Congenital hypothyroidism (CH) with delayed TSH elevation: the importance of the second-screening strategy and the evolution of CH in preterm infants

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There are no conflicts of interest

Introduction and objectives:

Preterm infants often present CH characterized by delayed TSH elevation. We describe the clinical and biochemical features and the evolution of CH in preterm infants with delayed TSH elevation, detected by the 2° screening for CH.

Methods:

All preterm infants born in Lombardy Region between 2007-2014 negative to the 1° screening (b-TSH<10 mcU/ml) at 2-5 days of life and positive to the 2° screening at 12-33 days (b-TSH ≥5 mU/L), diagnosed with CH and followed-up in a single tertiary Centre of paediatric endocrinology were included (figure 1).

According to the result of the thyroid function testing after the withdrawal of therapy, at 2-3 years, patients were divided into 3 groups, as shown in figure 2.

Figure 1: Study design

Total of infants born in Lombardy Region (Italy) between 2007 and 2014

⬇

Total of patients affected by CH and detected by the 2° screening (b-TSH ≥5 mU/L)

⬇

119 patients, detected by the 2° screening, were followed up at San Raffaele Hospital

46 of them were born preterm and included in the study

Figure 2: Clinical re-evaluation in patients with gland in situ at 2-3 years

After the withdrawal of therapy

- Permanent CH, if s-TSH >10 mcU/ml
- Persistent hyperthyrotropinemia, if s-TSH 5-10 mcU/ml
- Transient CH, if s-TSH < 5 mcU/ml

Results:

46 preterm patients were included in the study (26 males). At diagnosis, the neck ultrasound showed 1 ectopy, 1 hemiagenesis, and 44 cases of GIS. Treatment was started at a median age of 40 days (15-89).

Figure 3: Neonatal features at diagnosis

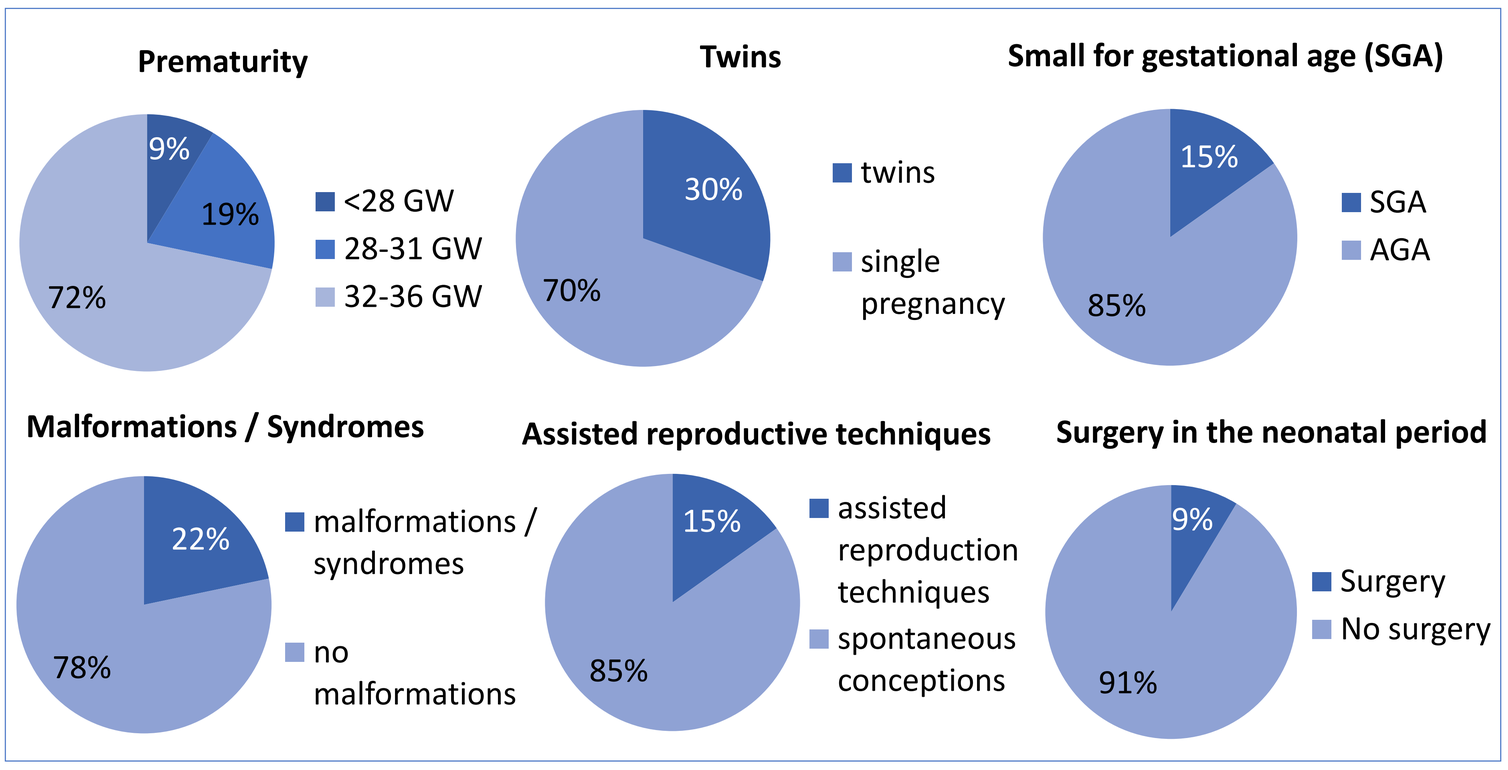


Table 1: s-TSH at diagnosis

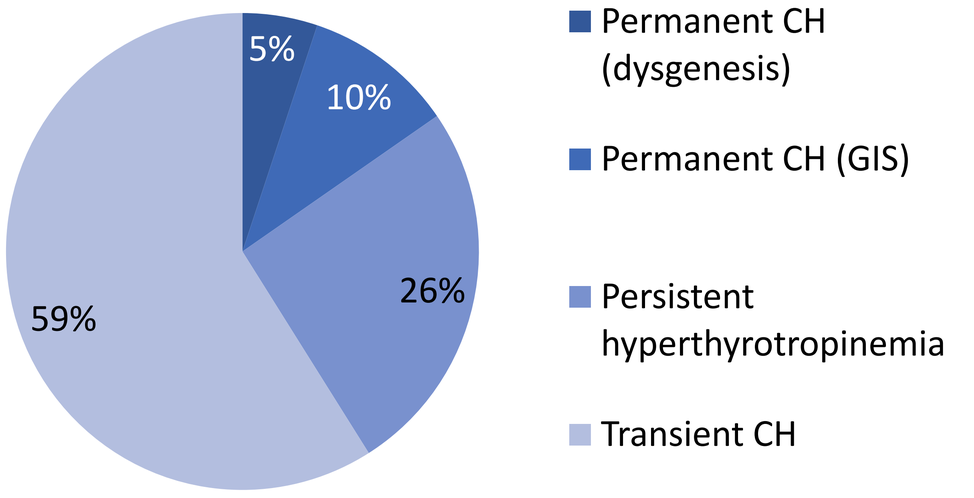
	s-TSH (mcU/ml)
Mimimum value	10,61
25° p.le	14,24
Median	19,34
75° p.le	49,28
Maximun value	756,60

Two patients had thyroid dysgenesis. Among the 44 patients with GIS, 37 were reevaluated. At reevaluation:

- 4 patients had permanent CH (s-TSH 17,9-24,1 mcU/ml), requiring the reintroduction of LT4
- 10 had persistent hyperthyrotropinemia
- 23 had transient CH

The 4 permanent cases with GIS were moderate-to-late preterm (32-36 GW), 2/4 were twins and in both cases the other twin (not included in the study) had hyperthyrotropinemia. Moreover, they showed only mild s-TSH elevation at diagnosis (s-TSH 14,40-19,77 mcU/ml).

Figure 4: Evolution of thyroid function



Conclusions:

• We confirmed the usefulness of the second-screening strategy for CH to detect preterm infants who otherwise would not be identified at the first screening.

• Although preterm infants very often have transient CH, many of them may have severe CH at diagnosis, which requires prompt treatment, and some others may have permanent CH (including thyroid dysgenesis), despite mild TSH elevation at diagnosis.