and non-coding sequences (introns); introns are spliced out of the messenger RNA prior to translation, leaving only the exons to ultimately encode the amino acid product.

Inversion: A chromosomal rearrangement in which a segment of genetic material is broken away from the chromosome, inverted from end to end, and re-inserted into the chromosome at the same breakage site. These may be balanced, with no loss or gain of genetic material, or unbalanced, with missing or extra genetic material.

Karyotype: A photographic representation of the chromosomes of a single cell, arranged in pairs based on their banding pattern and size according to a standard classification.

Kindred: An extended family; term often used in linkage studies to refer to large families.

Linkage analysis: Indirect DNA analysis. The use of several DNA sequence polymorphisms (normal variants) that are near or within a gene of interest to track the inheritance of a disease-causing mutation in that gene within a family.

Linkage disequilibrium: In a population, co-occurrence of a specific DNA marker and a disease at a higher frequency than would be predicted by chance alone.

Locus: The physical site or location of a specific gene on a chromosome.

Locus heterogeneity: A single disorder, trait, or pattern of traits caused by mutations in genes at different chromosomal loci.

Lyonization: In females, the phenomenon in which one X chromosome (either maternally or paternally derived) is randomly inactivated in early embryonic cells, with fixed inactivation in all descendant cells; first described by the geneticist Mary Lyon.

Mapped gene: A gene or phenotype whose relative position on a segment of DNA or on a chromosome has been established.

Marker: An identifiable segment of DNA with enough variation between individuals that its inheritance and co-inheritance with alleles of a given gene can be traced; used in linkage analysis.

Marker chromosome: A small chromosome, usually containing a centromere, occasionally seen in tissue culture, often in a mosaic state. A marker chromosome may be of little clinical significance however may create an imbalance for whatever genes are present. Clinical significance, particularly if found in a fetal karyotype, is often difficult to assess.

Maternal contamination: The situation which occurs in prenatal testing in which a fetal sample becomes contaminated with maternal cells, which can confound interpretation of the results of genetic analysis.

Meiosis: Specialized cell division in which a single diploid cell undergoes two nuclear divisions following a single round of DNA replication in order to produce four daughter cells, each that contain half the number of chromosomes as the original diploid cell. Meiosis occurs during the formation of gametes from diploid organisms.

Messenger RNA: RNA that serves as a template for protein synthesis.

Microdeletion syndrome: A syndrome caused by a chromosomal deletion spanning several genes that is too small to be detected under the microscope using conventional cytogenetic techniques. Other methods of DNA analysis can sometimes be employed to identify the deletion.

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.

Monosomy: The presence of only one chromosome from a pair; partial monosomy refers to the presence of only one copy of a segment of a chromosome.

Mosaicism: The post-fertilization occurrence of two or more cell lines with different genetic or chromosomal constitutions within a single individual or tissue.

Multifactorial inheritance: The combined contribution of one or more often unspecified genes and environmental factors, often unknown, in the causation of a particular trait or disease.

Mutation: Any alteration in a gene from its natural state; may be disease causing or a benign, normal variant.

New mutation: An alteration in a gene that is present for the first time in one family member as a result of a mutation in a germ cell (egg or sperm) of one of the parents or in the fertilized egg itself.

Non-disjunction: The failure of homologous chromosomes or chromatids to segregate during mitosis or meiosis, with the result that one daughter cell has both of a pair of parental chromosomes or chromatids, and the other has none.

Nonsense mutation: A single base pair substitution that prematurely codes for a stop in amino acid translation (stop codon).

Nucleotide: A molecule consisting of a nitrogenous base (adenine, guanine, thymine, or cytosine in DNA; adenine, guanine, uracil, or cytosine in RNA), a phosphate group, and a sugar (deoxyribose in DNA; ribose in RNA). DNA and RNA are polymers of many nucleotides.

Obligate carrier or obligate heterozygote: An individual who may be clinically unaffected but who must carry a gene mutation based on analysis of the family history; usually applies to disorders inherited in an autosomal recessive and X-linked recessive manner.

Oncogene: A gene that normally directs cell growth. If altered, an oncogene can promote or allow the uncontrolled growth of cancer. Alterations can be inherited or caused by an environmental exposure to carcinogens.

Oncosuppressor gene: Also called a tumor suppressor gene. Genes in the body that can suppress or block the development of cancer.

Paracentric inversion: A chromosomal inversion in which the breakpoints are confined to one arm of a chromosome; the inverted segment does not span the centromere.

Parentage testing: The process through which DNA sequences from a particular child and a particular adult are compared to estimate the likelihood that the two individuals are related; DNA testing can reliably exclude but cannot absolutely confirm an individual as a biological parent.

Pedigree: A diagram of the genetic relationships and medical history of a family using standardized symbols and terminology.

Penetrance: The proportion of individuals with a mutation causing a particular disorder who exhibit clinical symptoms of that disorder; a condition is said to have complete penetrance if clinical symptoms are present in all individuals who have the disease-causing mutation, and to have reduced or incomplete penetrance if clinical symptoms are not always present in individuals who have the disease-causing mutation.

Pericentric inversion: A chromosomal inversion in which the breakpoints occur on both arms of a chromosome and therefore the inverted segment spans the centromere.

Phenotype: The observable physical and/or biochemical characteristics of the expression of a gene; the clinical presentation of an individual with a particular genotype.

Pleiotropy: Multiple, often seemingly unrelated, physical effects caused by a single altered gene or pair of altered genes.

Point mutation: An alteration in DNA sequence caused by a single nucleotide base change, insertion, or deletion.

Polygenic: Genetic condition resulting from the combined action of alleles of more than one gene (e.g., heart disease, diabetes, and some cancers). Although such disorders are inherited, they depend on the simultaneous presence of several alleles; thus the hereditary patterns usually are more complex than those of single-gene disorders.

Polymerase chain reaction (PCR): A method of creating copies of specific fragments of DNA. PCR rapidly amplifies a single DNA molecule into many billions of molecules.

Polymorphism: Natural variations in a gene, DNA sequence, or chromosome that have no adverse effects on the individual and occur with fairly high frequency in the general population.

Polyploidy: An increase in the number of haploid sets (23) of chromosomes in a cell. Triploidy refers to three whole sets of chromosomes in a single cell (in humans, a total of 69 chromosomes per cell); tetraploidy refers to four whole sets of chromosomes in a single cell (in humans, a total of 92 chromosomes per cell).

Preimplantation diagnosis: A procedure used to genetically test one cell removed from early embryos conceived by in vitro fertilization. Typically performed to identify specific genetic abnormalities, and then transferring to the mother's uterus only those embryos determined not to have inherited the condition in question.

Premutation: In disorders caused by trinucleotide repeat expansions, an abnormally large allele that is not associated with clinical symptoms but that can expand into a full mutation when transmitted to offspring (full mutations are associated with clinical symptoms of the disorder).

Prenatal diagnosis: Testing performed during pregnancy to determine if a fetus is affected with a particular disorder. Chorionic villus sampling (CVS), amniocentesis, periumbilical blood sampling (PUBS), ultrasound, and fetoscopy are examples of procedures used either to obtain a sample for testing or to evaluate fetal anatomy.

Presymptomatic testing: Testing of an asymptomatic individual in whom the identification of a gene mutation indicates development of findings related to a specific diagnosis at some future point. A negative result excludes the diagnosis.

Private mutation: A distinct gene alteration observed in a single family.

Proband: The affected individual through whom a family with a genetic disorder is ascertained; may or may not be the consultand (the individual presenting for genetic counseling).

Rearrangement: A structural alteration in a chromosome, usually involving breakage and reattachment of a segment of chromosome material, resulting in an abnormal configuration, may be balanced or unbalanced. Examples include inversion and translocation.

Recessive: A gene which will be expressed only if there are 2 copies or, for a male, if one copy ispresent on the X chromosome.

Reciprocal translocation: A segment of one chromosome is exchanged with a segment of another chromosome of a different pair.

Recombination: The exchange of a segment of DNA between two homologous chromosomes during meiosis leading to a novel combination of genetic material in the offspring.

Recurrence risk: The likelihood that a trait or disorder present in one family member will occur again in other family members in the same or subsequent generations.

Reflex testing: Follow-up testing automatically initiated when certain test results are observed in the laboratory; used to clarify or elaborate on primary test results.

Restriction fragment length polymorphism (RFLP) analysis: Fragment of DNA of predictable size resulting from cutting of a strand of DNA by a given restriction enzyme. DNA sequence mutations that destroy or create the sites at which a restriction enzyme cuts DNA change the size and number of DNA fragments resulting from digestion by a given restriction enzyme.

Ring chromosome: Abnormal chromosomes in a circular configuration.

Risk assessment: Calculation of an individual's chance of having inherited a certain gene mutation, of developing a particular disorder, or of having a child with a certain disorder based upon analysis of multiple factors including genetic test results, family medical history, and/or ethnic background.

Robertsonian translocation: The joining of two acrocentric chromosomes at the centromeres with loss of their short arms to form a single abnormal chromosome; acrocentric chromosomes are the Ychromosome and chromosome numbers 13, 14, 15, 21, and 22.

Screening: Testing designed to identify individuals in a given population who are at higher risk of having or developing a particular disorder, or carrying a gene for a particular disorder.

Second-degree relative: A relative with whom one quarter of an individual's genes is shared (i.e., grandparent, grandchild, uncle, aunt, nephew, niece, half-sibling).

Segregation: The separation of the homologous chromosomes and their random distribution to the gametes at meiosis.

Sensitivity: The frequency with which a test yields a positive result when the gene mutation in question is actually present in the individual being tested.

Somatic mutation: Alterations in DNA that occur after conception. Somatic mutations can occur in any of the cells of the body except the germ cells (sperm and egg) and therefore are not passed on to children.

Specificity: The frequency with which a test yields a negative result when the gene mutation in question is not present in the individual being tested.

Sporadic: Denoting either a genetic disorder that occurs for the first time in a family due to a new mutation or the chance occurrence of a non-genetic disorder or abnormality that is not likely to recur in a family.

Stem cells: Relatively undifferentiated cells from which other types of cells can develop.

Substitution: A type of mutation due to replacement of one nucleotide in a DNA sequence by another nucleotide or replacement of one amino acid in a protein by another amino acid.

Telomere: The segment at the end of each chromosome which has a specialized structure and is involved in chromosomal replication and stability.

Transcript: A sequence of RNA produced by transcription from a DNA template.
Transcription factor: A protein that binds to regulatory regions and helps control gene expression.

Transfer RNA: Small RNA molecules that bond with amino acids and transfer them to the ribosomes, where proteins are assembled according to the genetic code carried by mRNA.

Translation: The process of forming a protein molecule at a ribosomal site of protein synthesis from information contained in messenger RNA.

Translocation: A chromosome alteration in which a whole chromosome or segment of a chromosome becomes attached to or interchanged with another whole chromosome or segment. These may be balanced, with no loss or gain of genetic material, or unbalanced, with extra or missing genetic material.

Trinucleotide repeat: Sequences of three nucleotides repeated in tandem on the same chromosome a number of times. A normal, polymorphic variation in repeat number with no clinical significance commonly occurs between individuals; however, repeat numbers over a certain threshold can, in some cases, lead to adverse effects on the function of the gene, resulting in genetic disease.

Trisomy: The presence of a single extra chromosome, yielding a total of three chromosomes of that particular type instead of a pair. Partial trisomy refers to the presence of an extra copy of a segment of a chromosome.

Trisomy rescue: The phenomenon in which a fertilized ovum initially contains 47 chromosomes but loses one of the trisomic chromosomes in the process of cell division such that the resulting daughter cells and their descendants contain the normal number of chromosomes.

Uniparental disomy: (UPD) The situation in which both members of a chromosome pair, or segments of a chromosome pair, are inherited from one parent and neither is inherited from the other parent. Uniparental disomy can result in an abnormal phenotype in some cases.

UPD testing: Testing used to identify whether specific chromosomes are maternally or paternally derived; can aid in confirming the clinical diagnosis of certain disorders for which UPD is a possible underlying etiology.

Variable expression: Variation in clinical features (type and severity) of a genetic disorder between individuals with the same gene alteration, even within the same family.

X-linked dominant: Describes a dominant trait or disorder caused by a mutation in a gene on the X chromosome.

X-linked lethal: A disorder caused by a dominant mutation in a gene on the X chromosome that is observed almost exclusively in females because it is almost always lethal in males who inherit the gene mutation.

X-linked recessive: A mode of inheritance in which a mutation in a gene on the X chromosome causes the phenotype to be expressed in males who are hemizygous for the gene mutation or in females who are homozygous for the gene mutation. Carrier females, who have only one copy of the mutation, do not usually express the phenotype, although differences in X-chromosome inactivation can lead to varying degrees of clinical expression in carrier females.

Zygosity testing: The process through which DNA sequences are compared to assess whether individuals born from a multiple gestation (twins, triplets, etc.) are monozygotic (identical) or dizygotic (fraternal); often used to identify a suitable donor for organ transplantation or to estimate disease susceptibility risk if one sibling is affected.

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