EHDI guidelines: hearing screening before one month of age, evaluation before 3 months of age and intervention before 6 months of age

A

AABR
automated ABR hearing test

AAC
Arizona Administrative Code

abnormal
a test value outside the reference range, a positive result

ABR
auditory brainstem response hearing test

ACHDNC

ACMG
American College of Medical Genetics

ACT sheets
ACTion sheets developed by ACMG for use by physicians as a quick reference to NBS results and needed actions for possible disorders
http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm

acylcarnitine
a fatty acyl ester of carnitine, the transport form for a fatty acid crossing the mitochondrial membrane. Acylcarnitines are named by the number of carbons in the acyl group (the side chain attached to carnitine):
- C0: no acyl group, free carnitine
- C3: 3 carbon acyl group attached to carnitine - propionylcarnitine
- C5OH: 5 carbon acyl group with hydroxyl on the 3"rd carbon - 3-hydroxyisovalerylcarnitine

ADHS
Arizona Department of Health Services

Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) in the Maternal and Child Health Bureau of HRSA, HHS. Source of the Recommended Uniform Screening Panel whose core disorders should be included in every newborn screening program. The secondary target disorders are defined as disorders that can be detected in the differential diagnosis of a core disorder. The ACHDNC maintains and periodically updates the panel.
http://www.hrsa.gov/heritabledisorderscommittee/uniformscreeningpanel.htm
alpha thalassemia
a hereditary hemolytic anemia marked by a decreased rate of synthesis of the α globin chains of hemoglobin. Four gene deletions produce alpha thalassemia major which is incompatible with life. Affected infants are stillborn or born with severe hydrops fetalis (abnormal accumulation of fluid in the entire body of the infant) and die quickly. If transfusions are given prenatally and continued throughout life, a baby can survive. Three gene deletions produce Hemoglobin H disease, two produce alpha thalassemia trait, and one the “silent carrier.”

alpha thalassemia trait
a condition in which an individual inherits two working genes for alpha globin chain synthesis. Individuals with this condition will have a mild microcytic, hypochromic anemia. This decrease in alpha globin chain production does not lead to health problems.

Asians with alpha thalassemia trait usually, but not always, have the two non-working genes on the same chromosome (cis-type) making it possible to have a child with hydrops fetalis. African Americans with alpha thalassemia trait usually, but not always, have the two non-working genes on different chromosomes (trans-type) and therefore can only transmit one non-working gene to their offspring.

American College of Medical Genetics (ACMG)
the professional organization for biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors and other health care professionals committed to the practice of medical genetics. ACMG provides education, resources and a voice for the medical genetics profession.  http://www.acmg.net

ACMG 29
29 original target disorders for newborn screening proposed by ACMG in the report entitled, Newborn Screening: Toward a Uniform Screening Panel and System (2006) and recommended by ACHDNC http://www.acmg.net/AM/Template.cfm?Section=Practice_Guidelines&Template=/CM/HTMLDisplay.cfm&ContentID=3671

analyte
the chemical compound being tested for (e.g. the analyte for CAH testing is 17-OHP)
anemia
   a condition in which the blood is deficient in the number of red blood cells, in the quantity of hemoglobin, or in the total volume of packed red blood cells and may result from excessive blood loss, excessive blood cell destruction or from decreased blood cell production, iron deficiency or bone marrow failure.

   hemolytic anemia - resulting from hemolysis of red blood cells
   hypochromic anemia - where hemoglobin is deficient and cells appear paler than normal
   macrocytic anemia - marked by abnormally large red blood cells
   microcytic anemia - marked by abnormally small red blood cells

Arizona Administrative Code (AAC)
   where the official rules of the state of Arizona that govern state agencies, boards and commissions are published. See Rules.

Arizona Department of Health Services (ADHS)
   the agency housing the State Laboratory and the Arizona Newborn Screening Program
   http://www.azdhs.gov

Arizona Health Care Cost Containment System (AHCCCS)
   Arizona’s Medicaid agency that provides health care to Arizonans that meet income and other eligibility requirements
   http://www.azahcccs.gov

Arizona Revised Statutes (ARS)
   The statute establishing the Arizona Newborn Screening Program is found in ARS 36-694 (Report of blood tests; newborn screening program; committee; fees; definitions)
   http://www.azleg.gov/ars/36/00694.doc

Arizona State Public Health Laboratory (ASL)
   in the Bureau of State Laboratory Services, Division of Public Health Services, ADHS – the designated laboratory for newborn screening in Arizona
   http://www.azdhs.gov/lab

ARS
   Arizona Revised Statutes

ASA
   argininosuccinic acidemia/argininosuccinic aciduria, a core disorder of the original Recommended Uniform Screening panel

ASDB
   Arizona State Schools for the Deaf and the Blind
ASL
American Sign Language

assay
a test or analysis

atresia
congenital absence or closure of a normal body opening or tubular structure

audiology
the branch of science that studies hearing, balance, and related disorders. Audiologists test hearing and make recommendations about treatment options (hearing aids, cochlear implants, surgery) and appropriate medical referrals

AutoDelfia®
an automatic immunoassay system from PerkinElmer – the method used for TSH, 17-OHP and IRT testing by the State Lab.

auditory brain stem response (ABR)
a method used for hearing screening which measures the brain’s response to sound. A device near the ear makes clicking sounds while earpieces in the ear canals conduct the sound. Electrode pads placed on the forehead and behind the ear track the sound moving through the ear to the brain. Results are averaged and compared with normal hearing. This method may also be used in a more extensive, modified form as a diagnostic test.

autosomal recessive inheritance
inheritance pattern carried on a pair of the non-sex determining chromosomes (the 22 pairs of autosomes) where matching genes for a condition must be inherited from both parents

All of the disorders on the newborn screening panel except congenital hypothyroidism are inherited in an autosomal recessive inheritance pattern.

AzEIP
Arizona Early Intervention Program (acronym pronounced Ay-zip)

B

BAER
brainstem auditory evoked response hearing test
Barts
an abnormal, usually transitory, type of hemoglobin that is not effective in oxygen transport. It contains 4 gamma globin chains. It is often present in small amounts in newborns but larger amounts indicate alpha thalassemia trait, Hemoglobin H disease or alpha thalassemia.

BCAA
branched chain amino acids (leucine, isoleucine, valine)

beta thalassemia (β thal)
a hereditary hemolytic anemia marked by a decreased rate of synthesis of the β globin chains of hemoglobin. The homozygous form (Cooley's, Mediterranean or erythroblastic anemia) in which β globin is completely absent is also referred to as β° thalassemia (or β° thal). The NBS hemoglobin result would be F only. When some beta chains are present but in decreased amount it is called β+ thalassemia (β+ thal). The NBS hemoglobin result would be FA or possibly F only. In an older individual, there would be increased amounts of Hb A2.
http://www.cooleysanemia.org

β thalassemia trait
also called β thalassemia minor

BIO
biotinidase deficiency, a core disorder of the original Recommended Uniform Screening panel

Bio-Rad
Bio-Rad Laboratories, supplier of the kit for the galactose-1-phosphate uridyl transferase assay to screen for galactosemia.
http://www.bio-rad.com

BKT
beta-ketothiolase deficiency, a core disorder of the original Recommended Uniform Screening panel

borderline
for some tests, an equivocal or borderline range is reported. It is outside the reference range but is not frankly abnormal. The program follows these results but not as immediately or aggressively (e.g. borderline TSH or 17-OHP results)

branched chain amino acids
leucine, isoleucine and valine
CAH
congenital adrenal hyperplasia, a core disorder of the original Recommended Uniform Screening panel

carrier
a heterozygote for a recessively inherited disorder where two copies of the disease-causing gene must be present in order for the disease to be expressed. Carriers or those with traits usually have no symptoms of disease or milder symptoms under stressful conditions.

CBAVD
congenital bilateral absence of the vas deferens, a common form of sterility in males with CF or CFTR mutations

Cbl A, B
methylmalonic acidemia (cobalamin defects), a core disorder of the original Recommended Uniform Screening panel

CDC
Centers for Disease Control and Prevention – an agency of HHS

Centers for Disease Control and Prevention (CDC)
an agency of HHS providing community health protection through health promotion, prevention of disease, injury and disability and preparedness for new health threats.
http://www.cdc.gov
http://www.cdc.gov/newbornscreening

CF
cystic fibrosis, one of the 29 core disorders of the original Recommended Screening panel

CFR
Code of Federal Regulations

CFTR gene
cystic fibrosis transmembrane conductance regulator gene

CH
congenital hypothyroidism, a core disorder of the original Recommended Uniform Screening panel

CIT-1
citrullinemia type I, a core disorder of the original Recommended Uniform Screening panel
CLIA
Clinical Laboratory Improvement Act of 1965 (Amendments 1988)

Clinical and Laboratory Standards Institute (CLSI)
www.clsi.org

clinical diagnosis
diagnosis based on signs, symptoms and laboratory findings

Clinical Laboratory Improvement Act of 1965 (CLIA) with amendments in 1988
In order to ensure quality laboratory testing, the Centers for Medicare & Medicaid Services (CMS) in HHS regulates all laboratory testing (except research) performed on humans in the U.S. through the Clinical Laboratory Improvement Amendments (CLIA). In total, CLIA covers approximately 200,000 laboratory entities including the Arizona State Laboratory. http://www.cms.hhs.gov/CLIA/01_Overview.asp#TopOfPage

CLSI
Clinical and Laboratory Standards Institute

CMV
cytomegalovirus

collection device
another name for collection kit, filter paper kit, dried bloodspot card or newborn screening card (sometimes called the Guthrie card in honor of its developer)

collection kit
the numbered card with special filter paper attached for collection of drops of blood that are then dried before shipping and testing. It has areas for demographic information, accession number, date stamp, bar code, etc. Each state designs their own card but all contain the same standardized filter paper

confirmatory/diagnostic test
a test to show whether a condition suspected because of screening test results is present or not (for screening using dried blood spots, this testing is from a specimen other than the screening specimen).

congenital
present from birth, but not necessarily genetic

congenital bilateral absence of the vas deferens
a common form of sterility in males with CF or CFTR mutations

Cooley’s anemia
thalassemia major, ß° thalassemia
CRS
Children’s Rehabilitative Services

CUD
carnitine uptake defect, a core disorder of the original Recommended Uniform Screening panel

cutoff
the first value outside the reference range (the point where a result is reported as abnormal or positive for the disease tested for)

Cystic fibrosis transmembrane conductance regulator gene (CFTR gene)
Mutations to this gene can cause cystic fibrosis (over 1000 mutations to this gene have been identified). The Arizona NBS program uses a DNA kit from Third Wave Technologies to detect 46 mutations to this gene.

DBS
dried blood spot

DHHS
United States Department of Health and Human Services

diagnosis
identification of a disease by history, lab tests and symptoms

dietary monitoring specimen
To monitor phenylalanine levels so that diets can be adjusted for individuals with PKU, regardless of their ages, the State Lab receives specimens and reports phenylalanine levels only, at no charge

DOB
date of birth

DOC
date of collection (of NBS sample)

DOR
date of receipt of NBS sample by the State Lab.

DPOAE
distortion product otoacoustic emissions

dried blood spot
blood collected usually from a heel stick and air dried on an approved filter paper card
ECMO 
extracorporeal membrane oxygenator – similar to a heart/lung bypass machine

EHDI 
Early Hearing Detection and Intervention (acronym pronounced “eddy”)

EI 
early intervention

ELBW 
extremely low birth weight

-emia (suffix) 
of the blood

endocrine 
relating to or affecting those glands that secrete their products (hormones) directly into the bloodstream.

enteral 
within or by way of the intestine.

enzyme 
a protein produced by living organisms that catalyzes (increases the rate of) a specific chemical reaction but is not consumed in the reaction or altered upon completion of the reaction

Enzymes are inactivated by heat and/or humidity and both enzyme assays (for biotinidase and GALT activity) in the newborn screening panel can have false positive results particularly in the summer if samples have been exposed to heat, strong sunlight and/or humidity.

extracorporeal membrane oxygenator (ECMO) 
similar to a heart/lung bypass machine, ECMO can be considered to be a continuous transfusion of red blood cells and other blood components and any NBS samples taken during ECMO are invalid.

extremely low birth weight (ELBW) 
birth weight less than 1000 g

false negative 
a negative (normal) result in an affected individual (one who has the disease)

false positive
a positive (abnormal) result in an individual without the disease

fatty acid
an organic saturated or unsaturated carboxylic acid containing a single carboxyl group and usually an even number of carbon atoms that can be combined with glycerol to form fats (glycerol plus 3 fatty acids make a triglyceride).

fatty acid oxidation
stepwise catabolism of fatty acids in which two-carbon fragments are successively removed from the carboxyl end of the chain

fatty acid oxidation disorders (FODs)
a group of genetic metabolic disorders in which the body is unable to oxidize (breakdown) fatty acids to make energy because an enzyme is either missing or not working correctly. When the supply of glucose, the main source of energy for the body, runs out, fat is broken down to supply energy but this can’t happen in individuals with one of these disorders.

FFP
fresh frozen plasma

filter paper
special standardized filter paper has been designed to collect blood spots for newborn screening tests. One large drop of blood will soak evenly through the filter paper to the back and then spread out in the circle so that punches from the circle will each contain the same amount of blood.

filter paper kit
another name for a specimen collection kit

first screen
the initial specimen collected from a newborn who is less than five days of age

follow-up
actions taken to ensure that a newborn whose screening test results are positive or unsatisfactory receives appropriate, prompt further testing and evaluation; and actions taken to ensure that the newborn screening system can evaluate the effectiveness of screening.
fresh frozen plasma

Transfusion of this blood product can produce false negative newborn screening results for biotinidase testing only. The effect is transitory and a valid repeat screen can be collected a few days to a week after transfusion.

G

GA-1

glutaric acidemia type 1, a core disorder of the original Recommended Uniform Screening panel

galactose

a simple sugar (an isomer of glucose) which in combination with glucose forms lactose

GALT

galactosemia, a core disorder of the original Recommended Uniform Screening panel

Gal-1-PUT

galactose-1-phosphate uridyltransferase: the enzyme whose absence or reduced activity causes galactosemia

GBYS

Guide By Your Side Program, Hands and Voices

genotype

the genetic material of an individual, the pair of genes present for a particular characteristic or protein

gestation

the length of time from conception to birth. The average gestation in humans, calculated from the first day of the last normal menstrual period is 280 days (40 weeks), with a normal range of 259 days (37 weeks) to 287 days (41 weeks). Infants born prior to the 37th week are considered premature and those born after the 41st week, postmature.

gestational age

the length of the pregnancy at birth measured from the first day of the last menstrual period, in weeks.

globin

the protein portion of hemoglobin (consisting of 2 alpha chains and 2 beta chains in hemoglobin A)
glucose
a simple sugar; also called D-glucose or dextrose, released by digestion of starch and other sugars, absorbed by the small intestine and circulated to the liver where excess is converted to glycogen. Within most cells, glucose is the primary energy source and is oxidized to carbon dioxide and water to produce energy.

Guthrie, Robert
U.S. microbiologist (1916-1995), the “father of newborn screening”

Guthrie test
another name for a newborn screening test or a dried bloodspot test where blood is collected on filter paper. This type of test was first developed for newborn screening by Dr. Robert Guthrie.

H

Hb S/ß thal
S, beta-thalassemia, a core disorder of the original Recommended Uniform Screening panel

Hb S/C
S, C disease, a core disorder of the original Recommended Uniform Screening panel

Hb SS
sickle cell disease or sickle cell anemia, a core disorder of the original Recommended Uniform Screening panel

HCY
homocystinuria, a core disorder of the original Recommended Uniform Screening panel

health care provider
a physician, physician assistant, registered nurse practitioner or midwife.

Health Insurance Portability and Accountability Act of 1996 (HIPAA)
a federal law creating national standards for recording and disclosing or keeping private protected health information (PHI).

45 CFR 164.512 contains the exception where covered entities are not required to obtain written authorization for the disclosure of PHI for public health purposes: when the uses and disclosures are required by law or where they are disclosed to a public health authority “for the purpose of preventing or controlling disease...”

Health Resources and Services Administration (HRSA)
an agency of HHS
http://www.hrsa.gov

HEAR
hearing loss, a core disorder of the original Recommended Uniform Screening panel
heelstick
a method for obtaining a blood sample from a newborn where the heel is punctured in the lateral or medial area of the plantar surface of the heel (avoiding the posterior curvature of the heel) to a depth of no more than 2.4 mm.

heme
an iron compound that is the portion of hemoglobin that carries oxygen. There is one heme associated with each of the four globin chains of a hemoglobin molecule.

hemoglobin
an iron-containing pigment of red blood cells that functions primarily in the transport of oxygen from the lungs to the tissues of the body, that consists of four polypeptide (globin) chains of which two are of the type designated alpha and two are of one of the types designated beta, gamma, or delta and each of which is linked to a heme molecule, that combines loosely and reversibly with oxygen in the lungs and with carbon dioxide in the tissues

hemoglobin A (Hb A)
the hemoglobin in the red blood cells of the normal human adult that contains two alpha and two beta globin chains, each with a heme attached

hemoglobin A2 (Hb A2)
a variant hemoglobin found in small quantities (1-2%) in normal human adults (two alpha and two delta globin chains)

hemoglobin C (Hb C)
an abnormal hemoglobin that differs from hemoglobin A in having a lysine residue substituted for the glutamic acid residue at position 6 of each of the two β globin chains in a hemoglobin molecule

hemoglobin C disease
homozygous Hb C disease: an inherited hemolytic anemia that occurs most often in individuals of African descent and is characterized especially by splenomegaly, arthralgias, abdominal pain and the presence of target cells and hemoglobin C in the blood

hemoglobin electrophoresis
a test that measures the different types of hemoglobin in the blood by their movement as charged particles suspended in a liquid on gel under the influence of an applied electric field.
hemoglobin F (Hb F)
fetal hemoglobin (containing 2 alpha and 2 gamma globin chains) is the predominant hemoglobin found in the fetus and makes up more than half of a newborn’s total hemoglobin at birth. The amount of Hb F decreases after birth and a small amount of fetal hemoglobin (< 2%) can be present in adult hemoglobin.

hemoglobin H (Hb H)
an abnormal, fast migrating hemoglobin occurring in the red blood cells when alpha globin chains are present in reduced amounts and excess beta chains come together in tetramers (4 beta chains)

hemoglobin H disease
a chronic inherited hemolytic anemia resulting from only one working copy of the gene for alpha globin synthesis (3 gene deletions or two gene deletions and one non-deletional mutation). It is marked by hypochromic red blood cells with inclusions that cause them to resemble golf balls.

hemoglobin H – Constant Spring disease
a chronic inherited hemolytic anemia resulting from only one working copy of the gene for alpha globin synthesis and a structural defect called Constant Spring in one of the non-working copies of the gene. It is a more severe form of Hemoglobin H disease with variable consequences up to transfusion dependency.

hemoglobinopathy
an inherited disorder of hemoglobin structure with characteristic clinical and laboratory abnormalities and often overt anemia.

hemoglobin S (Hb S)
an abnormal hemoglobin occurring in the red blood cells in sickle-cell anemia and sickle-cell trait and differing from hemoglobin A in having a valine residue substituted for the glutamic acid residue in position 6 of the two ß globin chains in the hemoglobin molecule.

hemolysis
breakdown or destruction of red blood cells with liberation of hemoglobin

HFK
Hear For Kids, a program of the EAR Foundation of Arizona

HHS
U.S. Department of Health and Human Services
high performance liquid chromatography (HPLC)
the method by which hemoglobin results are confirmed at the State Lab.
a form of column chromatography used to separate, identify and quantify compounds based on their polarities and interactions with the column’s stationary phase (the particles packed in the column). HPLC uses different types of stationary phase, a pump to move the mobile phase (sample dissolved in a liquid) through the column and a detector that records a retention time for the analyte.

HIPAA
Health Insurance Portability and Accountability Act of 1996

HI*TRACK
the data management system from NCHAM used by hospitals, audiologists and the Newborn Screening Program to record and report hearing screening, diagnosis and early intervention information
http://www.hitrack.org

HMG
3-hydroxy-3-methylglutaric aciduria, a core disorder of the original Recommended Uniform Screening panel

HOPS
hearing outpatient screening

HPLC
high performance liquid chromatography

HRSA
Health Resources and Services Administration – an agency of HHS

hydrolysis
any reaction in which water is one of the reactants; a chemical decomposition in which a substance is split into simpler compounds by the addition of the taking up of the elements of water.

17-α-hydroxyprogesterone
an adrenal hormone that builds up in the absence of 21-hydroxylase and is used as the analyte for CAH testing

hyper- (prefix)
abnormally increased, excessive

hyperplasia
abnormal increase in the number of normal cells in normal arrangement in an organ or tissue which increases its size or volume

hypo- (prefix)
deficient, low in quantity

t hypochromic
in red blood cells, a decrease in the quantity of hemoglobin present so that they are abnormally pale

hypoplasia
  underdevelopment of a tissue, organ or body

idiopathic
  pertaining to conditions without clear pathogenesis, or disease without recognizable cause, as of spontaneous origin.

IEF
  isoelectric focusing

IEM
  inborn errors of metabolism – another name for metabolic disorders

incidence
  the rate at which an event occurs per unit time per person in the members of a defined population at risk; the rate of occurrence of new cases of a particular disease in a population being studied

inconclusive
  a test result that cannot be determined to be either normal or abnormal because of some confounding factor. A red blood cell transfusion prior to collecting a newborn screening sample would give an inconclusive result for hemoglobin since the sample contains cells that belong to the donor and not exclusively to the baby tested.

Individuals with Disabilities Education Act (IDEA)
  a law enacted in 1975 ensuring services to children with disabilities. IDEA governs how states and public agencies provide early intervention, special education and related services to more than 6.5 million eligible infants, toddlers, children and youth with disabilities. Infants and toddlers with disabilities (birth-2) and their families receive early intervention services under IDEA Part C. Children and youth (ages 3-21) receive special education and related services under IDEA Part B.

infant
  a baby from birth until a year of age (NBS rules identify an infant as one from 29 days of age until a year of age – for the first four weeks an infant is defined as a newborn).
isolectric focusing (IEF) is the method used by the State Lab to identify variant hemoglobins. Results are then confirmed by HPLC. It is a technique for separating different proteins by their electrical charge differences (due to their relative content of acidic and basic side chains).

Samples are distributed over a gel medium that has a pH gradient. An electric current is passed through the medium, creating a positive and negative end. Charged hemoglobin molecules migrate toward the oppositely charged end through the changing pH gradient until they reach a pH point that corresponds to their isoelectric points (pI) - the point where they no longer have a net charge. No further migration takes place and the proteins become focused into bands at this point.

IVA
isovaleric acidemia, a core disorder of the original Recommended Uniform Screening panel

J
julian date
consecutive numbering of the days of the year beginning with 001 for January 1st and ending with 365 for December 31st (in non-leap years)

K
ketone
an organic compound with a carbonyl group (C=O) attached to two carbon atoms. Acetone (C₃H₆O) is a simple ketone. Ketones are formed from incomplete metabolism of fatty acids, usually from carbohydrate deficiency.

kit number
the unique number printed on the collection kits (under the bar code, in red on the upper right hand side of the demography entry sheets and above the circles on the filter paper). This number follows the designation AZ and is sometimes known as the AZ number.

L
lab number
unique identification number assigned by the State Lab to newborn screening bloodspot specimens (see accession number)

lactose
a disaccharide that on hydrolysis yields glucose and galactose

lactose formula
a milk-based formula with lactose as its carbohydrate source

LBW
low birth weight

LCHAD
long-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency, a core disorder of the original Recommended Uniform Screening panel

linked kits
double collection kits that can be taken apart at the middle perforation. The top kit is marked 1 and the bottom kit is marked 2. These kits are intended to be used by hospitals to collect a first specimen with the number 1 kit. The number 2 kit is to be placed in a bright pink envelope and sent home with the baby with instructions to take it to the first visit to the doctor (to be used to collect the second specimen). The kit numbers allow the two parts to be matched in the system.

lost to follow-up (LTF)
a follow-up case closed when a baby whose family cannot be located for follow-up activities (further testing, referral to specialist, etc.) despite completion of follow-up activities per protocol.

low birth weight (LBW)
birth weight of 2500 g (5.5 pounds) or less

LTF lost to follow-up

M
cailer
the lab report for each newborn screening test. A copy is mailed to the submitter of the specimen and to the doctor listed who ordered the test.

Maternal and Child Health Bureau (MCHB)
a bureau of HRSA, HHS that provides the Maternal and Child Health Block Grant to state maternal and child health programs under Title V of the Social Security Act of 1935
http://mchb.hrsa.gov

MCAD
medium chain acyl-CoA dehydrogenase deficiency, a core disorder of the original Recommended Uniform Screening panel

3-MCC
3-methylcrotonyl-CoA carboxylase deficiency, a core disorder of the original Recommended Uniform Screening panel
MCD
multiple carboxylase deficiency, a core disorder of the original Recommended Uniform Screening panel

MCH
maternal and child health

MCHB
Maternal and Child Health Bureau of HRSA, HHS

MCH Block Grant
funding provided by MCHB to states under Title V of the Social Security Act of 1935 and its amendments
http://mchb.hrsa.gov/programs/blockgrant/overview.htm

MCT oil
medium chain triglyceride oil

meconium
first feces of a newborn infant (greenish black and tarry)

meconium ileus
a blockage of the intestines with impacted meconium in a newborn – usually in infants with cystic fibrosis

metabolic disorders
disorders affecting metabolism – usually inherited defects of enzyme production where specific enzymes are inactive and a metabolic pathway is blocked and certain substances build up to toxic levels.

metabolic formula
a formula food designed to meet the needs of individuals with metabolic disorders (e.g. for PKU, a formula with most or all of the phenylalanine removed)

metabolic pathway
a sequence of connected, enzyme-catalyzed reactions in cells that either builds a complex molecule (anabolic pathway) or breaks down a complex molecule into simpler compounds (catabolic pathway).

metabolism
the sum of the processes in the buildup and destruction of living tissue, the chemical changes in living cells by which energy is provided for vital processes and activities and new material is assimilated (see anabolism and catabolism).

metabolite
a compound that is a starting material for a metabolic process, an intermediate product of metabolism (a product of one metabolic process that is essential to another process in the same organism) or a metabolic end product that is usually excreted

microcytic
microtia
unusually small and underdeveloped external ear

MOD
March of Dimes

MS/MS
tandem mass spectrometry

MSUD
maple syrup urine disease, a core disorder of the original Recommended Uniform Screening panel

MUT
methylmalonic acidemia – mutase deficiency, a core disorder of the original Recommended Uniform Screening panel

mutation
a permanent transmissible change in the genetic material (DNA)

N

National Newborn Screening and Genetics Resource Center (NNSGRC)
a cooperative agreement between the Maternal and Child Health Bureau (MCHB), Genetics Services Branch and the University of Texas Health Science Center at San Antonio, Department of Pediatrics that provides information and resources in the area of newborn screening and genetics
http://genes-r-us.uthscsa.edu

National Newborn Screening Information System (NNSIS)
housed at NNSGRC where states report their NBS statistics and national reports are published

NBHS
newborn hearing screening

NBS
newborn screening

negative result
a test value within the reference range, a normal result
neonatal
    of, relating to, or affecting the newborn and especially the human infant during the first month after birth

neonatologist
    a medical specialist (pediatrician) trained in the care of neonates that require intensive care.

neurotology
    a clinical subspecialty within otolaryngology (ENT) that focuses on the neurology and neurosurgery of the ear related to sensorineural hearing and balance disorders

newborn
    a neonate: a term applied to human infants less than a month of age

newborn screening (NBS)
    testing done within days of birth to identify infants at increased risk for specific disorders so that treatment can begin as soon as possible; when a newborn screening result is positive, further diagnostic testing is usually required to confirm the results.

Arizona Newborn Screening Program: http://www.aznewborn.com
State NBS contacts: http://genes-r-us.uthscsa.edu/state_contacts.pdf

Newborn Screening Advisory Committee
    A committee appointed by the Director of ADHS to provide recommendations and advice to the department regarding tests that the committee believes should be included in the Newborn Screening Program. It meets annually and includes the following members: seven physicians who represent the medical specialties of endocrinology, pediatrics, neonatology, family practice, otology and obstetrics; a neonatal nurse practitioner; an audiologist, a representative of an agency that provides services under Part C of the individuals with disabilities education act; at least one parent of a child with a hearing loss or a congenital disorder; a representative from the insurance industry; the director of AHCCCS or designee; and a representative of the hospital or health care industry. See Rules.

NICU
    neonatal intensive care unit

NIH
    National Institutes of Health – the medical research agency of HHS

NNSGRC
    National Newborn Screening and Genetics Resource Center

NNSIS
    National Newborn Screening Information System
OAE
otoacoustic emissions

OC不予
Office for Children with Special Health Care Needs (acronym pronounced ocean)

Office for Children with Special Health Care Needs (OCSHCN)
in the Division of Behavioral Health Services, ADHS, (602) 542-1860
The Children’s Rehabilitative Services (CRS) Program is administered through this office
http://www.azdhs.gov/phs/ocshcn/index.htm

17-OHP
17-α-hydroxyprogesterone, an adrenal hormone that builds up in the absence of 21-hydroxylase and is used as the analyte for CAH testing

organic acid disorders (organic acidemias)
rare inherited metabolic disorders (OAs) that occur when enzymes necessary in the breakdown of certain protein components and other substances are missing or not working properly. This results in the excretion of non-amino organic acids in urine.
Support group: Organic Acidemia Association: http://www.oaanews.org

otoacoustic emissions testing (OAE)
a method used for hearing screening which measures otoacoustic emissions generated by sound. An earpiece with a microphone measures the cochlea’s response to sound, listening for otoacoustic emissions (weak echo sounds made by the ear soon after it hears). If no otoacoustic emissions are recorded, it could mean possible hearing loss. The OAE does not require the baby to respond to sound. A pass result suggests that the cochlea is working normally. A baby who doesn’t pass may have a temporary hearing loss because of fluid or a permanent hearing loss.

P

packed red blood cells (PRBC)
Transfusion of red blood cells can cause false negative newborn screening results for hemoglobinopathies and galactosemia. A valid repeat screen can be collected 3 – 4 months after the last transfusion.

parent refusal
Parents are allowed to refuse a newborn screening test for their infant. For documentation, the physician obligated to order the test will have the parents sign a form acknowledging that they understand the possible consequences of not screening their infant. The collection kit has a box to check for parent refusal. The demographics should be filled out completely, the box checked and the kit sent without blood to the State Lab.

patient number
the lab number of the screen (usually a first screen) that a subsequent screen is linked to. When a screen is unlinked its patient number is its lab number. When a screen is linked to another screen, its patient number is the lab number of the screen it is linked to. If the patient number is different from the lab number it signifies that this screen is linked to another. If the patient number is the same as its lab number it shows that it is not linked to any other screen but other screens may be linked to it.

Perkin Elmer
Perkin Elmer, Inc. – supplier of the AutoDelphia automatic immunoassay systems, Waters Quattro Micro tandem mass spectrometers and assay kits for TSH, 17-OHP, hemoglobin, IRT and analytes for MS/MS. 
http://www.perkinelmer.com

PHI
protected health information (kept confidential under HIPAA privacy rules)

PKU
phenylketonuria, a core disorder of the original Recommended Uniform Screening panel

positive predictive value
the percentage of true cases of disease identified in a screening test (true positives) out of the total number of positive screens- the higher the percentage, the better the test results identify those with disease. For example, a test where there were 10 abnormals reported and one true positive would have a PPV of 10% (a good test) and a test where there were 100 abnormals and one true positive would have a PPV of 1% (not as good a test).

positive result
a test value outside the reference range, an abnormal result

practitioner
one who has met the professional and legal requirements necessary to provide a health care service, such as a physician, nurse, dentist or physical therapist.

premature
born at any time prior to completion of the 37th week of gestation

prevalence
the frequency of a trait in a population, the percentage of a population that is affected with a particular disease at a given time

PROP
propionic acidemia, a core disorder of the original Recommended Uniform Screening panel

prophylactic, prophylaxis
measures designed to preserve health and prevent the spread of disease; protective or preventive treatment
Propylthiouracil (PTU)
a medication that suppresses thyroid hormone production and is used in the treatment of hyperthyroidism. It crosses the placenta and if a pregnant woman is treated, her baby can have transient hypothyroidism.

PTU
propylthiouracil, a medication that suppresses thyroid hormone

Q
QNS
quantity not sufficient

R
RBC
red blood cell
recall screen
a sample submitted after an abnormal repeat screen. Only 3 circles of blood are needed since only the test for the previously abnormal analyte(s) is performed (at no charge by the State Lab)

Recommended Uniform Screening Panel
the list of 30 core disorders and 26 secondary disorders recommended by the Secretary’s Advisory Committee on Heritable Disorders of Newborns and Children (ACHDNC) for inclusion in every state newborn screening program. In May, 2010, SCID and related T-cell lymphocyte deficiencies were added to the core disorders and secondary disorders respectively. Arizona screens for the original 29 core disorders and occasionally detects a secondary disorder, but does not yet screen for SCID.
http://www.hrsa.gov/heritabledisorderscommittee/uniformscreeningpanel.htm

reference range
the range of normal values for a particular analyte or test

Rules

S
SACHDNC
HHS Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children (in the Maternal and Child Health Bureau of HRSA, HHS). See ACHDNC.
salt-wasting, salt-losing CAH
see congenital adrenal hyperplasia

sample
another name for specimen

SCID
Severe combined immunodeficiency disorder, a core disorder of the Recommended Uniform Screening panel (added to the panel in May, 2010). Arizona does not yet screen for SCID.

screening test
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 screen
a specimen collected from a newborn after a first specimen has been collected or from an infant at least five days old and not older than one year of age, regardless of whether a first specimen was collected.

sensorineural hearing loss
occurs when there is damage to the inner ear (cochlea) or to the nerves from the inner ear to the brain and is considered to be a permanent loss. This can be caused by diseases, birth injury, drugs and genetic syndromes or as a result of noise exposure, head trauma, aging and tumors.

S-HPFH
sickle/hereditary persistence of fetal hemoglobin – a much milder form of sickle cell disease

SNHL
sensorineural hearing loss

soy formula
infant formula with soy protein isolate as its protein source and usually sucrose as its carbohydrate source - used when babies cannot tolerate lactose in milk-based formulas
specificity
in a screening test – the proportion of people who are truly free of a specific disease and are so identified by the test. A test with high specificity would have a minimum of false positives.

specimen
a part of a thing intended to show kind and quality of the whole; for Newborn Screening, the filter paper containing dried blood spots and the identifying demographic information

specimen kit
the strip of filter paper for collecting a blood sample attached to a form for obtaining identifying information about the infant - another name for collection kit

specimen number
unique identification number assigned by the State Lab to each newborn screening bloodspot specimen (see accession number)

submitter
any entity that submits newborn screening specimens to the State Lab for testing. Each submitter is assigned an identifying code by the Demographic section of the Newborn Screening program. Results are mailed to submitters and physicians ordering the tests.

supplemental kits
single NBS collection kits, usually used for repeat tests but containing check boxes to identify what type of specimen is being submitted

T
tandem mass spectrometry (MS/MS)
an analytical method that can determine the substances present in a sample by separating them by their weights (masses) and then measuring the amounts of the substances present. This is done by ionizing chemical compounds to generate charged molecules or molecule fragments and measuring their mass-to-charge ratios. As a newborn screening method, MS/MS can test for amino acids and acylcarnitines and detect amino acid, fatty acid oxidation and organic acid disorders.

target cell
a red blood cell with a darker round central area surrounded by a paler ring, which in turn is surrounded by a darker ring making it look like a bulls eye target

TEOAE
transiently evoked otoacoustic emissions
testing fees

Arizona NBS specimens are billed to submitters at $30.00 for a first screen and to insurance companies or responsible parties at $40.00 for a second screen. These fees are set in Rules (R9-13-208)


TFP

trifunctional protein deficiency, a core disorder of the original Recommended Uniform Screening panel

thalassemias

a group of hereditary hemolytic anemias marked by a decreased rate of synthesis of one or more hemoglobin polypeptide (globin) chains classified according to the chain involved (α-thalassemia – absence or reduced number of alpha chains available for combination into the globin portion of hemoglobin; β-thalassemia – absence or reduced number of beta chains)

Third Wave

Third Wave Technologies, Inc. now Hologic, Inc., provider of the CFTR InPlex™ reagent kits used by the Arizona Newborn Screening Program to detect 46 mutations to the CFTR gene

http://www.twt.com

Title V

the section of the Social Security Act which funds State programs which improve health and welfare services for mothers and children. These funds are provider in the Maternal and Child Health Services Title V Block Grant.

ture positive

a positive (abnormal) result in an individual with the disease

TYR-1

tyrosinemia type 1, a core disorder of the original Recommended Uniform Screening panel

U

Uniform Screening Panel

see Recommended Uniform Screening Panel, ACHDNC, ACMG 29, ACMG 54

unilateral hearing loss

normal hearing in one ear and hearing loss in the other ear

Universal Precautions

an OSHA standard that protects employees who may be occupationally exposed to blood and other potential infectious materials like other body fluids

unsat
an unsatisfactory specimen which will be accessioned as specimen type 5 (if it is a first screen or specimen type 6 if it is a second screen – if it is made unsat after accessioning, it will have unsat results listed for all tests on the panel but its specimen type will remain whatever it was originally)

unsatisfactory specimen
   a specimen which cannot be tested – no results other than the reason the specimen could not be tested will be reported

urea cycle disorder
   an inherited disorder caused by a deficiency of one of the enzymes in the urea cycle which allows ammonia to build up in the blood where it can cause irreversible brain damage, coma and death.
   of the screened disorders, citrullinemia and argininosuccinic acidemia are urea cycle disorders

U.S. Department of Health and Human Services (HHS)
   the federal government’s principal agency for protecting the health of all Americans and providing essential human services,
   http://www.hhs.gov

V

very low birth weight (VLBW)
   birth weight less than 1500 g

VLCAD
   very long-chain acyl-CoA dehydrogenase deficiency, a core disorder of the original Recommended Uniform Screening panel

W

WAL
   within acceptable limits

WNL
   within normal limits