



VFN PRAHA
VŠEOBECNÁ FAKULTNÍ
NEMOCNICE

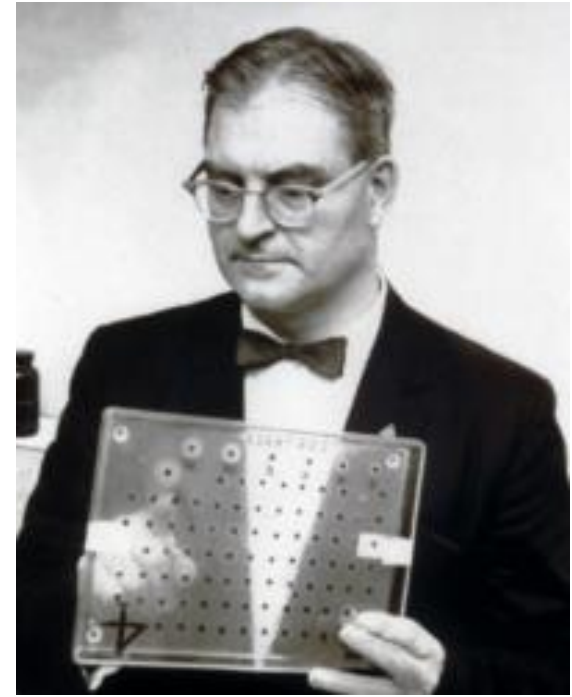
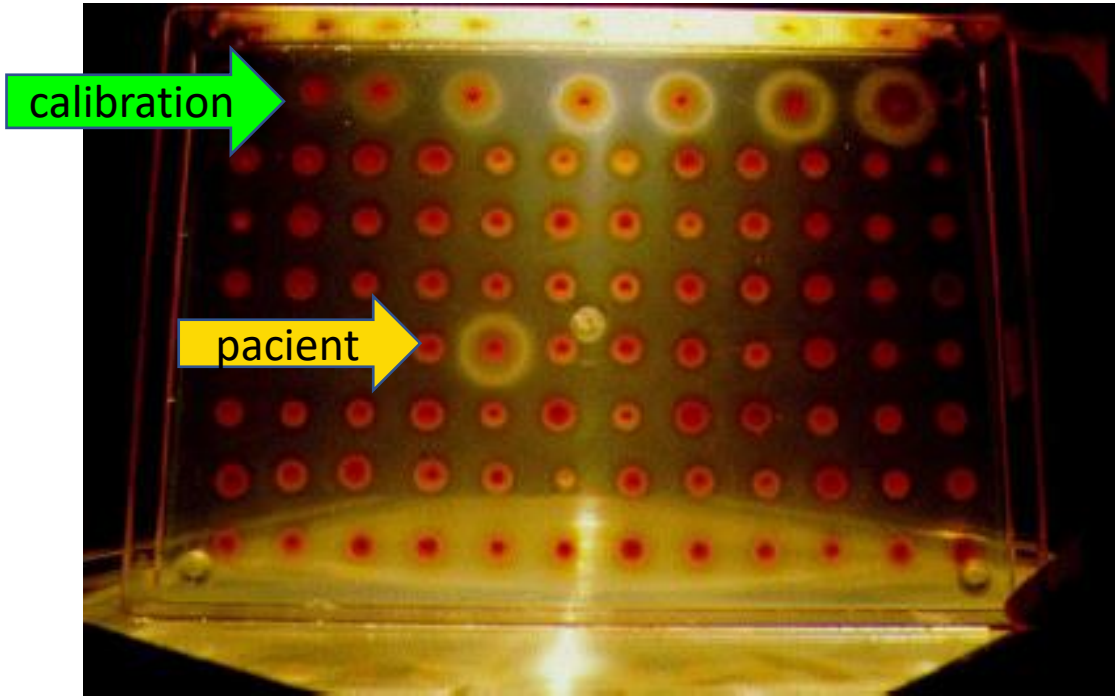
Screening in paediatrics

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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*



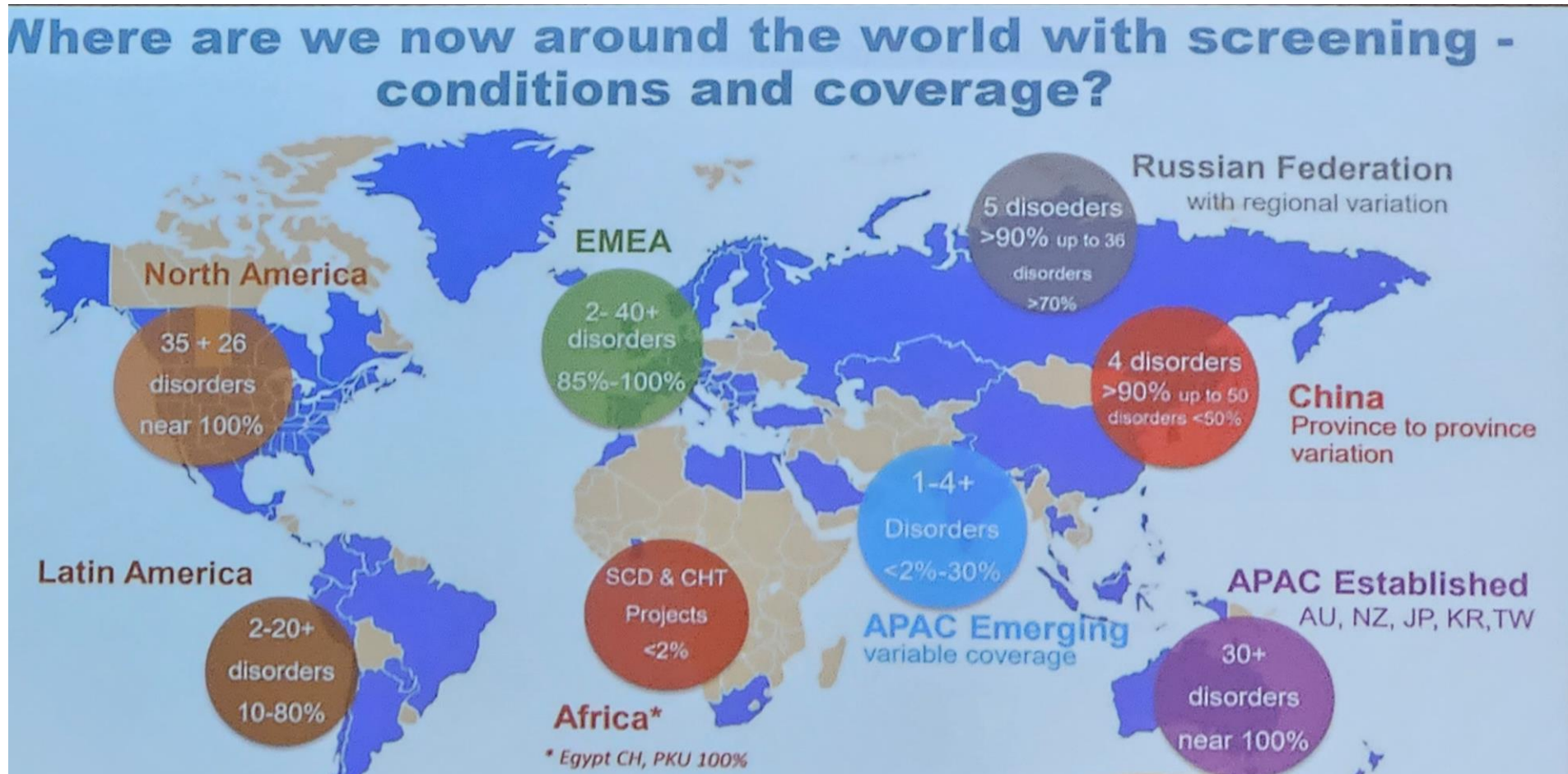
WORLD HEALTH ORGANIZATION

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**

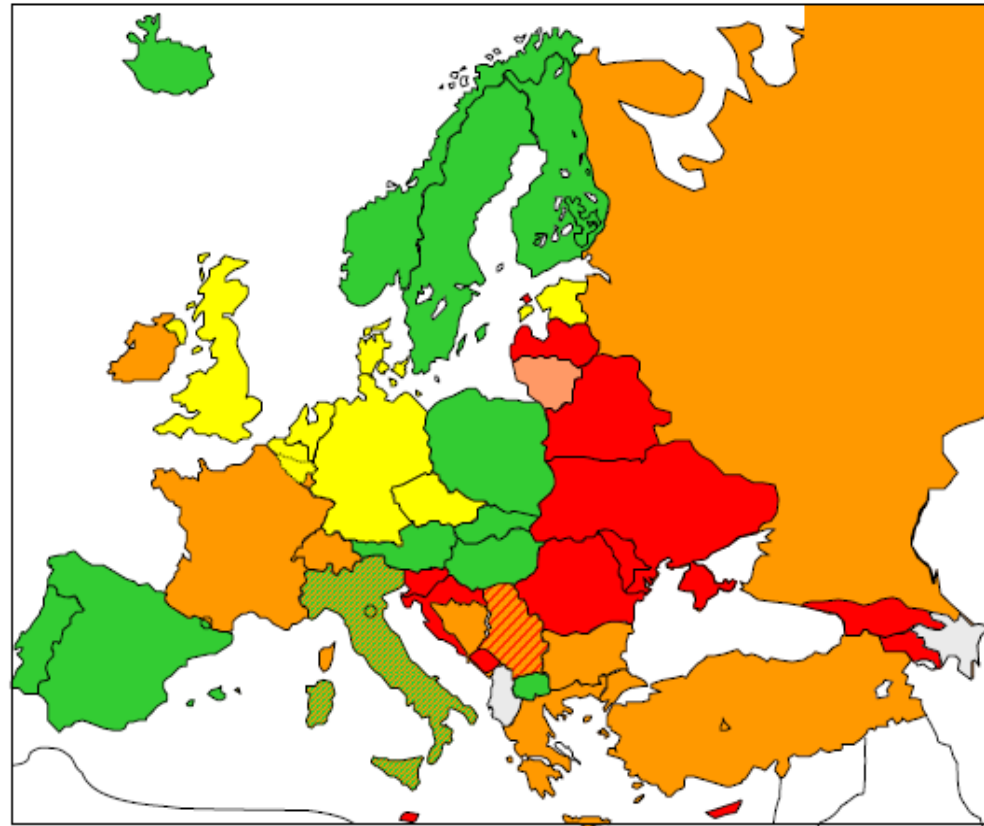
Coverage of populations with NBS



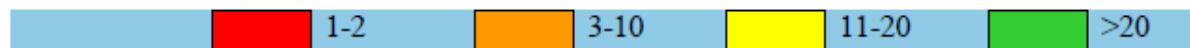
28 % of newborns born

Among-country variation of NBS

Number of conditions per country



2016, G.Loeber, ISNS



Newborn screening in the Czech Republic

Newborn screening (NBS) since 2024

n=20

Endokrinopathies

hypothyreosis
cong. adrenal
hyperplasia

Cystic fibrosis

SMA
SCID

IMD (IEM)

PKU/HPA

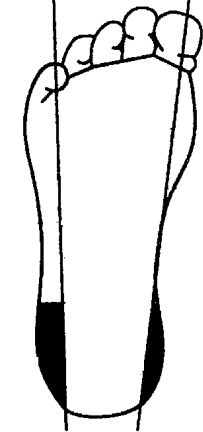
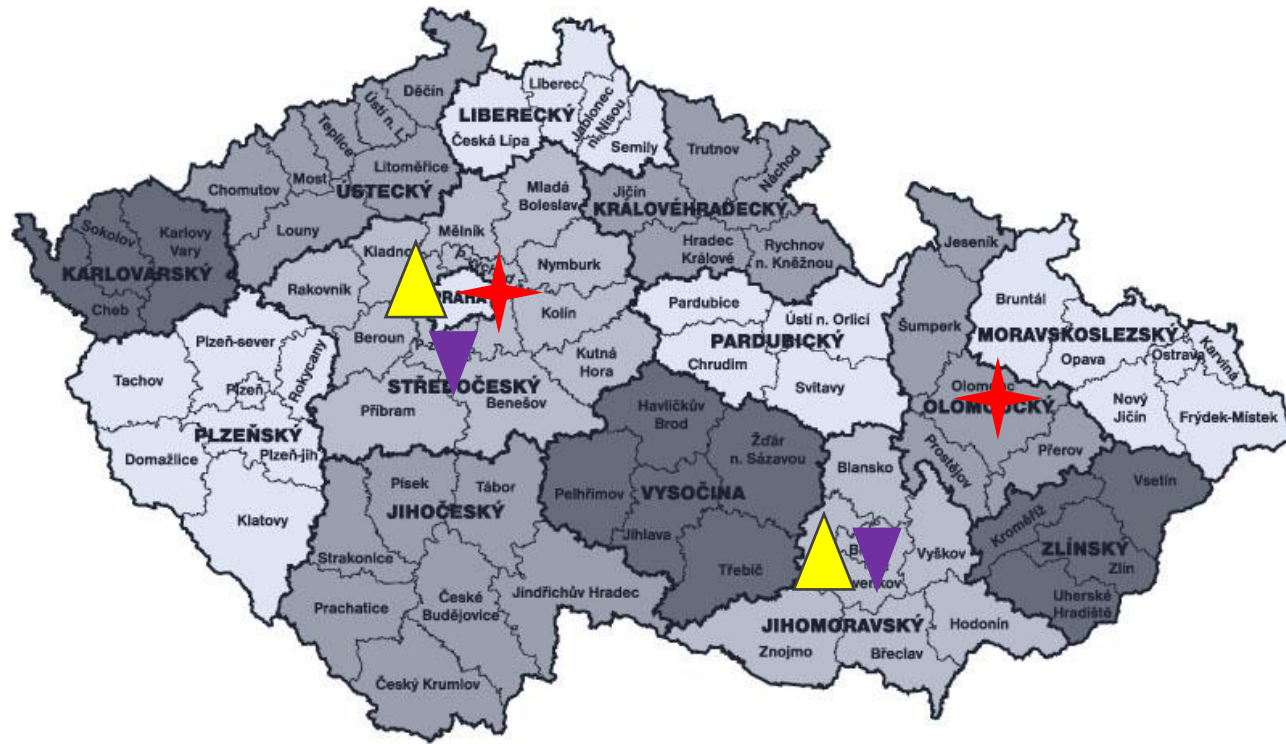
4 aminoacidopathies

3 organic acidurias

6 fatty acid
ox.disorders

*biotinidase
deficiency*

NBS blood sampling



endocrinopathies

CF genetics

IEMetab (Inborn Errors of Metabolism)

1.62 milion newborns (2010-2023)

Endokrinopathie	1:3,000
PKU/HPA	1:5,199
CF	1:6,000
BTD*	1:9,000
FAOD	1:15,500
Other IEM*	1:77,500
*since 2016	
SMA&	1:8,400
SCID£	1:59,100
&£ pilot study 2022-2023	

PILOT PROJECT FROM 1.1.2022

SCID – Severe
or T cell fu

A
A

SMA – Spi

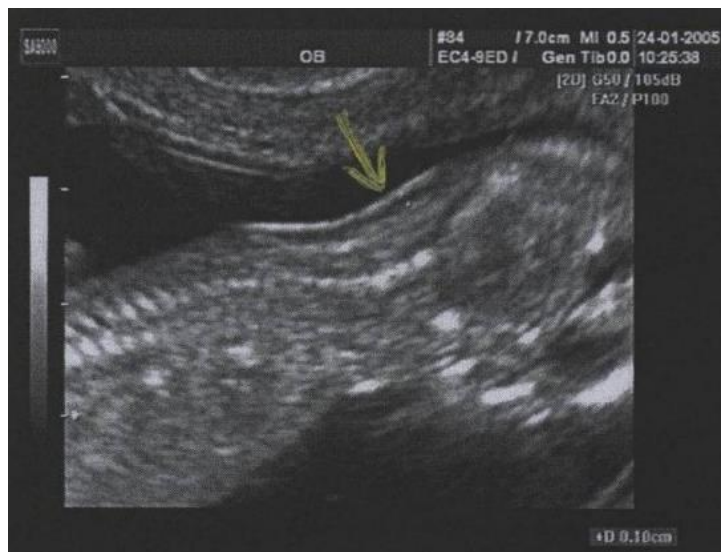
H

Nusinersen (S

Dg.	SMA	7
	SCID	1
	DiGeorgův syndrom	1
	Brutonova agamaglobulinemie	2
	AR agamaglobulinemie	2
	Sekundární iatrogenní deficit	5
	Tranzientní porucha vývoje B lymfocytů	1
INCIDENCE	SMA	1:8400
	SCID	1:59100
	DiGeorgův syndrom	1:59100
	Brutonova agamaglobulinemie	1:29500
	AR agamaglobulinemie	1:29500
	Primární imunodeficience	1:9800
	Sekundární imunodeficience	1:9800

disorder a

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



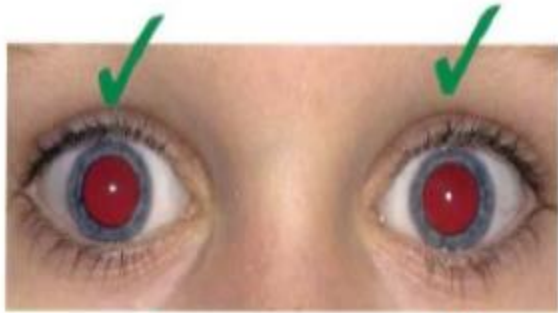
Screening – 2. trimestr

↑AFP - VVV nekryté kůží

↓AFP ↓ uE3 ↑ hCG riziko T21

↓AFP ↓ uE3 ↓ hCG riziko T18, T13

Screening in neonatology



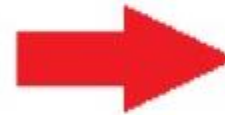
Normal reflex



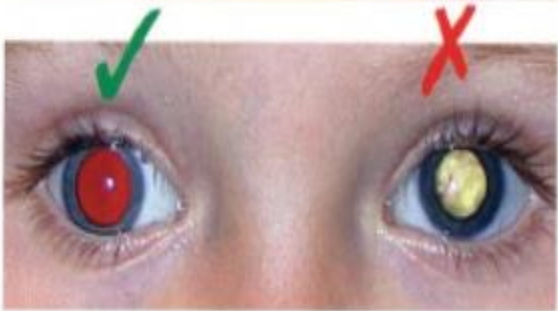
No action required



Red reflex absent



See your GP urgently



Red reflex abnormal

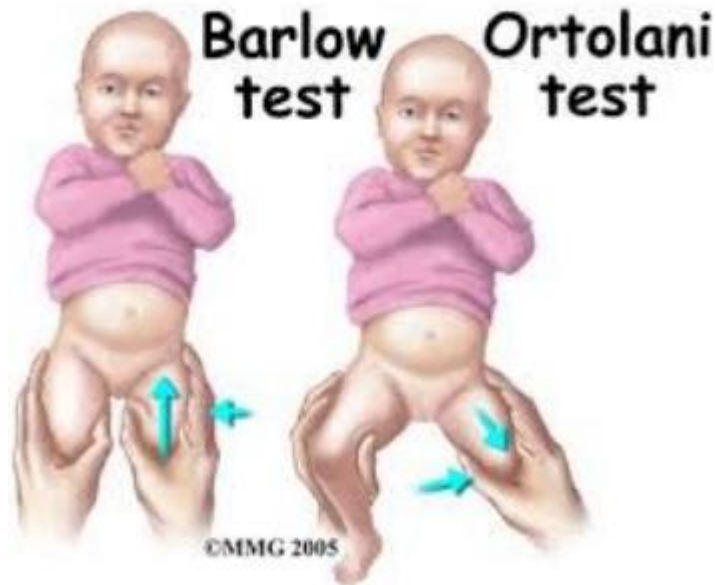


See your GP urgently

Screening in neonatology

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

Dislocation maneuver/provocative test Repositioning maneuver



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

Frejka pillow



Pavlik harness



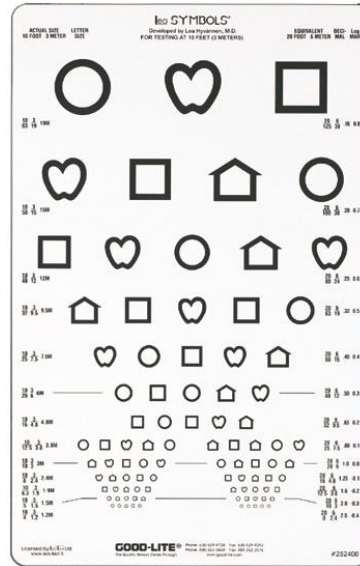
Screening in pediatrics

Plusoptix



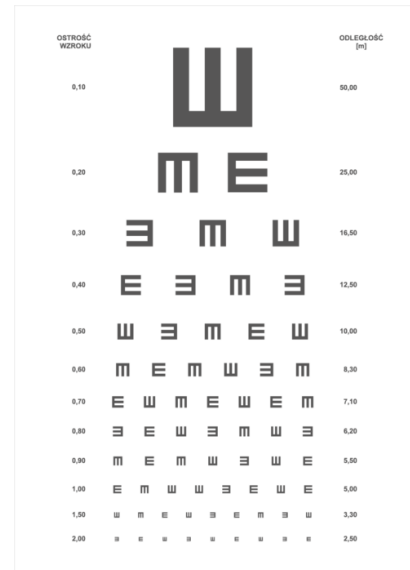
1-2 yrs

Lea symbols



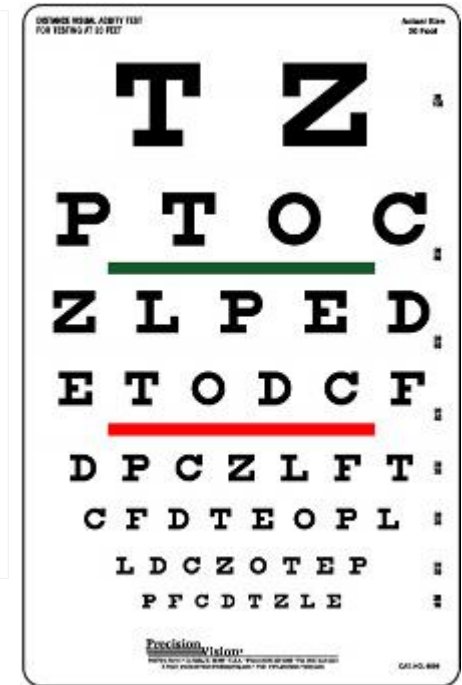
3 yrs

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