HUMAN HEALTH

ENVIRONMENTAL HEALTH

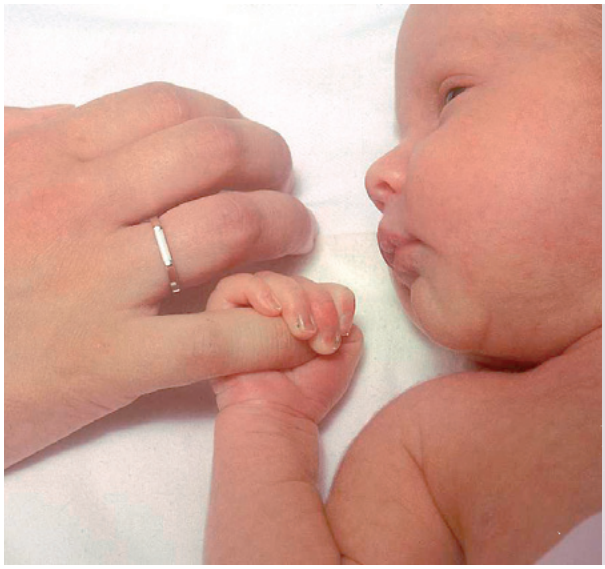
STEP FORWARD IN NEWBORN SCREENING

GSP® NEONATAL ASSAYS

for CH, PKU, biotinidase deficiency, galactosemia, CAH and CF screening

Brochure not for distribution in the USA

PerkinElmer®
For the Better



GSP® NEONATAL KITS

DELFIA® or enzyme-based fluorescence assays

GSP® is the new, automated neonatal screening system from PerkinElmer. It has multi-technology capability, which means that both DELFIA® and prompt fluorescence assays can now be run on a common platform. Its versatility and speed help it to accommodate present and future screening needs.

Kits available are

- **GSP Neonatal hTSH and GSP Neonatal Thyroxine (T4),** for congenital hypothyroidism (CH) screening
- **GSP Neonatal Phenylalanine,** for phenylketonuria screening
- **GSP Neonatal Biotinidase,** a time-resolved fluorescence assay for biotinidase deficiency screening
- **GSP Neonatal TGAL and GSP Neonatal GALT,** allowing automation in galactosemia screening
- **GSP Neonatal 17α-OH-progesterone,** a GSP version of the globally most widely used assay for 1st tier congenital adrenal hyperplasia (CAH)
- **GSP Neonatal IRT** for cystic fibrosis screening

Work with the leader in Newborn Screening

PerkinElmer is the global market leader in neonatal screening, currently serving customers in some 74 countries.

The company is a total solution provider offering complete systems based on a broad range of high quality, validated products, including newborn screening kits, consumables, instruments and software. Our global presence and comprehensive support philosophy mean that our expertise is available to you at all times.

55 babies saved every day

The first DELFIA neonatal kit was developed in 1985, to allow dried blood spot measurement of hTSH.

By 2013, some 415 million babies had been screened with PerkinElmer products. For every day of the year serious disorders are revealed in 55 babies so that treatment may be applied in time.

GSP NEONATAL ASSAYS

Reduced risk of error

All contents of GSP Neonatal kits, including reagents, plates, QC material and lot specific QC certificates are barcoded to reduce the risk of errors.

The robust assays are not affected by EDTA, citrate or heparin in samples.

Less work – faster results

To minimize the pre-analytical work, for most GSP assays, all reagents are ready to use. On the instrument itself, there are direct access water and waste lines, with automatic dilution of wash concentrate. The reagent storage compartment is temperature controlled to provide extended on board stability, and relieve the need to unload reagents once loaded.

For further time efficiency, a single calibration curve can be used for up to 24 hours. This enables more samples to be run on plates and easier reruns. Additionally, all calibrators and controls come in cassette format to support automatic punching.

SCREENING FOR CONGENITAL HYPOTHYROIDISM

using GSP Neonatal hTSH and GSP Neonatal Thyroxine (T4) assays

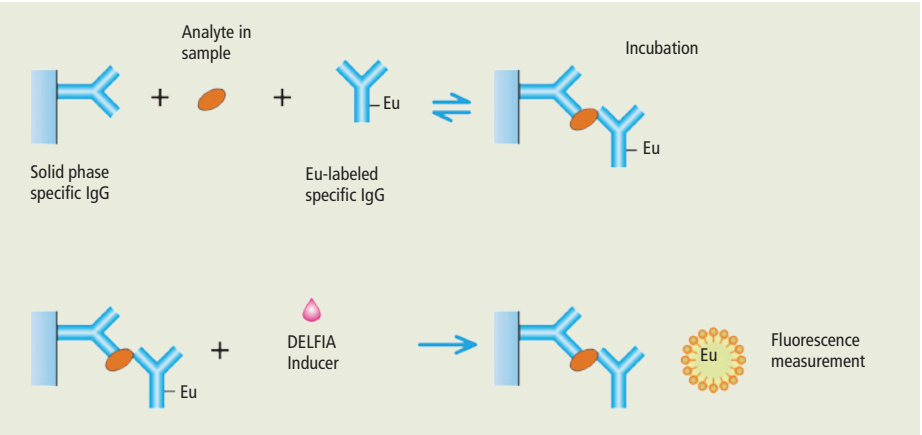
Congenital hypothyroidism (CH) occurs in 1 in 4,000 to 1 in 3,000 newborns.[1] CH results from a failure of the thyroid glands to produce thyroid hormones in adequate amounts. The condition can easily be treated with daily doses of thyroid hormones but clinical diagnosis is difficult to establish and the disease may continue unrecognized for a long time causing irreversible brain damage. However, increased thyroid stimulating hormone (hTSH) and decreased thyroxine are clear signs of CH, which have led to the establishment of large scale screening programs.

The PerkinElmer GSP Neonatal hTSH and GSP Neonatal Thyroxine (T4) assays are used with dried blood spot specimens as an aid in screening newborns for congenital hypothyroidism.

- The incubation time is only 3.5 h for hTSH, and 2 h for T4
- Sensitive, robust DELFIA chemistry for confidence in results
- The kits contain reagents and plates for 1152 tests (12 plates)

GSP Neonatal hTSH assay

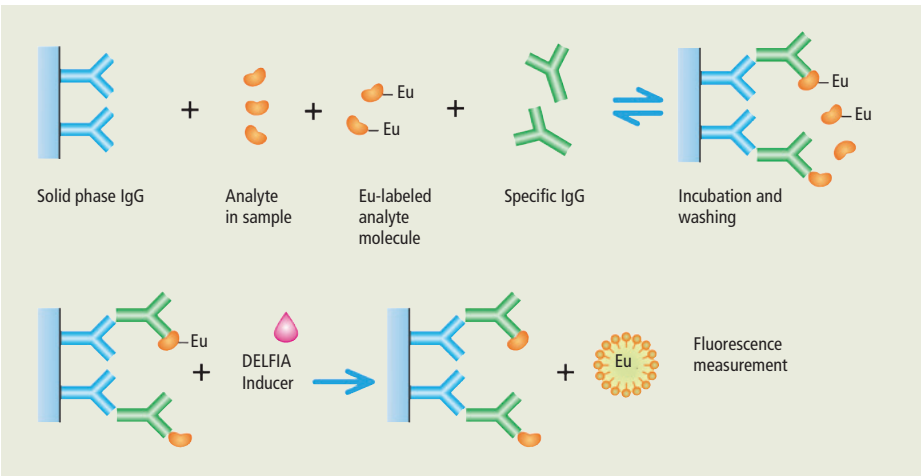
The GSP Neonatal hTSH assay is based on a direct sandwich technique where two monoclonal antibodies recognize separate antigenic determinants on the hTSH molecule. The fluorescence signal is proportional to the analyte concentration in the sample.



Sandwich-type assay design as used in the GSP Neonatal assay for hTSH.
The same design is also employed in the IRT assay described on page 11.

GSP Neonatal Thyroxine (T4) assay

In the GSP Neonatal Thyroxine (T4) assay the analyte competes with europium-labeled T4 for the binding sites on T4 specific monoclonal antibodies and the fluorescence signal is inversely proportional to the analyte concentration in the sample.



Competitive assay design as used in the GSP Neonatal assay for T4.
The same design is also employed in the 17OHP assay described on page 8.

AUTOMATED SCREENING FOR GALACTOSEMIA

GSP Neonatal Total Galactose (TGAL) and GSP Neonatal GALT

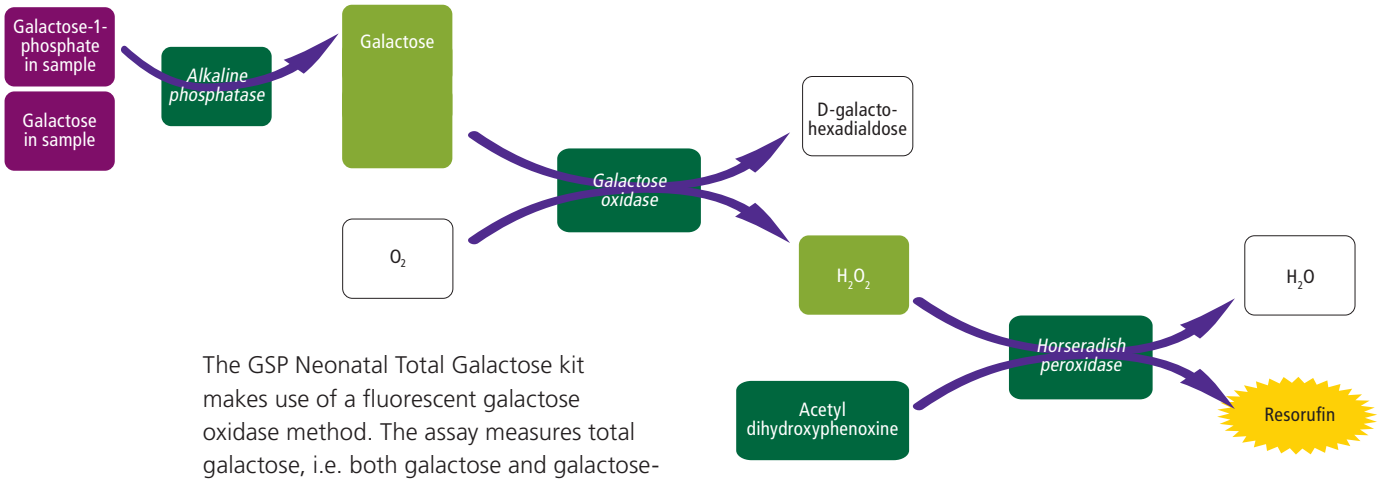
Galactosemia is an inherited disorder caused by a deficiency of one of three enzymes responsible for the metabolism of α-D-galactose. The most common form of the disease, galactose 1-phosphate uridyltransferase (GALT) deficiency occurs in approximately 1 in 30,000 - 60,000 live births.[2] This disorder is often referred to as classic galactosemia. If not diagnosed and treated within the newborn period, it can lead to diarrhea, dehydration, jaundice, hepatic failure, hypoglycemia, cataracts, developmental retardation, and death within a few weeks. Treatment of the disease consists of withdrawal of all foods containing lactose and galactose from the diet.

The GSP Neonatal Total Galactose (TGAL) assay and GSP Neonatal GALT assay are the first fully automated assays for galactosemia screening. The assays are used with dried blood spot specimens.

GSP Neonatal Total Galactose (TGAL) assay

The GSP Neonatal Total Galactose (TGAL) assay is used as an aid in screening for deficiency in any of these three enzymes involved in galactose metabolism.

- The first fully automated Total Galactose assay for galactosemia screening
- The reconstituted reagent stability is improved from one hour in the manual kit to several days
- Contains reagents for 1152 tests (12 plates), clear U-bottomed microplates should be ordered separately in a bulk pack of 100 (4091-0010)



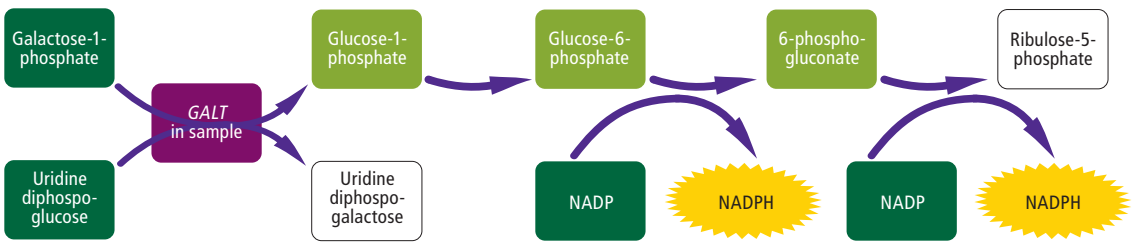
Design for the GSP Neonatal Total Galactose (TGAL) assay

The GSP Neonatal Total Galactose kit makes use of a fluorescent galactose oxidase method. The assay measures total galactose, i.e. both galactose and galactose-1-phosphate. The illustration summarizes the reactions that occur during the test procedure. The fluorescence is measured using an excitation wavelength of 505 nm and an emission wavelength of 580 nm.

GSP Neonatal GALT assay

The GSP Neonatal GALT assay is used as an aid in screening newborns for classic galactosemia.

- Faster assay - incubation time now only 2 h
- Improved precision compared to manual assays
- Provides results in today's preferred unit, U/dL
- Floating disks control
- Contains reagents for 1152 tests



Design for the GSP Neonatal GALT assay

In the GSP Neonatal GALT assay, GALT in the blood sample catalyzes a reaction between galactose- 1-phosphate and uridine diphosphoglucose contained in the assay substrate reagent. In the course of further reactions NADP (nicotinamide adenine dinucleotide phosphate) also contained in the assay substrate reagent is reduced to NADPH, a fluorescent substance that can be measured with excitation at 355 nm and emission detection at 460 nm.

SCREENING FOR CONGENITAL ADRENAL HYPERPLASIA

with GSP Neonatal 17α-OH-progesterone

Congenital adrenal hyperplasia (CAH) is a genetic disorder affecting approximately 1 in 16,000 live births in North America[1], and the most severe form of the disease can lead to a life threatening condition during the first weeks of life. The disease is caused by enzyme defects in steroid biosynthesis, the most frequent types being 21- and 11a-hydroxylase deficiency. These types represent 95% of CAH cases and in both, the 17α-OH-progesterone (17OHP), a precursor of cortisol, is increased. The determination of 17OHP is thus a useful screening method for 95% of all CAH cases.

The GSP Neonatal 17α-OH-progesterone assay is intended for the quantitative determination of 17OHP in dried blood spot specimens as an aid in screening newborns for CAH.

- Incubation time 3 h
- Sensitive, robust DELFIA chemistry for confidence in results
- Contains reagents and plates for 1152 tests (12 plates)

GSP Neonatal 17α-OH-progesterone assay

The GSP Neonatal 17α-OH-progesterone assay is based on the competitive binding of europium-labeled 17OHP and 17OHP in the sample to 17OHP-specific antibodies. The fluorescence signal is inversely proportional to the analyte concentration in the sample.

The assay principle is shown in the lower diagram on page 5.

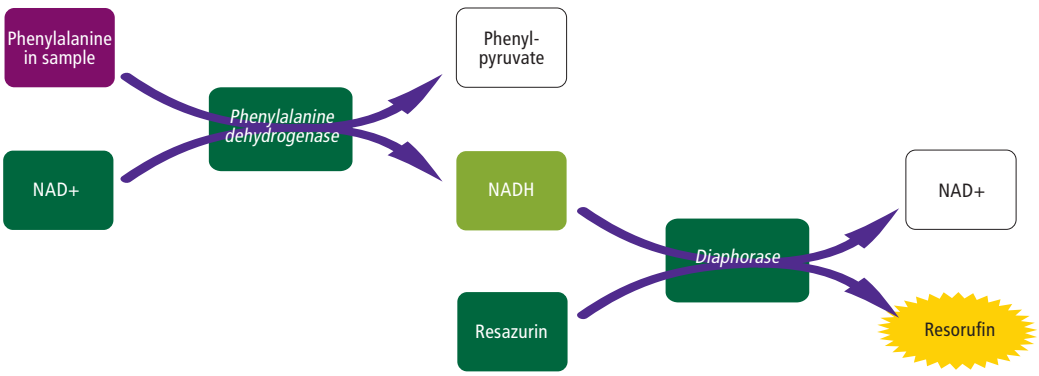
SCREENING FOR PKU WITH GSP NEONATAL PHENYLALANINE

- the automated enzymatic assay

Phenylketonuria (PKU) is a disorder of amino acid metabolism with reported incidence ranging from 1 in 19,000 to 1 in 13,500 newborn infants[1]. It is caused by an inability to convert phenylalanine to tyrosine due to deficient activity of the enzyme, phenylalanine hydroxylase. As a result, excessive amounts of phenylalanine and toxic metabolites accumulate causing various degrees of mental retardation. The symptoms can be clearly reduced with a diet low in phenylalanine, and early detection is critical in starting the treatment and ensuring normal brain development.

The GSP® Neonatal Phenylalanine (Phe) kit is intended for the quantitative determination of phenylalanine concentrations in blood specimens dried on filter paper as an aid in screening newborns for phenylketonuria.

- Automated assay
- Incubation time only 2 x 1h
- Improved precision and performance compared to manual assays
- 1152 tests (12 plates) or 5760 tests (60 plates) product versions, clear U-bottomed microplates should be ordered separately in a bulk pack of 100 (4091-0010)
- Calibrators and controls are in cassette format



Design for the GSP Neonatal Phenylalanine assay

In the first reaction of GSP Neonatal Phenylalanine assay, phenylalanine dehydrogenase converts phenylalanine to phenylpyruvate, generating a stoichiometric amount of NADH. In the presence of NADH, resazurin dye is reduced to fluorescent resorufin in a diaphorase catalyzed reaction. Resorufin fluorescence is read using an excitation wavelength of 505 nm and an emission wavelength of 580 nm. This method quantitatively measures the phenylalanine present in the sample.

SCREENING FOR BIOTINIDASE DEFICIENCY

with GSP Neonatal Biotinidase assay

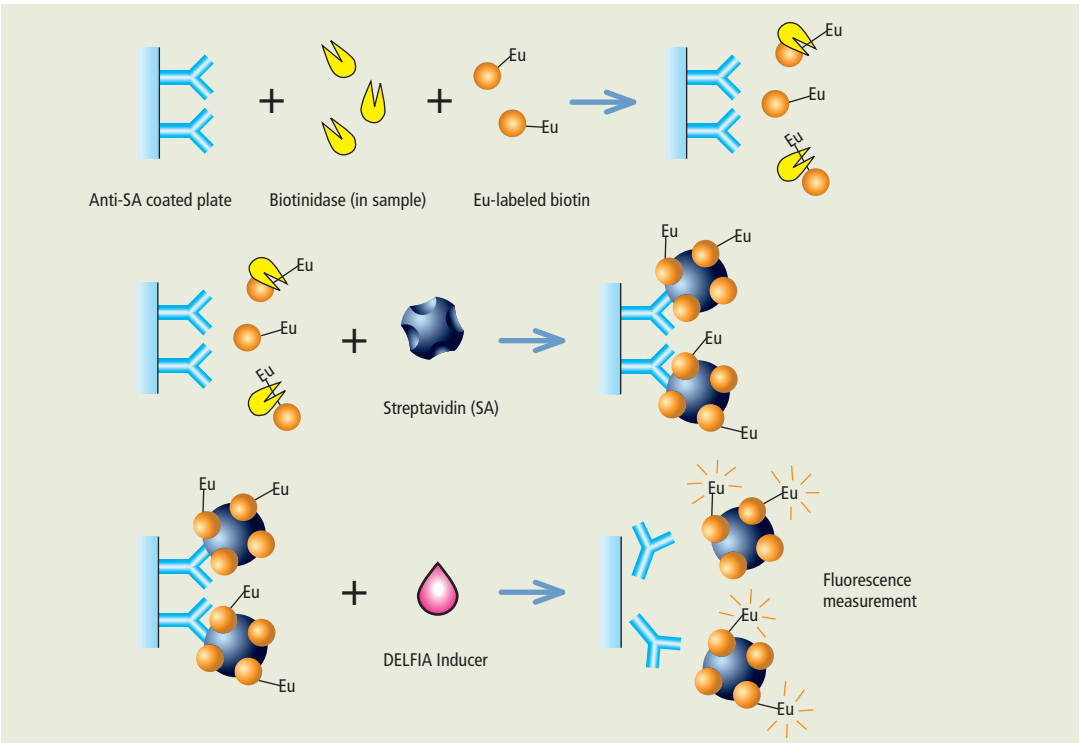
The incidence of biotinidase deficiency has been reported as follows: profound biotinidase deficiency (<10% activity) 1 in 112,000, partial deficiency (10%–30% activity) 1 in 129,000, and profound and partial deficiency together 1 in 60,000[1]. Symptoms include seizure and possible skin disorders, followed by developmental delays, speech problems and possible vision and hearing difficulties.

The GSP Neonatal Biotinidase assay is intended for the semi-quantitative determination of biotinidase activity using dried blood spot specimens.

- First fully automatic assay for biotinidase deficiency screening
- Improved performance
- Combination of enzymatic and immunoassay
- All reagents ready to use (no need for reconstitution)
- No ethanol precipitation in the assay

GSP Neonatal Biotinidase assay

The GSP Neonatal Biotinidase assay combines an enzyme reaction with a solid phase time-resolved immunofluorescence assay. The enzyme reaction is the cleavage by biotinidase of the amide bond in Eu-labeled biotin. The enzyme reaction is stopped by addition of streptavidin which has high affinity for biotin (either Eu-labeled or free biotin). The streptavidin-biotin complexes are captured by the solid phase monoclonal antibody directed against streptavidin. DELFIA® Inducer dissociates the molecules into the solution where the europium fluorescence is measured. The measured fluorescence is inversely proportional to the biotinidase activity of the sample.



Design for the GSP Neonatal Biotinidase assay

SCREENING FOR CYSTIC FIBROSIS

with GSP Neonatal IRT

Cystic fibrosis is a common genetic disorder affecting approximately 1 in 3,500 white newborn infants[1]. The diagnosis is often based on the symptoms which may cause considerable delays in the disease intervention, and evidence indicates that early attention may be important in determining the clinical outcome. The amount of the pancreatic enzyme, immunoreactive trypsin(ogen) (IRT) has been shown to be increased in blood of CF patients especially during the first weeks after birth.

The GSP Neonatal IRT assay allows the quantitative determination of IRT from dried blood spot specimens, and is intended as an aid in screening for cystic fibrosis.

- Linear calibration curve fitting - improved precision
- Sensitive, robust DELFIA chemistry for confidence in results
- Incubation time 2 h
- The kit contains reagents and plates for 1152 tests (12 plates)

GSP Neonatal IRT assay

The assay is based on a direct sandwich technique where two monoclonal antibodies bind to different epitopes on the target molecule. The fluorescence signal is proportional to the analyte concentration in the sample.

The assay principle is shown in the upper diagram on page 5.

ORDERING INFORMATION

3301-0010	GSP Neonatal hTSH kit
3302-0010	GSP Neonatal Thyroxine (T4) kit
3303-0010	GSP Neonatal GALT kit excluding plates, reagents for 12 plates
3305-0010	GSP Neonatal 17α-OH-progesterone kit
3306-0010	GSP Neonatal IRT kit
3307-0010	GSP Neonatal Biotinidase kit
3308-0010	GSP Neonatal Phenylalanine (Phe) kit
3309-0010	GSP Neonatal Total Galactose (TGAL) kit
3304-0010	DELFIA Inducer
4080-0010	GSP Wash concentrate
4076-0010	Clear microplates for GSP Neonatal GALT kit (50 plates)
4091-0010	Clear U-bottomed microplates for GSP Neonatal Phe kit or GSP Neonatal TGAL kit (100 plates)

All PerkinElmer neonatal products may not be available in all countries.

References:

[1] Kaye, CI. and the Committee on Genetics (2006)
Newborn Screening Fact Sheets.
Pediatrics 118; 934-963. DOI: 10.1542/peds.2006-1783.

[2] Fridovich-Keil, JL. (2006).
Galactosemia: The Good, the Bad, and the Unknown.
J Cell Physiol. 209; 701-705.

For the better

At PerkinElmer, we're working to improve the health and safety of people and their environment. From safer water to cleaner air to healthier babies, our solutions touch every part of your life.

For information on availability of PerkinElmer neonatal products please contact your local representative.

PerkinElmer, Inc.
940 Winter Street
Waltham, MA 02451 USA
Phone: (800) 762-4000 or
(+1) 203-925-4602
www.perkinelmer.com

PerkinElmer, Inc.
Wallac Oy
PO Box 10
20101 Turku, Finland
Phone: (+ 358) 22678-111
Fax: (+ 358) 22678-35757

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