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THYROID DISORDERS IN SIBLINGS OF CH PATIENTS WITH THYROID DYSGENESIS.

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INTRODUCTION

Thyroid dysgenesis has been considered a sporadic disease, but recent observations suggested a possible genetic basis.

OBJECTIVE

To evaluate the incidence of **hormonal** and **ultrasound thyroid anomalies** in siblings of CH patients with thyroid dysgenesis.

DESCRIPTION OF METHODS

In Emilia-Romagna Region (Italy) 328 CH infants were diagnosed by neonatal screening between January 2000 and December 2012. 122 cases of permanent CH due to thyroid dysgenesis were enrolled in this study (63 ectopic gland, 28 athyreosis and 31 hypoplasia).

INCLUSION CRITERIA

- Confirmation of CH diagnosis at our Screening Centre
- Parents' informed consent
- Availability of thyroid hormonal and US data in siblings of CH cases.

RESULTS

In 49/122 families 65 siblings (7 twins) over the CH patient were found.

19/65 siblings (29,2%) showed subclinical hypothyroidism.

CH patients with Thyroid Dysgenesis		Affected siblings: Thyroid Morphology	Age of diagnosis in affected siblings	TSH serum (mU/l)	FT4 (pg/ml)
1.	Athyreosis (M)	Ts (M)	4 years	8	11,2
2.	Ectopy (M)	Ts (F)	4 years	7,37	11,3
3.	Ectopy (F)°	Hypoplasia(F)	5 years	7,3	16,6
4.	Ectopy (F)	Ts* (F)	21 days	6,52	16,4
5.	Ectopy (F)	Hypoplasia* (M)	15 days	113,4	13,1
6.	Hypoplasia (M)	Hypoplasia* (M)	1 month	9,33	12,6
7.	Hypoplasia (M)	Hypoplasia* (M)	2 months	7,14	15,3
8.	Hypoplasia (F)	Ts* (F)	1 month	12,64	11,7
9.	Hypoplasia(F)°	Ts*(F)	6 days	5,74	11,8
10.	Hypoplasia(M)	Ts (M)	3 years	5,44	13,5
11.	Hypoplasia(F)	Hypoplasia (M)	6 months	10,27	15
12.	Hypoplasia(M)^{T}	Ts (M)	19 years	5,74	10,1
		Ts (F)	16 years	6,07	11,8
		Ts (M)	15 years	6,11	11,7
		Ts (F)	12 years	5,38	11,5
		Ts (M)	10 years	6,54	14,5
		Ts* (M)	1 month	8,27	15
13.	Hypoplasia(F)	Goiter (M)	6 years	7,29	14,2
		Ts (M)	4 years	5,83	13,6

Cut-off serum values: TSH 0,5-4.5 mU/l, FT4 9,0-17,0 pg/ml
**=twin*
°=Asian ethnic group
^{T}= Mutation of TSH-receptor P162A(exon6)

✓Thyroid anomalies were found in all twins examined.

✓L-Thyroxine therapy was needed in 7 cases (table, cases n° 2, 5, 6, 7, 8, 10, 11.)

✓CH screening test was negative in all siblings.

✓The frequency of positive history for thyroid diseases was similar in each group.

CONCLUSIONS

Monitoring of thyroid function is strongly recommended in siblings of patients with thyroid dysgenesis regardless of the result of the screening test. The type of thyroid disorders found in our sample of siblings seems to suggest a multifactorial origin of CH in which genetic and environmental risk factors can play a role.

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