Etiology and severity of congenital hypothyroid children detected through neonatal screening: a cut-off based analysis

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Background:
TSH cut-off (CO) levels for the detection of congenital hypothyroidism (CH) have been lowered progressively in many screening programs. Nevertheless, population detected with lower CO levels differs in severity and etiology.

Objective and hypotheses:
To describe the etiological characteristics and severity of children detected by neonatal screening related to CO TSH levels.

Methods
Data of CH neonatal screening performed between June 2013 and January 2016 in 4 maternities
TSH (IFMA-Delfia) measured in DBS obtained by heel puncture at maternity discharge (CO:8mUI/l blood)

Confirmation:
* serum TSH, T4, FT4 and TG
* Tc99 thyroid scan (when available)

Severity
Severe: FT4 <1 ng/dl
Mild: FT4≥1 ng/dl

Characterization:
athyreotic
eutopic
goiter

Findings were related to TSH levels intervals of detection:
>20 mU/l, 10-20 mU/l, 8-9.9 mU/l

Diagnostic efficiency (DE) was calculated for each TSH interval.

Results
20441 newborns
81 recalled
Rate: 0.4%

>20 mU/l
n = 18
Confirmed
18/18
DE: 100%

10-20 mU/l
n = 24
Confirmed
8/24
DE: 30%

8-9.9 mU/l
n = 39
Confirmed
2/39
DE: 5.1%

CH Characterization
Ectopic Eutopic Goiter Athyreotic

CH Severity
Severe Mild

Conclusion
➢ While higher TSH levels allowed detection of patients with dysgenesis and severe disorders with better efficiency, lower CO identified mainly mild thyroid disorders.
➢ Further evaluation will allow the better characterization of the hypothyroid spectrum and to delineate adapted guides on detection and follow up.