Newborn screening is the process of testing newborn babies for treatable genetic, endocrinologic, metabolic and hematologic diseases.[1][2] Robert Guthrie is given much of the credit for pioneering the earliest screening for phenylketonuria in the late 1960s using blood samples on filter paper obtained by pricking a newborn baby's heel on the second day of life to get a few drops of blood. [3] Congenital hypothyroidism was the second disease widely added in the 1970s.[4] The development of tandem mass spectrometry screening by Edwin Naylor and others in the early 1990s led to a large expansion of potentially detectable congenital metabolic diseases that affect blood levels of organic acids.[5] Additional tests have been added to many screening programs over the last two decades. Newborn screening has been adopted by most countries around the world, though the lists of screened diseases vary widely.

Contents

[edit] Disease qualification

Common considerations in determining whether to screen for disorders:

1. A disease that can be missed clinically at birth
2. A high enough frequency in the population
3. A delay in diagnosis will induce irreversible damages to the baby
4. A simple and reasonably reliable test exists
5. A treatment or intervention that makes a difference if the disease is detected early

[edit] Newborn Screening Program in the Philippines

The following tests are mandated in the R.A. 9288 or Newborn Screening program of 2004.Newborn screening is available in practicing health institutions (hospitals, lying-ins, Rural Health Units and Health Centers) with cooperation with DOH. If babies are delivered at

home, babies may be brought to the nearest institution offering newborn screening. A negative screen mean that the result of the test is normal and the baby is not suffering from any of the disorders being screened. In case of a positive screen, the NBS nurse coordinator will immediately inform the coordinator of the institution where the sample was collected for recall of patients for confirmatory testing. Babies with positive results should be referred at once to the nearest hospital or specialist for confirmatory test and further management. Should there be no specialist in the area, the NBS secretariat office will assist its attending physician. Disorders Screened:

Heel Prick Method for the newborn screening

- CH (Congenital hypothyroidism) - is a condition of thyroid hormone deficiency present at birth. Approximately 1 in 4000 newborn infants has a severe deficiency of thyroid function, while even more have mild or partial degrees. If untreated for several months after birth, severe congenital hypothyroidism can lead to growth failure and permanent mental retardation. Treatment consists of a daily dose of thyroid hormone (thyroxine) by mouth. Because the treatment is simple, effective, and inexpensive, nearly all of the developed world practices newborn screening to detect and treat congenital hypothyroidism in the first weeks of life.
- CAH (Congenital adrenal hyperplasia) - refers to any of several autosomal recessive diseases resulting from mutations of genes for enzymes mediating the biochemical steps of production of cortisol from cholesterol by the adrenal glands (steroidogenesis). Most of these conditions involve excessive or deficient production of sex steroids and can alter development of primary or secondary sex characteristics in some affected infants, children, or adults. Approximately 95% of cases of CAH are due to 21-hydroxylase deficiency.
- GAL (Galactosemia) - is a rare genetic metabolic disorder which affects an individual's ability to properly metabolize the sugar galactose. Lactose in food (such as dairy products) is broken down by the body into glucose and galactose. In individuals with galactosemia, the enzymes needed for further metabolism of galactose are severely diminished or missing entirely, leading to toxic levels of galactose to build up in the blood, resulting in hepatomegaly (an enlarged liver), cirrhosis, renal failure, cataracts, and brain damage. Without treatment, mortality in infants with galactosemia is about 75%.
- PKU (Phenylketonuria) - is an autosomal recessive genetic disorder characterized by a deficiency in the enzyme phenylalanine hydroxylase (PAH). This enzyme is necessary to metabolize the amino acid phenylalanine to the amino acid tyrosine. When PAH is deficient, phenylalanine accumulates and is converted into phenylpyruvate (also known as phenylketone), which is detected in the urine. PAH is found on chromosome number 12.Left untreated, this condition can cause problems with brain development, leading to progressive mental retardation and seizures. However, PKU is one of the few genetic diseases that can be controlled by diet. A diet low in phenylalanine and high in tyrosine can be a very effective treatment. There is no cure. Damage done is irreversible so early detection is crucial.

- G6PD Deficiency - is an X-linked recessive hereditary disease characterized by abnormally low levels of the glucose-6-phosphate dehydrogenase enzyme (abbreviated G6PD or G6PDH). It is a metabolic enzyme involved in the pentose phosphate pathway, especially important in red blood cell metabolism.
- Newborn screening results are available within three weeks after the NBS Lab receives and tests the samples sent by the institutions. Results are released by NBS Lab to the institutions and are released to your attending birth attendants or physicians.Parents may seek the results from the institutions where samples are collected. Christian Nieto,EACSN

[edit] Newborn screening in the United States

The following tests are mandated (required to be performed on every newborn born in the state) in most of the United States. According to the U.S. Centers for Disease Control, approximately 3,000 babies with severe disorders are identified in the United States each year using newborn screening programs at current testing rates. States vary, and not all tests are required in every state, and a few states mandate more than this. The first test to be universally mandated across the U.S. was the Guthrie test for phenylketonuria (PKU), and in many areas and hospitals, the newborn blood test is often erroneously referred to as a "PKU test", even though all states now universally test for congenital hypothyroidism, galactosemia, and increasing numbers of other diseases as well.

- Endocrine disorders: Congenital adrenal hyperplasia (CAH), Congenital hypothyroidism
- Blood cell disorders: sickle-cell disease (SS)
- Inborn errors of carbohydrate metabolism: Galactosemia
- Inborn errors of amino acid metabolism: Phenylketonuria (PKU), Maple syrup urine disease (MSUD), Homocystinuria
- Inborn errors of organic acid metabolism: Biotinidase deficiency

For a recent state-by-state list, see U.S. National Newborn Screening and Genetics Resource Center. According to this resource, the only tests mandated in every state are the following:

- CH - Congenital hypothyroidism
- H-HPE - Benign hyperphenylalaninemia
- PKU -- Phenylketonuria/hyperphenylalaninemia
- HEAR - Hearing
- GALT - Transferase deficient galactosemia

[edit] Usual procedures and responses to positive results

Heel blood on a filter paper card for the newborn screening

In nearly all of the United States, the newborn screening program is a division of the state health department. State law mandates collecting a sample by pricking the heel of a newborn baby to get enough blood (typically, two to three drops) to fill a few circles on filter paper labeled with names of infant, parent, hospital, and primary physician. It is usually specified that the sample be obtained on the second or third day of life, after protein-containing feedings (i.e., breast milk or formula) have started, and the postnatal TSH surge subsided. Every hospital in the state as well as independent midwives supervising home deliveries are required to collect the papers and mail each batch each day to the central laboratory.

The state health department agency in charge of screening will either run a laboratory or contract with a laboratory to run the mandated screening tests on the filter paper samples. The goal is to report the results within a short period of time. If screens are normal, a paper report is sent to the submitting hospital and parents rarely hear about it.

If an abnormality occurs, employees of the agency, usually nurses, begin to try to reach the physician, hospital, and/or nursery by telephone. They are persistent until they can arrange an evaluation of the infant by an appropriate specialist physician (depending on the disease). The specialist will attempt to confirm the diagnosis by repeating the tests by a different method or laboratory, or by performing other corroboratory tests. Depending on the likelihood of the diagnosis and the risk of delay, the specialist will initiate treatment and provide information to the family. Performance of the program is reviewed regularly and strenuous efforts are made to maintain a system that catches every infant with these diagnoses. Guidelines for newborn screening and follow up have been published by the American Academy of Pediatrics.[6]

[edit] Expanded screening and controversies

With the development of tandem mass spectrometry in the early 1990s, the number of detectable diseases quickly grew, especially in the categories of fatty acid oxidation disorders and organic acidoses. Screening tests for the disorders listed below (and an increasing number of others) are now available, though not universally mandated. There is considerable variability from state to state, and sometimes from hospital to hospital within a state, on disease that are screened. To make matters more confusing, some hospitals routinely obtain supplemental screening (most of the tests below) on all infants even if not mandated by the state or requested by parents. In recent years in the United States, expanded newborn screening with tandem mass spectrometry has become a profitable commercial venture.

Newborn screening tests have become a subject of political controversy in the last decade. Two California babies, Zachary Wyvill and Zachary Black, were both born with Glutaric acidemia type I. Wyvill's birth hospital only tested for the four diseases mandated by state law, while Black was born at a hospital that was participating in an expanded testing pilot program. Black's disease was treated with diet and vitamins; Wyvill's disease went undetected for over six months, and during that time the damage from the enzyme deficiency became irreversible. Birth-defects lobbyists pushing for broader and more universal standards for newborn testing cite this as an example of how much of an impact testing can have.

Instituting MS/MS screening often requires a sizable up front expenditure. When states choose to run their own programs the initial costs for equipment, training and new staff can be significant. To avoid at least a portion of the up front costs, some states such as Mississippi have chosen to contract with private labs for expanded screening. Others have chosen to form Regional Partnerships sharing both costs and resources. But for many states, screening is an integrated part of the department of health which can not or will not be easily replaced. Thus

the initial expenditures can be difficult for states with tight budgets to justify. Screening fees have also increased in recent years as healthcare costs rise and more states add MS/MS screening to their programs. (See Report of Summation of Fees Charged for Newborn Screening, 2001–2005) Dollars spent for these programs may reduce resources available to other potentially lifesaving programs. It has been recommended that one disorder, Short Chain Acyl-coenzyme A Dehydrogenase Deficiency, or SCAD, be eliminated from screening programs, due to a "spurious association between SCAD and symptoms. [7] However, recent studies suggest that expanded screening is cost effective (see ACMG report page 94-95 and articles published in *Pediatrics* [8],[9]. Advocates are quick to point out studies such as these when trying to convince state legislatures to mandate expanded screening.

Expanded newborn screening is also opposed by among some health care providers who are concerned that effective follow-up and treatment may not be available, that false positive screening tests may cause harm, and issues of informed consent[10].

[edit] Conditions and disorders

The following list includes most of the disorders detected by the expanded or supplemental newborn screening by mass spectrometry. This expanded screening is not yet universally mandated by most states, but may be privated purchased by parents or hospitals at a cost of approximately US$80. Perhaps one in 5,000 infants will be positive for one of the metabolic tests below (excluding the congenital infections).

The 29 marked with a "@" were recommended as "core panel" by the 2005 report of the American College of Medical Genetics (ACMG). The incidences reported below are from their report, pages 143-307, though the rates may vary in different populations. (WARNING: The file is a very large PDF.)

[edit] Blood cell disorders

- Glucose-6-phosphate dehydrogenase deficiency (G6PD)
- Sickle cell anemia (Hb SS) > 1 in 5,000; among African-Americans 1 in 400
- Sickle-cell disease (Hb S/C) > 1 in 25,000
- Hb S/Beta-Thalassemia (Hb S/Th) > 1 in 50,000

[edit] Inborn errors of amino acid metabolism

- Tyrosinemia I (TYR I) < 1 in 100,000
- Tyrosinemia II
- Argininemia
- Argininosuccinic aciduria (ASA) < 1 in 100,000
- Citrullinemia (CIT) < 1 in 100,000
- Phenylketonuria (PKU) > 1 in 25,000
- Maple syrup urine disease (MSUD) < 1 in 100,000
- Homocystinuria (HCY) < 1 in 100,000

[edit] Inborn errors of organic acid metabolism

- Glutaric acidemia type I (GA I) > 1 in 75,000
- Glutaric acidemia type II
- HHH syndrome (Hyperammonemia, hyperornithinemia, homocitrullinuria syndrome)

- Hydroxymethylglutaryl lyase deficiency (HMG) < 1 in 100,000
- Isovaleric acidemia (IVA) < 1 in 100,000
- Isobutyryl-CoA dehydrogenase deficiency
- 2-Methylbutyryl-CoA dehydrogenase deficiency
- 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC) > 1 in 75,000
- Beta-methyl crotonyl carboxylase deficiency
- 3-Methylglutaconyl-CoA hydratase deficiency
- Methylmalonyl-CoA mutase deficiency (MUT) > 1 in 75,000
- Methylmalonic aciduria, cblA and cblB forms (MMA, Cbl A,B) < 1 in 100,000
- Beta-ketothiolase deficiency (BKT) < 1 in 100,000
- Propionic acidemia (PROP) > 1 in 75,000
- Adenosylcobalamin synthesis defects
- Multiple-CoA carboxylase deficiency (MCD) < 1 in 100,000

[edit] Inborn errors of fatty acid metabolism

- Carnitine palmityl transferase deficiency type 2 (CPT)
- Long-chain acyl-CoA dehydrogenase deficiency (LCAD)
- Long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD) > 1 in 75,000
- Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
- Short-chain hydroxy Acyl-CoA dehydrogenase deficiency (SCHAD)
- Medium-chain acyl-CoA dehydrogenase deficiency (MCAD) > 1 in 25,000
- Very-long-chain acyl-CoA dehydrogenase deficiency (VLCAD) > 1 in 75,000
- Carnitine/acylcarnitine Translocase Deficiency (Translocase)
- Multiple acyl-CoA dehydrogenase deficiency (MADD)
- Trifunctional protein deficiency (TFP) < 1 in 100,000
- Carnitine uptake defect (CUD) < 1 in 100,000

[edit] Congenital infections

- Congenital toxoplasmosis
- HIV

[edit] Miscellaneous multisystem diseases

- Cystic fibrosis (CF) > 1 in 5,000
- Maternal vitamin B12 deficiency
- Congenital hypothyroidism (CH) > 1 in 5,000
- Biotinidase deficiency (BIOT) > 1 in 75,000
- Congenital adrenal hyperplasia (CAH) > 1 in 25,000
- Classical galactosemia (GALT) > 1 in 50,000

[edit] Newborn screening by other methods than blood testing

- Congenital deafness (HEAR) > 1 in 5,000

[edit] Newborn screening programs worldwide

Newborn screening has also been adopted by most countries in Europe and around the world, though the lists of screened diseases vary widely.

[edit] See also

- Inborn error of metabolism
- Genetic disorder

[edit] References

1. ^ Tarini BA (2007). "The current revolution in newborn screening: new technology, old controversies". *Archives of pediatrics & adolescent medicine* **161** (8): 767–72. doi:10.1001/archpedi.161.8.767. PMID 17679658.
2. ^ Kayton A (2007). "Newborn screening: a literature review". *Neonatal network : NN* **26** (2): 85–95. PMID 17402600.
3. ^ Clague A, Thomas A (2002). "Neonatal biochemical screening for disease". *Clin. Chim. Acta* **315** (1-2): 99–110. doi:10.1016/S0009-8981(01)00716-1. PMID 11728413.
4. ^ Klein AH, Agustin AV, Foley TP (1974). "Successful laboratory screening for congenital hypothyroidism". *Lancet* **2** (7872): 77–9. doi:10.1016/S0140-6736(74)91637-7. PMID 4137217.
5. ^ Chace DH, Kalas TA, Naylor EW (2003). "Use of tandem mass spectrometry for multianalyte screening of dried blood specimens from newborns". *Clin. Chem.* **49** (11): 1797–817. doi:10.1373/clinchem.2003.022178. PMID 14578311.
6. ^ Newborn Screening Expands: Recommendations for Pediatricians and Medical Homes-Implications for the System. Pediatrics121:1 192-217 January 2008[1]
7. ^ Newborn Screening for Metabolic Disorders. Journal of the American Medical Association 2006 PMID 16926360
8. ^ Expanded Newborn Screening for Inborn Errors of Metabolism by Electrospray Ionization-Tandem Mass Spectrometry: Results, Outcome and Implication PMID 12777559 [2]<
9. ^ Cost-Benefit Analysis of Universal Tandem Mass Spectrometry for Newborn Screening. Pediateics 2002 PMID 12359795 [3]
10. ^ Financial, Ethical, Legal, and Social Issues

[edit] External links

- U.S. National Newborn Screening and Genetics Resource Center
- About Newborn Screening
- Baily, M.A. and Murray, T.H. (2009).*Ethics and Newborn Genetic Screening*. Johns Hopkins University Press. ISBN 9780801891519
- PerkinElmer Genetics, Inc. (Commercial company that pioneered some of the screening procedures and offers testing directly to parents. Excellent set of links to other sites about metabolic diseases and screening.)
- Waldholz, Michael, "A Drop of Blood Saves One Baby; Another Falls Ill," *Wall Street Journal*, 17 June 2001, p. A1 (52k PDF)
- March of Dimes, The Difference is Black and Wyvill, 2004
- Save Babies Through Screening Foundation
- Organic Acidemia Association
- $Millions saved nationally by newborn screening per Delaware DPH
- The New England Consortium of Metabolic Programs
- **Newborn Screening Program** *for Mumbai, Navi-Mumbai and Thane*

Categories: Pediatrics | Inborn errors of metabolism | Epidemiology | Rare diseases

NNSGRC

National Newborn Screening & Genetics Resource Center

Contact Us | Events | Parents/Families | Professionals | NNSIS

Welcome to the National Newborn Screening and Genetics Resource Center web site: GeNeS-R-US, (Genetic and Newborn Screening Resource Center of the United States).

Health Improvement Institute
AESCULAPIUS AWARD
WWW SITE
2008 Award of Excellence

The National Newborn Screening and Genetics Resource Center (NNSGRC) is a cooperative agreement between the Maternal and Child Health Bureau (MCHB), Genetic Services Branch and the University of Texas Health Science Center at San Antonio (UTHSCSA), Department of Pediatrics.

We provide information and resources in the area of newborn screening and genetics to benefit health professionals, the public health community, consumers and government officials.

Search Search our site

Links of Special Interest

Papers and Reports----------

NEW **Message Board:** A discussion forum for consumers of newborn screening services including healthcare workers, parents, and others affiliated with newborn screening programs.

NEW **"The Story of Newborn Screening" by Harvey Levy, MD**: A 10-part webcast from the New England Regional Consortium describing the basics of newborn screening (may be viewed in its entirety or in individual segments).

NEW **"The President's council on Bioethics: The Changing Moral Focus of Newborn Screening: An Ethical Analysis"** (2009)

SACGHS Report: Oversight of Genetic Testing (2008)

ACT Sheets (ACMG)

ACMG Newborn Screening Report (2006) **- Executive Summary**

Fact Sheets **and Introduction** (AAP)

AAP Blueprint for Newborn Screening (2000)

Related Links----------

NEW **Newborn Screening Use Case** Use case documents developed by ONC
-Draft Use Case
Draft Resource Guide
Resource Database a web-based tool to allow the review of proposed standards for newborn screening condition and analyte terminology,

- Executive Summary

Screening Programs----------

US Newborn Screening Programs (clickable map)

US Genetics Programs (clickable map)

Regional Collaboratives (clickable map)

NBS Program Contact Information

Conditions screened by US programs
HTML, MS-Word, PDF
Maps of conditions screened (MOD)

Conditions screened by Canadian programs
PDF

codes, and mapping.

Brochure for Parents: Model developed for state use based on parent focus groups

Brochure for Providers: Model developed for state use based on provider focus groups

Brochure for Grandparents

Foreign Language Educational Materials

Laboratory Services: Additional non-state newborn screening laboratory services

ACHDNC: Secretary's Advisory Committee on Heritable Disorders in Newborns and Children

SACGHS: Secretary's Advisory Committee on Genetics,Health and Society

NNSIS: National Newborn Screening Information System: Program Information and Data from 2001 to Present

Copies of Written Reports 1996 1999 1997 2000 1998

PEAS: Performance Evaluation and Assessment Scheme

U.S. National Library of

Newborn Screening Coding
Terminology G

Data Standards for Electronic Re

The goal of the Newborn Screening Coding and Terminology Guide is to promote and facilitate the use of electronic health data standards in recording and transmitting newborn screening test results. The Web site includes standard codes and terminology for newborn tests and the conditions for which they screen, and links to other related sites. The codes and vocabulary standards are provided in a series of tables that you can view on the Web and/or download for your own use. These tables cover conditions recommended for screening by the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) or by a state within the U.S.

Use of these standards can speed the delivery of newborn screening reports, facilitate the care and follow-up of infants with positive test results, enable the use (and comparison) of data from different laboratories, and support the development of strategies for improving the newborn screening process.

Work is underway on guidance for creating an HL7 version 2.x message using these codes with examples, which will appear on this website in the future. If you would like us to notify you about this and other new content, please subscribe to the NBS-Announcements e-mail list from the U.S. National Library of Medicine.

You can reach these various resources by picking a choice below.

- Views: Generate customized Web views from the tables of conditions and analytes/measurements maintained by the U.S. National Library of Medicine (NLM®).
    - **Conditions** — Conditions that are targeted by newborn screening
    - **Analytes/Measurements** — Tests that are used as markers for newborn screening conditions
    - **Tailored Views** — Specify subsets, or see relationships between conditions and analytes/measurements
- Downloads: Download the tables of newborn screening conditions, of markers for these conditions and/or of mappings between conditions and their markers.
- Resources: Find additional information about newborn screening and related codes and data standards, including the Newborn Screening Draft Detailed Use Case that

was developed by the Office of the National Coordinator for Health Information Technology (ONC).

- Code and Terminology Standards: View terms of use and other information about codes and terminologies listed and referenced on this Web site, including Logical Observation Identifiers Names and Codes (LOINC®), Systematized Nomenclature of Medicine — Clinical Terms (SNOMED CT®), and others.
- About Us: Learn about the National Library of Medicine Lister Hill National Center for Biomedical Communications (LHNCBC) and gather more information about the Newborn Screening Coding and Terminology Guide.

Your questions and comments are welcome. Contact Us.

This graphic notice ( ) indicates a PDF document, best viewed with a PDF reader.
This graphic notice ( ) means the link leads to a Web site outside the domain of the US Government.
Site last updated: September 9, 2009

Analytes/Measurements View

The Analytes/Measurements View lists the analytes (chemical entities) and the hearing measurements that serve as markers for newborn screening conditions. This view includes the analytes you selected on a previous screen, a short name, and a Logical Observation Identifiers Names and Codes (LOINC®) Number that may be used in electronic laboratory reports. Click on an analyte name to view related conditions, or to see answer lists for measurements with categorical values. For measurements with numeric values, the view also includes the units of measurement.

Analyte Name Choose analyte to view related conditions	Analyte Short Name	LOINC Number	Units
Hearing			
Newborn hearing screen method	Hear-Meth	54106-0	Answer list
Newborn hearing screen of Ear - bilateral	Hear-Both	54107-8	Answer list
Newborn hearing screen of Ear - left	Hear-L	54108-6	Answer list

Analyte Name Choose analyte to view related conditions	Analyte Short Name	LOINC Number	Units
Newborn hearing screen of Ear - right	Hear-R	54109-4	Answer list
Hearing loss newborn screen interpretation		46770-4	
MS/MS			
Acyl-Carnitine			
Octenoylcarnitine (C8:1)	C8:1	53174-9	umol/L
Amino Acid			
5-Oxoproline+Pipecolate	OXOPRO + PIPA	53232-5	umol/L
5-Oxoproline+Pipecolate/Phenylalanine	[OXOPRO + PIPA] / PHE	53394-3	N/A
Alanine+Beta Alanine+Sarcosine	ALA + BALA + SARC	53150-9	umol/L
Alloisoleucine+Isoleucine+Leucine+Hydroxyproline	AILE + ILE + LEU + OHPRO	53152-5	umol/L
Alloisoleucine+Isoleucine+Leucine+Hydroxyproline/Alanine	[AILE + ILE + LEU + OHPRO] / ALA	53154-1	N/A
Alloisoleucine+Isoleucine+Leucine+Hydroxyproline/Phenylalanine	[AILE + ILE + LEU + OHPRO] / PHE	53153-3	N/A
Alloisoleucine+Isoleucine+Leucine+Hydroxyproline+Valine/Phenylalanine+Tyrosine	[AILE + ILE + LEU + OHPRO + VAL] / [PHE + TYR]	53393-5	N/A
Arginine	ARG	47562-4	umol/L
Arginine/Phenylalanine	ARG / PHE	53398-4	N/A
Argininosuccinate	ASA	53062-6	umol/L
Argininosuccinate/Arginine	ASA / ARG	53200-2	N/A
Asparagine+Ornithine	ASN + ORN	53155-8	umol/L
Asparagine+Ornithine/Phenylalanine	[ASN + ORN] / PHE	53396-8	N/A
Asparagine+Ornithine/Serine	[ASN + ORN] / SER	53395-0	N/A
Aspartate	ASP	4757	umol/

Analyte Name Choose analyte to view related conditions	Analyte Short Name	LOINC Number	Units
		3-1	L
Citrulline	CIT	42892-0	umol/L
Citrulline/Arginine	CIT/ARG	54092-2	N/A
Citrulline/Phenylalanine	CIT / PHE	53157-4	N/A
Citrulline/Tyrosine	CIT / TYR	53399-2	N/A
Glutamate	GLU	47623-4	umol/L
Glycine	GLY	47633-3	umol/L
Histidine	HIS	47643-2	umol/L
Homocitrulline	HOMOCIT	53158-2	umol/L
Lysine	LYS	47689-5	umol/L
Methionine	MET	47700-0	umol/L
Methionine/Alloisoleucine+Isoleucine+Leucine+Hydroxyproline	MET / [AILE + ILE + LEU + OHPRO]	53397-6	N/A
Methionine/Phenylalanine	MET / PHE	53156-6	N/A
Phenylalanine	PHE	29573-3	umol/L
Phenylalanine/Tyrosine	PHE / TYR	35572-7	N/A
Proline	PRO	47732-3	umol/L
Proline/Phenylalanine	PRO / PHE	53392-7	N/A
Serine	SER	47742-2	umol/L
Succinylacetone	SUAC	53231-7	umol/L
Threonine	THR	47784-4	umol/L
Tryptophan	TRP	53159-0	umol/L

Analyte Name Choose analyte to view related conditions	Analyte Short Name	LOINC Number	Units
Tyrosine	TYR	35571-9	umol/L
Valine	VAL	47799-2	umol/L
Valine/Phenylalanine	VAL/PHE	53151-7	N/A
Amino acidemias newborn screen interpretation		46733-2	
Fatty Acid Oxidase			
3-Hydroxybutyrylcarnitine (C4-OH)	C4OH	50102-3	umol/L
3-Hydroxydecenoylcarnitine (C10:1-OH)	C10:1OH	53182-2	umol/L
3-Hydroxydodecanoylcarnitine (C12-OH)	C12OH	53189-7	umol/L
3-Hydroxydodecenoylcarnitine (C12:1-OH)	C12:1OH	53188-9	umol/L
3-Hydroxyhexanoylcarnitine (C6-OH)	C6OH	53173-1	umol/L
3-Hydroxylinoleoylcarnitine (C18:2-OH)	C18:2OH	50109-8	umol/L
3-Hydroxyoleoylcarnitine (C18:1-OH)	C18:1OH	50113-0	umol/L
3-Hydroxypalmitoleylcarnitine (C16:1-OH)	C16:1OH	50121-3	umol/L
3-Hydroxypalmitoylcarnitine (C16-OH)	C16OH	50125-4	umol/L
3-Hydroxypalmitoylcarnitine (C16-OH)/Palmitoylcarnitine (C16)	C16OH / C16	53201-0	N/A
3-Hydroxystearoylcarnitine (C18-OH)	C18OH	50132-0	umol/L
3-Hydroxytetradecadienoylcarnitine (C14:2-OH)	C14:2OH	53196-2	umol/L
3-Hydroxytetradecanoylcarnitine (C14-OH)	C14OH	50281-5	umol/L
3-Hydroxytetradecenoylcarnitine (C14:1-OH)	C14:1OH	53197-0	umol/L
Carnitine.free (C0)+Acetylcarnitine (C2)+Propionylcarnitine (C3)+Palmitoylcarnitine (C16)+Oleoylcarnitine (C18:1)+Stearoylcarnitine (C18)/Citrulline	[C0 + C2 + C3 + C16 + C18:1 + C18] / CIT	53236-6	N/A
Carnitine free (C0)	C0	3848	umol/

Analyte Name Choose analyte to view related conditions	Analyte Short Name	LOINC Number	Units
		1-8	L
Carnitine free (C0)/Palmitoylcarnitine (C16)	C0 / C16	53233-3	N/A
Carnitine free (C0)/Palmityolcarnitine (C16)+Stearoylcarnitine (C18)	C0 / [C16 + C18]	53235-8	N/A
Carnitine free (C0)/Stearoylcarnitine (C18)	C0 / C18	53234-1	N/A
Decadienoylcarnitine (C10:2)	C10:2	53180-6	umol/L
Decanoylcarnitine (C10)	C10	45197-1	umol/L
Decenoylcarnitine (C10:1)	C10:1	45198-9	umol/L
Dodecanoylcarnitine (C12)	C12	45199-7	umol/L
Dodecenoylcarnitine (C12:1)	C12:1	45200-3	umol/L
Hexanoylcarnitine (C6)	C6	45211-0	umol/L
Linoleoylcarnitine (C18:2)	C18:2	45217-7	umol/L
Octanoylcarnitine (C8)	C8	53175-6	umol/L
Octanoylcarnitine (C8)/Acetylcarnitine (C2)	C8 / C2	53176-4	N/A
Octanoylcarnitine (C8)/Decanoylcarnitine (C10)	C8 / C10	53177-2	N/A
Oleoylcarnitine (C18:1)	C18:1	53202-8	umol/L
Palmitoleylcarnitine (C16:1)	C16:1	53198-8	umol/L
Palmitoylcarnitine (C16)	C16	53199-6	umol/L
Stearoylcarnitine (C18)	C18	53241-6	umol/L
Stearoylcarnitine (C18)/Propionylcarnitine (C3)	C18 / C3	53400-8	N/A
Tetradecadienoylcarnitine (C14:2)	C14:2	53190-5	umol/L
Tetradecanoylcarnitine (C14)	C14	53192-1	umol/L
Tetradecenoylcarnitine (C14:1)	C14:1	5319	umol/

Analyte Name Choose analyte to view related conditions	Analyte Short Name	LOINC Number	Units
		1-3	L
Tetradecenoylcarnitine (C14:1)/Acetylcarnitine (C2)	C14:1 / C2	53193-9	N/A
Tetradecenoylcarnitine (C14:1)/Dodecenoylcarnitine (C12:1)	C14:1 / C12:1	53194-7	N/A
Tetradecenoylcarnitine (C14:1)/Palmitoylcarnitine (C16)	C14:1 / C16	53195-4	N/A
Fatty acid oxidation defects newborn screen interpretation		46736-5	
Fatty Acid Oxidase-Organic Acid			
3-Hydroxyoctanoylcarnitine (C8-OH)+Malonylcarnitine (C3-DC)	C8OH + C3DC	53178-0	umol/L
3-Hydroxyoctanoylcarnitine (C8-OH)+Malonylcarnitine (C3-DC)/Butyrylcarnitine+Isobutyrylcarnitine (C4)	C8OH + C3DC / C4	53402-4	N/A
3-Hydroxyoctanoylcarnitine (C8-OH)+Malonylcarnitine (C3-DC)/Decanoylcarnitine (C10)	C8OH + C3DC / C10	53179-8	N/A
Acetylcarnitine (C2)	C2	50157-7	umol/L
Butyrylcarnitine+Isobutyrylcarnitine (C4)	C4	53166-5	umol/L
Butyrylcarnitine+Isobutyrylcarnitine (C4)/Acetylcarnitine (C2)	C4 / C2	53167-3	N/A
Butyrylcarnitine+Isobutyrylcarnitine (C4)/Octanoylcarnitine (C8)	C4 / C8	53169-9	N/A
Butyrylcarnitine+Isobutyrylcarnitine (C4)/Propionylcarnitine (C3)	C4 / C3	53168-1	N/A
Glutarylcarnitine (C5-DC)+3-Hydroxydecanoylcarnitine (C10-OH)	C5DC + C10OH	53183-0	umol/L
Glutarylcarnitine (C5-DC)+3-Hydroxydecanoylcarnitine (C10-OH)/3-Hydroxyisovalerylcarnitine (C5-OH)	C5DC + C10OH / C5OH	53184-8	N/A
Glutarylcarnitine (C5-DC)+3-Hydroxydecanoylcarnitine (C10-OH)/Butyrylcarnitine+Isobutyrylcarnitine (C4)	C5DC + C10OH / C4	53403-2	N/A
Glutarylcarnitine (C5-DC)+3-Hydroxydecanoylcarnitine (C10-OH)/Octanoylcarnitine (C8)	C5DC + C10OH / C8	53185-5	N/A
Glutarylcarnitine (C5-DC)+3-Hydroxydecanoylcarnitine (C10-OH)/Palmitoylcarnitine (C16)	C5DC + C10OH / C16	53186-3	N/A
Organic Acid			
3-Hydroxyisovalerylcarnitine (C5-OH)	C5OH	50106-4	umol/L
3-Hydroxyisovalerylcarnitine (C5-OH)/Carnitine.free (C0)	C5OH / C0	53171-5	N/A
3-Hydroxyisovalerylcarnitine (C5-OH)/Octanoylcarnitine	C5OH / C8	5317	N/A

Analyte Name Choose analyte to view related conditions	Analyte Short Name	LOINC Number	Units
(C8)		2-3	
Acrylylcarnitine (C3:1)	C3:1	53237-4	umol/L
Formiminoglutamate	FIGLU	53165-7	umol/L
Isovalerylcarnitine+Methylbutyrylcarnitine (C5)	C5	45216-9	umol/L
Isovalerylcarnitine+Methylbutyrylcarnitine (C5)/Acetylcarnitine (C2)	C5 / C2	53239-0	N/A
Isovalerylcarnitine+Methylbutyrylcarnitine (C5)/Carnitine.free (C0)	C5 / C0	53238-2	N/A
Isovalerylcarnitine+Methylbutyrylcarnitine (C5)/Octanoylcarnitine (C8)	C5 / C8	53401-6	N/A
Isovalerylcarnitine+Methylbutyrylcarnitine (C5)/Propionylcarnitine (C3)	C5 / C3	53240-8	N/A
Methylglutarylcarnitine (C6-DC)	C6DC	53187-1	umol/L
Methylmalonylcarnitine (C4-DC)	C4DC	45222-7	umol/L
Methylmalonylcarnitine (C4-DC)/3-Hydroxyisovalerylcarnitine (C5-OH)	C4DC / C5OH	53181-4	N/A
Propionylcarnitine (C3)	C3	53160-8	umol/L
Propionylcarnitine (C3)/Acetylcarnitine (C2)	C3 / C2	53163-2	N/A
Propionylcarnitine (C3)/Carnitine.free (C0)	C3 / C0	53162-4	N/A
Propionylcarnitine (C3)/Methionine	C3 / MET	53161-6	N/A
Propionylcarnitine (C3)/Palmitoylcarnitine (C16)	C3 / C16	53164-0	N/A
Tiglylcarnitine (C5:1)	C5:1	53170-7	umol/L
Organic acidemias newborn screen interpretation		46744-9	
Non-MS/MS			
Biotinidase			
Biotinidase	BIO	38478-4	%
Biotinidase deficiency newborn screen interpretation		46770-4	
Cystic Fibrosis			

Analyte Name Choose analyte to view related conditions	Analyte Short Name	LOINC Number	Units
CFTR gene mutations found	CFTR-Mut	54083-1	Specific alleles
DNA sequencing of the CFTR gene	CFTR-Seq		Specific mutations
Sweat chloride	Sweat-Cl	2077-6	mmol/L
Trypsinogen I Free	IRT	48633-2	umol/L
Cystic fibrosis newborn screen interpretation		46769-6	
Endocrine Disorders			
11-Deoxycorticosterone	11DOC	53347-1	ng/mL
11-Deoxycortisol	11DC	53338-0	ng/mL
17-Hydroxyprogesterone	17OHP	38473-5	ng/mL
17-Hydroxyprogesterone+Androstenedione/Cortisol	17OCHS+Androst/Cortis	53336-4	N/A
21-Deoxycortisol	21DC	53341-4	ng/mL
Androstenedione	Androst	53343-0	ng/mL
Cortisol	Cortis	53345-5	ng/mL
Thyrotropin	TSH	29575-8	mIU/L
Thyroxine	T4	31144-9	ng/dL
Congenital adrenal hyperplasia newborn screen interpretation		46758-9	
Primary congenital hypothyroidism newborn screen interpretation		46762-1	
Galactosemia			
Enzyme NADPH5	NADPH5		mg/dL
Total Galactose	Galact	54084-9	mg/dL
Galactosemia newborn screen interpretation		46737-3	

Analyte Name Choose analyte to view related conditions	Analyte Short Name	LOINC Number	Units
Hemoglobin Disorders			
Hemoglobin A/Hemoglobin.total	%Hb A	54072-4	%
Hemoglobin Barts/Hemoglobin.total	%Hb Barts	54069-0	%
Hemoglobin C/Hemoglobin.total	%Hb C	54073-2	%
Hemoglobin D/Hemoglobin.total	%Hb D	54070-8	%
Hemoglobin E/Hemoglobin.total	%Hb E	54071-6	%
Hemoglobin F/Hemoglobin.total	%Hb F	54074-0	%
Hemoglobin O - Arab/Hemoglobin.total	%Hb O-ARAB	54068-2	%
Hemoglobin pattern in Electrophoresis	Hb fract-IEF	54103-7	Answer list
Hemoglobin pattern in HPLC	Hb fract-HPLC	54104-5	Answer list
Hemoglobin pattern in Isoelectric focusing	Hb fract-Elph	54105-2	Answer list
Hemoglobin S/Hemoglobin.total	%Hb S	56476-5	%
Hemoglobin disorders newborn screen interpretation		46740-7	Answer list
Infectious Diseases			
HIV 1+2 IgG Ab	HIV IgG	54086-4	Pos or Neg
Toxoplasma gondii IgG Ab	Tox IgG	54087-2	Pos or Neg
Toxoplasma gondii IgM Ab	Tox IgM	54088-0	Pos or Neg
Other			
Glucose-6-Phosphate dehydrogenase	G6PD	33287-4	N/A

The above view reflects the criteria you selected on the previous screen.

The view includes derived measures.

Description of Analyte/Measurement Table

Analyte Name is the chemical whose concentration is being measured by the Dried Blood Spot testing, or the newborn hearing screening method.

Analyte Short Name is an abbreviation for the analyte.

LOINC Number is the unique and permanent code assigned by the Logical Observation Identifiers Names and Codes (LOINC®) Committee to identify the test measurement. LOINC codes are unique for different test methods and different units of reporting to enable interoperability and comparison of results from different labs. LOINC is a U.S. government standard for electronic health information exchange of laboratory tests and other measurements in Interoperability Specifications produced by the Healthcare Information Technology Standards Panel (HITSP).

Units: For quantitative tests with units of measure, the table lists the preferred reporting units. We use printable alphanumeric characters to avoid problems of interoperability that occur with Greek characters. Ratios and categorical variables do not have units of measure. The table shows UCUM units which have been adopted as the standard by the US for units reporting in Laboratory messages.

Ergebnisse **1** - **10** von ungefähr **1.020.000** für **"Newborn Screening" (16.9.09)**

- ***Newborn screening* - Wikipedia, the free encyclopedia**

- [Diese Seite übersetzen]

30 Jul 2009 **...** *Newborn screening* is the process of testing newborn babies for treatable genetic, endocrinologic, metabolic and hematologic diseases. **...**
*en.wikipedia.org/wiki/**Newborn_screening*** - Im Cache - Ähnlich

- ***Newborn Screening*: MedlinePlus**

- [Diese Seite übersetzen]

10 Jun 2009 **...** The primary NIH organization for research on *Newborn Screening* is the National Institute of Child Health and Human Development **...**

Overviews - Latest News - Specific Conditions - Related Issues

*www.nlm.nih.gov/.../**newbornscreening**.html* - Im Cache - Ähnlich

- ***Newborn Screening* Tests**

- [Diese Seite übersetzen]

Newborn screening tests look for harmful or potentially fatal disorders that aren't apparent at birth. Find out which tests are done and which disorders **...**
*kidshealth.org/.../**newborn_screening**_tests.html* - Im Cache - Ähnlich

- ***Newborn Screening* for endocrine and metabolic diseases**

Informationen der Deutschen Gesellschaft für das Neugeborenen-Screening auf endokrine und metabole Störungen sowie das Screening-Journal mit Newsletter.
www.neoscreening.de/ - Im Cache - Ähnlich

- ***Newborn Screening* News - *NewbornScreening*.Com**

- [Diese Seite übersetzen]

24 Apr 2005 **...** Regularly updated information and news pertaining to testing and screening. Explanation as to what it is, condition profiles, lab profiles **...**
*www.**newbornscreening**.com/* - Im Cache - Ähnlich

- **National *Newborn Screening* and Genetics Resource Center**

- [Diese Seite übersetzen]

Provides a large collection of resources on birth defects and on genetic/metabolic screening of infants as a component of public health.
genes-r-us.uthscsa.edu/ - Im Cache - Ähnlich

- **NHS Newborn Bloodspot Screening Programme Home Page**

- [Diese Seite übersetzen]

4 Aug 2009 **...** Welcome to the UK *Newborn Screening* Programme Centre. The UK *Newborn Screening* Programme Centre has responsibility for developing, **...**
newbornbloodspot.screening.nhs.uk/ - Im Cache - Ähnlich

- ***Newborn Screening* von PerkinElmer -**

Newborn screening is used to detect in babies congenital diseases that are treatable only when identified during the first days of life. **...**
las.perkinelmer.de/Catalog/default.htm?... - Im Cache - Ähnlich

- **State *newborn screening* programs advance, but most infants still ...**

Expanded *newborn screening* is now required by law in dozens of states, but most infants still are not covered by the full panel of 29 tests recommended by **...**
www.innovations-report.de/.../bericht-46475.html - Im Cache - Ähnlich

- ***NEWBORN SCREENING***

- [Diese Seite übersetzen]

21 Apr 2009 **...** *Newborn screening* programs are a collaborative effort between public health departments, hospitals, government agencies and the parents of **...**
*www.**newbornscreening**.info/* - Im Cache - Ähnlich