

Analytes, instrumentation and  
software for neonatal screening



# Complete solutions

for screening newborns

# Everything you need for efficient neonatal screening



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.

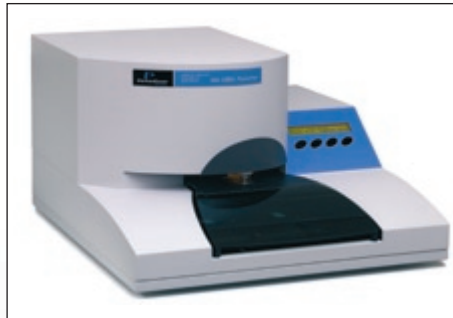
There are DELFIA® or AutoDELFI A® kits for T4, TSH, 17-hydroxyprogesterone, IRT, Hb\* and Toxoplasma\*, and fluorescence kits for Phenylalanine, GALT, TGAL\*, Biotinidase\* and G6PD\*. All tests are based on dry blood spot samples measured in 96-well microtitration plates. Kits have been developed specifically for neonatal screening, and employ proven fluorescence chemistries to assure excellent performance.

In addition to its new dual label Hb immunoassay\*, PerkinElmer offers hemoglobinopathy tests employing IEF.

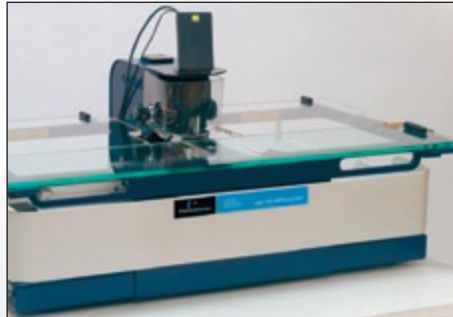
\* Product not available in the USA.

NeoGram™ MSMS reagent kits are designed specially for the newborn screening application using tandem mass spectrometry. Using either the traditional derivatized amino acid/acylcarnitines method or the novel non-derivatized amino acid/acylcarnitines method, more than 30 different metabolic diseases can be detected from a single dried blood spot sample in less than two minutes.

For measurement of tests, PerkinElmer offers various instrument solutions based on our plate readers, automation and the Micromass Quattro tandem mass spectrometer. Together with software for data management, a complete package can be created according to the needs of the screening program.



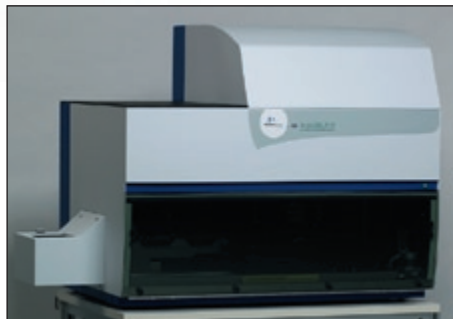
The Wallac DBS Puncher automatically punches dry blood spots into microtitration plates. It has a changeable head to allow the use of blood spots with diameters of 3.2, 4.7 or 6 mm. Two plates may be loaded to allow simultaneous preparation of different assays.



The Wallac MultiPuncher™ can handle up to six microtitration plates simultaneously. Two different disc sizes can be punched from the same spot.



The Wallac VICTOR<sup>2</sup>™ D is used for measurement of all Wallac neonatal fluorescence based assays. Together with Wallac devices such as the DBS Puncher, DELFIA Washer-Diskremove, DELFIA Plateshake and the Wallac incubator, it supports semi-automatic assay performance.



The Wallac AutoDELFLIA<sup>®</sup> immunoassay system allows automatic processing of the DELFLIA<sup>®</sup> Neonatal assays. It accepts worklists for microplates prepared using the Wallac DBS Puncher or the Wallac MultiPuncher.



The Micromass Quattro™ is a compact efficient tandem mass spectrometer. It is supported with reliable MSMS reagent kits and informatics.

# A wide range of **screening assays** to meet today's needs

The diseases screened today are varied. 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)**

The incidence of CH is 1:3000 – 1:6000. If untreated CH can lead to severe mental retardation. Treatment in time allows affected children to develop normally.

### **DELFI<sup>®</sup>/AutoDELFI<sup>®</sup> Neonatal TSH and DELFI<sup>®</sup>/AutoDELFI<sup>®</sup> Neonatal T4**

The DELFIA Neonatal TSH and the DELFIA Neonatal T4 kits offer two alternative screening strategies for CH. Both are time-resolved fluorometry-based assays using dried blood spot samples.

## **Galactosemia**

The incidence of galactosemia is 1:14000 – 1:60000. The untreated disorder will cause poor development, and in the worst case fatality. Treatment consists of a galactose-free diet and allows remission of symptoms.

### **Galactose-1-phosphate uridyl transferase (GALT) and Neonatal Total Galactose (TGAL)\***

The Neonatal GALT assay is intended for the determination of GALT 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  $\alpha$ -D-galactose metabolism.

## **Phenylketonuria (PKU)**

The incidence of PKU is 1:4500 – 1:20000. If untreated it can lead to mental retardation. Prompt treatment with a phenylalanine restricted diet allows normal development.

### **Neonatal Phenylalanine**

The Phenylalanine assay is based on the fluorescence of a phenylalanine-ninhydrin reaction product, which is enhanced by the dipeptide L-leucyl-L-alanine. The use of a succinate buffer and the addition of copper further improve the analytical specificity and sensitivity.

### **NeoGram<sup>™</sup> PKU**

For laboratories expanding into tandem mass spectrometry, the NeoGram PKU reagent kit provides a reliable alternative for measurement of phenylalanine and tyrosine.

## **Glucose-6-phosphate dehydrogenase (G6PD) deficiency**

G6PD deficiency is one of the most common newborn disorders. In the US it has an incidence of 0.6-2.9% and in some countries as high as 26%. Affected persons are sensitive to e.g. 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.

### **Neonatal G6PD\***

The neonatal G6PD assay is designed for the quantitative measurement of G6PD on dried blood spot samples.



## Cystic fibrosis (CF)

The incidence of CF is 1:2000 – 1:3000 in Caucasians. CF causes chronic obstructive lung disease, airway infections and gastrointestinal abnormalities. Early detection and treatment can significantly improve the quality of life.

### DELFIA®/AutoDELFIA® Neonatal IRT

DELFIA® 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.



## Hemoglobinopathies

Hemoglobinopathies represent the biggest group of recessive genetic disorders on earth. They comprise sickle-cell diseases, and thalassemias. As many as 1 in 10 African Americans is a carrier of the sickle cell allele, while in Southeast Asia alone, 30 million people suffer from thalassemia. Children with hemoglobinopathies are at risk in a number of ways. Those suffering from sickle cell diseases are, in particular, very susceptible to pneumococcal infections and acute chest syndrome (ACS). While an infant with a thalassemia will be subject to infections and intestinal problems and will not thrive.

### AutoDELFIA® Hb Immunoassay\*

The AutoDELFIA® Hb Immunoassay supports the detection of Sickle Cell Anemia and all its related traits from dried blood spot samples. Dual label technology is employed to allow simultaneous measurement of Hb-A and Hb-S hemoglobin forms.

### Resolve Neonatal Hemoglobin

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.

## Congenital adrenal hyperplasia (CAH)

The incidence of CAH is 1:10000 to 1:15000. CAH is a group of disorders, the most serious one being potentially fatal. For all classes of CAH, early treatment can greatly benefit the patient.

### DELFIA®/AutoDELFIA® Neonatal 17-OHP

The DELFIA Neonatal 17-OHP assay is a competitive immunoassay optimized for measurement of 17-OHP from dried blood spots.

## Biotinidase deficiency

Although the incidence of profound biotinidase is estimated to be lower than 1:100,000 worldwide, biotinidase deficiency (either profound or partial) may have incidence greater than 1:10,000 in some populations. 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.

### Neonatal Biotinidase\*

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

## Congenital toxoplasmosis

The incidence of congenital toxoplasmosis has been estimated to be 1:1000 to 1:10000. It is caused by the protozoa, *Toxoplasma gondii*. Acute infections in pregnant women can be transmitted to the fetus, later causing severe illness in the child. The illness is characterized by damage to the eyes, nervous system, skin, and ears. In the majority of affected children, symptoms are not obvious at birth. Congenital toxoplasmosis is a treatable disease, but must be detected at an early stage.

### AutoDELFIA® Neonatal Toxoplasma-Screen\*

The AutoDELFIA Neonatal Toxoplasma-Screen kit detects IgM antibodies to *Toxoplasma gondii* by means of immunoassay from dried blood spot samples.

\* Product not available in the USA. The availability of all products should be checked locally.

# Mass spectrometry means multiple analyte testing

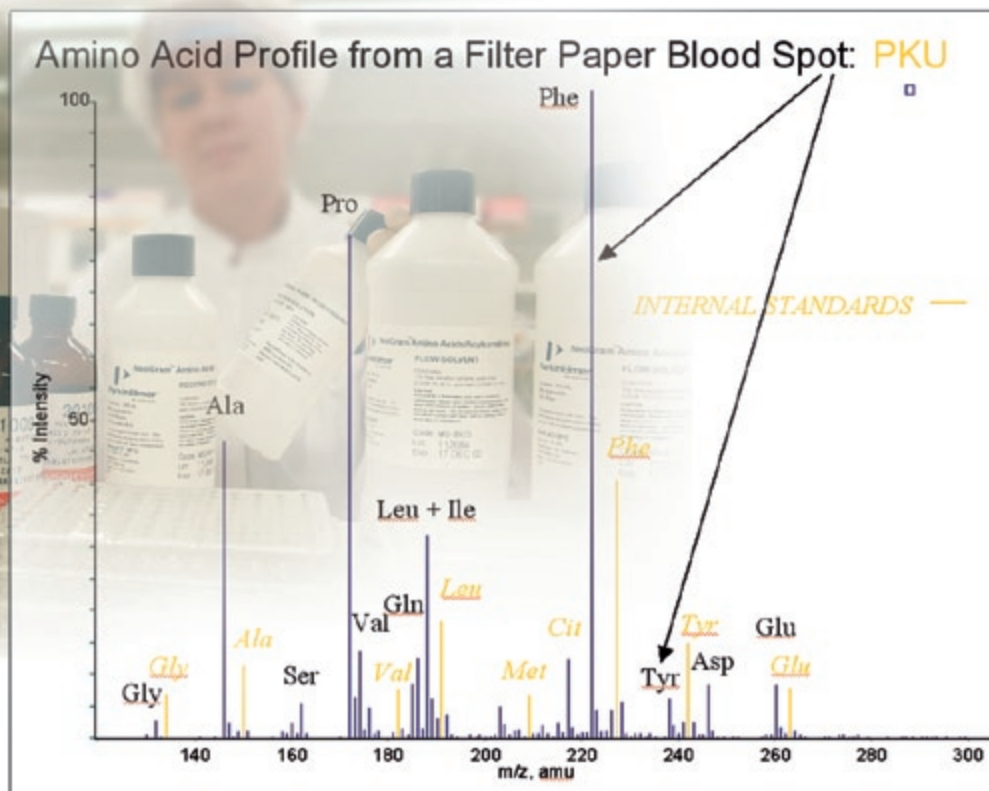
PerkinElmer is the pioneer in the commercial application of mass spectrometry for newborn screening. Our knowledge and commitment to newborn screening make us the best partner to expand your screening program. Using tandem mass spectrometry, multiple analytes are measured simultaneously. Only a single test is required; changing the concept of screening from “one test – one disorder” to the “one test – many disorders”.

PerkinElmer reagent kits have been specifically optimized for the quantitative measurement of amino acids and acylcarnitines from dried blood spot specimens in a high throughput screening environment. In combination with the tandem mass spectrometer and NeoGram software, a system can measure more than 30 analyte

parameters in less than two minutes from a single dried blood spot specimen. A kit includes all the reagent components necessary to produce results. Components have been carefully validated to work together to produce results so that the laboratory can have 100 % confidence.

## NeoGram™ derivatized AAAC kit

In addition to the NeoGram PKU kit, PerkinElmer supplies the NeoGram derivatized kit for amino acids (including glycine, alanine, valine, leucine, methionine, phenylalanine, tyrosine, ornithine, citruline and arginine) and acylcarnitines. The acylcarnitine internal standards include C0 (free carnitine), C2, C3, C4, C5, C5DC, C6, C8, C10, C12, C14, C16, C18.



With MS-MS you can effectively screen for all relevant amino acids and acylcarnitines, all from one single sample, in less than two minutes.

# Specimen Gate® informatics

based on a **higher level** of expertise

As the amount of patient demographic information continues to grow, no screening laboratory can function effectively without an information management system. And the strength of MSMS technology – the ability to multiplex - is increasing the data flood further, making the need for efficiency in the screening laboratory even more acute.

Your laboratory must be able to track all of the information required to process a sample and follow up a specimen. In addition, timely, accurate results have to be stored for future reference. While all this is going on you have to maintain and demonstrate your compliance with laboratory regulations.

PerkinElmer provides the solution in the form of its Specimen Gate® software suite. Specimen Gate has been under continuous development since 1994 and is

installed in more than 100 laboratories in over thirty countries worldwide. A full-time staff of highly trained professionals are dedicated solely to the development, implementation, and support of Specimen Gate software suite.

### Modular and scalable

Specimen Gate is modular. The main modules are Specimen Gate Laboratory, Specimen Gate LifeCycle™ and Specimen Gate PatientCare™. To meet the needs of all kinds of programs and laboratories, Specimen Gate Laboratory comes in alternative product editions. Furthermore, the software is scalable – you can start with the Laboratory module, and expand as your laboratory's needs grow to the full Specimen Gate software suite with LifeCycle and PatientCare products.

**Specimen Gate® Laboratory**

- Punching and Request/Repeat Management
- Quality Control
- Assay Review
- Result Code Assignment
- Cut-off Analysis

**Specimen Gate® LifeCycle**

- Data Entry
- Linking and Matching
- Reporting (including Patient Reports)
- Contact Management

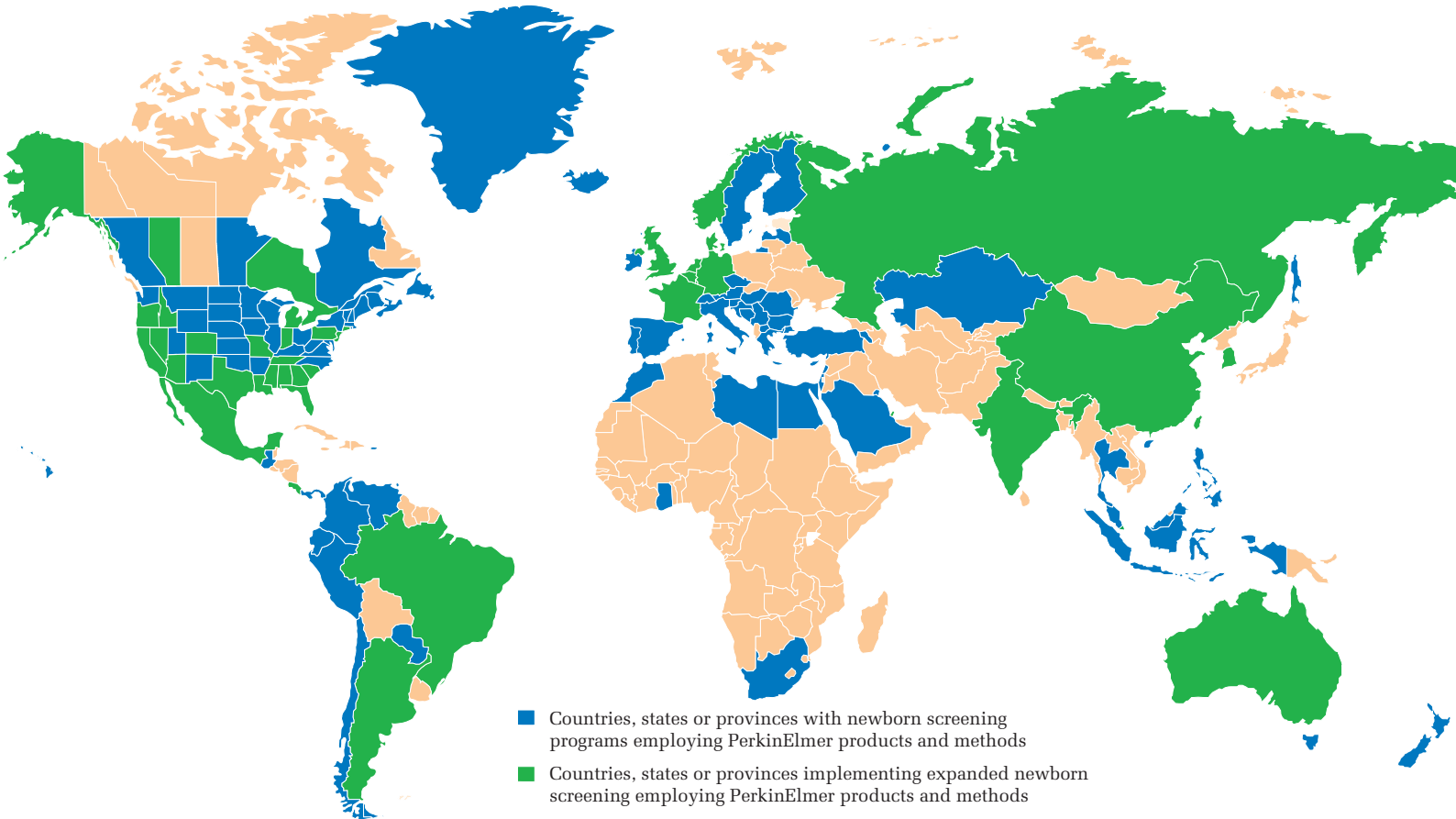
**Specimen Gate® PatientCare**

- Users track each action (e.g. phone call or letter)
- Configurable alerts show expired deadlines
- Configurable workflows mimic follow-up processes

Specimen Gate Laboratory is available in alternative product versions

- Enterprise Edition
- MSMS Data Suite Edition

# PerkinElmer is active in newborn screening in 74 countries and all US states



PerkinElmer Life and Analytical Sciences provides innovative total solutions for population screening and clinical diagnostics, as well as for drug discovery, and genomic and proteomic research. PerkinElmer population screening and clinical diagnostics products are used to protect the health of members of the population from before birth to old age.

Our key role is to help customers to identify susceptible members of the population, so that appropriate treatment may be applied at the right time, often before symptoms of a disease appear. With our wide range of test kits, instruments and software we offer the key components for cost-effective, successful screening programs.

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All products presented in this brochure may not be available in all countries. For information on availability please contact your local representative.



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