Mahidol University
Faculty of Medicine Siriraj Hospital

SiENDO
Division of Endocrinology and Metabolism
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Reviews in internal medicine R3

Reproductive and growth disorders

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4-4-26

Long case R3

Short stature
- Turner's syndrome
- Digeorge syndrome
- Pseudohypoparathyroidism
- Rickets
- Systemic diseases

Tall stature
- Hypogonadism
- Marfan syndrome
- Gigantism

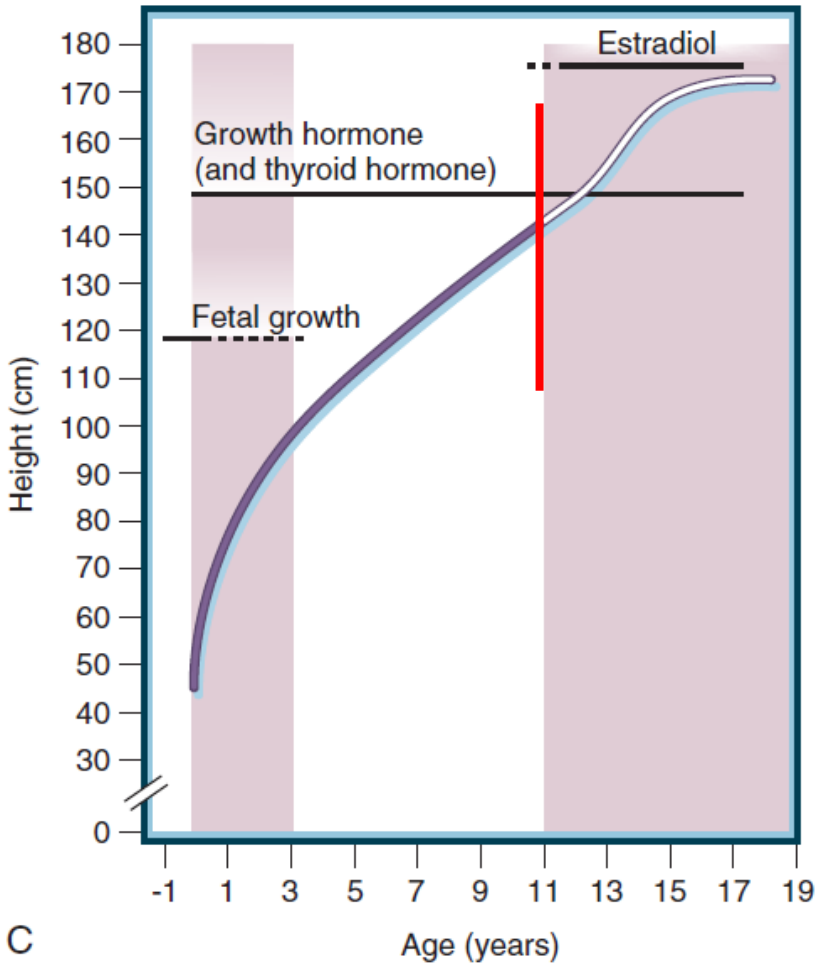
Gynaecomastia
- Hypogonadism
- Kennedy disease
- Feminizing ACC

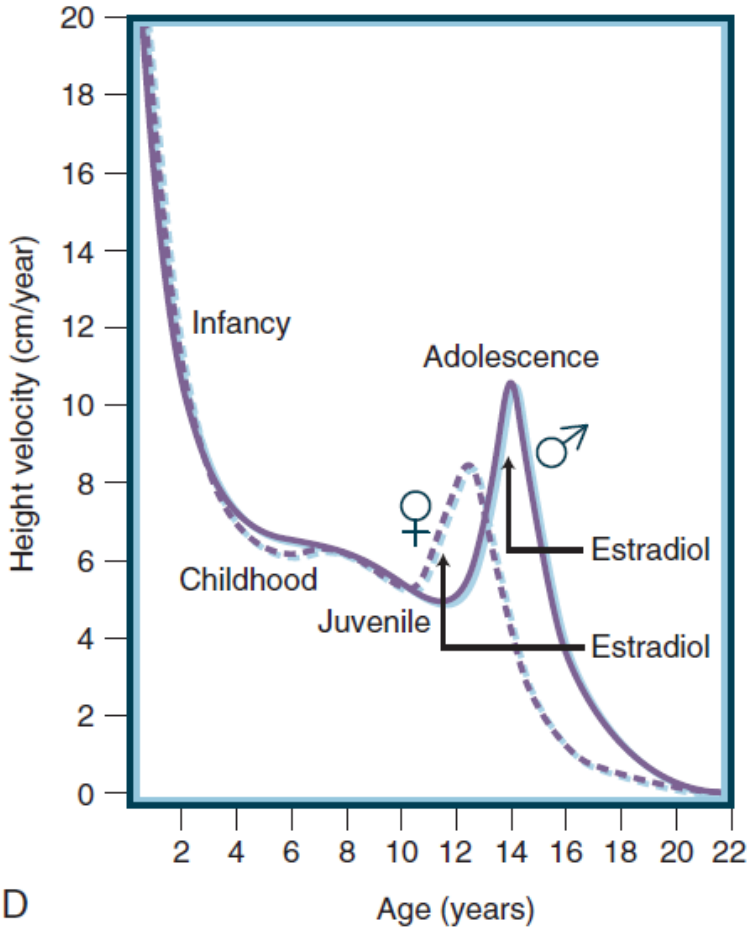
Androgen excess
- PCOS
- CAH: esp. 21OHD def
- Virilizing ACC

Physiology of growth

Nutrition
GH
Thyroid hormone

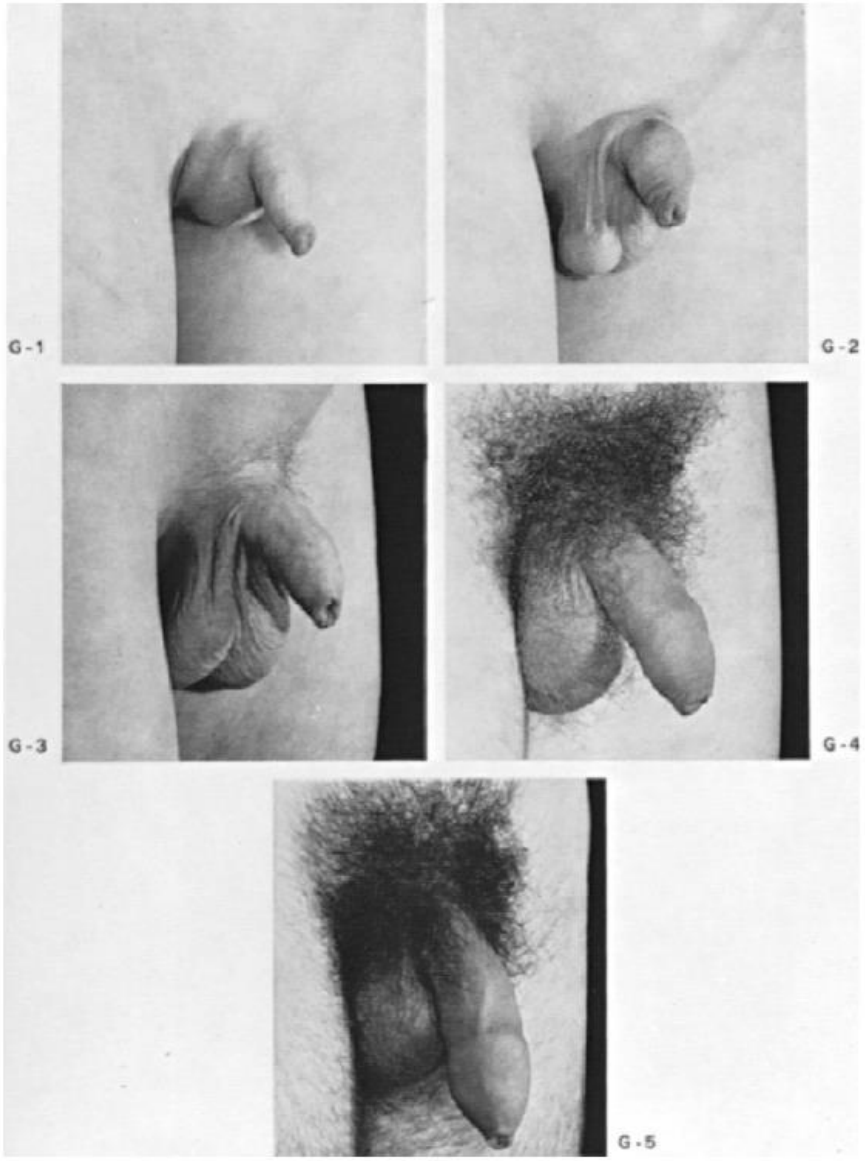
Sex steroids
- Increase GH
and IGF-1



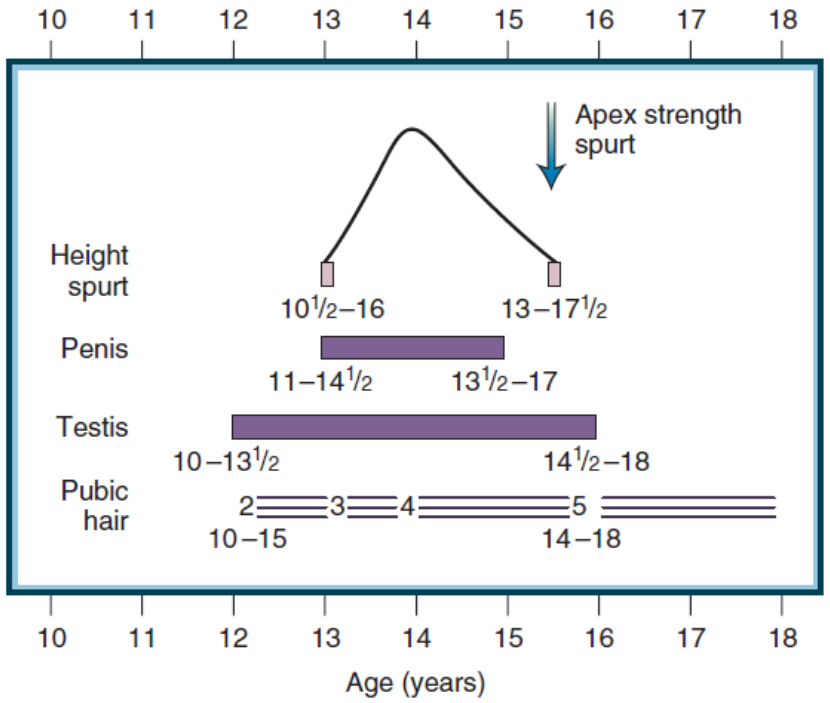


William textbook endocrinology

Physiology of growth



Male



Puberty : Boy

Testicular enlargement 4 mL or 2.5 cm

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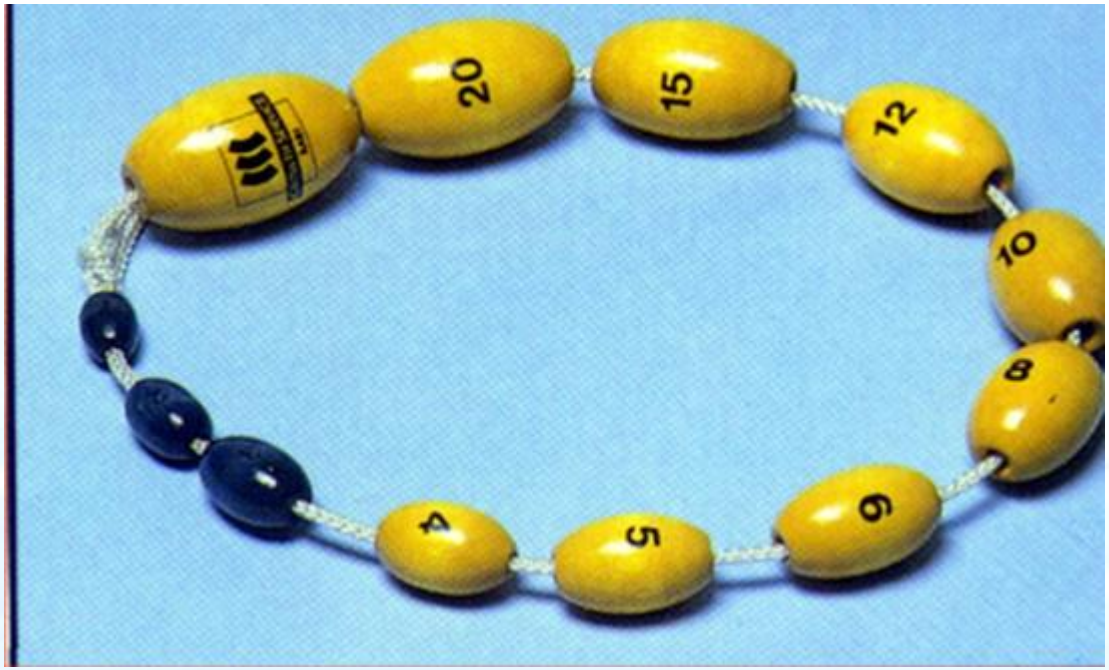
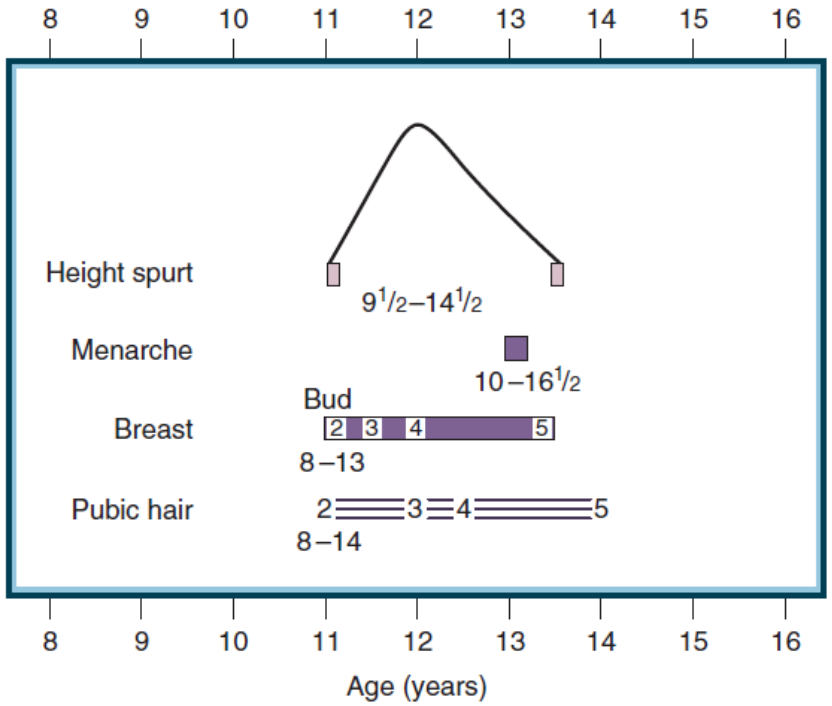
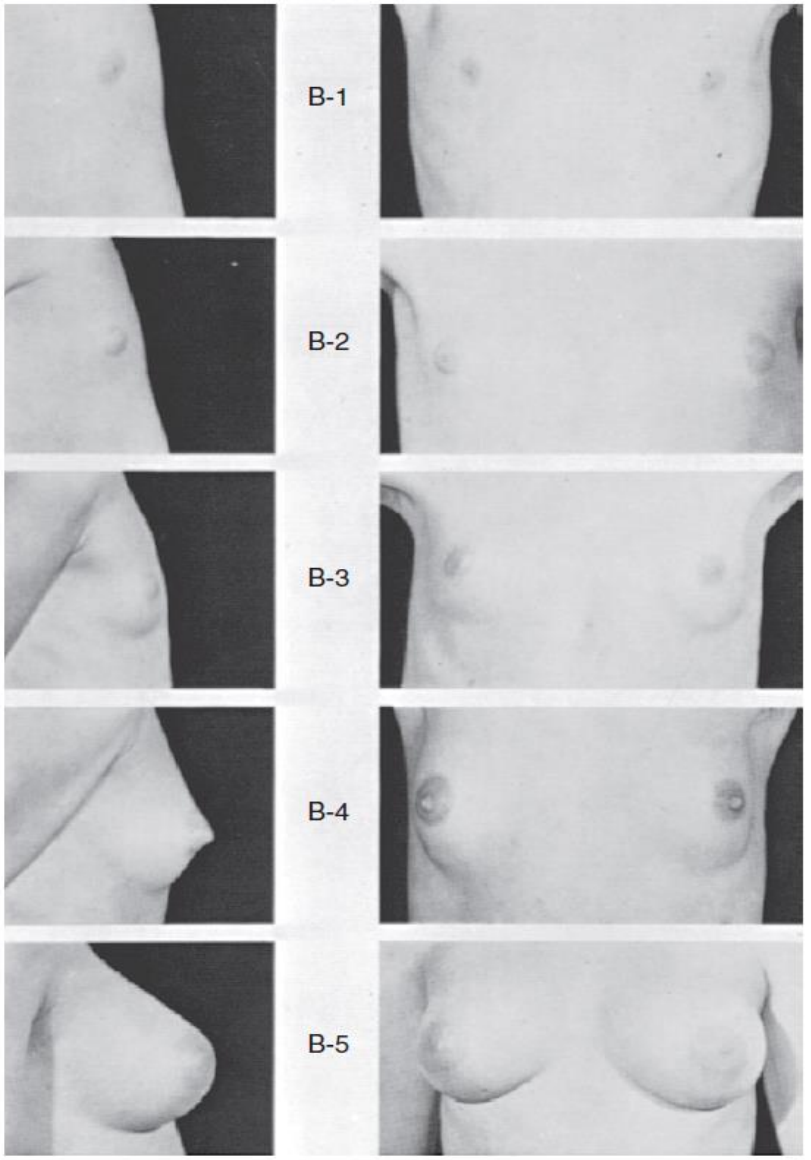
Prader orchidometer



Physiology of growth

Female





Puberty : Girl

Breast bud

William textbook endocrinology

Short stature

Definition

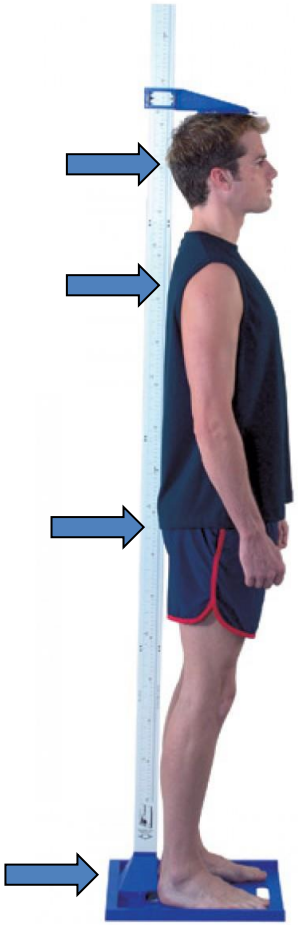
- ✓Height < 2 SD below the mean age
- ✓Height < 3rd percentile for chronological age

Important information

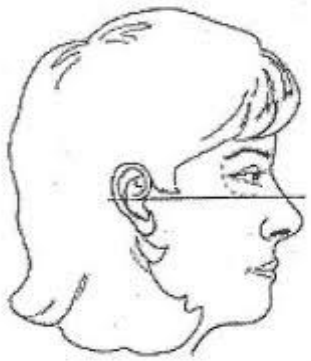
- ✓Accurate height measurement
- ✓Previous growth record
- ✓Height velocity
- ✓Mid-parental height
- ✓Upper:Lower segment ratio
- ✓Bone age

Accurate height measurement

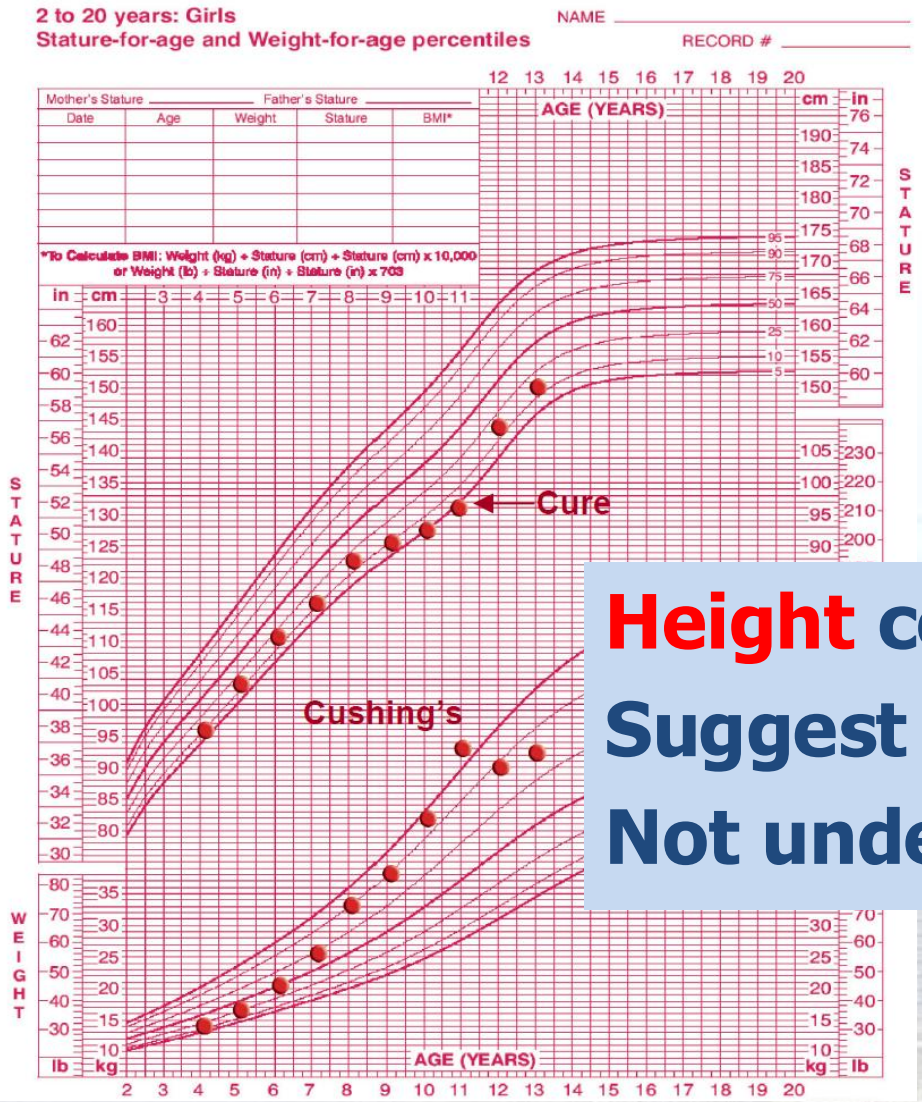
Harpenden stadiometer



- ✓ **Without foot wear**
- ✓ Heels, buttocks, scapula and occiput touching the wall
- ✓ Lower border of the eye socket in the same horizontal plane as external auditory meatus (**Frankfurt plane**)
- ✓ Looking straight ahead



Previous growth record



- ✓Cushing's syndrome
- ✓Growth hormone deficiency
- ✓Hypothyroidism
- ✓Albright's hereditary osteodystrophy

Height centile < Weight centile

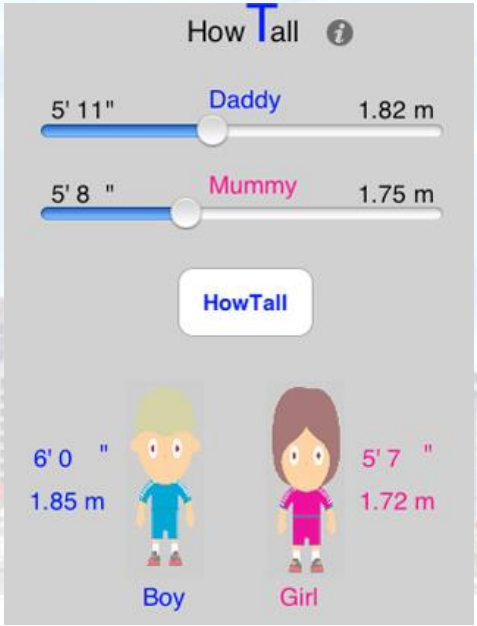
Suggest endocrine causes

Not undernutrition ★★★

Mid-parental height

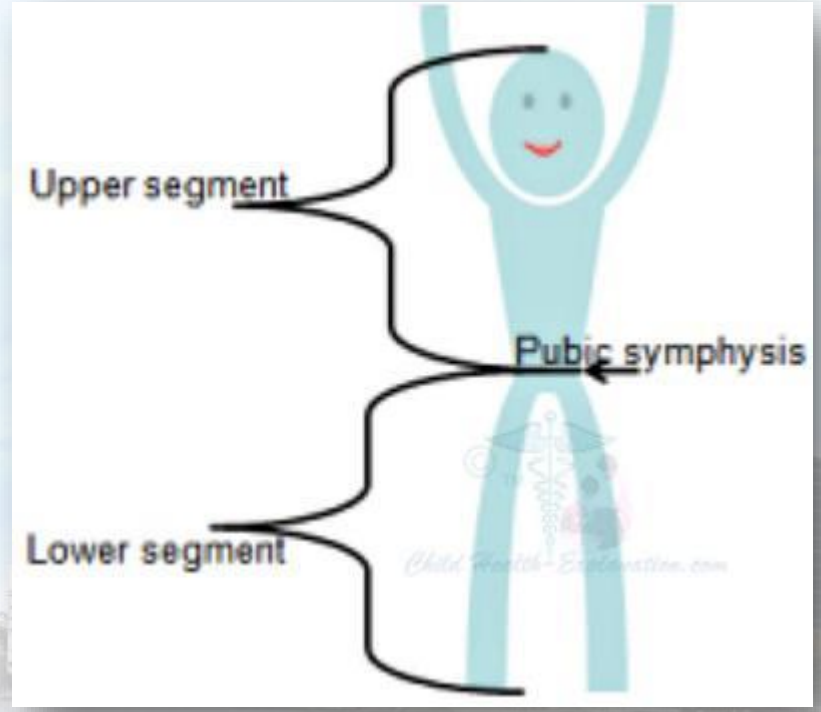
Girl = $\frac{\text{Father's height + Mother's height} - 13}{2}$ ± 5-8 cm

Boy = $\frac{\text{Father's height + Mother's height} + 13}{2}$ ± 5-8 cm



U:L segment ratio

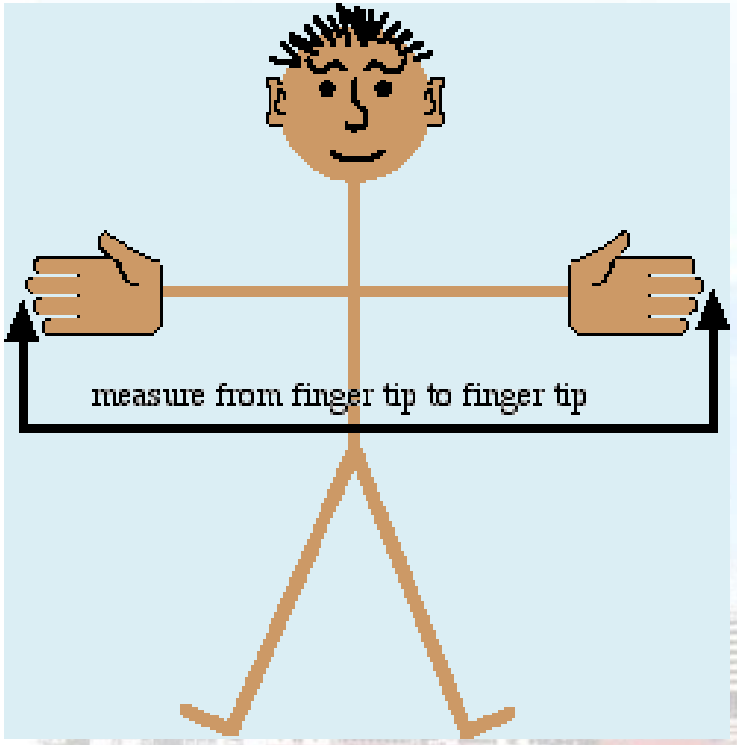
- **Lower segment** = วัดจาก upper border ของ pubic symphysis ถึงพื้น
- **Upper segment** = Height – Lower segment
- First step : Height measurement



Age	U/L segment
Birth	1.7
6 mo	1.6
1 yr	1.5
2 yr	1.4
3 yr	1.3
4 yr	1.25
5 yr	1.2
10 yr	**1.0**

Arm span

- วัดจากปลายนิ้วกลางข้างหนึ่งไปสู่ปลายนิ้วกลางอีกข้างหนึ่ง เมื่อเหยียดแขนออกไปทั้งสองข้างขนานกับพื้น



Boy age 10-11yr **arm span < height**
Man **arm span – height= 5.3cm**

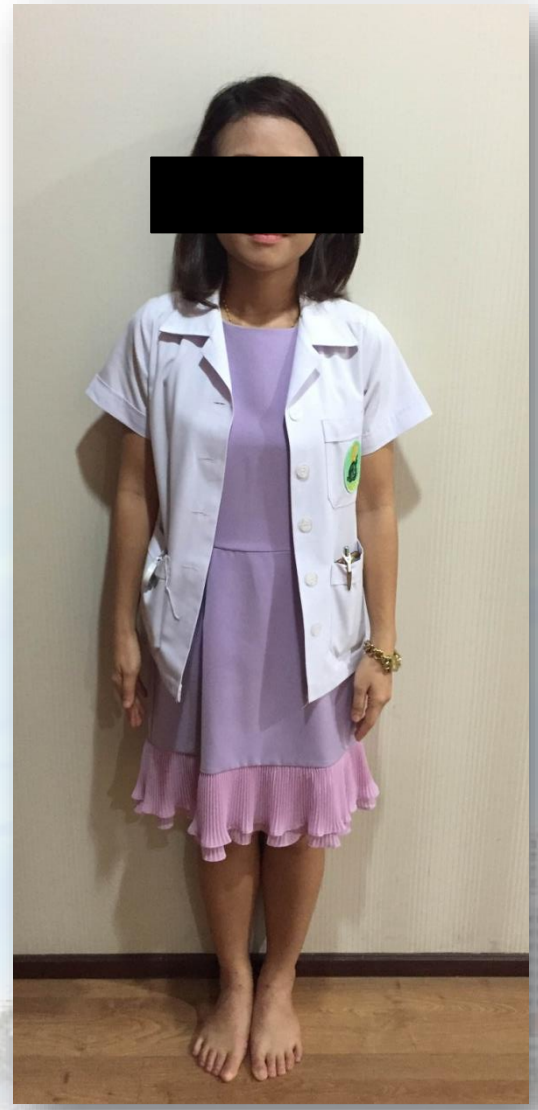
Girl age 11-14yr **arm span < height**
Woman **arm span – height = 1.2cm**

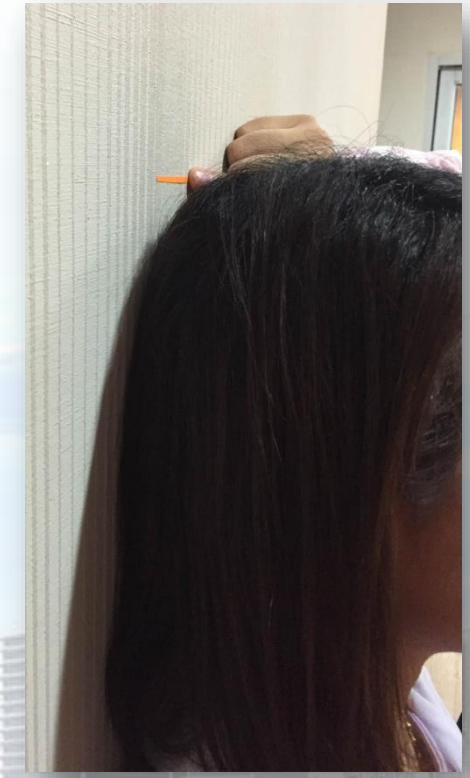
Arm span > Height 5 cm
“Eunuchoid appearance”

When to concern : short stature

- Severe short stature
- Poor height velocity
- Dysmorphic features
- Signs and symptoms of systemic illness
- Height centile < MPH centile

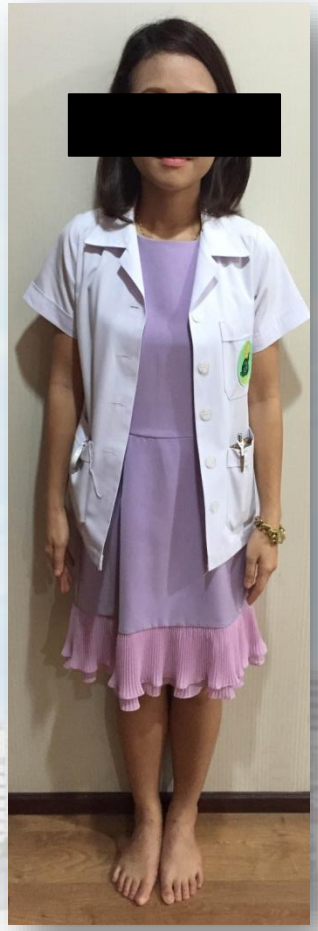
Height measurement





Upper : Lower segment ratio

Height – Lower segment = Upper segment

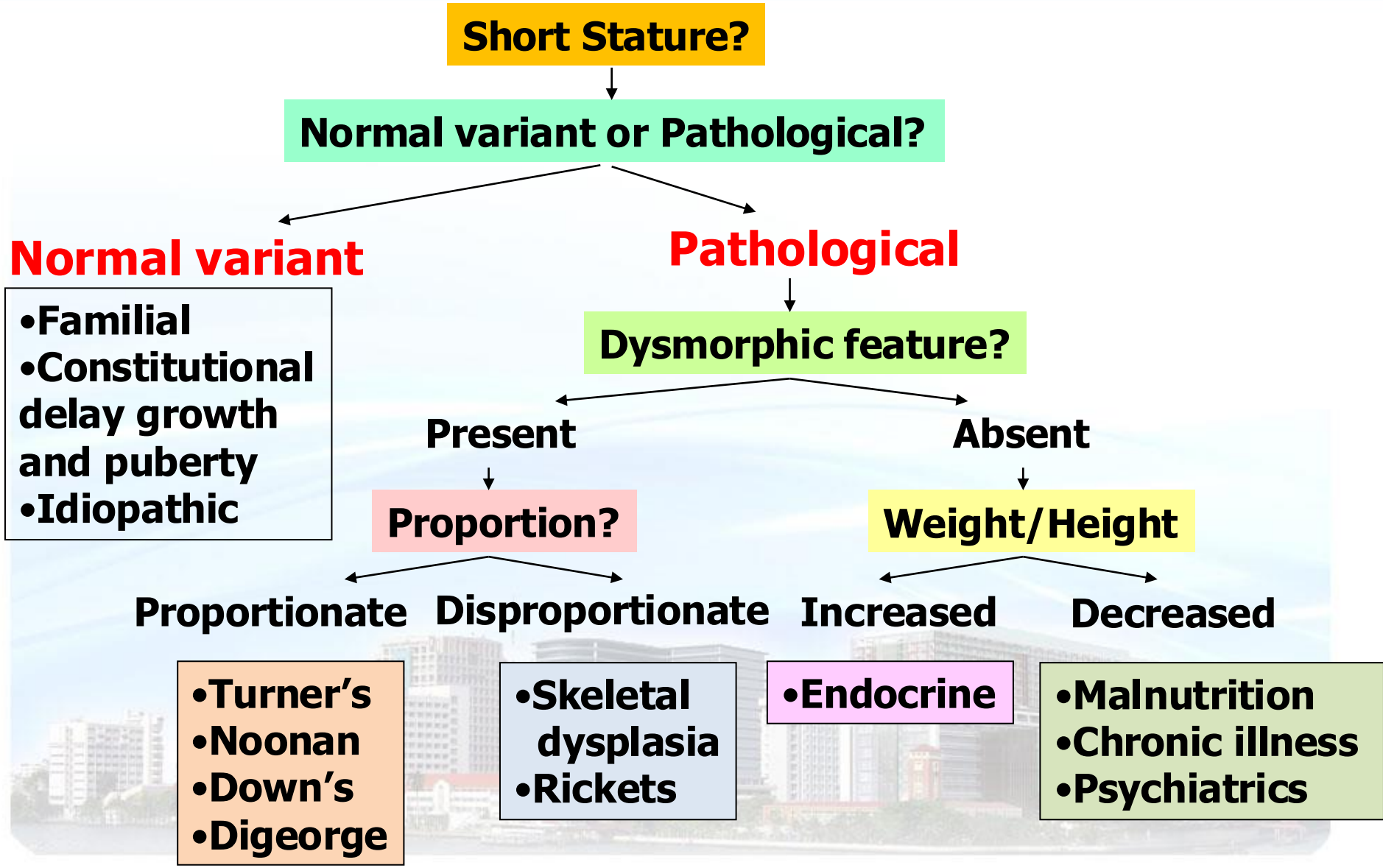


150-75 = 75 cm
Upper : Lower = 75:75 = 1:1

Arm span

Arm span > Height 2 cm (<5 cm)

Practical approach: Short stature



Practical approach: Short stature

Short Stature?

↓

Normal variant or Pathological?

Normal variant

- **Familial**
- **Constitutional delay growth and puberty**
- **Idiopathic**

Pathological

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When to concern : short stature

- Severe short stature
- Poor height velocity
- Dysmorphic features
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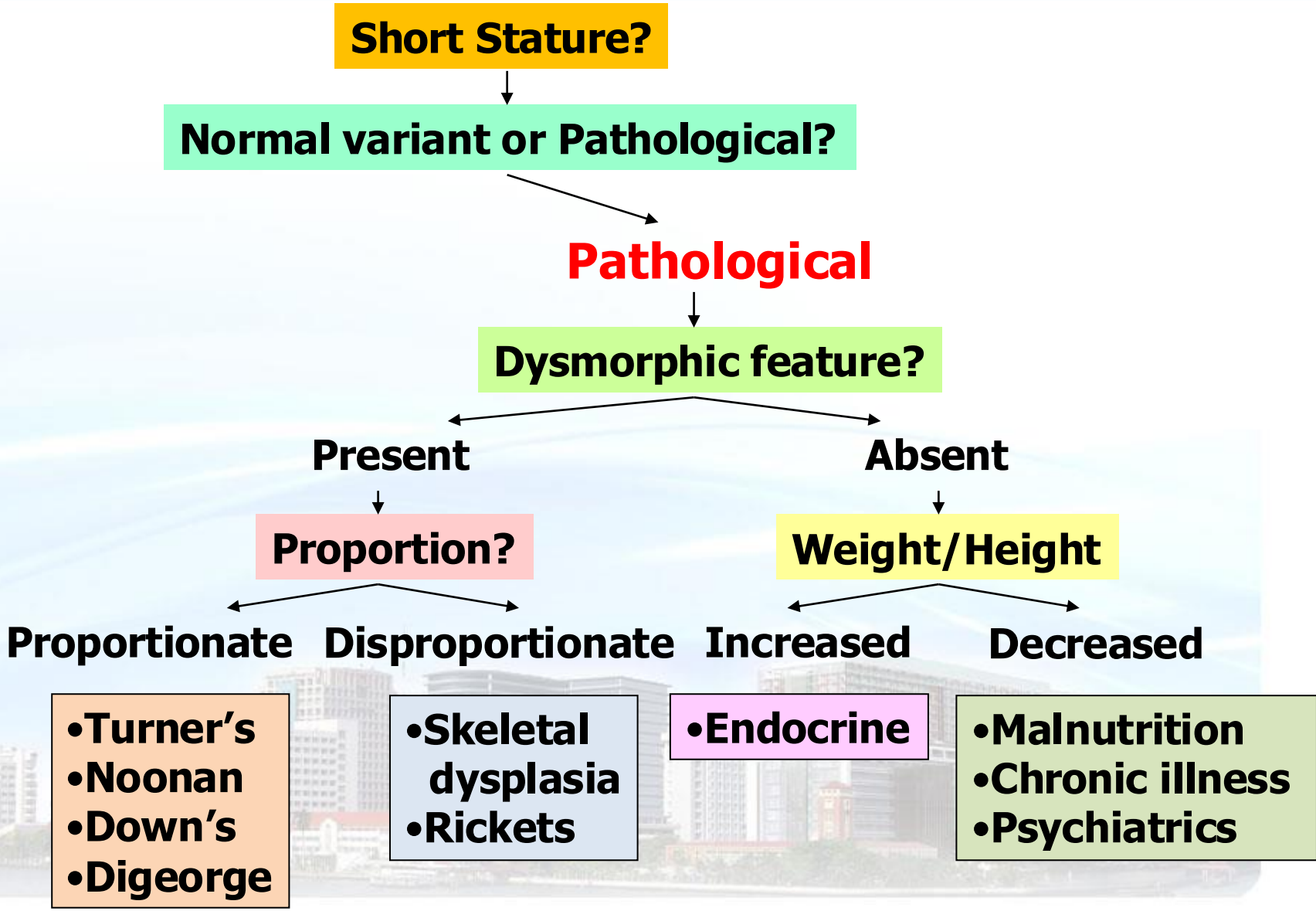
Practical approach: Short stature



Disproportionate

Short Limbs
-Skeletal dysplasia
- Rickets

U>L

L>U

Short Trunk
- Scoliosis

Rickets

Achondroplasia

Proportionate

Prenatal Onset
- IUGR
- Syndromes
- Chromosome dis

Postnatal Onset
- Malnutrition
- Chronic disease
- Drugs
- Psychological
- Endocrine disease

Proportionate

Prenatal Onset
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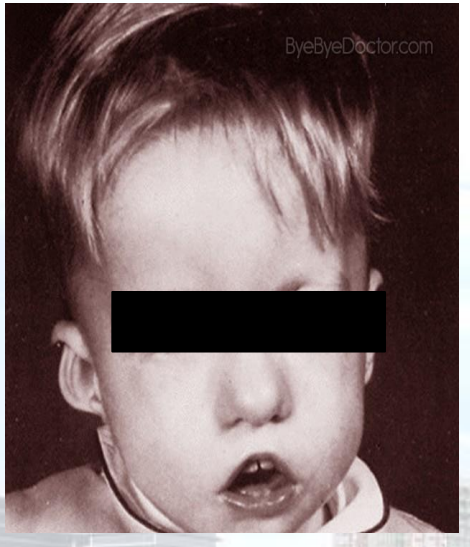
Turner syndrome

Noonan syndrome



Digeorge syndrome

Turner's syndrome

- ✓ Important cause of **short stature** in girls and **primary amenorrhea** in young women
- ✓ Loss of part or all of an X-chromosome

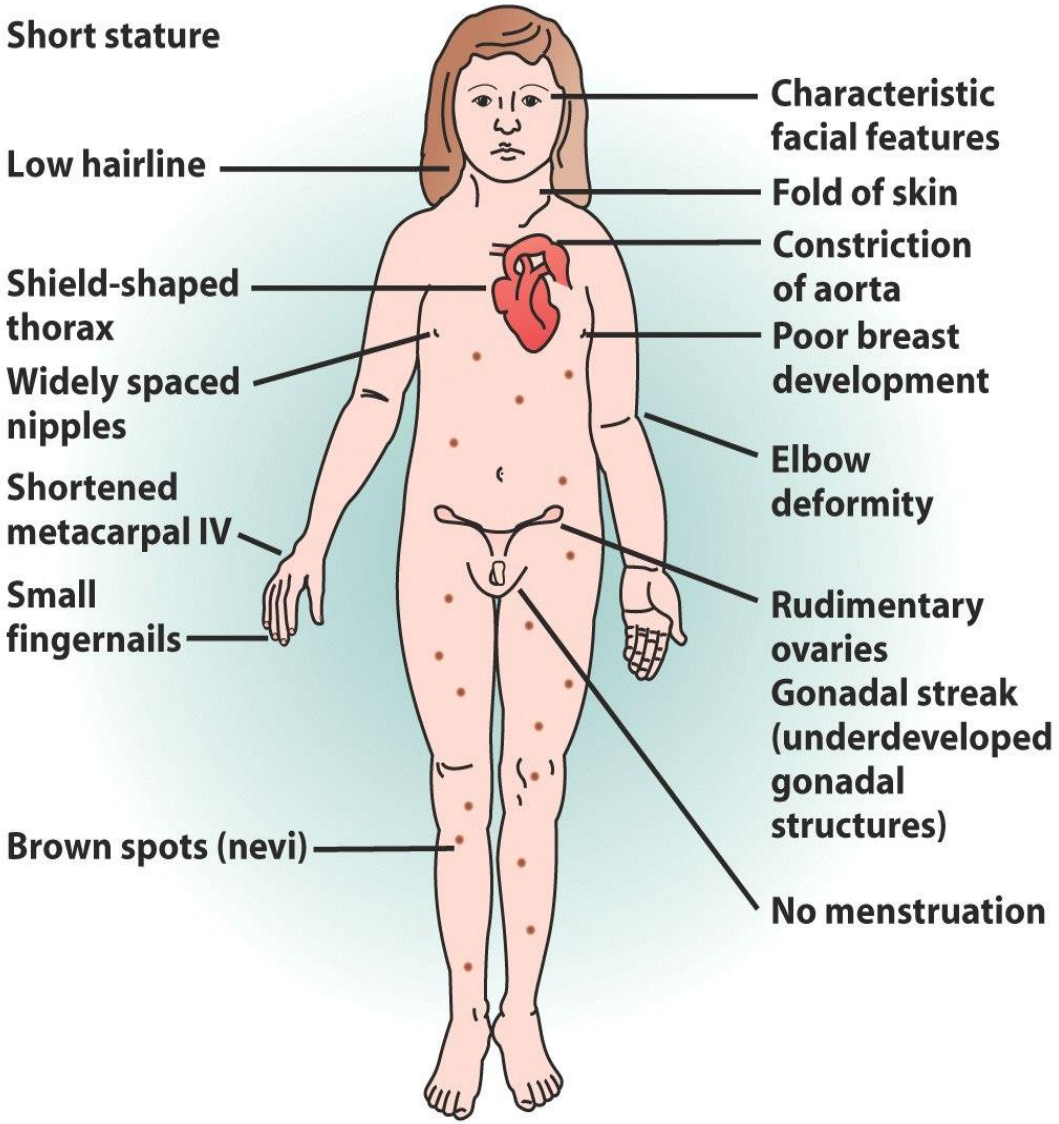
Karyotype: 45 X

Turner's syndrome: Karyotype

- 1:2,500-5,000 female live births
- Karyotype: the only method of confirming TS diagnosis

Karyotype	No.	%	Phenotype
45,X	95	48	Most severe phenotype. Highest incidence of structural cardiac and renal abnormalities.
46,Xi(Xq)	36	18	Structural abnormalities uncommon. Increased risk of autoimmunity, particularly thyroiditis and IBD, and deafness.
45,X/46,XX	21	11	Least severe phenotype. Increased mean height. Spontaneous puberty and menses in up to 40%.
46,Xr(X)	19	10	Spontaneous menses in 33%. Congenital abnormalities uncommon. Cognitive dysfunction in those with a small ring chromosome.
45,X/46,XY	11	6	Increased risk of gonadoblastoma.
45,X/46,X,idic(Y)	2	1	Increased risk of gonadoblastoma.
46,XXp-	3	1.5	Similar phenotype to 45,X monosomy.
46,XXq-	6	3	Variable phenotype.
other	3	1.5	

Turner's syndrome



Clinical features of Turner's syndrome

Feature	Frequency (%)
Short stature	98
Gonadal failure	95
Micrognathia	60
Cubitus valgus	47
Low posterior hairline	42
Short neck	40
High arched palate	38
Short fourth metacarpal	37
Multiple naevi	25
Webbed neck	25
Lymphedema of hands and feet	22
Nail dysplasia	13
Scoliosis	11
Madelung deformity	7

Primary: elevated FSH,LH and low E2

Turner's syndrome

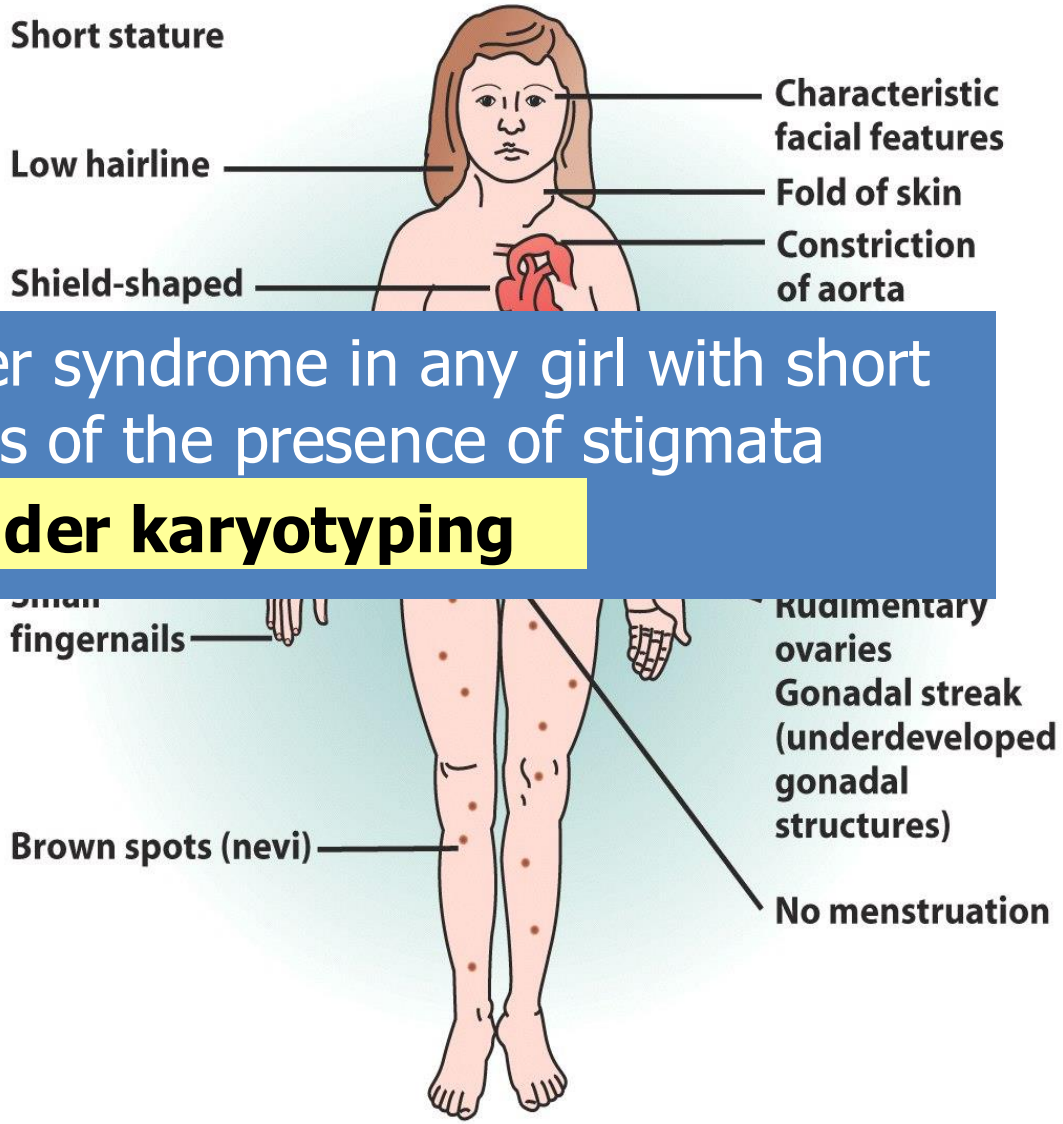
Cardiac abnormalities

	No. (%)[a]
Total no. of patients assessed	1,126
Structural abnormalities	
Bicuspid aortic valve	132 (12)
Coarctation of aorta	103 (9)
Aortic stenosis/regurgitation	38 (3.4)
Partial anomalous venous drainage of the pulmonary veins	26 (2.3)
Other	17 (1.5)
Total	316 (28)

Other abnormalities

- ✓ Hypothyroidism
- ✓ Osteoporosis
- ✓ Insulin resistance & T2DM
- ✓ Kidney abnormalities
- ✓ Hearing loss

Turner's syndrome



Always think of turner syndrome in any girl with short stature regardless of the presence of stigmata

Consider karyotyping

Indications for genetic testing to diagnose TS

As the only clinical feature:

- Fetal cystic hygroma, or hydrops, especially when severe
- Unexplained short stature
- Left-sided outflow congenital heart defects (excluding BAV)[a]
- Unexplained delayed puberty/menarche, failure to progress puberty or secondary amenorrhea
- Infertility
- Characteristic physical features[b]

As least two of the following:

- Renal anomaly (horseshoe, absence, or hypoplasia)
- Madelung deformity
- Neuropsychologic problems, and/or psychiatric issues
- Multiple typical or melanocytic nevi
- Dysplastic or hyperconvex nails
- Other congenital heart defects (including BAC)[c]
- Hearing impairment <40 years of age together with short stature

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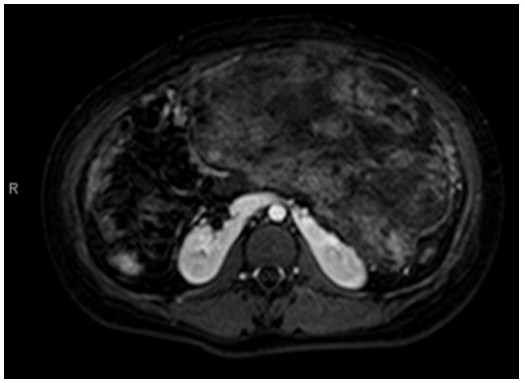
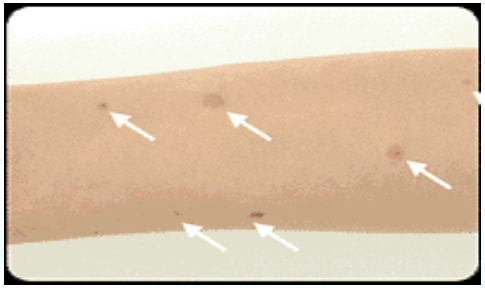
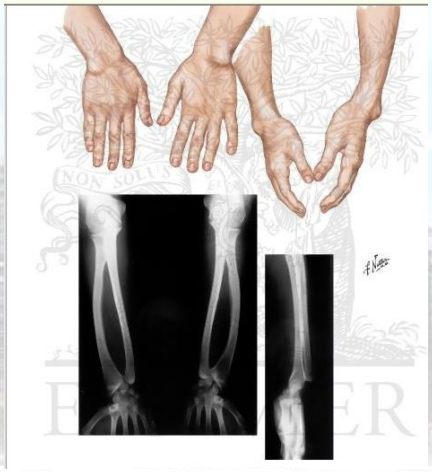
[a]Coarctation; aortic stenosis; mitral valve anomalies; hypoplastic left heart syndrome.

[b]Down-slanted palpebral fissures; epicanthal folds; low-set anomalous pinnae; micrognathia; narrow palate; short broad neck; webbing of the neck.

[c]Partial anomalous pulmonary venous return/connection; atrial septal defect, secundum type; ventricular septal defects, muscular or membranous; BAV (bicuspid aortic valves).





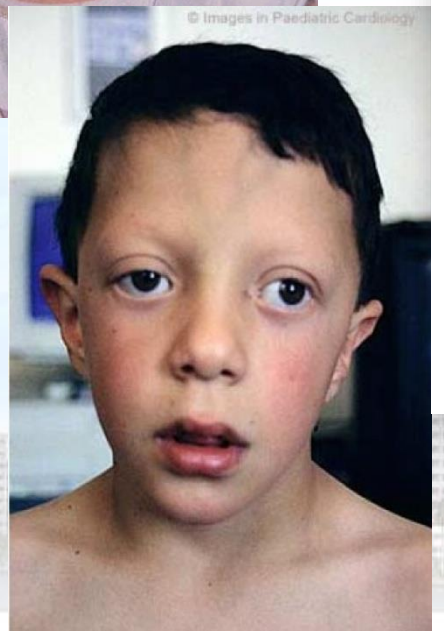


Gravholt CH. Eur J Endocrinol. 2024 Jun 5;190(6):G53-G151.

Noonan syndrome

- **Autosomal dominant** that affects both males and females
- One of the most common genetic syndromes associated with **congenital heart disease**

Noonan syndrome: features



Short stature

- Up to 83% of patients have short stature[1]

Characteristic facial features[3]

- Broad, high forehead
- Hypertelorism
- Low-set, posteriorly rotated ears with a thick helix
- High-arched palate
- Micrognathia
- High-arched eyebrows[1]
- Short neck with excess nuchal skin
- Epicanthal folds
- Downward-slanting palpebral fissures
- Low posterior hairline

Congenital heart defects[3]

- Pulmonary valve stenosis
- Hypertrophic obstructive cardiomyopathy
- Atrial and ventricular septal defects
- Persistent ductus arteriosus

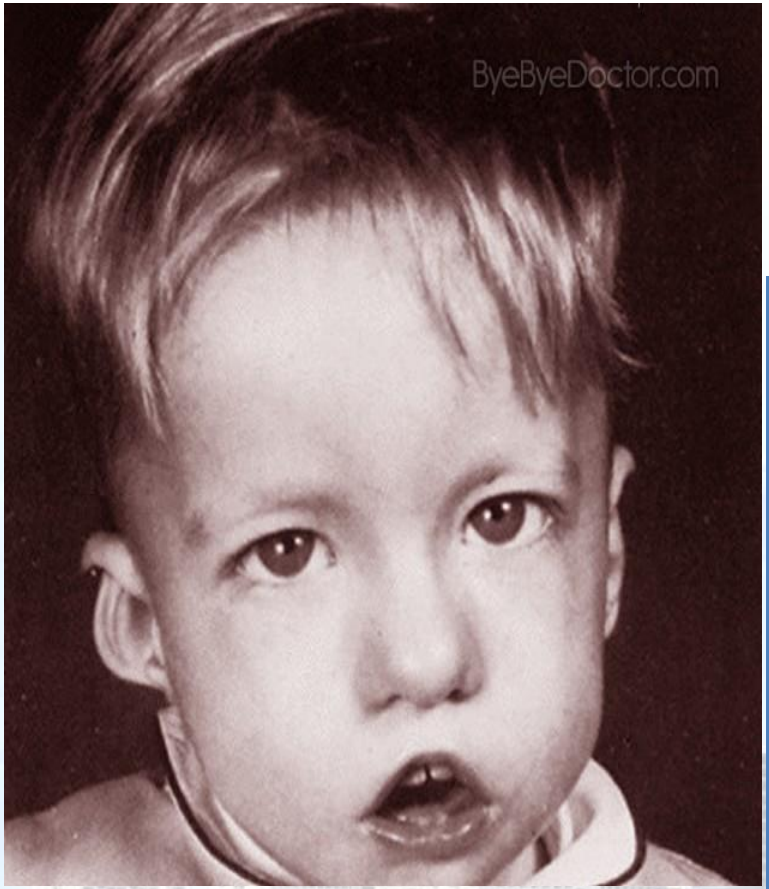
Other clinical manifestations[3-5]

- Pectus carinatum, pectus excavatum
- Scoliosis
- Cryptorchidism
- Lymphatic abnormalities
- Coagulopathy
- Cognitive/learning disabilities
- Ophthalmological issues
- Arnold-Chiari malformation
- Seizures

Noonan syndrome

Characteristic	Turner's syndrome	Noonan's syndrome
Incidence	1:1500-2500 female live birth	1:1000-2500 live birth
2nd sex characteristic	No/Incomplete	Normal
Webbed neck	Posterior	Anterior
IQ	Normal	Mild MR
Family history	Positive	AD
CVS	Coarction of aorta Bicuspid AV	PS, HOCM, PDA, ASD, VSD
Chromosomal study	45,x/mosaic	46,XY 46,XX

Digeorge syndrome



- 22q11.2 deletion syndrome
- The features of this syndrome vary widely

Facial abnormalities:

- **Hypertelorism**
- **Micrognathia**
- **short philtrum with fish-mouth appearance**
- **antimongoloid slant**
- **telecanthus**

Otolaryngic

- **low-set ears, with defective pinna**
- **cleft palate**

Case presentation

Total calcium 6.8 mg/dL
Phosphorus 4.9 mg/dL
Serum Cr 0.85 mg/dL

A 24-year-old man
Presented with
refractory seizure

Approach to hypocalcemia

Hypocalcemia

Serum phosphorus

High serum phosphorus

- Phosphorus loading
- Hypomagnesemia
- Hypoparathyroidism
  - postsurgical(neck)

- Chronic renal failure
- Hypoparathyroidism
  - hereditary
  - infiltrative disease
- Pseudohypoparathyroid

Normal serum phosphorus

- Critical illness :
  gram negative sepsis
- Acute pancreatitis
- Massive Blood transfusion

- Malabsorption
- Osteoblastic metastasis

Low serum phosphorus

- Hungry bone syndrome after parathyroidectomy

- Vitamin D deficiency
- Malabsorption
- Medication : Phenytoin

Acute

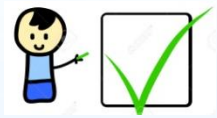
Chronic

Case presentation

Hypocalcemia

Total calcium 6.8 mg/dL
Phosphorus 4.9 mg/dL
Serum Cr 0.85 mg/dL

Serum phosphorus



High serum phosphorus

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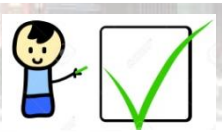


Acute

Chronic

Back to this patient

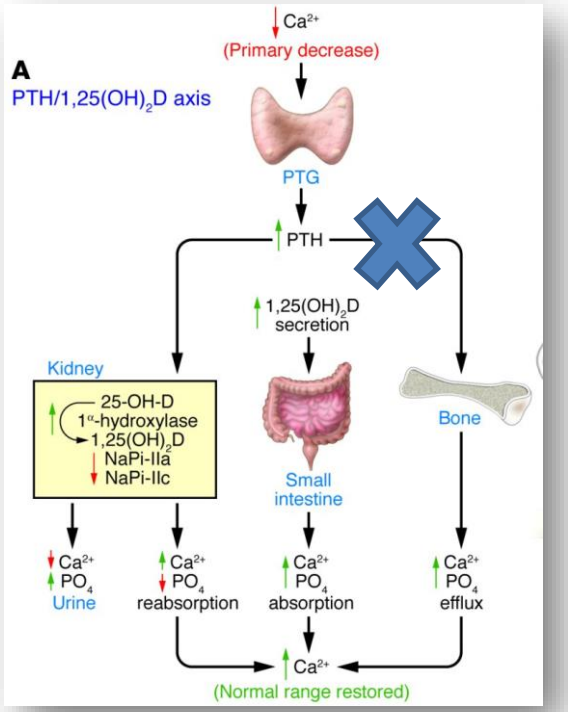
Chronic hypocalcemia with hyperphosphatemia

- Chronic renal failure
- Hypoparathyroidism
  - hereditary
  - infiltrative disease
- Pseudohypoparathyroidism

Serum creatinine 0.85

Hypoparathyroidism vs PseudohypoPTH

Hypoparathyroidism



Resistance to PTH action

AHO features

Short stature, obesity

Rounded face,

Short 4th MCP

	Serum calcium	Serum phosphorus	PTH level
Hypoparathyroidism	↓	↑	↓
PseudohypoPTH	↓	↑	↑

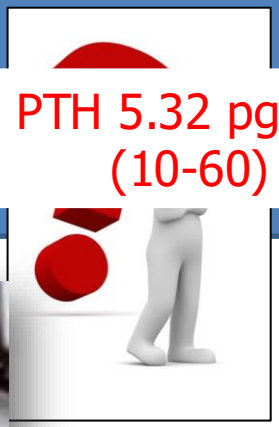
Back to this patient

Chronic hypocalcemia with hyperphosphatemia

- Chronic renal failure
- Hypoparathyroidism
  - hereditary
  - infiltrative disease
- Pseudohypoparathyroidism

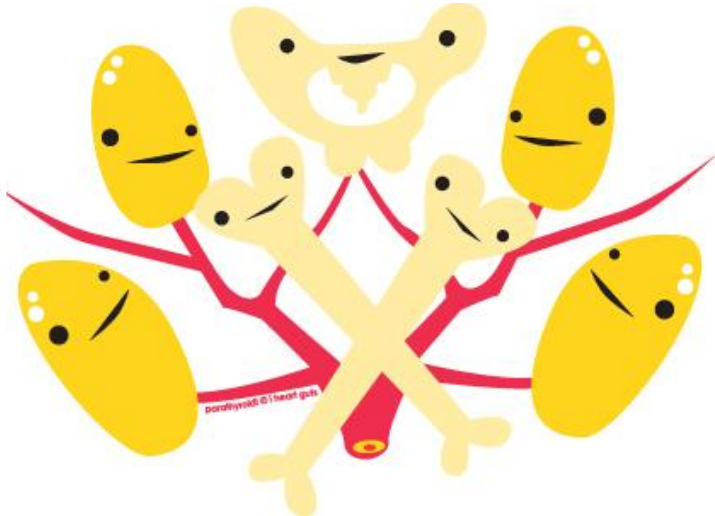
Serum creatinine 0.85

PTH 5.32 pg/mL
(10-60)



Hypoparathyroidism : Causes

1. Genetic syndromes: Digeorge syndrome
   PTH gene mutation
2. Autoimmune disorders: isolated, APS1
3. Infiltrative disease: Hemochromatosis,
   Wilson disease, metastatic cancer
4. Post surgery, irridiation
5. Hypomagnesemia

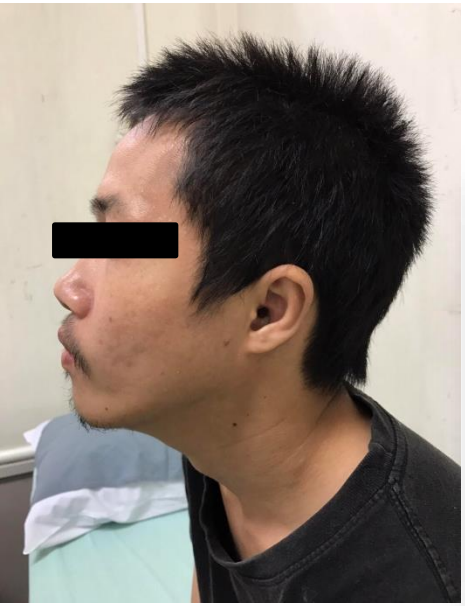


A thorough history taking and physical examination

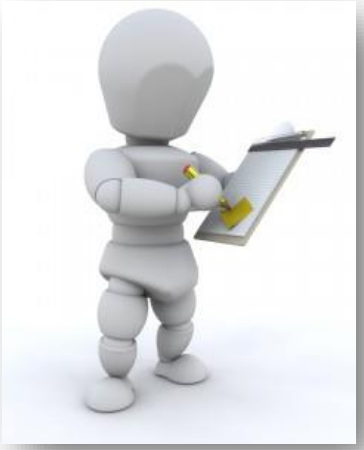
Developmental delay and mild mental retardation

History of Chronic otitis media with left mastoiditis S/P mastoidectomy: **CMI defect**

Abnormal facies: long face, bulbous nose, low set ear, small earlobes



Chronic hypoparathyroidism: onset since birth



Chromosome 22q11.2 deletion syn
Velocardiofacial syndrome
DiGeorge syndrome

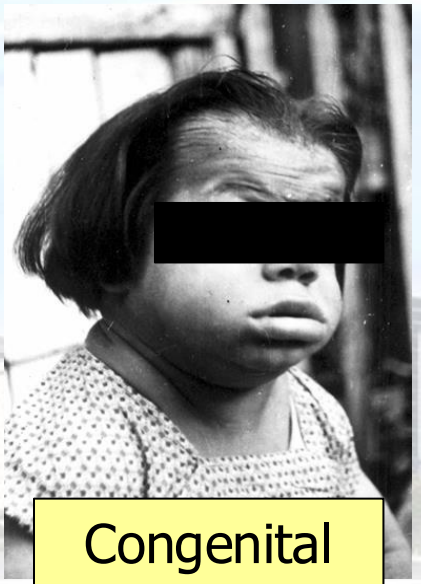
Proportionate

Prenatal Onset
- **IUGR**
- **Syndromes**
- **Chromosome dis**

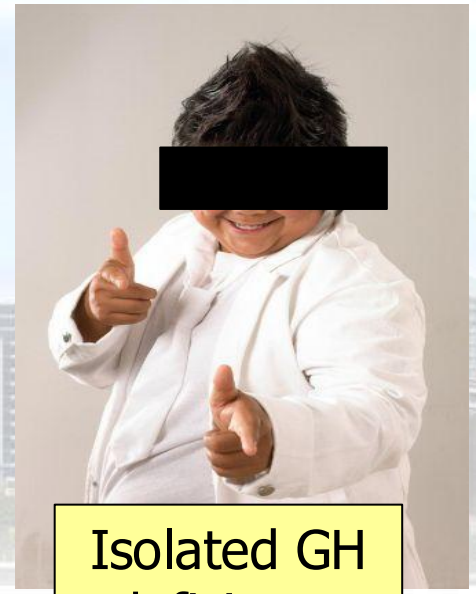
Postnatal Onset
- **Malnutrition**
- **Chronic disease**
- **Drugs**
- **Psychological**
- **Endocrine disease**

Endocrine diseases: เตี้ย+อ้วน

Cushing syndrome



Congenital hypothyroid



Isolated GH deficiency

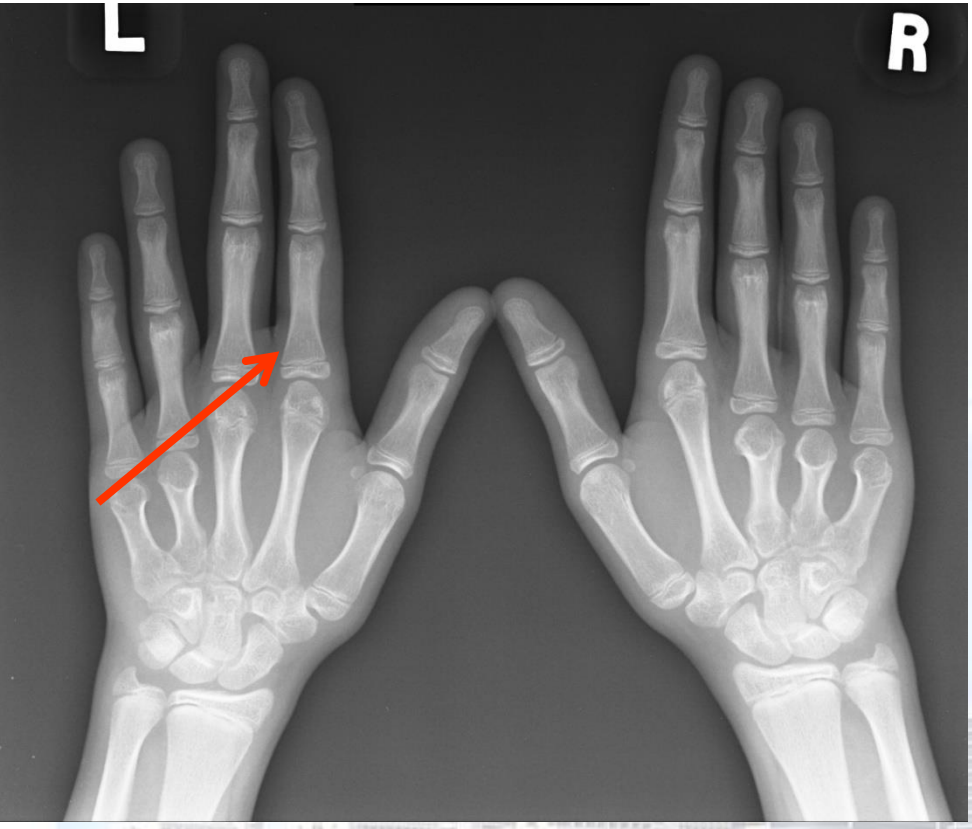
Albright's hereditary osteodystrophy

Albright's Hereditary Osteodystrophy

- A lack or responsiveness to PTH (**PTH resistance**)
  Low cal, High Po_4, High PTH
- Short stature, **Short 4th, 5th MCP**, round face, mild mental retardation
- PseudohypoPTH type 1A and pseudopseudohypoPTH

Condition		Appearance	PTH levels	Calcitriol	Calcium	Phosphates	Imprinting
Hypoparathyroidism		Normal	Low	Low	Low	High	Not applicable
Pseudohypoparathyroidism	Type 1A	Skeletal defects	High	Low	Low	High	Gene defect from mother (GNAS1)
	Type 1B	Normal	High	Low	Low	High	Gene defect from mother (GNAS1 and STX16)
	Type 2	Normal	High	Low	Low	High	?
Pseudopseudohypoparathyroidism		Skeletal defects	Normal	Normal	Normal[9]	Normal	gene defect from father

Albright's Hereditary Osteodystrophy



Short 4th, 5th MCP

Knuckle-dimple sign

Proportionate: Endocrine disease

Endocrine disease: เตี้ย+อ้วน

Cushing syndrome

- ✓Signs of cushing syndrome

Congenital hypothyroid

- ✓84% thyroid agenesis
- ✓ Mental retardation
- ✓ Cretinous features

Isolated GH deficiency

- ✓Decreased growth rate after the age of 6 months
- ✓ Small voice, frontal bossing
- ✓ younger-appearance than chronological age

Proportionate

Prenatal Onset
- **IUGR**
- **Syndromes**
- **Chromosome dis**

เตี้ย+ผอม

Weight for Height ↓

Weight for Height ↑

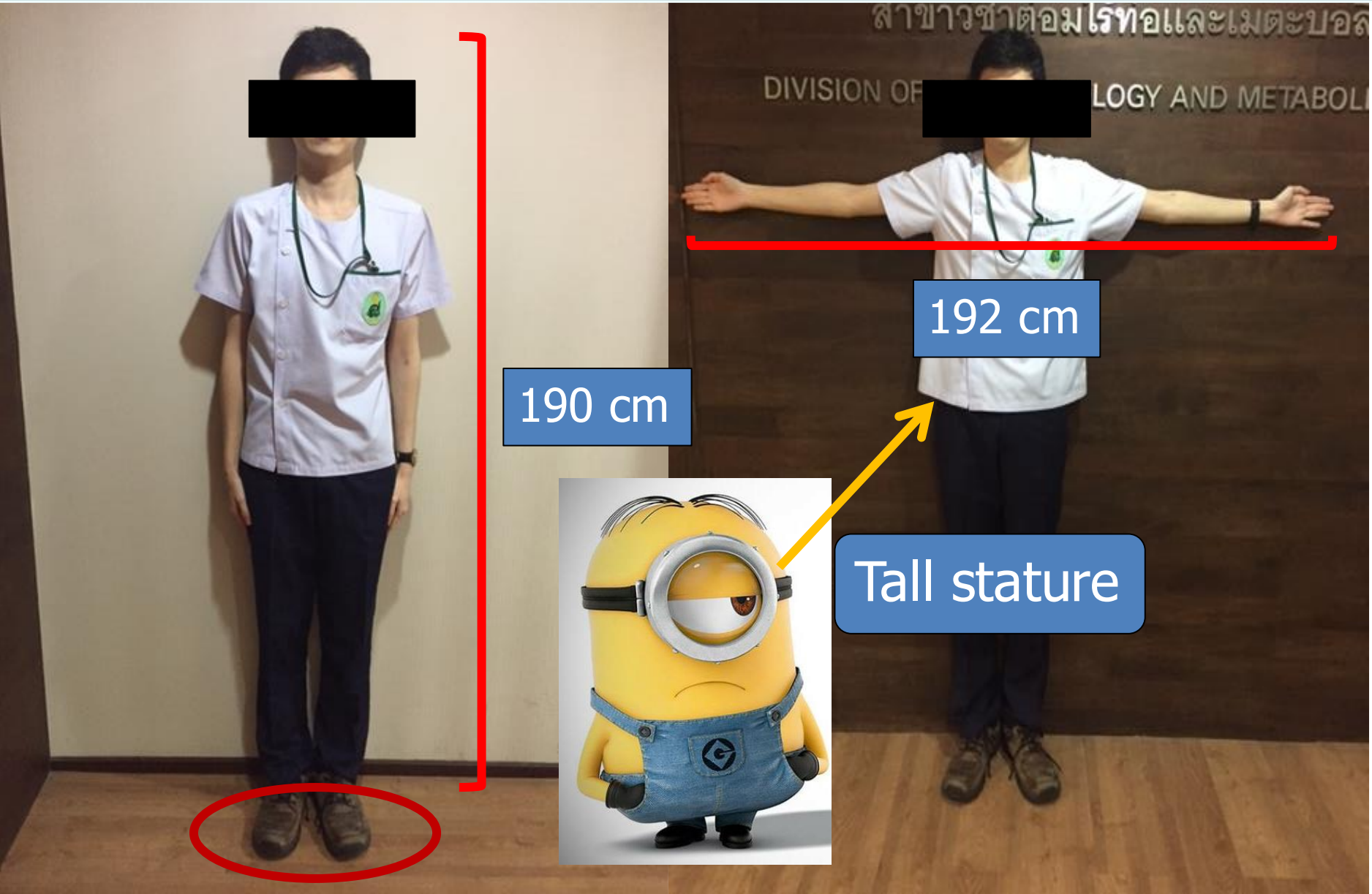
เตี้ย+อ้วน

Postnatal Onset
- **Malnutrition**
- **Chronic disease**
- **Drugs**
- **Psychological**
- **Endocrine disease**

Undernutrition
Systemic illness
-RS: Asthma
-CVS: Congenital heart disease
-GI: Malabsorption
-Renal: **RTA**, renal failure
-Neuro: Brain tumors
-Hemato: **Thalassemia**
-Autoimmune diseases

Endocrine involvement in thalassemia

Endocrine complications	Recommendations
Short stature	Monitor height and weight
Delayed puberty	Monitor pubertal progression
Low bone mass	BMD
Vitamin D deficiency	25-OHD every 6 months
Glucose intolerance and DM	FPG or OGTT annually
Hypothyroidism	TFT annually
Adrenal insufficiency	Serum cortisol
Hypoparathyroidism	Calcium, phosphate, and PTH annually


DIVISION OF
LOGY AND METABOL
192 cm
190 cm
Tall stature

Tall stature

Definition

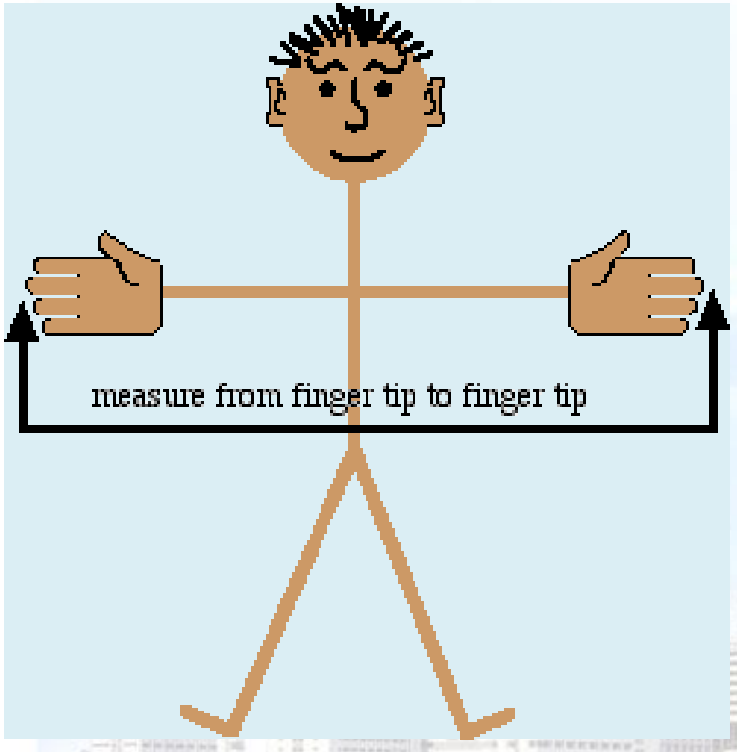
- ✓**Height > 2 SD above the mean age**
- ✓**Height > 2 SD above mid-parental height**

Important information

- ✓Accurate height measurement
- ✓Previous growth record
- ✓Height velocity
- ✓Mid-parental height
- ✓Upper:Lower segment ratio
- ✓Bone age
- ✓Secondary sex characteristics

Arm span

- วัดจากปลายนิ้วกลางข้างหนึ่งไปสู่ปลายนิ้วกลางอีกข้างหนึ่ง เมื่อเหยียดแขนออกไปทั้งสองข้างขนานกับพื้น

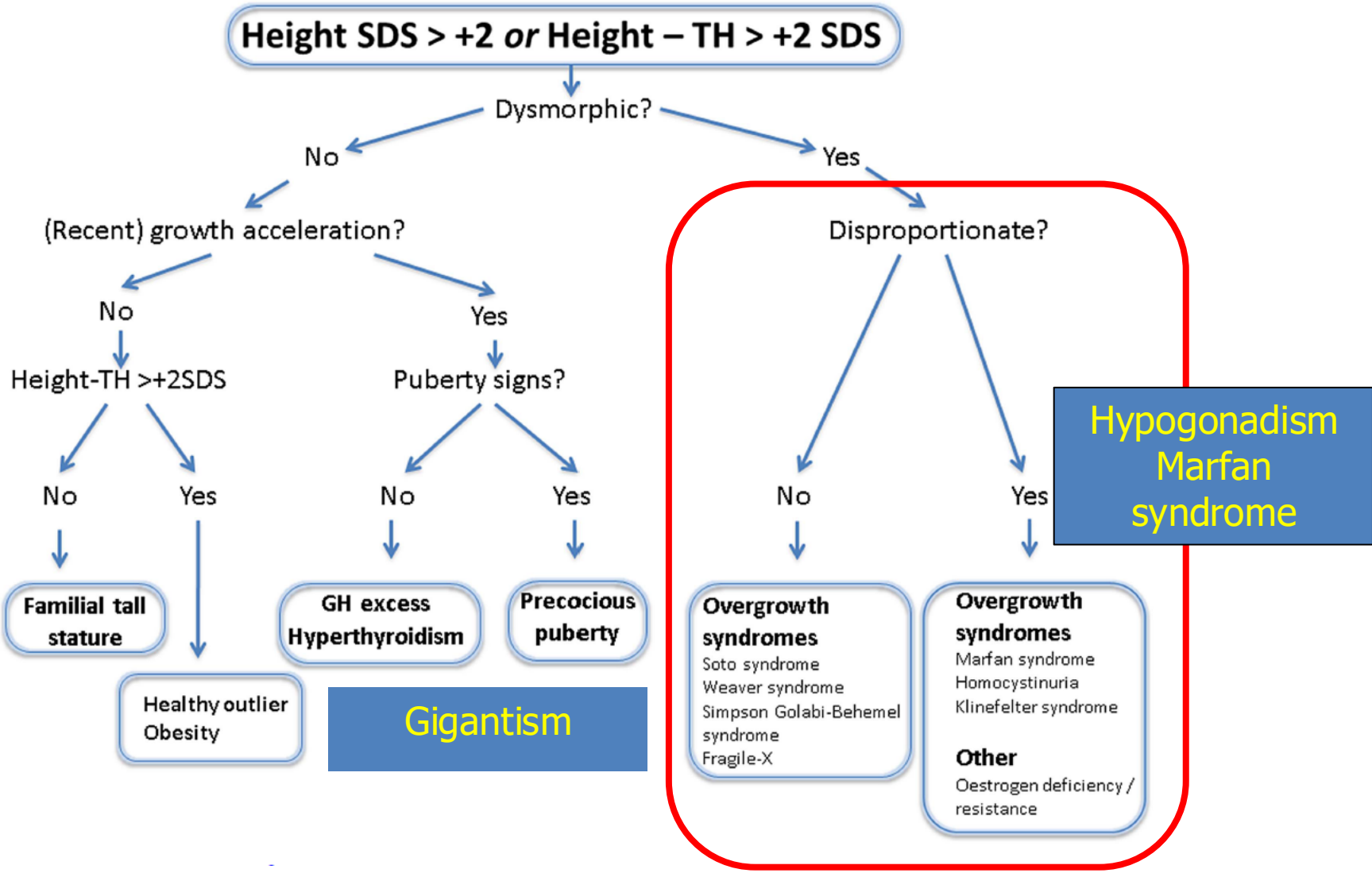


Boy age 10-11yr **arm span < height**
Man **arm span – height= 5.3cm**

Girl age 11-14yr **arm span < height**
Woman **arm span – height = 1.2cm**

Arm span > Height 5 cm --> suspected eunuchoid appearance

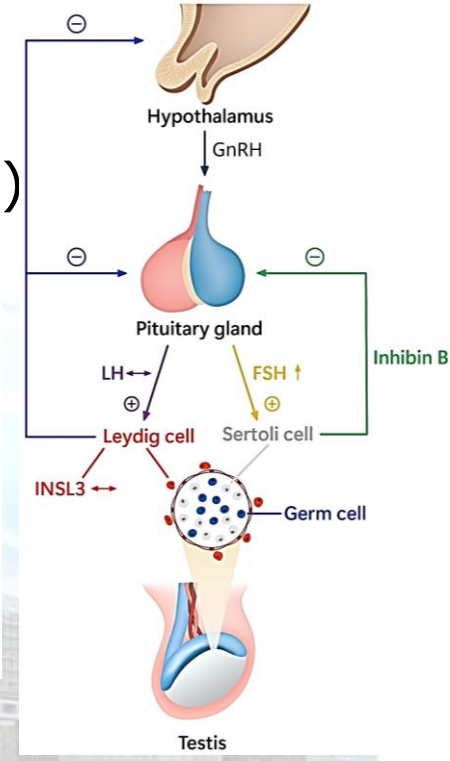
Tall stature



Male hypogonadism : Definition

- A decrease in either of the **two major functions** of the testes:
  - sperm production
  - testosterone production
- Can result from disease
  - Testis (Primary hypogonadism)
  - Pituitary or Hypothalamus (Secondary hypogonadism)

	Primary hypogonadism	Secondary hypogonadism
Testosterone And/or sperm count	Low	Subnormal/Low
FSH/LH	**High**	**Normal/Low**



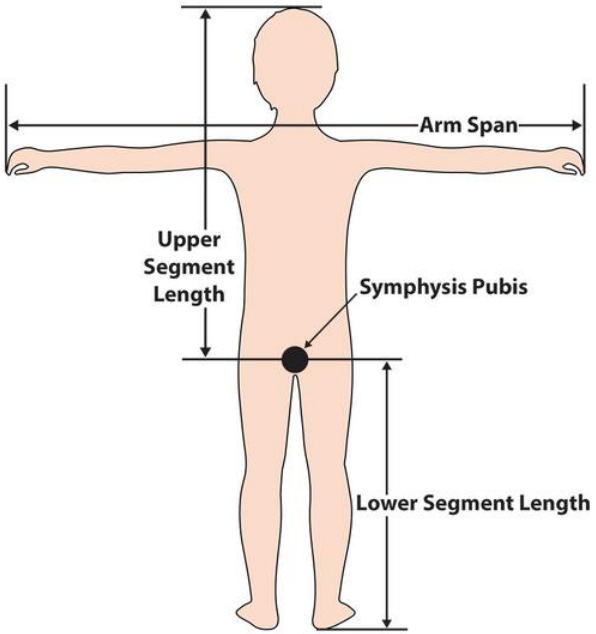
Symptoms

- **Primary or secondary hypogonadism**
  - **Primary (testis)**: มีอาการของ testosterone deficiency อย่างเดียว
  - **Secondary (Pituitary)** : มีอาการของ testosterone deficiency ร่วมกับขาด hormone อื่น ๆ ด้วย เช่น thyroid, cortisol, GH
- **Prepubertal onset or postpubertal onset**
  - **Prepubertal** : enuchoidism or enuchoid habitus
  - **Postpubertal** :
    - normal pubertal maturation, normal body proportion, normal voice and temporal hair recession
    - decreased hair distribution, ↓ **libido, impotence, infertility**, ↓ muscle strength, soft testis, gynaecomastia

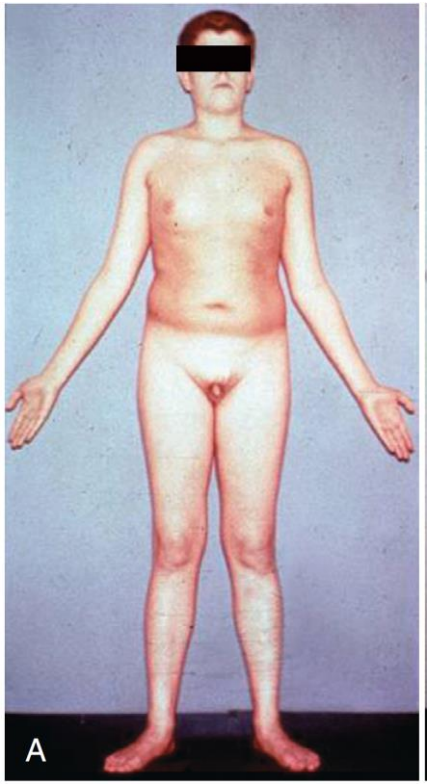
Prepubertal vs. Postpubertal onset

	Prepubertal onset	Postpubertal onset
Symptoms	No pubertal development Delayed puberty	**Sexual symptoms** - Decreased libido - Erectile dysfunction **Non-specific symptoms** - Decreased energy, weakness, decreased physical performance - Hot flushes, sweating - Poor memory and concentration - Minimal trauma fracture, height loss ↓ shaving frequency
Signs		
Eunuchoid body proportion	Present	Absent
Facial, axillary, pubic, body hair	No-sparse	Decreased
Gynecomastia	Present or absent	Present or absent
Penis	Small	Normal
Testis	Small, cryptorchidism	Shrinking testes (longstanding hypogonadism)
Voice	High-pitched	Normal voice pitch

Measurement of body proportion



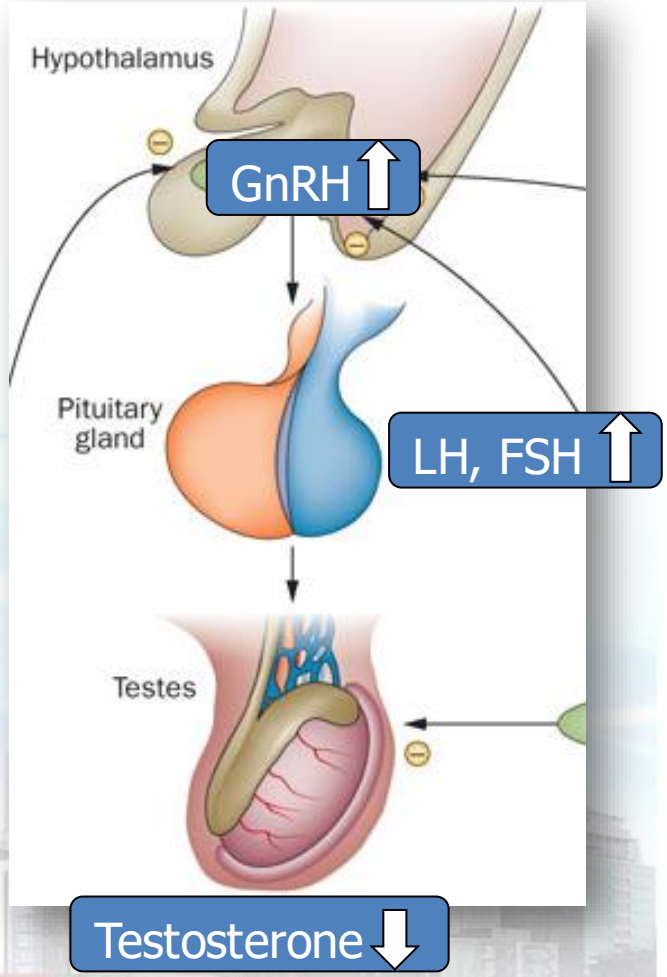
- **Normal**
  - Upper: lower segment = 0.9-1
  - Arm span exceeds height < 5 cm
- **Eunuchoid body proportion**
  - Upper: lower segment <0.9
  - Arm span > height ≥ 5 cm
  - Prepubertal onset of hypogonadism
  - Marfan syndrome



Fernandes JA. Cambridge University Press.
Williams Textbook of Endocrinology 14th Edition.

Primary hypogonadism

- **Prepubertal onset**
  - Klinefelter's syndrome
- **Postpubertal onset**
  - Mumps orchitis
  - Autoimmune orchitis
  - Trauma, radiation, surgery



Secondary hypogonadism

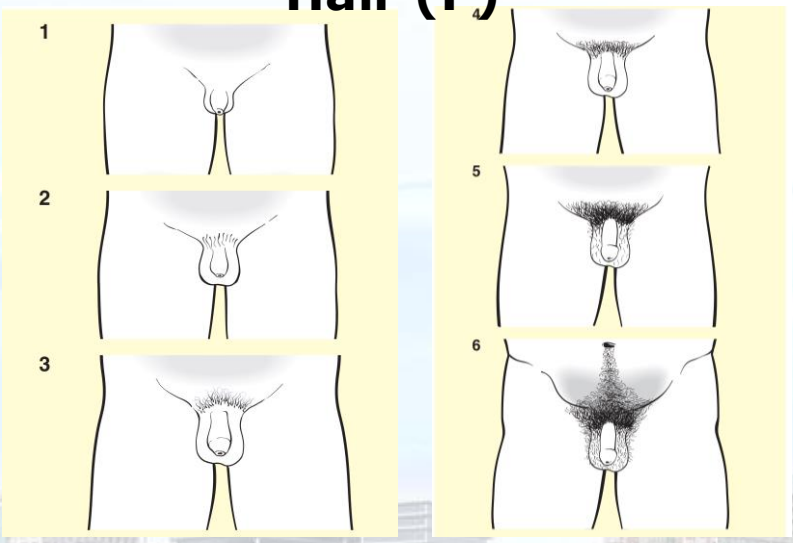
Prepubertal onset	Postpubertal onset
•**Kallmann's syndrome** •Idiopathic hypogonadotropic hypogonadism •Pituitary tumor (Craniopharyngioma) •Uremia •Severe systemic illness •Cranial radiation •Hyperprolactinemia	•Acquired idiopathic hypogonadotropic hypogonadism •**Pituitary macroadenoma** •Uremia •Severe systemic illness •Cranial radiation •Hyperprolactinemia •Hemochromatosis •Cushing's syndrome •Cirrhosis •Morbid obesity

Genital examination

Testicular volume Orchidometer

Puberty: >3 mL
Normal adult size: 15-25 mL

Tanner staging of genitalia (G) and pubic Hair (P)



Testicular consistency

- Normal: rubbery – slightly firm
- Soft: 2° hypogonadism
- Firm: Klinefelter syndrome
- Hard: malignancy

Figure from Nieschlag. Andrology 2010 based on Marshall and Tanner 1970.

Laboratory tests in male hypogonadism

Testosterone

Morning 7.00-10.00 am, fasting

Normal serum T

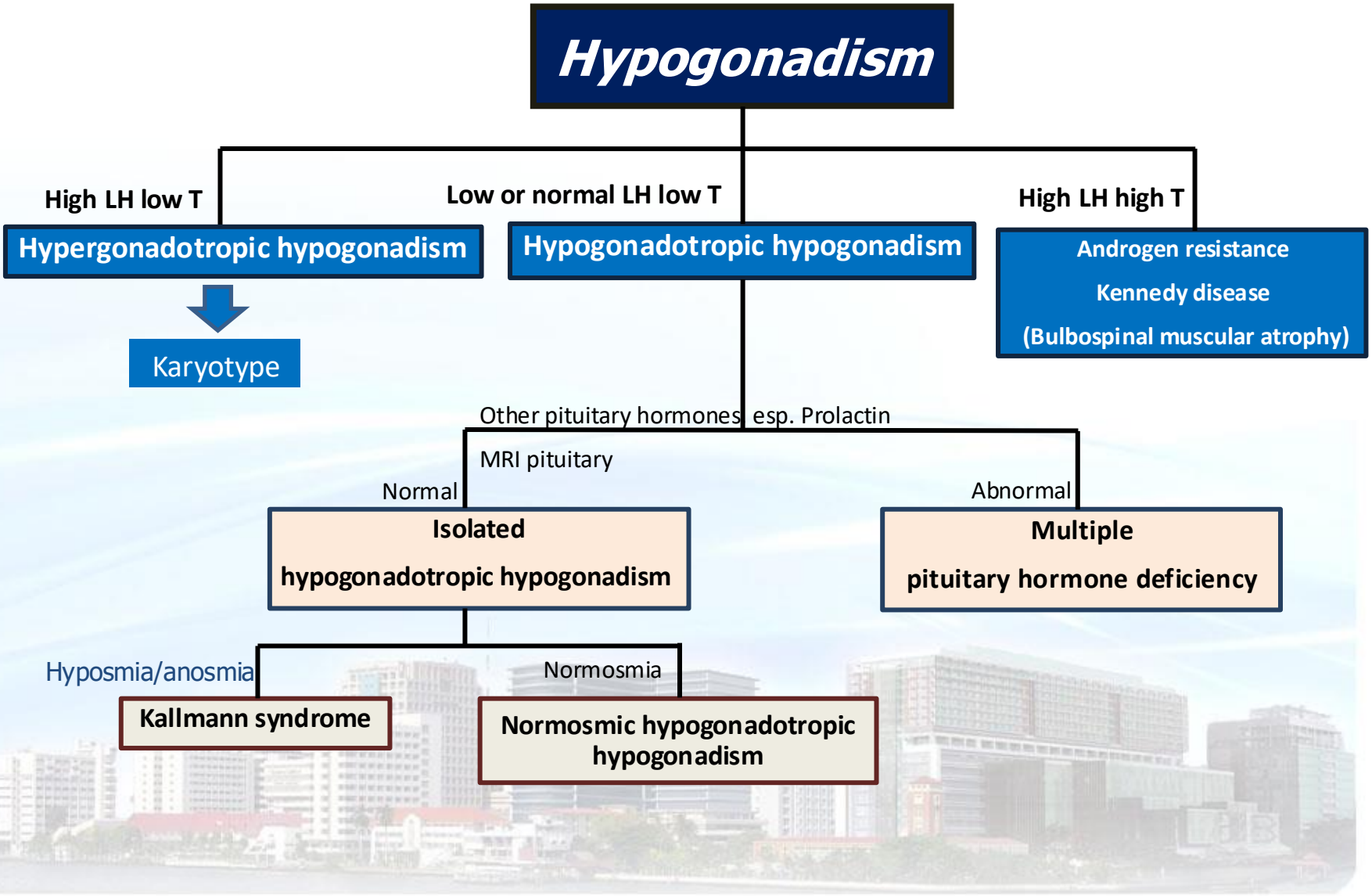
3.0-10.0 ng/mL
(300-1000 ng/dL)

Androgen deficiency

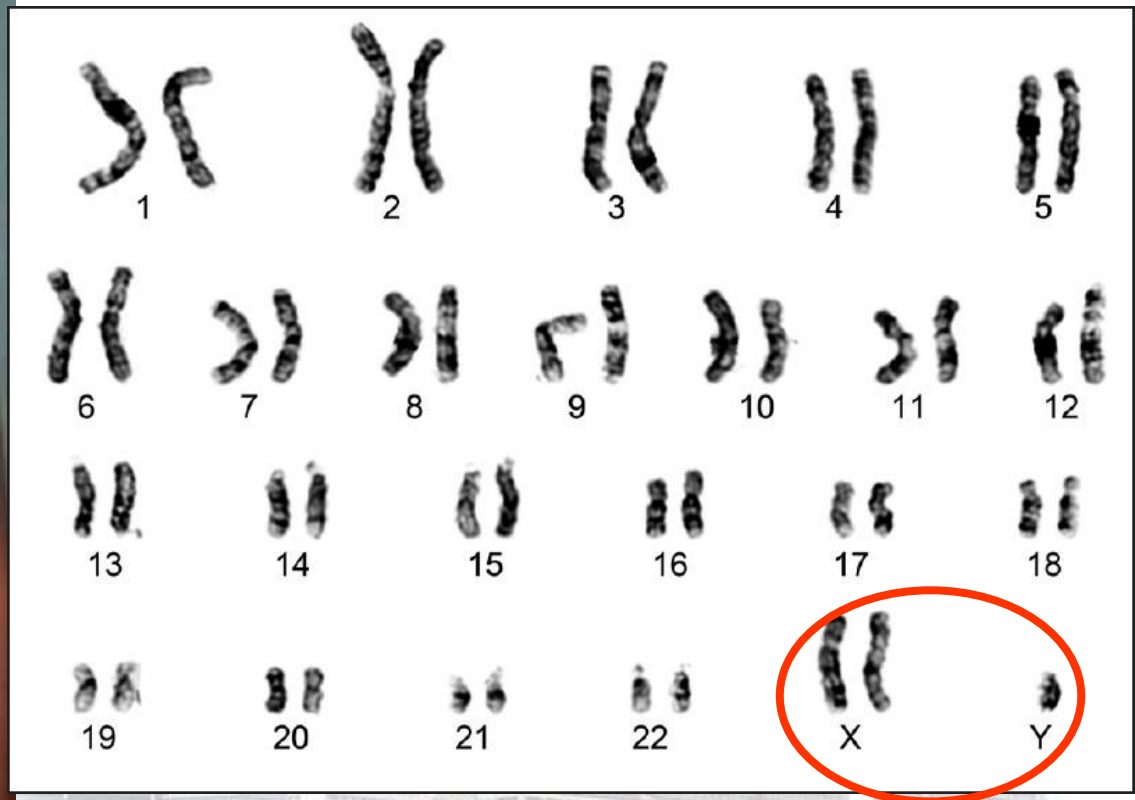
Total T < 264 ng/dL (2018)
> 1 measurements

Semen analysis	
Azoospermia	No sperm
Oligozoospermia	Sperm concentration < 16 million/mL

6th edition of WHO laboratory manual, 2021.

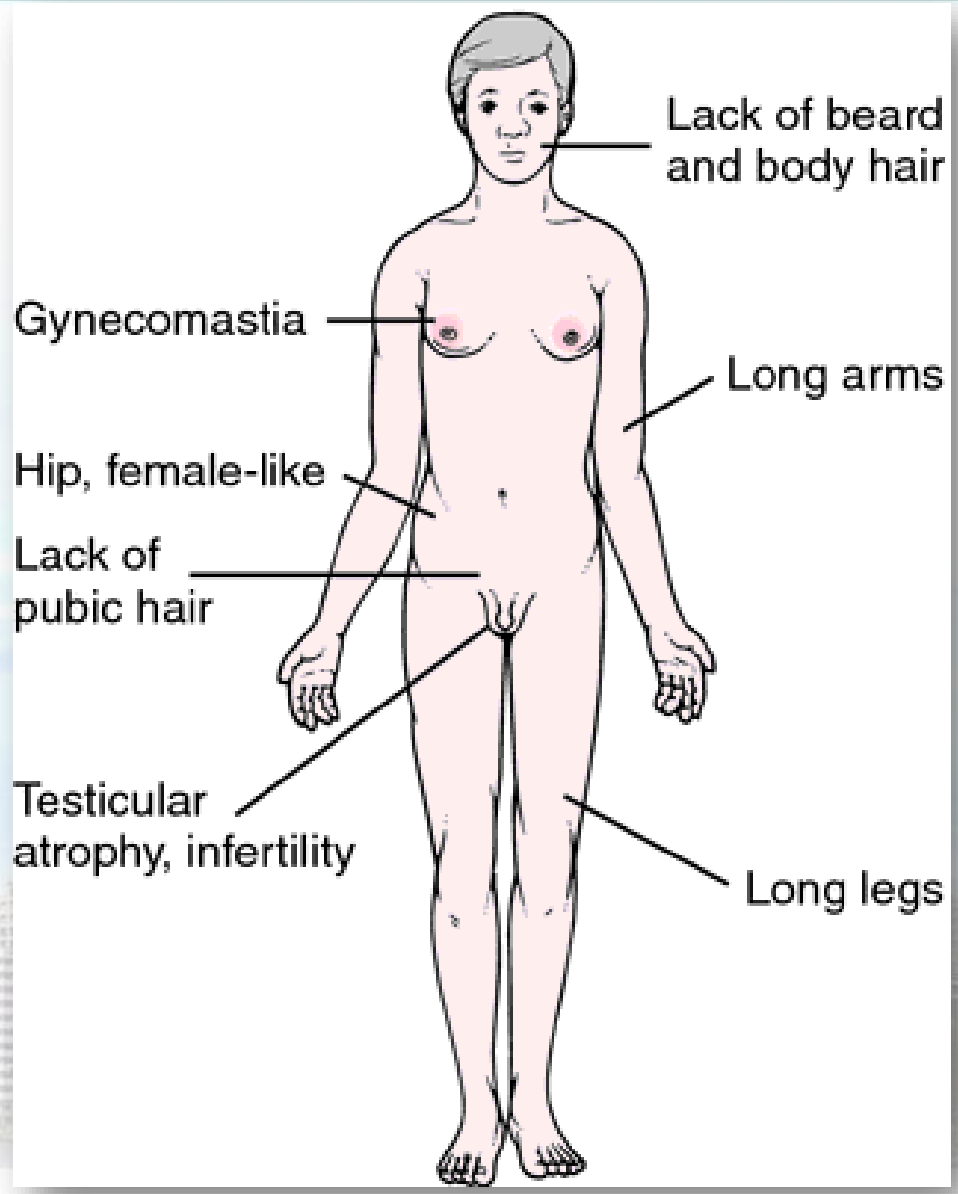

Hypogonadism
High LH low T
Hypergonadotropic hypogonadism
Karyotype
Low or normal LH low T
Hypogonadotropic hypogonadism
High LH high T
Androgen resistance
Kennedy disease
(Bulbospinal muscular atrophy)
Other pituitary hormones esp. Prolactin
MRI pituitary
Normal
Isolated
hypogonadotropic hypogonadism
Abnormal
Multiple
pituitary hormone deficiency
Hyposmia/anosmia
Kallmann syndrome
Normosmia
Normosmic hypogonadotropic hypogonadism

What is the most likely diagnosis?

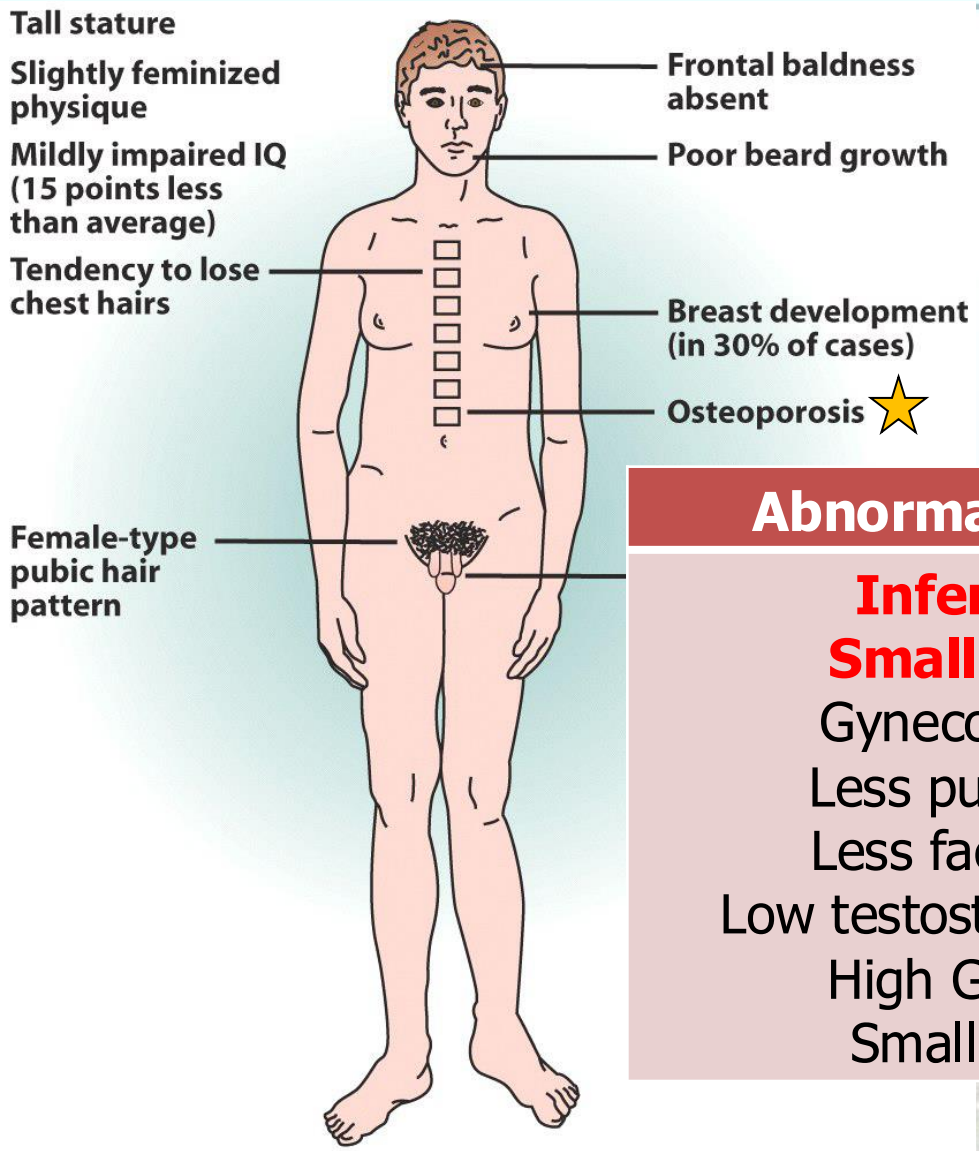


47XXY Klinefelter's syndrome

Klinefelter's syndrome

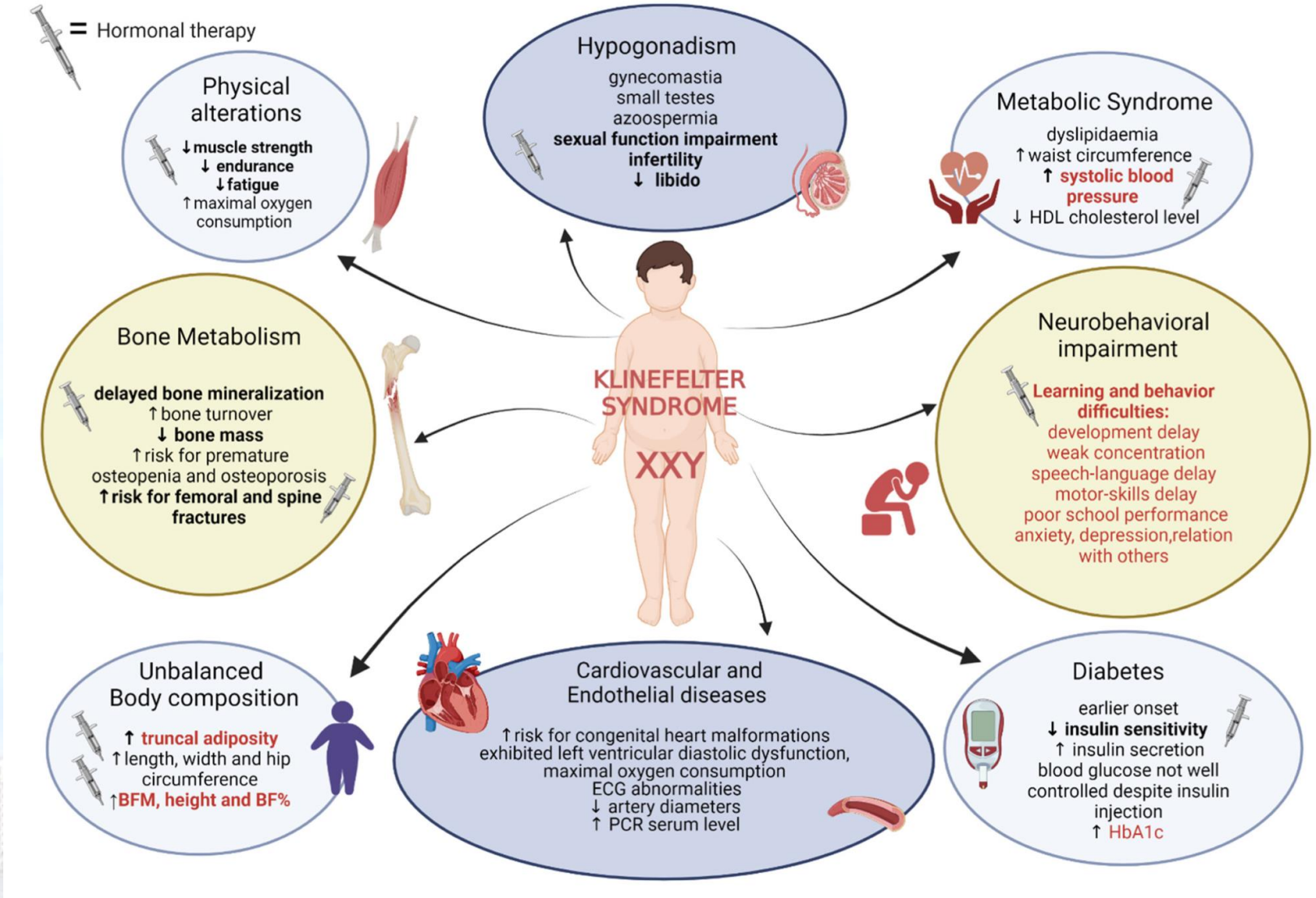


Klinefelter's syndrome

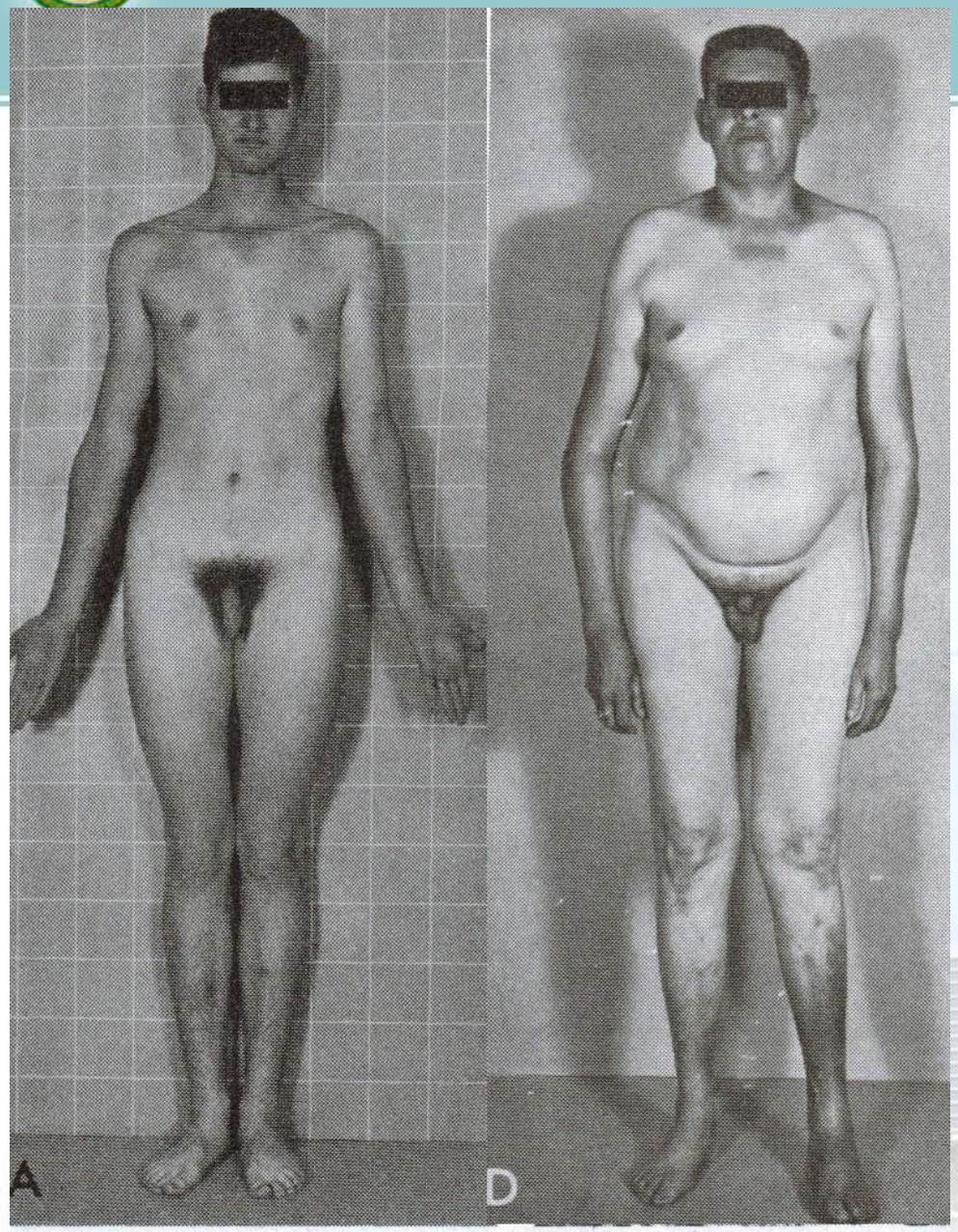


Abnormal finding	%
Infertility	**99 – 100**
Small testis	**99 – 100**
Gynecomastia	50 -75
Less pubic hair	30 – 60
Less facial hair	60 – 80
Low testosterone level	65 – 85
High Gn level	90 – 100
Small penis	10 – 25

Characteristics of KS



Nutrients **2022**, *14*, 2107



- Tall stature, Infertile

  LH <0.5

  FSH <0.5

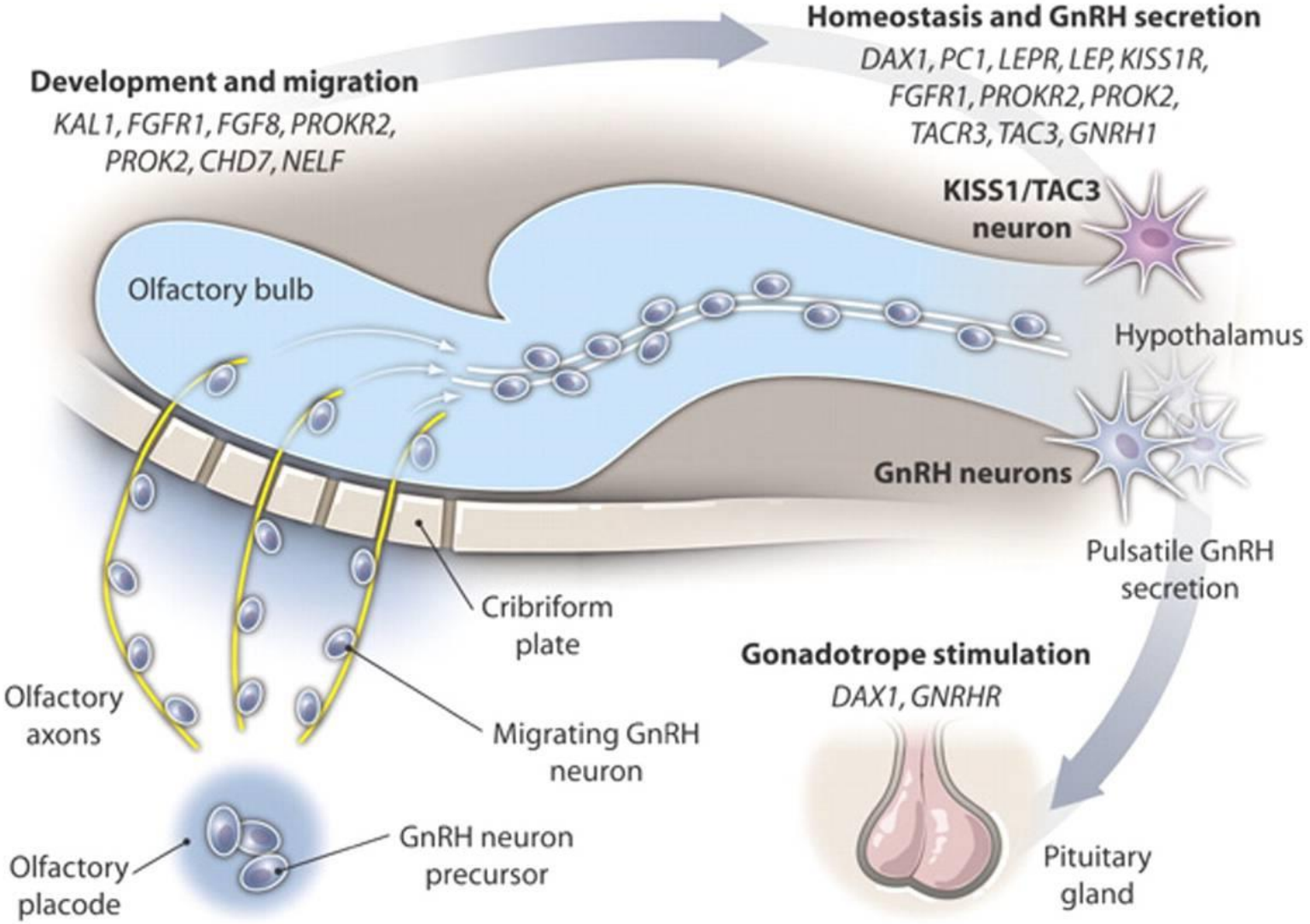
  testosterone <0.025

  Anosmia

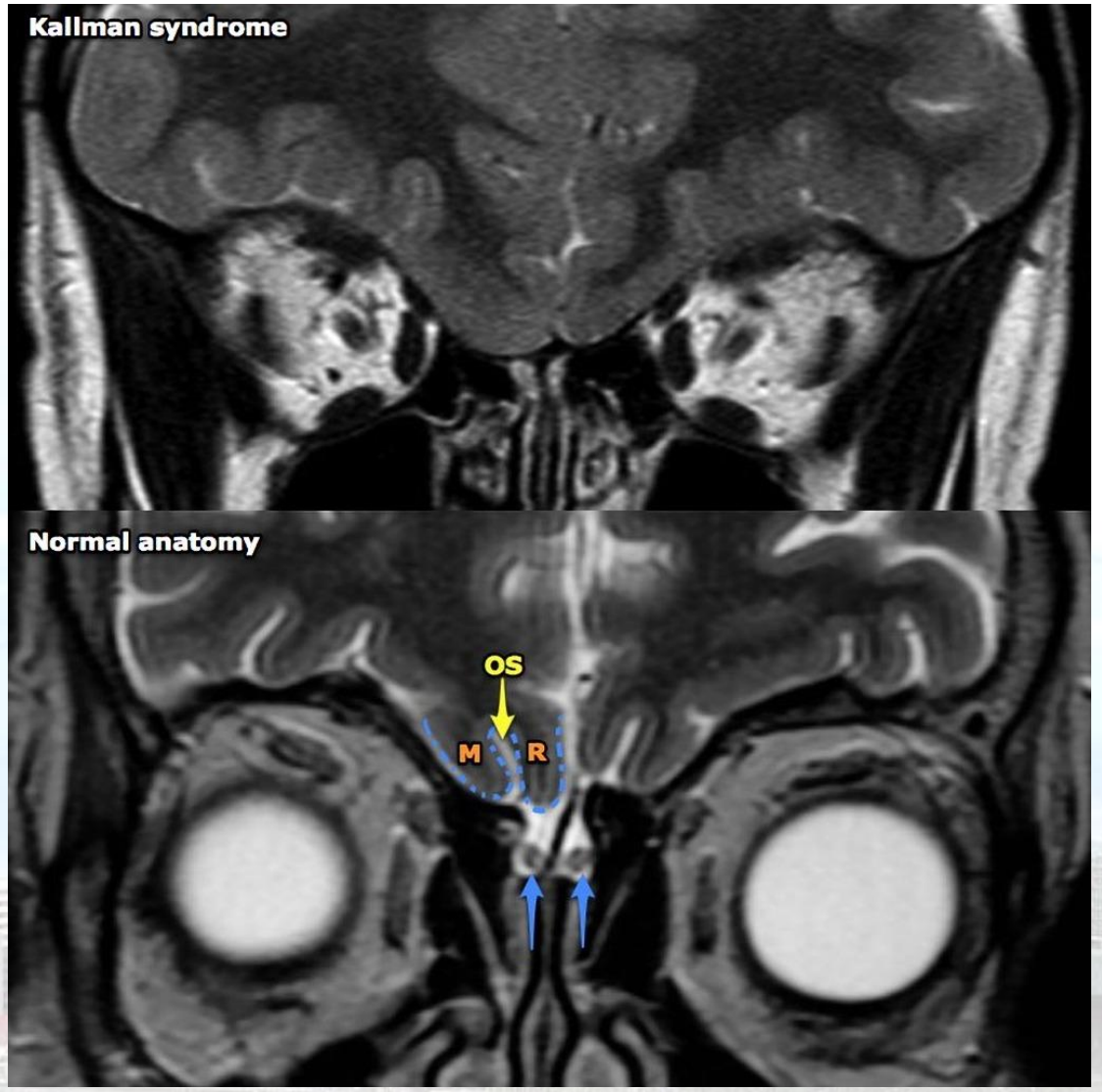
- What is the most likely diagnosis?

Kallmann syndrome

Isolated Hypogonadotrophic Hypogonadism


Homeostasis and GnRH secretion
DAX1, PC1, LEPR, LEP, KISS1R, FGFR1, PROKR2, PROK2, TACR3, TAC3, GNRH1
Development and migration
KAL1, FGFR1, FGF8, PROKR2, PROK2, CHD7, NELF
KISS1/TAC3 neuron
Olfactory bulb
Hypothalamus
GnRH neurons
Pulsatile GnRH secretion
Cribriform plate
Gonadotrope stimulation
DAX1, GNRHR
Olfactory axons
Migrating GnRH neuron
Olfactory placode
GnRH neuron precursor
Pituitary gland

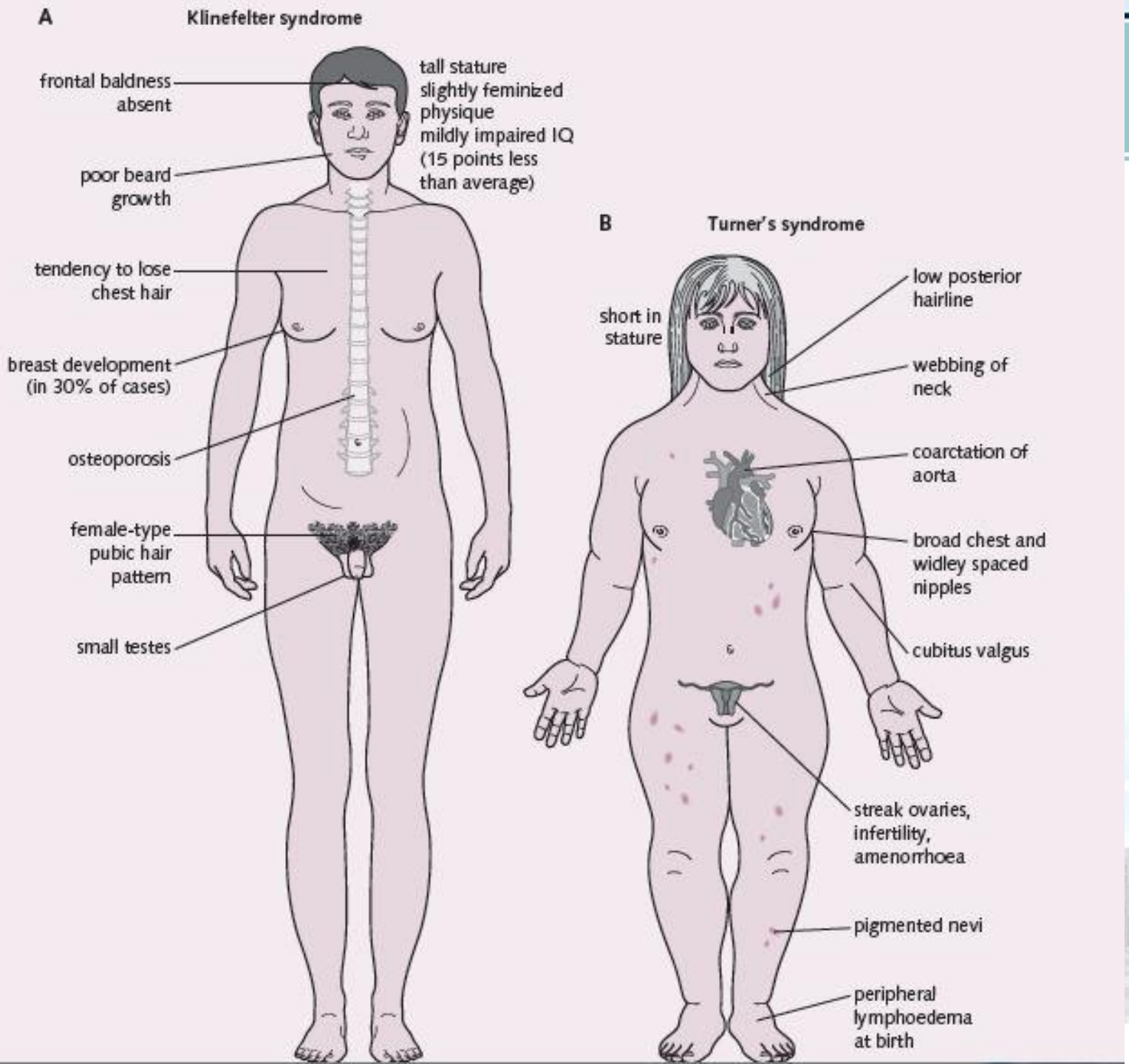
MRI: Kallman syndrome



	Klinefelter	Kallmann
Inheritance	-	Most (2/3) : sporadic (new mutation) , others : AD, AR, X-linked
Gynecomastia	Common	Rare (usu s/e of androgen replacement)
Testis	Small, **firm**	Small, **rubbery** or atrophic
Stature	Normal – tall Long leg	Normal No pubertal growth spurt Long arms and legs
Gn levels	**High**	**Usually undetectable**
GnRH test LH response	Hyperresponse at puberty	Prepubertal or no response
Plasma gonadal steroids	Low or normal	Low
Olfaction	**Normal**	**Anosmia or hyposmia**

	Klinefelter	Kallmann
Incidence	1:1,000 live male birth	1: 10,000 in male 1:50000 in female
Karyotype	47XXY or variants -48 XXXY -46XY, 46 XXY mosaicism -46XX male	Normal
Associated congenital anomalies		Midline facial defect, red-green color blindness, urogenital tract anomalies, synkinesis, neurosensory hearing loss
Intellectual function	Many with impairment Psychosocial abnormality marked lack of insight poor judgment impaired ability to learn from adverse experience Deficit in the ability to sustain attention	

	Klinefelter	Kallmann
Morbidities and mortality	Pulmonary diseases chronic bronchitis Bronchiectasis emphysema Cancers **germ cell tumors** (particularly extragonadal tumors involving the mediastinum) **breast cancer** possibly non-Hodgkin lymphoma varicose veins, leading to leg ulcers SLE, probably due to the extra X chromosome Diabetes mellitus	Congenital heart diseases Neuropsychiatric problems


A
Klinefelter syndrome
frontal baldness absent
poor beard growth
tendency to lose chest hair
breast development (in 30% of cases)
osteoporosis
female-type pubic hair pattern
small testes
tall stature
slightly feminized physique
mildly impaired IQ (15 points less than average)
B
Turner's syndrome
short in stature
low posterior hairline
webbing of neck
coarctation of aorta
broad chest and widley spaced nipples
cubitus valgus
streak ovaries, infertility, amenorrhoea
pigmented nevi
peripheral lymphoedema at birth

SIENDO

Faculty of Medicine Siriraj Hospital