NEONATAL SCREENING FOR HYPOTHYROIDISM IN ROMANIA

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Introduction

As thyroid hormones are essential for the newborn neurodevelopment, neonatal screening is an effective method for early detection of congenital hypothyroidism. This heterogenous disease requires the prompt initiation of the treatment, in order to prevent the subsequent neuropsychiatric and developmental impairments, such as intellectual disability, spasticity, disturbances of gait and coordination.

In Romania, neonatal screening for hypothyroidism was initiated as a pilot project in 1979, has been expanded since 2002, and is currently conducted through the National Health Program "Neonate Screening, Diagnosis, Confirmation and Application specific diet in PKU and congenital hypothyroidism" financed by the Ministry of Health. The screening of the hypothyroidism is performed by TSH levels measurement.

Objectives

We aimed to assess the current state of hypothyroidism screening, the management of this condition, and to determine the incidence of hypothyroidism at birth in 2013 in Romania.

Subjects and methodology

We analysed data reported to the Implementation Unit of Health Programs in 2013 by the 4 regional centers (**Bucharest - IOMC, Cluj, Iasi and Timisoara**) (Figure 1) and the parameters registered in the Hypothyroidism National Registry (in Bucharest and Iasi).

The informed consent of the mother was obtained before any investigation was performed.

In the case of normal weight newborns, fed with breast milk or formula, the blood was collected in the 3rd-5th day of life or on discharge day. In premature, underweight newborns, and in all cases with delayed enteral feeding, the blood was collected in the 3rd -5th day from dairy food initiation.

TSH levels were assessed by immunofluorimetric method in 3 regional centers and by Elisa technique in the fourth. The test was considered for a neo-TSH cut-off of 15mIU/L.

The diagnosis was locally confirmed by TSH, T4 and free-T4 levels in patients serum.

All the newborns were investigated by the "dry spot" method; capillary blood samples were collected from the heels. One drop of blood is placed on the marked area of the strip (15 - 16 mm).

All the measurements were done in duplicate. The date of sampling is mentioned in the discharged summaries.

If TSH levels are ≥ 15 mIU/L the test was considered "positive " and the newborn family was contacted for further confirmatory tests (free thyroid hormones).

Results

In 2013, in **Romania,** there were registered **167076** neonates; **154357** neonates were screened for congenital hypothyroidism (**92,76%**) (Fig1). We identified **96 positive patients (0,07%)**.

Further tests confirmed congenital hypothyroidism **in 41 newborns (0.03%).**

In the **Bucharest** screening center – from **106947 registrated newborns, 99375 were screened (92,92%), with 31 cases confirmed congenital hypothyroidism (0,03%).**

In confirmed cases, a multidisciplinary team (endocrinologist, paediatrician, psychologist, neuropsychiatrist) treated and followed-up the outcome (thyroid re-evaluation, growth, psychomotor development and school progression).

The families refused the diagnosis investigations in 3 patients (0.002%).

The hypothyroidism incidence in this study is **1/3409 infants**.

Fig. 1 - Number of newborns screened for hypothyroidism in Romania between 2002 and 2013

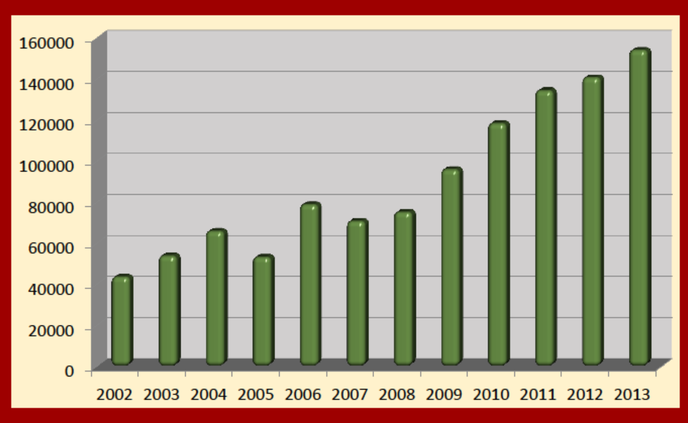
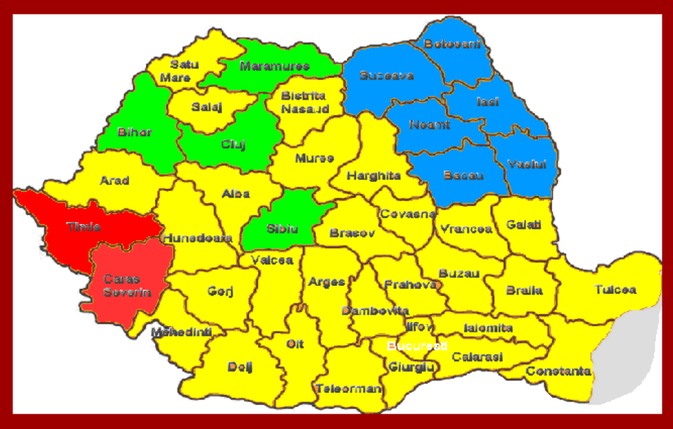
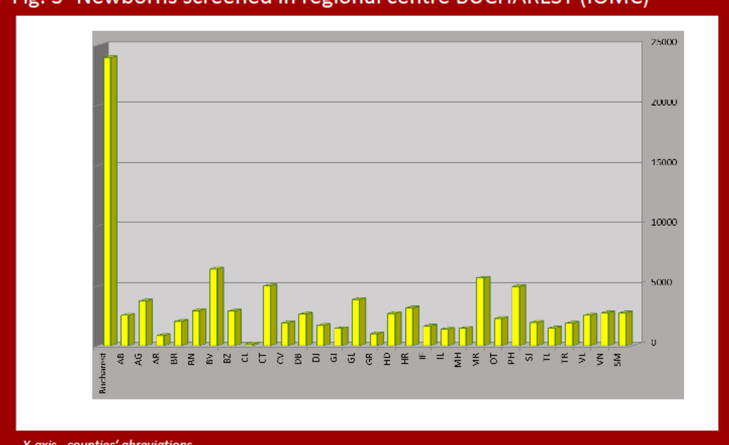


Fig.2- Romanian regional centres for congenital hypothyroidy screening



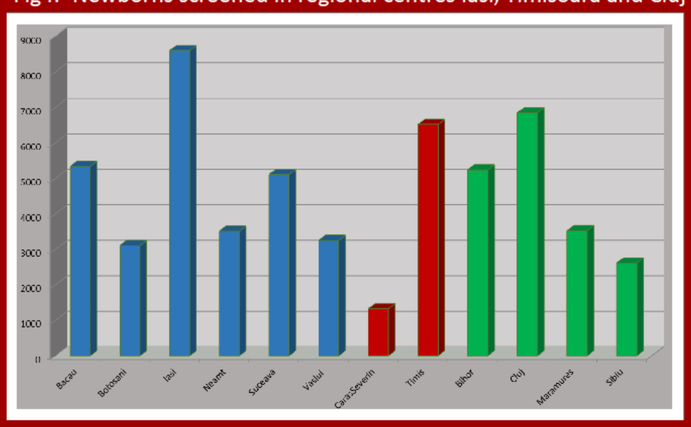
Romanian regional centers for congenital hypothyroidism screening: BUCHAREST, IASI, TIMISOARA, CLUJ

Fig. 3- Newborns screened in regional centre BUCHAREST (IOMC)



X-axis - counties' abreviations

Fig4.- Newborns screened in regional centres Iasi, Timisoara and Cluj



Conclusions

- ❖ **National neonatal screening program for congenital hypothyroidism demonstrates highly social and economical benefits, due to newborns being diagnosed early on.**
- ❖ **The incidence of congenital hypothyroidism in Romania is within the limits reported in literature.**
- ❖ **Early diagnosis and initiation of therapy ensures a proper neurodevelopment of affected infants and prevents morbidity.**
- ❖ **We attempt to increase the number of the newborns tested for hypothyroidism by raising awareness about screening benefit among the families.**
- ❖ **Taking into account the false positive cases, we estimate that ongoing studies will enable the increase of the neo-TSH cut-off value at 17 mIU/l.**

References

Juliane Léger et al. J Clin Endocrinol Metab. 2014 Feb; 99(2): 363–384.

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