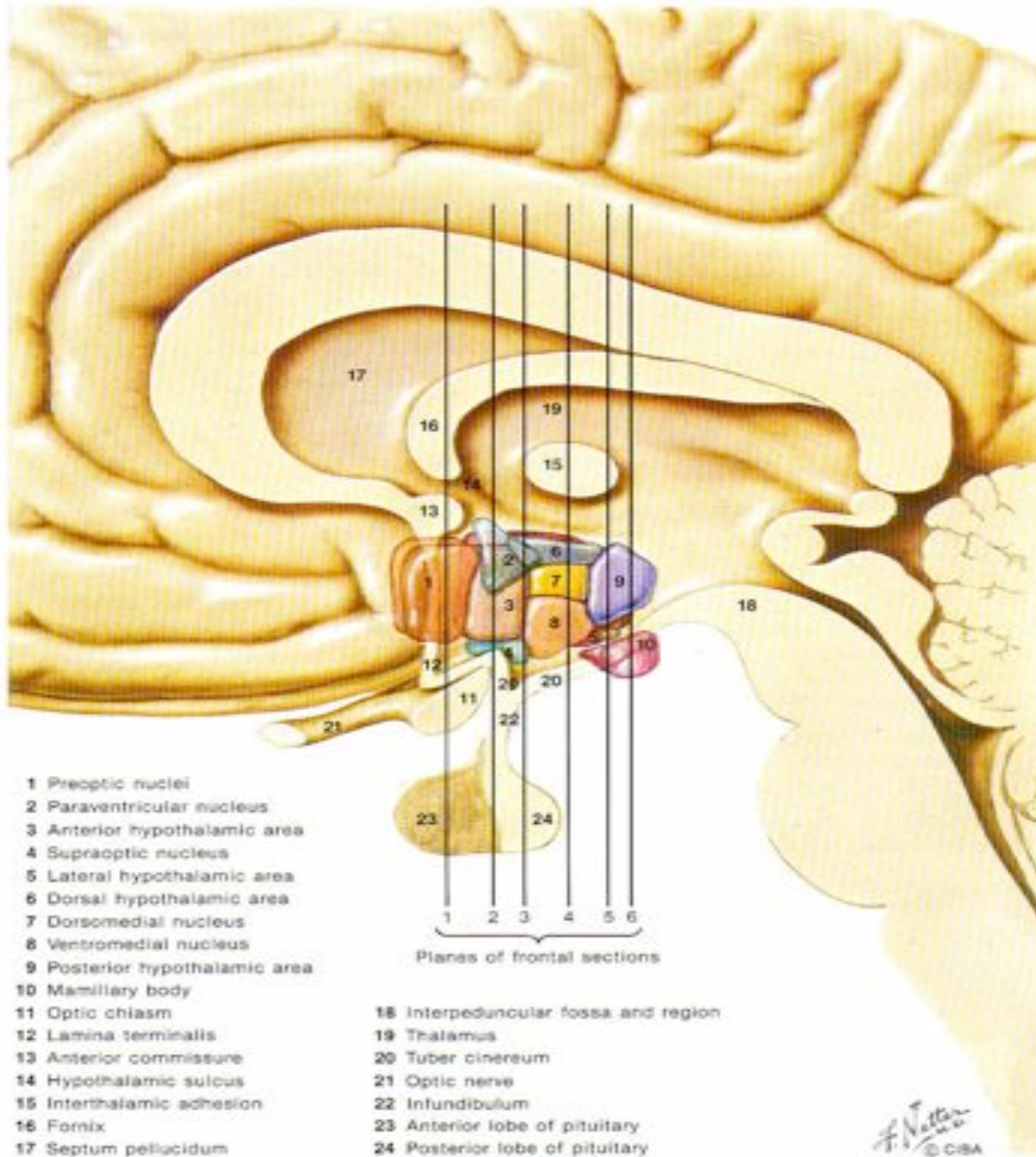


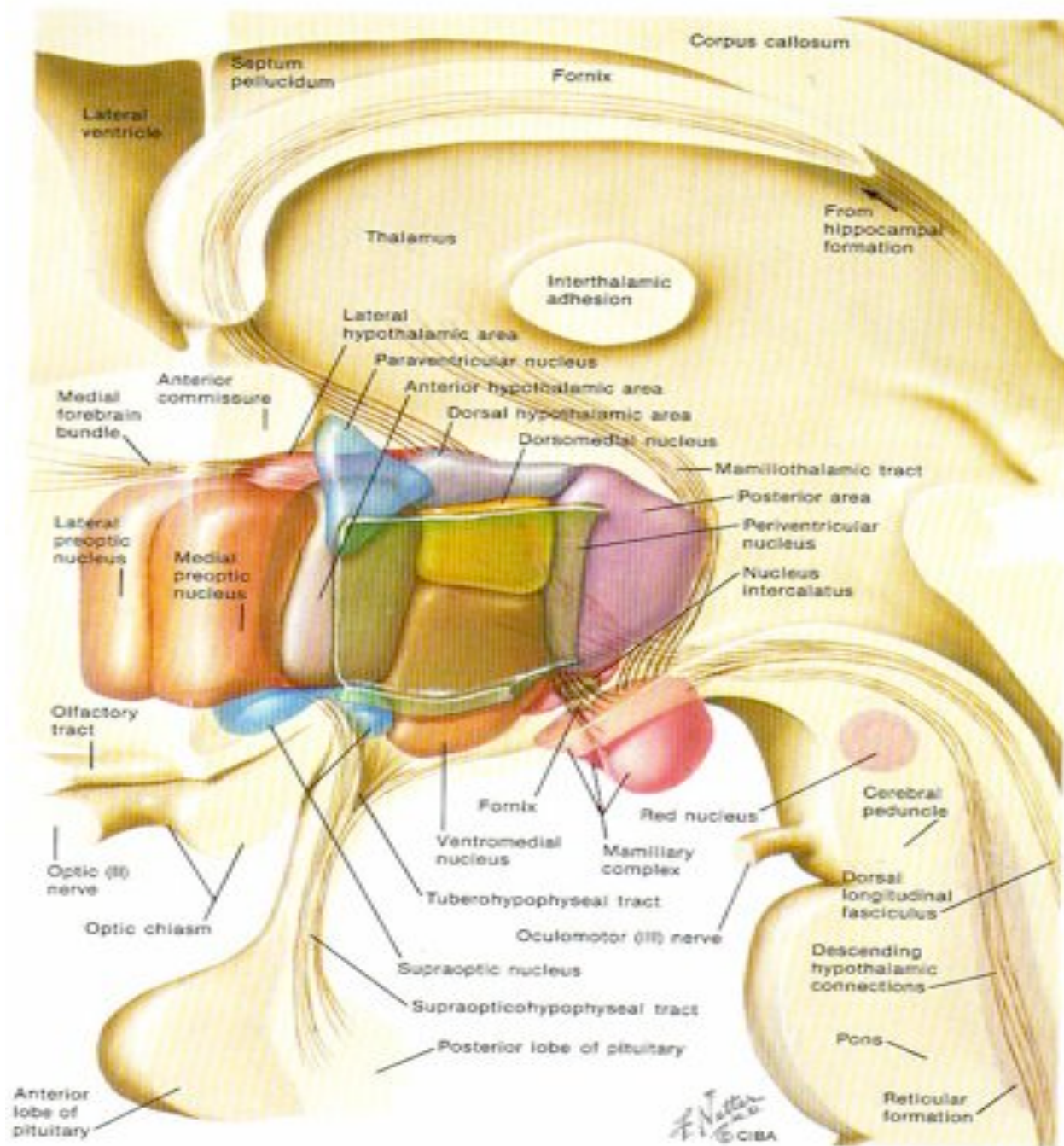
# Endocrinology and CHARGE Syndrome

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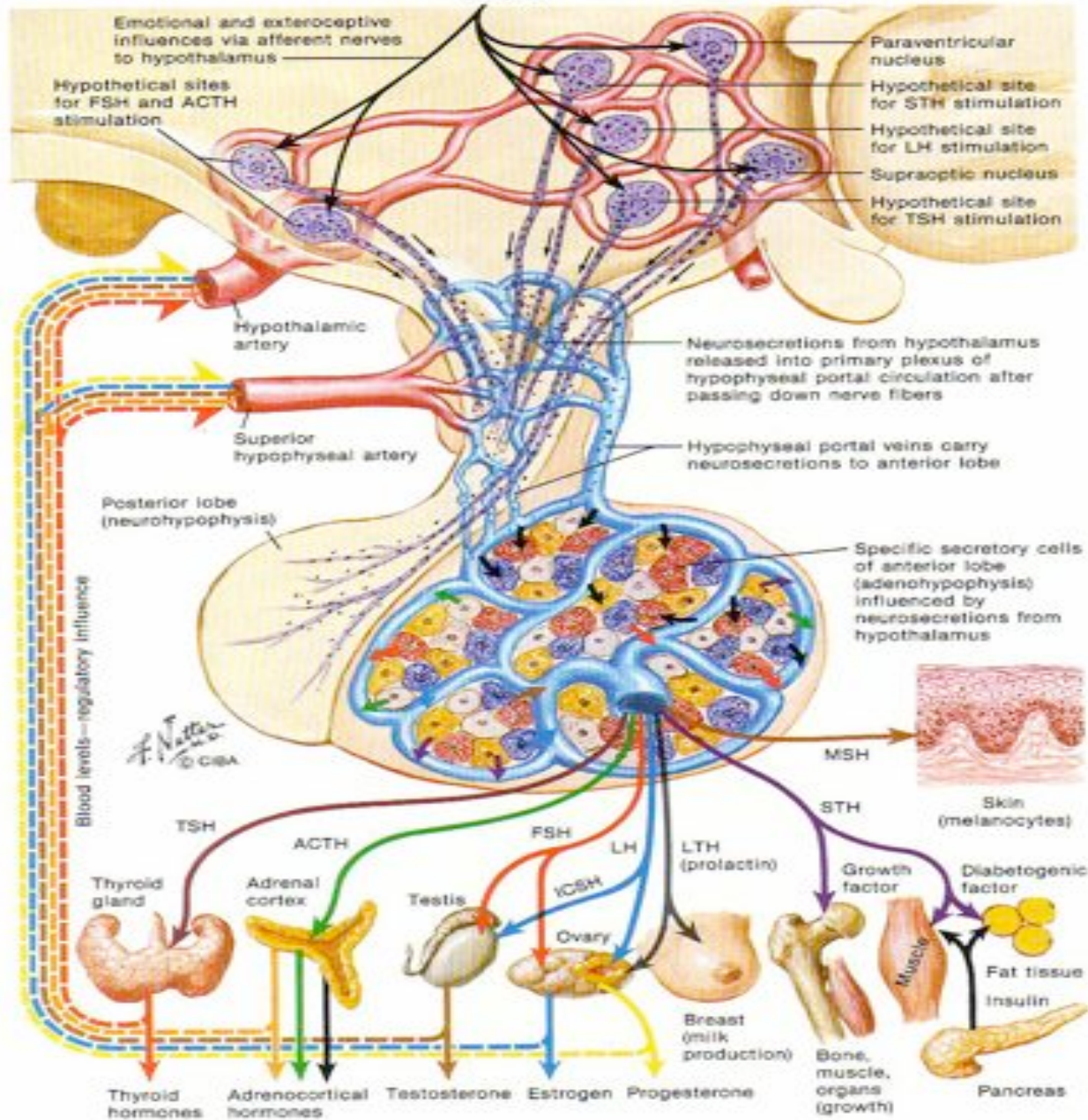
### General Topography of Hypothalamus



### Schematic Reconstruction of Hypothalamus



### Adenohypophyseal Hormones



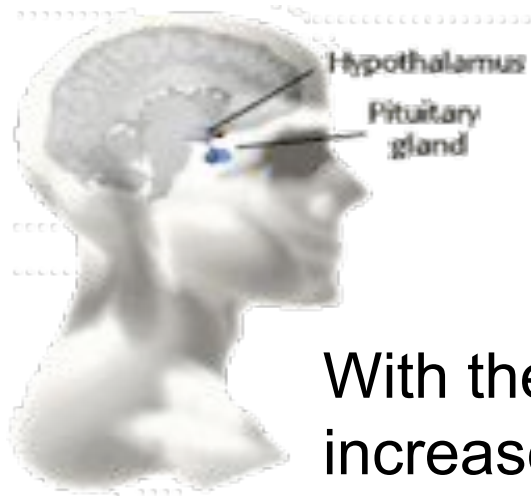
# Overview

- Hypoglycaemia, Hypoadrenalism
- Micropenis, undescended testes
- Small labiae
- Small stature
- Hypothyroidism
- Puberty delay or absence
- Osteoporosis

in boys

Foetal penis grows by testosterone stimulation by anterior pituitary LH

When this does not occur, this causes the micropenis






With the onset of puberty, the hypothalamus increases the release of gonadotropin releasing hormone (GnRH). GnRH is needed for sexual maturity and normal reproduction

GnRH stimulates the anterior pituitary gland to release hormones. Among these are the gonadotropins 'follicle stimulating hormone' (FSH) and 'luteinizing hormone' (LH)

These gonadotropins act by stimulating the production of sex hormones in the testis and ovary

**TABLE 1**  
Pubertal Milestones in Girls

Tanner stage	Breasts*	Standard	Public hair*	Standard	Growth	Other
1	Prepubertal elevation of papilla only		Prepubertal, villus hair only	—	Basal: about 5.0 to 6.0 cm (2.0 to 2.4 in) per year	Adrenarche Ovarian growth
2	Breast bud appears under enlarged areola (11.2 years)		Sparse growth of slightly pigmented hair along the labia (11.9 years)		Accelerated about 7.0 to 8.0 cm (2.8 to 3.2 in) per year	Clitoral enlargement Labia pigmentation Uterus enlargement

3

Breast tissue grows beyond areola without contour separation (12.4 years)



Hair is coarser, curled and pigmented; spreads across the pubes (12.7 years)



Peak velocity: about 8.0 cm (3.2 in) per year (12.5 years)

Axillary hair (13.1 years)  
Acne (13.2 years)

4

Projection of areola and papilla forms a secondary mound (13.1 years)



Adult-type hair but no spread to medial thigh (13.4 years)



Deceleration < 7.0 cm (2.8 in) per year

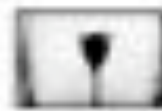
Menarche (13.3 years)  
Regular menses (13.9 years)

5

Adult breast contour with projection of papilla only (14.5 years)





Adult-type hair with spread to medial thigh but not up linea alba (14.6 years)



Cessation at about 16 years

Adult genitalia

**TABLE 2**  
**Pubertal Milestones in Boys**

<u>Tanner stage</u>	<u>Standard</u>	<u>Genitalia*</u>	<u>Pubic hair*</u>	<u>Growth</u>	<u>Other</u>
1		Prepubertal Testes: < 2.5 cm (1.0 in)	Prepubertal, villus hair only	Basal, about 5.0 to 6.0 cm (2.0 to 2.4 in) per year	Adrenarche
2		Thinning and reddening of scrotum (11.9 years)  Testes: 2.5 to 3.2 cm (1.0 to 1.28 in)	Sparse growth of slightly pigmented hair at base of penis (12.3 years)	Basal, about 5.0 to 6.0 cm (2.0 to 2.4 in) per year	Decrease in total body fat

3



Growth of penis, especially length (13.2 years)

Testes: 3.3 to 4.0 cm (1.32 to 1.6 in)

Thicker, curlier hair spreads to the mons pubis (13.9 years)

Accelerated: about 7.0 to 8.0 cm (2.8 to 3.2 in) per year

Gynecomastia (13.2 years)

Voice break (13.5 years)

Muscle mass increase

4



Growth of penis and glands, darkening of scrotum (14.3 years)

Testes: 4.1 to 4.5 cm (1.64 to 1.8 in)

Adult-type hair but no spread to medial thigh (14.7 years)

Peak velocity: about 10.0 cm (4.0 in) per year (13.8 years)

Axillary hair (14.0 years)

Voice change (14.1 years)

Acne (14.3 years)

5



Adult genitalia (15.1 years)

Testes: > 4.5 cm (1.8 in)

Adult-type hair with spread to medial thighs but not up linea alba (15.3 years)

Deceleration and cessation (about 17 years)

Facial hair (14.9 years)

Muscle mass continues to increase after Stage 5

# Hypogonadotropic Hypogonadism

- Lack of development at puberty
- Prepubertal testicular size in adolescence
- Absence of secondary sexual development
- Short stature
- Anosmia

# **Bone health is based on**

genetics

physical activity

nutrition

sex hormones of puberty

sun, vitamin D, pigmentation

fractures

# USEFUL WEBSITES and RESOURCES

## **Delayed puberty**

[www.pituitary.asn.au](http://www.pituitary.asn.au)

[www.bsped.org.uk/NN/DELAYP.htm](http://www.bsped.org.uk/NN/DELAYP.htm)

Patient booklet from Serono at  
[oceania@seronosymposia.org](mailto:oceania@seronosymposia.org)

# THYROID DISORDERS

- [www.thyroid.org.au](http://www.thyroid.org.au)
- [www.thyroidfoundation.com.au](http://www.thyroidfoundation.com.au)
- [www.chw.edu.au/prof/services/newborn/ch.htm](http://www.chw.edu.au/prof/services/newborn/ch.htm)
- [www.thyroidfoundation.org](http://www.thyroidfoundation.org)
- [www.endocrineweb.com](http://www.endocrineweb.com)
- [www.nevdgp.org.au/geninf/topics/thyroid\\_resources.htm](http://www.nevdgp.org.au/geninf/topics/thyroid_resources.htm)
- [www.thyroidmanager.org](http://www.thyroidmanager.org)

# GROWTH DISORDERS

- [www.magicfoundation.org](http://www.magicfoundation.org)
- [www.hgfound.org](http://www.hgfound.org)
- [http://groups.msn.com/  
KidsWithInhibitedGrowthEmailSupport](http://groups.msn.com/KidsWithInhibitedGrowthEmailSupport)
- [www.kidshealth.org/kid/grow/body\\_stuff/growth\\_  
disorders.html](http://www.kidshealth.org/kid/grow/body_stuff/growth_disorders.html)
- [www.hgfound.org/chat\\_CHAT.html](http://www.hgfound.org/chat_CHAT.html)

# GROWTH HORMONE THERAPY

- [www.pituitary.asn.au](http://www.pituitary.asn.au)
- [www.humangrowthhormonemedinfo.com](http://www.humangrowthhormonemedinfo.com)
- [www.howkidsgrow.com](http://www.howkidsgrow.com)
- [www.magicfoundation.org/divisions/ghd\\_ch/htm](http://www.magicfoundation.org/divisions/ghd_ch/htm)
- [www.mediaprowebcast.com/magic/events/webcast2.htm](http://www.mediaprowebcast.com/magic/events/webcast2.htm)
- Growth Problems in Children – information booklet from Serono at [oceania@seronosymposia.org](mailto:oceania@seronosymposia.org)

# HORMONE REPLACEMENT THERAPY

- [www.hormone.org](http://www.hormone.org)
- [www.menopause.org.au](http://www.menopause.org.au)
- <http://www.anedgp.com.au/alerts/hrtstudy/HRT%20Fact%20Sheet.pdf>
- [www.nps.org.au/Topics/nps.org.au\\_hrt.html](http://www.nps.org.au/Topics/nps.org.au_hrt.html)
- [www.npan.com.au](http://www.npan.com.au)
- Testosterone Implants Information Leaflet from Organon at [organon@au.organon.com](mailto:organon@au.organon.com)

# OSTEOPOROSIS

- [www.osteoporosis.org.au](http://www.osteoporosis.org.au)  
Osteoblast Magazine 1800 242 141
- [www.noca.asn.au](http://www.noca.asn.au) at [osteoprosis@nocasn.au](mailto:osteoprosis@nocasn.au)
- [www.osteofound.org](http://www.osteofound.org)
- [www.fore.org](http://www.fore.org)