Screeningul neonatal și prevenția din primele zile de viață

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“There is a need to expand the current panel of diseases included in the screening programs to achieve a national improvement in the quality of life for newborns at risk and to achieve a better use of financial resources within the Romanian health system.
The costs for carrying out national screening programs have proven to be considerably lower than those for late treatment of the conditions included in the screening.”

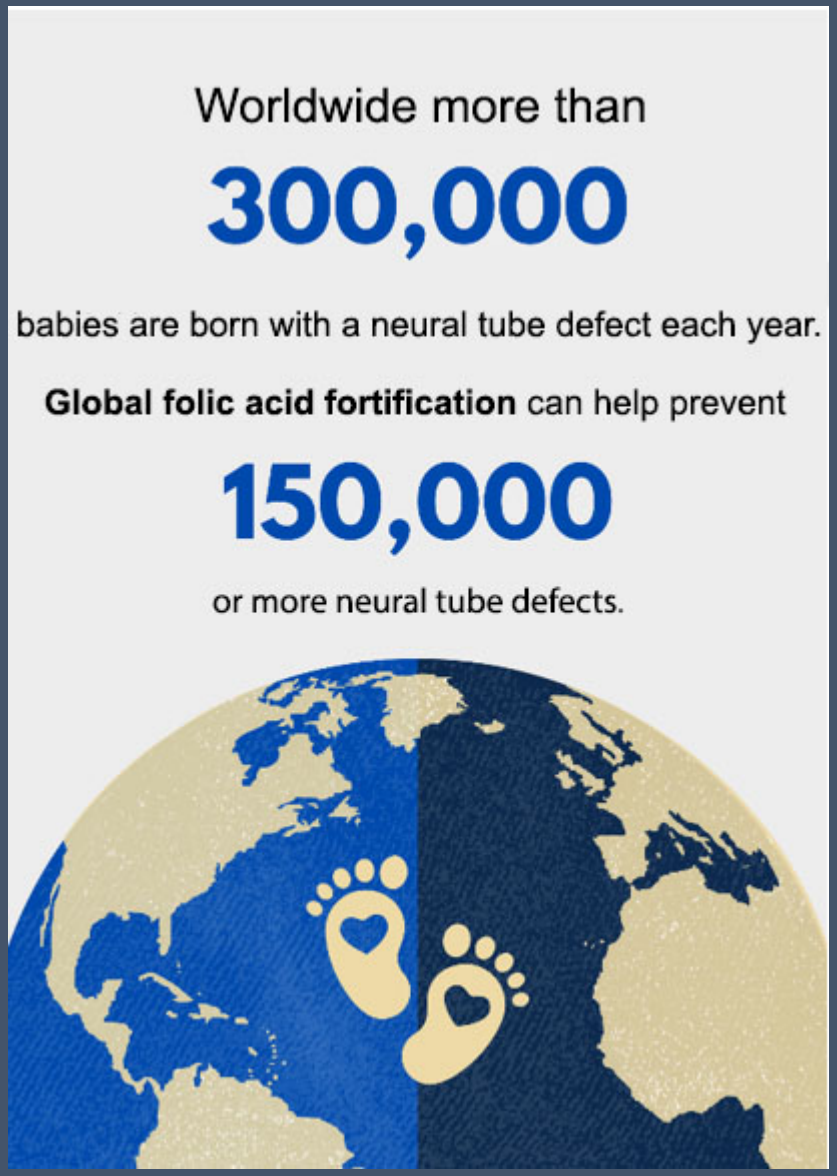
Newborn screening in Romania – present and future
Vlad DIMA –Romanian Journal of Preventive Medicine, volume 1, issue 1
ISSN 2971 – 902X

De ce administrăm acid folic antenantal?

Folic Acid and the Prevention of Birth Defects: 30 Years of Opportunity and Controversies

Annual Review of Nutrition
Vol. 42:423-452 (Volume publication date August 2022)
https://doi.org/10.1146/annurev-nutr-043020-091647

Krista S. Crider,[1] Yan Ping Qi,[1] Lorraine F. Yeung,[1] Cara T. Mai,[1] Lauren Head Zauche,[2] Arick Wang,[1] Kelicia Daniels,[3] and Jennifer L. Williams[1]



Utilizarea acidului folic în perioada preconcepțională și în sarcină

Doza recomandată în primele luni de sarcină
5 mg/zi

Femeile cu sarcini anterioare cu DTN
4000 µg/zi

Doza recomandată în perioada preconcepțională
400 µg/zi

Gravide cu epilepsie, pe tot parcursul sarcinii primesc
0,4 mg/zi

Viswanathan M, Treiman KA, Doto JK, Middleton JC, Coker-Schwimmer EJL, Nicholson WK. Folic Acid Supplementation: An Evidence Review for the U.S. Preventive Services Task Force [Internet]. Rockville (MD): Agency for Healthcare Research and Quality (US); 2017 Jan. Report No.: 14-05214-EF-1. PMID: 28151610.

De ce administrăm corticosteroizi antenatal?

Royal College of Obstetricians & Gynaecologists

Antenatal corticosteroids to reduce neonatal morbidity and mortality

Green-top Guideline No. 74

July 2022

Please cite this paper as: Stock SJ, Thomson AJ, Papworth S; the Royal College of Obstetricians, Gynaecologists. Antenatal corticosteroids to reduce neonatal morbidity and mortality. BJOG 2022;

Jing *et al. BMC Pregnancy and Childbirth* (2021) 21:50
https://doi.org/10.1186/s12884-020-03510-w

BMC Pregnancy and Childbirth

RESEARCH ARTICLE Open Access

Single-course antenatal corticosteroids is related to faster growth in very-low-birth-weight infant

Jiajia Jing[1†], Yiheng Dai[2†], Yanqi Li[3†], Ping Zhou[4], Xiaodong Li[5], Jiaping Mei[6], Chunyi Zhang[7], Per Trop Sangild[3,8], Zhaoxie Tang[1], Suhua Xu[1], Yanbin Su[1], Xiaoying He[1] and Yanna Zhu[1*]

ELSEVIER

Early Human Development

Volume 74, Issue 2, November 2003, Pages 83-88

The effect of antenatal corticosteroids on gut peptides of preterm infants—a matched group comparison: Corticosteroids and gut development

C. Costalos [a], A. Gounaris [b], S. Sevastiadou [a], Z. Hatzistamatiou [a], M. Theodoraki [b], E.N. Alexiou [b], E. Constandellou [c]

Cochrane Library

Cochrane Database of Systematic Reviews

Antenatal corticosteroids for accelerating fetal lung maturation for women at risk of preterm birth (Review)

McGoldrick E, Stewart F, Parker R, Dalziel SR

Utilizarea antenatală a corticosteroizilor

O cură de CS la VG= 24s -33s6z pt. gravidele cu risc de naștere prematură

Se poate repeta cura dacă se menține amenințarea de naștere prematură (>14 zile)

O singură cură se va efectua în cazul apariției PROM

Betametazonă
2 doze
12 mg IM la 24 h

Dexametazonă
4 doze
6 mg IM la 12 h

În cazul RCIU dozele rămân aceleași

Pentru VG=34s – 36s6z
NU
se recomandă o cură de CS

NU se recommendă pentru op cezariană electivă, la termen
(corect >39 săpt.)

La gravidele obeze, la cele cu diabet și în sarcinile multiple NU se fac doze suplimentare

Dagklis T, Sen C, Tsakiridis I, Villalaín C, Allegaert K, Wellmann S, Kusuda S, Serra B, Sanchez Luna M, Huertas E, Volpe N, Ayala R, Jekova N, Grunebaum A, Stanojevic M. The use of antenatal corticosteroids for fetal maturation: clinical practice guideline by the WAPM-World Association of Perinatal Medicine and the PMF-Perinatal Medicine foundation. J Perinat Med. 2022 Mar 11;50(4):375-385. doi: 10.1515/jpm-2022-0066. PMID: 35285217.

Screening și prevenție în Neonatologie

PREZENT

Prevenirea sdr. hemoragic al nou-născutului (vit K)

Prevenirea infecției cu HVB
(vaccin AHB + imunoglobulină umană specifică unde e cazul)

Vaccinare împotriva tuberculozei (vaccin BCG)

Prevenirea infecției cu VSR (prematuri, MCC, BDP) – anticorpi monoclonali (palivizumab)

Screening metabolic (PKU, HTC, FC + AMS*)

Screening hipoacuzie congenitală

Screening ROP (prematuri)

Screening luxație congenitală de șold**

*Testare AMS doar în 7 maternități în prezent
** Ecografie de șold – recomandare în prezent; nu există program de screening național

ACTA ENDOCRINOLOGICA (BUC)

The International Journal of Romanian Society of Endocrinology / Registered in 1938

in Web of Science Master Journal List

Acta Endocrinologica(Bucharest) is live in PubMed Central

Journal Impact Factor - click here.

Year 2021

Contents: July - September 2021, Issue 3

Actualities in medicine

Dima V

Actualities in Neonatal Endocrine and Metabolic Screening

Acta Endo (Buc) 2021 17: 394-399 doi: 10.4183/aeb.2021.394 [Abstract] [Full Text]

doi: 10.4183/aeb.2021.416

ACTUALITIES IN NEONATAL ENDOCRINE AND METABOLIC SCREENING

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"Filantropia" Clinical Hospital of Obstetrics and Gynecology, Bucharest, Romania

Abstract

Neonatal metabolic screening has proven to be an important tool for the early detection of innate metabolic errors. Despite the fact that simple and effective methods of testing for metabolic diseases have been identified since the middle of the twentieth century, no consensus has been reached so far on the content of neonatal metabolic screening panels. There are large differences between countries in the number of metabolic diseases identified through national metabolic screening programs, ranging from zero to several tens, the most common testing being for phenylketonuria and congenital hypothyroidism (including in Romania). Given the fact that rare but treatable diseases have been identified in recent decades, reducing the financial burden on the health system, it would be useful to include them in the national neonatal metabolic screening program.

Keywords: neonatal metabolic screening, innate metabolic errors, costs, consensus, expansion.

INTRODUCTION

Inborn errors of metabolism (IEM) are a category of inherited diseases caused mainly by genetic defects, which conduct to functional defects of some enzymes and other proteins necessary to maintain normal metabolism. IEM results in disruption of biochemical pathways, accumulation of intermediate metabolites, or lack of terminal metabolites. There are several forms of IEM, including disorders of amino acid metabolism (sulfur amino acid metabolism disorders, urea cycle disorders, branched-chain amino acid metabolism abnormalities), organic acidemia, fatty acid oxidation disorders, glucose metabolism disorders, and other conditions. Many genetic metabolic diseases, such as amino acid metabolic disorders, organic acid metabolic disorders, and fatty acid oxidation disorders can be simultaneously detected with high accuracy by determining the amino acid and acylcarnitine content of dried blood on filter paper. A large number of genetic metabolic diseases can be detected at an early stage in newborns due to the widespread popularity of tandem mass spectrometry (MS/MS). The incidence rate of IEM varies between different countries and populations around the world (1).

Hereditary metabolic diseases have high morbidity and mortality, a high risk of recurrence in affected families, but there is the possibility of detecting asymptomatic newborns through neonatal metabolic screening (NBS) programs. NBS programs are internationally recognized secondary prevention interventions that would have a positive impact in the "field of public health". These screening programs aim at the precocious detection of asymptomatic newborns suffering from certain rare diseases, to find a final diagnosis and apply the appropriate treatment to prevent further complications and sequelae.

History of neonatal metabolic tests

The most significant finding in the history of neonatal metabolic screening was the discovery of phenylketonuria (PKU) by Dr. Asbjørn Folling in 1934 (2). The discovery of phenylketonuria marked the first finding of a biochemical explanation for mental retardation (3).

Guthrie and Susi (4) developed a simple, inexpensive, and effective metabolic test to determine if newborns have phenylketonuria (PKU) in the mid-20th century. To define the requirements for additional diseases to be included in screening programs, the World Health Organization published the Wilson and Jungner screening criteria in the Disease Screening Principles and Practices in 1968. These criteria were intended to ensure that these programs meet the main objective: maximum benefit with minimum cost. A screening test is cost-effective when it reduces costs or when the cost of the test, lifelong treatment, and follow-up is offset by the health benefits for the patient (5).

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Newborn screening in Romania – present and future

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Abstract

Neonatal screening has proven its usefulness over time by discovering conditions that can profoundly affect the quality of life and the incidence of mortality and morbidity in the pediatric age. Current neonatal screening in Romania consists of metabolic screening (congenital hypothyroidism, phenylketonuria, and mucoviscidosis), screening for hearing loss, and screening for congenital hip dislocation. A separate category is represented by premature newborns who were admitted to the neonatal intensive care unit and subjected to retinopathy screening. Neonatal metabolic screening has helped in the early detection of inborn errors of metabolism.

Since the middle of the 20th century, even though simple and effective test methods for the detection of metabolic diseases have been identified, currently no worldwide consensus has been found regarding the content of neonatal metabolic screening panels. There are vast differences between countries in terms of the number of metabolic diseases identified through the national metabolic screening programs, the number varying from zero to several dozen, the most frequent testing being done for phenylketonuria and congenital hypothyroidism (including in Romania). Given that rare diseases recently identified can be treated effectively, reducing the financial burden on the health system, we can count on their introduction into the national neonatal metabolic screening program.

Keywords: neonatal screening, hearing loss, retinopathy, congenital dislocation of the hip, inborn errors of metabolism, costs, consensus, extension

History of neonatal screening

Metabolic tests - The most significant event in the history of neonatal screening was the discovery of phenylketonuria (PKU) by Dr. Asbjørn Folling in 1934. The discovery of phenylketonuria was the first biochemical explanation for mental retardation [1,2]. Guthrie and Susi developed a simple, inexpensive, and effective metabolic test to determine whether newborns have phenylketonuria (PKU) in the mid-20th century. Over time, new laboratory tests have been developed for the detection of additional diseases, among which we should emphasize congenital hypothyroidism (ChT). Phenylketonuria and congenital hypothyroidism are the diseases most commonly tested in neonatal metabolic screening programs, and there is substantial variation in the other diseases that are included in national screening programs [3,4].

Screening for Congenital Hearing Loss - The use of some form of hearing testing as a screening for all newborns was first pioneered by Larry Fisch in 1957, and by the end of the 20th century hearing loss screening had become standard in most countries [5].

Hearing loss has been recognized as a factor inducing significant morbidity, which has prevented affected children from reaching their full social, cognitive and economic potential. The effect of early hearing loss was documented in the Ebers papyrus around 1534 BC. Deafness is described as follows: "When deaf, the mouth cannot be opened, that is, it cannot speak" [6]. In the 16th and 17th centuries, in monasteries and later in the secular world there were developed various forms of sign language as instructional methods for the severely deaf child.

Screening și prevenție în Neonatologie

Propuneri pentru viitor

- **Screening MCC prin pulsoximetrie**
- **Screening AMS la nivel național**
- **Introducerea de noi boli în panelul de screening metabolic național (*până la un total de maxim 15*)**
- **Reactivarea Registrului Național al Nou-Născuților Prematur**

(cu obligativitatea înrolării tuturor prematurilor)

- **Înființarea unui program național de follow-up pentru nou-născuții prematur**

Propunere panel screening metabolic

(utilizând datele obținute în perioada 2012-2022 de la Cytogenomic Medical Laboratory)

Congenital Hypothyroidism (TSH)
Methylmalonic acidemia
Congenital adrenal hyperplasia (17-OHProgesterone)
PKU
Galactosemia (Tgal)
Cystic Fibrosis (IRT)
SMA
Non-Ketotic Hyperglycinemia
LCHAD
Hyperornithinemia
MethylcrotonylCoA carboxylase Defficiency
Biotinidase Defficiency
SCAD
G6PD deficiency
Tyrosinemia

Dar de ce este atât de importantă prevenția?

SOCIETATEA ROMÂNĂ DE MEDICINĂ PREVENTIVĂ

drivers
FOR HEALTH EQUITY

Îmbunătăţirea echităţii în domeniul sănătăţii prin acţiuni pe parcursul vieţii

Rezumatul privind statisticile şi recomandările din proiectul DRIVERS

https://eurohealthnet.eu/wp-content/uploads/publications/before-2016/drivers_recommendations_romanian.pdf

https://preventioncentre.org.au/resources/the-value-of-prevention/

Vă mulțumesc!