

Clinical Features of Newborn with Congenital Hypothyroidism Diagnosed by Neonatal Screening: Single Center Experience

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Background: It is well known that congenital hypothyroidism (CH) is the most important cause of preventable mental and motor retardation. Neonatal screening programs are necessary in the diagnosis of the disease. Our aim was to compare of clinical features of newborns with and without CH who were detected in screening programme between 2009 and 2014.

Material and Methods: The study enrolled 710 infant (348 girls) referred from CH-screening programme to our clinic. All newborns were examined. Serum TSH and fT4 was measured. The patients who have Serum TSH value smaller than 10 $\mu\text{IU/mL}$ ($\text{TSH} < 10 \mu\text{IU/mL}$) and free T4 values larger than 0.7 ng/mL ($\text{fT4} > 0.7 \text{ ng/mL}$) are accepted as euthyroid; the ones who have $\text{TSH} \geq 10 \mu\text{IU/mL}$ and $\text{fT4} \geq 0.7 \text{ ng/mL}$ subclinical hypothyroid; and others who have $\text{TSH} \geq 10 \mu\text{IU/mL}$ and $\text{sT4} < 0.7 \text{ ng/mL}$ are regarded as overt hypothyroidism L-Thyroxin (LT4) was prescribed and thyroid ultrasonography (USG) was performed.

Results of All Screening Group:

Median age was 23 (18) days in study-population. Although, 68.7% of children are brought to hospital within first month of their life; 25.5 % were brought between 30 and 90 days of age and 5.63% of them were brought while they were older than 91 days of age (Table 1). When we compare infants with and without CH, although significant difference was found in admission weight ($4292 \pm 1151 \text{ gr}$ vs $3950 \pm 782 \text{ gr}$, $p < 0.001$), length ($53.9 \pm 4 \text{ cm}$ vs $52.1 \pm 4 \text{ cm}$, $p = 0.012$) and age [22 (17) days vs 28.1(21) days, $p = 0.01$] between infants with and without CH, birth weights of the infants were similar each other ($3271 \pm 468 \text{ gr}$ vs $3186 \pm 624 \text{ gr}$, respectively). There was positive correlation between gender and birth weight ($\rho: 0.111$, $p = 0.007$). Infants who's younger than 31 days of age at admission, weight at admission ($p < 0.001$), birth weight ($p = 0.004$) and L-T4 dosage at the beginning of therapy ($5.86 \pm 2.6 \mu\text{g/kg/day}$ for girls vs $6.17 \pm 1 \mu\text{g/kg/day}$ for boys, $p = 0.023$) were statistically different among girls and boys.

Results of Infants with Congenital Hypothyroidism:

CH was diagnosed in 16.7 % of infants. Although significant difference was found in admission weight ($p < 0.001$), length ($p = 0.012$) and age ($p = 0.01$) between infants with and without CH, birth weights of the infants were similar each other. Birth weight of infant with CH whose admission age more than 91 days was different than infant with or without CH (Figure 1). There was a negative correlation between birth weight and fT4 in infants who admitted to clinic before 30 days of their life ($r: -0.297$, $p = 0.009$). Of the 69% infants with CH was diagnosed in first month.

Thyroid USG (n:72) revealed hypoplasia 64.6%, dyshormonogenesis 23.5% and agenesis 7.4%. Mean L-T4 dose was $8.7 \pm 2.4 \mu\text{g/kg/d}$ for agenesis group, $7.5 \pm 2.99 \mu\text{g/kg/d}$ for hypoplasia group and $7 \pm 1.9 \mu\text{g/kg/d}$ for dyshormonogenesis group (Figure 2). L-T4 dose was significantly correlated with serum TSH ($r: 0.505$) and fT4 ($r: -0.230$).

There was negative correlation between the age at the beginning of the therapy and serum TSH levels ($\rho: -0.399$; $p < 0.001$). Serum fT4 level and LT4 dose for normalization of serum TSH level were negatively correlated ($\rho: -0.550$; $p < 0.001$). Correlation of LT4 dose at the beginning of therapy with fT4 level ($\rho: -0.566$; $p < 0.001$), with age at the beginning of therapy ($\rho: -0.238$; $p = 0.017$) and with TSH normalization age ($\rho: -0.238$; $p = 0.021$) were negative; on the other hand correlation of LT4 dose at the beginning of therapy with serum TSH positive ($\rho: 0.381$; $p < 0.001$). In addition to that; LT4 dose at the beginning of therapy was positively correlated with TSH normalization dose ($\rho: 0.791$; $p < 0.001$).

Conclusion: Our study determined that one fifth of infants with CH weren't diagnosed before than first month. Weight and length of the infants with CH were lower than the healthy infants. Deterioration of cognitive function can be seen in untreated children with CH. Presence of high neonatal TSH in screening, serum TSH should be obtained and treatment should be initiated as soon as possible.

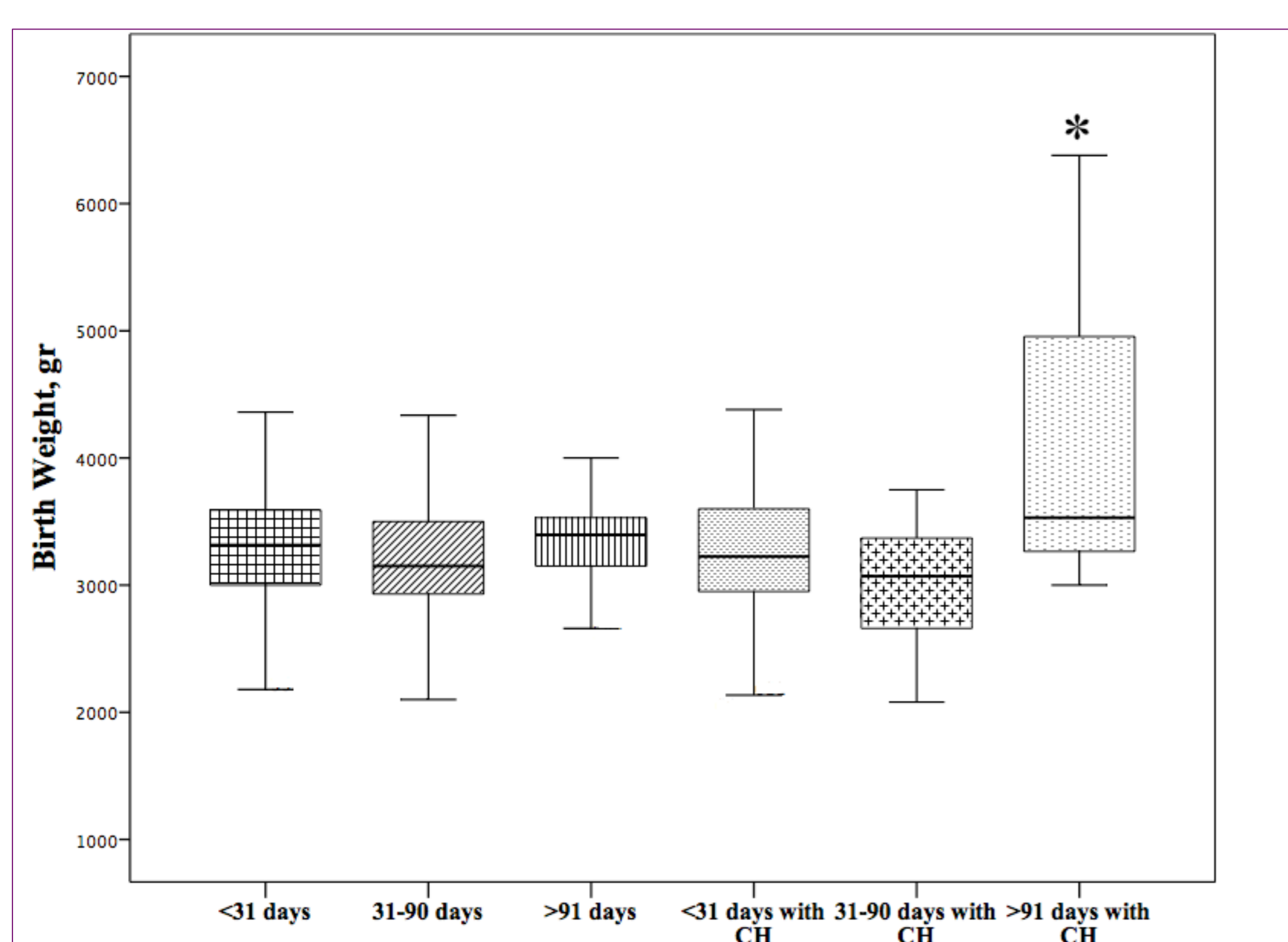


Figure 1: Birth weight and admission age of the infants with and without congenital hypothyroidism (* $p = 0.01$)

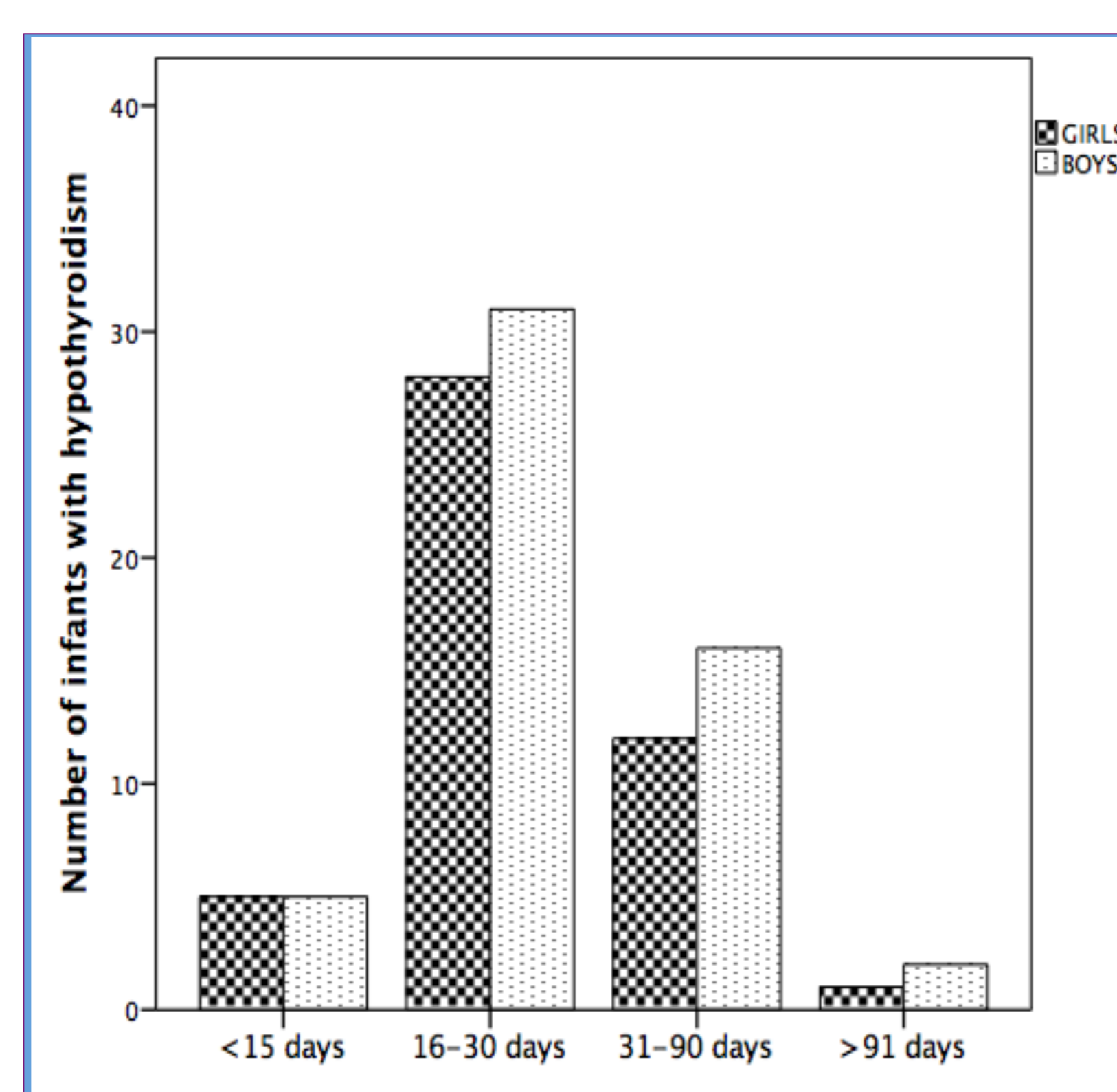


Figure 2: Distributions of the infants' age according the sexes

Table 1. Clinical and Laboratory Features of The Study Population According to Admission Age

	Group 1 (<31 days) n:489	Group 2 (31-90days) n:181	Group3 (>91days) n:40	p
Age at admission, day	19 \pm 5.9	47.3 \pm 15	162 \pm 88	
Weight at admission, gr	3872 \pm 621	4756 \pm 897	6947 \pm 1817	<0.001
Length at admission, cm	53.3 \pm 22	58.1 \pm 40	64 \pm 4.7	<0.001
Head circumference at admission, cm	37.5 \pm 21	37.5 \pm 1.8	41.7 \pm 4.7	
Birth weight, gr	3277 \pm 473	3131 \pm 549	3441 \pm 659	0.007
First capillary TSH, mIU/mL	27.9 \pm 23	15.8 \pm 12	28.3 \pm 23	0.008
Serum TSH, mIU/mL	4.2(2.8)*	3.7(2.74)*	2.62 \pm 1.41	<0.001
fT4 ng/mL	1.07 \pm 0.29	1.05 \pm 0.28	1.0 \pm 0.16	0.004
Age at the beginning of therapy, day	26.5 \pm 17	48.9 \pm 4	71 \pm 69	
LT4 dose, $\mu\text{g/kg/day}$	7.92 \pm 2.6	5.77 \pm 2.2	10.8 \pm 3.4	
Age at normalization of serum TSH level, day	65.3 \pm 44	82.5 \pm 40	138 \pm 63	
Dose at normalization of serum TSH level, $\mu\text{g/day}$	30.8 \pm 10	24.5 \pm 9.3	37.5 \pm 17	

* median (IQR)

