

Reference List for ASC Orientation Plan

Topic Area: General Program Knowledge **Core Competencies 1 – 8**

Required Reading

[Larsson A, Therrell BL. \(2002\). Newborn screening: the role of the obstetrician. *Clin Obstet Gynecol*, 45 \(3\), 697-710; discussion 730-732.](#)

Recommended Articles

[Watson MS, Mann MY, Lloyd-Puryear MA, Rinaldo P, Howell RR, American College of Medical Genetics Newborn Screening Expert Group. \(2006\). Newborn Screening: Toward a Uniform Screening Panel and System – Executive Summary. *Pediatrics*, 117 \(5 Pt 2\), S296-307.](#)

[American Academy of Pediatrics Newborn Screening Authoring Committee. \(2008\). Newborn screening expands: recommendations for pediatricians and medical homes – implications for the system. *Pediatrics*, 121 \(1\), 192-217.](#)

Other Articles

[Blood-Siegfried J, Lieder HS, Deary K. \(2006\). To Screen or Not to Screen: Complexities of Newborn Screening in the 21st Century. *The Journal for Nurse Practitioners – JNP*, 2 \(5\), 300-307.](#)

[Botkin JR, Clayton EW, Fost NC, Burke W, Murray TH, Baily MA, Wilfond B, Berg A, Ross LF. \(2006\). Newborn screening technology: proceed with caution. *Pediatrics*, 117 \(5\), 1793-1797.](#)

[Green NS, Dolan SM, Murray TH. \(2006\). Newborn screening: complexities in universal genetic testing. *Am J Public Health*, 96 \(11\), 1955-1959.](#)

[Kaye CI, Committee on Genetics, Accurso F, La Franchi S, Lane PA, Hope N, Sonya P, G Bradley S, Michele ALP. \(2006\). Newborn screening fact sheets. *Pediatrics*, 118 \(3\), e934-964.](#)

[Kaye CI, Committee on Genetics, Accuro F, La Franchi S, Lane PA, Northrup H, Pang S, Schaefer GB. \(2006\). Introduction to the newborn screening fact sheets. *Pediatrics*, 118 \(3\), 1304-1312.](#)

[Opdal SH, Rognum TO. \(2004\). The sudden infant death syndrome gene: does it exist? *Pediatrics*, 114 \(4\), e506-512.](#)

[Spahis JK, Bowers NR. \(2006\). Navigating the maze of newborn screening. *MCN Am J Matern Child Nurs*, 31 \(3\), 190-196.](#)

[Therrell BL, Buechner C, Lloyd-Puryear MA, van Dyck PC, Mann MY. \(2008\). What's new in newborn screening? *Pediatric Health*, 2 \(4\), 411-429.](#)

[Watson MS, Lloyd-Puryear MA, Mann, MY, Rinaldo P, Howell RR. \(2006\). Newborn screening: toward a uniform screening panel and system. *Genet Med*, 8 Suppl 1, 1S-252S.](#)

Websites for Competencies 1 – 8

State of California NBS Web Site

<http://www.cdph.ca.gov/programs/GDSP/Pages/default.aspx>
<http://www.cdph.ca.gov/programs/nbs/Pages/NBSFutureAdditions.aspx>
<http://www.cdph.ca.gov/programs/nbs/Pages/NBSProgrOVforParents.aspx>
<http://www.cdph.ca.gov/programs/nbs/Pages/NBSProgrOVforProviders.aspx>

California Children's Services

<http://www.dhcs.ca.gov/services/ccs>

March of Dimes

<http://www.marchofdimes.com>

Newborn Screening Quality Assurance Program

Centers for Disease Control and Prevention

<http://www.cdc.gov/nceh/dls/newborn.htm>

CDC information about laboratory quality assurance methods for NBS dried blood spot analysis in state labs. Methodology and quarterly reports are available to the public.

Save Babies

<http://www.savebabies.org/>

Patient NBS advocacy foundation for providers and parents. Provides NBS disorder descriptions and state-by-state comparisons of NBS screening panels, and recommendations for disorders to be included on NBS panels.

A New Era In Newborn Screening - Saving Lives, Improving Outcomes

<http://www2a.cdc.gov/phtn/webcast/newborn/default.asp>

Originally aired September 19, 2002 - 2:00-4:00 PM ET (Requires Real Player to View)

Websites specifically for Competency 2

Regulations: Barclays

<http://www.calregs.com>

Websites specifically for Competency 4

Program Mission:

<http://www.cdph.ca.gov/programs/GDSP/Pages/GDSPMission.aspx>

Topic Area: Disorders

Core Competencies 9 – 10

Website specifically for conditions screened

Genetic Home Reference from the NIH

<http://ghr.nlm.nih.gov/>

Provides many brief, surface-level descriptions of various genetic conditions, including most found on NBS (search for each by name). Also gives many general links to other web resources, glossary of terms and genes, as well as a handbook describing general genetics concepts (under the Handbook tab on top).

National Newborn Screening & Genetics Resource Center

<http://genes-r-us.uthscsa.edu/index.htm>

Quick overview of the purpose and procedures of newborn screening. Patient-oriented information, and up-to-date state-to-state comparisons of conditions tested on their NBS panels. Publication listing of recent articles related to NBS for providers. Other NBS resources (brochures, links, fact sheets) provided also.

Phenylketonuria (PKU)

Required Reading

[Peterson RM, Koch R, Schaeffler GE, Wohlers A, Acosta PB, Boyle D. \(1968\). Phenylketonuria. Experience at one center in the first year of screening in California. *Calif Med*, 108 \(5\), 350-354.](#)

Text

Guthrie R, Whitney S.(1964) Phenylketonuria detection in the newborn infant as a routine hospital procedure: a trial of a phenylalanine screening method in 400,000 infants. *Children's Bureau Publication 419. Washington (DC): U.S. Department of Health, Education and Welfare.*

Other Articles

[Guthrie R, Susi A. \(1963\). A simple phenylalanine method for detecting phenylketonuria in large populations of newborn infants. *Pediatrics*, 32, 338-343.](#)

[Lorey FW, Cunningham GC. \(1994\). Effect of specimen collection method on newborn screening for PKU. *Screening*, 3, 57-65.](#)

[Rouse B, Matalon R, Koch R, Azen C, Levy H, Hanley W, Trefz F, de la Cruz F. \(2000\). Maternal phenylketonuria syndrome: congenital heart defects, microcephaly, and developmental outcomes. *J Pediatr*, 136 \(1\), 57-61.](#)

[Velazquez A, Bilbao G, Gonzalez-Trujillo JL, Hernandez D, Perez-Andrade ME, Vela M, Ciceron I, Loera-Luna A, Cederbaum S, Phoenix B. \(1996\). Apparent](#)

[higher frequency of phenylketonuria in the Mexican state of Jalisco. *Hum Genet*, 97 \(1\), 99-102.](#)

Website

National PKU News

<http://www.pkunews.org>

Newsletter and patient-oriented information/resource website for parents of and patients with PKU. Comprehensive overview of aspects of PKU course.

Videos

Parent Interviews with George Cunningham, MD, MPH

Interviews of 4 mothers with children who have PKU. Copies available at GDSP on DVD and ASCs on VHS.

California's First Maternal PKU Camp – For Professionals

Highlights of the 1987 MPKU Camp showing girls in cooking class and peer support groups. Copy available at GDSP on VHS.

PKU and You: Young Women Share Their Thoughts

Copies available at GDSP and ASCs on VHS.

Message to PKU Parents

Copies available at GDSP and ASCs on VHS.

Galactosemia

Required Reading

TBD

Text Chapter

Segal S, Berry G, T. (1995) Disorders of galactose metabolism. In: Scriver, C.R., Beaudet, A.L., Sly, W.S., Valle, D. Editors, *The Metabolic and Molecular Basis of Inherited Disease.*, (seventh ed), McGraw-Hill, New York., 967-1000.

Other Articles

[Elsas LJ, Langley S, Steele E, Evinger J, Fridovich-Keil JL, Brown A, Singh R, Fernhoff P, Hjelm LN, Dembure PP. \(1995\). Galactosemia: a strategy to identify new biochemical phenotypes and molecular genotypes. *Am J Hum Genet*, 56 \(3\), 630-639.](#)

[Kaufman FR, Xu YK, Ng WG, Donnell GN. \(1988\). Correlation of ovarian function with galactose-1-phosphate uridylyl transferase levels in galactosemia. *J Pediatr*, 112 \(5\), 754-756.](#)

[Nelson CD, Waggoner DD, Donnell GN, Tuerck JM, Buist NR. \(1991\). Verbal dyspraxia in treated galactosemia. *Pediatrics*, 88 \(2\), 346-350.](#)

Primary Congenital Hypothyroidism

Required Reading

[American Academy of Pediatrics, Rose SR, Section on Endocrinology and Committee on Genetics, American Thyroid Association, Brown RS, Public Health Committee, Lawson Wilkins Pediatric Endocrine Society, Foley T, Kaplowitz PB, Kaye CI, Sundararajan S, Varma SK. \(2006\). Update of newborn screening and therapy for congenital hypothyroidism. *Pediatrics*, 117 \(6\), 2290-2303.](#)

Other Articles

[Bongers-Shokking JJ, Koot HM, Wiersma D, Verkerk PH, de Muinck Keizer-Schrama SM. \(2000\). Influence of timing and dose of thyroid hormone replacement on development in infants with congenital hypothyroidism. *J Pediatr*, 136 \(3\), 292-297.](#)

[Conrad SC, Chiu H, Silverman BL. \(2004\). Soy formula complicates management of congenital hypothyroidism. *Arch Dis Child*, 89 \(1\), 37-40.](#)

[Fisher DA. \(2000\). The importance of early management in optimizing IQ in infants with congenital hypothyroidism. *J Pediatr*, 136 \(3\), 273-274.](#)

Congenital Adrenal Hyperplasia

Required Reading

TBD

Other Articles

[Allen DB. \(1993\). Newborn screening for congenital adrenal hyperplasia in Wisconsin. *Wis Med J*, 92 \(2\), 75-8.](#)

[Allen DB, Hoffman GL, Fitzpatrick P, Laessig R, Maby S, Slyper A. \(1997\). Improved precision of newborn screening for congenital adrenal hyperplasia using weight-adjusted criteria for 17-hydroxyprogesterone levels. *J Pediatr*, 130 \(1\), 128-133.](#)

[Pang S, Clark A. \(1993\). Congenital adrenal hyperplasia due to 21-hydroxylase deficiency: Newborn screening and its relationship to the diagnosis and treatment of the disorder. *Screening*, 2, 105-139.](#)

[Section on Endocrinology and Committee on Genetics. \(2000\). Technical Report: Congenital Adrenal Hyperplasia. *Pediatrics*, 106 \(6\), 1511-1518.](#)

Website

CARES Foundation-Parent Support group for CAH

<http://www.caresfoundation.org>

Research, education, advocacy, and patient-support website with patient-oriented material on many aspects of CAH manifestations, life-course, and treatment. May also serve as a broad overview of CAH for health providers. Also has up-to-date links to current clinical trials.

Videos

Congenital Adrenal Hyperplasia (CAH)

Five-hour professional staff training in 2005 with Dr. Mitchell Geffner and staff from Children's Hospital of Los Angeles Endocrinology. Copies available at GDSP on VHS and at ASCs on DVD.

Cystic Fibrosis

Required Reading

[Grosse SD, Boyle CA, Botkin JR, Comeau AM, Kharrazi M, Rosenfeld M, Wilfond BS. \(2004\). Newborn Screening for Cystic Fibrosis: Evaluation of Benefits and Risks and Recommendations for State Newborn Screening Programs. *CDC MMWR Recommendations and Reports*, 53 \(RR13\), 1-36.](#)

Recommended Articles

[Ross LF. \(2008\). Newborn screening for cystic fibrosis: a lesson in public health disparities. *J Pediatr*, 153 \(3\), 308-313.](#)

[Castellani C, Cuppens H, Macek M Jr, Cassiman JJ, Kerem E, Durie P, Tullis E, Assael BM, Bombieri C, Brown A, Casals T, Claustres M, Cutting GR, Dequeker E, Dodge J, Doull I, Farrell P, Ferec C, Girodon E, Johannesson M, Kerem B, Knowles M, Munck A, Pignatti PF, Radojkovic D, Rizzotti P, Schwarz M, Stuhmann M, Tzetis M, Zielenski J, Elborn JS. \(2008\) Consensus on the use and interpretation of cystic fibrosis mutation analysis in clinical practice. *J Cyst Fibros*, 7 \(3\), 179-196.](#)

Other Articles

[Ciske DJ, Haavisto A, Laxova A, Rock LZ, Farrell PM. \(2001\). Genetic counseling and neonatal screening for cystic fibrosis: an assessment of the communication process. *Pediatrics*, 107 \(4\), 699-705.](#)

[Farrell PM, Rosenstein BJ, White TB, Accurso FJ, Castellani C, Cutting GR, Durie PR, Legrys VA, Massie J, Parad RB, Rock MJ, Campbell PW 3rd; Cystic Fibrosis Foundation. \(2008\). Guidelines for diagnosis of cystic fibrosis in newborns through older adults: Cystic Fibrosis Foundation consensus report. *J Pediatr*, 153 \(2\), S4-S14.](#)

[Mischler EH, Wilfond BS, Fost N, Laxova A, Reiser C, Sauer CM, Makhholm LM, Shen G, Feenan L, McCarthy C, Farrell PM. \(1998\). Cystic fibrosis newborn screening: impact on reproductive behavior and implications for genetic counseling. *Pediatrics*, 102 \(1 Pt 1\), 44-52.](#)

Websites

Cystic Fibrosis Foundation

<http://www.cff.org>

Extensive research, education, support, and advocacy organization for Cystic Fibrosis. Information about life-course, treatment, research/clinical trials, and support groups. Accredits CF care centers where specialized CF providers can be located.

Videos

Cystic Fibrosis

Two-day professional staff training in 2007. Copies available at GDSP and ASCs on DVD.

Biotinidase Deficiency

Required Reading

[Wolf B, Heard GS. \(1990\). Screening for biotinidase deficiency in newborns: worldwide experience. *Pediatrics*, 85 \(4\), 512-517.](#)

Text Chapter

Wolf B. (2001). Disorders of biotin metabolism: treatable neurological syndromes. In R. Rosenberg, S. DiMauro, H. L. Paulson, L. Ptacek, & E. J. Nestler (Eds.), *The Molecular and Genetic Basis of Neurologic and Psychiatric Disease* (pp. 739-745). PA: Wolters Kluwer-Lippincott Williams and Wilkins.

Videos

Biotinidase Deficiency

Two-hour professional staff training in 2007 conducted by Dr. Bruce Barshop of UC San Diego. Copy available at GDSP on DVD. Powerpoint slides available at ASCs.

Hemoglobinopathies

Required Reading

TBD

Texts

Sickle Cell Counselor Training and Certification Course Manual, CDPH, Genetic Disease Screening Program (2008)

[NHLBI. \(2002\). *The management of sickle cell disease*. Array \[Bethesda, Md.\]: National Institutes of Health, National Heart, Lung, and Blood Institute, Division of Blood Diseases and Resources.](#)

Other Articles

[Inati A, Koussa S, Taher A, Perrine S. \(2008\). Sickle cell disease: new insights into pathophysiology and treatment. *Pediatr Ann*, 37 \(5\), 311-321.](#)

[Reed W, Lane PA, Lorey F, Bojanowski J, Glass M, Louie RR, Lubin BH, Vichinsky EP. \(2000\). Sickle-cell disease not identified by newborn screening because of prior transfusion. *J Pediatr*, 136 \(2\), 248-250.](#)

[Section on Hematology/Oncology Committee on Genetics; American Academy of Pediatrics. \(2002\). Health supervision for children with sickle cell disease. *Pediatrics*, 109 \(3\), 526-535.](#)

[Wojciechowski EA, Hurtig A, Dorn L. \(2002\). A natural history study of adolescents and young adults with sickle cell disease as they transfer to adult care: a need for case management services. *J Pediatr Nurs*, 17 \(1\), 18-27.](#)

Websites

Clinical Practical Guidelines for Management of Thalassemia Patients (2008)
<http://www.thalassemia.com/documents/thalhandbook2008.final.pdf>

Cooley's Anemia Foundation

<http://www.thalassemia.org>

Research-oriented support organization for Thalassemias. Resource for up-to-date clinical trial information.

Northern California Comprehensive Thalassemia Center
(Children's Hospital Oakland)

<http://www.thalassemia.com>

Extensive Thalassemia resource with alpha/beta/trait descriptions, patient-oriented life-course and treatment information, provider-oriented management guidelines, genetic counseling issues, US provider contacts, clinical trial and research information, support group contacts, and translated brochures in Cambodian, Chinese, Farsi, Tagalog, Vietnamese, and Thai.

Sickle Cell Disease Foundation of California

<http://www.scdfc.org/>

Organization with camps/programs, and support-group programs for parents of children with sickle cell disease (BabySteps), and listings of National and California-specific medical, health insurance, and support resources for children with sickle cell disease.

Sickle Cell Information Center
(Emory Univ., Grady Health Systems &
Morehouse School of Medicine, Atlanta, GA)

<http://www.scinfo.org>

Extensive information and links for professionals, families, & public on Sickle Cell.

Sickle Cell Disease Association of America, Inc.

<http://www.sicklecelldisease.org>

Parent organization of SCDFC (above), provides information and support resources in other locations across the United States for Sickle Cell.

Thalassemia Support Foundation (Southern California)

<http://www.HelpThals.org>

Support organization for individuals with Thalassemia. Brief information descriptions provided, has events for affected individuals.

Videos

Sickle Cell Counselor Training and Certification Course

A self-instructed video course of Module I "Introduction to Sickle Cell Disease, Sickle Cell Trait and Common Hemoglobinopathies" available on VHS or DVD at GDSP and ASCs.

Metabolic Conditions

Required Reading

[Banta-Wright SA, Steiner RD. \(2004\). Tandem mass spectrometry in newborn screening: a primer for neonatal and perinatal nurses. *J Perinat Neonatal Nurs*, 18 \(1\), 41-58; quiz 59-60.](#)

Recommended Text

Scriver, C. R., et al (2001) *The Metabolic and Molecular Bases of Inherited Disease*, (8th edition) McGraw-Hill, New York.

Other Text

Nyhan, W. L. et al., (2005). *Atlas of Metabolic Diseases*, (2nd ed), Hodder Arnold, London

Other Articles

[Chace DH, Kalas, TA, Naylor EW. \(2002\). The application of tandem mass spectrometry to neonatal screening for inherited disorders of intermediary metabolism. *Annu Rev Genomics Hum Genet*, 3, 17-45.](#)

[Ficicioglu C, Payan I. \(2006\). 3-Methylcrotonyl-CoA carboxylase deficiency: metabolic decompensation in a noncompliant child detected through newborn screening. *Pediatrics*, 118 \(6\), 2555-2556.](#)

[Hsu HW, Zytovicz TH, Comeau AM, Strauss AW, Marsden D, Shih VE, Grady GF, Eaton RB. \(2008\). Spectrum of medium-chain acyl-CoA dehydrogenase deficiency detected by newborn screening. *Pediatrics*, 121\(5\), e1108-1114.](#)

[Morris AA, Hoffmann GF, Naughten ER, Monavari AA, Collins JE, Leonard JV. \(1999\). Glutaric aciduria and suspected child abuse. *Arch Dis Child*, 80 \(5\), 404-405.](#)

[Rinaldo P, Matern D, Bennett MJ. \(2002\). Fatty acid oxidation disorders. *Annu Rev Physiol*, 64, 477-502.](#)

[Waisbren SE, Levy HL, Noble M, Matern D, Gregersen N, Pasley K, Marsden D. Short-chain acyl-CoA dehydrogenase \(SCAD\) deficiency: an examination of the medical and neurodevelopmental characteristics of 14 cases identified through newborn screening or clinical symptoms. *Mol Genet Metab*, 95 \(1-2\), 39-45.](#)

Websites

CDPH – NBS Reference Ranges

<http://www.cdph.ca.gov/programs/nbs/Documents/NBS-ReferenceRanges-May%202008.pdf>

California Children's Services

<http://www.dhcs.ca.gov/services/ccs>

Links Links and more Links (Society of Inherited Metabolic Disorders)

<http://www.simd.org/Links/>

Long, non-specific listing of links to medical societies, patient support groups, research institutions and journals that focus on inherited metabolic and mitochondrial disease. Links may or may not be up-to-date.

FOD Family Support Group

(Fatty Acid Oxidation Disorders – SCADD, LCHADD, etc.)

<http://www.fodsupport.org>

Videos

Metabolic Disorders (Disorders detectable by MS/MS)

Three-day professional staff training in 2005. Copies available at GDSP on VHS and ASCs on DVD.

Newborn Screening Expansion

California State Capitol Press Conference on 7/26/2005. Copies available at ASCs on DVD.

Topic Area: NBS Collection

Core Competency 12

Required Reading

TBD

Recommended Text

[CLSI \(2007\) Blood collection on filter paper for newborn screening programs; Approved standard – fifth edition, LA04-A5.](#)

Other Articles

American Academy of Pediatrics. (2000). Serving the family from birth to the medical home. A report from the Newborn Screening Task Force convened in Washington DC, May 10-11, 1999. *Pediatrics*, 106 (2 Pt 2), 383-427

[Preface](#), [Executive Summary](#), [Blueprint for the Future](#)

[Pass KA, Lane PA, Fernhoff PM, Hinton CF, Panny SR, Parks JS, Pelias MZ, Rhead WJ, Ross SI, Wethers DL, Elsas LJ 2nd. \(2000\). US newborn screening system guidelines II: follow-up of children, diagnosis, management, and evaluation. Statement of the Council of Regional Networks for Genetic Services \(CORN\). *J Pediatr*, 137 \(4 Suppl\), S1-46.](#)

Videos

Making a Difference Through Newborn Screening:
Blood Collection on Filter Paper (California Version)
NCCLS Video California Version

Video about the collection and process of using dried blood spot specimens for newborn screening. Copies are available at GDSP on DVD and ASCs on VHS.

Topic Area: Health Education

Core Competency 28 - Cultural Competency

Required Reading

TBD

Texts

Lynch, E. and Hanson, M. (1998). *Developing Cross-Cultural Competence: A Guide for Working with Young Children and Their Families, Second Edition*. Paul H. Brooks Publishing Co, Inc., Baltimore. 35-59.

<http://www.brookespublishing.com/store/books/lynch-7446/index.htm>

[Schilling B. U.S. Dept. of Agriculture, and U.S. Dept. of Health and Human Services, \(1986\). *Cross-Cultural Counseling: A Guide for Nutrition and Health Counselors.*](#)

Handouts

California Department of Public Health, Genetic Disease Screening Program Sickle Cell Counselor Training and Certification Course (2008), Socio-Cultural Assessment Tool, Strategies for Cultural Assessment, and Quick Guide for Cross-Cultural Counseling.

Websites

Office of Minority Health, NIH

<http://www.omhrc.gov/>

Provides data, statistics, and law/policy information on minority health. Provides links to cultural competence modules, programs, training, continuing education.

Culturally Competent Nursing Care: A Cornerstone of Caring

<https://ccnm.thinkculturalhealth.org/>

Free online educational program designed specifically for nurses and is accredited by the American Nurses Credentialing Center and the National Association of Social Workers.

A Physician's Practical Guide to Culturally Competent Care

<https://cccm.thinkculturalhealth.org/>

Free online educational program accredited for physicians, physician assistants, and nurse practitioners.

Health Care Language Services Implementation Guide

<https://hclsig.thinkculturalhealth.org/user/home.rails>

Web-based interactive tool that can assist health care organizations in planning, implementing, and evaluating language access services to better serve their limited English proficiency patient population and decrease disparities in access to health care

Nursing Spectrum/Nurse Week Continuing Educations

<http://www.nurse.com/ce/>

Non-specific, online, continuing education resources/classes for nurses. Requires a fee.

Topic Area: Evaluation, QI of ASCs – Processes and Tools

Core Competencies 45 – 50

Required Reading

[Hall LW, Moore SM, Barnsteiner JH. \(2008\). Quality and nursing: moving from a concept to a core competency. *Urol Nurs*, 28 \(6\), 417-425.](#)

Additional Websites on NBS and Genetics

Dolan DNA Learning Center

<http://www.dnalc.org/ddnalc/about/>

General genetics learning resource. Click on “Gene Almanac” or “Websites” link at the top for links to their modules on broad, genetics topics.

GeneReviews

<http://www.genetests.org>

Click tab at top of page for GeneReviews. Search by condition name. Provides comprehensive clinical information about natural history, inheritance, treatment, genetic counseling, molecular testing for many conditions. Authored by experts, funded by the NIH.

Genetic and Rare Disease (GARD)

<http://rarediseases.info.nih.gov/GARD>

Searchable NIH-run information and resource database for genetic and rare disease (including all NBS disorders) that provides links to descriptions, support groups, clinical trials, and services specific to that disorder.

National Human Genome Research Institute

<http://www.genome.gov/>

NIH Institute for genetic research, very research-oriented in clinical & bench science, policy/ethics implications, and genetic discrimination. Educational resources tab provides teaching materials for general public about genetics, genomics, testing, and genomic research.

National Organization for Rare Disorders (NORD)

<http://www.rarediseases.org>

Organizes financial (drug) assistance programs to individuals with some rare disorders (PKU, homocystinuria, tyrosinemia) who are unable to qualify for Medicaid, but may still have difficulty paying for medications. Of note is the PKU Medical Food Assistance Program for diagnosed individuals with family incomes up to 500% federal poverty level.

Online Mendelian Inheritance in Man (OMIM)

<http://www.ncbi.nlm.nih.gov/sites/entrez?db=omim>

Exhaustive research-oriented database of all documented genes and conditions in the literature about each specific disease and its variants. Focuses on molecular and clinical research: gene identification, symptomatology, epidemiology: links to references in PubMed.

Pediatrix

<http://www.pediatrix.com/>

Provider of MFM, newborn, and pediatric subspecialty services. Also provides newborn hearing screening. Of note is the Parent & Patient Information section, which provides patient-oriented summaries of disorders, and all aspects of NICU care (provider, test, procedure descriptions). Also has Spanish-language fact sheets of the same information.

STAR-G Screening Technology and Research in Genetics

<http://www.newbornscreening.info/index.html>

Multi-state collaboration to develop parent-oriented fact sheets on all NBS disorders. Fact sheets were used to develop the Parents' Guide booklets provided by CDPH-GDSP. Also has brief genetics overview for professionals. Contains list of annotated links addressing general NBS issues as well as Financial, Ethical, Legal, and Social Issues of NBS.