INCIDENCE AND ETIOLOGY OF PRIMARY CONGENITAL HYPOTHYROIDISM IN FLANDERS

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INTRODUCTION

In several European countries, the incidence of primary congenital hypothyroidism (PCH) seems to increase in the last decade, especially in countries that have lowered the newborn screening TSH cutoff.

In Flanders, two centers manage since 2012 the newborn screening for PCH and these maintained the same TSH assay and cutoff (15 mU/L).

AIM

We studied in Flanders

- Current incidence of PCH
- Etiology of PCH
- Clinical characteristics of infants with PCH
- Initiation of thyroxine treatment

METHOD

Inclusion: All screened infants between 2012 and 2017

Single center (VCBMA), covering about half of the newborns in Flanders.

Collected data when diagnosed with PCH:

- Birth data,
- Results of hormonal measurements at screening and at recall
- Scintigraphy and ultrasound results at initial evaluation
- Data on thyroxine treatment initiation

RESULTS

Table 1: Incidence of CHT

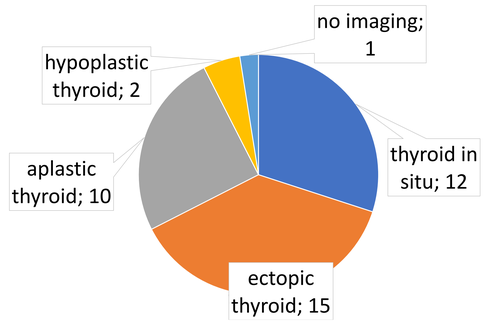
Year	Screened children	Children with CHT	Incidence of CHT
2012	28511	4	1/7128
2013	28571	5	1/5714
2014	28384	3	1/9461
2015	27535	13	1/2118
2016	27904	5	1/5581
2017	26748	10	1/2675
Total	167653	40	1/4191

During the study period, 167,653 live newborns were screened for PCH, and thyroxine treatment was started in 40 (22 females [F] and 18 males [M]) newborns after confirmed PCH (serum TSH > 20 mU/L at recall).

The yearly number of treated newborns varied greatly (between 3 and 15 per year), but without a clear increase.

A low serum FT4 (< 15 pmol/L) was present in 36 newborns.

Figure 1: Etiology of CHT



Based on scintigraphy (n = 29) and/or ultrasound (n= 32), a diagnosis of gland in situ, ectopy, aplasia and hypoplasia and no imaging was made in respectively 12 (5F), 15 (11 F), 10 (5 F) and 2 (0 F) and 1 (0 F) newborns.

Table 2: Clinical characteristics and treatment of CHT

	CHT median (min-max)	Starting dose median (min-max)
Number of cases	40	11.8 µg/kg/dag (7.7-31.6)
Gestational age (weeks)	40 (25; 42)	
Birth weight (SDS)	0.00 (-2.31- 2.57)	
Birth length (SDS)	-0.14 (-1.93-2.89)	
Birth head circumference (SDS)	0.30 (-2.70-2.60)	

No fetal growth differences were seen between the different etiologies.
Thyroxine was started at a median (range) age of 10 (3-18) days (n = 35 before day 14). The thyroxine starting dose ranged between 25 (n=6) and 50 µg (n = 10). Newborns with apparent aplasia had the lowest FT4 concentrations (median (range) 3 (0.4 -18) pmol/l) and received the highest starting dose (12.6 (7.6-20.4) µg/kg/day) when compared with the other etiologies.

CONCLUSIONS

The overall incidence of PCH is 1 in 4191 live newborns.

Abnormalities in thyroid morphogenesis are diagnosed in 69 %.

No fetal growth difference is observed between the different etiologies of PCH , but a female gender excess is present in the newborns with ectopy.

Newborns with apparent thyroid aplasia receive the highest dose. Thyroxine treatment is initiated before the age of 14 days in 87.5 % of the newborns.

REFERENCES

1. J. Leger. European Society of Paediatric Endocrinology consensus guidelines on screening, diagnosis, and management of congenital hypothyroidism. Hormone research in Paediatrics. 2014 ; 81: 80-103
2. American Academy of Pediatrics, Susan R. Rose, American Thyroid Association, Rosalind S. Brown and Lawson Wilkins Pediatric Endocrine Society. Update of Newborn Screening and Therapy for Congenital Hypothyroidism. Pediatrics. 2006; 117;2290.
3. Edgar J. Schoen, Wesley Clapp, Trinh T. To and Bruce H. Fireman. The Key Role of Newborn Thyroid Scintigraphy With Isotopic Iodide (123I) in Defining and Managing Congenital Hypothyroidism. Pediatrics, 2004, 114 (6) e683-e688
4. Il Soon Park, Jong Seo Yoon, Cheol Hwan So, Hae Sang Lee, Jin Soon Hwang. Predictors of transient congenital hypothyroidism in children with eutopic thyroid gland. J Pediatr Endocrinol Metab 2017; 22: 115-118.

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