



Missed Cases of Congenital Hypothyroidism Detected By Screening Program in Central Serbia (1983–2014)



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Background

There are a lot of reasons for missing the diagnosis in neonatal screening for congenital hypothyroidism (CH), but errors in processing samples and reporting results are the most frequent!

In Central Serbia screening for CH was instituted in 1983 by determination of the TSH level in dried filter-paper blood spots.

All samples are analyzed at one central laboratory.

The average number of specimens that are annually screened is 50 000.

The screening process was divided into:

- specimen collection,
- laboratory procedure,
- follow-up phase.

A missed case was defined as one not identified through the standard protocol of a neonatal screening.

Objective and hypotheses

The aim of the study is to determine the extent of the problem of missed cases

Method

To gather data on missed cases of CH, in 2013 we did the retrospective study that included:

- investigation of case notes of all patients with CH who were treated and followed up in our hospital
- questionnaires that were sent to the pediatricians in the primary health care and to pediatric endocrinologists in Central Serbia.

Results

During 30 years of screening program in Central Serbia 1 547 122 newborns are screened.

Incidence of CH is 1:3728

The diagnosis of CH:

- confirmed in **434** newborns
- missed in **12** cases

1983 – 2002

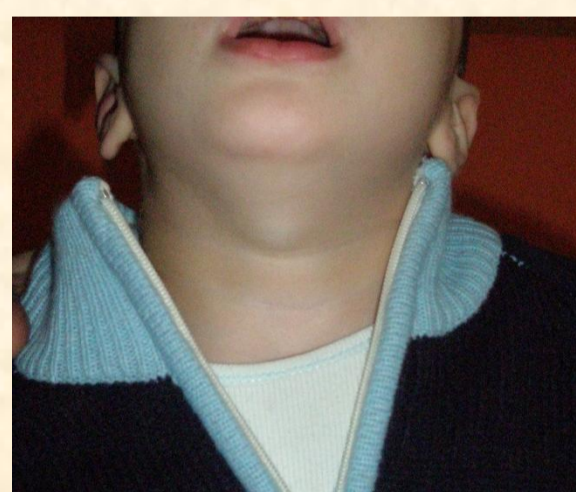
- 7 specimens were not received in the laboratory
- 1 specimen was exchanged in maternity ward

2003 – 2007

no new cases

2008 – 2010

- 3 cases of false-negative results
- discovered in the age of 3.5 years, 6 years and 15 months
- in all the diagnosis was thyroid ectopy



2012

- 1 specimen was not collected
- patient was transferred from maternity ward to ICU to another hospital because of perinatal asphyxia
- diagnose: athyreosis



2013 – 2014

no new cases

Presenter disclosure information

The author disclosed no conflict of interest

Results

For the period of 30 years:

- 8 specimens were not received in the laboratory
- 1 specimen was exchanged in maternity ward
- 3 false-negative results

According to our knowledge

1 missed case of CH

for every 36 detected.

Conclusion

For the great majority of infants, neonatal screening program for CH has been successful.

Because standard screening procedures may not detect every case of CH, physicians should be clinically vigilant with regard to signs and symptoms of CH.

Special attention should be paid to children who are transferred from one hospital to another.

False -negative results are a problem that can not be avoided.

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