



## TITLE: Case series on Spectrum of disorders of Thyroid dysgenesis in congenital Hypothyroidism at a tertiary care centre

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### ABSTRACT (upto 300 words)

**AIM AND OBJECTIVE:** 1) PRIMARY OBJECTIVE: TO REINFORCE THAT THYROID DYSGENESIS HAS VARIED AGE OF PRESENTATION AND VARIED SEVERITY OF PRESENTATION

2) SECONDARY OBJECTIVE : TO REINFORCE THE IMPORTANCE OF NEW BORN SCREENING.

#### MATERIALS AND METHODS:

**Place of study :** Niloufer hospital

• **Duration of study :** 18 months

• **No. of subjects :** 15

• **Inclusion criteria :**

1) Patients presenting with symptoms of hypothyroidism

2) patients less than 5 years of age

• **Exclusion criteria :** 1) patient with other congenital anomalies or syndromes.

#### RESULTS & DISCUSSION:

Total **15 cases** of thyroid dysgenesis are included in this case series

• Out of which : **10 cases (66.6%)** of thyroid agenesis, **4 cases (26.6%)** of ectopic thyroid, **1 case (6.6%)** of thyroid hypoplasia.

- Male:Female ratio - 1.1:1

- **8 cases (53.3%)** - less than 1 year and **7 cases (46.6%)** - more than 1 year

- **12 cases (80%)** - developmental delay with other symptoms like constipation, short stature.

**2 cases (13.3%)** - prolonged jaundice and

**1 case (6.6%)** - cough and difficulty while swallowing.

- **9 cases** are in followup, among those cases

**TSH levels got normalised after 1 month** of starting levothyroxine and clinical improvement was been observed in most of the cases.

• In our study **most of the cases presented to us with developmental delay** and they were been **missed from newborn screening** during early infancy **except 2 cases hence they had better outcome.**

• Cases detected to have hypothyroidism were been started with levothyroxine and followed up for once in 2 weeks for first 3 months, once a month after 3 months of starting therapy.

**CONCLUSION:** This study is to emphasise that thyroid dysgenesis has varied age of presentation and varied severity of presentation.



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- To emphasise the importance of newborn screening and early detection of congenital hypothyroidism and for prevention and treatment of intellectual disability.
- In our study most of the cases has showed clinical improvement after starting with levothyroxine.

