



VFN PRAHA
VŠEOBECNÁ FAKULTNÍ
NEMOCNICE

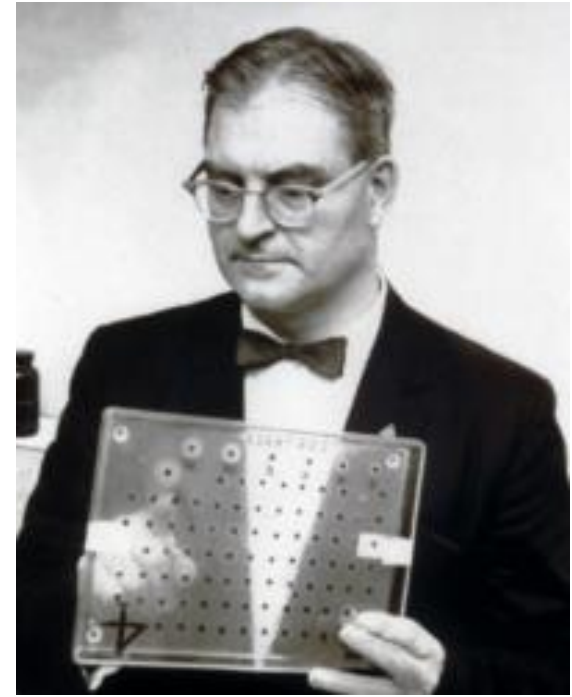
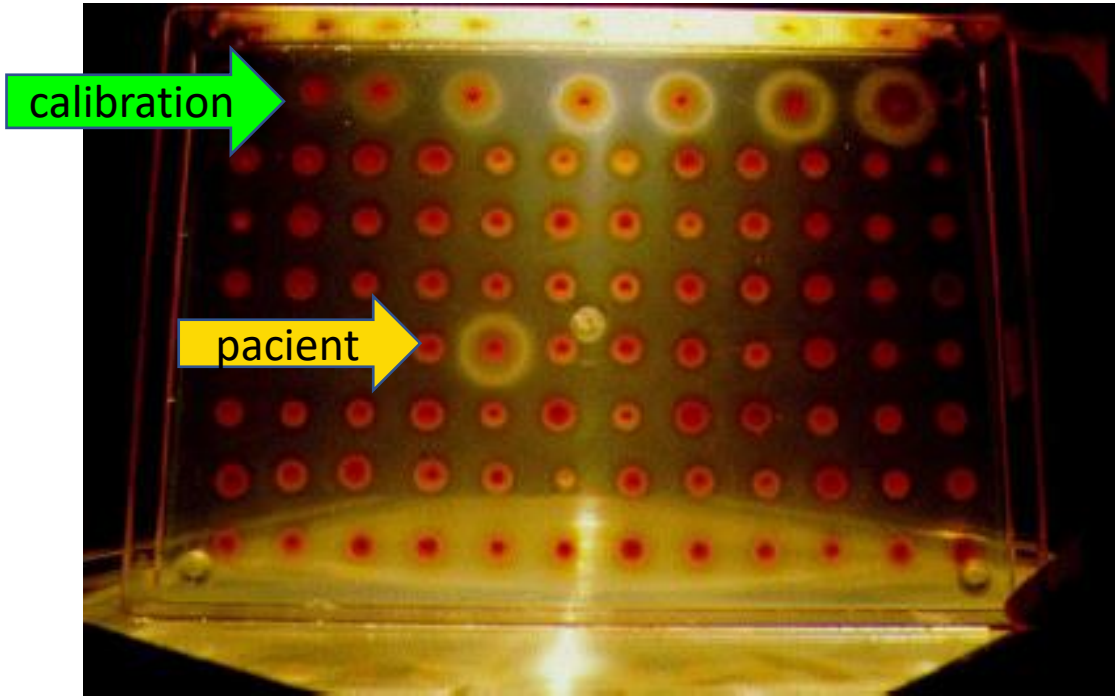
Screening in paediatrics

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First Faculty of Medicine, Charles University and General University
Hospital in Prague

Laboratory newborn screening (NBS)

First screening program for neonates



- Dry blood spot testing
- Beta-2-thienylalanin - B.subtilis growth inhibition
- ↑Phe enables bacteria to growth

Prof. Robert Guthrie 1916-1995

Pediatrics 1963

Principles of screening for diseases

PRINCIPLES AND PRACTICE OF SCREENING FOR DISEASE

J. M. G. WILSON

*Principal Medical Officer, Ministry of Health,
London, England*

G. JUNGNER

*Chief, Clinical Chemistry Department, Sahlgren's Hospital,
Gothenburg, Sweden*



WORLD HEALTH ORGANIZATION

GENEVA

1968

- **An important health problem and there should be a recognisable latent stage**
- **A suitable and acceptable test to the population**
- **An accepted treatment and agreed policy on whom to treat**
- **The cost of screening should be economically balanced in relation to expenditure on medical care**

Quality assurance in screening programmes

J A Muir Gray and June Austoker

Institute of Health Sciences, Oxford, UK

All screening programmes do harm; some also do good. The responsibility of the policy-maker is to decide which programmes do more good than harm at reasonable cost and then introduce them, once they are confident that the screening programme could and will reach the standard of quality required for success. The ratio of benefit to harm is not, however, constant and this relationship demonstrates a shifting balance.

Newborn screening in Czechia

1975	Phenylketonuria (PKU/HPA)
1985	cong. hypothyreosis (CH)
2006	cong. adrenal hyperplasia (CAH)
2009	Cystic fibrosis (CF) + 9 IEM
2016	+ 5 IEM
2024	+ SMA, SCID

SMA AND SCID

SCID – Severe combined immunodeficiency) – maturation disorder a or T cell function (1:58 000)

Absence TREC – T cell receptor excision circles

Absence KREC – k-deleting recombination excision circles

SMA – Spinal muscular atrophy – *SMN1* gene
(1:6000-10 000)

Homozygous deletion of exon 7. *SMN1*

Nusinersen (Spinraza) or onasemnogen abeparvovec (Zolgensma)

Pilot study SMA a SCID

2022-2023

185,355 newborns

22 SMA

18 PID

Newborn screening (NBS) since 2024

n=20

Endokrinopathies

hypothyreosis
cong. adrenal
hyperplasia

Cystic fibrosis

SMA
SCID

IMD (IEM)

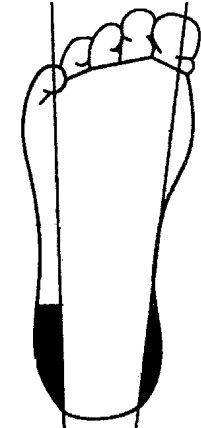
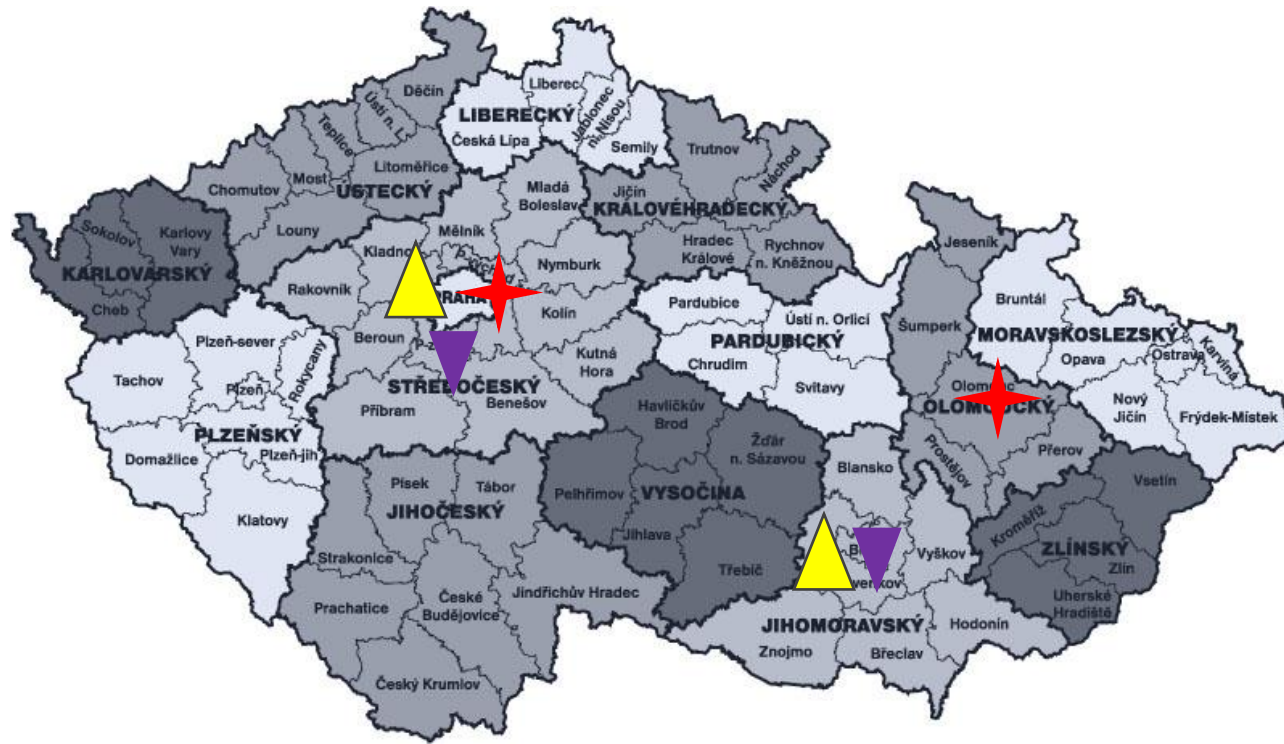
PKU/HPA

4 aminoacidopathies

3 organic acidurias

6 fatty acid
ox.disorders
*biotinidase
deficiency*

NBS blood sampling



Card 1: 0050598

Kartičku vyplnit před odběrem Nedotýkat se oblasti pro kapky krve Při poškození kartičku nepoužít	
Jméno novorozence	153173
Rodné číslo, pečlivě	075902/1972
Datum a čas narození	03.06.00
Datum a čas odběru	29.06.00
Kódové číslo odběru	64.000
Praktický ošetřitel	M. Novotná
Jméno matky	M. Novotná
Telefon matky (rodina)	77
Adresa matky (pobytu)	Čes. 11111111
Odeslatel vzorku	Podolská 157/1 PČ 147 00 PRAHA - PODOLÍ pediatrická pracoviště / POK /

Card 2: 0050598

Kartičku vyplnit před odběrem Nedotýkat se oblasti pro kapky krve Při poškození kartičku nepoužít	
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endocrinopathies

CF genetics

IEMetab (Inborn Errors of Metabolism)

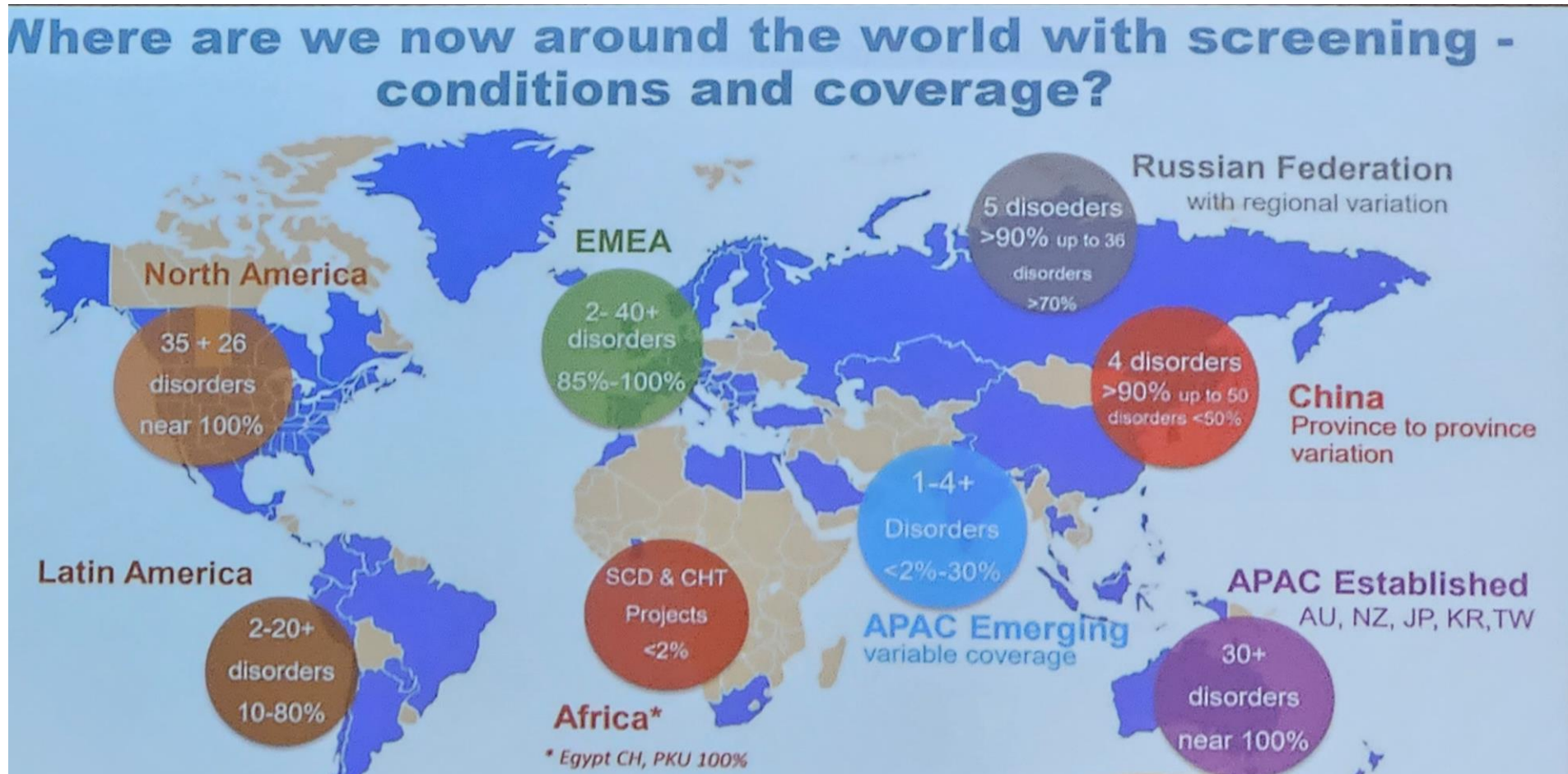
1.53 milion newborns (2010-2023)

CH	1:2,800
PKU/HPA	1:4,856
CF	1:6,218
BTD	1:9,905
FAOD	1:15,500
Other IEM	1:77,500
SMA	1:6,800
SCID	1:59,100

CSF-SPID	84
Hydroxyprolinemia	4
Argininosucc.aciduria	4
Vit B12 deficiency	5
MTP deficiency	1

Neonatal prevalence 1:1072

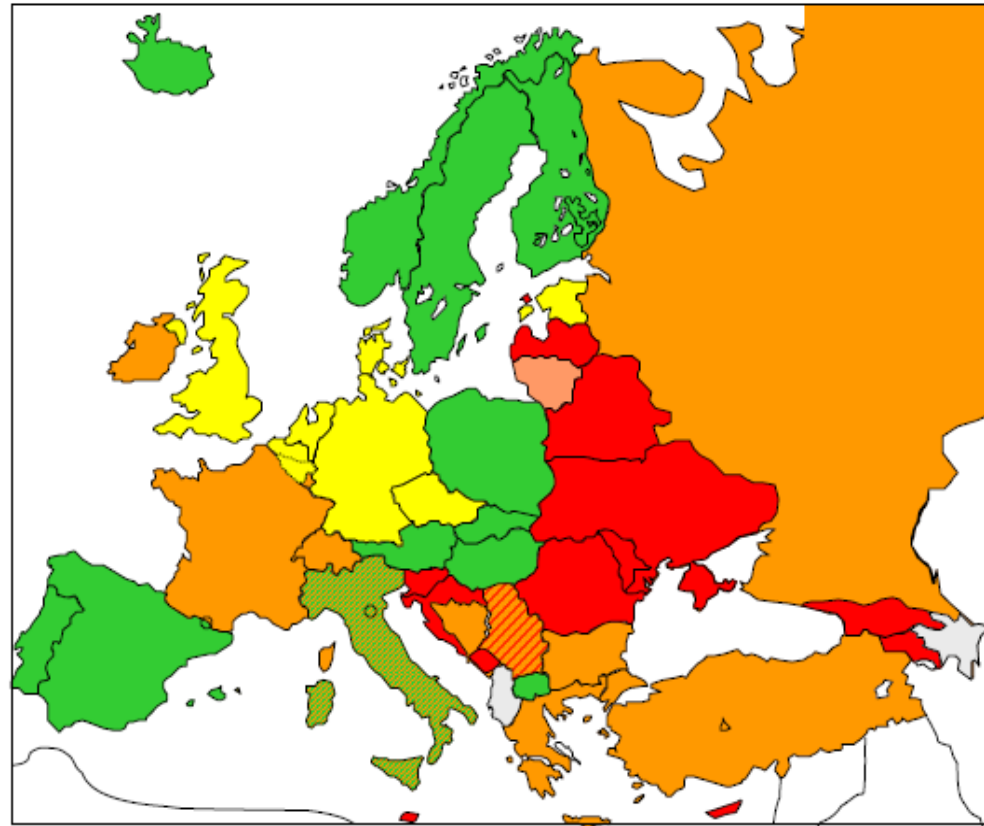
Coverage of populations with NBS



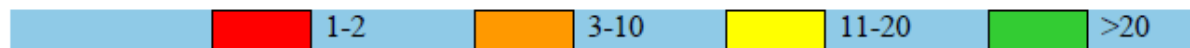
28 % of newborns born

Among-country variation of NBS

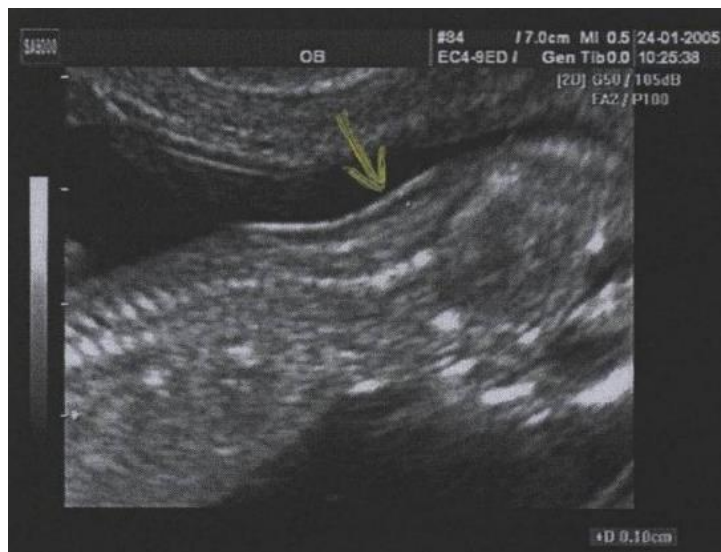
Number of conditions per country



2016, G.Loeber, ISNS



Screening in neonatology and paediatrics



PAPP-A, AFP, bHCG
estriol/inhibin A



Screening – 1. trimestr

	Euploidní	T21	T18	T13
NT mm	2,0	3,4	5,5	4,0
Fb hCG	-	↑	↓	↓
PAPP-A	-	↓	↓	↓
Abs.nosní kosti %	1,4	69	53	45



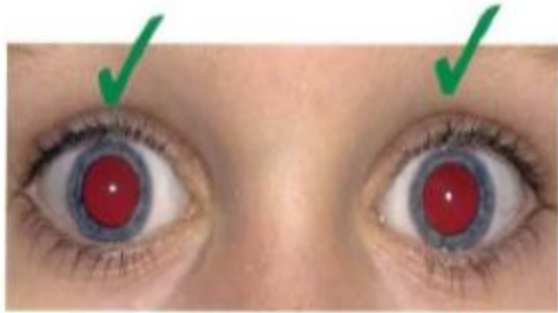
Screening – 2. trimestr

↑AFP - VVV nekryté kůží

↓AFP ↓ uE3 ↑ hCG riziko T21

↓AFP ↓ uE3 ↓ hCG riziko T18, T13

Screening in neonatology



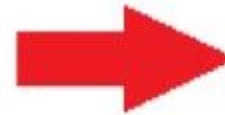
Normal reflex



No action required



Red reflex absent



See your GP urgently



Red reflex abnormal

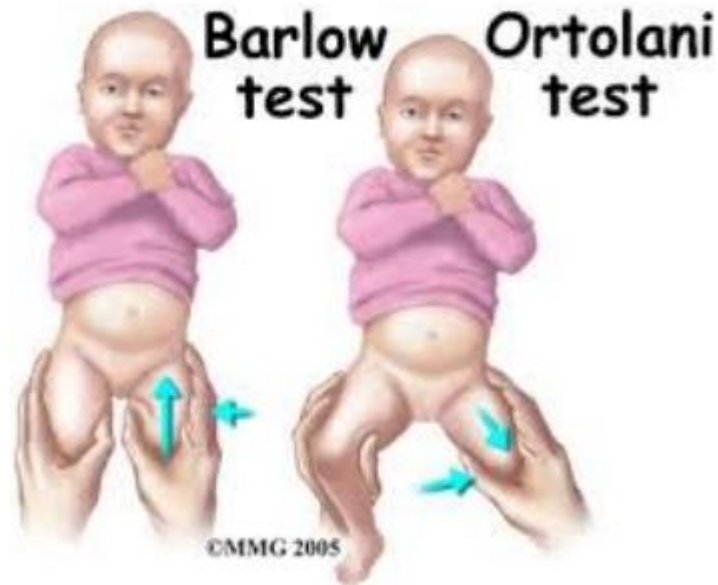


See your GP urgently

Screening in neonatology

Developmental Displasia of the hip – the „triple sieve“ method

Dislocation maneuver/provocative test Repositioning maneuver



Frejka pillow



Pavlik harness



According to USG: (bony acetabulum, labrum, femoral epiphysis)
Ia, Ib – fyz.; IIa+ – immature hip; IIa-, IIb – mild displasia;
IIc – displasia; IId, IIIa, IIIb, IV – decentration

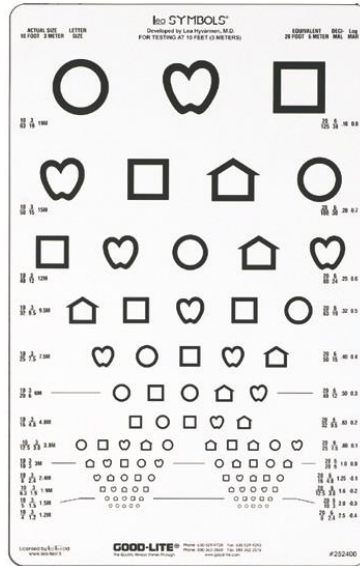
Screening in pediatrics

Plusoptix



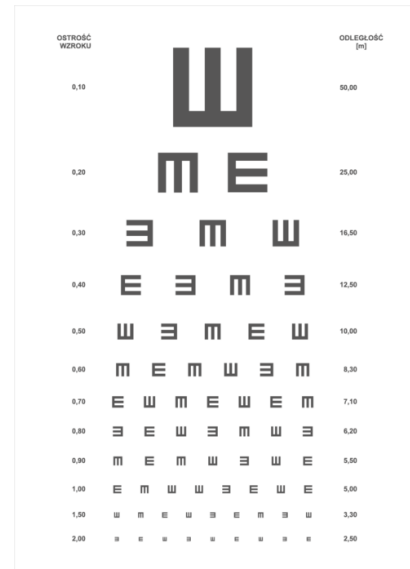
1-2 yrs

Lea symbols



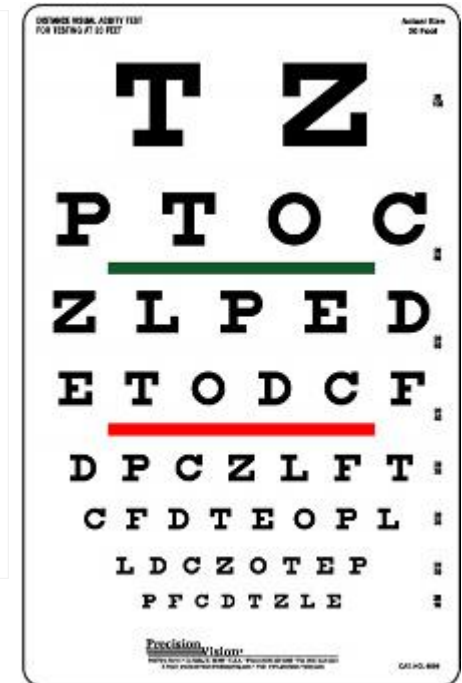
3 yrs

Pflüger hooks



4-6 yrs

Snellen optotype



7 yrs

- Otoacoustic emissions
- USG of the kidney
- hearing and sight examination in GP
- Evaluation of psychomotor dev., blood pressure in GP
- Lipidogram at 5yrs and 13yrs if pos. family history
- Screening for obesity

Modifikovaný dotazníkový test pro záchyt poruchy autistického spektra u batolat

Přeloženo se souhlasem autora z anglického originálu:

Modified Checklist for Autism in Toddlers, Revised, with Follow-Up

(M-CHAT-R/F)TM

Diana L. Robins, Ph.D.
Deborah Fein, Ph.D.
Marianne Barton, Ph.D.

0-2 points low risk of ASD
3-7 moderate risk of ASD
8-20 high risk of ASD

At the 18-month health maintenance visit

M-CHAT-RTM

Please answer these questions about your child. Keep in mind how your child usually behaves. If you have seen your child do the behavior a few times, but he or she does not usually do it, then please answer **no**. Please circle **yes** or **no** for every question. Thank you very much.

1. If you point at something across the room, does your child look at it? (FOR EXAMPLE , if you point at a toy or an animal, does your child look at the toy or animal?)	Yes	No
2. Have you ever wondered if your child might be deaf?	Yes	No
3. Does your child play pretend or make-believe? (FOR EXAMPLE , pretend to drink from an empty cup, pretend to talk on a phone, or pretend to feed a doll or stuffed animal?)	Yes	No
4. Does your child like climbing on things? (FOR EXAMPLE , furniture, playground equipment, or stairs)	Yes	No
5. Does your child make <u>unusual</u> finger movements near his or her eyes? (FOR EXAMPLE , does your child wiggle his or her fingers close to his or her eyes?)	Yes	No
6. Does your child point with one finger to ask for something or to get help? (FOR EXAMPLE , pointing to a snack or toy that is out of reach)	Yes	No
7. Does your child point with one finger to show you something interesting? (FOR EXAMPLE , pointing to an airplane in the sky or a big truck in the road)	Yes	No
8. Is your child interested in other children? (FOR EXAMPLE , does your child watch other children, smile at them, or go to them?)	Yes	No
9. Does your child show you things by bringing them to you or holding them up for you to see – not to get help, but just to share? (FOR EXAMPLE , showing you a flower, a stuffed animal, or a toy truck)	Yes	No
10. Does your child respond when you call his or her name? (FOR EXAMPLE , does he or she look up, talk or babble, or stop what he or she is doing when you call his or her name?)	Yes	No
11. When you smile at your child, does he or she smile back at you?	Yes	No
12. Does your child get upset by everyday noises? (FOR EXAMPLE , does your child scream or cry to noise such as a vacuum cleaner or loud music?)	Yes	No
13. Does your child walk?	Yes	No
14. Does your child look you in the eye when you are talking to him or her, playing with him or her, or dressing him or her?	Yes	No
15. Does your child try to copy what you do? (FOR EXAMPLE , wave bye-bye, clap, or make a funny noise when you do)	Yes	No
16. If you turn your head to look at something, does your child look around to see what you are looking at?	Yes	No
17. Does your child try to get you to watch him or her? (FOR EXAMPLE , does your child look at you for praise, or say "look" or "watch me"?)	Yes	No
18. Does your child understand when you tell him or her to do something? (FOR EXAMPLE , if you don't point, can your child understand "put the book on the chair" or "bring me the blanket"?)	Yes	No
19. If something new happens, does your child look at your face to see how you feel about it? (FOR EXAMPLE , if he or she hears a strange or funny noise, or sees a new toy, will he or she look at your face?)	Yes	No
20. Does your child like movement activities? (FOR EXAMPLE , being swung or bounced on your knee)	Yes	No

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Answer NO – risk of ASD
in 2, 5 a 12 – YES risk of ASD