

ORGANIZATIONS

The following is an alphabetical compilation of organizations listed in the *Resources* section of the main body entries. Although the list is comprehensive, it is by no means exhaustive. It is a starting point for gathering further information. Many of the organizations listed provide information for multiple disorders and have links to additional related websites. Email addresses and web addresses listed were provided by the associations; Thomson Gale is not responsible for the accuracy of the addresses or the contents of the websites.

5p– Society

7108 Katella Ave. #502
Stanton, CA 90680
Phone: (888) 970-0777
Website: <http://www.fivepminus.org>

22q13 Deletion Syndrome Foundation

250 East Broadway
Maryville, TN 37804
Phone: (800) 932-2943
Website: <http://www.22q13.com>

A

A-T Children's Project

668 South Military Trail
Deerfield Beach, FL 33442
Phone: (800) 5-HELP-A-T
Website: <http://www.atcp.org>

A-T Medical Research Foundation

5241 Round Meadow Road
Hidden Hills, CA 91302
Website: <http://www.pathnet.medsch.ucla.edu/people/faculty/gatti/gatsign.htm>

AboutFace International

123 Edwards Street, Suite 1003
Toronto, ON M5G 1E2
Canada
Phone: (800) 665-FACE
E-mail: info@aboutfaceinternational.org
Website: <http://www.aboutfaceinternational.org>

AboutFace USA

PO Box 458
Crystal Lake, IL 60014
Phone: (312) 337-0742 or (888) 486-1209
E-mail: aboutface2000@aol.com
Website: <http://www.aboutface2000.org>

Achromatopsia Network

C/O Frances Futterman
PO Box 214
Berkeley, CA 94701-0214
Website: http://www.achromat.org/how_to_join.html

Acid Maltase Deficiency Association (AMDA)

PO Box 700248
San Antonio, TX 78270-0248
Phone: (210) 494-6144 or (210) 490-7161
Fax: (210) 490-7161 or (210) 497-3810
Website: <http://www.amda-pompe.org>

Agensis of the Corpus Callosum (ACC) Network

University of Maine
Merrill Hall, Room 18, 5749
Orono, ME 04469-5749
Phone: (207) 581-3119
E-mail: um-acc@maine.edu

Aicardi Syndrome Awareness and Support Group

29 Delavan Ave.
Toronto, ON M5P 1T2
Canada
Phone: (416) 481-4095

Aicardi Syndrome Foundation

450 Winterwood Dr.
Roselle, IL 60172
Phone: (800) 373-8518.
Website: <http://www.aicardi.com>

AIS Support Group (AISSG)

PO Box 269, Banbury
Oxon, OX15 6YT
United Kingdom
Website: <http://www.medhelp.org/www/ais>

AKU Hotline

Website: <http://www.goodnet.com/~ee72478/enable/hotline.htm>

Alcoholics Anonymous World Services

PO Box 459, Grand Central Station
New York, NY 10163
Phone: (212) 870-3400

Alexander Graham Bell Association for the Deaf, Inc.

3417 Volta Place NW
Washington, DC 20007-2778.
Phone: (800) 432-7543
Website: <http://www.agbell.org>

Allergy and Asthma Network. Mothers of Asthmatics, Inc.

2751 Prosperity Ave., Suite 150
Fairfax, VA 22031
Phone: (800) 878-4403
Fax: (703) 573-7794

Alliance of Genetic Support Groups

4301 Connecticut Ave. NW, Suite 404
Washington, DC 20008

Phone: (202) 966-5557
 Fax: (202) 966-8553
 Website: <http://www.geneticalliance.org>

Alpha 1 National Association
 8120 Penn Ave. South, Suite 549
 Minneapolis, MN 55431
 Phone: (612) 703-9979 or (800) 521-3025
 E-mail: julie@alpha1
 Website: <http://www.alpha1.org>

Alpha One Foundation
 2937 SW 27th Ave., Suite 302,
 Miami, FL 33133.
 Phone: (305) 567-9888 or (877) 228-7321.
 E-mail: mservern@alphaone.
 Website: <http://www.alphaone.org>

Alpha to Alpha
 RR#5 Box 859
 Warsaw, MO 65355
 Phone: (660) 438-3045
 Website: <http://www.alpha2alpha.org>

AlphaNet
 Phone: (800) 557-2638
 Website: <http://www.alphanet.org>

ALS Association of America (ALSA)
 27001 Agoura Road, Suite 150
 Calabasas Hills, CA 91301-5104
 Phone: (818) 800-9006
 Fax: (818) 880-9006
 Website: <http://www.alsa.org>

Alzheimer's Association
 919 North Michigan Ave., Suite 1000
 Chicago, IL 60611-1676
 Phone: (800) 272-3900

Alzheimer's Disease Education and Research (ADEAR)
 National Institute on Aging, P.O. Box 8250
 Silver Spring, MD 20907-8250
 Phone: (800) 4388-4380
 Website: <http://www.alzheimers.org>

Alzheimer's Disease International
 45/46 Lower Marsh
 London, SE1 7RG
 United Kingdom
 Phone: (+44 20) 7620 3011
 E-mail: adi@alz.co.uk
 Website: <http://www.alz.co.uk>

Ambiguous Genitalia Support Network
 PO Box 313
 Clements, CA 95227-0313
 Phone: (209) 727-0313
 Fax: (209) 727-0313
 E-mail: agsn@jps.net
 Website: <http://www.stepstn.com>

AMD Alliance International
 PO Box 550385
 Atlanta, GA 30355
 Phone: (877) 263-7171
 Website: <http://www.amdalliance.org>

American Academy of Allergy, Asthma & Immunology
 611 E. Wells Street
 Milwaukee, WI 53202
 Phone: (414) 272-6071
 Fax: (414) 272-6070
 Website: <http://www.aaaai.org>

American Academy of Dermatology
 PO Box 4014, 930 N. Meacham Road
 Schaumburg, IL 60168-4014
 Phone: (847) 330-0230
 Fax: (847) 330-0050
 Website: <http://www.aad.org>

American Academy of Ophthalmology
 PO Box 7424
 San Francisco, CA 94120-7424
 Phone: (415) 561-8500
 Website: <http://www.eyenet.org>

American Academy of Orthopaedic Surgeons
 P.O. Box 2058
 Des Plaines, IL 60017
 Phone: (800) 824-2263
 Website: <http://www.aaos.org>

American Academy of Pediatrics
 141 Northwest Point Boulevard
 Elk Grove Village, IL 60007-1098
 Phone: (847) 434-4000
 Fax: (847) 434-8000
 Website: <http://www.aap.org/visit/contact.htm>

American Association for Klinefelter Syndrome Information and Support (AAKSIS)
 2945 W. Farwell Ave.
 Chicago, IL 60645-2925
 Phone: (773) 761-5298 or (888) 466-5747
 Fax: (773) 761-5298
 Website: <http://www.aaksis.org>
 E-mail: aaksis@aaksis.org

American Association for Pediatric Ophthalmology and Strabismus
 Website: <http://med-aapos.bu.edu/>

American Association of the Deaf-Blind
 814 Thayer Ave., Suite 302
 Silver Spring, MD 20910
 Phone: (301) 588-6545, (301) 495-4403
 TTY Phone: (301) 495-4402
 Fax: (301) 495-4404
 Email: info@aadb.org
 Website: <http://www.aadb.org>

American Association of Kidney Patients
 3505 E. Frontage Rd., Ste. 315
 Tampa, FL, 33607-1796
 Phone: (800) 749-2257, (813) 636-8100
 Fax: (813) 636-8122
 Email: info@aakp.org
 Website: <http://www.aakp.org>

American Association on Mental Retardation (AAMR)
 444 North Capitol Street NW, Suite 846
 Washington, DC 20001-1512
 Phone: (800) 424-3688.
 Website: <http://www.aamr.org>

American Cancer Society
 1599 Clifton Road NE
 Atlanta, GA 30329
 Phone: (800) 227-2345
 Website: <http://www.cancer.org>

American Cleft Palate-Craniofacial Association
 104 South Estes Dr., Suite 204
 Chapel Hill, NC 27514
 Phone: (919) 993-9044
 Fax: (919) 933-9604
 Website: <http://www.cleftline.org>

American College of Obstetricians and Gynecologists
 PO Box 96920, 409 12th Street SW
 Washington, DC 20090-6920
 Website: <http://www.acog.org>

American College of Rheumatology
 1800 Century Place, Suite 250
 Atlanta, GA 30329
 Phone: (404) 633-3777
 Website: <http://www.rheumatology.org>

American Council of the Blind
 1155 15th Street NW, Suite 720
 Washington, DC 20005
 Phone: (202) 467-5081 or (800) 424-

8666
Website: <http://www.acb.org>

American Diabetes Association
1701 N. Beauregard Street
Alexandria, VA 22311
Phone: (703) 549-1500 or (800) 342-2383
Website: <http://www.diabetes.org>

American Epilepsy Society
342 North Main Street
West Hartford, CT 06117
Phone: (860) 586-7505
Fax: (860) 586-7550
E-mail: info@aesnet
Website: <http://www.aesnet.org>

American Foundation for the Blind
11 Penn Plaza, Suite 300
New York, NY 10001
Phone: (800) 232-5463

American Foundation for Urologic Disease, Inc.
1128 North Charles Street
Baltimore, MD 21201-5559
Phone: (410) 468-1808
Website: <http://www.afud.org>

American Hair Loss Council
Phone: (888) 873-9719
Website: <http://www.ahlc.org>

American Heart Association
7272 Greenville Ave.
Dallas, TX 75231-4596
Phone: (214) 373-6300 or (800) 242-8721
E-mail: inquire@heart
Website: <http://www.americanheart.org>

American Hemochromatosis Society, Inc.
777 E. Atlantic Ave., PMB Z-363
Delray Beach, FL 33483-5352
Phone: (561) 266-9037 or (888) 655-IRON (4766)
E-mail: ahs@emi.net
Website: <http://www.americanhs.org>

American Kidney Fund
Suite 1010, 6110 Executive Boulevard
Rockville, MD 20852
Phone: (899) 638-8299

American Liver Foundation
75 Maiden Lane, Suite 603
New York, NY 10038
Phone: (800) 465-4837 or (888) 443-7222

Website: <http://www.liverfoundation.org>

American Lung Association
1740 Broadway
New York, NY 10019-4374
Phone: (212) 315-8700 or (800) 586-4872
Website: <http://www.lungusa.org>

American Macular Degeneration Foundation
PO Box 515
Northampton, MA 01061-0515
Phone: (413) 268-7660
Website: <http://www.macular.org>

American Medical Association
Washington Office
1101 Vermont Ave. NW
Washington, DC 20005
Phone: (202) 789-7400
Website: <http://www.ama-assn.org>

American Nystagmus Network
PO Box 45
Jenison, MI 49429-0045
Website: <http://www.nystagmus.org>

American Optometric Association
243 North Lindbergh Boulevard
St. Louis, MO 63141
Phone: (314) 991-4100
Website: <http://www.aoanet.org>

American Porphyria Foundation
PO Box 22712
Houston, TX 77227
Phone: (713) 266-9617
Website: <http://www.enterprise.net/apf/>

American Pseudo-Obstruction & Hirschsprung's Society
158 Pleasant Street
North Andover, MA 01845
Phone: (978) 685-4477

American Psychiatric Association
1400 K Street NW
Washington, DC 20005
Phone: (202) 682-6220

American Sleep Disorders Association
1610 14th Street NW, Suite 300
Rochester, MN 55901
Phone: (507) 287-6006

American Society for Deaf Children
PO Box 3355
Gettysburg, PA 17325

Phone: (800) 942-ASDC or (717) 334-7922 (v/tty)
Email: asdc@deafchildren.org
Website: <http://www.deafchildren.org>

American Society for Dermatologic Surgery
1567 Maple Ave.
Evanston, IL 60201
Phone: (708) 869-3954

American Society for Reproductive Medicine
1209 Montgomery Highway
Birmingham, AL 35216-2809
Phone: (205) 978-5000
E-mail: asrm@asrm
Website: <http://www.asrm.org>

American Society of Human Genetics
9650 Rockville Pike
Bethesda, MD 20814-3998
Phone: (301) 571-1825
Website: <http://www.faseb.org/genetics/ashg/ashgmenu.htm>

American Society of Hypertension
515 Madison Ave., Suite 1212
New York, 10022
Phone: (212) 644-0600
Website: <http://www.ash-us.org>

American Syringomyelia Project
PO Box 1586
Longview, TX 75606-1586
Phone: (903) 236-7079

American Thyroid Association
Townhouse Office Park, 55 Old Nyack Turnpike, Suite 611
Nanuet, NY 10954
Website: <http://www.thyroid.org>

Anemia Institute for Research and Education
151 Bloor Street West, Suite 600
Toronto, ON M5S 1S4
Canada
Phone: (877) 99-ANEMIA
Website: <http://www.anemiainstitute.net>

Angelman Syndrome Foundation
414 Plaza Dr., Suite 209
Westmont, IL 60559-1265
Phone: (630) 734-9267 or (800) 432-6435
Fax: (630) 655-0391
E-mail: info@angelman.org
Website: <http://www.angelman.org>

Anxiety Disorders Association of America

11900 Parklawn Dr., Suite 100
Rockville, MD 20852
Phone: (301) 231-9350
Fax: (301) 231-7392
E-mail: anxdis@adaa.org

Apert Syndrome Support Group

8708 Kathy
Street Louis, MO 63126
Phone: (314) 965-3356

Aplastic Anemia Foundation

PO Box 613
Annapolis, MD 21404-0613
Phone: (800) 747-2820
Website: <http://www.aplastic.org>

Arc (A National Organization on Mental Retardation)

1010 Wayne Ave., Suite 650
Silver Spring, MD 20910
Phone: (800) 433-5255
Website: <http://www.tharclink.org>

Arc of the United States (formerly Association for Retarded Citizens of the US)

500 East Border Street, Suite 300
Arlington, TX 76010
Phone: (817) 261-6003
Website: <http://thearc.org>

Arc's Fetal Alcohol Syndrome Resource Guide

The Arc's Publication Desk
3300 Pleasant Valley Lane, Suite C,
Arlington, TX 76015
Phone: (888) 368-8009
Website: <http://www.thearc.org/misc/faslist.html>

Arthritis Foundation

1330 West Peachtree Street
Atlanta, GA 30309
Phone: (800) 283-7800
Website: <http://www.arthritis.org>

Arthrogryposis Group (TAG)

1 The Oaks, Gillingham
Dorset, SP8 4SW
United Kingdom
Phone: 01-747-822655
Website: <http://tagonline.uk>

Association for Glycogen Storage Disease (United Kingdom)

Phone: 0131 554 2791
Fax: 0131 244 8926
Website: <http://www.agsd.uk>

Association for Frontotemporal Dementias (AFTD)

P.O. Box 7191
St. David's, PA 19087-7191
Email: info@ftd-picks.org
Website: <http://www.ftd-picks.org>

Association for Macular Diseases, Inc.

210 East 64th Street
New York, NY 10021
Phone: (212) 605-3719
E-mail: 2020@nei.nih.gov
Website: <http://www.macula@macula.org>

Association for Neuro-Metabolic Disorders

5223 Brookfield Lane
Sylvania, OH 43560-1809
Phone: (419) 885-1497

Association for Science in Autism Treatment

175 Great Neck Road, Suite 406
Great Neck, NY 11021
Phone: (516) 466-4400
Fax: (516) 466-4484
Email: asat@autism-treatment

Association for Spina Bifida and Hydrocephalus

42 Park Road
Peterborough, PE1 2UQ
United Kingdom
Phone: 0173 355 5988
Fax: 017 3355 5985
E-mail: postmaster@asbah
Website: <http://www.asbah.demon.co.uk>

Association for the Bladder Exstrophy Community

PO Box 1472
Wake Forest, NC 27588-1472
Phone: (919) 624-9447
Website: <http://www.bladderexstrophy.com/support.htm>

Association of Birth Defects in Children

930 Woodcock Road, Suite 225
Orlando, FL 32803
Phone: (407) 895-0802
Website: <http://www.biethdefects.org>

Association Strumpell-Lorain

7 D rue des Granges, Besancon, Intl
25000, France
Phone: (038) 150 2391
Website: <http://perso.wanadoo.fr/asl.spastic/anglasl/englindx.htm>

Asthma and Allergy Foundation of America (AAFA)

1233 20th Street NW, Suite 402
Washington, DC 20036
Phone: (800) 7-ASTHMA
Fax: (202) 466-8940
Website: <http://www.aafa.org>

The A-T Project

3002 Enfield Road
Austin, TX 78703
Phone: (512) 472-4892
Website: <http://www.atproject.org>

Ataxia MJD Research Project, Inc.

875 Mahler Road, Suite 161
Burlingame, CA 94010-1621
Phone: (650) 259-3984
Fax: (650) 259-3983
Website: <http://www.ataxiamjd.org>

Autism Research Institute

4182 Adams Ave.
San Diego, 92116
Fax: (619) 563-6840

Autism Society of America

7910 Woodmont Ave. Suite 300
Bethesda, MD 20814-3015
Phone: (301) 657-0881 or (800) 3-AUTISM
Website: <http://www.autism-society.org>

AVENUES National Support Group for Arthrogryposis Multiplex Congenita

PO Box 5192
Sonora, CA 95370
Phone: (209) 928-3688
Email: avenues@sonnet.com
Website: <http://www.sonnet.com/avenues>

B**Bachmann-Strauss Dystonia & Parkinson Foundation, Inc.**

Mount Sinai Medical Center, One Gustave L. Levy Place, Box 1490
New York, NY 10029
Phone: (212) 241-5614
Website: <http://www.dystonia-parkinsons.org>

Battens Disease Support and Research Association

2600 Parsons Ave.
Columbus, OH 43207
Phone: (800) 448-4570
Website: <http://www.bdsra.org>

BCCNS Life Support Network

PO Box 321
Burton, OH 44021
Phone: (866) 834-1895 (Toll Free),
(440) 635-0078
Fax: (440) 635-0078
Email: info@bccns.org
Website: <http://www.bccns.org>

Beckwith-Wiedemann Support Network

2711 Colony Road
Ann Arbor, MI 48104
Phone: (734) 973-0263 or (800) 837-2976
Website: <http://www.beckwith-wiedemann.org>

Blind Children's Fund

4740 Okemos Road
Okemos, MI 48864-1637
Phone: (517) 347-1357
Website: <http://www.blindchildrensfund.org>

Boys Town National Research Hospital

555 N. 30th Street
Omaha, NE 68131
Phone: (402) 498-6749
Website: <http://www.boystown.org/Btnrh/Index.htm>

**Canadian Hemophilia Society**

625 President Kennedy, Suite 1210
Montreal, QUE H3A 1K2
Canada
Phone: (514) 848-0503
Fax: (514) 848-9661
E-mail: chs@hemophilia.ca
Website: <http://www.hemophilia.ca/english/index.html>

Canadian Multiple Endocrine Neoplasia Type 1 Society, Inc. (CMEN)

PO Box 100
Meota, SK S0M 1X0
Canada
Phone: (306) 892-2080

The Canadian National Institute for the Blind

National Office, 1929 Bayview Avenue
Toronto, ON M4G 3E8
Phone: (416) 486-2500
Fax: (416) 480-7677
Website: <http://cnib.ca>

Canadian Opitz Family Network

Box 892
Errington, BC V0R 1V0
Canada
Phone: (250) 954-1434
Fax: (250) 954-1465
E-mail: opitz@apollos.net
Website: <http://www.apollos.net/arena/opitz/start.html>

Canadian Society for Mucopolysaccharide and Related Diseases

PO Box 64714
Unionville, ON L3R-OM9
Canada
Phone: (905) 479-8701 or (800) 667-1846
Website: <http://www.mpsociety.ca>

Canavan Foundation

320 Central Park West, Suite 19D
New York, NY 10025
Phone: (212) 877-3945

Canavan Research Foundation

Fairwood Professional Building
New Fairwood, CT 06812
Phone: (203) 746-2436
E-mail: canavan_research@hotmail.com
Website: <http://www.canavan.org>

Cardiac Arrhythmias Research and Education Foundation, Inc.

2082 Michelson Dr., #301
Irvine, CA 92612-1212
Phone: (949) 752-2273 or (800) 404-9500
Email: care@longqt.org
Website: <http://www.longqt.org>

Cardio-Facio-Cutaneous Syndrome Foundation

3962 Van Dyke Street
White Bear Lake, MN 55110
Website: <http://www.cfcfoundation.com>

CardioFacioCutaneous Support Network

157 Alder Ave.
McKee City, NJ 08232
Phone: (609) 646-5606

Cardiofaciocutaneous Syndrome Family Network

183 Brown Road
Vestal, NY 13850
Phone: (607) 772-9666
Website: <http://www.cfcysndrome.org>

Center for Loss in Multiple Birth, Inc. (CLIMB)

PO Box 1064
Palmer, AK 99654
Phone: (907) 222-5321

Center for Neurologic Study

9850 Genesee Ave., Suite 320
Lajolla, CA 92037
Phone: (858) 455-5463
Fax: (858) 455-1713
E-mail: cns@cts.com
Website: <http://www.cnsonline.org>

Center for Study of Multiple Birth

334 E. Superior Street, Suite 464
Chicago, IL 60611
Phone: (312) 266-9093
Website: <http://www.multiplebirth.com>

Centers for Disease Control

GDP Office, 4770 Buford Highway NE
Atlanta, GA 30341-3724
Phone: (770) 488-3235
Website: <http://www.cdc.gov/genetics>

Changing Faces

1 & 2 Junction Mews
London, W2 1PN
United Kingdom
Phone: 0207 706 4232
E-mail: info@changingfaces.co.uk
Website: <http://www.changingfaces.co.uk>

Charcot Marie Tooth Association (CMTA)

2700 Chestnut Parkway
Chester, PA 19013
Phone: (610) 499-9264 or (800) 606-CMTA
Fax: (610) 499-9267
E-mail: cmtassoc@aol.com
Website: <http://www.charcot-marie-tooth.org>

CHARGE Family Support Group

82 Gwendolen Ave.
London, E13 ORD
United Kingdom
Phone: 020-8552-6961
Website: <http://www.widerworld.co.uk/charge>

INDEX

Individual volume references are listed before colons; numbers following a colon refer to specific page numbers within that particular volume. **Boldface** page numbers indicate main topical essays. *Italicized* page numbers indicate an illustration or a photographic image. A lowercase *t* following a page number indicates a table on that page.

3-methylcrotonylglycemia, 2: 946,
2: 947, 2: 948
4H syndrome. *See* Pallister-Hall
syndrome
4p deletion syndrome.
See Wolf-Hirschhorn syndrome
4p- syndrome. *See* Wolf-Hirschhorn
syndrome
5,10-methylenetetrahydrofolate
reductase (MTHFR), 2: 1212–1213
5-alpha-reductase deficiency, 1: 540
5-OH-indole-3-acetic acid, 1: 297
5p deletion syndrome. *See* Cri du
chat syndrome
7-dehydrocholesterol (7-DHC), 2: 1203
11q deletion syndrome. *See* Jacobsen
syndrome
17-hydroxyprogesterone, 1: 279, 1: 280,
1: 281
21-hydroxylase deficiency (CAH21),
1: 279–280
21-hydroxylation enzyme defect, 1: 540
22q11 deletion syndrome. *See* Deletion
22q11 syndrome
22q13 deletion syndrome, 1: **1–3**
47,XXY syndrome. *See* Klinefelter
syndrome
1278T gene, 1: 642, 1: 644

A

AACAP (American Academy of Child
and Adolescent Psychiatry), 1: 132
AAMI (Age-associated memory
impairment), 1: 338
Aarskog, Dagfinn, 1: 3
Aarskog syndrome, 1: 3, 1: **3–5**, 1: 4
Aase syndrome, 1: **5–7**
ABCA1 gene, 2: 1239
ABCA1 protein, 2: 1239
ABCA4 gene, 2: 1229
ABCC6 gene, 2: 1080, 2: 1081
ABCC8 gene, 1: 351
Abciximab, 2: 1264
ABCR gene, 1: 277, 2: 1229
Abdominal desmoid tumors, 1: 444,
1: 445, 1: 446
Abdominal mass, with pyloric stenosis,
2: 1083
Abdominal organs, 1: 118

Abdominal pain
familial Mediterranean fever,
1: 450
pancreatitis, 1: 618–619
paroxysmal nocturnal
hemoglobinuria, 2: 992
Abdominal ultrasound
ambiguous genitalia, 1: 541
Beckwith-Wiedemann syndrome,
1: 154
Peutz-Jeghers syndrome, 2: 1013
Von Hippel-Lindau syndrome,
2: 1320
Abdominal wall abnormalities
amyoplasia, 1: 82
Beckwith-Wiedemann syndrome,
1: 152, 1: 154
prenatal ultrasound, 2: 1057
Shprintzen-Goldberg
craniosynostosis syndrome,
2: 1180
triploidy fetus, 2: 1277
See also Omphalocele
Abdominal x rays, conjoined twin
embryos, 1: 291
Abducens nerve absence, 1: 375
Abduction
distal arthrogyriposis syndrome,
1: 362
Duane retraction syndrome, 1: 375,
1: 376
A-beta amyloid neuritic plaque
formation, 1: 70, 1: 71
A-beta amyloid protein, 1: 68
Abetalipoproteinemia (ABL), 1: **7–9**
Ablation, liver cancer, 1: 599
Abnormal, *vs.* normal, 1: 535
Abscesses, with severe combined
immunodeficiency, 2: 1174
Absence seizures, 1: 39, 1: 422
Absent corpus callosum. *See* Agenesis
of corpus callosum
Absorptiometry, dual energy x-ray,
2: 964
Abstract thinking difficulties, 1: 337
Acamprosate, 1: 52
Acanthocytes, 1: 7, 1: 8
Acanthocytosis. *See*
Abetalipoproteinemia
Acanthosis nigricans
Beare-Stevenson cutis gyrata
syndrome, 1: 151

Crouzon syndrome, 1: 312, 1: 313,
1: 314
fibroblast growth factor receptor 3
gene mutation, 1: 313
polycystic ovary syndrome, 2: 1036
Acarbose, for type II diabetes, 1: 356
Acardia, 1: **9–12**, 1: 10
Acardius acephalus, 1: 10
Acardius acornus, 1: 10
Acardius amorphus, 1: 10
Acardius anceps, 1: 10
ACC. *See* Agenesis of corpus callosum
ACC (Aplasia cutis congenita),
1: 32
ACC with chorioretinal abnormality.
See Aicardi syndrome
Accelerometers, 1: 429
Accutane, 1: 12–15
Accutane embryopathy, 1: **12–16**,
1: 238
Accutane Pregnancy Prevention
Program (PPP), 1: 14
ACE inhibitors. *See* Angiotensin-
converting enzyme inhibitors
Acentric chromosomes, 1: 257
Acephaly. *See* Anencephaly
Acetaminophen
gastroschisis and, 1: 503
for Marfan syndrome, 2: 786
for osteoarthritis, 2: 958
Acetazolamide, for hydrocephalus,
1: 651
Acetylcholine, Alzheimer disease and,
1: 70
Acetylcholinesterase (ACHE),
2: 935
Acetylcholinesterase inhibitors, for
frontotemporal dementia, 1: 487
Acetyl-CoA-alpha-glucosaminide
acetyltransferase deficiency, 2: 831
Acetylcysteine, for cystic fibrosis,
1: 320
Achilles tendon contractures, 1: 413
Achondrogenesis, 1: **16–19**, 1: 17,
1: 390
Achondrogenesis type 1, 1: 16, 1: 18,
2: 1231
Achondrogenesis type 1A, 1: 16, 1: 17
Achondrogenesis type 1B, 1: 16, 1: 17,
1: 358
Achondrogenesis type 2, 1: 16,
1: 17–18, 1: 658–659, 1: 660

- Achoondroplasia, 1: **19–23**, 1: 21, 1: 22, 1: 390, 1: 392
 autosomal dominant inheritance, 1: 20, 1: 685–686
 fibroblast growth factor receptor related, 1: 467–468, 1: 468*t*, 2: 1197, 2: 1261
 vs. pseudoachondroplasia, 2: 1077
- ACHOO syndrome, 1: **23–25**, 1: 24
- Achromatopsia, 1: 274–275
- Acid lipase, 2: 1353
- Acid maltase, 2: 1041–1042
- Acid maltase deficiency. *See* Pompe disease
- Acidemias, organic, 2: 946–949
- Acne treatment, 1: 12
- ACOG. *See* American College of Obstetricians and Gynecologists
- ACP. *See* Familial adenomatous polyposis
- ACPS (Acrocephalopolysyndactyly disorders), 1: 215
- ACPS II. *See* Carpenter syndrome
- ACPS IV. *See* Carpenter syndrome
- Acquired angioedema, 1: 600, 1: 601
- Acquired immunity, 2: 772–773
- Acquired immunodeficiency syndrome. *See* AIDS
- Acquired long QT syndrome, 1: 758
- Acquired mutations. *See* Somatic mutations
- Acquired myopia, 2: 873
- Acquired nephrogenic diabetes insipidus, 2: 892, 2: 893, 2: 894
- Acquired ochronosis, 1: 58
- Acquired sideroblastic anemia, 1: 91, 1: 92
- Acral dysostosis with facial and genital abnormalities, 2: **1132–1134**, 2: 1133
- Acrania. *See* Anencephaly
- Acrocallosal syndrome, 1: **25–27**, 1: 26
- Acrocephalopolysyndactyly disorders (ACPS), 1: 215
See also specific acrocephalopolysyndactyly disorders
- Acrocephalopolysyndactyly type I. *See* Pfeiffer syndrome
- Acrocephalopolysyndactyly type II. *See* Carpenter syndrome
- Acrocephalosyndactyly type III. *See* Saethre-Chotzen syndrome
- Acrocephalysyndactyly. *See* Apert syndrome
- Acromatopsia. *See* Color blindness
- Acromegaly, 1: **28–31**, 1: 29, 1: 30, 2: 797–798, 2: 963
- Acromelic dysplasia, 2: 1197
- Acromesomelic brachymelia, 2: 1133
- Acromesomelic dysplasia, 2: 1197
- Acroparesthesias, 1: 433, 1: 434
- Acroscyphodysplasia, metaphyseal, 2: 811
- Acrocephalosyndactyly type I. *See* Apert syndrome
- ACS. *See* American Cancer Society
- ACTH (Adrenocorticotrophic hormone), for lissencephaly, 1: 756
- ACTH stimulation test (Adrenocorticotrophic hormone stimulation test), 1: 280
- Activated charcoal, for congenital erythropoietic porphyria, 2: 1050
- Activated protein C, 1: 438, 1: 439
- Activin receptor-like kinase 1 (ALK1) gene, 2: 954
- Actonel, for osteoporosis, 2: 965
- Acute chest syndrome, 2: 1184, 2: 1186
- Acute encephalopathy, 1: 192
- Acute hyperammonemia, 2: 951
- Acute intermittent porphyrias (AIP), 2: 1045, 2: 1048, 2: 1049–1051
- Acute lymphocytic leukemia (ALL), 1: 746
- Acute myelogenous leukemia (AML), 1: 458, 1: 746, 2: 993
- Acute porphyrias, 2: 1046–1051
- ACVR2B gene, 1: 728, 1: 730
- AD1 gene defect, 1: 68
- AD3 gene defect, 1: 68
- AD4 gene defect, 1: 68
- ADA (Adenosine deaminase) gene, 2: 1173, 2: 1174, 2: 1197
- Adalimumab, for rheumatoid arthritis, 2: 1120
- Adams-Oliver syndrome, 1: **31–34**
- Adapin, for depression, 1: 346
- Adaptive functioning, fetal alcohol syndrome, 1: 463
- ADD (Attention deficit disorder), 2: 794, 2: 1267
- Adderall, for attention deficit hyperactivity disorder, 1: 132
- Addison disease. *See* Adrenoleukodystrophy
- Adducted thumb-clubfoot syndrome, 1: 109, 1: 110
- Adduction, 1: 375, 1: 376
- Adenocarcinomas, 1: 200, 2: 981
- Adenomatous polyposis coli (ACP). *See* Familial adenomatous polyposis
- Adenomatous polyposis, familial. *See* Familial adenomatous polyposis
- Adenomatous polyposis of the colon (APC). *See* Familial adenomatous polyposis
- Adenomatous polyps, 1: 442, 1: 443, 1: 444, 1: 603
- Adenosine deaminase deficiency, 1: 515, 2: 810, 2: 811
- Adenosine deaminase (ADA) gene, 2: 1173, 2: 1174, 2: 1197
- Adenosine triphosphate (ATP) enzyme deficiencies and, 2: 1275
- pyruvate carboxylase deficiency and, 2: 1085
- pyruvate dehydrogenase complex deficiency and, 2: 1087
- Adenosylcobalamin, 2: 814–815
- Adenoviruses, gene therapy and, 1: 514, 1: 516, 1: 517
- ADH (Antidiuretic hormone), 1: 349, 2: 891, 2: 892
- ADHD. *See* Attention deficit hyperactivity disorder
- Adjuvant therapy, cancer, 1: 204–205
- ADOI (Autosomal dominant osteopetrosis I), 2: 1160
- ADOII (Autosomal dominant osteopetrosis II), 2: 1160
- Adolescents, bipolar disorder, 1: 167–168
- Adopted children
 alcoholism, 1: 48
 schizophrenia, 2: 1148
- ADP (ALA dehydratase deficiency porphyria), 2: 1045, 2: 1048
- ADPKD (Autosomal dominant polycystic kidney disease), 1: 452–456
- Adrenal crisis, 1: 279, 1: 280
- Adrenal gland tumors, 2: 939, 2: 1317, 2: 1318
- Adrenal glands, 1: 34, 2: 798
- Adrenal insufficiency, 1: 35, 2: 976
- Adrenalectomy, 1: 281
- Adrenergic antagonists, for essential hypertension, 1: 427
- Adrenocortical carcinoma, 1: 746, 1: 746*t*, 1: 748
- Adrenocorticotrophic hormone (ACTH), for lissencephaly, 1: 756
- Adrenocorticotrophic hormone stimulation test (ACTH stimulation test), 1: 280
- Adrenoleukodystrophy (ALD), 1: **34–38**, 2: 1008
- Adrenomyeloneuropathy (AMN), 1: 35, 1: 37
- Adriamycin, birth defects and, 2: 1313
- Adult alpha-mannosidosis, 2: 781
- Adult celiac disease, 1: **221–224**, 2: 774*t*
- Adult genetic counseling, 1: 520–521
- Adult glaucoma, 1: 544, 1: 544*t*, 1: 545, 1: 546–547
- Adult GM1-gangliosidosis, 1: 556–557
- Adult hemoglobin (HbA). *See* Hemoglobin A
- Adult hypophosphatasia, 1: 666, 1: 667
- Adult Pompe disease, 2: 1042, 2: 1043
- Adult Refsum disease, 1: 681–682
See also Refsum disease
- Adult-onset polycystic kidney disease, 2: 1032–1035
- Adult-onset subacute necrotizing encephalomyelopathy, 1: 737, 1: 738
- Advil
 gastroschisis and, 1: 503
 with Osler-Weber-Rendu syndrome, 2: 956
 for rheumatoid arthritis, 2: 1119
 telogen effluvium from, 1: 571
- Adysplasia syndrome, urogenital, 2: 1302–1304

- AEC syndrome. *See* Hay-Wells syndrome
- AFAP. *See* Attenuated familial adenomatous polyposis
- Aflatoxin
liver cancer and, 1: 597
pancreatic cancer and, 2: 981
- AFP. *See* Alpha-fetoprotein
- African Americans, 2: 1068, 2: 1069, 2: 1070
See also specific disorders
- A-gammaglobulinemia tyrosine kinase, 1: **186–189**
- Aganglionic megacolon.
See Hirschsprung disease
- Age
acromegaly, 1: 31
adrenoleukodystrophy, 1: 35
alcoholism, 1: 49
Azorean disease, 1: 140
Becker muscular dystrophy, 1: 381, 1: 383
bipolar disorder, 1: 167
color blindness, 1: 275
dentatorubral-pallidolusian atrophy, 1: 342
depression, 1: 345
developmental milestones, 1: 229
Duchenne muscular dystrophy, 1: 381, 1: 383
dystonia, 1: 394
Fahr disease, 1: 441
hereditary colorectal cancer screening, 1: 606
myopia, 2: 874
panic disorder, 2: 985
polycystic kidney disease, 1: 452
prostate cancer, 2: 1068
schizophrenia, 2: 1151
trisomy 18, 2: 1289
See also Maternal age
- Age-associated memory impairment (AMMI), 1: 338
- Agenesis of clavicles and cervical vertebral and talipes equinovarus, 1: 306–307
- Agenesis of corpus callosum (ACC), 1: **299–303**, 1: 300, 1: 329, 2: 1360
- Agenesis of corpus callosum with chorioretinal abnormality. *See* Aicardi syndrome
- Agenesis of fetal kidneys, 1: 536
- Age-related macular degeneration (AMD), 2: **767–772**, 2: 770, 2: 1230
- Aging, premature, 2: 1337–1338
- Agnosia, 1: 337
- Agnosognosia, 1: 68
- Agonists, centrally acting, 1: 427
- Agoraphobia, 2: 985, 2: 986, 2: 987–988
- AHD (Arteriohepatic dysplasia), 1: **40–43**
- Aicardi, Jean, 1: 38
- Aicardi syndrome, 1: 38, 1: **38–40**, 1: 559
- AICDs (Automatic implanted cardioverter defibrillators), 1: 697, 1: 762
- AIDS (Acquired immunodeficiency syndrome)
dementia, 1: 339
gene therapy, 1: 516
schizophrenia, 2: 1151
- AIDS virus. *See* Human immunodeficiency virus (HIV)
- AIP. *See* Acute intermittent porphyrias
- Air pressure changes, Marfan syndrome and, 2: 786
- Airway constriction, 1: 602, 2: 832
- AIS (Androgen insensitivity syndrome), 1: **87–90**, 1: 89r
- Akathisia, 2: 1151
- Akinesia, fetal, 1: 81
- AKU (Alkaptonuria), 1: **54–60**, 1: 58
- ALA dehydratase deficiency porphyria (ADP), 2: 1045, 2: 1048
- ALAD (Delta-aminolevulinatase dehydratase) gene, 2: 1048
- Alagille syndrome, 1: **40–43**
- Alagille-Watson syndrome, 1: **40–43**
- Alanine aminotransferase, 1: 62
- ALAS2 (Delta-aminolevulinatase synthase), 1: 91
- Albinism, 1: 43, 1: **43–46**, 1: 45
Griscelli syndrome and, 1: 563
vs. Waardenburg syndrome, 2: 1328
See also Hermansky-Pudlak syndrome
- Albuterol, for facioscapulohumeral muscular dystrophy, 1: 437
- Alcaptonuria. *See* Alkaptonuria
- Alcohol
cancer and, 1: 201
cleft lip and palate with maternal use, 1: 257
congenital heart disease with maternal use, 1: 285
essential tremor management, 1: 430
holoprosencephaly with maternal use, 1: 636
osteoporosis and, 2: 965
panic attacks from, 2: 986
as teratogen, 1: 462, 2: 1247
- Alcohol concentration, blood, 1: 49
- Alcohol injections, cerebral palsy, 1: 230
- Alcohol Use Disorder Identification Test (AUDIT), 1: 50, 1: 51
- Alcoholism, 1: **47–54**, 1: 50, 1: 51
amelia, 1: 73
color blindness, 1: 275
dementia, 1: 67, 1: 336, 1: 337, 1: 339, 1: 340
depression, 1: 49, 1: 344
fetal alcohol syndrome, 1: 461–464, 1: 717, 2: 1248
hyperlipoproteinemia testing, 1: 657
Lebers hereditary optic atrophy, 1: 735
teratogen, 2: 1247
- Alcohol-sensitizing medications, 1: 52
- ALD. *See* Adrenoleukodystrophy
- ALD deficiency (Argininosuccinase acid lyase deficiency), 2: 1301
- ALD gene, 1: 34–35
- Aldactone, for polycystic ovary syndrome, 2: 1038
- Aldehyde dehydrogenase deficiency, 1: 48
- Aldosterone, congenital adrenal hyperplasia and, 1: 278–279, 1: 281
- Aldrich, R. A., 2: 1346
- Aldrich syndrome. *See* Wiskott-Aldrich syndrome
- Alendronate, for osteoporosis, 2: 965
- Aleve
with Osler-Weber-Rendu syndrome, 2: 956
for rheumatoid arthritis, 2: 1119
- Alexander's disease, 1: 743, 1: 744–745
- ALK1 (Activin receptor-like kinase 1) gene, 2: 954
- Alkaline phosphatase (ALP), 1: 51, 1: 664
- Alkaptonuria (AKU), 1: **54–60**, 1: 58
- Alkyl dihydroxyacetone phosphate synthase, 2: 1123
- ALL (Acute lymphocytic leukemia), 1: 746
- Alleles, 1: 256, 1: 381
Chediak-Higashi syndrome, 1: 239–240
diabetes mellitus, 1: 349, 1: 350
gene pool, 1: 511
gene structure, 1: 508
genetic disorder inheritance, 1: 524
genotype, 1: 542
inheritance, 1: 685
- Allen Brown criteria, 2: 1094
- Allergen immunotherapy, 1: 125
- Allergens, 1: 120, 1: 123
- Allergies
ACHOO syndrome, 1: 24–25
asthma, 1: 123, 1: 125
vs. hereditary angioneurotic edema, 1: 602
infections from, 2: 1174
- Allopurinol, for Lesch-Nyhan syndrome, 1: 742
- Alobar holoprosencephaly, 1: 635, 1: 636, 1: 637, 1: 638
- Alopecia. *See specific disorders*
- Alopecia areata, 1: 222–223, 1: 571–574
- ALP (Alkaline phosphatase), 1: 51, 1: 664
- Alpha agonists, for glaucoma, 1: 547
- Alpha globin genes, 2: 1250, 2: 1251, 2: 1253
- Alpha spectrin gene, 1: 626
- Alpha thalassemia, 1: 64, 1: 65, 1: 654, 2: 1249–1252
- Alpha thalassemia major, 2: 1249, 2: 1250, 2: 1252, 2: 1253, 2: 1255

- Alpha thalassemia mental retardation syndrome, 2: 1251–1252
- Alpha thalassemia trait, 2: 1249
- Alpha-1 antitrypsin, 1: **60–64**, 1: 596
- Alpha-1 antitrypsin protein augmentation therapy, 1: 63
- Alpha/beta blockers, for essential hypertension, 1: 427
- Alpha-blockers, 1: 427
- Alpha-fetoprotein (AFP), 1: 75
- amelia, 1: 74
 - amniocentesis, 1: 77
 - anencephaly, 1: 94
 - ataxia-telangiectasia, 1: 128
 - gastroschisis, 1: 503
 - genetic counseling, 1: 520
 - hepatoblastoma screening, 1: 154
 - Meckel-Gruber syndrome, 2: 802
 - spina bifida, 2: 1213–1214
- See also* Maternal serum alpha-fetoprotein test
- Alpha-galactosidase A, Fabry disease and, 1: 433, 1: 434, 1: 435
- Alphaglucoosidase inhibitors, for type II diabetes, 1: 356
- Alpha-L-iduronidase, 2: 830, 2: 833, 2: 834, 2: 835
- Alpha-mannosidosis, 2: 780, 2: 781
- Alpha-N-acetylneuraminidase, 2: 901, 2: 905
- Alpha-neuraminidase deficiency. *See* Neuraminidase deficiency
- Alpha-thalassemia X-linked mental retardation syndrome, 1: **64–67**, 2: 1201
- ALPL gene, 2: 1197
- Alport syndrome, 1: 452–456, 1: 609, 1: 610, 1: 612
- ALS. *See* Amyotrophic lateral sclerosis
- Alsln gene, 1: 622
- Alternative treatments
- arthrogryposis multiplex congenita, 1: 111
 - cancer, 1: 205
 - cystic fibrosis, 1: 321
 - dystonia, 1: 394
 - Ehlers-Danlos syndrome, 1: 407
 - multiple sclerosis, 2: 858
 - myopia, 2: 877
 - osteogenesis imperfecta, 2: 962
 - Parkinson disease, 2: 991
 - porphyrias, 2: 1050–1051
 - sickle cell disease, 2: 1186
- Altmann, Richard, 1: 364
- Aluminum-containing antacids, osteoporosis from, 2: 963
- Alzheimer, Alois, 1: 67, 1: 484
- Alzheimer disease, 1: **67–72**, 1: 69, 1: 70
- color blindness from, 1: 275
 - as dementia, 1: 336, 1: 337
 - diagnosis, 1: 69–70, 1: 339
 - with Down syndrome, 1: 67–68, 1: 336, 1: 371
 - vs. frontotemporal dementia, 1: 486, 1: 487
 - presymptomatic testing, 1: 530
- Amantadine
- for Friedreich ataxia, 1: 481
 - for Parkinson disease, 2: 990
- Ambiguous genitalia, 1: **539–542**
- AMC. *See* Arthrogryposis multiplex congenita
- AMD. *See* Age-related macular degeneration
- Amelia, 1: **72–75**
- Amelogenesis imperfecta, 1: 277
- American Academy of Child and Adolescent Psychiatry (AACAP), 1: 132
- American Academy of Neurology, 1: 487
- American Academy of Pediatrics, 2: 1021
- American Cancer Society (ACS)
- cancer screening, 1: 203
 - cancer signs, 1: 202
 - gastric cancer, 1: 498, 1: 500
 - liver cancer, 1: 596
 - Pap smears, 1: 389
 - prostate cancer, 2: 1070, 2: 1072
- American College of Medical Genetics, 1: 197
- American College of Obstetricians and Gynecologists (ACOG)
- amniocentesis, 1: 76
 - Canavan disease, 1: 197
 - prenatal ultrasound, 2: 1056–1057
- American Diabetes Association, 1: 351–352
- American Heart Association, 1: 286
- American Institute of Ultrasound Medicine Bioeffects Committee, 2: 1056
- American Journal of Medical Genetics*, 2: 1209
- American Optical/Hardy, Rand, and Ritter Pseudoisochromatic test, 1: 275
- Americans with Disabilities Act of 1990, 1: 133
- Amiloride, for nephrogenic diabetes insipidus, 2: 893
- Amino acids
- dibasic, 1: 325, 1: 326
 - metabolism disorders, 2: 946–949, 2: 1019–1024
 - methylmalonic CoA mutase deficiency and, 2: 816
 - for Parkinson disease, 2: 991
- Aminocaproic acid therapy, for nosebleeds, 2: 955–956
- Aminoglycoside antibiotics, 1: 321
- Aminoglycosides, hearing loss from, 1: 612
- Aminopterin syndrome sine aminopterin, 1: 306
- Amitriptyline
- for depression, 1: 346
 - for rheumatoid arthritis, 2: 1121
- AML (Acute myelogenous leukemia), 1: 458, 1: 746, 2: 993
- AMN (Adrenomyeloneuropathy), 1: 35, 1: 37
- Amniocentesis, 1: **75–81**, 1: 79, 1: 531
- achondroplasia, 1: 21
 - adrenoleukodystrophy, 1: 37
 - aftercare, 1: 532
 - alpha-thalassemia X-linked mental retardation syndrome, 1: 66
 - Apert syndrome, 1: 102
 - Beals syndrome, 1: 148
 - biotinidase deficiency, 1: 165
 - vs. chorionic villus sampling, 1: 78
 - chromosomal abnormalities, 1: 254
 - clubfoot, 1: 262
 - congenital adrenal hyperplasia, 1: 281
 - cystinosis, 1: 323
 - deletion 22q11 syndrome, 1: 334
 - dentatorubral-pallidolulian atrophy, 1: 342–343
 - Down syndrome, 1: 372–373
 - ectrodactyly-ectodermal dysplasia-clefting syndrome, 1: 403
 - Ehlers-Danlos syndrome kyphoscoliosis type, 1: 406
 - encephalocoeles, 1: 417
 - facioscapulohumeral muscular dystrophy, 1: 437
 - fragile X syndrome, 1: 474
 - genetic counseling, 1: 520
 - GM1-gangliosidosis, 1: 557
 - harlequin fetus, 1: 580
 - hemophilia, 1: 594
 - hypochondrogenesis, 1: 660
 - hypochondroplasia, 1: 662–663
 - hypophosphatasia, 1: 666, 1: 667
 - ichthyosis, 1: 674
 - infantile Refsum disease, 1: 683
 - Klinefelter syndrome, 1: 716
 - Krabbe disease, 1: 724
 - Leigh syndrome, 1: 739–740
 - Menkes syndrome, 2: 807, 2: 808
 - miscarriages with, 1: 76, 1: 78–79, 1: 343, 1: 373, 1: 532, 2: 1185, 2: 1291
 - mucopolysaccharidosis type I, 2: 835
 - myotonic dystrophy, 2: 880
 - neurofibromatosis, 2: 910
 - omphalocele, 2: 935
 - Opitz syndrome, 2: 943
 - osteogenesis imperfecta, 2: 961
 - Pelizaeus-Merzbacher disease, 2: 1001
 - Peutz-Jeghers syndrome, 2: 1012
 - Pfeiffer syndrome, 2: 1016
 - Prader-Willi syndrome, 2: 1054
 - propionic acidemia, 2: 1066
 - pseudoachondroplasia, 2: 1078
 - Refsum disease, 2: 1098
 - rhizomelic chondrodysplasia punctata, 2: 1123
 - Rieger syndrome, 2: 1127

- risks, 1: 532
severe combined immunodeficiency, 2: 1175
sickle cell disease, 2: 1185
Sjögren-Larsson syndrome, 2: 1195
Smith-Lemli-Opitz syndrome, 2: 1203
spina bifida, 2: 1213
spinocerebellar ataxia, 2: 1221
spondyloepiphyseal dysplasia, 2: 1224
thalassemia, 2: 1252
triploidy, 2: 1278
trisomy 18, 2: 1291
Turner syndrome, 2: 1299
tyrosinase-negative oculocutaneous albinism, 1: 45
ultrasound use in, 2: 1059
Williams syndrome, 2: 1341
Wolman disease, 2: 1354
XX male syndrome, 2: 1368–1369
XYY syndrome, 2: 1371
Zellweger syndrome, 2: 1373
See also Prenatal testing
- Amniocytes, 1: 77
Amnioinfusion, 2: 932
Amnion, 1: 73, 1: 75
Amniotic band syndrome, 1: 416
Amniotic bands, 1: 536, 2: 1059, 2: 1170
Amniotic fluid, 1: 75
 Beckwith-Wiedemann syndrome, 1: 152
 genetic testing, 1: 77–78
 leakage and oligohydramnios, 2: 930
 See also Polyhydramnios
Amniotic sac, 1: 75
Amoxicillin
 for Cornelia de Lange syndrome, 1: 298
 for Marfan syndrome, 2: 785
Amphetamine-like stimulant drugs, for narcolepsy, 2: 891
Amphetamines
 for attention deficit hyperactivity disorder, 1: 132
 motor and vocal tics and, 2: 1267
 panic attacks and, 2: 986
Ampicillin, myasthenia gravis and, 2: 872
Amsler grid test, 2: 770
Amsterdam criteria, hereditary nonpolyposis colorectal cancer, 1: 604
Amsterdam Dwarf Syndrome of de Lange. *See* Cornelia de Lange syndrome
Amyloidosis, 1: 293, 1: 294
 familial Mediterranean fever and, 1: 449, 1: 450
 nephrogenic diabetes insipidus from, 2: 892
Amyoplasia, 1: 81–83, 1: 109, 1: 110
Amyotrophic lateral sclerosis (ALS), 1: 83–87, 1: 85, 1: 86, 1: 485, 1: 488, 1: 623
Anabolic steroids, liver cancer and, 1: 597
Anadysplasia, metaphyseal, 2: 810
Anaftranil
 for depression, 1: 346
 for trichotillomania, 1: 573
Anagen effluvium, 1: 571, 1: 573, 1: 574
Anakinra, for rheumatoid arthritis, 2: 1120–1121
Anal atresia, 2: 1313, 2: 1314
Analgesics
 for Gaucher disease, 1: 506
 for long QT syndrome, 1: 762
 for multiple epiphyseal dysplasia, 2: 852
 for pseudoachondroplasia, 2: 1079
 for sickle cell disease, 2: 1186
Andersen cardiodysrhythmic periodic paralysis. *See* Long QT syndrome 7
Andersen syndrome. *See* Long QT syndrome 7
Andersen's disease, 1: 549, 1: 550, 1: 551, 1: 553
Andersen-Tawil syndrome. *See* Long QT syndrome 7
Anderson, W. French, 1: 515
Anderson, William, 1: 433
Andre syndrome, 2: 966, 2: 967
Androgen insensitivity syndrome (AIS), 1: 87–90, 1: 89*t*
Androgen receptor (AR) gene
 androgen insensitivity syndrome, 1: 87–88, 1: 89
 Kennedy disease, 1: 713
Androgen-dependent hair loss. *See* Androgenetic alopecia
Androgenetic alopecia, 1: 569–570, 1: 570, 1: 572, 1: 574
Androgens
 congenital adrenal hyperplasia and, 1: 278
 for Fanconi anemia, 1: 459
 for hereditary angioneurotic edema, 1: 602
 for partial androgen insensitivity syndrome, 1: 90
 in polycystic ovary syndrome, 2: 1036
 See also Testosterone
Androstenedione, 1: 279
Anemia
 abetalipoproteinemia, 1: 8
 alpha thalassemia, 2: 1249, 2: 1253
 alpha-thalassemia X-linked mental retardation syndrome, 1: 64, 1: 65, 1: 66
 aplastic, 1: 457–458, 2: 993, 2: 1183
 beta thalassemia, 1: 157, 2: 1249, 2: 1253
 bilateral renal agenesis, 2: 1102–1103
 congenital dyserythropoietic, 2: 993
 Fanconi, 1: 457–460, 1: 458, 2: 1315
 fetal, 2: 1253
 hemolytic, 1: 626, 2: 1091, 2: 1249, 2: 1253, 2: 1255
 hereditary spherocytosis, 1: 625, 1: 626
 hydrops fetalis from, 1: 653
 Osler-Weber-Rendu syndrome, 2: 954
 paroxysmal nocturnal hemoglobinuria, 2: 992
 renal failure, 2: 1104
 sideroblastic X-linked, 1: 90–93
 treatment, 1: 506, 2: 956, 2: 1253, 2: 1255
 triose phosphate isomerase deficiency, 2: 1274–1275
 See also Congenital hypoplastic anemia; Sickle cell anemia
Anencephaly, 1: 93–95, 1: 94, 1: 415, 2: 898, 2: 899, 2: 899, 2: 900
aminopterin syndrome, 1: 306
Crane-Heise syndrome, 1: 306
multifactorial inheritance, 1: 93, 2: 843
Anesthesia
 with congenital spondyloepiphyseal dysplasia, 2: 1225
 Freeman-Sheldon syndrome and, 1: 479
 with long QT syndrome, 1: 762
 malignant hyperthermia with, 2: 776, 2: 1018
 with mucopolysaccharidoses, 2: 832
 with muscular dystrophy, 2: 866–867
 with myotonic dystrophy, 2: 881
 saddle, 2: 1213
 trismus-pseudocamptodactyly syndrome and, 2: 1281
Aneuploidy, 1: 76, 1: 250, 1: 250–251, 1: 251*t*
 acardia, 1: 10
 first-trimester screening, 2: 1056–1057
 fluorescent in situ hybridization and, 1: 471
 maternal age and, 1: 254
 trisomy 18, 1: 250, 2: 1288
 ultrasound markers for, 2: 1058
Aneurysms
 aortic, 1: 160, 1: 161
 brain, 2: 1033
Angelman syndrome (AS), 1: 95–98, 1: 97
 chromosome deletions, 1: 95, 1: 97, 1: 252, 2: 1053
 imprinting, 1: 252, 1: 677
Angioedema, acquired, 1: 600, 1: 601
 See also Hereditary angioneurotic edema
Angiofibromas, facial, 2: 1296
Angiokeratoma
 Fabry disease, 1: 433, 1: 434
 galactosialidosis, 2: 907
Angiomas, leptomeningeal, 2: 1234, 2: 1235