

NEWBORN SCREENING

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What's the newborn screening?

- The nationwide newborn screening (NS)
= **preventive program of testing newborns** for increased risk of certain rare diseases
- NS tests are done by **measuring metabolites** in samples of whole blood collected on special filter paper („newborn card“ or „Guthrie card“) – **analysis of dry blood spots**

Test card

WhaTman 903™
6131313W113

WhaTman 903™
LOT 6961313W113 SN 1207715CZ
2016-09

Nedotýkejte se testovací zóny s tečkami.
Poškozenou kartičku nepoužívejte.

Kartičku vyplňte před odběrem. Použijte kulčičkové pero a dostatečný tlak k samoprůpisu. Oddělené kartičky odešlete po odběru na adresy příslušných laboratoří: jeden list na DMP a jeden list na KH+CAH+CF.

Odběr první: Důvod opakování (rescreeningu):
Opakovaný:

Novorozenec Jméno + Příjmení Pohlaví Ž M

Rodné číslo	Dítěte nebo matky - nutno vyplňat!	Pojišťovna	Porodní hmotnost g	Gest.věk týdny

Datum DD.MM. Datum a čas narození DD.MM.RRRR - HH.MM Datum a čas odběru DD.MM.RRRR - HH.MM

Kód Kódové číslo odběru Kód oddělení (AAA) + pořadí odběru (113) -AAA113 Praktický dětský lékař Jméno, telefon, město

Matka novorozence Jméno + Příjmení

Telefon matky (rodiny) Adresa matky (pobytu)

Odesílatel vzorku Čitelné razítko, jmenovka, podpis

CE IVD REP 10539735 Rev.AF GE Healthcare Ltd Forest Farm Industrial Estate, Cardiff, CF14 7YT, UK 14 Webster Way Westborough, MA 01581-0119, USA

History

- NS debuted in US in **1960**
- **Robert Guthrie** (1916 – 1995)
- American microbiologist, developed a simple method using a bacterial inhibition assay that could detect high levels of phenylalanine in blood shortly after a baby was born.
- Guthrie also used the collection of blood on filter paper which could be easily transported.



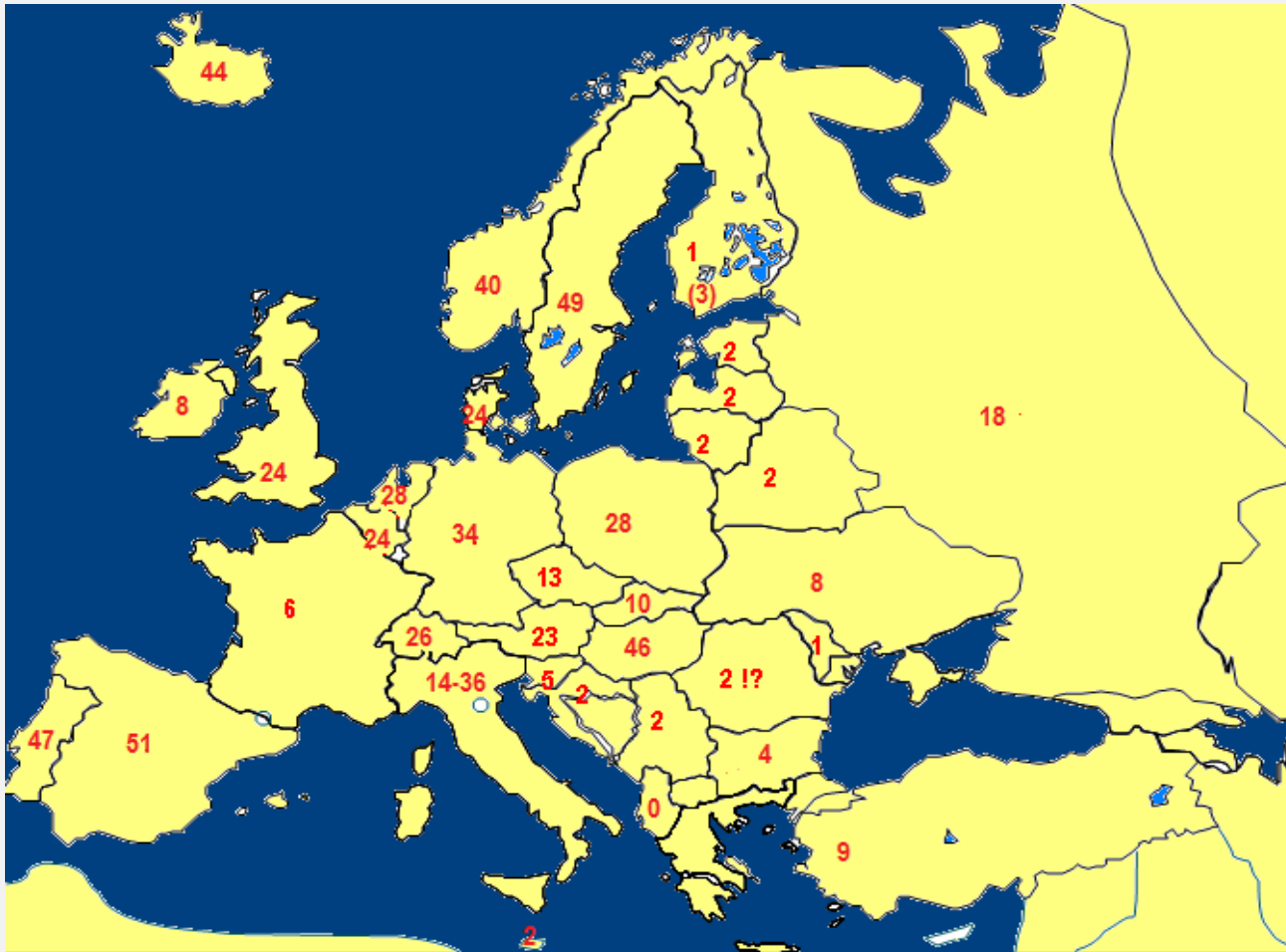
History of NS in the Czech Republic

- 1975 - Phenylketonuria
- 1985 - Congenital hypothyroidism
- 2006 - Congenital adrenal hyperplasia
- 2009 - Cystic fibrosis and 9 additional metabolic diseases
- 2016 – further expansion of NS program (5 diseases: argininemia, citrulinemia, 2 types of homocystinuria, biotinidase deficiency)

At present there are **18 diseases tested** in the Czech Republic.

NS program is covered by health insurance

NS Europe - 2015



•Zdroj: Therrell BL et al: Current status of newborns screening worldwide: 2015. Seminars₆ in Perinatology 2015;39: 171-87

Methods of analysis

- **Fluoroimmunoassays (method DELFIA)**
 - congenital hypothyroidism (TSH)
 - congenital adrenal hyperplasia (17 OHP)
 - cystic fibrosis (IRT)
- **Tandem mass spectrometry (MS/MS)**
 - phenylketonuria and other disturbances of amino acid metabolism (aminoacids)
 - disorders of fatty acid oxidation (acylcarnitines)
- **Molecular techniques**
 - second step of NS CF

Automatic analyzer AutoDELFIA



Tandem mass spectrometer



Newborn screening Laboratories in the Czech Republic

For immunoanalytic method

- University Hospital Kralovské Vinohrady, Prague
- University Hospital, Brno

For tandem mass spectrometry

- General University Hospital, Prague
- University Hospital, Olomouc

Sample collection

- At the age of **48-72 hours after birth** several drops of capillary blood from the heel of the child are sampled on a special paper (newborn card)
- After the collection of blood it has to **dry well, avoid contamination**
- These cards are sent for analysis to the screening laboratories



15°C
- 22 °C



Sample collection



Screening results

Evaluation of results – **cut off = limit value**

- Negative result – no suspicion for any the screened disease
- Positive result – **indicates only increased risk of a disease**
 - It is necessary to confirm this positive result, usually by use further laboratory tests

Results of NS in the Czech Republic 2010-2018

CH	334
PKU/HPA	184
CF	150
CAH	84
MCADD	43
LCHADD	13
VLDADD	5
GA I	5
IVA	5
MSUD	5
CPT I	0
CPTII/CACT	0
ARG *	0
CIT *	0
BTD *	29
CBS *	1
MTHFR *	0
Total	858

* Diseases included to NS program since June 1st, 2016

Case – positive NS CF

- newborn – birth in term, birth weight 3800g
- results of NS:
 - TSH < 2,5 mIU/L → NS CH negative
 - 17OHP = 16 nmol/L → NS CAH negative
 - **IRT = 230 ng/mL** > cut off → molecular tests
 - **2 mutations in CFTR gene** [F508del]+[F508del]
 - sweat test – positive, **Cl = 106 mmol/L** (cutt off 30)
 - **dg. CF positive**

Case – positive NS CAH

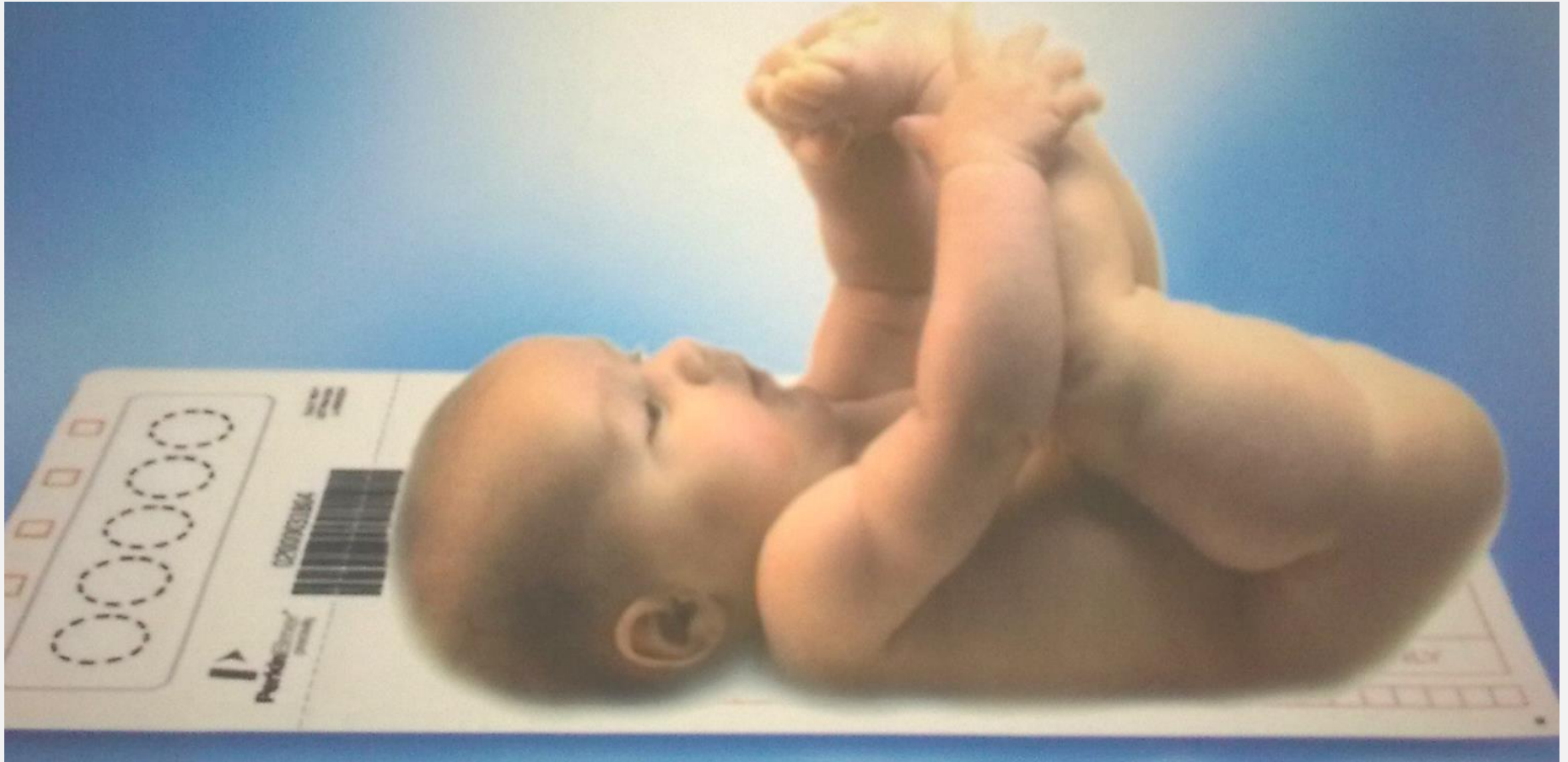
- newborn – birth in term, birth weight 3350g
- results of NS:
 - TSH < 2,5 mIU/L → NS CH negative (cut off=15)
 - IRT = 12,8 ng/mL → NS CF negative (cut off=65)
 - **17OHP > 315 nmol/l (700 nmol/l) → positive (cut off 20)**
 - **S/Na = 128 mmol/l, S/K = 5,6 mmol/l, ACTH = 872 pg/ml, 17 OHP > 300 nmol/l (in venous blood)**
 - **dg. CAH, v.s. type SW**

Website

For more information about newborn screening program in the Czech Republic you can contact website:

www.novorozeneckyscreening.cz

(English version)



Thank you for your attention