HYPOTHYROIDISM

DEFINITION: thyroid hormone *deficiency*

Congenital: thyroid gland development defect (dysgenesis) (85%), thyroid hormone synthesis defect (10-15%)

Acquired: autoimmune (Hashimoto's), drugs (antiepileptics, etc), thyroid injury (radiation), iodine deficiency (rare)

More common with family history of autoimmune conditions, Turner Syndrome, and Down's Syndrome

Most common **preventable** cause of intellectual disability

PATHOPHYSIOLOGY

Normal regulation of HPT axis

Hypothalamus
↓ **TRH**
Anterior Pituitary
↓ **TSH**
Thyroid
↓ **T4→T3**
Bloodstream

Dysregulation of HPT axis in hypothyroidism

3º hypothyroidism – hypothalamic dysfunction (↓ TRH)

2º hypothyroidism – pituitary dysfunction (↓/N TSH)

1º hypothyroidism – inflammation/destruction of the thyroid (↑ TSH, ↓T4)

CLINICAL MANIFESTATIONS

Infants (often asymptomatic)

Early signs:
- Prolonged jaundice
- Pallor
- Large anterior fontanelle
- Hypotonia
- Edema
- Hypothermia

Late signs:
- Poor feeding
- Poor growth
- Umbilical hernia
- Developmental delay
- Macroglossia
- Lethargy

Children & Adolescents

- ***Short stature***
- Delayed bone age
- Puberty delay
- Menstrual irregularity
- Goiter
- Fatigue
- Constipation
- Dry Skin

DIAGNOSIS

- **Routine newborn screening** used before clinical signs develop and if positive, confirm with thyroid function tests
- In children, consider ordering a **TSH** if there is clinical suspicion of hypothyroidism

INVESTIGATIONS

Type	TSH	T4	Abnormality
Primary	↑	↓	Thyroid
Secondary/ Tertiary	↓	↓	Pituitary/ Hypothalamus
Subclinical	↑	-	Thyroid

MANAGEMENT

Congenital

- Start Levothyroxine **immediately**
- Confirm etiology with **thyroid ultrasound**
- Monitor TSH frequently

Acquired

- Start Levothyroxine & treat until TSH returns to normal
- Check **antithyroglobulin antibodies**
- Monitor TSH every 6-12 months & after dose change

- **Subclinical hypothyroidism**: continue to monitor and treat if symptoms or a goiter develop

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