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 infants (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 µIU/mL (TSH>10 µIU/mL) and free T4 values larger than 0.7 ng/mL (FT4>0.7 ng/mL) are accepted as euthyroid; the ones who have TSH>10 µIU/mL and FT4 ≥ 0.7 ng/mL subclinical hypothyroid; and others who have TSH>10 µIU/mL and ST4 <0.7 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's are brought to hospital within first month of their life; 25.5 % were brought between 30 and 90 days of age and 5.6% 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±1151gr vs 3950±782 gr, p<0.001), length (53.9±4cm vs 52.1±4 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±468 gr vs 3186±624 gr, respectively). There was positive correlation between gender and birth weight (p: 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±0.6 µg/kg/day for girls vs 6.17±1 µg/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 prescribed and thyroid ultrasonography (USG) was performed.

Conclusion: Our study determined that one fifth of infants with CH weren't diagnosed before than first month. Weight and lenght 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.