Congenital Hypothyroidism incidence and dysgenesis or dyshormonogenesis prevalence in a large infants cohort from south of France

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Congenital hypothyroidism (CH) is the most frequent endocrine disease in infants with prevalence ratio in the range of 1:2000 to 1:4000 new-borns. The disorder can be permanent (CHP) or transient (CHT) .CH is classified into 2 main groups: Dysgenesis, which accounts classically for 80–85 % of cases and Dyshormonogenesis for remaining 15–20 %. From the last decade, studies described a upward trend for CH prevalence and changes in groups' proportions.

OBJECTIVES

To determine the updating prevalence of CH, CHP, CHT, Dysgenesis and Dyshormonogenesis in infants confirmed with CH after newborn screening in a large French region (Midi-Pyrénées 31,000 births /yr.)

METHODS

-Dysgenesis and Dyshormonogenesis were defined as proposed in ESPE consensus and based on neonatal thyroid ultrasonography, thyroglobulin level and scintigraphy.

-CHP categorised dysgenesis or dyshormonogenesis as infants with LT4 replacement necessary after 3 years of life and CHT when LT4 replacement is stopped between 1 and 3 years of life, with normal thyroid lab tests, ultrasonography and scintigraphy with perchlorate discharge test.

-Retrospective study for all 100 newborn confirmed at day 10 for CH in our region (between 1/11/02 & 31/10/11). Prospective study with reevaluation after treatment discontinuation after 3 years of age

RESULTS

•Between November 2002 and October 2011, 100 new-borns were confirmed with HC after TSH screening (62%females), **Incidence is 1:2828 new-borns**.

•Repartition was 51 dysgenesis (61% ectopy, 35% athyreosis, 4% hypoplasia) and 49 eutopic gland (20 CHP and 29 CHT).

•Incidence for CHP (after 3 years) was 1: 3983 infants.

•Congenital malformations in the whole group were found in 11% of neonates but none in the dysgenesis group.

•In the CHP group with dyshormonogenesis, molecular genetic studies identified **7 genes mutations (NIS, 3 Thyroglobulin, 2 TPO, GNAS).**

•In the CHT group, interestingly 1/3 of new-borns were premature babies and 2/3 were admitted in neonatal unit. **Investigations confirmed the use of iodized antiseptic in these situations (for caesarean, neonatal surgery or umbilical venous catheter).**

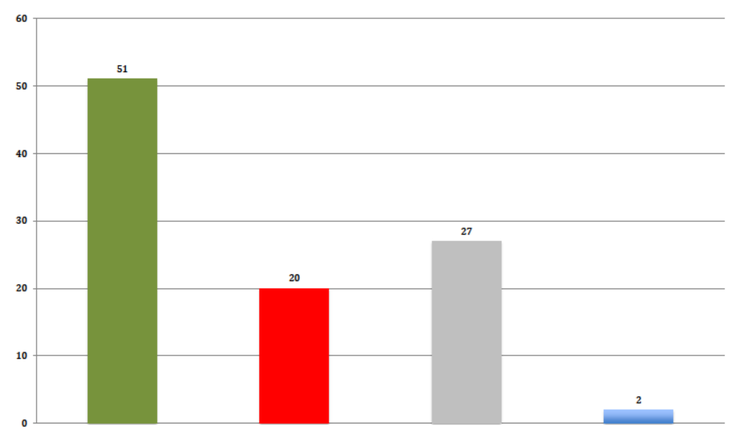

Fig 1:*Repartition between all diagnostics in the total population (n=100): Dysgenesis (51), Dyshormonogenesis(20),transient CH (27), maternal origin (2)*

Table 1:*Birth clinical parameters and biology at diagnosis; average (standart deviation) in total population and subgroups*

	Sexe G/B	Term (AW)	Weight (g)	Lenght (cm)	freeT4 pg/ml	freeT3 pg/ml	TSH> 150 en %
Total population n=100	1,63	38,6 (3,4)	3106 (825)	48,3 (4,1)	6,83(4,6)	2,9(1,5)	63
Dysgénesis n=51	2	39,9(1,5)	3366 (544)	49,6(2,2)	5,26(3,25)	2,4(1,2)	92
Euthopic gland n=49	1,33	37,3 (4,3)	2836 (978)	46,8	8,48 (5)	3,5 (1,8)	35
Permanent CH n= 20	1,22	38,5 (2,9)	2986 (785)	47,8 (4,8)	8,21 (5)	3,5 (1,9)	45
Transient CH n= 27	1,41	36,44 (4,8)	2733 (1174)	46 (5,35)	8,67 (5,4)	3,48 (1,74)	24

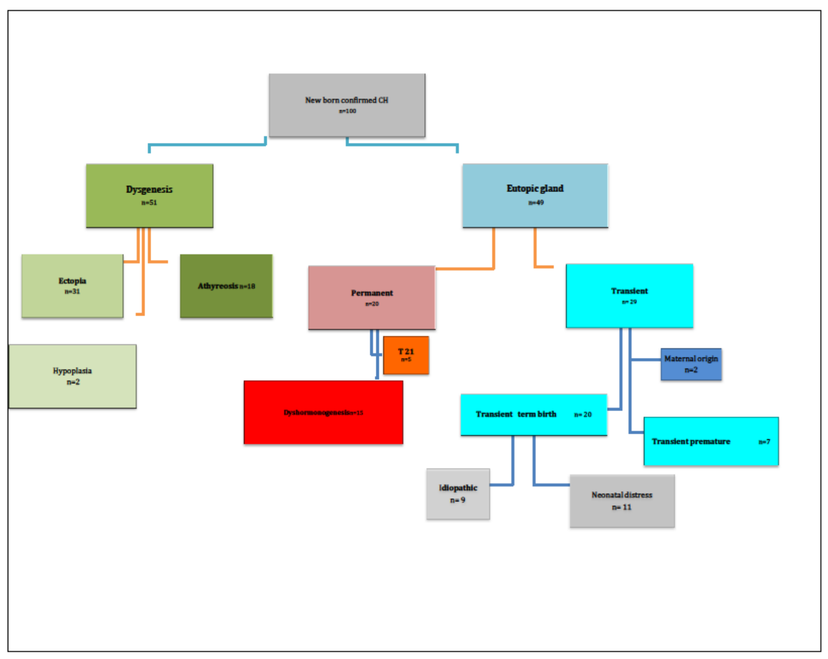

Fig 2*: Distribution between permanent and transient forms CH and all different diagnosis performed*

CONCLUSIONS

We found in our large French region a CH incidence as closed to 1:3000 new-borns, similar to French CH incidence. We confirmed the trend to an increased proportion of eutopic gland compared to dysgenesis. When regarding only CHP, this proportion also remains higher to the one classically described.

Références

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Pituitary

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