New England Regional Genetics Group, Inc.

genetics resources: a regional directory

FOR MORE INFORMATION, COMMENTS OR QUESTIONS, PLEASE CONTACT:

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Mary-Frances Garber M.S., C.G.C
Executive Assistant
P.O. Box 920288
Needham, MA 02492-0288
Phone: 781-444-0126
Fax: 781-444-0127
Email: mfgnergg@verizon.net
Web site: www.nergg.org

This resource directory is a supplement to and not a substitute for medical advice. Patients with specific questions about genetic counseling or testing should contact their healthcare provider or a genetics clinic.

NERGG, Inc.

Continuing the mission of the NEW ENGLAND REGIONAL GENETICS GROUP

Thomas Brewster, MD
Project Director
Regional Genetics Services and NBS Collaborative

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NEW ENGLAND REGIONAL GENETICS AND NEWBORN SCREENING COLLABORATIVE
HRSA GRANT # 1U22MC03959
09/01/2004-05/31/2007
Thomas Brewster, MD
Project Director

Dear Reader:

I am pleased to present the New England Regional Genetics Group, Inc.'s latest endeavor, Genetics Resources: A Regional Directory.

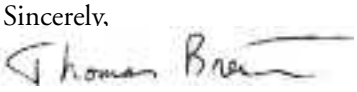
Advances in genetics hold the promise of great benefits to the health of New England residents. Yet there is a growing need for enhanced genetics knowledge among physicians, other health care providers, and the general public. To help meet this need, the Department of Public Health in Connecticut developed a genetics resources directory, and the health departments within our region supplemented it with their information. By joining forces, this wealth of information was created for accessing local, regional, and national genetics resources. This directory was developed to help New England 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 listings for national genetics organizations. It also includes support groups for various conditions and lists specialized resources and supports for each condition whenever possible.

Each state has a listing of its genetic services, including clinical, public health and laboratory.

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,



Thomas Brewster, MD

ACKNOWLEDGEMENTS

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PREFACE

Rapid developments in genetics research have led increasingly to the integration of genetics into medical practice. 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 affect people in unique ways. Such information can predict events that occur years in the future, or not at all. It may predict the future of other family members, and has the potential to challenge, and even stigmatize both individuals and families.

Genetics Resources: A Regional Directory, is the product of a multi-year genetics planning initiative at the Connecticut Department of Public Health. Its development was prompted by concern that a lack of general, current genetics knowledge among the public and medical professionals could result in missed health promotion opportunities. The expansion of the directory to include resources outside Connecticut was undertaken by the New England Public Health Genetics Education Collaborative of the NERGG, Inc. Updating this directory is an important part of a strategy to increase genetics literacy for all.

This directory is intended to help 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 to locate and access resources. The information and resources in this Directory extend beyond those related to traditional public health efforts in screening newborns for inherited disorders. Rather they encompass the impact of genetics across the entire lifespan. Wherever possible, additional sources of published information accompany listings related to a particular diagnosis or genetic condition.

This second draft of Genetics Resources: A Regional Directory, will require revision and updating with new resources as the "genomics revolution" continues to expand, creates new challenges and necessitates the creation of new resources in response to them.

As planning and preparation continue to address the impact of genetic developments on public health in New England, the need for sharing timely, accurate and useful information will grow. Public health departments will continue to play a critical role in addressing these challenges.

*Preface originally prepared by the CT Department of Public Health, 2004

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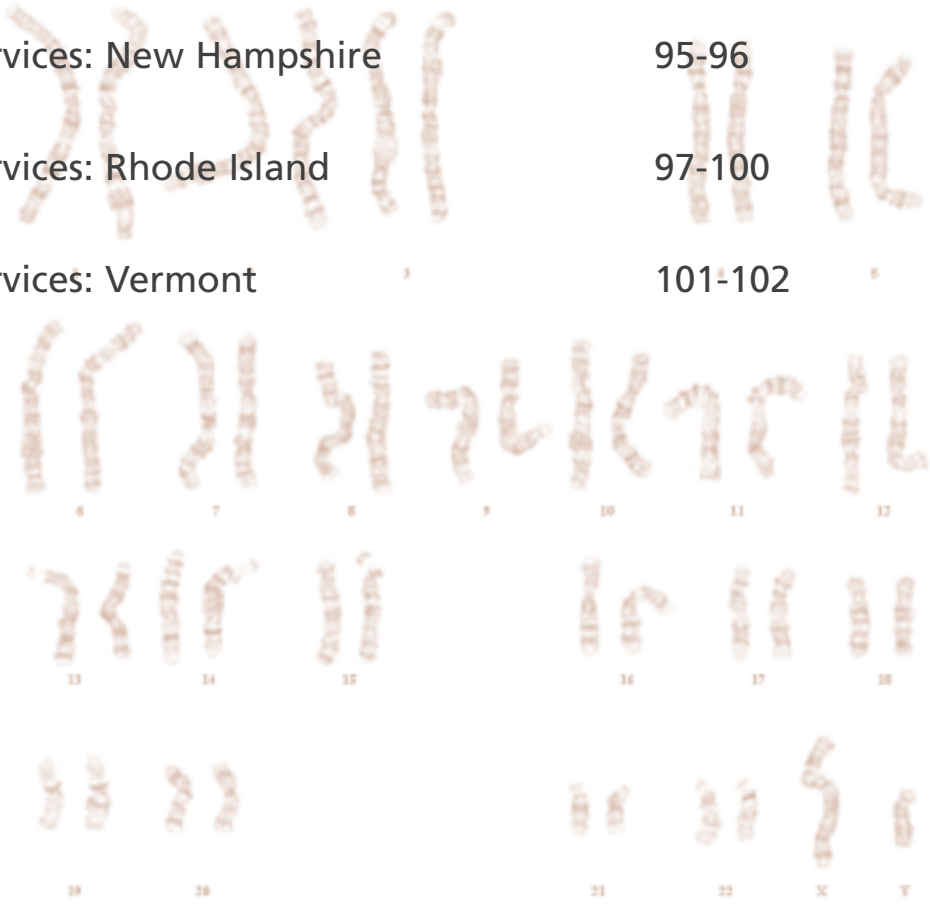
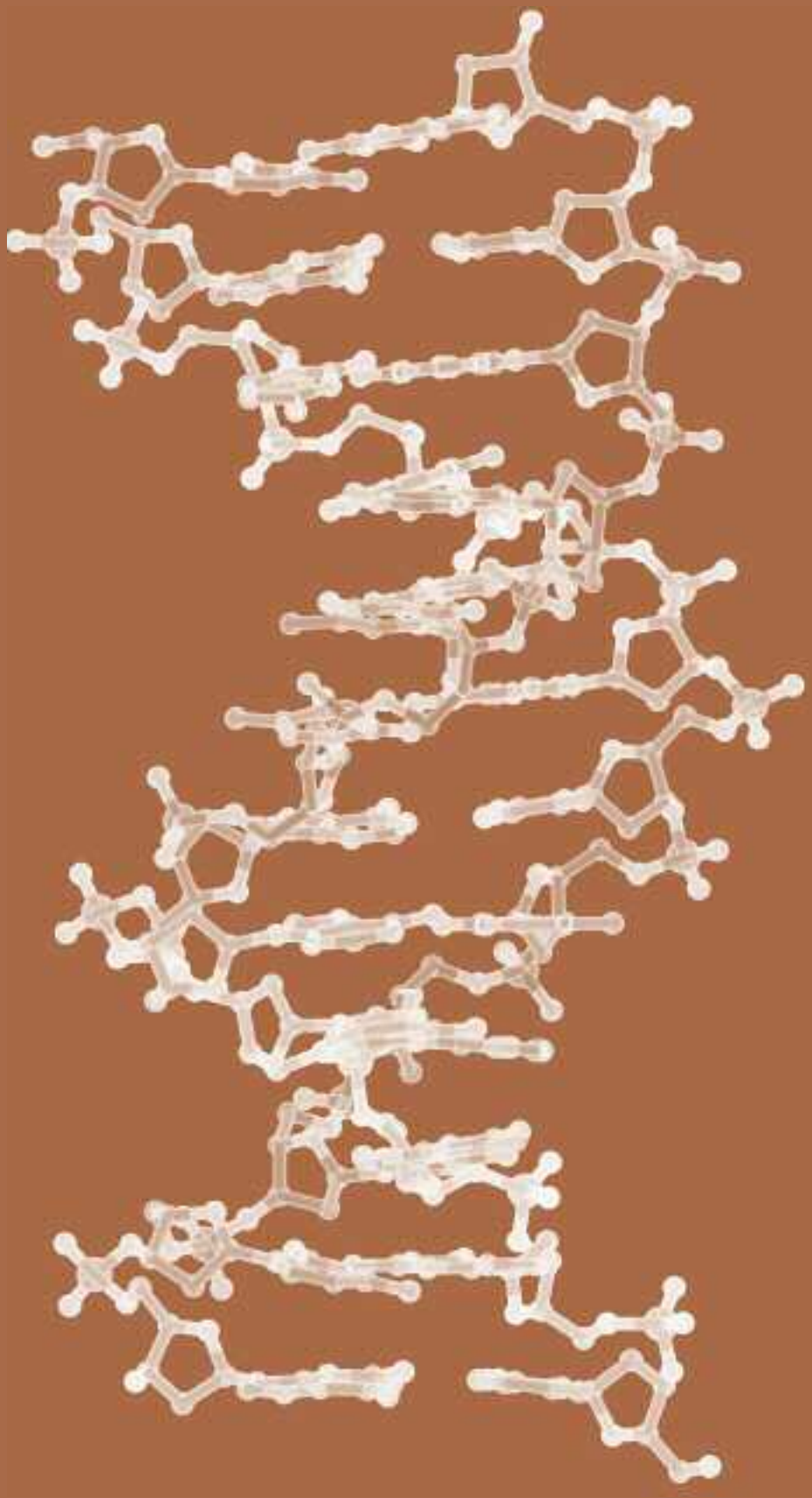




I.

GENETICS THROUGHOUT THE LIFESPAN

Human genetics. Understanding ourselves and our make-up from the most basic perspective can be overwhelming. The study of genetics (single genes and their effects), once considered obscure has now been confounded by genomics--the study of all the genes in an individual and how they interact with each other and with the environment to create the complexity of life. Even today, with the completion of the Human Genome Project and the related media exposure, the risk for birth defects or genetic disease is often viewed as a "one in-a-million" possibility. Many people believe that genomics is unrelated to their daily lives. The reality, however, is that genomic medicine-- using genetic information to diagnose and treat disease--is here to stay and most people will benefit from genetic testing and information at some time in their lives.

Genetics is the study of heredity. It concerns the transfer of qualities or traits from one generation to the next. The basic units of heredity are genes. Genes are made of DNA (deoxyribonucleic acid) and come in pairs, and are packaged in structures called chromosomes, which collectively form an organism's genome.

Most cells in an individual contain 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 contains only about 20,000-25,000 genes. Humans have a total of 46 chromosomes representing 23 pairs. We get 23 chromosomes from our mother, via the egg, and 23 chromosomes from our father, via the sperm. The joining of egg and sperm at conception results in a fertilized egg with its full complement of chromosomes. Of the 23 pairs of chromosomes, 22 pairs are called the autosomes. The 23rd pair consists of the sex chromosomes. Females have two "X" chromosomes, and males have one "X" chromosome and one "Y" chromosome.

Most things about us, including, in part, our health, are determined by our genes. Our genetic blueprint not only determines whether or not our children might have a serious genetic disorder, but may also determine our chances of developing a chronic disease or our susceptibility to an infectious disease.

Traditionally, genetic testing and counseling have centered on prenatal and pediatric genetic diagnoses. This is changing: genetics is rapidly expanding into other medical fields, including adult medicine. However, there are key times throughout the lifespan that genetic issues are more likely to be encountered.

Historically, pregnant 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, regardless of their age. Parental carrier testing is routinely available for a variety of genetic diseases that are more common in specific racial and ethnic groups. Sperm and egg donors are often asked to submit to a variety of genetic tests. When couples are known to be at risk for a baby with genetic disease, pre-implantation genetic diagnosis affords the option of testing fertilized eggs in-vitro prior to transfer to the uterus.

Each year, millions of newborns receive newborn screening for various genetic, metabolic, and infectious disorders. The goal of State newborn screening programs is to identify affected newborns before the onset of symptoms. Medical treatment can be promptly initiated to prevent irreversible medical problems or even death. Information about each state's newborn screening program is available on its health department's web site, (see Genetic Services section of this directory).

The identification of susceptibility genes for common adult-onset genetic diseases is moving the field of genetics in a new and demanding direction. Diabetes, certain cancers, and Alzheimer's disease are a few conditions for which a genetic component has been identified. Of the top ten leading causes of death in the U.S., all but one (injuries) has a genetic component.

Pre-symptomatic and susceptibility genetic testing have created an entirely new concept-- Healthy individuals who are expected to develop or have an increased risk to develop a disorder. The psychosocial implications of this information represent a continuing challenge to the genetics profession.

Genetics professionals have valid ethical concerns about genetic testing and genomic medicine. They worry about the possible misuse of information about genetic traits as a basis for denial of health coverage or employment. Other ethical issues in the field of genetics include: gene patents, the storage of DNA samples, and testing minors for adult-onset conditions. Given these concerns, detailed counseling, informed consent, and confidentiality are essential components of the genetic testing process.

II.

UMBRELLA ORGANIZATIONS & REFERENCES

A. Umbrella Organizations and References:

Genetic Alliance

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

The Genetic Alliance is an international coalition comprising 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: 888-663-4637or 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.

Connecticut Chapters:
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

Maine Chapters:
60 Gray Road, Unit #8
Falmouth, ME 04105-2024
Phone: 207-878-1199
Fax: 207-878-1166
Email: ME376@marchofdimes.com
www.marchofdimes.com/maine/

Maine Satellite:
51 Fern Street
Turner, ME 04282
Phone: 207-225-6155
Fax: 207-225-6156

Northern Division:
12 Acme Road
Brewer, ME 04412
Phone: 207-989-3376
Fax: 207-989-5500

Massachusetts Chapter:
114 Turnpike Road, Suite 202
Westborough, MA 01581
Phone: 508-366-9066
Fax: 508-366-6634
Email: MA625@marchofdimes.com
www.marchofdimes.com/massachusetts/

Southeastern Division:
895 Mary Dunn Rd.
Hyannis, MA 02601
Phone: 508-790-1093
Fax: 508-790-1094

New Hampshire Chapter:
22 Bridge Street
Concord, NH 03301
Phone: 603-228-0317
Fax: 603-228-0318
Email: NH634@marchofdimes.com
www.marchofdimes.com/newhampshire/

Rhode Island Chapter:
260 West Exchange Street, Suite 002
Providence, RI 02903
Phone: 401-454-1911
Fax: 401-454-1970
Email: RI440@marchofdimes.com
www.marchofdimes.com/rhodeisland/

Vermont Chapter:
107 N Main Street
Barre, VT 05641
Phone: 802-479-3265
Fax: 802-479-0957
Email: VT478@marchofdimes.com
www.marchofdimes.com/vermont/

New England Regional Genetics Group, Inc.

PO Box 920288
Needham, MA 02492
Phone: 781-444-0126
Fax: 781-444-0127
Email: mfgnergg@verizon.net
www.nergg.org

NERGG, Inc., formerly known as the New England Regional Genetics Group, is a consortium of genetic health services providers, representatives of the New England public health community, and support groups and individuals with interest in genetics. NERGG, Inc.'s purpose is to promote the health of both children and adults by increasing the awareness of genetic concerns, the understanding of the role of genetics in health care, and the availability of appropriate services. NERGG, Inc. has a particular interest in programs with regional perspectives and activities. NERGG, Inc. is committed to, and actively involved in: public and professional education, data collection and dissemination, quality improvement in clinical care, application of new technologies, and the social and ethical issues associated with human genetics.

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 (e.g., tobacco and alcohol), and promotion of breastfeeding.

National Organization for Rare Disorders (NORD)

55 Kenosia Avenue
P.O. Box 1968
Danbury CT 06813
Phone: 800-999-6673 or 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" diseases 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.

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 Newborn Screening Status Report

www.genes-rus.uthscsa.edu/nbsdisorders.pdf

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, and drug information.

National Library of Medicine

www.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 web site for support information about genetic conditions, disease summaries, gene and chromosome summaries, and a glossary of genetics and medical terminology.

National Human Genome Research Institute

www.genome.gov

An on-line listing of information for researchers, consumers, support and advocacy groups, health professionals, and policy makers. Includes links to clinical genetics resources, search engines and family history tools, support 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

This comprehensive on-line service, based at University of Kansas Medical Center, provides clinical, research, and educational resources for genetic counselors, clinical geneticists and medical geneticists. This site 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; listings of upcoming professional meetings in genetics; and information on careers in human genetics.

C. Professional Genetics Societies

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 ACMG 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.

American Society for Human Genetics

9650 Rockville Pike
Bethesda, MD 20814
Phone: 866-HUM-GENE or 301-634-7300
Email: society@ashg.org
www.ashg.org

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

National Society of Genetic Counselors (NSGC)

Executive Office
401 N. Michigan AVenue
Chicago, IL 60611
Phone: 312-321-6834
Fax: 312-673-6972
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.

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

NCHPEG is 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, support 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

Covers 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 of 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. Information about every known inherited disease.

The Practical Guide to the Genetic Family History
by Bennett, R. L.
Wiley-Liss, 1999

Thorough and well-organized with a complete index and references; especially useful for the primary care physician.

Practical Genetic Counseling
by Harper, P. S.
Arnold Publishers, 6th Edition, 2004

This book provides up to date guidance through the profusion of new genetic information and the associated psychosocial and ethical considerations and concerns.

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., Blackmon, L. and Rosenatien, B.
Textbook paperback, 2002

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.

III.

SUPPORT ORGANIZATIONS & RESOURCES

This section lists contact information for support groups, including New England state chapters where appropriate. If your state of interest does not appear to have a chapter for a particular group, we suggest contacting that group's national office for assistance.

Little People of America

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

District 1 (CT, ME, MA, NH, RI, VT)
Director, Casey Hubelbank
Phone: 203-641-9760
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:

Little People: Learning to See the World Through My Daughter's Eyes

by Kennedy, D.
Rodale Books, 2003

Written by the father of a child with achondroplasia, this book confronts the deepest of parental fears: What if my child is different? His search for an answer provides a penetrating look at how our culture of diversity clashes with the reality of disability and the belief that we have a right to the so-called perfect child.

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

Connecticut Contact: Eileen Corey
Phone: 203-283-3546
Email: ecorey76@hotmail.com

Maine Contact: Rachel Bennett
Phone: 207-549-5583
Email: rbennett_sw@yahoo.com

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
10500 SW Starr Drive
Tualatin, OR 97062
Phone: 503-885-0455
Email: alagille@earthlink.net
www.alagille.org

ADDITIONAL WEB RESOURCES:

Cincinnati Children's Hospital Medical Center page on Alagille syndrome
www.cincinnatichildrens.org/svc/pro/liver/diseases/alagille-syndrome.htm

Information on genetics, signs and symptoms, diagnosis, treatment, and prognosis of Alagille syndrome.

ALBINISM AND HYPOPIGMENTATION

National Organization for Albinism and Hypopigmentation (NOAH)
P.O. Box 959
East Hampstead, NH 03826
Phone: 800-473-2310 or 603-887-2310
Fax: 800-648-2310
Email: info@albinism.org
www.albinism.org
For local resources, call national office.

Hermansky-Pudlak Syndrome Network, Inc.
1 South Road
Oyster Bay, NY 11771
Contact: Donna Appell
Phone: 800-789-9477 or 516-922-4022
Fax: 516-624-0640
Email: dappell@hps.network.org
www.hermansky-pudlak.org
www.hpsnetwork.org

Chediak Higashi Syndrome Association
A division of the Hermansky-Pudlak Syndrome Network.
www.chediak-higashi.org

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 the goal of understanding the causes and effects of albinism and other forms of pigment loss in humans.

ADDITIONAL LITERATURE:

Too White to be Black and Too Black to be White
by Edwards, L.G.; Authorhouse, 2001

Expresses the true emotion and life experiences of the 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 or 312-335-8700
Fax: 312-335-1110
Email: info@alz.org
www.alz.org

Connecticut Chapter:
279 New Britain Road
P.O. Box 454
Kensington, CT 06037
Phone numbers:
Helpline: 866-363-6679
Business line: 860-282-2828
Fax: 860-828-2417
www.alzct.org (links to 98 support groups)

Maine Chapter:
163 Lancaster St; Suite 160B
Portland, ME 04101
Helpline: 800-660-2871
Business Line: 207-772-0115
Fax: 207-772-0354
www.mainealz.org

Massachusetts Chapter:
311 Arsenal Street
Watertown, MA 02472
Helpline(s): 800-548-2111
Business Line: 617-868-6718
Fax: 617-868-6720
www.alzmass.org

Rhode Island Chapter:
245 Waterman Street, Suite 306
Providence, RI 02906
Helpline(s): 800-244-1428
Business Line: 401-421-0008
Fax: 401-421-0115
www.alz-ri.org

Vermont and New Hampshire Chapter:
10 Ferry Street Suite 42B
Concord, NH 03301
Helpline(s): 800-272-3900
Business Line: 802-477-7000
Fax: 802-477-7003
www.alzvtnh.org

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
(Members section links to local support groups by state)

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 information 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

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 Mifflin, 2003

Provides hundreds of practical tips to ease life for everyone coping with Alzheimer's disease.

AMYOTROPHIC LATERAL SCLEROSIS

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

Connecticut Chapter:
4 Oxford Road
Unit D1
Milford, CT 06460
Phone: 877-257-2281 or 203-874-5050
Fax: 203-874-7070
Email: jargento@alsact.org
Contact Person - Joan Argento
www.alsact.org

Massachusetts Chapter:
75 McNeil Way, Ste. 310
Dedham, MA 02026
Phone: 781-326-8884
Fax: 781-326-4940
Contact Person - Judy Teplow
Emails: Judy.Teplow@als-ma.org
www.als-ma.org

Northern New England Chapter: (covers Maine, Vermont and New Hampshire)
The Concord Center
10 Ferry St., Ste. 438
Box 314
Concord, NH 03301
Phone: 603-226-8855
Fax: 603-226-8890
Contact Person - Cheryl Flanders
Email: **nh@alsanne.org**
www.alsanne.org

Rhode Island Chapter:
Gateway Plaza
1637 Warwick Ave.
Warwick, RI 02889
Phone: 401-732-1609
Fax: 401-732-2577
Contact Person - Lynn McGovern
Email: alsarilm@aol.com
www.alsari.org

ASTHMA

Allergy and Asthma Network—Mothers of Asthmatics, Inc.

2751 Prosperity Avenue
Suite 150
Fairfax, VA 22031
Phone: 800-878-4403 or 703-641-9595
Fax: 703-573-7794
Email: info@aanma.org
www.breatherville.org

American Lung Association

61 Broadway, 6th floor
New York, NY 10006
Phone: 800-586-4872 or 212-315-8700
Fax: 212-315-8800
Email: tlestrange@lungusa.org
www.lungusa.org

Connecticut Chapter:
American Lung Association of Connecticut
45 Ash Street
East Hartford, CT 06108
Phone: 800-586-4872 or 860-289-5401
FAX: 860-289-5405
Email: bcase@alact.org
www.alact.org

Massachusetts Chapters:
American Lung Association of Massachusetts
460 Totten Pond Road, Suite 400
Waltham, MA 02451-1991
Phone: 781-890-4262
Fax: 781-890-4280
Email: info@lungma.org
www.lungusa2.org/massachusetts/index.html

American Lung Association of Massachusetts (Western Area)
393 Maple Street
Springfield, MA 01105-1954
Phone: 413-737-3506
Fax: 413-737-3511
Email: info@lungma.org
www.lungusa2.org/massachusetts/index.html

Maine Chapter:
American Lung Association of Maine
122 State Street
Augusta, ME 04330
Phone: 207-622-6394
Fax: 639-426-2919
EMail: Cgagne@MaineLung.org
www.mainelung.org

New Hampshire Chapter:
American Lung Association of New Hampshire
9 Cedarwood Drive, Unit 12
Bedford, NH 03110
Phone: 603-669-2411
Fax: 603-645-6220
Email: info@nhlung.org
www.nhlung.org/

Rhode Island Chapter:
American Lung Association of Rhode Island
298 West Exchange Street
Providence, RI 02903-3700
Phone: 401-421-6487
Fax: 401-331-5266
Email: ALARI@lungri.org
www.lungusa2.org/rhodeisland/index.html

Vermont Chapter:
American Lung Association of Vermont
30 Farrell Street
South Burlington, VT 05403-6196
Phone: 802-863-6817
Fax: 802-863-6818
Email: dhunt@vtlung.org
www.lungusa2.org/vermont/

American Academy of Allergy Asthma and Immunology
555 East Wells Street
Suite 110
Milwaukee, WI 53202
Phone: 800-822-2762 or 414-272-6071
Fax: 414-272-6070
Email: info@aaaai.org
www.aaaai.org

Asthma and Allergy Foundation of America, Inc.
1233 20th Street NW
Suite 402
Washington, DC 20036
Phone: 800-727-8462 or 202-466-7643
Fax: 202-466-8940
Email: info@aafa.org
www.aafa.org

New England Chapter:
220 Boylston Street
Chestnut Hill, MA 02467
Phone: 877-2-ASTHMA or 617-965-7771
Fax: 617-965-8886
Email: info@asthmaandallergies.org
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:

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

Discusses diagnosis, symptoms, individual management plans, current therapies, and pros and cons of medications.

Ataxia Telangiectasia

Ataxia Telangiectasia Children s Project
6685 Military Trail
Deerfield Beach, FL 33442
Phone: 800-543-5728 or 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 91302
Phone: 818-704-8146
Fax: 818-704-8310
Email: becca4435@aol.com
Contact: Pamela Smith

ADDITIONAL WEB RESOURCES:

Ataxia Telangiectasia Mutation Database
www.vmresearch.org/bri_investigators/atm.htm

Scientific site containing listings of AT mutations associated with cancer, AT polymorphisms and variants, and nucleotide sequence of AT gene. Includes clinical information on AT.

AUTISM

Autism Research Institute
4182 Adams Avenue
San Diego, CA 92116
Phone: 619-281-7165
Fax: 619-563-6840
www.autismresearchinstitute.com

Autism Society of America
7910 Woodmont Avenue
Suite 300
Bethesda, MD 20814
Phone: 800-3AUTISM or 301-657-0881
Fax: 301-657-0869
www.autism-society.org

Connecticut Chapter:
P.O. Box 1404
Guilford, CT 06437
Phone: 888-453-4975
Email: asconn@sbcglobal.net
www.autismsocietyofct.org

Maine Chapter:
72B Main Street
Winthrop, ME 04364
Phone: 800-273-5200
Email: info@asmoline.org
www.asmonline.org/

Massachusetts Chapter:
47 Walnut Street
Wellesley Hills, MA 02481
Phone: 781-237-0272
Email: asamasschapter@hotmail.com
www.geocities.com/asamasschapter/

New Hampshire Chapter:
PO Box 68
Concord, NH 03302
Phone: 603-679-2424
Email: info@nhautism.com
www.nhautism.com/

Rhode Island Chapter:
PO Box 16603
Rumford, RI 16603
Phone: 401-595-3241

Vermont Chapter:
PO Box 978
White River Junction, VT 05001
Phone: 800-559-7398

National Alliance for Autism Research
Research Park
99 Wall Street
Princeton, NJ 08540
Phone: 888-777-6227 or 609-430-9160
Fax: 609-430-9163
Email: naar@naar.org
www.naar.org

NAAR — New England Chapter
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

A web site dedicated to helping families who are living with the 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 and special problems and needs of the adult with autism. Provides a 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 carefullly investigated all promising treatment approaches.

Family Pictures: A Novel
by Miller, S.
Perennial, 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.
Crown, 1992

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.
Crowns, 1994

Sequel to "Nobody Nowhere," the author describes her life as a published author and graduate student.

BATTEN DISEASE

Batten Disease Support and Research Association
120 Humphries Drive
Suite 2
Reynoldsburg, OH 43068
Phone: 800-448-4570 or 740-927-4298
Fax: 614-445-4191
Email: bdsra1@bdsra.com
www.bdsra.org

New England Chapter:
Diane Burl
22 Carver Street
Granby, MA 01033
Phone: 413-467-2294

ADDITIONAL WEB RESOURCES:

The Natalie Fund
www.nataliefund.org

Striving to find a treatment and cure for Batten 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
www.beckwith-wiedemann.org

Canavan Disease

Canavan Foundation
450 West End Avenue #10C
New York, NY 10024
Phone: 877-4-CANAVAN or 212-873-4640
Fax: 212-873-7892
Email: info@canavanfoundation.org
www.canavanfoundation.org

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

Connecticut Chapter:
(Serving Connecticut, New Jersey and New York)
1202 Lexington Avenue #288
New York, NY 10028
Phone: 888-354-7788 or 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 or 404-320-3333
Email available through web site only.
www.cancer.org

CONNECTICUT CHAPTERS
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

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

Maine Chapter:
One Main Street; Suite 300
Topsham, ME 04086
Phone: 207-373-3700
Fax: 207-725-6680

Massachusetts Chapters:
18 Tremont St
Suite 700
Boston, MA 02108
Phone: 617-556-7400
Fax: 617-263-6825

1115 W Chestnut St
Brockton, MA 02301
Phone: 508-584-9600
Fax: 508-584-9699

59 Bobala Rd
Holyoke, MA 01040
Phone: 413-734-6000
Fax: 413-493-2199

9 Riverside Rd
Weston, MA 02493
Phone: 781-894-6633
Fax: 781-314-2699

350 Plantation St
Worcester, MA 01604
Phone: 508-270-4600
Fax: 508-751-8095

New Hampshire Chapter:
360 Route 101, Unit #8
The Gail Singer Memorial Building
Bedford, NH 03110
Phone: 603-472-8899
Fax: 603-472-7093

Rhode Island Chapter:
222 Richmond St.
Suite 200
Providence, RI 02903
Phone: 401-722-8480
Fax: 401-421-0535

Vermont Chapter:
121 Connor Way, Ste 240
Williston, VT 05495
Phone: 802-872-6300
Fax: 802-872-6399

Candlelighters Childhood Cancer Foundation

P.O. Box 498
Kensington, MD 20895
Phone: 800-366-2223 or 301-962-3520
Fax: 301-962-3521
Email: staff@candlelighters.org
www.candlelighters.org

New Hampshire Chapter:
Childhood Cancer Lifeline of New Hampshire
P.O. Box 395
Hillsboro, NH 03244
Contact: Sylvia Pelletier, President
Phone: 603-645-1489
info@childhoodcancerlifeline.org

Services: Candlelighters national books, support group meetings, in-hospital visits.

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 containing 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

An essential overview of human cancer genetics.

Counseling About Cancer: Strategies for Genetic Counseling

by Schneider, K.
Wiley-Liss, 2nd Edition, 2001

A comprehensive resource including medical and psychological issues, cancer risk counseling, predisposition testing, and essential references.

Healing Lessons

by Winawer, S.J. and Taylor, N.
Rutledge, 1999

The 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 American family typical only in the diagnosis of breast cancer that hangs like a dark cloud over all its women.

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 860847
Plano, TX 75086
Phone: 800-250-5273
Fax: 972-633-8088
Email: cdgaware@aol.com
www.cdgs.com

Cerebral Palsy

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

Connecticut Chapters:
UCP of Eastern Connecticut
42 Norwich Road
Quaker Hill, CT 06375
Phone: 860-443-3800
Fax: 860-443-8272
Email: mmorisson@ucpect.org
www.ucpect.org

UCP of Greater Hartford
80 Whitney Street
Hartford, CT 06105
Phone: 860-236-6201
Fax: 860-236-6205
Email: jmcmahon@sunrisegroup.org
www.ucpect.org

UCP of Southern Connecticut
94-96 South Turnpike Road
Wallingford, CT 06492
Phone: 203-269-3511
Fax: 203-269-7411
Email: ucpasouthernct@yahoo.com
www.ucpasouthernet.com

Massachusetts Chapters;
UCP of Berkshire County
208 West Street
Pittsfield, MA 01201
Phone: 413-442-1562
Fax: 413-499-4077
E-mail: info@ucpberkshire.org
www.ucpberkshire.org

UCP of MetroBoston
71 Arsenal Street
Watertown, MA 02472
Phone: 617-926-5480
Fax: 617-926-3059
E-mail: ucpbost@aol.com
www.ucpboston.org

Maine Chapter:
UCP of Northeastern Maine
700 Mt. Hope Avenue, Suite 320
Bangor, ME 04401
Phone: 207-941-2952
Fax: 207-941-2955
E-mail: bobbijo.yeager@ucpofmaine.org
www.ucpofmaine.org

Rhode Island Chapter:
UCP of Rhode Island
200 Main Street, Suite 210
P.O. Box 36
Pawtucket, RI 02862
Phone: 401-728-1800
Fax: 401-728-0182
E-mail: ucprisupport@ucpri.org
www.ucpri.org

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

This overview of CP explains medical procedures and medical and psychological implications, discusses advocacy, provides recommended reading and a list of resources.

CHARCOT- MARIE- TOOTH

Charcot-Marie-Tooth Association
2700 Chestnut Street
Chester, PA 19013
Phone: 800-606-2682
Fax: 610-499-9267
Email: CMTassoc@aol.com
www.charcot-marie-tooth.org
www.cmtinfo.org

Connecticut Chapater:
Contact Beverly Wurzel
Phone: 845-783-2815
Email: cranomat@frontiernet.net

ADDITIONAL WEB RESOURCES:

CMTnet
www.users.rcn.com/smith.ma.ultr-ranet/CMTneto.html

Repository of information on research and treatment of CMT for both 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: 561-395-4252
Fax: 561-395-4252 call first
Email: info@chromodisorder.org
www.chromodisorder.org

4p- Support Group
P.O. Box 1676
Gresham, OR 97030
Phone: 503-661-7546
Email: fourthchromosome@aol.com
www.4p-supportgroup.org

5p- Society
P.O. Box 268
Lakewood, CA 90714
Phone: 888-970-0777or 562-804-4506
Fax: 562-920-5240
Email: director@fivepminus.org
www.fivepminus.org or
www.criduchat.org

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
New patient: 937-641-4000
Fax: 937-463-5325
Email: callif-daleyF@childrensdayton.org
www.cmc-dayton.org

Chromosome 9p- Network
P.O. Box 54
Stanley, ID 83278
Phone: 435-574-1121
Fax: 435-574-2000
Email: pduffy006@verizon.net
www.9pminus.org

11q Research and Resource Group
83 Lantern Hill Road
Mystic, CT 06355
Phone: 860-599-4015
Fax: 860-441-6159
http://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

IDEAS (Isodicentric 15 Exchange, Advocacy, and Support)
18 Kings Road
Canton, MA 02021
Phone: 508-253-2872
Email: info@idic15.org
www.idic15.org

Disorders of Chromosome 16 Foundation
1321 Marcy Street
Iowa City, IA 52240
Email: danalex@avalon.net
www.trisomy16.org

Support Organization for Trisomy 18, 13 and Related Disorders (SOFT)
2982 South Union Street
Rochester, NY 14624
Phone: 800-716-7638 or 585-594-4621
Fax: 716-594-1957
Email: barbsoft@rochester.rr.com
www.trisomy.org

Connecticut contact:
Lisa and Robert Handel
1473 Forbes Street
East Hartford, CT 06118
Phone: 860-568-0171
Email: lisahandel@sbcglobal.net

Massachusetts and Maine contact:
Michael and Pamela Healey
18 Richard Rd.
Lexington, MA 02421
Phone: 781-862-8273
Email: Healeylex@aol.com

New Hampshire contact:
Marie Primeau
19 West Ridge Drive
Peterborough, NH 03458
Phone: 603-924-3603
Email: mandbprimeau@adelphia.net

Trisomy 18 Support Foundation, Inc.
4491 Cheshire Station Plaza
Suite 157
Dale City, VA 22193
Phone: 703-878-7013
Fax: 703-878-7013
Email: info@trisomy18support.org
www.trisomy18support.org

Chromosome 18 Registry and Research Society
7155 Oakridge Drive
San Antonio, TX 78229
Phone: 210-657-4968
Fax: 210-657-4968
Email: office@chromosome18.org
www.chromosome18.org

New England Regional Coordinator:
Deb Ammann
Phone: 631-223-3039
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

Maine Contact:
Sharon Smith
RR 2 Box 8780
Canaan, ME 04924
Phone: 207-474-5374
Email: mainelypsych@prexar.com

New Hampshire Contact:
Maureen Anderson
2 Lansing Drive
Salem, NH 03079
Phone: 613- 898-6332
Email: MLADJA@aol.com

Ring Chromosome 22 Email Discussion List
14 Westwood Acres
Morris, MN 56267
Phone: 320-589-1050
Email: r22@maelstrom.stjohns.edu
www.maelstrom.stjohns.edu/archives/r22.htm

ADDITIONAL LITERATURE:

Chromosome Abnormalities and Genetic Counseling
by Gardner, RT.J.M. and Sutherland, G.R. Oxford University Press, 3rd Edition, 2003

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 158
South Beloit, IL 61080-0158
Phone: 888-486-1209
Fax: 630-761-2985
Email: info@aboutfaceusa.org
www.aboutfaceusa.org
Call main office or check web site for local support groups.

American Cleft Palate — Craniofacial Association
1504 East Franklin Street
Suite 102
Chapel Hill, NC 27514
Phone: 919-933-9044
Fax: 919-933-9604
Email: info@acpa-cpf.org
www.cleftpalate-craniofacial.org

Children s Craniofacial Association
13140 Coit Road
Suite 307
Dallas, TX 75240
Phone: 800-535-3643
Fax: 214-570-8811
Email: contactcca@ccakids.com
www.ccakids.com

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

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

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 dealing with the challenges associated with cleft-ing.

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. Available used or at libraries.

Babyface: A Story of Heart and Bones
by McDermott, J.
Penquin Books, 2002

The story of a child with Apert syndrome. Available used or at libraries.

Cleft Lip and Palate: From Origin to Treatment
by Wyszynski, D.F.
Oxford University Press, 2002

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
Email: cclsfoundation@yahoo.com
www.clsf.info

Maine Contact:
Mark & Lynn Richards
90 Hunts Meadow Road
Whitefield, ME 04353
Phone: 207-549-5511
Email: lynndeluxe05@yahoo.com

Massachusetts Contact:
Ronald and Deborah Gallagher
45 Harrison Avenue
Williamstown, MA 01267
Phone: 413-458-2213
Email: Ronald.L.Gallagher@williams.edu

CONGENITAL ADRENAL HYPERPLASIA

CARES Foundation, Inc. (Congenital Adrenal Hyperplasia, Research, Education and Support)
189 Main Street
2nd floor
Millburn, NJ 07041
Phone: 866-227-3737 or 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@charter.net

Maine contact:
Jeff & Jody Spear
Phone: 207-832-2006
Email: jrspear@adelphia.net

Additional web resources:

CAH Education and Support Network
www.congenitaladrenalhyperplasia.org

Includes: "What Is CAH?", message boards, medical links, glossary, and Frequently Asked Questions.

Congenital Hypothyroidism

American Foundation of Thyroid Patients
P.O. Box 4914
Odessa, TX 79760
Email: thyroid@flash.net
www.thyroidfoundation.org

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

Additional web resources:

Educating Parents of Extra-Special Children
www.epeconline.com/Congenital Hypothyroidism.html

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: 800-223-8355 or 860-676-8166
Fax: 860-676-8337
Email: info@cdlsusa.org
www.cdlsusa.org

Additional web resources:

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

Crohn s Disease and Colitis

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

Connecticut Chapters:

Central Connecticut Chapter
P.O. Box 185431
Hamden, CT 06518
Phone: 203-393-8964
Fax: 203-248-7832
Email: ctccfa@aol.com

Fairfield/Westchester Chapter
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

New England Chapter:
Contact: Stacey Snyder
Phone: 800-314-3459 ext. 13
or 781-449-0324
Fax: 781-449-0325
Email: ssnyder@ccfa.org

ADDITIONAL WEB RESOURCES:

Crohn s Disease Resource Center
www.healingwell.com/ibd

This site includes a resource directory, message boards, chat rooms, and links to 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 resources, 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

An older text but a good general introduction to the diseases.

Cystic Fibrosis

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

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

Massachusetts/Rhode Island Chapter:
Natick, MA
Phone: 508-655-6000
Email: mass-ri@cff.org
www.cff.org/Chapters/massachusetts/

Northern New England Chapter: (Maine, New Hampshire and Vermont)
Nashua, NH
Phone: 603-598-8191
Email: no-new-eng@cff.org
www.cff.org/Chapters/newengland/

ADDITIONAL WEB RESOURCES:

Cystic Fibrosis Mutational Database
www.genet.sickkids.on.ca/cftr

This database of mutations in CFTR gene provides CF researchers and related professionals with up-to-date information about individual mutations.

Cystic Fibrosis.com
www.cysticfibrosis.com

This site incudes CF information, frequently asked questions, a "Just for Kids" section, and links to CF associations and clinical trials.

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, and 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. Available used and at libraries.

Cystic Fibrosis: A Guide for Patient and Family
by Orenstein, D.M.
Lippincott, Williams and Wilkins, 3rd Edition, 2003

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: email@cystinosis.com
www.cystinosisfoundation.org

Cystinosis Research Network
10 Pine Avenue
Burlington, MA 01803
Phone: 866-276-3669 or 781-229-6182
Fax: 781-229-6030
Email: CRN@cystinosis.org
www.cystinosis.org

ADDITIONAL WEB RESOURCES:

Cystinosis Central
www.medicine.ucsd.edu/cystinosis/INDEX.htm

Up-to-date site with 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 or 202-337-5220
Fax: 202-337-8314
Email: agbell2@aol.com
www.agbell.org
With links to state representatives and web sites.

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

National Association of the Deaf
8630 Fenton Street
Suite 820
Silver Spring, MD 20910-3819
Phone: 301-587-1788
Fax: 301-587-1791
Email: NADinfo@nad.org
www.nad.org
Has email links to current presidents of each state affiliate.

Connecticut Association of the Deaf
http://www.deafct.org/

Massachusetts Association of the Deaf
http://www.msad.org/

Rhode Island Association of the Deaf
http://www.riadeaf.org/

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, this site 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, 2002

Informs reader about 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, the author has written a candid, affecting, and often funny memoir.

DIABETES

American Diabetes Association

1701 North Beauregard Street
Alexandria, VA 22311
Phone: 800-DIABETES or 888-DIABETES
(for local contact)
Fax: call for faxing information
Email: ASKADA@diabetes.org
www.diabetes.org

Connecticut Chapter:
306 Industrial Park Road, Suite 105
Middletown, CT 06457
Phone: 203-639-0385
Fax: 860-632-5098

Maine Chapter:
163 Lancaster Street, Suite 98R
Portland, ME 04101
Phone: 207-774-7717
Fax: 207-774-7714

Massachusetts Chapter:
330 Congress Street, 5th Floor
Boston, MA 02210
Phone: 617-482-4580
Fax: 617-482-1824

New Hampshire Chapter:
249 Canal Street
Manchester, NH 03101
Phone: 603-627-9579
Fax: 603-669-1477

Rhode Island Chapter:
222 Richmond St., Suite 204
Providence, RI 02903
Phone: 401-351-0498
Fax: 401-351-1674

Vermont Chapter:
1 Kennedy Drive, Suite L8
South Burlington, VT 05403
Phone: 802-654-7716
Fax: 802-658-9145

Juvenile Diabetes Research Foundation

120 Wall Street, 19th floor
New York, NY 10005
Phone: 800-533-2873
Fax: 212-785-9595
Email: info@jdrf.org
www.jdrf.org

Connecticut Chapters:
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

New England/Maine Chapter:
33 Silver St.
Portland, ME 04101
Phone: 207-761-0133
Fax: 207-761-1687
Email: maine@jdrf.org
www.jdrf.org/maine

New England/New Hampshire Chapter:
2 Wellman Ave., Suite 340
Nashua, NH 03064
Phone: 603-595-2595
Email: newhampshire@jdrf.org
www.jdrf.org/newhampshire

New England/Rhode Island Chapter:
2374 Post Road, Suite 203
Warwick, RI 02886
Phone: 401-738-9898
Fax: 401-738-7162
Email: rhodeisland@jdrf.org
www.jdrf.org/rhodeisland

ADDITIONAL WEB RESOURCES:

Children with Diabetes
www.childrenwithdiabetes.com

An online community for kids, families and adults with diabetes.

Joslin Diabetes Center
www.joslin.org

A Harvard Medical School affiliate and internationally recognized treatment, research, and education institution headquartered in Boston, Massachusetts.

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

A 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.
For Dummies, 2nd edition,2004

Addresses Type 1 and Type 2 diabetes, including: causes, symptoms, treatment, importance of diet and exercise, finding the right practitioner, building a support team, and finding 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, this book provides information about 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 or 770-604-9500
Fax: 212-979-2873 or 770-604-9898
Email: info@ndsccenter.org
www.ndsccenter.org

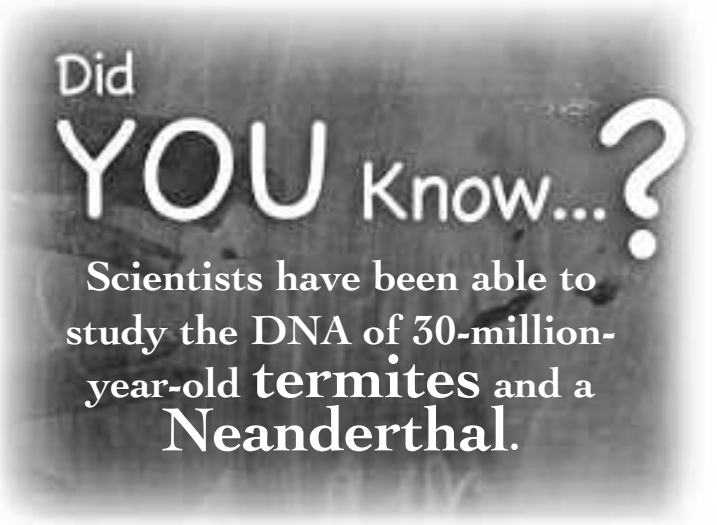
Connecticut Chapter:
Connecticut Down Syndrome Congress
263 Farmington Avenue
MC-6222
Farmington, CT 06030
Phone: 888-486-8537
Email: manager@ctdownsyndrome.org
www.ctdownsyndrome.org

Maine Affiliate (serving all of Maine):
Sourthern Maine Down Syndrome Family Network
P.O. Box 705
Windham, ME 04062
Phone: 866-571-2223
Fax: 866-571-2223
Email: smdsfn@downsyndromemaine.org
www.downsyndromemaine.org

Massachusetts Down Syndrome Congress
P.O. Box 866
Melrose, MA 02176
Phone: 800-664-6372
Email: mdsc@mdsc.org
www.mdsc.org

New Hampshire Chapter:
Northern New England Down Syndrome Congress
P.O. Box 1234
Concord, NH 03302
Phone: 603-622-6904
www.nnedsc.org

Rhode Island Chapter:
Down Syndrome Congress of Rhode Island
99 Bald Hill Road
Cranston, RI 02920
Phone: 401-463-5751
www.dssri.org



National Down Syndrome Society
666 Broadway
Suite 810
New York, NY 10012
Phone: 800-221-4602 or 212-460-9330
Fax: 212-979-2873
Email: info@ndss.org
www.ndss.org
Mulitple resources listed by state.

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

Additional web resources:

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

This web site includes a brief history of the genetic basis of Down syndrome and growth charts for children with and without Down syndrome.

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.

www.ds-health.com/ds_sites.htm

Includes a list of useful links to a wealth of resources about Down syndrome. Compiled by a physician.

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

Front cover: The child with Down syndrome very often can be raised to become independent. This book offers parents and guardians sound advice on managing healthcare, dealing with siblings, coping with finances, and choosing education, recreation, adult living, and employment options. Available used and in libraries.

Teaching the Infant with Down Syndrome: A Guide for Parents and Professionals
by Hanson, M.J.
Pro-Ed, 2nd Edition, 1996

A comprehensive curriculum for children with Down syndrome from birth to 24 months. Includes helpful advice.

Expecting Adam: A True Story of Birth, Rebirth, and Everyday Magic
by Beck, M.
Crown, 1999

Autobiographical tale of an 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 faed with by abnormal prenatal 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 or 312-755-0198
Fax: 312-803-0138
Email: dystonia@dystonia-foundation.org
www.dystonia-foundation.org

Connecticut Chapters:
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

Massachusetts Chapter:
Lahey Clinic Support Group
Contact: Ann Lebrun
Phonel: 978-688-2789
Email: annlebrun@comcast.net

New Hampshire Chapter:
New Hampshire Support Group
Diane Dinsmoor
Phone: 603-524-7861
Email: ddinsmoor@metrocast.net

Vermont Chapter:
Vermont Support Group
Marcia Kenworthy
Phone: 802-223-2526
Email: coolestzero@aol.com

ADDITIONAL WEB RESOURCES:

International Dystonia On-line Support Group
www.dystonia-support.org

Created by affected individuals,this site includes medical information, personal accounts, chat rooms, information for parents, and a 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: research techniques to find general information, research techniques for specific topics in dystonia, and a guide to the latest scientific research.

EHLERS- DANLOS SYNDROME

Ehlers-Danlos National Foundation
3200 Wilshire Boulevard
Suite 1601, South Tower
Los Angeles, CA 90010
Phone: 213-368-3800
Fax: 213-427-0057
Email: staff@ednf.org
www.ednf.org

Local support group information is accessible through web site.

EDS Today
P.O. Box 88814
Seattle, WA 98138
Phone: 253-835-1735
Fax: 253-835-1735
Email: info@edstoday.org
www.edstoday.org

ADDITIONAL WEB RESOURCES:

Ehlers-Danlos Syndrome
www.orthop.washington.edu/arthritis/types/ehlersdanlos/01

Web site of the University of Washington Orthopedics and Sports Medicine Group, with information about 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 or 301-459-3700
Fax: 301-577-2684
Email: postmaster@efa.org
www.epilepsyfoundation.org

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

Massachusetts and Rhode Island Affiliate:
Epilepsy Foundation Massachusetts & Rhode Island
540 Gallivan Boulevard, 2nd Floor
Boston, MA 02124-5401
Phone: 888-576-9996 or 617-506-6041
www.epilepsyfoundation.org/massri/

Vermont Affiliate:
Epilepsy Foundation Vermont
PO Box 6292
Rutland, VT 05702-6292
Phone: 800-565-0972 or 802-775-1686
www.epilepsyvt.org

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

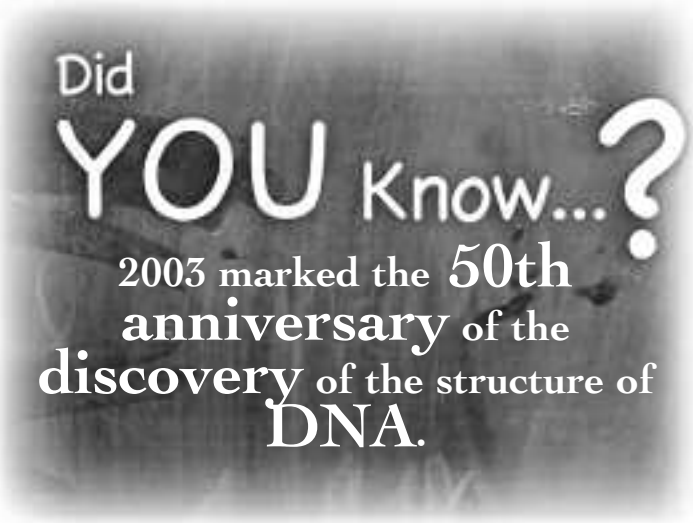
Dysautonomia Foundation, Inc.
315 West 39th Street
Suite 701
New York, NY 10018
Phone: 212-279-1066
Fax: 212-279-2066
Email: info@familialdysautonomia.org
www.familialdysautonomia.org

National Dysautonomia Research Foundation
P.O.Box 301
Red Wing, MN 55066
Phone: 651-267-0525
Fax: 651-267-0524
Email: ndrf@ndrf.org
www.ndrf.org

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
www.fanconi.org

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.
Eurekan.com Inc., 2004

Medical textbook.

Fatty Acid Oxidation Disorders

FOD Family Support Group
1559 New Garden Road, 2E
Greensboro, NC 27410
Phone: 336-547-8682
Fax: call for faxing information
Email: deb@fodsupport.org
www.fodsupport.org

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

Fragile X Syndrome

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

Connecticut Affiliate:
Fragile X Society of Connecticut
Contact: Tammy Selinger
Phone: 860-233-1904
Email: xfamily@aol.com

Maine Affiliate:
Maine Fragile X Resource Group
Cheryl Peterson
Home Phone: 207-224-2035
Julie Gosselin
Home Phone: 207-657-2395
Email: mainefragilex@yahoo.com
www.fragilex.org/html/maine.htm

Massachusetts Affiliates:
Fragile X Resource Group of Western Massachusetts
Denise Devine
Phone: 413-584-1859
Email: Devinehdly@aol.com

FraX'em (FX Resource Group of E. MA)
Contact: Sandra Morse
Phone: 978-927-3669
Email: wishlist@hotmail.com
lhttp://communities.msn.com/TheWorldofFragileX

New Hampshire Affiliate:
New Hampshire Fragile X Resource Group
Contact: Debbie White
Phone: 603-329-4632
Email: nhfragx@yahoo.com
www.fragilex.org/html/new_hampshire.htm

Rhode Island Affiliate:
Rhode Island Fragile X Resource Group
Contact: Robert & Anne McInerney
Phone: 401-467-5135
Fax: 401-467-5135
Email: RWM1954@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

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 seeking 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, and gene therapy.

GALACTOSEMIA

Parents of Galactosemic Children, Inc.

1519 Magnolia Bluff Drive
Gautier, MS 39553
Phone: 775-626-0885
Email: president@galactosemia.org
www.galactosemia.org

ADDITIONAL WEB RESOURCES:

Galactosemia Resources and Information

www.galactosemia.com

Includes general information on galactosemia, galactose content in foods, and contact information for other affected families.

GAUCHER DISEASE

National Gaucher Foundation, Inc.

61 General Early Drive
Harpers Ferry, WV 25425
Phone: 800-925-8885
Fax: 304-725-6429
Email: ngf@gaucherdisease.org
www.gaucherdisease.org

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

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, a 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 about where and how to look for information. Also useful for doctors, caregivers, and other health professionals.

Glycogen Storage Diseases

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

The Children s Fund for Glycogen Storage Disease Research
917 Bethany Mountain Road
Cheshire, CT 06416
Phone: 203-272-CURE or 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 or 212-688-1000
Fax: 212-483-8179
Email: info@liverfoundation.org
www.liverfoundation.org

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

New England Chapter:
88 Winchester Street
Newton, MA 02461
Phone: 800-298-6766 or 617-527-5600
Email: info@liverfoundation-ne.org
www.liverfoundation-ne.org

Growth Disorders

Human Growth Foundation
997 Glen Cove Avenue
Glen Head, NY 11545
Phone: 800-451-6434 or 516-671-4041
Fax: 516-671-4055
Email: hgf1@hgfound.org
www.hgfound.org

MAGIC Foundation for Children s Growth
6645 West North Avenue
Oak Park, IL 60302
Phone: 800-362-4432 or 708-383-0808
Fax: 708-383-0899
Email: mary@magicfoundation.org
www.magicfoundation.org

Heart Disorders

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

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

Maine Chapter:
51 US Route 1
Suite M
Scarborough, ME 04074
Phone: 207-879-5700
Fax: 207-879-5918

Massachusetts Chapters:
20 Speen Street
Framingham, MA 01701
Phone: 508-620-1700
Fax: 508-620-6157

1111 Elm Street, Suite 9A
West Springfield, MA 01089
Phone: 413-827-0400
Fax: 413-827-9390

2 White's Path
South Yarmouth, MA 02664
Phone: 508-760-6818
Fax: 508-760-6824

New Hampshire Chapter:
2 Wall Street
Manchester, NH 03101
Phone: 603-669-5833
Fax: 603-669-6745

Rhode Island Chapter:
222 Richmond Street, Suite 108
Providence, RI 02903
Phone: 401-330-1700
Fax: 401-330-1720

Vermont Chapter:
434 Hurricane Lane
Williston, VT 05495
Phone: 802-878-7700
Fax: 802-878-7850

Congenital Heart Anomalies Support, Education, and Research

2112 North Wilkins Road
Swanton, OH 43558
Phone: 419-825-5575
Fax: 419-825-2880
Email: chaser@compuserve.com
www.csun.edu/~hcmth011/chaser/chaser-news.html

Mended Hearts Inc.

7272 Greenville Avenue
Dallas, TX 75231
Phone: 888-432-7899 or 214-360-6149
Fax: 214-360-6145
Email: info@mendedhearts.org
www.mendedhearts.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

Little Hearts, Inc.

P.O. Box 171
Cromwell, CT 06416
Phone: 860-635-0006 or 866-435-4673
Fax: 860-635-0006
Email: info@littlehearts.org
www.littlehearts.net

ADDITIONAL WEB RESOURCES:

Texas Heart Institute

www.texasheartinstitute.org

Information about the heart institute at St. Luke's Episcopal Hospital in Houston, Texas.

Congenital Heart Disease Center

www.heartcenteronline.com

Provides patient guides on many congenital heart anomalies, includes animated videos showing how the heart works.

ADDITIONAL LITERATURE:

Cardiac Kids: A Book for Families Who have a Child with Heart Disease

by Elder, V. and King, A.
Tenderhearts Publishing Company, 1994

For parents and children to read together, this 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

A resource that succinctly addresses and provides answers for these complex disorders. Also explains infections, tests, treatments, and risk factors.

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

Written by two professors in pediatric cardiology, along with pediatric nurse. The authors have included how the heart develops in a child and show what can go wrong during the growing process. They also discuss the risk factors for heart defects, explain how heart problems are recognized, and cover treatment options.

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 whose child has been recently diagnosed with a congenital heart defect (CHD). The authors knowledgeably discuss both the medical and the emotional issues to be addressed, from diagnosis through surgery (if needed) to recovery on to adulthood. Includes an illustrated overview of 16 of the most common defects.

Fetal Echocardiography

by Drose, J.A.
W. B. Saunders Company, 1998

An essential reference for anyone involved in fetal echocardiography.

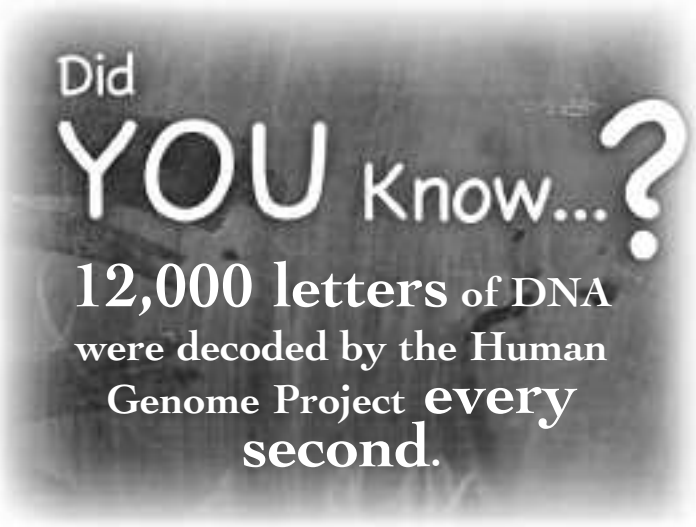
Hemochromatosis

American Hemochromatosis Society

4044 West Lake Mary Boulevard
Suite 104, PMB 416
Lake Mary, FL 32746
Phone: 888-655-4766 or 407-829-4488
Fax: 407-333-1284
Email: mail@americanhs.org
www.americanhs.org

American Liver Foundation

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



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

New England Chapter:
88 Winchester Street
Newton, MA 02461
Phone: 800-298-6766 or 617-527-5600
Email: info@liverfoundation-ne.org
www.liverfoundation-ne.org

ADDITIONAL WEB RESOURCES:

Hemochromatosis Information Society

www.hemoinfo.org

Information on the condition, its treatment, getting an at-home test kit, and links for further information.

ADDITIONAL LITERATURE:

Living with Hemochromatosis

by Everson, G. and Weinberg, H.
Hatherleigh Press, 2003

Patient guide covers signs, symptoms, diagnosis, treatment options, and new areas of research, as well as 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

Informs patients, caregivers and health professionals about looking for information on hemochromatosis.

Hemophilia

National Hemophilia Foundation

116 West 32nd Street, 11th floor
New York, NY 10001
Phone: 800-424-2634 or 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

Resource for hematology education & news.

Huntington s Disease

Huntington s Disease Society of America

505 Eighth Avenue, Suite 902
New York, NY 10018
Phone: 800-345-4372 or 212-242-1968
Fax: 212-239-3430
Email: hdsainfo@hdsa.org
www.hdsa.org

New England Chapter:
1253 Worchester Road
Suite 202
Framingham, MA 01701
Phone: 888-872-8102 or 508-872-8102
Fax: 508-872-8103
Email: virginia@hdsa-ne.org
www.hdsa-ne.org

Hereditary Disease Foundation (focuses on Huntington s Disease)

1303 Pico Boulevard
Santa Monica, CA 90405
Phone: 310-450-9913
Fax: 310-450-9532
Email: cures@hdfoundation.org
www.hdfoundation.org

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, and 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 or 562-402-3523
Fax: 562-924-6666
Email: hydrobrat@earthlink.net
www.nhfonline.org

Hydrocephalus Association

870 Market Street, Suite 705
San Francisco, CA 94102
Phone: 888-598-3789 or 415-732-7040
Fax: 415-732-7044
Email: info@hydroassoc.org
www.hydroassoc.org

Guardians of Hydrocephalus Research Foundation

2618 Avenue Z
Brooklyn, NY 11235
Phone: 718-743-4473
Fax: 718-743-1171
Email: GHRF2618@aol.com
www.ghrf.homestead.com/ghrf.html

Hydrocephalus Foundation

910 Rear Broadway, Route 1
Saugus, MA 01906
Phone: 781-942-1161
Email: HyfII@netscape.net
www.hydrocephalus.org

Additional web resources:

The Hydrocephalus Center

www.patientcenters.com/hydrocephalus

Comprehensive site with links to resources for families with children with hydrocephalus.

NIH Hydrocephalus links

www.nlm.nih.gov/medlineplus/hydrocephalus.html

Listing of resources on hydrocephalus compiled by the U.S. National LIbrary of Medicine and the National Institutes of Health.

Additional literature:

Hydrocephalus: A Guide for Patients, Families and Friends

by Toporek, C. Robinson, K. and Lamb, L.
Patient Center Guide, Inc., 1999

Addresses: selecting a skilled neurosurgeon, treatments, and support.

Pediatric Hydrocephalus

by Cinalli, G., Maixner, W.J. and Sainte-Rose, C.
Springer-Verlag, 2006

Medical reference text for health care professionals including classification, etiology, pathophysiology, genetics, and recent advances.

Jewish Genetic Diseases

Jewish Genetic Disease Consortium

315 West 39th Street, Suite 701
New York, NY 10018
Phone: 866-370-4363
Email: info@JewishGeneticDiseases.org
www.jewishgeneticdiseases.org

The Jewish Genetic Disease Consortium was created as a means by which smaller, individual organizations could join together to heighten awareness of Jewish genetic diseases. The target audience includes medical professionals, rabbis, and the Ashkenazi Jewish population at large.

Klinefelter Syndrome

American Association for Klinefelter Syndrome Information and Support

2945 West Farwell Avenue
Chicago, IL 60645
Phone: 888-466-5747 or 773-761-5298
Fax: 773-761-5298
Email: ksinfo@aaksis.org
www.aaksis.org

Northeast/Boston area Regional Support Group:
Co-chairs of the Support Group
Dalene Basden
Phone: 781-599-9288
Email: ptahsw@verizon.net

Bill Mulkern
Phone: 617-742-4247

Klinefelter Syndrome and Associates

11 Keats Court
Coto De Caza, CA 92679
Phone: 888-999-9428
Fax: 949-858-3443
Email: help@genetic.org
www.genetic.org

ADDITIONAL WEB RESOURCES:

Klinefelter Syndrome Support Group Home Page

www.klinefeltersyndrome.org

Includes links to prenatal diagnosis information, support group information, research studies, medical conferences, and other useful web sites.

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, and information on Internet resources. Designed for physicians, medical students, medical researchers, and patients.

KLIPPEL- TRENAUNAY- WEBER

Klippel-Trenaunay Support Group

5404 Dundee Road
Edina, MN 55436
Phone: 952-925-2596
Fax: 612-677-1338
Email: ktnewmembers@yahoo.com
www.k-t.org

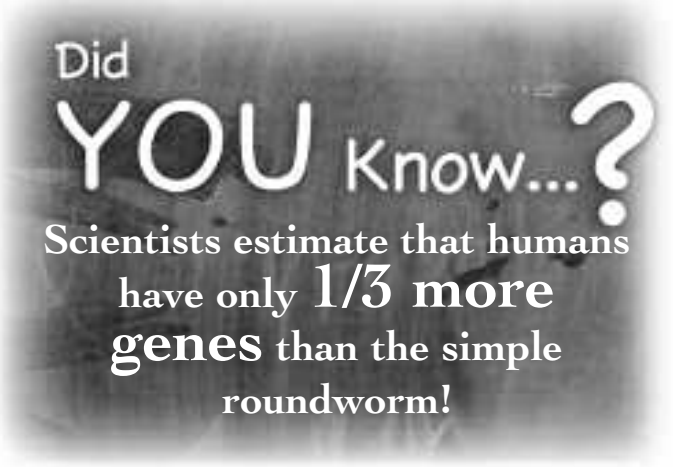
LESCH- NYHAN SYNDROME

Lesch-Nyhan Syndrome Registry

New York University School of Medicine
Department of Psychiatry
New Bellevue 18 18E13
Bellevue Hospital
New York, NY 10016
Phone: 212-263-6458
Fax: 212-629-9523
Email: lta1@nyu.edu
www.lndinfo.org

Lesch-Nyhan Syndrome Children s Research Foundation

210 South Green Bay Road
Lake Forest, IL 60045
Phone: 847-234-3154
Fax: 847-234-3136



ADDITIONAL WEB RESOURCES:

National Institute of Neurological Disorders and Stroke; Lesch-Nyhan Syndrome Information Page

www.ninds.nih.gov/disorders/lesch_nyhan/lesch_nyhan.htm

Includes a description of Lesch-Nyhan syndrome, treatment options, prognosis and information on research.

LEUKODYSTROPHY

United Leukodystrophy Foundation

2304 Highland Drive
Sycamore, IL 60178
Phone: 800-728-5483 or 815-895-3211
Fax: 815-895-2432
Email: office@ulf.org
www.ulf.org

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 educational documents. For medical professionals, reference tools and texts have detailed technical information.

LUPUS

American Autoimmune Related Diseases Association, Inc.
22100 Gratiot Avenue
East Detroit, MI 48021
Phone: 586-776-3900
Fax: 586-776-3903
Email: aarda@aarda.org
www.aarda.org

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

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

Connecticut and Rhode Island Chapter:
97 South Street
Suite 110
West Hartford, CT 06110
Phone: 800-699-6967 or 860-953-0387
Fax: 860-953-0483
Email: CTLFA@sbcglobal.net
www.lupusct.org

New Hampshire and
Vermont Chapter:
Waterbury, VT
Phone: 802-244-5988

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, and email list.

ADDITIONAL LITERATURE:

Coping with Lupus: A Guide to Living With Lupus for You and Your Family
by Phillips, R.H.
Avery Publishing Group, 3rd Edition, 2001

Includes new research, treatments, and information.

The Lupus Handbook for Women
by Dibner, R.
Fireside, 1994

"Must-read" book of common sense advice and tips for daily living, as well as an entire chapter on pregnancy.

MARFAN SYNDROME

National Marfan Foundation
22 Manhassett Avenue
Port Washington, NY 11050
Phone: 800-862-7326 or 516-883-8712
Fax: 516-883-8040
Email: staff@marfan.org or
support@marfan.org
www.marfan.org

Connecticut Contact:
Richard Paul
Phone: 203-268-7559
Email: richcpaul@hotmail.com

Maine Contact:
Merrill Henderson
Phone: 207-839-8637
Email: mhender1@maine.rr.com
Massachusetts Contacts:
Suzanne Kouri (Lexington)
Phone: 781-862-6398 or
781-248-8743(Cell)
Email: kouri34@aol.com
Sandra La Pan
Phone: 413-739-2950

Rhode Island Contact:
Adrienne Shafer
Phone: 401-658-1719
Email: ashafer28@cox.net

ADDITIONAL WEB RESOURCES:

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

Answers general questions about Marfan syndrome. It describes the characteristics of the disorder, the diagnostic process, and ways to manage symptoms.

ADDITIONAL LITERATURE:

Connective Tissue and Its Heritable Disorders: Molecular, Genetic and Medical Aspects
by Royce, P.M. and Steinmann, B. Wiley-Liss, 2nd Edition, 2002

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 or 301-565-3842
Fax: 301-565-5342
Email: info@thearc.org
www.thearc.org

Connecticut chapters:
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: info@mwsinc.org
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
www.starinc-lightingtheway.org

ARC of New London County
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: marclizw@snet.net
www.marcweb.org

Litchfield County ARC
314 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

Maine Chapters:
(listed alphabetically by town)

DEH Operating Co.
d/b/a Downeast Horizons
1200 State Highway 3
Bar Harbor, ME 04609
Phone: 207-288-4234
Email: info@dehi.org
www.dehi.org

Independence Association
P.O. Box 642
Brunswick, ME 04011
Phone: 207-725-4371
Email: webmaster@indepassoc.org
www.independenceassociation.org

Work First, Inc.
P.O. Box 86
Farmington, ME 04938
Phone: 207-778-3200
Community Living Association
45 School Street
Houlton, ME 04730
Phone: 207-532-9446
www.cla-maine.org/

Green Valley Arc
PO Box 127
Island Falls, ME 04747
Phone: 207-463-2156

Katahdin Friends Inc.
1024 Central Street, Suite A
Millinocket, ME 04462-2111
Phone: 207-723-9466
www.kfimaine.org

Central Aroostook Arc
26 Lombart Street
P.O. Box 1245
Presque Isle, ME 04769
Phone: 207-764-0134

Arc - Oxford County
85 Lincoln Avenue
Rumford, ME 04276
Phone: 207-369-0141

Arc - Waban Projects, Inc.
5 Dunaway Drive
Sanford, ME 04073
Phone: 207-324-7955

Sebasticook Farms Arc
P.O. Box 65
St Albans, ME 04971
Phone: 207-938-4615

Arc - Northern Aroostook
267 Main Street
Van Buren, ME 04785
Phone: 207-868-5203
Email: naainc@verizon.net

Ken-A-Set Arc
P.O. Box 334
Waterville, ME 04903-0334
Phone: 207-872-6484

Massachusetts Chapters (listed alphabetically by town):

The Arc of Northern Bristol County
141 Park Street
Attleboro, MA 02703
Phone: 508-226-1445
Email: nfo@arcnbc.org
www.arcnbc.org/

Greater Boston Arc
221 North Beacon Street
2nd Floor
Brighton, MA 02135
Phone: 617- 783-3900
Email: GBARc1@msn.com
www.gbarc.org/

Brockton Area Arc
1250 West Chestnut Street
Brockton, MA 02301
Phone: 508-583-8030
Email: BAARC@comcast.net
www.brocktonareaarc.org

Minute Man Arc for Human Services
1269 Main Street
Concord, MA 01742
Phone: 978-287-7932
Email: postmaster@minutemanarc.org
www.minutemanarc.org/

North Shore Arc
64 Holten Street
Danvers, MA 01923
Phone: 978-762-4878
www.nsarc.org

The Arc of Greater Fall River
P.O. Box 1943
Fall River, MA 02722
Phone: 508-679-0001
Email: danarc29A@msa.com

ARC Community Services
564 Main Street
Fitchburg, MA 01420
Phone: 978-343-6662 Ext 131
Email: tikennedy@arminc.info
www.arccommunityservices.org

The United Arc of Franklin & Hampshire Counties
111 Summer Street
Greenfield, MA 01301
Phone: 413-774-5558

The Arc of Northern Essex County, Inc.
57 Wingate Street, Suite 301
Haverhill, MA 01832
Phone: 978-373-0552
Email: arc.nec@verizon.net

The Arc of Cape Cod
P.O. Box 428
Hyannis, MA 02601
Phone: 508-790-3667
Email: arcofcapecod@hotmail.com
www.arcofcapecod.org

The Arc of Greater Lawrence
One Parker Street
Lawrence, MA 01843
Phone: 978-975-8587
Email: info@classinc.org
www.classinc.org/arc_page.htm

Charles River Arc
59 East Militia Heights Drive
Needham, MA 02492-1313
Phone: 781-444-4347
Email: development@crarc.org
www.crarc.org/

The Arc of The South Shore
371 River Street
North Weymouth, MA 02191
Phone: 781-335-3023
Email: info@arcsouthshore.org
www.arcsouthshore.org

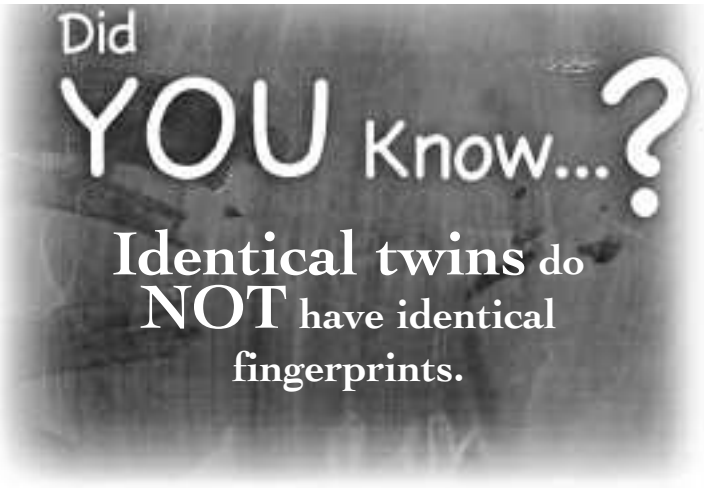
Berkshire County Arc
395 South Street
Pittsfield, MA 01201
Phone: 413-499-4241
Email: bcarc@bcarc.org
www.bcarc.org/

The Arc of Greater Plymouth, Inc.
Cordage Commerce Center
10 Cordage Park Circle, Suite 208
Plymouth, MA 02360
Phone: 508-732-9292
Email: info@thearcofgp.com
www.thearcofgp.com

East Middlesex Arc
20 Gould Street
Reading, MA 01867-2927
Phone: 781-942-4888
www.theemarc.org/

Southern Worcester County Arc
P.O. Box 66
100 Foster 1N Street
Southbridge, MA 01550
Phone: 508-764-4085

The Arc of Massachusetts
217 South Street
Waltham, MA 02453-2710
Phone: 781-891-6270
Email: arcmass@arcmass.org
www.arcmass.org



Greater Waltham ARC
56 Chestnut Street
Waltham, MA 02453
Phone: 781-899-1344
Email: gwarc@gwarc.org
www.gwarc.org

South Norfolk County Arc
789 Clapboardtree Street
Westwood, MA 02090
Phone: 781-762-4001
www.sncarc.org/

Central Middlesex Arc
147 New Boston Street
Woburn, MA 01801
Phone: 781-935-7057

New Hampshire Chapters: (listed alphabetically by town)

Concord Regional Arc, Inc.
P.O. Box 1173
Concord, NH 03302-1173
Phone: 603-228-8279

The Arc of Greater Manchester
P.O. Box 3363
Manchester, NH 03105-3363
Phone: 603-434-2738
Email: president@arcmanchester.org
www.arcmanchester.org

Salem Arc, Inc.
8 Centerville Drive
Salem, NH 03079
Phone: 603-893-9889

Rhode Island Chapters:
(listed alphabetically by town)

Bristol County Chapter Arc
P.O. Box 711
Bristol, RI 02809
Phone: 401-253-5900

Arc - Down Syndrome Society of Rhode Island
99 Bald Hill Road
Cranston, RI 02920
Phone: 401-463-5751
Email: coordinator@dssri.org
www.dssri.org

Cranston Arc, Inc.
111 Comstock Parkway
Cranston, RI 02921
Phone: 401-941-1112
www.cranstonarc.org

Rhode Island Arc (RIARC)
99 Bald Hill Road
Cranston, RI 02920
Phone: 401-463-9191
Email: RhodeIslandArc@cs.com

Newport County Arc
P.O. Box 4390
Middletown, RI 02842
Phone: 401-846-4600

Greater Providence Arc
220 Woonasquatucket Avenue
North Providence, RI 02911
Phone: 401-353-7000
www.fogartycenter.org

The Arc of Blackstone Valley
115 Manton Street
Pawtucket, RI 02861
Phone: 401-727-0150
Email: contact@bvcriarc.org
www.bvcriarc.org

The Arc South County Chapter
238 Robinson Street
Wakefield, RI 02879
Phone: 401-789-4386

Kent County Arc
3445 Post Road
Warwick, RI 02886
Phone: 401-739-2700
www.kentcountyarc.org

Westerly - Chariho Chapter
Frank A. Olean Center
93 Airport Road
Westerly, RI 02891
Phone: 401-596-2091

The Arc of Northern Rhode Island
320 Main Street
Woonsocket, RI 02895
Phone: 401-765-3700
Email: info@arcofnri.org
www.arcofnri.org

American Association on Mental Retardation

444 North Capitol Street NW
Suite 846
Washington, DC 20001
Phone: 800-424-3688 or 202-387-1968
Fax: 202-387-2193
Email: dcroser@aamr.org
www.aamr.org
Connecticut Contact:
David B. Scott
Email: dscott@ccaoh.org

Maine Contact:
Scott Jones
Email: sjones@ohimaine.org

Massachusetts Contact:
Jean M. Phelps
Email: jphelps@shorecollaborative.org

New Hampshire Contact:
Deborah G. Larochelle
Email: Debglrri@aol.com

Rhode Island Contact:
Donna Martin
Email: dmartin@osarr.net

Vermont Contact:
Diane Blais
Email: dblais@nkhs.net

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

New England Chapter of UMDF
39 Bay Farm Drive
Plymouth, MA 02360
Phone: 508-224-7165
Email: NEngChapter@umdf.org

Mitochondria Research Society

P.O. Box 1952
Buffalo, NY 14221
Phone: see web site for individual contacts.
Email: mitoresearch@mitoresearch.org
www.mitoresearch.org

Additional web resources:

Mitochondrial Disease in Perspective: Symptoms, Diagnosis and Hope for the Future

www.mitoresearch.org/treatmentdisease.html

Online lecture covering symptoms, diagnosis, management, research and potential 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

Additional web resources:

Moebius Syndrome

www.moebius1.org

Information, resource links and support.

ADDITIONAL LITERATURE:

My Face
by Abbott, M. and Abbott, B.
Forward Face, 1998

Children's book written by the mother of an 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 or 212-986-3240
Fax: 212-986-7981
Email: generalmailbox@nmss.org
www.nationalmssociety.org

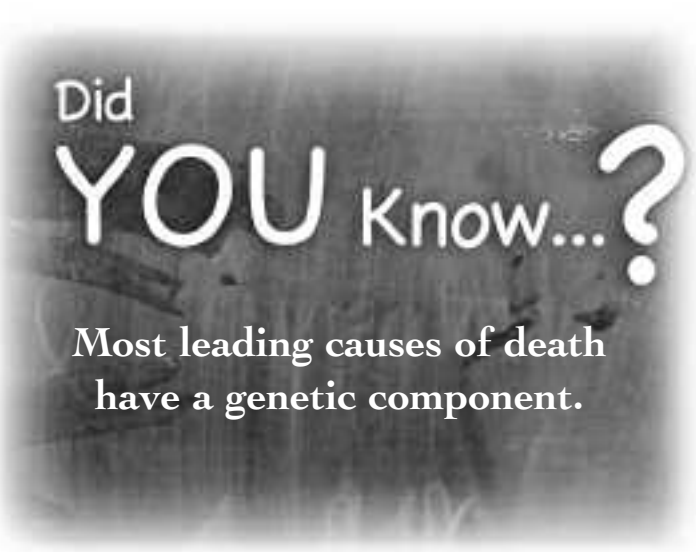
Connecticut Chapters:

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: info@msswct.org
www.msswct.org

Maine Chapter:
170 US Route One, Suite 200
Falmouth, ME 04105
Phone: 1-800-FIGHT MS
Email: info@msmaine.org
www.msmaine.org

Central New England (Massachusetts and New Hampshire)
101A First Avenue, Suite 6
Waltham, MA 02451-1115
Phone: 800-493-9255 or 781-890-4990
Email:communications@mam.nmss.org
www.msnewengland.org



Rhode Island Chapter:
205 Hallene Road, Suite 209
Warwick, RI 02886
Phone: 1-800-FIGHT-MS or 401-738-8383
Email: emily.murphy@rir.nmss.org
www.nationalmssociety.org/rir

Vermont Chapter:
75 Talcott Road
Williston, VT 05495
Phone: 1-800-344-4867 or 802-862-0912
Email: vtn@nmss.org
www.nationalmssociety.org/vtn

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

Rhode Island Contact:
Brenda Berube
West Warwick, RI
Phone: 401-823-0216

ADDITIONAL WEB RESOURCES:

All About Multiple Sclerosis
www.mult-sclerosis.org

Accurate and comprehensive medical data about multiple sclerosis written by people living with the disease. Includes archives of 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

This insightful, informative, and empathic resource discusses traditional and complementary therapies, explains medical terminology and diagnosis, and addresses lifestyle issues.

MUSCULAR DYSTROPHY

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

Connecticut contact:
Phone: 860-633-4466

Maine/New Hampshirecontact:
Phone: 207-854-3749

Massachusetts contact:
Phone: 781-575-1881

Rhode Island contact:
Phone: 401-732-1910

Vermont contact (in Albany, NY):
Phone: 518-489-5495

Muscular Dystrophy Family Foundation
3951 North Meridian Street, Suite 100
Indianapolis, IN 46208
Phone: 800-544-1213 or 317-923-6333
Fax: 317-923-6334
Email: mdff@mdff.org
www.mdff.org

ADDITIONAL WEB RESOURCES:

Parent Project Muscular Dystrophy
www.parentprojectmd.org

Founded by parents of children with Duchenne and Becker muscular dystrophies, this group funds research and disseminates information on research, supports an international conference, and provide a newsletter. Site includes links to information, resources, breaking news, and treatments, and addresses emotional issues.

ADDITIONAL LITERATURE:

Muscular Dystrophy in Children: A Guide for Families
by Siegel, I.M.
Demos Medical Publishing, 1999

A text for families, including information on symptoms, medical treatments, psychosocial issues, probable disease course and therapies.

Muscular Dystrophy: The Facts
by Emery, A.E.H.
Oxford University Press, 2nd Edition, 2000

Easy to understand book explains the complexities of muscular dystrophy, including daily life issues.

Moonrise: One Family, Genetic Identity, and Muscular Dystrophy
by Wolfson, P.
St. Martin's Press, 2004

Personal account, written by the mother of a son with Duchenne muscular dystrophy. Explores special education, prenatal diagnosis, and genetics. Used or in libraries.

NEUROFIBROMATOSIS

The Children s Tumor Foundation
95 Pine Street, 16th floor
New York, NY 10005
Phone: 800-323-7938 or 212-344-6633
Fax: 212-747-0004
Email: info@ctf.org
www.ctf.org

Northern New England Chapter (serves CT, ME, MA, NH, RI, VT)
75 McNeil Way
Suite 201
Dedham, MA 02026
Phone: 888-585-5316 or 508-879-5638
Fax: 781-326-4940
Email: mbraden@ctf.org

Neurofibromatosis, Inc.

P.O. Box 18246
Minneapolis, MN 55148
Phone: 800-942-6825 or 301-918-4600
Email: nfinfo@nfinc.org
www.nfinc.org

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

Listing of neurofibromatosis information on the web, including general information, genetics, support, news groups, home-pages, and information for children and adolescents.

ADDITIONAL LITERATURE:

Living with A 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 and professionals. Includes: Clinical aspects; gene structure, expression, and mutation; animal models; and disease treatment and prevention.

Niemann- Pick Disease

National Niemann-Pick Disease Foundation, Inc.

P.O. Box 49
401 Madison Avenue, Suite B
Fort Atkinson, WI 53538
Phone: 877-287-3672 or 920-563-0930
Fax: 920-563-0931
Email: nnpdf@idcnet.com
www.nnpdf.org

ADDITIONAL WEB RESOURCES:

International Center for Types A and B Niemann-Pick Diseases Mt. Sinai School of Medicine

www.mssm.edu/niemann-pick/

Includes information on the disease, its natural history, genetics.

ADDITIONAL LITERATURE:

The Official Parent s Sourcebook on Niemann-Pick Disease: A Revised and Updated Directory for the Internet Age

by Parker, J.N. and Parker, P.M., (eds)
Icon Health, 2002

Guide to looking for information online, finding a doctor, and locating the latest research.

Organic Acidemias

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

Osteogenesis Imperfecta

Osteogenesis Imperfecta Foundation, Inc.

804 West Diamond Avenue Suite 210
Gaithersburg, MD 20878
Phone: 800-981-2663 or 301-947-0083
Fax: 301-947-0456
Email: bonelink@oif.org
www.oif.org

Massachusetts contact:
Cheryl & Richard Manduca
Phone: 781-545-2521

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

ADDITONAL WEB RESOURCES:

The Bones Page
www.geocities.com/dr_plotkin

Basic information & links to other resources.

PARKINSON S DISEASE

American Parkinson Disease Association
135 Parkinson Avenue
Staten Island, NY 10305
Phone: 800-223-2732 or 718-981-8001
Fax: 718-981-4399
Email: apda@apdaparkinson.org
www.apdaparkinson.org

Connecticut Chapter:
27 Allendale Drive
North Haven, CT 06473
Phone: 888-400-2732 or 203-789-3936
Fax: 203-288-0546
Email: gladkt@hotmail.com
www.ctapda.com
See Connecticut web site for >15 local contacts.

Maine Chapter:
17 Blueberry Lane, Cottage 21
Falmouth, ME 04105
Carl Barker, President
Phone: 207-781-3070
Email: cnbarker@maine.rr.com

Massachusetts Chapter:
715 Albany Street, C329
Boston, MA 02118
Keith Ciccone, President
Phone: 800-651-8466 or 617-638-8466
Fax: 617-638-5354
Email: information@apdama.org
Alt. Email: Keith.ciccone@bmc.org
www.apdama.org
See Massachusetts site for > 15 local contacts.

New Hampshire Chapter:
P.O. Box 6212
Nashua, NH 03063
George McHugh, President
Phone: 603-305-0398 or 603-459-8040
Email: gmchugh@comcast.net

Rhode Island Chapter:
P.O. Box 41659
Providence, RI 02940-1659
Athol Cochrane, President
Phone: 401-823-5700
Email: RichapAPDA@aol.com
www.parkinsonsdisease-rhodeisland.org

Vermont Chapter:
Fletcher Allen Health Care
1 South Prospect Street
Burlington, VT 05401
Phone: 802-847-3366 or 888-763-3366
Email: Parkinsonsvt@surfglobal.net
www.parkinsonsvt.org
See Vermont web site for > 15 local contacts.

Parkinson s Disease Foundation, Inc.
1359 Broadway, Suite 1509
New York, NY 10018
Phone: 800-457-6676 or 212-923-4700
Fax: 212-923-4778
Email: info@pdf.org
www.pdf.org

ADDITIONAL WEB RESOURCES:

Parkinson s Information
www.parkinsonsinfo.com

Information about Parkinson's disease, a directory of 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. Available used and in libraries.

Parkinson s Disease: A Guide for Patient and Family
by Duvoisin, R.C. and Sage, J.
Lippincott, Williams & Wilkins, 5th Edition, 2001

In depth information on diagnosis, medications, management, and genetics.

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

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 relevant legislation and policies.

Low Protein Recipes
www.lowprotein.com

Delicious low protein recipes created by a mother of a child with homocystinuria.

National Coalition for PKU and Allied Disorders
www.pku-allieddisorders.org

Organization composed 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, 3rd Edition, 1997

Provides recipes for American style cooking, instructions for calculating nutrient content, and tips on handling special circumstances such as kid's parties.

PORPHYRIA

American Porphyria Foundation
4900 Woodway
Suite 780
Houston, TX 77056
Phone: 713-266-9617
Fax: 713-840-9552
Email: porphyrus@aol.com
www.porphyriafoundation.com

PRADER-WILLI SYNDROME

Prader-Willi Syndrome Association
5700 Midnight Pass Road, Suite 6
Sarasota, FL 34242
Phone: 800-926-4797 or 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
Email: pwsactchapter@yahoo.com

Prader-Willi Association of New England
New England Region (ME, MA, RI, NH, VT)
Sherie Bombardier
2 Ernest Street
Webster, MA 01570
Phone: 508-943-1400

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.

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, this book provides information on the condition and its management including medical, nutritional, psychological, educational, social, and therapeutic issues.

Prune Belly Syndrome

Prune Belly Syndrome Network, Inc.
P.O. Box 154
Beloit, WI 53512
Email: postmaster@prunebelly.org
www.prunebelly.org

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: NAPEStLouis@sbcglobal.net
www.pxenape.org

PXE, International
4301 Connecticut Avenue NW, Suite 404
Washington, DC 20008
Phone: 202-362-9599
Fax: 202-966-8553
Email: info@pxe.org
www.pxe.org

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

Rare Conditions

Rare Kids, Inc.
PO Box 69
East Walpole, MA 02320
Phone: 508-668-2850
Email: labruno33@comcast.net
www.Rarekids.org

Nonprofit organization established to support children afflicted with rare diseases. Rare Kids has also supported many charitable organizations which provide support and research for those afflicted with rare diseases including Families of SMA, The A-T Children's Project, Foundation for Ichthyosis and more.

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

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

Massachusetts - Chapter:
Lynn E. Donnelly, President
232 Winchester Street # 2
Brookline, MA 02446-2767
Phone: 617-739-3169
Email: Lynn_Donnelly@gillette.com

Massachusetts – Support Group:
Contact: Tina Kurys
Email: vzeeiz32@verizon.net

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

Connecticut Council of the Blind:
Alice Jackson
191 Centrebrook Road
Hamden, CT 06518
Email: ajackson212@comcast.net
members.tripod.com/~dmclean/ccb.html

Maine Chapter:
Mel Clarrage, President
221 Longfellow Street, Unit 2
Westbrook, ME 04092
Email: mclarrage1@maine.rr.com

Bay State Council of the Blind
Jerry Berrier, President
108 Bumble Bee Circle
Shrewsbury, MA 01545
Email: jerry.berrier@townisp.com
www.acb.org/baystate/

Vermont Council of the Blind:
Harriet G. Hall, President
95 Pine Grove Cemetery Road
Newport, VT 05855
Email: greenharriet@yahoo.com

American Foundation for the Blind
11 Penn Plaza, Suite 300
New York, NY 10001
Phone: 800-232-5463 or 212-502-7600
Fax: 212-502-7777
Email: afbinfo@afb.net
www.afb.org

ADDITIONAL WEB RESOURCES:

Visionchannel
www.visionchannel.net/retinitis
Includes overview of retinitis pigmentosa including 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 a 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 or 301-856-3334
Fax: 301-856-3336
Email: irsa@rettsyndrome.org
www.rettsyndrome.org

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 pertaining to 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
300 Rosewood Drive
Suite 105
Danvers, MA 01923
Phone: 800-722-4673 or 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)
59 Front Street
Binghamton, NY 13905
Phone: 800-867-0885 or 607-723-2239
FAX: 607-723-2039
Email: sdtrristate@aol.com
www.scleroderma.org/chapter/tristate/

New England Chapter (includes MA, ME, NH, VT, RI):
462 Boston Street, Suite 1-1
Topsfield, MA 01983
Tom Curran, Executive Director
Phone: 978-887-0658 or 888-525-0658
Email: tcurran@sfnewengland.org
www.scleroderma.org/chapter/newengland/

Scleroderma Research Foundation
220 Montgomery Street
Suite 1411
San Francisco, CA 94104
Phone: 800-441-2873 or 415-834-9444
Fax: 415-834-9177
Email: info@sclerodermaresearch.org
www.srfcure.org/srf/home.htm

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

Practical information provided by a leading expert in the field.

Scleroderma: A New Role for Patients and Families
by Brown, M.
Scleroderma Press, 2nd Edition, 2002

Written from the perspective of a patient and family, this is a useful resource for both medical and non-medical issues.

SICKLE CELL ANEMIA

Sickle Cell Disease Association of America
165 South Calvert Street, Suite 600
Baltimore, MD 21202
Phone: 800-421-8453 or 410-528-1555
Fax: 410-528-1495
Email: scdaa@sicklecelldisease.org
www.sicklecelldisease.org

Connecticut Chapters:
Southern Regional
Sickle Cell Association, Inc.
177 State Street, 3rd floor
Bridgeport, CT 06604
Phone: 888-745-2327 or 203-366-8710
Fax: 203-368-9071
Email: srsca2@aol.com

SCDAA- Connecticut Chapter
The Urban League Building
140 Woodland St., Suite 102
Hartford, CT 06511
Phone: 800-379-0119 or 860-527-0147 x145
Email: scdaa@iconn.net
www.sicklecellct.org

Satellite Offices:
226 Dixwell Avenue, 2nd Floor
New Haven, CT 06511
Phone: 203-498-4051
Fax: 203-498-4054
Email: scdaanh@iconn.net

Citizens for Quality Sickle Cell Care
P.O. Box 702
100 Arch Street
New Britain, CT 06050
Phone: 860-223-7222
Email: info@cqscc.org
www.CQSCC.org

Massachusetts Chapter:
Community Sickle Cell Support Group, Inc.
1542 Tremont St.
Roxbury, MA 02120
Phone: 617-427-4100
Fax: 617-262-3190
Email: cscsginc@aol.com
www.cscsginc.org

ADDITIONAL WEB RESOURCES:

Sickle Cell Information Center, Atlanta, GA
www.scinfo.org

Provides both the patient and the professional with 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, disease management , and new developments in the field.

SJOGREN S SYNDROME

Sjogren s Syndrome Foundation
8120 Woodmont Avenue
Suite 530
Bethesda, MD 20814
Phone: 800-475-6473 or 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

In Massachusetts:
Greater Boston Support Group Leader;
Lynn Epstein, MD
Phone: 617-636-3932 (through Dr. Athena Papas' office)

ADDITIONAL WEB RESOURCES:

SjS World
www.sjsworld.org

Online community that offers patients and families a place to meet and share experiences, books, and, medical information. Includes and email groups and chat rooms.

ADDITIONAL LITERATURE:

The New Sjogren Syndrome Handbook
by Wallace, D.J., Bromet, E.J., and the Sjogren Syndrome Foundation
Oxford University Press, 2005

A comprehensive and authoritative guide that has been extensively revised from their original edition. Designed for people with Sjogren syndrome and for physicians, it provides readers with the best medical and practical information available on this disorder. Includes information on symptoms, diagnosis, and treatment options and offers tips for daily living.

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 Foundation

P.O. Box 212
Georgetown, MA 01833
Phone: 978-352-5885
Contact: Cynthia Gold
Email: cgold@smithlemliopitz.org or info@smithlemliopitz.org
www.smithlemliopitz.org

Sotos Syndrome

Sotos Syndrome Support Association

P.O. Box 4626
Wheaton, IL 60189
Phone: 888-246-7772
Email: sssa@well.com
www.well.com/user/sssa
Email support groups available.

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 or 202-944-3285
Fax: 202-944-3295
Email: sbaa@sbaa.org
www.sbaa.org

Spina Bifida Association of Connecticut
P.O. Box 2545
Hartford, CT 06146
Phone: 800-574-6274 or 860-653-1976
Fax: 860-345-2600
Email: sbac@sbac.org
www.sbac.org

Massachusetts Spina Bifida Association
733 Turnpike Street, #282
North Andover, MA 01845
Phone: 888-479-1900 or 617-742-2574
Fax: 978-649-8725
Email: packard44@comcast.net
www.msbaweb.org

Additional web resources:

Children with Spina Bifida: A Resource Page for Parents

www.waisman.wisc.edu/~rowley/sb-kids/sb-awareness.html

Links to spina bifida organizations, articles, web sites, online discussion group. Information about 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

Provides parents with information, guidance and support to help meet their child's often intensive needs from birth through childhood.

Spinabilities: A Young Person s Guide to Spina Bifida

by Lutkenhoff, M. and Oppenheimer, S.G.
Woodbine House, 1997
This book helps young people who have spina bifida attain as much control over their own care and achieve as much independence as possible.

Tay-Sachs Disease

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 CT, NJ and NY)
1202 Lexington Avenue #288
New York, NY 10028
Phone: 888-354-7788 or 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

Follow links to Tay-Sachs Disease. Includes disease description and information about natural history, genetics and testing.

Additional literature:

Tay-Sachs Disease
by Desnick, R.J. and Kaback, M.M. Academic Press, 2001

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 or 718-321-2873
Fax: 718-321-3340
Email: info@cooleysanemia.org
www.cooleysanemia.org

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

Massachusetts Contact:
Rudi Viscomi
Phone: 617-332-5952

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, disease 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 or 718-224-2999
Fax: 718-279-9596
Email: ts@ts-usa.org
www.tsa-usa.org

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

Maine and New Hampshire Chapter:
Phone: 877-368-9800
Email tourette@thezac.com
www.thezac.com/tourette

Massachusetts Chapter:
Phone: 617-277-7589
Email Swiftcape@aol.com
www.tsamass.org

Rhode Island Chapter:
Phone: 401-301-9980
Email: tsri@ride.ri.net
www.ri.net/tsari

For Vermont info. contact National TSA.

ADDITIONAL WEB RESOURCES:

Tourette-Syndrome.com
www.tourette-syndrome.com

Comprehensive online community devoted to children and adults with Tourette syndrome.

ADDITIONAL LITERATURE:

Tourette Syndrome: Finding Answers and Getting Help
by Waltz, M.
Patient Center Guides, 2001

A support guide offering help to families living and dealing with Tourette syndrome.

Icy Sparks
by Rubio, G.H.
Penguin Books, 2001

This book is about a young girl who has Tourette, 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 Connection
P.O. Box 156
Boston, MA 02131
Contact Person: Judy (located in North Carolina)
Phone: 704-545-1921
Email: tom@tcconnection.org or judy@tcconnection.org
www.tcconnection.org

ADDITIONAL WEB RESOURCES:

Reflections on Treacher Collins Syndrome
www.treachercollins.org

Web site 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 or 301-562-9890
Fax: 301-562-9870
Email: info@tsalliance.org
www.tsalliance.org

Contact main office for support contacts in Connecticut, Massachusetts, New Hampshire, and Rhode Island.

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
MacKeith Press, 2003

Discusess correlation between new genetic and basic science data and the clinical presentation.

Turner Syndrome

Turner Syndrome Society of the United States
14450 TC Jester
Suite 260
Houston, TX 77014
Phone: 800-365-9944 or 832-249-9988
Fax: 832-249-9987
Email: tssus@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

Massachusetts and Southern New England Turner Syndrome Society
Contact: Beth Wheeler
1034 Maple Street
Mansfield, MA 02048
Phone: 508-339-3022
Email: Beth_Wheeler@comcast.net

Northern New England Turner Society (serving New Hampshire, Maine and Vermont)
Contact: Lori-Ann Pawlowski
38 Beaman Street
Laconia, New Hampshire 03246
Phone: 603-524-6011
Email: tssnnepa@hotmail.com

Rhode Island Chapter:
Contact: Debbie Pomerantz
24 Turner St. Unit 3
Warwick, Rhode Island 02886
Phone: 401-732-2136
Email: deb_pomerantz@hotmail.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

Additional web resources:

Urea Cycle Disorders
www.meadjohnson.com/metabolics/ureacycle.html

Detailed overview of the conditions and therapies.

Velo-Cardio-Facial Syndrome

Velo-Cardio-Facial Syndrome Educational Foundation, Inc.
P.O. Box 874
Milltown, NJ 08850
Phone: 866-823-7335 or 732-238-8803
Fax: 315-464-6593
Email: info@vcfsef.org
www.vcfsef.org
Contact main office for local chapters.

Northeast VCFS Support Group
Contact Maureen Anderson
2 Lansing Drive
Salem, NH 03079
Phone: 603-898-6332
Email: mladja@aol.com

Von Hippel-Lindau Syndrome

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

In Connecticut:
Email: northeast@vhl.org

In Maine:
Email: us-me@vhl.org

In Massachusetts:
Email: us-ma@vhl.org

In New Hampshire, Rhode Island and Vermont:
Email: usnortheast@vhl.org

Williams Syndrome

Williams Syndrome Association
P.O. Box 297
Clawson, MI 48017
Phone: 800-806-1871 or 248-244-229
Fax: 248-244-2230
Email: info@williams-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 on behavioral issues and research.

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 on the disorder including 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 linking genes with human behavior.

WILSON S DISEASE

Wilson s Disease Association International
1802 Brookside Drive
Wooster, OH 44691
Phone: 800-399-0266 or 330-264-1450
Fax: 509-757-6418
Email: wda@sssnet.com
www.wilsonsdisease.org

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

ADDITIONAL WEB RESOURCES:

Wilson s Disease Resources and Information
www.acsu.buffalo.edu/~drstall/wilsons.html

Web site written by physician who has Wilson's disease.

LOW COPPER DIET FOR WILSON S DISEASE
www.gicare.com/pated/edtgs17.htm

Gastroenterologist's web site 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

A guide for the physician with information about symptoms, diagnosis and treatment options for patients with Wilson's disease.

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

A guide for individuals with Wilson's disease. Helpful advice about diagnosis, treatment and quality of life issues.

IV.

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, is 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:
Describes a trait or disorder requiring the presence of only one copy 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.

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 cytogenetic 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:
In DNA, 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. Carriers are usually asymptomatic.

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:
Alteration in a gene present for the first time in one family member as a result of a mutation in the egg or sperm cell leading 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 through DNA analysis.

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. 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 person's 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 usually by the insertion of 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:
Analysis of DNA to look for a genetic alteration.

Genomics:
The study of the sequence, structure, and function of the genome. The complete DNA sequence in an individual or species.

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, (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 or 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.

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.

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 family.

Intron:
Non-coding sequence of DNA removed from mature 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 or 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 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 containing 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, 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 or 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 at least partly 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:
A natural variation in a gene, DNA sequence, or chromosome that has no adverse effect and occurs 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, before transferring to the mother's uterus only those embryos determined not to have inherited the abnormality 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 the development of a specific condition at some future point in time. 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, and resulting in an abnormal configuration which 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 is present on the X chromosome.

Reciprocal translocation:
A chromosomal rearrangement in which 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.

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 Y chromosomes and the acrocentric chromosomes 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 are 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 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 messenger RNA.

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.

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.

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.

SOURCES:

Genetics Home Reference — Your Guide to Understanding Genetic Conditions
A service of the U.S. National Library of Medicine Available at www.ghr.nlm.nih.gov. Accessed September 2004.

GeneTests: Medical Genetics Information Resource (database online)
Educational materials: Glossary. Copyright, University of Washington, Seattle. 1993-2004. Updated weekly. Available at http://www.genetests.org. Accessed September 2004.

Prepared by the CT Department of Public Health

V.

GENETIC SERVICES: CONNECTICUT

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

Maternal Fetal Medicine/Genetics
Phone: 203-384-3049 or 203-384-4724
Fax: 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 prevent irreversible problems ordeath.

*(CT Newborn Screening Designated Genetic Treatment Centers will be indicated by ** in listings.)*

CT NEWBORN SCREENING DESIGNATED ENDOCRINOLOGY REGIONAL TREATMENT CENTERS

CONNECTICUT CHILDREN'S MEDICAL CENTER
Pediatric Endocrinology Department
282 Washington Street
Hartford, CT 06106
Phone: 860-545-9370
Fax: 860-545-9371

YALE UNIVERSITY SCHOOL OF MEDICINE
Yale Department of Pediatric Endocrinology
3103, LMP
P.O. Box 208064
New Haven, CT 06520-8064
Phone: 203-764-9199
Fax: 203-764-9149

CT NEWBORN SCREENING DESIGNATED SICKLE CELL REGIONAL TREATMENT CENTERS

CT CHILDREN'S MEDICAL CENTER
Comprehensive Hemoglobin Disorders Treatment Center
282 Washington Street
Hartford, CT 06106
Phone: 860-545-9630
Fax: 860-545-9622

YALE SICKLE CELL SERVICE
Yale University School of Medicine
333 Cedar Street
LMP 4089
P.O. Box 208064
New Haven, CT 06520
Phone: 203-785-6662
Fax: 860-737-2461

Greenwich Hospital
5 Perryridge Road
Greenwich, CT 06830
Phone: 203-863-3000
www.greenhosp.org

Perinatal Genetic Counseling
Phone: 203-863-3917
Fax: 203-863-3467

Norwalk Hospital
Maple Street
Norwalk, CT 06856
www.norwalkhosp.org

Perinatology
Phone: 203-852-3354
Fax: 203-852-3610

Stamford Hospital
30 Shelburne Road
Stamford, CT 06904
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, First Floor
West Hartford, CT 06106
Phone: 860-523-6464
Administrative Phone: 860-523-6499
Fax: 860-523-6465
www.uchc.edu/genetics

GENERAL GENETICS CONSULTATION SERVICE **
Provides: up-to-date information about the heritability and cause of genetic disorders, such as birth defects, mental retardation and late onset adult diseases; evaluation, diagnosis and management of persons with genetic disorders; information about the recurrence risk to families with a genetic disorder; and supportive counseling to patients and their families.

HEREDITARY CANCER PROGRAM
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
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
Provides diagnostic evaluation of metabolic disorders that are detected by the Connecticut State Newborn Screening Program or by other evaluations.

OTHER COLLABORATIVE PROGRAMS
Hemophilia
Huntington Disease
Neurogenetics Program
Neuromuscular Disorders
Newborn Cystic Fibrosis Screening Program

EDUCATION
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
Provides information about the inheritance of genetic conditions, the cause for various birth defects, and the tests available to detect whether an individual or couple is at risk 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

OCCUPATIONAL AND ENVIRONMENTAL REPRODUCTIVE HAZARDS CLINIC
UConn Health Center
Dowling North Building, 3rd floor
Farmington, CT 06032
Phone: 800-325-5391 or 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

CONNECTICUT PREGNANCY EXPOSURE INFORMATION SERVICE
Phone: 800-325-5391 or 860-523-6419
Fax: 860-523-6465
wwwdocdb.uchc.edu/genetics/PregnancyExposure.htm

A state-funded program that provides 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.

Yale University School of Medicine

DEPARTMENT OF GENETICS
333 Cedar Street
New Haven, CT 06520
Phone: 203-785-2660
Fax: 203-785-3404
www.info.med.yale.edu/genetics

GENETIC CONSULTATION SERVICE **
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

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

GENETIC INBORN ERRORS OF METABOLISM SERVICE
Diagnosis, management and clinical care for patients with inborn errors of amino acid, organic acid, fatty acid metabolism and mitochondrial disorders. Newborn screening follow-up and assessment with rapid laboratory assessment and collaborative management with primary care physicians. Lysosomal storage disorders diagnosis and management. Molecular genetic testing with assessment of family members at risk.

EDUCATION
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
Appointment Phone: 203-785-5682
Administrative & Consult line: 203-785-2661
Fax: 203-785-7673

Provides a full array of prenatal genetic testing services.

Patients also seen at:

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

Waterbury Hospital
64 Robbins Street
Waterbury, CT 06721
www.waterburyhospital.org

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 or 203-401-4300
Fax: 203-785-5694

CANCER GENETIC COUNSELING PROGRAM
Yale Cancer Center
55 Church Street
Suite 402
New Haven, CT 06510
Phone: 203-764-8400
Fax: 203-764-8401
www.yalecancercenter.org/genetics/

Patients also seen at:

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

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
http://info.med.yale.edu/genetics/BDD/index.php

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

CYTOGENETICS (INCLUDING MOLECULAR CYTOGENETICS)
http://info.med.yale.edu/genetics/cytogenetics/cytogenetics.php

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

DNA DIAGNOSTICS LABORATORY
Phone: 203-785-5745
Fax: 203-785-7227

Testing for fragile X syndrome, Duchenne and Becker muscular dystrophies, Gorlin syndrome, Hemophilia A, MCAD , multiple endocrine neoplasia, and OTC.

VI.

GENETIC SERVICES: MAINE

Maine Genetics Program (MGP)

Ellie Mulcahy, RNC, Director
Division of Family Health
11 State House Station
Augusta, ME 04333-0011
Phone: 207-287-5357 or 1-800-698-3624
Fax: 207-287-4743

The Maine Genetics Program (MGP) is a statewide program that provides grants to agencies to assure the availability of comprehensive genetic services for the citizens of Maine. Services include risk assessment, laboratory and clinical diagnosis, counseling, case management and referral, and education and training to providers and consumers. The MGP also coordinates the Newborn Bloodspot and Newborn Hearing Screening Programs. The Newborn Bloodspot Screening Program screens all newborns for 28 conditions, which if left untreated, would cause mental retardation, other serious health problems or death. The Newborn Hearing Screening Program tests all newborns for possible hearing loss. Both the Newborn Bloodspot Screening Program and the Newborn Hearing Screening Program make referrals for treatment and comprehensive follow-up care.

Eastern Maine Medical Center Genetics Programs

PEDIATRIC SPECIALTY CLINICS
417 State Street
Webber East Suite 305
Bangor, ME 04401
Appointment Phone: 207-973-7559
or 1-877-366-3662 x7559
Administrative Phone: 207-973-7553
or 1-877-366-3662 x7553
http://emmc.org/Patient+Services/Child+and+Adolescent+Care+Center/default.htm

The Genetics Program provides consultation and management services through a number of specialty clinics at EMMC. The Program provides initial genetic evaluation, assessment and ongoing management. In addition to the general Genetics Clinic, the Genetics Program coordinates specialty clinics for the following conditions:

- Inborn Errors of Metabolism
- Spina Bifida
- Cystic Fibrosis
- Cleft Lip and Palate
- Hemophilia
- Muscular Dystrophy

The Genetics Program also provides Medical Genetics consultations and prenatal and preconception counseling for area physicians and women's health programs, including the Maternal Fetal Medicine Program at EMMC, as well as Developmental Evaluation Clinics.

All clients are seen regardless of ability to pay for services.

In addition to clinical services, the Genetics Program offers education and training to health professionals and other groups throughout the state.

Cancercare of Maine

417 State Street, Suite 20
Bangor, ME 04401-6600
Phone: 207- 973-7476 or 1-800-987-3005

The Cancer Risk Counseling Clinic provides assessment for individuals and families at risk for a variety of hereditary cancer syndromes. Genetic testing options, screening and preventative strategies are reviewed. An oncologist is available for consultation.

Maine medical Center, The Division of Genetics at the Barbara Bush Children s Hospital

MAINE PEDIATRIC SPECIALTY GROUP AND ENDOCRINOLOGY
887 Congress Street, Suite 320
Portland, ME 04102
Phone: 207-662-5522 or 1-800-860-6277
Fax: 207-662-5528
http://www.mmc.org/mmc_bush/clinical_services_prog_genetics.htm

The Division of Genetics at the Barbara Bush Children's Hospital provides Genetics services for individuals from primarily southern Maine and New Hampshire. Consultative and management services include dysmorphology evaluations, ongoing management of complex medical care for individuals with known diagnoses; family risk assessment; provision of genetic testing,; genetic counseling; evaluation, diagnosis and management of inborn errors of metabolism; enzyme replacement therapy services for lysosomal storage diseases; and prenatal consultations in complex cases. The Division of Genetics also provides essential education to primary care physicians and other subspecialists at MMC, as well as throughout the states of Maine and New Hampshire.

The Division is also responsible for the Barbara Bush Children's Hospital Metabolism Program and provides support for the Southern Maine Cleft Lip and Palate Program, the Cystic Fibrosis Clinic, the Hemophilia Clinic, the Maine Children's Cancer Program and the Spina Bifida Program. Medical Genetics consultations are also provided to the Division of Maternal Fetal Medicine at MMC and the Genetics staff at the Maine Center for Cancer Medicine.

All clients are seen regardless of ability to pay for services.

Maine Medical Center, Division of The Cancer Genetics Program

CANCER RISK AND PREVENTION CLINIC
AT MAINE MEDICAL CENTER
100 US Route 1
Scarborough, ME 04074
Phone: 207-885-7787
Fax: 207-885-8500
http://www.mmc.org/mmc_services/mainecenterforcancermedicine.htm

The Cancer Risk and Prevention Clinic provides a family history evaluation and genetic risk assessment for individuals and families primarily in Southern Maine and New Hampshire. A visit includes a review of an individual's personal and/or family history of cancer in order to evaluate possible hereditary cancer susceptibility disorders (HCSDs). The genetic counselor explains genetics, inheritance, genetic syndrome differentials, available testing, possible testing outcomes, and research possibilities. A visit continues with the physician who provides a personalized risk assessment. While direct patient care is the main focus, the Cancer Genetics Program also provides essential services and education to primary care physicians and other subspecialists at MMC, as well as throughout the states of Maine and New Hampshire.

Other Related Services with a Genetic Component

CHILDREN WITH SPECIAL HEALTH CARE NEEDS PROGRAM, (CSHN)
Toni Wall, MPA, Director
Division of Family Health
11 State House Station
Augusta, Maine 04333
Phone 207-287-5139 or 1-800-698-3624.
http://www.maine.gov/dhhs/boh/cshn

The Children with Special Health Needs Program (CSHN) serves children who have a serious physical condition(s) that requires extensive health and related services.

Chronic physical conditions that are covered by the CSHN Program include, but are not limited to, the following: asthma and other respiratory disorders, blood disorders, cardiac disorders, childhood cancer, chronic ear infections, cleft

lip and/or cleft palate, other craniofacial anomalies, cystic fibrosis, developmental delays, diabetes and other endocrine disorders, gastrointestinal disorders, genitourinary disorders, juvenile arthritis, orthopedic disorders, PKU and other inborn errors of metabolism, seizure disorders, skin disorders, spina bifida, and vision disorders.

The Program pays for diagnosis and medical care for children who meet the medical and income guidelines.

In addition, the Program supports and/or administers the following clinics:

Developmental Evaluation Clinics are available at Cary Medical Center, Eastern Maine Medical Center, MaineGeneral Medical Center in Waterville, and The Child Health Center in Auburn.

Maine Cleft Lip and Palate Clinic is supported and administered by CSHN Program in two regional sites: Bangor and Portland.

Maine Medical Center, Hemophilia Treatment Center

Maine Children's Cancer Program
100 U.S. Route One
Scarborough, ME 04074
Phone: 207-885-7565
Fax: 207-885-7577

Maine Medical Center provides comprehensive clinical services to individuals with hemophilia and other coagulation disorders. Genetic counseling services and coordination of DNA analysis for carrier testing and prenatal diagnosis are arranged when indicated.

The Foundation for Blood Research

Division of Genetics
8 Nonesuch Road
Scarborough, Maine 04074
Phone: 207-883-4131
Fax: 207-885-0807
www.fbr.org/genetics/genetics.html

The Division of Genetics at the Foundation for Blood Research (FBR) focuses on the integration of genetics into public health and private healthcare systems in Maine. Immediately available to primary and allied healthcare personnel are CME/CEU lectures and presentations offered either in person or via interactive TV (telemedicine), as well as publications.

The molecular diagnostic laboratory is involved in research to test the feasibility of DNA-based population screening protocols for genetic disorders such as cystic fibrosis and hereditary hemochromatosis.

Pregnancy Environmental Hotline:

Phone: 781-466-8474 or 800-322-5041 (in MA)
(also serves MA, NH and RI)

VII.

GENETIC SERVICES: MASSACHUSETTS

Massacusetts Department of Public Health

Office of Genomics and Health
250 Washington Street, 4th floor
Boston, MA 02108
Phone: 617-624-5070

The Massachusetts Genetics Program (MGP) is based within the Office on Health and Disability of the Massachusetts Department of Public Health. The MGP aims to inform consumers and professionals about medical and scientific developments in human genetics and associated ethical, legal and social issues that affect infants, children, adolescents and adults. The MGP strives to ensure that all individuals have access to culturally and linguistically appropriate, family-centered genetics services.

Baystate Medical Center

759 Chestnut Street
Springfield, MA 01199

CLINICAL AND REPRODUCTIVE GENETICS
Department of Obstetrics and Gynecology
Administrative Phone: 413-794-8890
Appointment Phone: 413-794-2222
Fax: 413-794-1666
Services: Prenatal, Pediatric, Adult, Cancer Genetics

The Genetics program at Baystate Medical Center offers comprehensive clinical genetics care, laboratory services and education for patients and healthcare professionals.

The clinical program includes services across the lifespan including prenatal and preconception assessment, pediatric and adult evaluations, and cancer risk assessment consultations.

Genetic laboratory services at Baystate Medical Center include cytogenetics, maternal serum screening, and molecular diagnostics.

Beth Israel Deaconess Hospital

330 Brookline Avenue
Boston, MA 02115

Clinical Genetics Program
Phone: 617-667-7110
Fax: 617-667-1551
Services: Prenatal diagnosis

Cancer Risk Assessment Program
Phone: 617-667-1905
Fax: 617-667-1551

Boston University Medical Center

715 Albany Street
W-4th Floor
Boston, MA 02118
(Mailing address)

THE CENTER FOR HUMAN GENETICS
700 Albany Street
Building CABR, Suite 408
Boston, MA 02118
(Clinical Site)
Phone: 617-638-7083
Fax: 617-638-7092
Services: Prenatal, pediatric, adult genetics; cytogenetic, biochemical, maternal serum screening, and molecular testing
http://www.bumc.bu.edu/hg

BOSTON COMPREHENSIVE SICKLE CELL CENTER
820 Harrison Ave., FGH 2nd Floor
Boston, MA 02118
Phone: 617-414-5727
Fax: 617-414-5739
Services: Testing, counseling for sickle cell anemia and other hemoglobinopathies, education, research

CENTER OF EXCELLENCE IN SICKLE CELL DISEASE
88 East Newton Street
Evans 248
Boston, MA 02118
Phone: 617-414-1020
Fax: 617-414-1021
www.bu.edu/sicklecell

Services: Research studies involving sickle cell anemia and other hemoglobinopathies

Brigham & Women s Hospital

75 Francis Street
Boston, MA 02115

CENTER FOR FETAL MEDICINE AND PRENATAL GENETICS
Phone: 617-732-4208
Fax: 617-264-6310
http://www.brighamandwomens.org/mfm/center4fetalgeneticshome.asp

The center focuses on the diagnosis and treatment of fetal disease with a comprehensive genetic counseling service, a fetal treatment program, and expansion of pre-implantation genetic diagnosis.

OUTREACH CLINICS:

Newton Wellesley Hospital

Maternal Fetal Medicine
2014 Washington Street
Newton, MA 02462
Phone: 617-243-5909
Fax: 617-243-5775

Women's Health Center

One Hutchinson Drive
Danvers, MA 01923
Phone: 978-739-6900
Fax: 978-774-9635

Exeter Hospital

Millbrook Office Park
118 Portsmouth Avenue, Suite 102
Stratham, NH 03885
Phone: 603-777-5002
Fax: 603-778-9290

Children s Hospital Boston

300 Longwood Avenue
Boston, MA 02115

CLINICAL GENETICS AND METABOLISM
Phone: 617-355-6394 or 617-355-4695
Fax: 617-730-0466
http://www.childrenshospital.org

The Division of Genetics at Children's Hospital Boston offers state-of-the-art diagnostic and clinical management for children, adolescents, and adults with genetic conditions, including inborn errors of metabolism. Patients are evaluated by a multi-disciplinary team of physicians, nurses and counselors who specialize in genetics, syndrome identification, metabolic disease, and birth defects.

OUTREACH SITES:

South Shore Hospital

851 Main Street
Weymouth, MA 02190
Phone: 781-331-4715
Fax: 781-335-5628

Lexington Medical Center

482 Bedford Street
Lexington, MA 02420
Phone: 781-672-2100
Fax: 781-672-2145

OTHER SERVICES:

Cardiovascular Genetics

Phone: 617-355-2079
Fax: 617-739-3784
Services: Evaluation for cardiac birth defects and inherited cardiac malformations

Dana-Farber Cancer Institute

44 Binney Street
Boston, MA 02115

CANCER RISK AND PREVENTION
Phone: 617- 632-2178
Fax: 617-632-6811
www.dana-farber.org/pat/cancer/

A team of specialists combines the latest medical and scientific knowledge with the most advanced surveillance, diagnostic, and prevention methods. Provides thorough evaluation and risk assessment, including genetic testing for cancer susceptibility, and educates patients about cancer risk and its implications for themselves and family members.

Harvard Vanguard Medical Associates

133 Brookline Avenue
Boston, MA 02215

GENETICS DEPARTMENT
Phone: 617-421-3320
Fax: 617-421-2989
www.harvardvanguard.org
Services: Prenatal, Pediatric, Adult and Cancer Genetics

Massachusetts General Hospital

Warren 801
55 Fruit Street
Boston, MA 02114-2696
www.hpcgg.org/LMM/

GENETICS & TERATOLOGY UNIT
Phone: 617-726-1742
Fax: 617-724-1911
Services: pediatric, prenatal counseling, adult genetics

CENTER FOR CANCER RISK ANALYSIS
55 Fruit Street
Boston, MA 02114
Phone: 617-724-1971
Fax: 617-726-9418

PARTNERS CENTER FOR HUMAN GENETICS AND GENOMICS
Administrative Office:
Simches Research Center
Room 2222
185 Cambridge Street
Boston, MA 02114

Clinical Office:
Yawkey Center for Outpatient Care
6th Floor 6C
55 Fruit Street
Boston, MA 02114
Phone: 617-726-1561
Fax: 617-726-1566
Services: Pediatric and adult genetics

DEVELOPMENTAL NEUROGENETICS
Administrative Office:
CRP Building North
5th Floor Suite 5240
185 Cambridge Street
Boston, MA 02114

Clinical Office:
Yawkey Center for Outpatient Care
6th Floor 6B
55 Fruit Street
Boston, MA 02114
Phone: 617-726-5732
Fax: 617-724-9620
Services: Evaluation and testing for neurogenetic disorders, including Fabry disease, Gaucher disease and mitochondrial disorders

National Birth Defects Center

40 Second Avenue, Suite 520
Waltham, MA 02451
Phone: 781-466-9555
Fax: 781-487-2361
www.thegenesisfund.org
Services: Diagnostic evaluation and testing, prenatal counseling

PREGNANCY ENVIRONMENTAL HOTLINE:
Phone: 781-466-8474 or 800-322-5041 (in MA)
(also serves ME, NH and RI)

New England Medical Center

750 Washington Street
Boston, MA 02111

CLINICAL GENETICS & METABOLISM SERVICES AT FLOATING HOSPITAL FOR CHILDREN
Phone: 617-636-5462
Fax: 617-636-0745
www.tufts-nemc.org/home/departments/pedi/pedgen.htm
Services: Pediatric genetic evaluation

OUTREACH SITES:

Worcester Medical Center

Laurie Demmer, MD
Phone: 617-636-7790

St. Anne s Hospital

Jodi Hoffman, MD
Fall River, MA
Phone: 508-235-5285

CENTER FOR PERINATAL DIAGNOSIS
Phone: 617-636-4277
Fax: 617-636-4273

OUTREACH SITES:

Lowell General Hospital

Maternal-Fetal Medicine Department
295 Varnum Avenue
Lowell, MA 01854
Phone: 978-937-6000
Fax: 978-937-6809

Lawrence General Hospital

Maternal-Fetal Medicine Department
46 Prospect Street
Lawrence, MA 01841
Phone: 978-683-4000 x 2901
Fax: 978-946-8184

South Shore Hospital

Maternal-Fetal Medicine Department
55 Fogg Road
South Weymouth, MA 02190
Phone: 781-340-8430
Fax: 781-340-4344

MetroWest Medical Center

Maternal-Fetal Medicine Department
115 Lincoln Street
Framingham, MA 01702
Phone: 508-383-1436
Fax: 508-383-1497

Melrose/Wakefield Office

Maternal-Fetal Medicine Department,
Perinatal Diagnosis
50 Rowe Street Suite 700
Melrose, MA 02176
Phone: 781-662-4688
Fax: 781-662-5394

Women s Medical Arts Building, 3rd floor

Maternal-Fetal Medicine Department
85 Herrick Street
Beverly, MA 01915
Phone: 978-922-3000 x 4010

University of Massachusetts memorial Medical Center

55 Lake Avenue North
Worcester, MA 01605

GENETICS CLINIC
Phone: 508-856-3949
Appointment Phone: 508-856-5695
Fax: 508-334-3525
Services: Prenatal, pediatric, cancer and adult genetic counseling

This program provides comprehensive services including diagnosis and management of patients with congenital malformations, genetic syndromes, chromosomal disorders and inborn errors of metabolism. Diagnostic evaluation of children with developmental delay and mental retardation is available. In addition, genetic counseling is provided for individuals, couples and families who may be at a risk for inherited diseases. Specific diagnostic, carrier, and/or prenatal testing is arranged as indicated.

Providers work closely with the Cytogenetics and Molecular Diagnostic Laboratories in the Department of Pathology at the University of Massachusetts Medical Center. Close alliance is also shared with the departments of Neonatology and Perinatal Medicine at the Medical Center of Central Massachusetts.

OUTREACH SITES:

Milford-Whitinsville Regional Hospital

Women's Pavilion
14 Prospect Street
Milford, MA 01757
Phone: 508-473-1190 x 2859
Fax: 508-473-1257

Health Alliance Hospital

60 Hospital Road
Leominster, MA 01453
Phone: 978-466-2660

VIII.

GENETIC SERVICES: NEW HAMPSHIRE

New Hampshire Department of Health

Newborn Screening Program
Marcia Lavochkin, RN BSN
Program Coordinator
29 Hazen Drive
Concord, NH 03301
Phone: 603-271-4225
Fax: 603-271-4519
www.dhhs.state.nh.us or www.nh.gov

The Newborn Screening Program monitors the blood screening of all infants born in NH for potentially serious disorders and ensures immediate follow-up on abnormal result.

Dartmouth- Hitchcock Medical Center

One Medical Center Drive
Lebanon, NH 03756

Medical Genetics Clinic: 603-653-6044
Prenatal Diagnosis Program: 603-653-6025
Neurometabolism Program: 603-653-9668
Familial Cancer Program: 603-653-3541
or 800-639-6918
Fax: 603-653-3585
http://www.dhmc.org/webpage.cfm?site_id=2&org_id=136&gsec_id=0&sec_id=0&item_id=3692

Elliott Hospital

275 Mammoth Road
Suite One
Manchester, NH 03109
Phone: 603-663-8611
Fax: 603-668-0164
www.elliothospital.org/services/genetic_counseling.html

Services include cancer genetic counseling, prenatal genetic counseling, pediatric and adult genetic counseling on a limited basis.

Pregnancy Environmental Hotline:

Phone: 781-466-8474 or 800-322-5041 (in MA)
(also serves MA, ME and RI)

IX.

GENETIC SERVICES: RHODE ISLAND

Pregnancy Environmental Hotline:

Phone: 781-466-8474 or 800-322-5041 (in MA)
(also serves ME, NH and RI)

Rhode Island Department of Health

NEWBORN SCREENING PROGRAMS
3 Capitol Hill, Room 302
Providence, RI 02908-5097
Ellen Amore, MS, Newborn Screening Program Manager
Phone: 401-222-4601
Family Health Information Line 1-800-942-7434
www.health.ri.gov/genetics/newborn.php

The Rhode Island Department of Health (HEALTH) has integrated genetics into many different programs. The department has two Genetics Committees. Information is available at **http://www.health.ri.gov/genetics/committees.php**

- Genetics Core Team — Includes representatives from a wide variety of programs involving genetics. The team coordinates implementation of the State Genetics Plan.

- Genetics Advisory Committee — Meets quarterly to advise the Director of HEALTH on genetic issues.

Rhode Island Hospital and Hasbro Children s Hospital

593 Eddy Street
Providence, RI 02903

GENETIC COUNSELING CENTER
Phone: 401-444-8361
Fax: 401-444-3288
www.lifespan.org

Provides genetic counseling and diagnostic services for children, adults and families with genetic disorders or birth defects. Genetic counseling for hereditary cancer families and cancer risk assessment is provided in collaboration with the Comprehensive Cancer Center at Rhode Island Hospital.

CENTER FOR SPECIAL CHILDREN (CSC)
Phone: 401-444-5685
Fax: 401-444-6115

The Center for Special Children (CSC) at Hasbro Children's Hospital is a site for the evaluation and treatment of children with neurological, developmental, behavioral, metabolic, and genetic disorders.

Specialty providers include Child Neurology, Developmental/Behavioral Pediatrics, Genetics, Metabolic Disorders, Neurosurgery, Neuro-rehabilitation,Orthopedics, Urology, and Psychology. Providers work in collaboration with occupational, physical and speech therapists, as well as social workers and parent consultants.

CYSTIC FIBROSIS CENTER
Phone: 401-793-8560
Fax: 401-793-8561

SICKLE CELL AND HEMATOLOGICAL DISORDERS
Phone: 401-444-5241
Fax: 401-444-3872

PEDIATRIC ENDOCRINOLOGY AND METABOLISM
Phone: 401-444-5504
Fax: 401-444-2534

Women and Infants Hospital

PRENATAL DIAGNOSIS CENTER
79 Plain Street
Providence, RI 02903
Phone: 401-453-7510
Fax: 401-453-7517
http://www.womenandinfants.com/body.cfm?id=92&action=detail&ref=44

The Prenatal Diagnosis Center provides genetic counseling before conception and during pregnancy as well as screening and diagnostic testing during pregnancy. Genetic counselors, nurses and physicians staff the center. Satellite clinics are located at South County Hospital (Wakefield, RI), Charlton Memorial Hospital (Fall River, MA), and New Bedford, MA.

PROGRAM IN WOMEN S ONCOLOGY
101 Dudley Street
Providence, RI 02905
Phone: 401-453-7540
Fax: 401-453-7785
http://www.womenandinfants.com/body.cfm?id=187

Provides cancer risk assessment and prevention counseling at Women and Infants Hospital, Kent Hospital, Westerly Hospital and in New London, Connecticut. Patients receive a comprehensive analysis of their cancer risk based upon medical, familial, and lifestyle factors. A personal approach to cancer prevention may include recommendations for medical screenings, lifestyle modifications, genetic testing and chemopreventive strategies.

Patients also seen at:

NEW LONDON CLINIC:
Phone: 860-439-1770
Fax: 860-447-2854

X.

GENETIC SERVICES: VERMONT

Vermont Department of Health

Newborn Screening and Universal
Newborn Hearing Screening
Children with Special Health Needs (CSHN)
P.O. Box 70
108 Cherry Street
Burlington, VT 05402
Newborn Screening Phone: 802-951-5180
Universal Newborn Hearing Screening
Phone: 802-865-1330
Consumer Scholarships Phone: 802-863-7338
Fax: 802-951-1218
http://www.healthyvermonters.info/hi/cshn

CSHN also has clinic services including: cardiology, child development, craniofacial/cleft lip and palate, cystic fibrosis, epilepsy and neurology, hand structure and function, hearing outreach program, hemophilia, juvenile rheumatoid arthritis, metabolic, muscular dystrophy, myelomeningocele, orthopedics, and rhizotomy (cerebral palsy).

The Vermont Regional Genetics Center

112 Colchester Avenue
Burlington, VT 05401
Phone: 802-847-4310
Fax: 802-847-4664

Offers prenatal, pediatric, adult and cancer genetic counseling and testing, as well as a free teratogen information service.

Vermont Pregnancy Risk Information

Phone: 800-932-4609