

CP 1.1 Glossary - Newborn Screening & GDSP Terms & Abbreviations

Term/Abbreviation	Definition
AAP	American Academy of Pediatrics
Acquired hypothyroidism	Hypothyroidism that develops after birth
Adequate specimen	A specimen that has been collected according to the instructions accompanying the collection form, that reaches the laboratory with proper identification within the specified time limit after collection, and that will yield acceptable results.
AFP	Alpha FetoProtein
Allele	One of the variant forms of a gene at a particular locus (or location) on a chromosome. Different alleles produce variation in inherited characteristics, such as hair color or blood type. In an individual, one form of the allele (the dominant one) may be expressed more than the other form (the recessive one).
Amino Acids	A building block of proteins. There are 20 different kinds of naturally occurring amino acids.
ASC	Area Service Centers – an institution, corporation, hospital or university medical center which has contracted with GDB to provide follow-up and referral or screen-positive babies and education, consultation and technical assistance to providers in the region. Prior to July 1, 2000, these centers were called Area Genetic Centers (AGCs).
BH4	Tetrahydrobiopterin, enzyme cofactor for PAH
Biopterin Defects	Defects in phenylalanine metabolism involving the production and utilization of tetrahydrobiopterin (BH4). BH4 is the cofactor of phenylalanine hydroxylase in converting phenylalanine to tyrosine.
Borderline	A result that is not classified as positive, but is near the cut-off for a positive. The California NBS Program requires follow-up on newborn with positive results. Follow-up on infants with borderline values is up to the professional judgment of the health care providers based upon family history, age at collection, and any clinical symptoms.
Business Objects (BO)	The computer program which collects and houses elements of SIS and is used to develop reports.
C of C	Confirmation of Contact – entered by the NAPS lab or GDL into SIS when results of a newborn screen are completed and results are available.
CAP	Corrective Action Plan
Carrier	An individual who possesses one copy of a mutant allele that causes

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	disease only when two copies are present. Although carriers are not affected by the disease, two carriers can produce a child who has the disease.
CDHS	CA Department of Health Services
CHDP	Child Health and Disability Prevention Program - overseen by CMS. (See Section 2.11 for description of services).
CMS/CCS	California Medical Services/California Children's Services (See Section 2.11 for description of services)
Chromosome	A structure found in the cell nucleus that contains the genes; chromosomes are composed of DNA and proteins. Each parent contributes one chromosome to each pair, so children get half of their chromosomes from their mothers and half from their fathers.
Classical Galactosemia (GG)	Caused by a deficiency of the transferase enzyme called Galactose-1-phosphate uridyl transferase (GALT) . In severe deficiency where GALT is absent, galactose level increases in the blood, Gal-1-phosphate increases in the tissues, and excess galactose is excreted in the urine. Symptoms associated with galactosemia include failure to thrive, vomiting, diarrhea, hepatomegaly, jaundice, sepsis, and retardation.
Classical PKU	Persistent deficiency of the enzyme phenylalanine hydroxylase leading to developmental delay if untreated. Confirmed by level of phenylalanine in blood and affected siblings or relatives
CLIA	Clinical Laboratory Improvement Amendments
Congenital	Any trait or condition that exists from birth
Congenital hypothyroidism	Hypothyroidism present at birth
CPS	Child Protective Services
Cut-off level	The analytical value or test result that establishes the dividing line between results that are classified as negative and those that are deemed positive.
DBS	Dried Blood Spot
Demo	Demographic portion of the Test request form
Deoxyribonucleic Acid (DNA)	The substance of heredity; a large molecule that carries the genetic information necessary for all cellular functions, including the building of proteins. DNA is composed of the sugar deoxyribose, phosphate, and the bases adenine, thymine, guanine, and cytosine.
DHPR	Dihydropteridine reductase, enzyme involved in the recycling of BH4
Diagnostic test	A test that is used to establish a definitive diagnosis of some condition in an affected newborn.
Dominant	A gene that almost always results in a specific physical characteristic even though the genetic material only has one copy. The chance of passing on a dominant gene is 50 –50 in each pregnancy.
Duarte Galactosemia (DG)	The combination of the Duarte allele and the classical galactosemia allele conferring 25% GALT enzyme activity. Felt by most clinicians to be benign, but some cite evidence that there is some clinical

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	symptomatology.
Early Collection	Newborn Specimen that is collected prior to 12 hours of age, requires second specimen be collected.
Early Discharge	The release of a newborn from the facility of birth at less than 48 hours of age.
Enzyme	A protein that encourages a biochemical reaction, usually speeding it up.
EPSDT	Early and Periodic Screening, Diagnosis and Treatment Program.
Frequency	The rate at which a disorder occurs in some defined population. In newborn screening it is the same as birth prevalence and is computed by dividing the total births and still births by the number of cases, giving a one per "X" number of births
Gal-1-p	A metabolite of galactose metabolism that accumulates as a result of classical galactosemia
GALT	Galactose 1 phosphate uridyl transferase, an enzyme involved in the metabolism of galactose. It is the enzyme deficient in Classical Galactosemia.
GDSP	Genetic Disease Screening Program
GDL	Genetic Disease Laboratory
GDPS	Genetic Disease Project Specialist
Gene	The functional and physical unit of heredity passed from parent to offspring. Genes are pieces of DNA, and most genes contain the information for making a specific protein.
Genetic Code (ATGC)	The language in which DNA's information is written. It consists of triplets of nucleotides, with each triplet corresponding to one amino acid in a protein's structure or to a signal to start or stop protein production.
Genetic Counseling	A short-term educational counseling process for individuals and families who have a genetic disease or who are at risk for such a disease. Genetic counseling provides patients (or parents) with information about their condition (or their risk for having a subsequent pregnancy with a genetic condition) and in order for them to make make informed decisions.
Genetic screening	Testing a population group to identify a subset of individuals at high risk for having or transmitting a specific genetic disorder.
Genotype	The genetic identity of an individual that does not show as outward characteristics.
GTPCH	Guanosine triphosphate-cyclohydrolase, one of the enzymes in BH4 synthesis
Guthrie test	Bacterial inhibition assay that can also be used in NBS for PKU.
HEPP Report	Hospital Evaluation Performance Profile – a report generated in BO and sent to hospitals to report their proficiency in meeting NBS requirements (completing TRFs, adequacy of specimens, etc).
Heterozygous	Possessing of two different forms of a particular gene, one inherited from each parent.

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HIPAA	Health Insurance Portability and Accountability Act
Homozygous	Possessing of two identical forms of a particular gene, one inherited from each parent.
Hormone	A “messenger” molecule of the body that helps coordinate the actions of various tissues; it is made in one part of the body and transported via the bloodstream, to other parts, where it has a specific effect on cells.
HRSA	Health Resources and Services Administration – Federal agency that provides funding for research.
Hyperphenylalaninemia	A condition characterized by an elevation of phenylalanine levels in the blood.
IIP	Newborn screening pamphlet given to parents called I mportant I nformation for P arents about the Newborn Screening Test.
Inadequate specimen	A specimen that has been improperly collected, improperly handled and/or transported to the laboratory such that the specimen is determined by the lab to be unsuitable in quality or quantity to perform NBS for one or more of the disorders.
Incidence	The probability of developing a new case of a disease over a specified time period among people who did not have the disease the beginning of the time interval. The true occurrence rate of a disease over the entire period of time that the disease could develop. Should not be used in newborn screening.
Inherited	Transmitted through genes from parents to offspring
Initial Positive Test	Newborn screening result from the initial specimen defined as positive for reporting and follow-up.
Initial Specimen	The first specimen collected for the newborn screening program.
Initial test	The first valid analysis of a newborn screening specimen or sample for the mandated tests.
Karyotype	The chromosomal complement of an individual, including the number of chromosomes and any abnormalities. The term is also used to refer to a photograph of an individual’s chromosomes.
Metabolism	The process by which the body breaks down more complex molecules into simpler molecules to facilitate their use, or to eliminate them.
Modified McCaman-Robins	Automated flourometric analytical method used in NBS for PKU in California.

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MPKU	Maternal PKU, a condition in which high levels of phenylalanine in pregnant women with PKU can put their newborns at risk of having disabilities.
MS/MS	Tandem Mass Spectrometry
Mutation	A permanent structural alteration in DNA. In most cases, DNA changes either have no effect or cause harm, but occasionally a mutation can improve an organism's chance of surviving and passing the beneficial change on to its descendants.
NAPS Labs	Newborn and Prenatal Screening Laboratory
NBS	Newborn screening
NBS-DC	Newborn Screening Diagnosed Case report
NBS-MR	Newborn Screening Missing Results, used to report missing NBS results.
NBS-NO	Newborn Screening Not Obtained – form used to report that no NBS specimen was obtained.
NBS-OH	Newborn Screening Out-of-Hospital – form to report an NBS specimen from a birth that occurred outside of the hospital setting
NBSP	Newborn Screening Program
NBSB	Newborn Screening Branch
NBS-TR	Newborn Screening Test Refusal, used to report refusal to participate in the newborn screening program.
Negative predictive value	The probability that a child with a negative test is truly negative (i.e. not affected by the disease).
Negative Test	A laboratory result on a satisfactory specimen which is designated as having insufficient risk for disease to justify follow-up action.
Newborn Screening Test	A laboratory or clinical procedure performed on a population of newborns to detect those at sufficiently increased risk for a particular condition to justify follow-up actions and diagnostic test procedures.
PAH	Phenylalanine hydrozylase, enzyme involved in the conversion of phenylalanine to tyrosine.
Panel Count	Number of initial specimens drawn on newborns/number of newborns drawn.
Panic Values	Positive test results requiring immediate follow-up, situation may be life-threatening, i.e. necessitates immediate notification of those responsible for the newborns care.

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PCH	Primary Congenital Hypothyroidism
PDC	Prenatal Diagnosis Center
PDEB	Program Development and Evaluation Branch of the GDSP
Phe	Short for phenylalanine (pronounced “fee”)
Phenotype	The observable traits or characteristics of an organism, for example, hair color, weight, or the presence or absence of disease.
PKU	Phenylketonuria.
Polymorphism	A common variation in the gene sequence of DNA among individuals.
Positive predictive value	The probability that a newborn with a positive test will have the disease. Refers to the percent of total referrals, i.e. the positives that are affected.
Positive Test	A laboratory result on a satisfactory specimen which is designated as having a high risk for a disease requiring a follow-up action.
Prevalence	The rate of occurrence of a specific disorder at a given point in time per a designated denominator (i.e. 5 per 10,000). In newborn screening this is birth prevalence.
Primary congenital hypothyroidism	A disorder of thyroid function present in utero and at birth. Caused by failure of thyroid gland development reducing thyroid hormone secretion and consequently causing a deficiency in circulating thyroxine. Primary denotes abnormalities directly affecting the thyroid’s ability to secrete active thyroid hormones (T4 or T3). Thyroid hormone deficiency can result from absence of the thyroid gland, incomplete gland development in utero, thyroid inflammation from autoimmune disease, hereditary defects in thyroid hormone synthesis, or inability to synthesize thyroxine because of dietary iodine deficiency during pregnancy.
PTPS	6-pyruvoyltetrahydropterin synthase, one of the enzymes involved in BH4 synthesis
Recall (confirmatory) test	A test performed on a recall specimen to determine the validity of a previous positive test. Recommendations from NBS standardization workshop suggested that the term “confirmatory” not be used.
Recall Positive	A positive laboratory result that is obtained from analysis of a recall specimen (disease confirmed).
Recall Specimen	A specimen collected from a newborn following the newborn screening laboratory’s report that the initial collected specimen was positive for any of the disorders screened. A recall specimen is a redrawn specimen and is tested only for those analytes positive in the initial test.
Recessive	A genetic condition that appears only in individuals who have received two copies of a mutant gene, one from each parent.

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Repeat Specimen	A specimen collected from a newborn following the newborn screening laboratory's report that the initial collected specimen was unsatisfactory (inadequate), or that the test results were inconclusive, too early, or otherwise unreliable. The repeat specimen is a redrawn sample and is tested for all analytes.
Result Mailer	Newborn screening laboratory result mailed to the baby's pediatrician and to the baby's hospital of birth.
Retest	An analysis performed on another sample of the same specimen analyzed in the initial test.
Ribonucleic Acid (RNA)	A single stranded molecule that carries genetic information. RNA is composed of the sugar ribose, phosphate, and the bases adenine, uracil, guanine and cytosine.
Sample	A part of a specimen taken that is representative of the whole specimen at a specific point in time. A sample is representative of all of the blood or urine, etc., in the specimen at the time of collection, and since the entire specimen is seldom used up in the test, a sample can also refer to the portion of the specimen used in the test (synonym – aliquot).
SCID	Severe Combined Immunodeficiency
Screening	The attempted identification initiated by the provider of services of an as yet undetected disease or condition by the use of tests, exams, or other procedures which can be performed on at risk populations. These tests sort out apparently well persons who have a sufficiently high probability of having the disease/condition to justify subsequent diagnostic tests or procedures. A screening test is not designed to be diagnostic. Persons with positive or suspicious findings must be referred for diagnosis and necessary treatment.
Sensitivity	The probability that a newborn with disease will have a positive test result, a true positive rate.
Sex Chromosomes	One of the two chromosomes that specify an organism's genetic sex. Humans have two kinds of sex chromosomes, one called X and the other Y. Sex chromosome disorders occur when the number of sex chromosomes varies from XX or XY (i.e. XO, or XXY).
SIS	Screening Information System – computer system used to track progress of specimens in both the NBS and the XAFP programs.
Specificity	The probability that a newborn without the disease will have a negative test, the true negative rate.
Specimen	A fluid or tissue collected from the newborn to be tested (e.g. blood, urine, stool, sputum).
Specimen Collection Card	The filter paper card on which the dried blood spot specimen is collected.

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Stale Dated List	List of initial positives and inadequate specimens that have not been resolved after a specified period of time (depending on the disorder) i.e., 15 days, or 30 days
T3	Triiodothyronine, a form of the thyroid hormone that is found in tissues
T4	Thyroxine, the hormone secreted by the thyroid gland
TBG	T hyroid B inding G lobulin, the protein that transports T4 in the bloodstream
Test	A procedure or analysis used to determine the presence or absence and/or the quantity of a substance of interest (analyte).
TG	T hyro g lobulin, a protein precursor of T4 and T3.
Transient Hyperphenylalaninemia	An elevation that does not persist on repeated testing in the absence of specific intervention
TRF	T est R equ E st F orm, which includes all the demographic information on the infant screened.
TRH	T hyrotropin R eleasing H ormone, released by the hypothalamus to stimulate the pituitary gland to release TSH.
TSH	T hyroid S timulating H ormone, released by the pituitary to stimulate the thyroid gland to release T4; also known as Thyrotropin.
Validity (Analytical Validity)	The probability that test will be positive when a particular analyte is present and the probability that the test will be negative when the analyte is absent.
Variant PKU	Less severe enzyme deficiency leading to persistent but lower elevation of plasma phe levels. May not lead to mental retardation. May be benign or malignant, benign variants are not associated with symptoms while malignant variants are associated with symptoms. Also referred to as non-PKU hyperphenylalaninemias.
Very Early Discharge	The release of a newborn from the facility of birth at less than 24 hours of age.
WIC	Supplemental nutrition program for low income W omen, I nfants and C hildren.
XAFP	Expanded AFP