Congenital hypothyroidism (CH) detected by the second newborn screening in Lombardia region: incidence and evolution of CH

Silvana Caiulo[1], Maria Cristina Vigone[1], Marianna Di Frenna[1], Simona De Angelis[2], Daniela Rotondi[2], Gaia Vincenzi[1], Simona Lucchi[3], Luisella Alberti[3], Graziano Barera[1], Carlo Corbetta[3], Antonella Olivieri[2], Giovanna Weber[1]

1 Vita-Salute San Raffaele University, Paediatric Department San Raffaele Hospital, Milan, Italy

2 Italian National Institute of Health, Rome, Italy

3 Regional Newborn Screening Laboratory, Children's Hospital V.Buzzi, Milan, Italy

Objective

To assess the incidence of CH in Lombardia region and the percentage of patients identified by the 2nd NBS. To describe the clinical features and evolution of CH patients detected by the 2nd NBS.

Methods

The 1st NBS (cut-off blood-TSH>10mU/L) was performed at 2–5 days in all neonates born in Lombardia region.
The 2nd NBS (cut-off blood-TSH>5mU/L) was performed in selected cases: prematurity, weight<2000g, malformations/syndromes, admission in NICU, twins, steroid treatment, maternal thyroid disease, borderline 1st NBS and associated risk factors.
Clinical data at diagnosis and reevalutation of patients detected by the 2nd NBS (period 2007-2014) and followed-up at a single tertiary centre for pediatric endocrinology were collected. At 2–3 years, patients with gland in situ (GIS) underwent reevaluation and were classified as permanent CH (s-TSH>10 mU/L), persistent hyperthyrotropinemia (HT) (s-TSH 5–10 mU/L), and transient CH (s-TSH<5 mU/L).

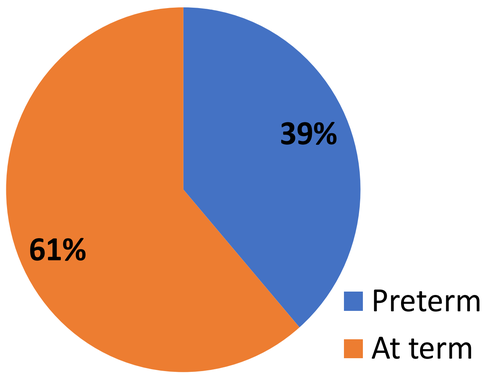
Results

Incidence of CH

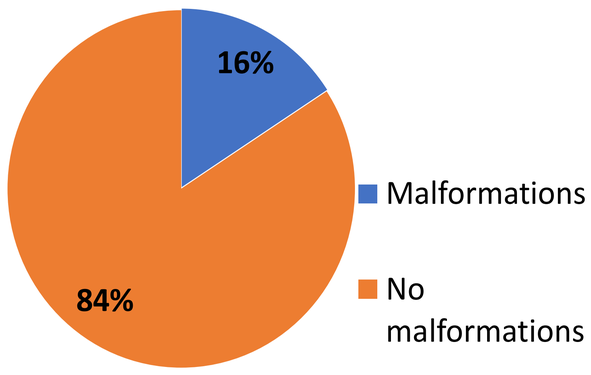
767.157 neonates born in Lombardia between 2007-2014 → **842** patients affected by CH (**1:911**), detected by the NBS → **273** patients detected by the 2nd NBS (**32.4% of CH**) → **101** patients followed-up at San Raffaele Hospital (96 GIS, 5 dysgenesis)

Clinical features

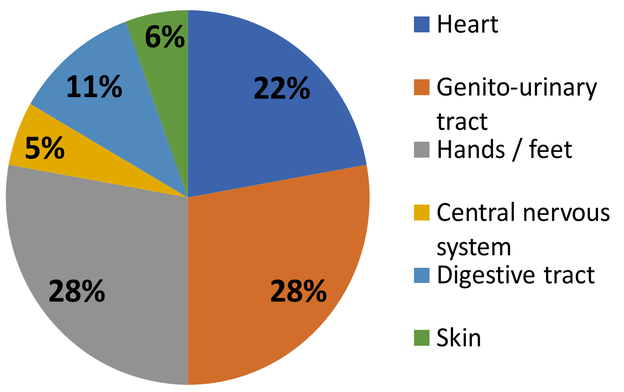
Prematurity



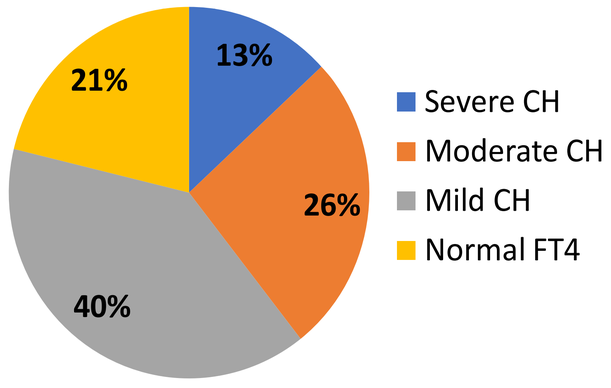
Malformations



Type of malformation



CH severity at diagnosis

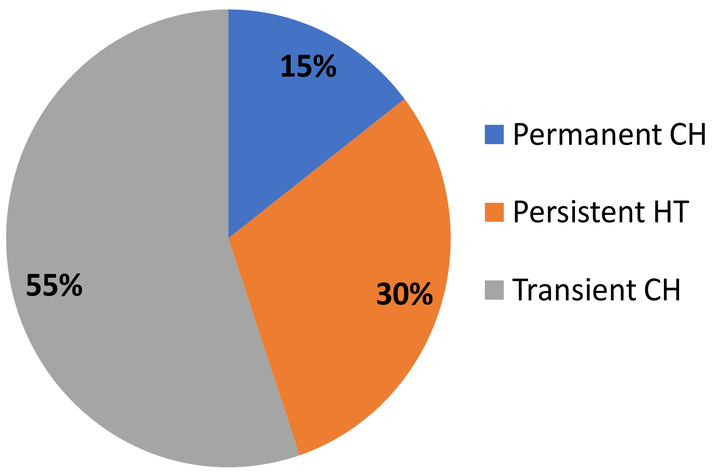


Patients with thyroid dysgenesis

	1st… 2nd b-TSH	Sieric TSH and FT4 at diagnosis	Age at start of therapy	Diagnosis
1	6,83…8,93 mU/L	TSH 22,9 mU/L FT4 1,1 ng/dl	26 days	Thyroid ectopy
2	8,27..37,24 mU/L	TSH 110 mU/L FT4 0,43 ng/dl	20 days	Thyroid hemiagenesis
3	9,33..13,36 mU/L	TSH 18,2 mU/L FT4 1,1 ng/dl	31 days	Dysmorphic thyroid tissue
4	4,10..17,25 mU/L	TSH 138,5 mU/L FT4 1,09 ng/dl	29 days	Thyroid hemiagenesis
5	5,33..5,16 mU/L	TSH 22,9 mU/L FT4 1 ng/dl	28 days	Thyroid hemiagenesis

Diagnostic reevaluation of patients with GIS

Diagnostic reevaluation



Genetic analysis

96 patients with GIS
↓
23 genetic analysis
↓
12 patients showed pathogenetic variants:
↓
9 patients: DUOX2
2 patients: TSHR
1 patient: PAX8

Conclusions

- Between 2007 and 2014 the **incidence of CH** confirmed at birth in Lombardia region was **1:911** (permanent and transient).
- In the absence of the 2nd NBS the incidence would have been 1:1348; **32.4% of patients were detected by the 2nd NBS**.
- In our cohort, **preterm infants were 39%**, indicating that other risk factors can contribute to delayed TSH elevation.
- **The frequency of malformations was higher than expected**.
- Although the majority of patients identified by the 2nd NBS had transient CH and HT, **the 2nd NBS allowed the identification of severe cases of CH, cases of thyroid dysgenesis, and cases of CH caused by genetic variants**.

58ESPE P1-423
Thyroid
Silvana Caiulo
Poster presented at: ESPE 2016 VIENNA
PosterSessionOnline.com