European Union
European Social Fund
Operational Programme Employment

Czech National Coordination Centre for Prevention of Serious Diseases
CZ.03.2.63/0.0/0.0/15_039/0006904

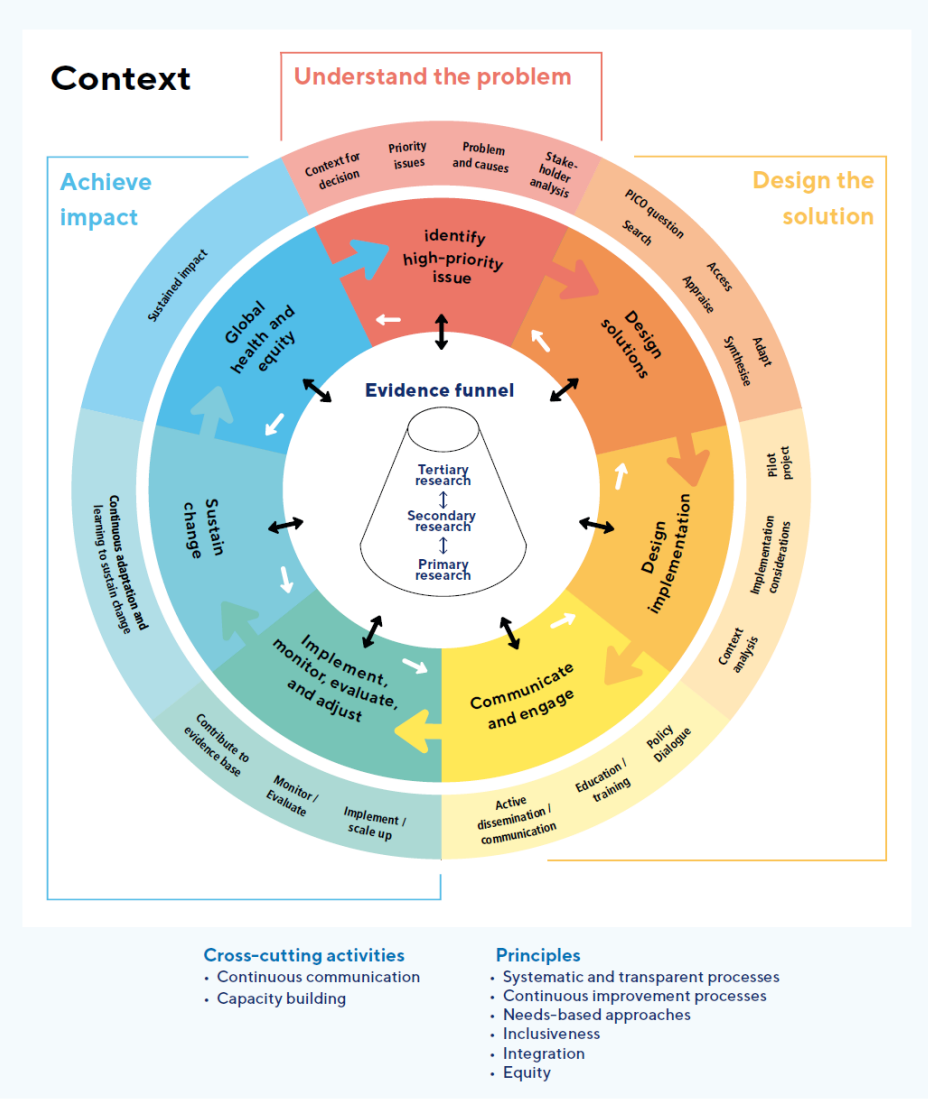
Key indicators of newborn screening: international context and future perspectives for cooperation

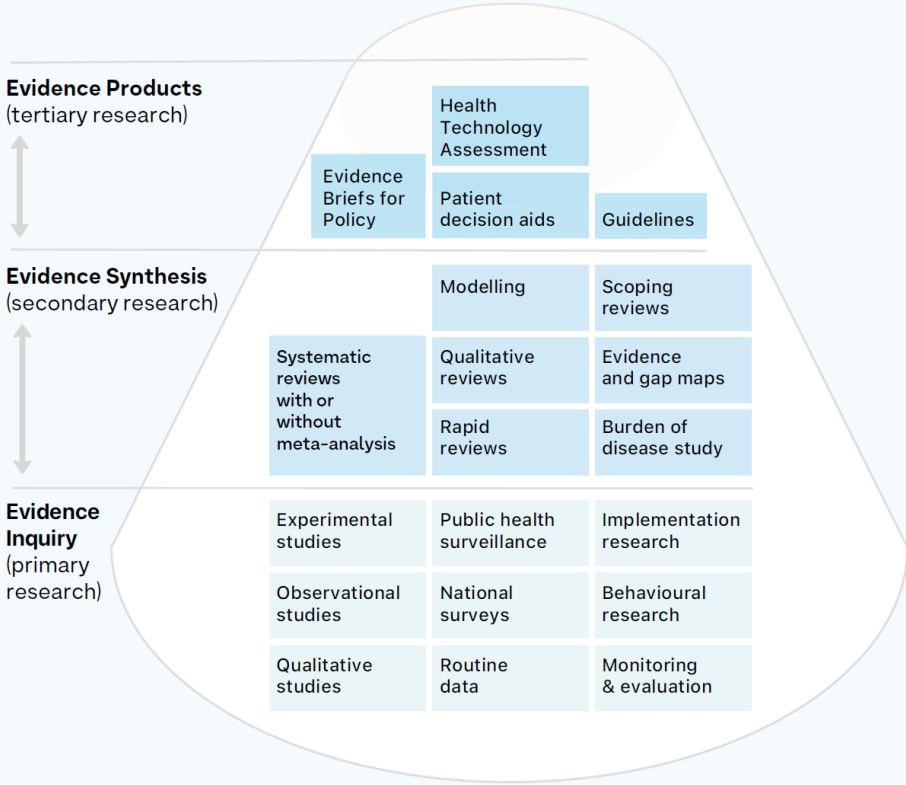
Ondřej Májek, Kateřina Hejcmanová, Renata Chloupková, Ondřej Ngo, Milan Macek, Viktor Kožich, Karel Hejduk

ÚZIS ČR
Ústav zdravotnických informací a statistiky České republiky
Institute of Health Information and Statistics of the Czech Republic

nsc.uzis.cz

Using evidence for impact in health systems





adapted from Reveiz 2020

Evidence, policy, impact. WHO guide for evidence-informed decision-making. Geneva: World Health Organization; 2021. Licence: CC BY-NC-SA 3.0 IGO.

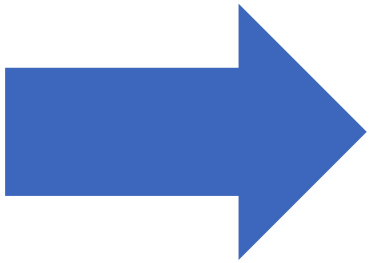
Newborn screening should be designed as an integrated system

- **Elements for effective operation of NBS programmes**
  - **Selection of (new) conditions in NBS panels should be based on published criteria**, the procedures should be standardised, open to public scrutiny and the result of deliberations should be published.
  - Laboratories and programmes should be able to produce data on **key performance indicators** relating to the **entire NBS process**, including blood sampling, transport conditions, blood spot quality, time to generate a laboratory result and refer screen positive cases.
  - Plans to **assess long term outcome data** should be in place and reported.
  - …

Scarpa, M., Bonham, J.R., Dionisi-Vici, C., Prevot, J., Pergent, M., Meyts, I., Mahlaoui, N. and Schielen, P.C., 2022. Newborn screening as a fully integrated system to stimulate equity in neonatal screening in Europe. *The Lancet Regional Health–Europe*, *13*.

Possible performance goals for the screening process

- Increasing coverage
- Increasing positive predictive values
- Optimize laboratory time to result
- Minimising clerical errors
- Secure follow-up of patients



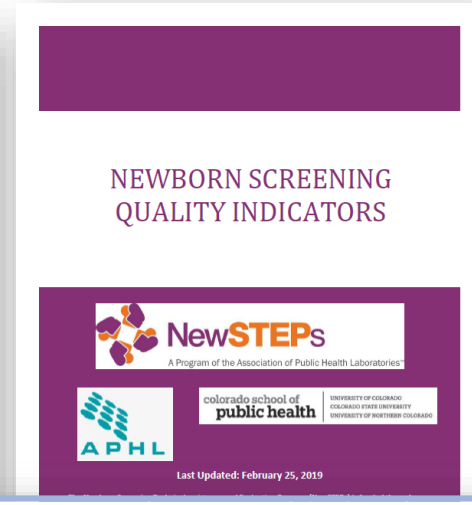
Multidisciplinary screening information system for monitoring screening results and possibly clinical outcomes

Loeber, J.G., Platis, D., Zetterström, R.H., Almashanu, S., Boemer, F., Bonham, J.R., Borde, P., Brincat, I., Cheillan, D., Dekkers, E. and Dimitrov, D., 2021. Neonatal screening in Europe revisited: an ISNS perspective on the current state and developments since 2010. *International journal of neonatal screening*, *7*(1), p.15.

Possible path towards common set of indicators in Europe

Initial desk research of newborn screening quality indicators: US Example

Quality Indicator 1	Percent of dried blood spot specimens that were unacceptable due to improper collection and/or transport
Quality Indicator 2	Percent of dried blood spot specimens with at least one missing state-defined essential data field upon receipt at the lab
Quality Indicator 3	Percent of eligible newborns not receiving a newborn screen, reported by dried blood spot or point-of-care screen(s)
Quality Indicator 4	Percent of infants that have no recorded final resolution (confirmed diagnosis or diagnosis ruled out by an appropriate medical professional) with the newborn screening program
Quality Indicator 5	Timeliness of newborn screening activities
Quality Indicator 6	Percent of infants with an out-of-range newborn screen result requ clinical diagnostic workup reported by disorder category
Quality Indicator 7	Percent of disorders detected by newborn screening with a confirm diagnosis by an appropriate medical professional
Quality Indicator 8	Percent of missed cases, reported by disorder



Proportion of specimens/screens that were obtained during the following process intervals:

a) Time from birth to specimen collection/ point-of-care testing.
b) Time from specimen collection to receipt at your state's newborn screening laboratory.[1]
c) Time from specimen receipt at your state's newborn screening laboratory to reporting out specimen results.
d) Time from birth to reporting out specimen results.
e) Time from reporting out-of-range results to medical intervention by an appropriate medical professional for infants with a confirmed clinical diagnosis.
f) Time from birth to confirmation of clinical diagnosis by an appropriate medical professional.
g) For infants with an out-of-range newborn screen result requiring a clinical diagnostic workup by an appropriate medical professional, time from birth to determining if a result was a false positive.

Indicators used in the programmes: initial findings from 3 systems

Blue indicators shared among the three systems (US, UK, NZ)

Screening programme intensity

- Coverage by screening examinations (US, UK, NZ)

Sample and record quality

- Proportion of samples received by the laboratory with adequate quality (US, UK, NZ)
- Proportion of dried blood spot specimens with at least one missing defined essential data field upon receipt at the laboratory (US)

Screening examination results and accuracy

- Percent of infants with an out-of-range newborn screen result (US)
- …

Timeliness of newborn screening activities

- Proportion of samples taken at an adequate time interval after birth (US, UK, NZ)
- Proportion of samples delivered to the laboratory within an adequate time interval after collection (US, UK, NZ)
- Proportion of newborns with a confirmed clinical diagnosis admitted to medical intervention within an adequate time interval after reporting out screening result (US, UK, NZ)
- …

Possible further steps: international survey

- Performance indicators not included in recent survey (Loeber et al., 2021)
- To guide further development of international consensus, a more specific survey on performance metrics used would be beneficial
- Survey is being discussed within Screen4Rare working group
- Potential contents of a survey
  - Governance and legal framework
  - Centralised registry elements (registries and variables)
  - Performance indicators used

The identification of potential indicators is a first step only: further refinements are needed regarding importance, appropriateness of definition and feasibility (Yusuf, 2019)

International policy example:
EU Council recommendation on cancer screening

RECOMMENDATION

L 327/34 EN Official Journal of the European Union 16.12.2003

COUNCIL RECOMMENDATION
of 2 December 2003
on cancer screening

(2003/878/EC)

ANNEX

SCREENING TESTS WHICH FULFIL THE REQUIREMENTS OF THE RECOMMENDATION (*):

— pap smear screening for cervical cancer precursors starting not before the age of 20 and not later than the age of 30;

— mammography screening for breast cancer in women aged 50 to 69 in accordance with European guidelines on quality assurance in mammography;

— faecal occult blood screening for colorectal cancer in men and women aged 50 to 74.

GUIDELINES

REPORTING