

## The successful “Guthrie test” celebrates its 10<sup>th</sup> birthday in Brazil!

The National Neonatal Screening (PNTN) is celebrating ten years of its implementation! It was instituted in Brazil through the Decree GM/MS 822 on June 6, 2001. In Brazil, the program is recognized by the people with the name of “Neonatal heel prick” and the adherence to testing is high and widely sought by the mothers of all social levels. The PNTN provides the diagnosis of four diseases: congenital hypothyroidism, PKU, hemoglobinopathies and cystic fibrosis.

The history of the Newborn Screening Program began in the sixties with the development of the technique for determination of phenylalanine in small samples of dried blood collected on filter paper cards, allowing early diagnosis of phenylketonuria (PKU). PKU was discovered in 1934 and its prevalence is about 1:15,000 live births. This metabolic disease has serious effects on the child’s development if not treated at the correct time. The accumulation of phenylalanine has toxic effects on the nervous system in formation, leading to mental retardation that can be avoided with a specific early diet of low levels of this amino acid. Later, in 1970s, it was developed in Canada, a technique for measurement of thyroxine (T4) and thyrotropin (TSH) on filter paper allowing neonatal screening for hypothyroidism. Hypothyroidism can lead to mental retardation and has a worldwide prevalence of around 1: 3,500 live births. The treatment when instituted until the end of the second week of life is able to guarantee the normal neurological development even in severe cases. Early diagnosis of hemoglobinopathies and cystic fibrosis also allow interventions to prevent complications, increasing the life expectancy of affected children.

Gradually, this type of newborn screening has evolved from a single disease laboratory test - PKU - for a number of other diseases that require a more comprehensive and complex control actions. Neonatal screening is based on laboratory testing in the first days of life of the newborn, if the tests are done on time and in an appropriate manner, it allows the beginning of the treatment to occur within a time window in which it is possible to avoid delays in child development and mental retardation, besides the possibility of genetic counseling that, in the context of health promotion, can help to reduce the incidence of such diseases. The time for collection shall not be less than 48 hours of protein feeding (breast) and not exceeding 30 days, ideally between the 3rd and 7th day of life.

The diseases targeted by neonatal screening programs have gained importance with the changes in the profile of infant morbidity and mortality in the so-called “demographic and epidemiological transition”, in which child mortality associated with infectious diseases and malnutrition were gradually replaced by other complications related to the perinatal period and genetic. In general, diseases in which early intervention can change a poor outcome and for which there is treatment available are those that comprise the Guthrie test in public health.

However, the coverage rate of newborn screening as well as the time of the results of that test are still different in various regions of Brazil. The access of children actually affected by some type of diagnosed diseases is complex, requiring a well-organized network so those who have problems can be quickly identified and the diagnosis of the disease confirmed by more sophisticated exams.

In short, despite the blood collection from the heel of the newborn is widely held in the country at the present time, the time between collection and the institution of the treatment needs to be reduced so that the ultimate goal is achieved: the improvement of the quality of life of the children.

Maria Elizabeth Moreira Lopes

Instituto Fernandes Figueira, Fundação Oswaldo Cruz