

HUMAN HEALTH

ENVIRONMENTAL HEALTH



# WHAT IS NEWBORN SCREENING?

A GUIDE FOR PARENTS



**SAVING  
LIVES WITH  
PERKINELMER  
NEWBORN  
SCREENING**



# SIMPLE BLOOD TESTS FOR ALL BABIES

Newborn screening is a form of preventive health care in which babies are tested within the first days of their life to discover evidence of disorders for which the principal symptoms may not yet be apparent.

There are a number of different disorders that are regularly screened, though the exact number will depend on your country, region and health care provider. The tests for the various disorders are all performed using the same blood sample, taken from your baby a few days after birth.

## Timely action can be of utmost importance

The disorders that are being tested for are rare diseases. Congenital hypothyroidism and phenylketonuria, for example, are two commonly-screened disorders that affect only one child in thousands or even one in tens of thousands. The diseases are, however, potentially very serious. Unless treatment is started within a week or two of birth there may be irreversible damage to the child's development.

As long as they are detected in time, the disorders can usually be treated effectively so that adverse symptoms do not develop, and the affected child will go on to enjoy a normal healthy life.



A blood sample is taken from a heel prick when the baby is a few days old. The sample is spotted onto absorbent paper and dried. Small disks of the sample material are then punched out from the paper. The PerkinElmer Panthera-Puncher™ 9 a punching device being operated here performs sample preparation for up to 9 different tests simultaneously .





# WHY YOUR CHILD SHOULD RECEIVE NEWBORN SCREENING

## 50 children, every day, every year, somewhere

Because the diseases in question are rare, the probability of your child actually being affected by one of them is rather small. However,

consider this: PerkinElmer, as the world's largest producer of newborn screening tests estimates that on average, for every day of every year, its newborn screening tests find and allow treatment of 50 babies that would otherwise have developed severe diseases.

As a result of the newborn screening all of these individual children are permitted to move forward to a normal healthy childhood. Without the screening, many of the 50 would have died in infancy, most of the remainder would have lived with physical sickness and would have spent the whole of their lives in a wheelchair; it is unlikely that any of the 50 children would ever have been able to enter the regular education system.

There are 50 such children, every day, every year, somewhere.



The picture shows two children, both positive for congenital hypothyroidism. The 7-year old girl on the right was successfully treated after newborn screening highlighted the disorder. The 14-year old boy was not screened, and the lack of timely treatment resulted in developmental retardation. Illustration courtesy of the University of the Philippines-National Institutes of Health (UP-NIH) and PhilHealth, the Philippine Health Insurance Corporation.

## HISTORY OF NEWBORN SCREENING

**1908** Sir Archibald Garrod introduced the term "inborn error of metabolism" in an historic series of lectures before the Royal College of Physicians in London.

**1962** Dr. Robert Guthrie developed the first simple, sensitive, and inexpensive screening test for PKU.

**1960's** Taking blood drop samples from babies' heels and drying these onto filter paper became the established sampling method in newborn screening, in use ever since.

**1970's** The arrival of the radioimmunoassay technology made it possible to develop tests for congenital hypothyroidism screening.

**2000's** Expanded screening using tandem mass spectrometry made it possible to screen for 30 or more disorders simultaneously.



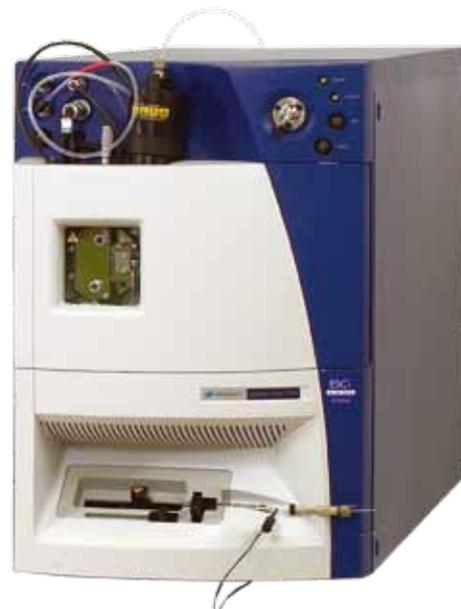
## More than 30 disorders now recommended for screening

In order for screening to be possible a simple and reliable test must exist. Also, there must be a treatment that makes a difference when the disease is detected early.

In 2006 the American College of Medical Genetics published national recommendations for a uniform screening panel. As a result, expanded newborn screening programs, not just in the USA, but in other countries too, provide screening for more than 30 disorders. This multiplication of the number of disorders screened has been made possible by the advent of tandem mass spectrometry. This allows multiple disorders to be screened simultaneously from the same dried blood spot sample.



PerkinElmer offers tandem mass spectrometry-based expanded newborn screening product to allow simultaneous detection of more than 30 disorders from one blood spot.





# COMMONLY- SCREENED DISEASES

The screened diseases are varied – what they all have in common is that without timely treatment they will cause severe harm to the child

## **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<sup>1</sup>. Symptoms include seizure and possible skin disorders, followed by developmental delays, speech problems and possible vision and hearing difficulties.

## **CONGENITAL ADRENAL HYPERPLASIA (CAH)**

Congenital adrenal hyperplasia (CAH) affects approximately 1 in 16,000 live births in North America, with somewhat higher incidences reported for Europe<sup>2</sup>. The most severe form of the disease can lead to a life threatening condition during the first weeks of life. For all classes of CAH, early treatment can greatly benefit the patient.

## **CONGENITAL HYPOTHYROIDISM (CH)**

Congenital hypothyroidism (CH) occurs in 1 in 4,000 to 1 in 3,000 newborns<sup>3</sup>. CH results from a failure of the thyroid glands to produce thyroid hormones in adequate amounts. Untreated, the disease is likely to cause irreversible brain damage. The condition can easily be treated with daily doses of thyroid hormones.

## **CYSTIC FIBROSIS (CF)**

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

## **GALACTOSEMIA**

Galactosemia is an inherited disorder caused by a deficiency of one of three enzymes responsible for the metabolism of  $\alpha$ -D-galactose. The most common form of the disease, galactose 1-phosphate uridylyltransferase (GALT) deficiency occurs in approximately 1 in 47,000 newborn infants<sup>4</sup>. 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. Treatment of the disease consists of withdrawal of all foods containing lactose and galactose from the diet.

## **GLUCOSE-6-PHOSPHATE DEHYDROGENASE (G6PD) DEFICIENCY**

Glucose-6-phosphate dehydrogenase (G6PD) deficiency is the most common of all clinically significant enzyme defects. Affected persons are sensitive to anti-malarial drugs, fava beans, sulfa drugs and large doses of vitamin C. In neonates the untreated condition can result in brain damage or death.

## **HEMOGLOBINOPATHIES**

Hemoglobinopathies comprise sickle-cell disease and thalassemias. 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<sup>5</sup>.

## METABOLIC DISORDERS

Metabolic disorders are faults in the biochemical pathways relating to structural growth or energy utilization. For example, certain amino acid disorders involve a damaging deficiency or excess of an amino acid or its precursor. Alternatively a fault in fatty acid oxidation interferes with the work of the cells' powerhouses - the mitochondria.

While several of these disorders (including Biotinidase deficiency, G6PD deficiency, PKU, etc.) are screened using dedicated tests, many programs nowadays screen for multiple disorders - perhaps as many as 30 - by using expanded newborn screening and tandem mass spectrometry.

## PHENYLKETONURIA

Phenylketonuria (PKU) is a genetic disorder with reported incidence ranging from 1 in 19,000 to 1 in 13,500 newborn infants<sup>3</sup>. It is caused by an inability to convert phenylalanine to tyrosine. 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.

## SCID

Severe Combined Immunodeficiency (SCID) is a group of disorders characterized by a severe defect in T cell production and function, such that affected infants die of infections by age 2 years unless immunity is reconstituted by treatment. Although the precise incidence of SCID is not known, it affects more than 1 in 100,000 babies<sup>6</sup>.

## References

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Your doctor or the staff of your antenatal clinic will be able to tell you more about newborn screening and the options available to you. There are also a number of national and international organizations that are keen to help spread newborn screening in the world by sharing knowledge.

- The American College of Medical Genetics (ACMG) <http://www.acmg.net>
- The Association of Public Health Laboratories (APHL) <http://www.aphl.org>
- Centers for Disease Control and Prevention (CDC) <http://www.cdc.gov>
- Climb - Children Living with Inherited Metabolic Diseases <http://www.climb.org.uk>
- The International Society for Neonatal Screening (ISNS) <http://www.isns-neoscreening.org>
- March of Dimes <http://www.marchofdimes.com>
- The Society for the Study of Inborn Errors of Metabolism (SSIEM) <http://www.ssiem.org>

## PerkinElmer, Inc.

PerkinElmer is a global scientific leader providing an extensive range of technology solutions and services to address the most critical issues facing humanity. From screening of babies to critical research, and from environmental testing to industrial monitoring, we are actively engaged in improving health and enhancing quality of life all around the world.

Recognized as the leader in newborn screening, PerkinElmer touches over 27 million pregnancies and births every year.

Our first products for newborn screening were introduced in the mid 1980's. Today we are active in 74 countries, offering instruments, test kits and laboratory software allowing programs to screen newborns for up to 50 different disorders.

In the USA, as well as being suppliers to almost all State newborn screening programs, we also provide newborn screening services through our PerkinElmer Genetics state-of-the-art newborn screening laboratory. PerkinElmer Genetics offers one of the world's most comprehensive programs for detecting clinically significant inherited disorders.

In addition to newborn screening, we are also committed to pregnancy monitoring and are continuously developing new products to support health care professionals as they strive for better maternal, fetal and newborn health. PerkinElmer also provides proven cord blood stem cell storage for families wishing to have an improved treatment option for nearly 80 life-threatening diseases.

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