Newborn blood spot screening fact sheet

What does the newborn blood spot screening involve?

In the first few days after birth, all babies are offered a newborn blood spot screening. The child's blood is screened for 19 of rare, serious, often hereditary disorders. The timely tracing of these disorders can prevent or minimise serious damage to its physical and cognitive development. These disorders cannot be cured but they can be treated. Screening for sickle cell anaemia can also detect disease carrier status. Parents can choose not to receive any information about carrier status.

1. Clinical picture

Newborn screening (newborn blood spot screening) can trace rare, serious disorders in newborns, making it possible to prevent the child from suffering irreversible health damage. Most disorders are hereditary, they can not be cured but they can be treated, with medication or a special diet, for example.

The following disorders are currently in the screening programme:

Thyroid disorder:

- Congenital hypothyroidism (**CH**): group of disorders in which the thyroid gland produces insufficient amounts of thyroid hormone (thyroxine, T4). CH is usually permanent and usually not hereditary. T4 plays an important role in regulating metabolism and is essential for growth and development. T4 deficiency at a young age has a negative impact on brain development, with a risk of permanent learning disability and limited motor ability. Early treatment with T4 can prevent this (almost) entirely. Treatment: lifelong course of daily thyroxine tablets. Prevalence: about 70-90 children per year.

Adrenal disorder:

- Adrenogenital syndrome (**AGS**): life-threatening, hereditary disorder affecting hormone production by the adrenal glands. The deviation leads to a cortisol deficiency, often also an aldosterone deficiency and an overproduction of androgens. In newborns, excessive salt loss leads to dehydration. At birth, girls have different degrees of masculinisation of the external genitals. Early treatment can prevent serious disruption of water and salt metabolism. Treatment: lifelong course of corticosteroid medication and other complementary medication. Prevalence: about 10-15 children per year.

Metabolic disorders:

- Biotinidase deficiency (**BIO**): hereditary metabolic disorder in which too little biotin (vitamin H) is produced. Untreated it results in skin problems, epileptic attacks, occasional baldness (partial or complete), delayed development and muscle problems. Early treatment can prevent all symptoms. Treatment: lifelong course of biotin. Prevalence: about 2-4 children per year.
- Galactosaemia (**GAL**): hereditary metabolic disorder in which galactose (component of milk sugar, also known as lactose) is not broken down sufficiently. Lactose is found in breast milk and in many food products for infants. Leads to severe jaundice, infection, cataracts (an eye disease) and death. Despite good treatment, galactosemia might lead to developmental delay and reduced fertility in of girls. Treatment: strict lifelong diet low in galactose. Prevalence: about 2-4 children per year.

- Glutaric acidaemia type I (**GA-I**): hereditary metabolic disorder, in which the amino acids lysine and tryptophan are not broken down properly. Untreated, this can lead to brain damage. A diet and medicinal treatment can prevent brain damage. Treatment: lifelong dietary protein restriction, with 'amino acid preparation' and medication. Prevalence: about 1 child per year.
- HMG-CoA-lyase deficiency (**HMG**): hereditary metabolic disorder in which the amino acid leucine is not broken down properly, resulting in suboptimal fatty acid oxidation leading to an energy deficiency. Problems arise during fasting, overnight sleeping, operations, periods of diarrhea and vomiting. If left untreated, the disease can cause vomiting, weakness and drowsiness, loss of consciousness, neurological problems and impaired development. Treatment: sometimes medication (carnitine) and a diet. Prevalence: about 1 child every 10 years.
- Isovalerian acidity (**IVA**): hereditary metabolic disorder in which the amino acid leucine is not broken down properly. Leads to vomiting, loss of consciousness, severe developmental retardation and death. Treatment: lifelong dietary protein restriction, 'amino acid preparation' and medication. Prevalence: about 2 children per year.
- Maple syrup urine disease (**MSUD**): hereditary metabolic disorder in which the breakdown of the amino acids leucine, isoleucine and valine is impaired. Untreated, the urine of the child and the child itself smell sweet. Lack of timely treatment leads to vomiting, loss of consciousness, severe developmental retardation and death. Treatment: lifelong protein-poor diet and an 'amino acid preparation'. Prevalence: about 1 child every two years.
- Phenylketonuria (**PKU**): hereditary metabolic disorder in which the amino acid phenylalanine is not broken down. Leads to severe developmental retardation. Treatment: lifelong strict dietary protein restriction, with 'amino acid preparation'. In some cases medication is prescribed. Prevalence: about 12-15 children per year.
- 3-Methylcrotonyl-CoA carboxylase deficiency (**3MCC deficiency**): hereditary metabolic disorder in which certain proteins containing the amino acid leucine are not broken down sufficiently. Can lead to convulsions, developmental retardation and loss of consciousness. Most children only have symptoms when they are ill. Treatment: dietary advice during illness. In some cases, lifelong dietary protein restriction and medicines is needed. . Prevalence: about 1-2 children per year.
- Tyrosinaemia type 1 (**TYR-1**): hereditary metabolic disorder in which the amino acid tyrosine is not broken down properly. Can lead to liver failure, kidney problems, nerve disorders, liver cancer and death. Treatment: lifelong strict protein restriction, amino acid supplements and medication. Sometimes, liver transplantation is necessary. Prevalence: about one child per year.
- Multiple CoA carboxylase deficiency (**MCD**): hereditary metabolic disorder in which proteins in the diet can not be properly converted into useful substances. Can lead to dehydration, loss of consciousness, skin abnormality, baldness, neurological problems, epileptic attacks and immune system defects. Treatment: lifelong administration of vitamin H, sometimes supplemented with dietary protein restriction (or moderate dietary protein restriction). Prevalence: very rare.
- Long-chain 3-hydroxyacyl-coenzyme a dehydrogenase deficiency (**LCHADD**): hereditary metabolic disorder in which long-chain fatty acids cannot be used as an energy source. Problems arise during fasting, overnight sleeping, operations, periods of diarrhoea and vomiting. The low blood sugar level may lead to sleepiness, drowsiness and loss of consciousness, and to muscle problems and cardiac muscle problems. Treatment: prevent that patients do not go for too long without eating, tight mealtime schedule, diet that includes extra carbohydrates and special fats. Prevalence: about 1 child per year.
- Medium-chain acyl-CoA dehydrogenase deficiency (**MCADD**): hereditary metabolic disorder in which medium-chain fatty acids cannot be used as an energy source. Problems arise during

fasting, overnight sleeping, operations, periods of diarrhoea and vomiting. The low blood sugar level may lead to sleepiness, drowsiness, loss of consciousness finally resulting in death. Treatment: ensure that patients do not go for too long without eating. Sometimes extra nutrition and medication is needed. Prevalence: about 10-15 children per year.

- Very long-chain acyl-coenzyme A dehydrogenase deficiency (**VLCADD**): hereditary metabolic disorder in which very long-chain fatty acids cannot be used for energy. Problems arise during fasting, overnight sleeping, operations, periods of diarrhoea and vomiting. The low blood sugar level may lead to sleepiness, drowsiness, loss of consciousness finally resulting in death. Treatment: prevent that patients do not go for too long without eating, tight mealtime schedule, diet that includes extra carbohydrates and special fats. Prevalence: about 2-4 children per year.

Blood disorders:

- Sickle cell anaemia (**SZ**): hereditary haemoglobin abnormality; at low oxygen tension, this leads to abnormally shaped red blood cells, which may clog small capillaries. This results in severe bone pain and organ infarctions (cerebral infarction and pulmonary infarction), plus an increased chance of serious infections, as the spleen does not work properly. The accelerated breakdown of blood results in anaemia. Treatment: analgesics, extra fluids and antibiotics. Blood transfusions may occasionally be required. Prevalence: about 35 children per year.
  Screening for sickle cell anaemia can also detect carrier status for this disease. The parents are informed, if they so wish (affects an average of 850 children per year).
- Alfa-thalassemia (**HbH**): hereditary disorder in which insufficient alpha-globin chains are produced. Children suffer from medium anaemia directly after birth. Treatment: folic acid supplements, blood transfusion. In case a patient is dependent on blood transfusions, stem cells transfusion can be considered. Prevalence: about 1 child per 2 years.
- Bèta-thalassemia major: hereditary disorder in which none or insufficient beta-globin chains are produced. From the third month after birth, progressive severe anemia will arise, which can be life threatening. Treatment: chronical blood transfusion and deferrization, daily folic acid supplements. In case a patient is dependent on blood transfusions, stem cell transfusion can be considered. Prevelance: about 2-5 children each year.

Pulmonary disorder:

- Cystic Fibrosis (**CF**): hereditary disorder in which mucus that is thicker and stickier than normal is produced in various parts of the body. The thick and sticky mucus causes problems in the airways and gastrointestinal tract. Early treatment can help to prevent or diminish these problems. Treatment: medication, caloric diet and physiotherapy. Prevalence: about 30-35 children per year.

2. Target group

The newborn blood spot screening is intended for all newborn babies until the age of six months. Approximately 175,000 newborn blood spot screening tests take place each year. The newborn blood spot screening is carried out as soon as possible, between 72 and 168 hours after birth. In a combined implementation with the newborn hearing screening, this screening is performed from 96 hours after birth. When the blood spot screening test is carried out later than 168 hours after birth, the results may be less reliable.

3. Figures

Each year approximately 175,000 newborn blood spot screening tests are carried out, of which 1/6th of them in hospitals. Participation rate of the blood spot screening has remained stable over time; in 2016 it was 99.2%.

Newborn blood spot screening (Screening in 2016)	Figures
Disease burden (incidence)	About 180-190 children per year.
Size of target group (2016 monitoring report)	174,085
Number of screening tests (2016 monitoring report)	172,754
Participation rate (2016 monitoring report)	99.2%
Number and percentage of referrals: (Source: Tables 3 to 7, MZ is the sum of the values in Table 5, 2016 monitoring report)	Total 647 (0.37%) 27 (0.02%) AGS, 357 (0.21%) CH, 158 (0.09%) MZ, 60 (0.04%) SZ, 45 (0.03%) CF
Detection rates by disorder (per 1000 screened) (Source: 2016 monitoring report)	Total 1.035 0.046 AGS, 0.411 CH, 0.278 MZ, 0.127 SZ, 0.168 CF
Positive predictive value of an anomalous result (Source: 2016 monitoring report)	Total 33% 33% AGS, 22% CH, 30% MZ, 96% SZ, 67% CF
False positives (per 1,000 screened) (source: 2016 monitoring report)	2.2 (n=375)
False negative screenings (per 1,000 screened) (2016 monitoring report)*	0.023 (n=4)
Missed patients (per 1,000 screened) (2016 monitoring report)**	0% (n=0)
Timeliness 1st newborn blood spot screening (% live births where the 1st newborn blood spot screening was carried out <168 hours after birth)	98.8%
Timely intervention (evaluationreport 2016)	Totaal 92% 100% AGS, 93% CH, 92% MZ, 92% SCZ, 93% CF

* Number of children with a newborn blood spot screening disease who were not traced by screening but who did participate in the screening.

** Number of children with a newborn blood spot screening disease who were not traced by screening, where something went wrong during the process that cannot be attributed to the test.

- Disease burden (incidence): number of newly diagnosed cases per year
- Participation rate: the percentage of those invited who actually undergo a screening test.
- Percentage of referrals: the percentage of screened individuals who receive a referral to a hospital for follow-up diagnostic testing.
- Detection rate: the number of abnormalities detected, expressed per number of individuals screened. This is a measure of the probability that a relevant abnormality will be traced.
- Positive predictive value of a referral: the probability that an abnormality (that is clinically relevant) will actually be found following a referral to the hospital.
- False-positives: the number of people given a referral to the hospital (expressed per number of individuals screened), but where no abnormality (that is clinically relevant) is found. In other words, the probability of someone being sent to the hospital unnecessarily.

- False negatives: the probability that, following a negative screening, a relevant abnormality will nevertheless be found (following clinical diagnosis).

  The table below lists all the children traced by blood spot screening in the period from 2007 to 2016.

	2007	2008	2009	2010	2011	2012	2013	2014	2015	2016
AGS	11	8	10	7	8	8	8	6	3	8
CH	57	90	65	95	80	69	72	78	78	71
MZ	70	97	87	61	71	51	47	60	50	48
SZ	41	30	30	41	39	34	35	34	25	22
CF					22	27	21	15	21	29
Total	**179**	**225**	**192**	**204**	**217**	**189**	**183**	**193**	**177**	**178**

4. Implementation

Process

Selection

- After its birth has been officially registered by its parents, RIVM's regional offices (DVP) receive the child's details from the local authority's primary administration. The RIVM's regional offices can also receive the birth registration and child information via administration of midwives and the Dutch Council for Refugees.

Invitation

- The RIVM regional offices inform the contracted YHC (Youth Health Care) organisation, which then phones the parents to make an appointment.
- In the third trimester of pregnancy, the obstetric care providers (midwives, gynaecologists and general practitioners who attend deliveries) provide information about the newborn blood spot screening, based on a national information consultation checklist for midwives. At this point, the information leaflet is handed to the expectant parent (or parents). The choice of whether or not to be informed about the child's carrier status for sickle cell anaemia is specifically discussed at this stage, as is the opportunity to object to the use of blood spot material for scientific research.

The screening test

- The newborn blood spot screening is carried out between 72 and 168 hours after birth, most often simultaneously with the newborn hearing screening. The newborn blood spot screening is performed at the place where the child is located at that time. Most children are pricked at home.
- During the newborn blood spot screening, a special device (lancet) is used to take a few drops of blood from the baby's heel, which are then placed on a heel prick blood test card.
- The heel prick blood test card is then sent to one of the five screening laboratories. The laboratory carries out the tests and reports the results to RIVM's regional office.
- About 100 times a year, as part of the screening method for cystic fibrosis, the submitted blood is sent for extensive DNA analysis of the CFTR gene to VU University Medical Center, Department of Clinical Genetics.
- When the results of the newborn blood spot screening are favourable, the parents are not notified.

Referral

- In the case of an anomalous result, the RIVM regional office's medical advisor contacts the general practitioner, who then arranges a referral to an academic centre (except in the case of CH, for which the children are also referred to general hospitals).
- A definite diagnosis is made at the medical centre, and treatment is started.

Who is involved in the population screening programme?

- At national level, the screening programme is organised by the RIVM's Centre for Population Screening (CvB), on behalf of the Dutch Ministry of Health, Welfare and Sport. The document 'Policy framework of the prenatal and neonatal screening' provides insight into the legal and policy based frameworks of the screening programs. The document describes the cooperation of all preparatory, executive and decision-making parties involved in the pre- and neonatal screening. Please refer to the following website: https://www.rivm.nl/en/Documents_and_publications/Scientific/Reports/2018/mei/Policy_framework_for_Prenatal_and_Neonatal_Screening.
- RIVM DVP's regional offices organise the regional implementation.
- RIVM-DVP buys and distributes the materials needed and manages the information system Praeventis.
- The obstetric care provider (midwife, gynaecologist or general practitioner who attends deliveries) informs the expectant parent (or parents) about the newborn blood spot screening and hands over a copy of the national leaflet.
- An official of the civil affairs department hands over another copy of the leaflet when the birth is officially registered.
- The drops of blood are collected by screeners from YHC organisations. In regions Gelderland and Zuid-Holland, the newborn blood spot screening is also carried out by midwives working under the responsibility of the YHC organisation. In Twente, the newborn blood spot screening is also carried out by maternity carers under responsibility of the YHC organisation.
- If, during the period when drops of blood have to be collected, a child is in the hospital, then the newborn blood spot screening will be carried out by a member of the hospital's staff.
- The blood test is carried out by five contracted screening laboratories. The RIVM-GZB serves as a reference laboratory.
- About 100 times a year, when screening for cystic fibrosis, supplementary DNA diagnosis of the CFTR gene takes place at VU University Medical Center, Department of Clinical Genetics.
- The annual monitoring of the screening is carried out by TNO Child Health, at the instigation of RIVM CvB.
- The Programme Committee for Newborn Blood Spot Screening, which was established by the RIVM-CvB, advises the RIVM regarding the programme's national co-ordination. The Programme Committee is composed of experts from relevant professional groups and organisations who are recognised authorities in their own field or network, and who have professional contacts in the field.
- The board of the Dutch Paediatrics Society (NVK) has established the following advisory committees for the newborn blood spot screening: the AGS-CH Advisory Committee (ANS-AGS-CH), the Metabolic Disorders Advisory Committee (ANS-MZ), the Haemoglobinopathies Advisory Committee (ANS-HbP) and the Cystic Fibrosis Advisory Committee (ANS-CF). These committees also advise the Programme Committee and are responsible for setting up the guidelines for diagnosis and treatment.

Link to care

In principle, all children who are referred from screening are treated at the academic centres (or, in the case of CH, at the general hospitals as well).
In case of an anomalous result regarding AGS, the child will be seen by the pediatrician-endocrinologist as soon as possible, but no later than 12:00 AM upcoming day[1].

[1] For AGS and CH applies: the result of the first blood spot screening can be negative (no action), anomalous (referral) or non-conclusive. When the result is non-conclusive, a second heel prick will take place. The result of the second blood spot screening can be negative (no action) or anomalous (referral).

In case of an anomalous result regarding CH, the child will be seen by the pediatrician-endocrinologist at the same day but no later than 12:00 AM of the upcoming day. Collegial consultation can take place when necessary [1].
In case of an anomalous result regarding CF, the child will be seen by a pediatrician-pulmonogist of the CF-Centre within 1 week, dependent on the planned date of the sweat test.
In case of an anomalous result regarding a metabolic disease, the child will be seen by the pediatrician-metabolic diseases as soon as possible.
In case of an anomalous results regarding sickle cell disease, alpha-thalassemia (HbH-disease) and beta-thalassemia major, the child will be seen before the age of 4 weeks by the pediatrician-hematologist.

If the result indicates carrier status for sickle cell anaemia, the parents of the child are invited by the general practitioner for an informative consultation within 4 weeks. During this consultation, the referral to the department of Clinical Genetics is discussed. .

Benefits and disadvantages of participation in the programme

Benefits

The newborn blood spot screening traces rare but serious disorders. These disorders can not be cured but they can be treated, provided that they are traced in time. This will prevent or limit any damage to the child's health, resulting in substantial health gains. An added benefit is that this avoids a lengthy, burdensome diagnostic process.

Disadvantages

Soon after its birth, parents are confronted with the fact that their child may have a serious, rare disorder. Moreover, in the case of a suspected disorder, the child will usually be quickly seen by a paediatrician. This causes anxiety for the family. The result in question may be a false-positive, in which case there may – ultimately – be nothing wrong with the child. Also false-negative cases can occur which means that sick children are missed. Details of these missed patients are registered by the RIVM regional office's medical advisor in the national database NEORAH. Paediatricians report details of missed patients by the screening in a national registry, via the Dutch Paediatric Surveillance Centre at The Dutch Paediatrics Society at the NVK. These reports are included in the annual evaluation of the programme by TNO Child Health, at the instigation of RIVM-CvB.

Secondary findings

Carnitin Transporter (OCTN2) deficiency is a secondary finding in the neonatal blood spot screening. In addition, screening for sickle cell anaemia can also detect carriers. From the child's point of view, this can be seen as a disadvantage. From the parents' point of view, and that of other relatives, this can be seen as a benefit, with a view to possible future pregnancies. Yet it is also a disadvantage, as this information forces parents to face a choice that they might not have wanted to face.

5. History

Screening for phenylketonuria (PKU), using blood from the newborn blood spot screening, has been carried out in the Netherlands since 1974. Since 1981, this blood is also tested for congenital hypothyroidism (CH) and, since 2000, for adrenogenital syndrome (AGS).
In November 2005, State Secretary Ross of the Dutch Ministry of Health, Welfare and Sport announced that tests for additional diseases are to be introduced. Based on a Health Council of the Netherlands' advisory report submitted in August 2005, screening for 14 additional disorders would commence on 1 January 2007. In 2010, the Health Council of the Netherlands recommended that cystic fibrosis (CF) be added to the blood spot screening. With effect from 1 May 2011, CF was also added to the screening. Screening for homocystinuria (HCY) was

terminated with effect from 1 April 2016, at the recommendation of the Health Council of the Netherlands. It had been suspended since October 2010, due to the large number of false-negative results obtained by the screening method in use at that time. Per 1 January 2017, alfa-thalassemia (HbH-disease) and beta-thalassemia major were added to the blood spot screening.

6. Developments

Fewer false-positives
Due to the optimisation of the screening methods, the number of children wrongly referred for diagnosis based on an anomalous newborn blood spot screening result has declined in recent years.

Implementation of blood spot screening in the Caribbean Netherlands
In October 2013, the Dutch Minister for Health, Welfare and Sport decided to introduce blood spot screening in the Caribbean Netherlands. The RIVM CvB has prepared the implementation of blood spot screening in the Caribbean Netherlands, at the instigation of the Dutch Ministry of Health, Welfare and Sport. Newborn blood spot screening commenced on Bonaire on 1 January 2015. St. Eustatius and Saba followed in October of that year.

Policy on the further use of bodily material
In anticipation of general legal regulations on the use (or further use) of bodily material (Control over Body Materials Act), clear policies should be established for population screening programmes with respect to the retention and use of bodily material taken in the context of screening programmes for objectives that fall within the scope of screening (primary diagnosis and follow-up diagnostic testing, internal quality control and improvement, education and training) and other purposes (further use). The current situation regarding the further use of bodily material is that, for non-traceable bodily material, if no objection is made then consent is assumed. In the case of the further use of traceable bodily material, seeking consent is mandatory. Procedures for use of the remainder of blood spot material collected during the heel prick, are stated on the website www.rivm.nl/hielprik/professionals.
At the instigation of the RIVM CvB, a description has been prepared of the legal conditions governing the storage and use of bodily material taken in the context of population screening programmes. In 2012/2013, the CvB submitted an advisory report to the Dutch Ministry of Health, Welfare and Sport concerning working with bodily material in screening programmes. Last May and June 2017, the Dutch Ministry of Health, Welfare and Sport has organized an internet consult regarding the law proposal about bodily material. In this period, discussions and meetings took place between RIVM-CvB and the Dutch Ministry of Health, Welfare and Sport regarding population screening programmes. Pending the decision of the Dutch Ministry of Health, Welfare and Sport about this new law proposal, the existing procedure will need to be modified accordingly.

Health Council of the Netherlands advisory report entitled 'Neonatal screening: new recommendations'.
On 8 April 2015, the Health Council of the Netherlands issued a wide-ranging advisory report on blood spot screening. The Health Council of the Netherlands recommends that the newborn blood spot screening should be extended with another fourteen disorders, that screening for homocystinuria should be terminated, that no non-treatable disorders should be included in the screening programme, and that the procedure of reporting carrier status for sickle cell anaemia should be terminatedOn 9 July 2015, the former Dutch Ministry of Health, Welfare and Sport published a policy position paper entitled 'Newborn blood spot screening'. The

Minister at that time, planned to extend the newborn blood spot screening by including another 14 disorders. However, the Minister asked RIVM CvB to carry out a feasibility study first, to determine the practicalities involved. To support the fast implementation of the alpha- and beta-thalassemia to the screening programme, the RIVM-CvB produced the first part of the feasibility report in November 2016. Since 1 January 2017, both diseases are included to the blood spot screening.
The second part of the feasibility report, which has been set up in close cooperation with all relevant parties, is provided to the Ministry of Health, Welfare and Sport on 6 July 2017. On 21 December 2017, the minister informed the house of representatives about the decision to extend the blood spot screening with 12 diseases. The implementation of these diseases will take place in phases, distributed over the period of 2018-2022. For each of these 12 diseases, additional research is needed. On 1 April 2018, the regional pilot study (SONNET) was started in order to investigate the implementation of the very severe and rare immune deficiency disease (SCID).
In the policy statement of 9 July 2015, it was requested to investigate the possibilities of inclusion of untreatable diseases to the blood spot screening. On 21 December 2018, the Minister of Health, Welfare and Sport stated that it is too early to start a feasibility study about the introduction of untreatable diseases. The minister asks the Health Council to advice on the conditions among which such diseases could be included in the programme.
Regarding the procedure of reporting carrier status for sickle cell anaemia, the former policy will not be changed. This means that the carrier status will be reported in case parents do not object. The minister will not yet change this policy and will come back to this topic later on.

Flexible Decision-making
The former Minister indicated in the policy statement of 9 july 2015 that, in the case of newborn blood spot screening, she wants to be able to respond more quickly to innovations to achieve rapid health gains. For this reason, she plans to seek the Health Council of the Netherlands' advice more frequently, as and when this is necessitated by new developments.

7. Financial

Since 1 January 2015, carrying out blood spot tests has been financed from the State Budget. In 2018, the charge for a newborn blood spot screening set was €3.62. Based the number of live births, it was estimated that 176,163 newborn blood spot screenings would be carried out. The implementation costs include the cost of collecting the blood from the child and the cost of laboratory tests. The YHC organisation receives €20.71 per child to cover the cost of collecting blood. Laboratory testing costs €52.14 per heel prick blood test card. RIVM-DVP's organisational costs are estimated to be about €4 million, while RIVM's co-ordination role costs around €1.76 million per year. The programme's total annual turnover is approximately €18.8 million. Referrals for diagnosis and treatment are financed as mainstream care.

8. International

In terms of their content, newborn screening programmes can sometimes differ substantially from one country to another, and even from one part of a country to another. In North America and most of Europe, as well as parts of Latin America, Japan, Australia and New Zealand, newborn screening is part and parcel of mainstream health provision. The number of clinical pictures covered by these screening programmes varies from two to more than 40.
In the rest of Europe, Latin America, the Middle East/North Africa and some Asian countries, work is in progress to bring such provision up to standard, but it will probably still take some considerable time before this can be achieved.

Throughout most of Africa, virtually nothing is being done in terms of newborn screening, nor is any action on this matter expected in the near future.
Given its current package (supplemented with hearing screening), the Netherlands ranks in Europe among the countries that screen for the largest number of disorders, along with Germany, Austria and Spain. France screens on five disorders (PKU, CH, AGS, SZ and CF), as does Great Britain (PKU, CH, SZ, CF and MCAD).

9. Websites

www.rivm.nl/hielprik (public)
www.rivm.nl/hielprik-professionals (professional)

10. Contact

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