Molecular Genetics and Metabolism 113 (2014) 42–45

Contents lists available at ScienceDirect

Molecular Genetics and Metabolism

journal homepage: www.elsevier.com/locate/ymgme

Newborn screening in southeastern Europe

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ARTICLE INFO

Article history:
Received 16 July 2014
Received in revised form 21 July 2014
Accepted 22 July 2014
Available online 1 August 2014

Keywords:
Newborn screening
Survey
Southeastern Europe

ABSTRACT

The aim of our study was to assess the current state of newborn screening (NBS) in the region of southeastern Europe, as an example of a developing region, focusing also on future plans. Responses were obtained from 11 countries. Phenylketonuria screening was not introduced in four of 11 countries, while congenital hypothyroidism screening was not introduced in three of them; extended NBS programs were non-existent. The primary challenges were identified. Implementation of NBS to developing countries worldwide should be considered as a priority.

1. Introduction

Newborn screening (NBS) programs were first introduced around 50 years ago, based on a groundbreaking work of Dr. Robert Guthrie [1]. Currently, NBS is a well established practice in most developed countries worldwide, while it is less uniformly implemented in developing countries, which represent most of the southeastern Europe [2–11]. Timely identification and appropriate measures in the case of a positive screening result can help in reducing disabilities and even mortalities in the affected newborns [12,13]. With the right choice of screened disorders NBS is clearly cost-effective, also in the context of a developing country [14–16]. A NBS program consists of six components: screening test, short term follow-up, diagnosis, treatment, management and evaluation [17].

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http://dx.doi.org/10.1016/j.ymgme.2014.07.020
1096-7192/

Table 1
Demographics and newborn screening program characteristics in southeastern Europe.

	Total pop. (Mil.)	GDP per cap. in 2012 (USD)	Screened/all Nb in 2012	No. of screening centers	Diseases mandatory screened (y of introduction)	Age when screened (h)	Lab. methods in NBSP	NBSP costs (per Nb)	Plans to expand NBSP in next 5 y	Which diseases planned to be added to NBSP	Main obstacle(s) perceived in expanding NBSP	Perceived urgency of expanding NBSP (5 = highest; 1 = lowest)[a]
Albania	2.83	9403	−/35,295	0	None	–	–	–	Yes	CH; PKU	FR; LW	5
Bosnia–Federation of Bosnia and Herzegovina (without Sarajevo)	2.07	9392[b]	16,915/nd	1	CH (2005); PKU (2005)[d]	48–96	D (CH); F (PKU)	8 EUR	In 2 y	CAH; CF	FR	3
Bosnia–Rep. of Srpska	1.33	9392[b]	9907/9978	1	CH (2007); PKU (2007)[e]	48–72	D (CH); F (PKU)	8 EUR	In 3–4 y	CAH, CF, GALT	FR; SI; LM;	4
Bosnia–Sarajevo	0.44	9392[b]	5152/nd	1	CH (2000); PKU (2006)	24 (96 if CS)	D (CH); F (PKU)	12 EUR	In 1 y	CF	FR; O; LS	2
Bulgaria	7.36	16,041	62,496/69,121	1	CH (1993); PKU (1979); CAH (2010)	72	D (CH); F (PKU)	3 EUR	After 2 y	CF	FR; LS; LM	3
Croatia	4.28	20,961	41,606/41,700	1	CH (1985); PKU (1986)[f]	72	D (CH); G (PKU)	5 EUR	In 1 y	CUD; GA1; IVA; LCHADD; MSUD; MCADD; VLCADD;	FR	5
Kosovo	1.74	8436	−/34,262	0	None	–	–	–	In 1 y	First CH; later PKU, CAH	FR; O	5
Macedonia	2.06	11,834	nd/23,752	1	CH (2002)	48–96	D (CH)	<2 EUR	Np	Np	nd	4
Moldova	3.50	4219	36,654/39,641	1	CH (1989–1993); PKU (1989)	>48	D (CH); F (PKU)	1 EUR	In 1 y	CH (again)	FR; LS	5
Montenegro	0.63	14,358	−/8156	1	CH (2007)	48–96	D (CH)	1.6 EUR	Np	–	FR; SI; LS	4
Romania	18.91	18,063	159,039/ 201,104	5	CH (1979); PKU (1979; whole country 2011)	48–72	D (CH); F (PKU)	2.5 EUR	In 2 y	GALT, CAH	FR; LS	3
Serbia	7.18	11,801	52,094/67,257[c]	2	CH (1983); PKU (1983)	48–72	D (CH); F (PKU)	4 EUR	Np	Np	FR; LS; O	4
Slovenia	2.05	28,476	21,888/21,938	1	CH (1981) PKU (1979)[g]	48–72	D (CH); F (PKU)	10.6 EUR	In 3 y	GA I; IVA; LCHADD; MCADD; MSUD; VLCADD	FR; O; LW	5

List of abbreviations: h—hours; cap.—capita; Lab.—laboratory; NBSP—newborn screening program; No.—number; Mil.—millions; Nb—newborn; Np—not planned; nd—no data; CW—country wide; FR—financial resources; LS—lack of staff; O—organization; LM—later management; SI—small incidences; LW—lack of (political) will; y—year; CS—cesarean section; D—Delfia method; F—fluorimetric method; G—Guthrie's test; CAH—congenital adrenal hyperplasia; CH—congenital hypothyroidism; CF—cystic fibrosis; CUD—carnitine uptake defect; GALT—classic galactosemia; GAI—glutaric acidaemia type I; IVA isovaleric acidaemia (IVA)/2-methylbutyrylglycinuria; LCHADD—long-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency/trifunctional protein; MCADD—medium-chain acyl-CoA dehydrogenase deficiency; MSUD—maple syrup urine disease; PKU—phenylketonuria; VLCADD—very long-chain acyl-CoA dehydrogenase deficiency.

[a] On a scale from 1 to 5; 1 meaning the lowest urgency and 5 the highest urgency.
[b] Data for all Bosnia and Herzegovina.
[c] Number of newborns in Serbia without Vojvodina region in 2012 was 49,325.
[d] CH from 2000 in Tuzla region; PKU from 2001 in Tuzla region.
[e] PKU from 1986 in Banja Luka region.
[f] PKU from 1978 in Zagreb region.
[g] PKU from 1975 in Trbovlje region.