



ENDOCRINE FACIES

OVERVIEW

- **Distinctive facial expression or appearance** associated with a specific medical condition
- Endocrine facies: distinct facial features due to hormone excess or deficiency

ANTERIOR PITUITARY GLAND

1. Dwarfism due to **Panhypopituitarism**:

a) Generalized decrease in secretion of all anterior pituitary hormones

b) Can cause **dwarfism**; rate of development of body parts is slowed down.

E.g. a child who has reached the age of 10 years may have the bodily development of a child aged 4 to 5 years; **No puberty as no gonadotropins are released.**

C) 1/3 of such cases can be due to **GH deficiency only**; undergo puberty. Dwarfs who have pure GH deficiency can be completely cured if treated early in life.

D) **Laron dwarfism and African pygmies**: normal GH secretion but mutation in GH receptor or inability to form IGF-1.



• Pituitary dwarfism vs Achondroplasia:

- The latter is due to **FGFR3 mutation**; AD inheritance. **Impaired growth of long bones.**



Types of Dwarfism

Disproportionate Dwarfism

Some average-size parts of the body and some shorter-than-normal parts of the body



Achondroplasia is the most common disproportionate dwarfism, causing a normal-size torso but short limbs

Proportionate Dwarfism

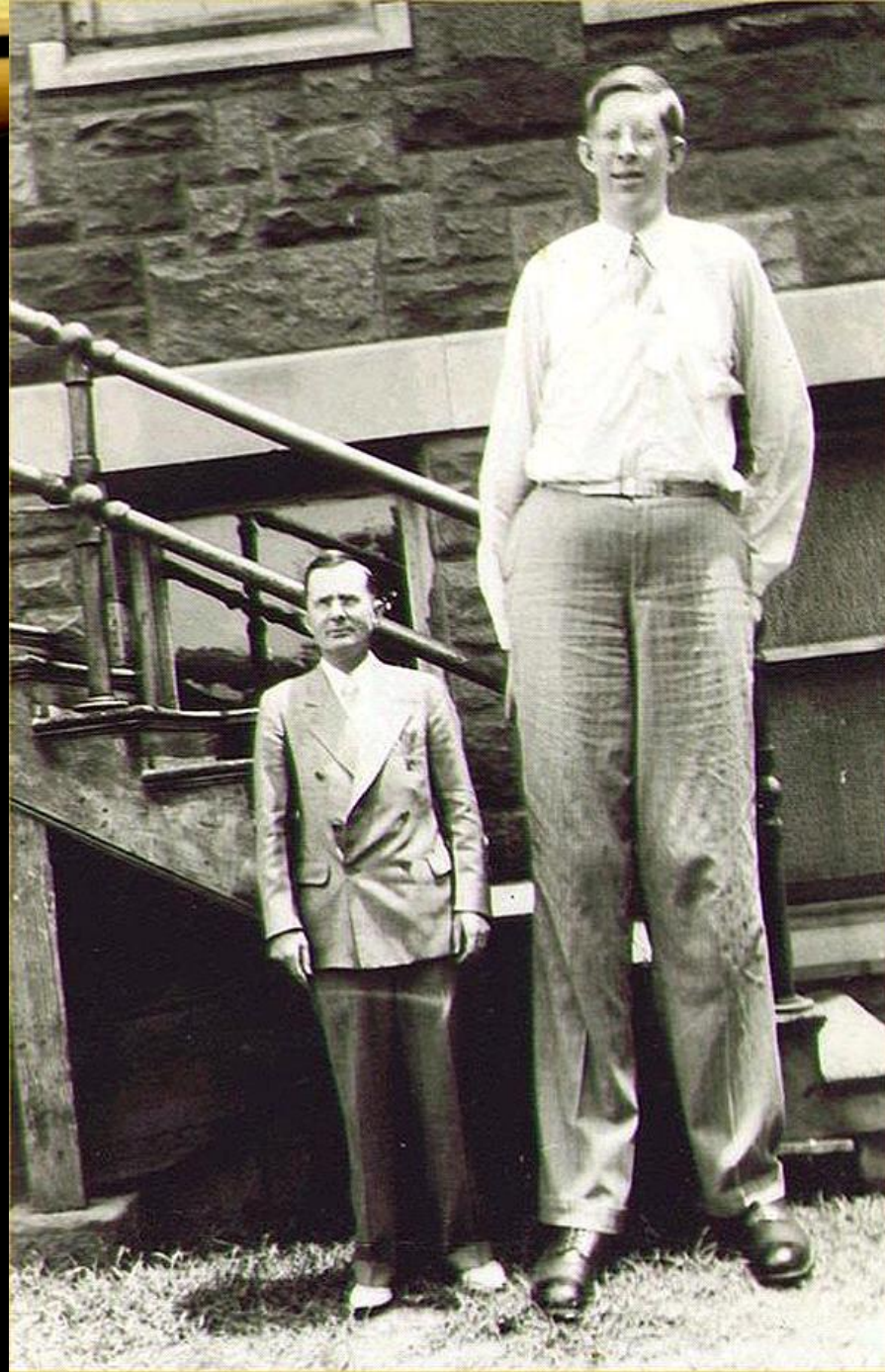
The individual is smaller than average all over



Includes growth-hormone deficiency dwarfism, primordial dwarfism, and Seckel syndrome

GIGANTISM

- Excess growth hormone secretion before adolescence.
- Occurs before the epiphyses of the long bones have become fused with the shafts, height increases so that the person becomes a giant—up to 8 feet tall.
- Normal facial features.



ACROMEGALY

- Excess growth hormone secretion after puberty; after epiphyses are fused → no linear growth → no increase in height. But bones can become thicker.
- Enlargement is especially marked in the bones of the **hands and feet**
- **Membranous bones**, including the **cranium, nose, bosses on the forehead, supraorbital ridges, lower jawbone, and portions of the vertebrae.**

- Consequently:
the **lower jaw protrudes forward**

the **forehead slants forward** because of excess development of the supraorbital ridges

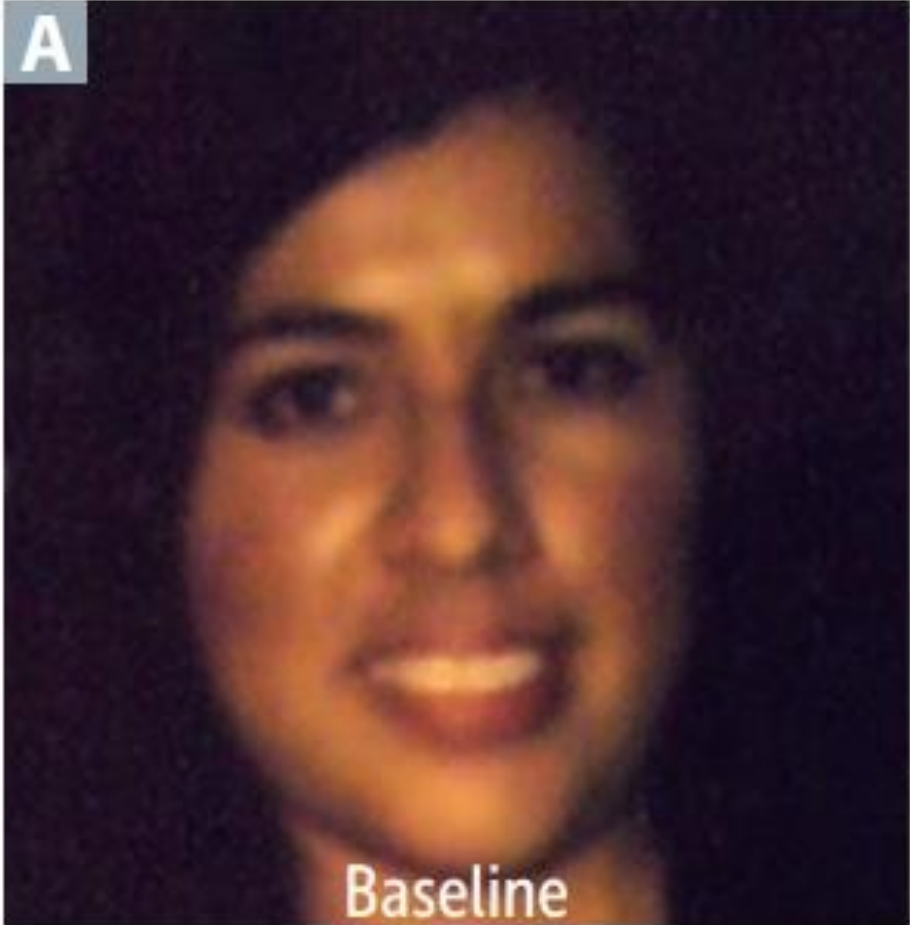
the nose increases to as much as twice normal size

the feet require size 14 or larger shoes, and the fingers become extremely thickened so that the hands are almost twice normal size

Changes in the vertebrae ordinarily cause a hunched back, which is known clinically as kyphosis.



A



Baseline



RU

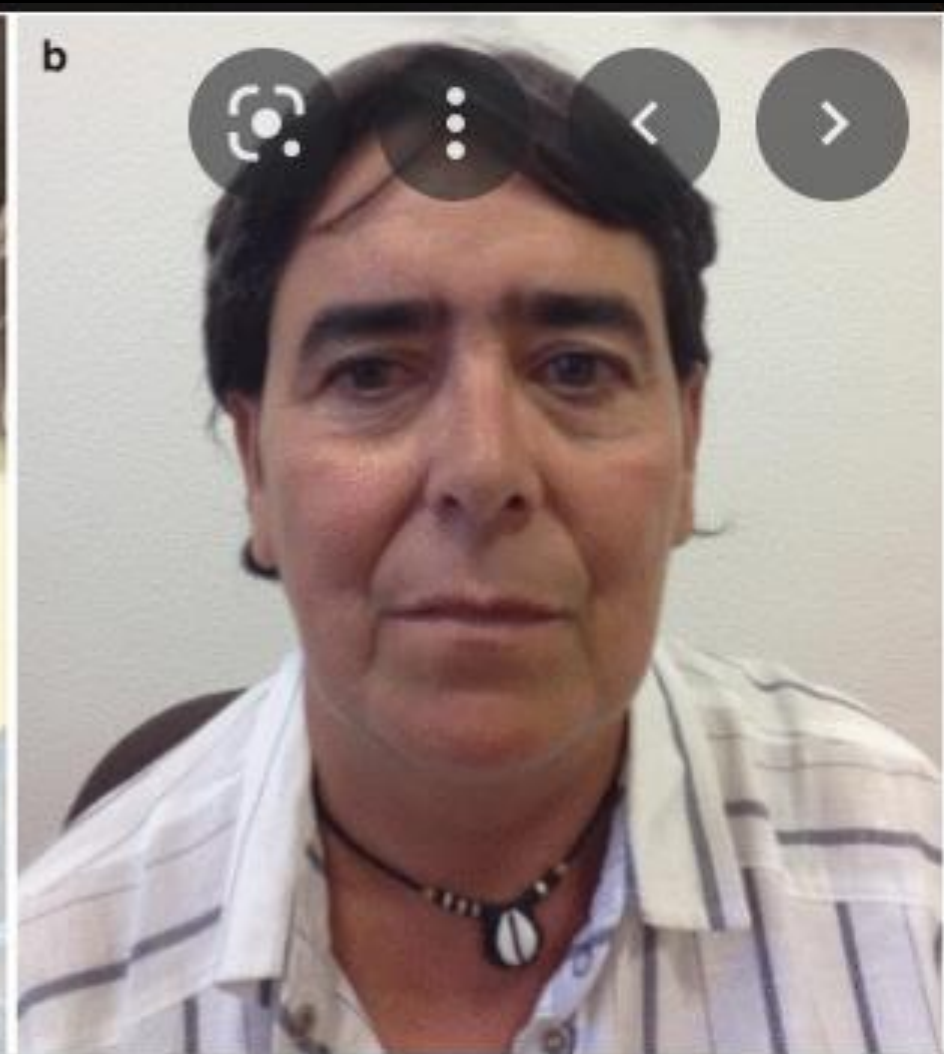
GRAVES DISEASE

- Most common cause of hyperthyroidism
- Hyperthyroidism can cause Exophthalmos—protrusion of the eyeballs and retraction of upper eyelids.
- Protrusion of eyeball can become severe such that eyelids do not close during blinking and sleeping— drying of cornea with eventual ulceration and blindness.
- Due to edematous swelling of retroorbital soft tissue and extraocular muscles.



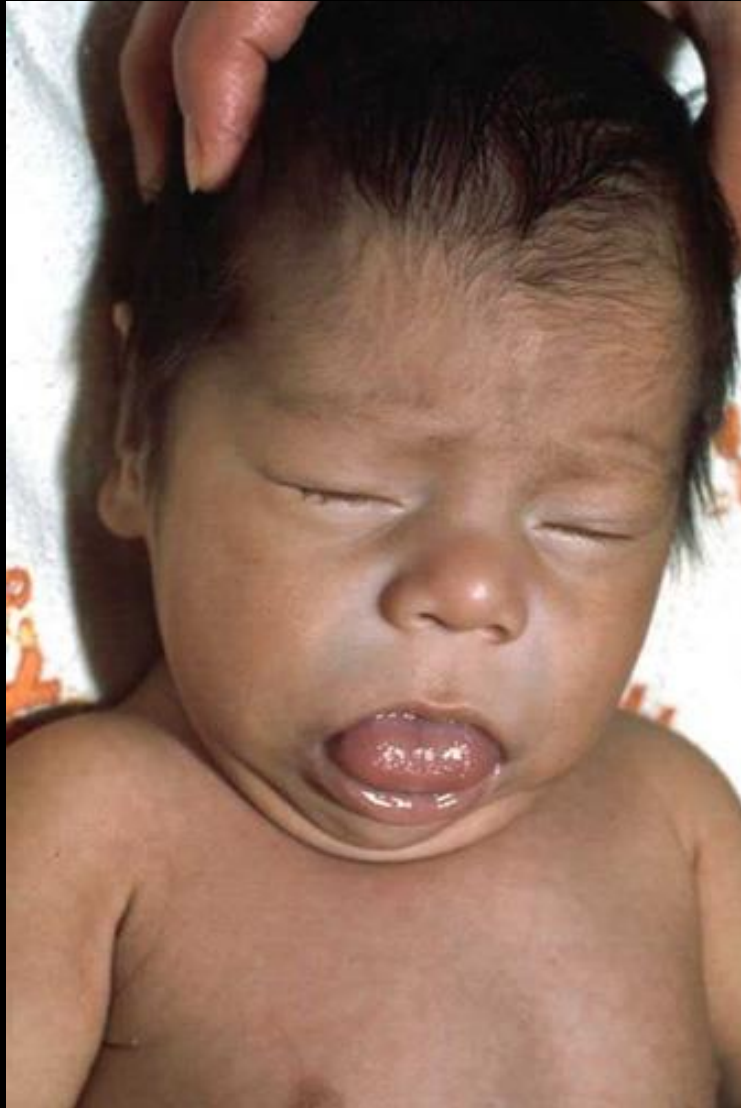
MYXEDEMA

- Due to hypothyroidism
- Bagginess under the eyes and swelling of the face.
- Due to excess GAGs accumulation in the interstitial spaces; nonpitting edema.



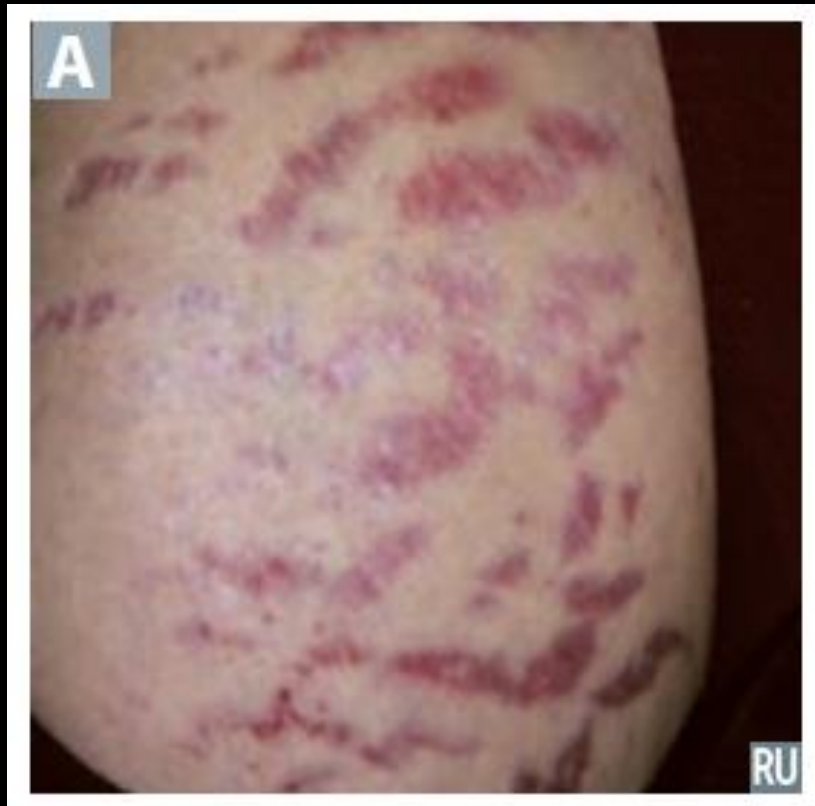
CRETINISM

- Extreme **hypothyroidism** during **fetal life, infancy, or childhood**



CUSHING SYNDROME

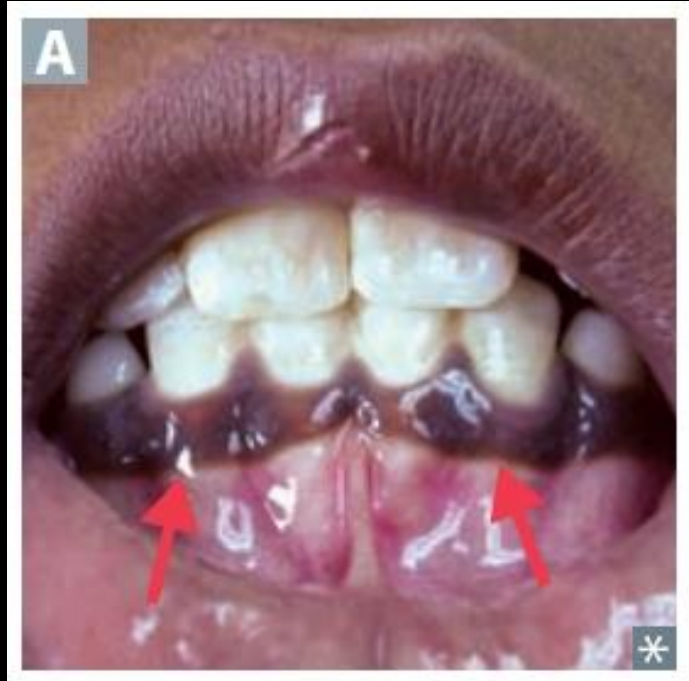
- Due to excess glucocorticoids (aka cortisol)
- Cause moon facies and buffalo hump
- Skin striae





ADDISON DISEASE

- Primary adrenal insufficiency
- Cause feedback increases in ACTH secretion
- skin/mucosal hyperpigmentation (increased melanin synthesis due to increased MSH, a byproduct of POMC cleavage)





THANK YOU