COMPLETE SOLUTIONS FOR SCREENING NEWBORNS

ANALYTES, INSTRUMENTATION AND SOFTWARE FOR NEONATAL SCREENING

PerkinElmer®
For the Better
EVERYTHING YOU NEED FOR EFFICIENT NEONATAL SCREENING

Screening before symptoms appear
PerkinElmer provides you with the tools you need to run a successful and cost-effective neonatal screening program. The test kit range covers most of the congenital disorders for which neonates are currently screened.

Our three conventional assay platforms are GSP®, AutoDELFIA® and VICTOR™. For these platforms, and subject to local registration requirements, we supply automated or manual assays for T4, TSH, 17α-hydroxyprogesterone, IRT and Hb, as well as for Phenylalanine, GALT, TGAL, Biotinidase and G6PD. Kits have been developed specifically for neonatal screening, and employ proven fluorescence chemistries to assure excellent performance.

All tests are based on dried blood spot samples measured in 96-well microtitration plates. To prepare the plates a variety of punching devices are available, providing the level of automation desired by the laboratory.

For hemoglobinopathy testing PerkinElmer offers an additional range of products based on the RESOLVE® system and isoelectric focusing.

PerkinElmer is a pioneer in the application of tandem mass spectrometry (MSMS) to newborn screening, supplying instrumentation, informatics and reagents. These products include the NeoBase™ MSMS reagent kit, which has 25 internal standards and supports screening for more than 30 disorders.

GSP and manual assays for Phenylalanine and G6PD, GSP assays for TGAL and Biotinidase, and immunoassays for Hb are not available in the USA.
ASSAY PROCESSING

Conventional assay platforms

GSP® is a multitechnology instrument for neonatal dried blood spot assays employing DELFIA® chemistry as well as prompt fluorescence or absorbance-based measurement technologies. From plate loading to results output, GSP performs all assay stages automatically. For laboratories requiring enhanced efficiency with higher throughput, the GSP instrument is compatible with Panthera-Puncher™ 9 and with Specimen Gate® informatics.

The VICTOR®_D Multilabel Counter is used for measurement of all manual neonatal fluorescence based assays. It is supported with a range of devices for sample preparation including the DBS Puncher, DELFIA Washer-Diskremove, DELFIA Plateshake and/or TriNest™ Incubator Shaker.

Expanded screening platform

The TQD MSMS instrument is used with the 2777C Sample Manager and 1525μ Binary Pump. Employing well-tested and highly reliable Waters triple quadrupole mass spectrometer technology, TQD offers the proven stability of the Waters Quattro™ micro, but superior transition times due to T-Wave™ collision cell technology. The TQD instrument has a very small footprint (35.5 cm x 84.8 cm, or 14”x 33.5”).

Sample preparation devices

With Panthera-Puncher™ 9, up to 9 microtitration plates can be prepared simultaneously, and up to 3 of the plates can be deep-well plates. There are four different punch head sizes (diameters, 1.5, 3.2, 4.75 and 6.0 mm) and 2 punch heads may be installed at a time so plates requiring different blood spot sizes can be punched during the same run. There is a camera to provide a clear full-color view of the punching area in real-time, and an automatically adaptable punch pattern that adapts to the shape and size of the blood spot.

The DBS Puncher has a changeable head to allow the use of disks with diameters of 3.2, 4.7 or 6 mm. Two plates may be loaded to allow simultaneous preparation of different assays.
A FULL RANGE OF SCREENING ASSAYS TO MEET TODAY’S NEEDS

The diseases screened today vary. What they have in common is that without timely treatment they will cause severe retardation of a child’s development. This represents a high cost to the child and their family, to the health care provider and to society in general. PerkinElmer’s assays help secure the first stage of the process to find affected individuals in time.

Congenital hypothyroidism (CH)

Congenital hypothyroidism (CH) occurs in 1 in 4,000 to 1 in 3,000 newborns. If untreated CH can lead to severe developmental or intellectual delay. Treatment in time allows affected children to develop normally.

PerkinElmer Neonatal hTSH and Neonatal T4 assays offer two alternative screening strategies for CH. The assays are time-resolved fluorometry-based assays using dried blood spot samples.

Available on these platforms:

<table>
<thead>
<tr>
<th>Assay</th>
<th>Platform 1</th>
<th>Platform 2</th>
<th>Platform 3</th>
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<tbody>
<tr>
<td>Neonatal hTSH</td>
<td>GSP</td>
<td>AutoDELFIA</td>
<td>VICTOR'D</td>
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<tr>
<td>Neonatal T4</td>
<td>GSP</td>
<td>AutoDELFIA</td>
<td>VICTOR'D</td>
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Galactosemia

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 uridylytransferase (GALT) deficiency occurs in approximately 1 in 47,000 newborn infants. If not diagnosed and treated within the newborn period, this disorder can lead to diarrhea, dehydration, jaundice, hepatic failure, hypoglycemia, cataracts, developmental retardation, and death within a few weeks.

PerkinElmer’s Neonatal GALT assay is intended for the determination of GALT (galactose-1-phosphate uridylytransferase) concentrations as an aid in screening for classic galactosemia. PerkinElmer’s Neonatal TGAL assay is intended for the determination of total galactose (galactose and galactose-1-phosphate). This measurement provides an aid in screening for deficiency of any of the three enzymes contributing to α-D-galactose metabolism.

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<tr>
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<td>GSP</td>
<td>VICTOR'D</td>
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<tr>
<td>Neonatal TGAL</td>
<td>GSP*</td>
<td>VICTOR'D</td>
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Phenylketonuria (PKU)

Phenylketonuria (PKU) is a genetic disorder with reported incidence ranging from 1 in 19,000 to 1 in 13,500 newborn infants. If untreated it can lead to developmental or intellectual delay. Prompt treatment with a phenylalanine restricted diet allows normal development.

PerkinElmer offers both automatic and manual Neonatal Phenylalanine assays, and these are based on fluorescence techniques.

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</thead>
<tbody>
<tr>
<td>Neonatal Phenylalanine</td>
<td>GSP*</td>
<td>VICTOR'D*</td>
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Glucose-6-phosphate dehydrogenase (G6PD) deficiency

G6PD deficiency is one of the most common newborn disorders, and is caused by a class of mutation on the long arm of the X chromosome. According to a WHO estimate 2.5% of newborns worldwide are G6PD deficient (2.1% male hemizygotes, 0.01% female homozygotes, 0.4% G6PD deficient female heterozygotes). Affected persons are sensitive to anti-malarial drugs, fava beans, sulfa drugs and large doses of vitamin C. These substances may trigger an oxidative stress that may cause jaundice, fatigue, tachycardia and enlarged spleen. In neonates and young children there can be a buildup of unconjugated bilirubin in the brain that may result in mental retardation or death.

The PerkinElmer Neonatal G6PD assay is designed for the quantitative measurement of G6PD from dried blood spot samples.

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Cystic fibrosis (CF)
Cystic fibrosis is a common genetic disorder affecting approximately 1 in 3,500 white newborn infants. CF causes chronic obstructive lung disease, airway infections and gastrointestinal abnormalities. Early detection and treatment can significantly improve the quality of life.

PerkinElmer’s Neonatal IRT allows measurement of immunoreactive trypsin (IRT) from dried blood spots. Measurement of IRT can be used as an aid in identifying newborns at increased risk of having CF.

Available on these platforms:
- Neonatal IRT
- GSP
- AutoDELFIA
- VICTOR’D

Hemoglobinopathies
Hemoglobinopathies include sickle-cell disease and thalassemias. According to a WHO estimate roughly 0.25% of all children born every year suffer one or other of these diseases. Children with hemoglobinopathies are at risk in a number of ways. Those suffering sickle-cell disease are very susceptible to pneumococcal infections and acute chest syndrome (ACS), while those with thalassemia will be subject to infections and intestinal problems, and will not thrive.

PerkinElmer’s Hb Immunoassay is designed to detect sickle-cell anemia and all its related traits from dried blood spot samples. Dual label DELFIA technology allows simultaneous measurement of Hb-A and Hb-S hemoglobin forms.

The RESOLVE Neonatal Hemoglobin test kit is designed to separate dried blood spot or cord blood hemoglobins by IEF on a thin layer gel to allow determination of hemoglobin variants and, for example, differentiation between sickle cell anemia and sickle cell trait.

Available on these platforms:
- Neonatal Hb immunoassay
- AutoDELFIA*
- Neonatal Hb IEF test kit
- RESOLVE

Biotinidase deficiency
Based on outcomes for infants screened between 1983 and 1990 it has been estimated that Biotinidase deficiency has an incidence of 1 in roughly 60,000. Symptoms include seizure and possible skin disorders, followed by developmental delays, speech problems and possible vision and hearing difficulties. Biotinidase deficiency is ranked 5th by the ACMG (American College of Medical Genetics) in their list of screenable core conditions.

The PerkinElmer Neonatal Biotinidase assay is intended for the semi-quantitative determination of biotinidase activity.

Available on these platforms:
- Neonatal Biotinidase
- GSP*
- assay in development
- VICTOR’D

Metabolic and amino acid disorders
Using tandem mass spectrometry, this technology laboratories can measure more than 30 amino acid and acylcarnitine analytes in less than two minutes from a single dried blood spot specimen.

For sensitive, reliable amino acid and acylcarnitine assay without a derivatization stage, our NeoBase™ MSMS reagent kit, has 25 internal standards and 23 high and low controls. The kit components have been carefully validated to work together to produce results to assure the laboratory 100% confidence.

Available on these platforms:
- Neonatal AA/AC
- TQD MSMS Instrument

For Tyrosine type 1
By ordering PerkinElmer’s Succinylacetone Assay Solution and adding this to samples at the same time as you add internal standard, you can extract succinylacetone along with amino acids and acylcarnitines. This allows simultaneous screening of succinylacetone alongside the other key metabolic disease markers with the NeoBase kit.

References

* Product not available in the USA or Canada
† Product not available in Canada

All of the PerkinElmer products mentioned on this page are not available in every country. For information on availability in your country please talk to your local PerkinElmer representative.
As the amount of patient demographic information continues to grow, no screening laboratory can function effectively without an information management system. PerkinElmer Specimen Gate® software is used by over 75 customers worldwide to process more than 7 million specimens each year. Our informatics solutions are characterized by insight and functionality, providing intuitive and practical design.

The software makes it possible for a laboratory to track all of the information required to process a sample and follow up a specimen. In addition, results are stored for future reference, and compliance with laboratory regulations is demonstrable at every stage.

**Specimen Gate users can:**

- Quickly find information about specimens, patients, and contacts without losing their place/current work.
- View and enter multiple specimens at once, saving work automatically, and choose between a variety of built-in validation rules.
- Tailor searches to individual needs and create shortcuts to key searches and specimens
- See, and drill down into the details of specimens and demographic information
- Automatically highlight QC and Cutoff violations, streamlining the process of reviewing and documenting assays
- Monitor and evaluate QC materials, trends, standards and controls (including automatic generation of Levey-Jennings plots and statistics such as mean, median, and standard deviation)
- Perform cutoff analysis based on the customer’s patient population

In PerkinElmer software both the on-screen functions and patient reports are available in local language versions. As part of a full range of data import and export capabilities support is provided for HL7, LOINC, and SNOMED.
Electronic results reporting for efficiency and cost savings

As one of several Specimen Gate special modules, PerkinElmer’s eReports tool harnesses the flexibility of the web to provide a faster and more efficient method for screening laboratories to report results. eReports allows laboratories using Specimen Gate applications considerable flexibility as to the design and extent of the information presented in patient reports. Account holders can then access these reports, old or new, via a standard web browser, at any time.

- Cost savings – reduced manual work and no postage
- Lab personnel can free up time and focus on other tasks
- Dedicated information centre enhances requester confidence
- Faster reporting leads ultimately to better patient care
Work with the leader in Newborn Screening
PerkinElmer is the global market leader in neonatal screening, currently serving customers in more than 90 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.

50 babies saved every day
The first DELFIA neonatal kit was developed in 1985, to allow dried blood spot measurement of hTSH. By 2012, some 381 million babies had been screened with PerkinElmer products. For every day of the year serious disorders are revealed in 50 babies so that treatment may be applied in time.

All of the PerkinElmer products mentioned in this brochure are not available in every country. For information on availability in your country please talk to your local PerkinElmer representative.