

GRAND ROUND ENDOCRINOLOGY

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DO YOU KNOW RICKETS ?

**What is the
RICKETS ?**

RICKETS type ?

Management and Follow -up

**Approach to
RICKETS**



Case 01 presentation 1



Case 1 A 2 year- 4-months old girl

Chief complaint : ไม่เดิน 6 เดือนก่อนมาโรงพยาบาล

Present illness

- เดิมเริ่มเดินได้ตั้งแต่อายุ 1 ปี
- 6 เดือนก่อนมา รพ. เริ่มไม่ยอมเดิน งอแงตอนผู้ป่วยครองจับให้เดิน ปฏิเสธประวัติ อุบัติเหตุก่อนหน้านี้นั้น หลังจากนั้นmarดาสังเกตเห็นว่าผู้ป่วยมีขาโก่งและบวมบริเวณรอบหัวเข่าทั้ง 2 ข้าง ข้อมือบวมทั้ง 2 ข้าง น้ำหนักขึ้นช้า รับประทานได้น้อยลงจากปกติ ไม่มีไข้ ไม่มีผื่นขึ้นตามตัว ไม่มีไอน้ำมูกน้ำมาก่อน
- 4 เดือนก่อนมา รพ. ไปพบแพทย์ที่ รพ.รัฐบาลแห่งหนึ่ง ตรวจพบ กะโหลกศีรษะ บริเวณหน้าผากบวมโน ไม่เจ็บไม่ร้อน กระดูกซีโครงผิดปกติร่วมกับอาการเข่นเดียวกันกับ 6 เดือนก่อนมาโรงพยาบาล และ ส่งตัวรับการรักษาตามสิทธิ์

Case 1 A 2 year- 4-months old girl

Personal History

- No U/D
- No history of trauma
- Term AGA, BBW 3,150 gm, no complication
- Vaccination – completed EPI program, no non-EPI vaccination
- Diet – ข้าว 3 มื้อ กินเนื้อสัตว์เนื้อแดงมีละ 5-6 ช้อนโต๊ะ ไม่กินผักผลไม้ กินนมจีดวันละ 3 กล่อง
- Development
 - GM เดินวิ่งได้ ขึ้นลงบันได ส่องเท้าได้ (ก่อนป่วย)
 - FM ขีดเส้นยุ่งๆได้
 - PS ตักข้าวกินเองได้
 - EL พูด combine word มีความหมาย
 - RL ทำตามคำสั่งง่ายๆ 1 คำสั่งโดยไม่ต้องซึ่งอก

Case 1 A 2 year- 4-months old girl

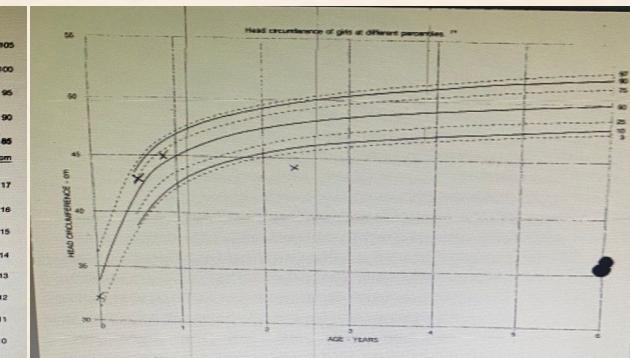
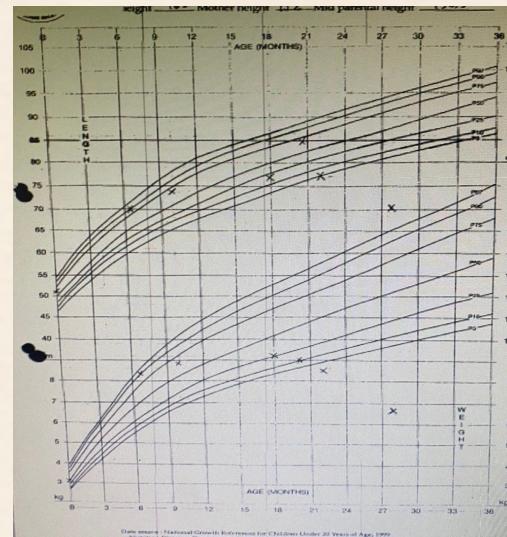
Physical examination

- BW 6.7 kg (<P3), Height 70 cm (<P3)
- BT 37.1 c, PR 140/min, RR 30/min, BP 98/55 mmHg
- GA: alert, active, good orientation
- HEENT: **protruded forehead**, no pale, no icteric sclera, no neck mass, no dysmorphic feature, no injected pharynx, no exudate
- RS : **rachitic rosary ribs both sides**, no harrison grooved sign, lung clear, equal breath sound
- Heart: regular normal S1S2, no murmur
- Abdomen: soft, normoactive bowel sound, no mass, liver and spleen can't be palpated
- Female genitalia, no ambiguous genitalia

Case 1 A 2 year- 4-months old girl

Physical examination

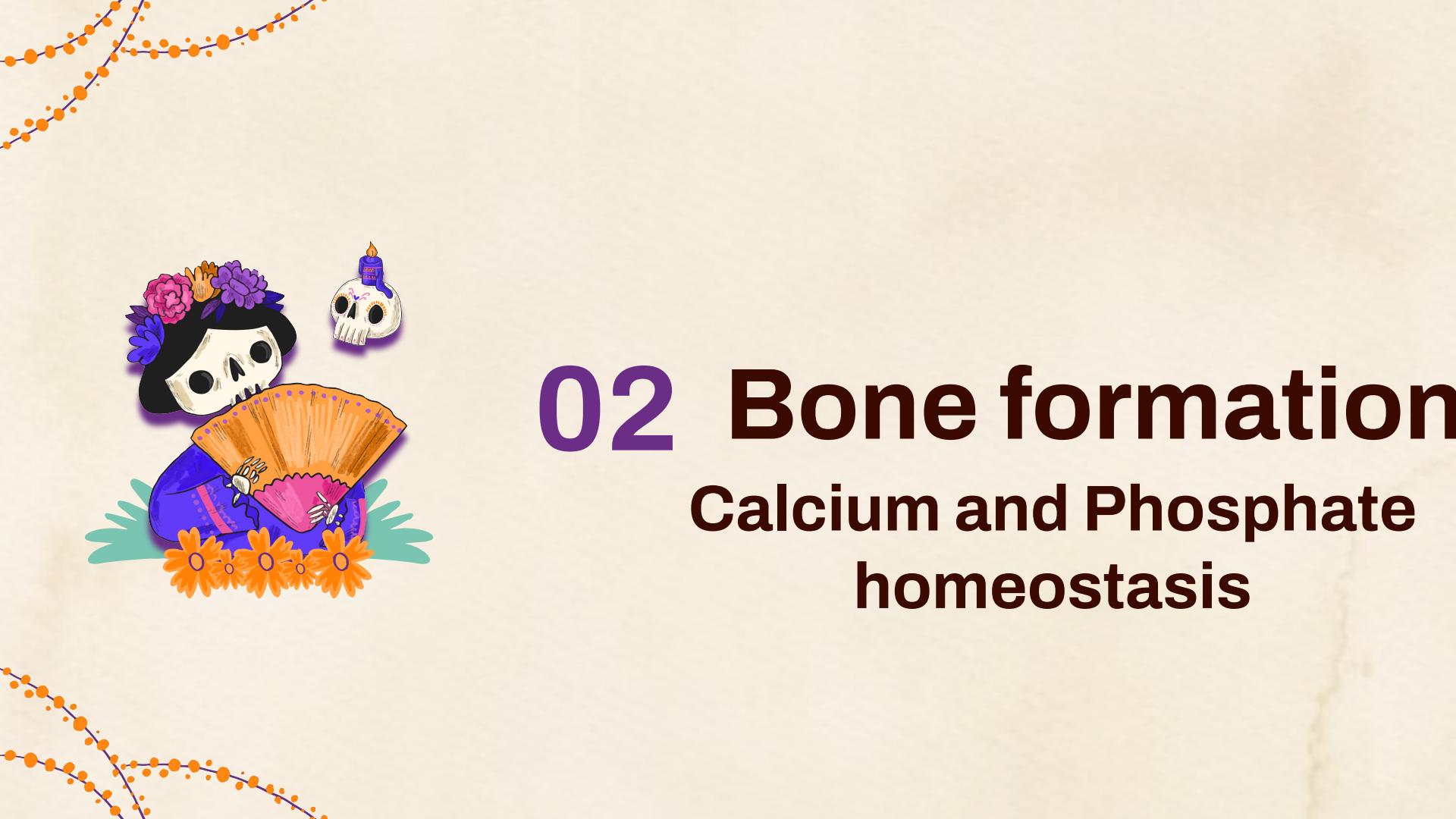
- Back : no spine deformity, no dimple, no mass
- **Extremities: bow leg both sides, muscle dystrophy at lower limb, widening wrist both sides**
- CNS: good orientation, no CN palsy, sensation- grossly intact, DTR 2+ All



Problem list

2 years 4 months old girl presented with
Clinical rickets and failure to thrive

Frontal bossing, Rachitic rosari, bowlegs and windening of wrist joint



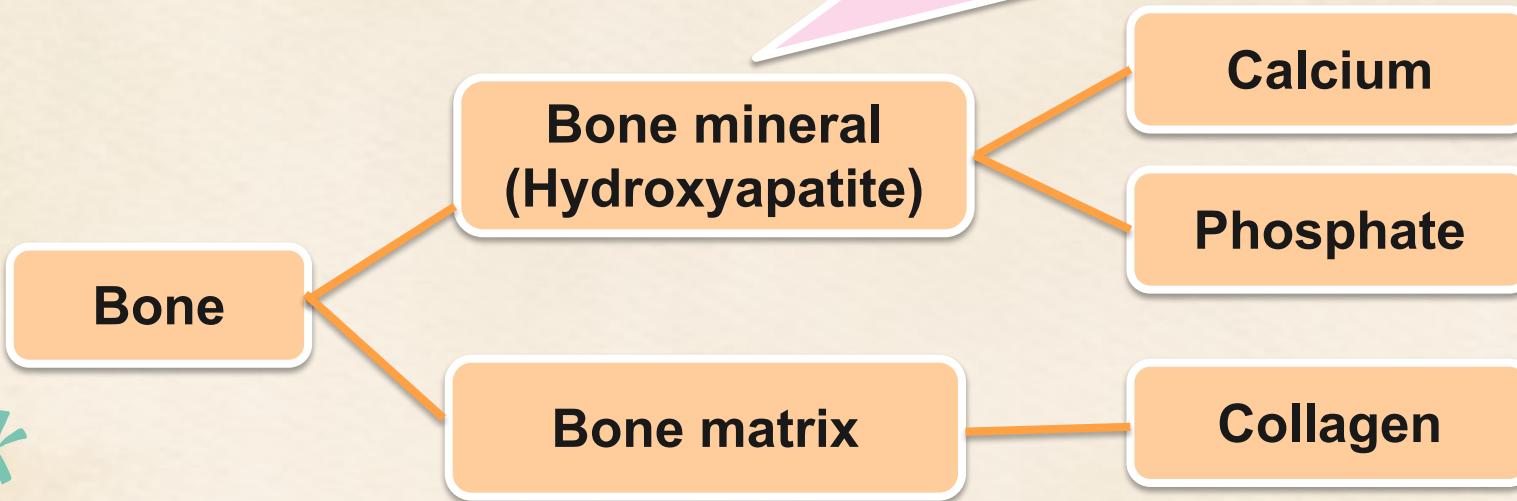
02 Bone formation

Calcium and Phosphate homeostasis



Bone composition

- **Rickets :**
 - Defective mineralization of Cartilage in the growth plate
- **Osteomalacia :**
 - Defective mineralization in bone cortex& trabeculae

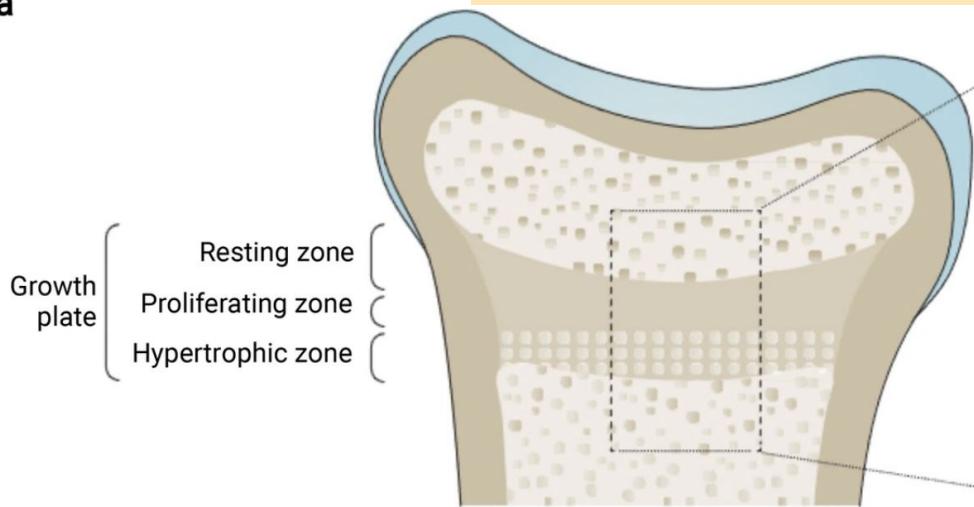


- **Osteogenesis imperfecta :**
 - Defective of collagen in bone cortex& tracbeculae

Bone formation

a

a, b. Normal healthy growth plate

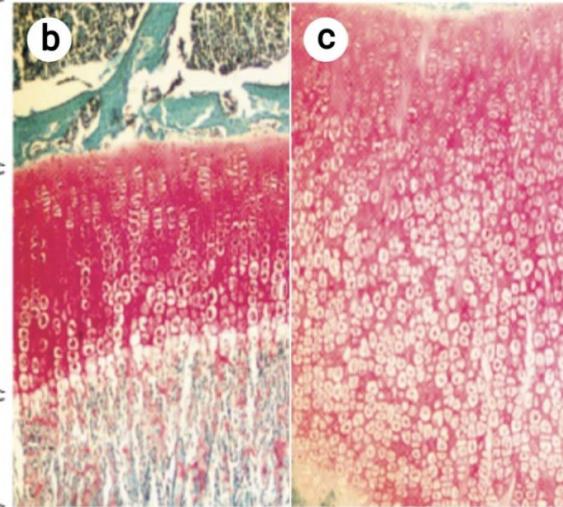


Epiphysis

Growth plate

Metaphysis

c. Growth plate in rickets.



* The growth plate is characterized by maturation of chondrocytes progressively from the epiphysis □ the metaphysis.

Rickets is caused by impaired apoptosis of hypertrophic chondrocytes and impaired mineralization of the cartilage matrix surrounding the apoptotic chondrocytes □ widening of the growth plates in bones and is usually associated with osteomalacia

Calcium and Phosphate Homeostasis

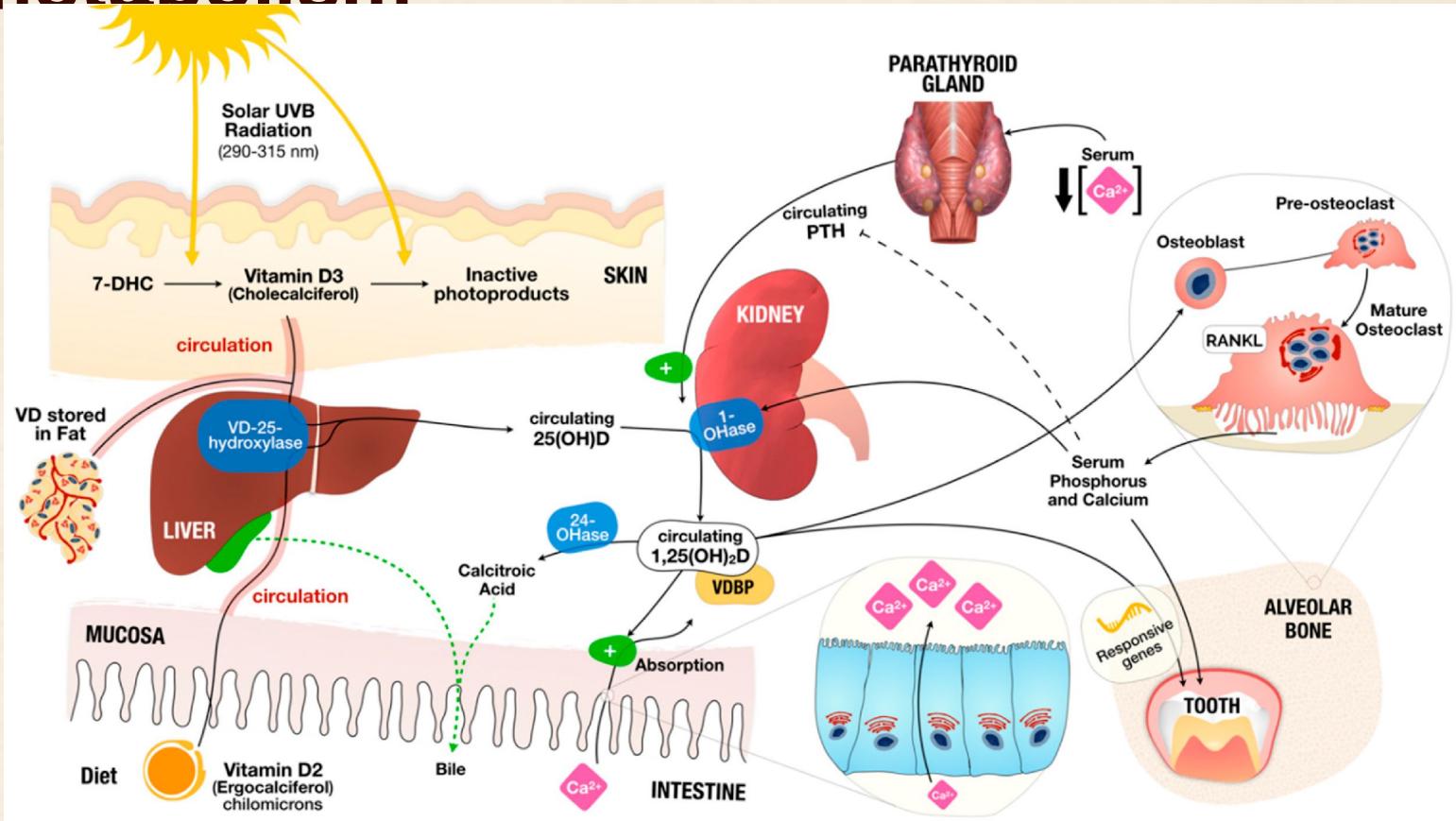
Hormone

- Parathyroid hormone (PTH)
- Vitamin D
- Fibroblast growth factor 23 (FGF23)
- Calcitonin

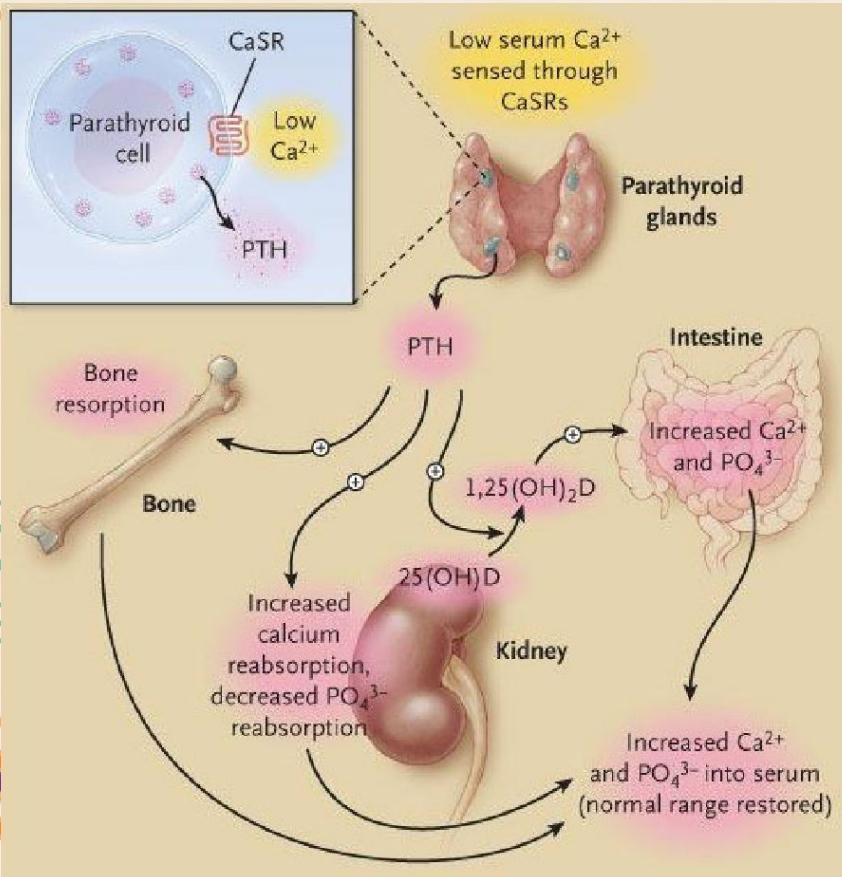
Organs

- Bone
- Liver
- Kidney
- Gastrointestinal system
- Parathyroid gland
- Skin

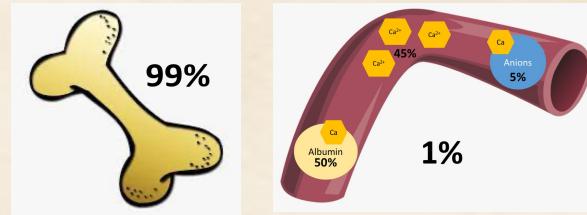
Calcium Homeostasis and vitamin D metabolism



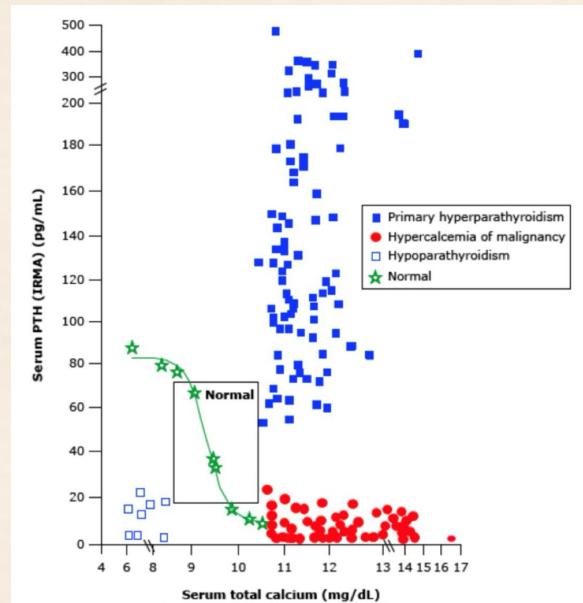
Calcium Homeostasis and parathyroid hormone



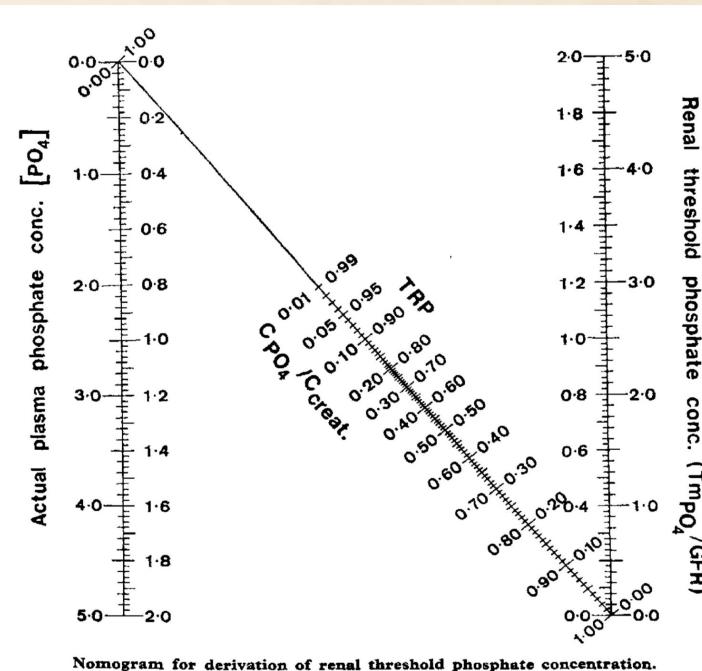
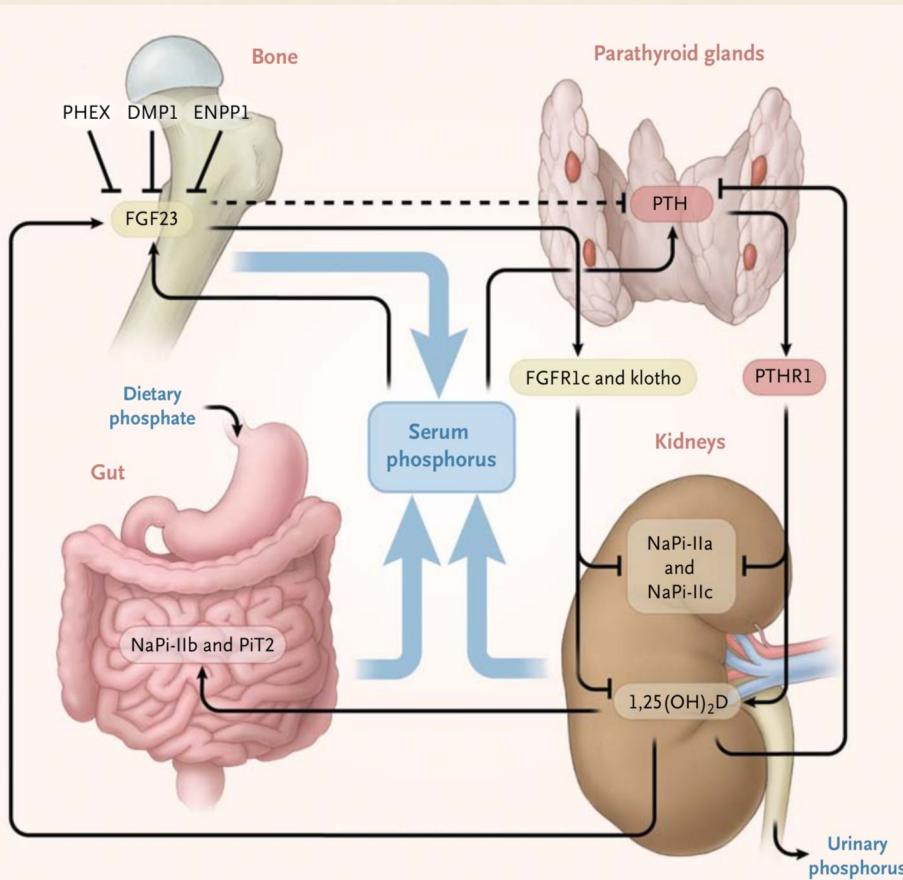
Calcium distribution



Serum PTH in hypercalcemia & hypocalcemia



Phosphate homeostasis



Tubular resorption of phosphate
Normal TRP 80 – 90% (85-95%)

Effect of PTH, vitamin D and Calcitonin

	PTH	Vitamin D	Calcitonin
Bone	Stimulate Osteoclast ↑Bone resorption	Accumulation of Ca in bone Stimulate Osteoclast	Inhibit osteoclast
Kidney	↑Ca reabsorption ↓PO ₄ reabsorption ↑1A-OH activity	↑Ca reabsorption ↑PO ₄ reabsorption	↓Ca reabsorption ↓PO ₄ reabsorption
GI	No direct effect	↑Ca absorption ↑PO ₄ absorption	No direct effect
Total Effect	↑Ca ↓PO ₄	↑Ca ↑PO ₄	↓Ca

03

Approach to Rickets



Clinical presentation

General

Failure to thrive
Listlessness
Protruding abdomen
Muscle weakness(esp.proximal)
Hypocalcemic dilated cardiomyopathy
Pathologic fracture
Increased ICP

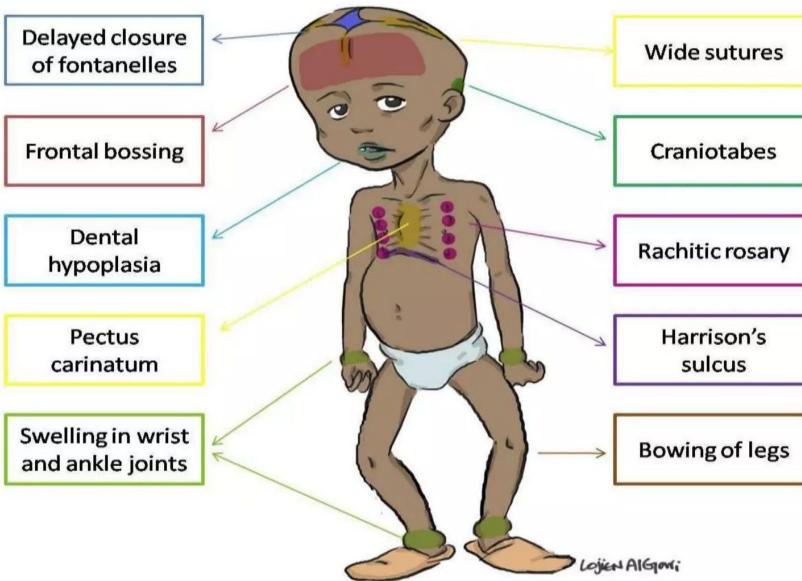
Head

Craniotabes, frontal bossing, delayed fontanel closure, delay dentition, caries, craniosynostosis

Chest

Rachitic rosary, harrison groove, respiratory infections and atelectasis

10 important clinical features in Rickets



Back

Scoliosis, kyphosis, lordosis

Extremities

Enlargement of wrist and ankle
Valgus or varus deformities
Windswelt deformity
Anterior bowing of leg
Leg pain
Short stature(short limb)

Hypocalcemic symptoms

Tetany, seizure,
stridor(laryngeal spasm)

Clinical presentation



B



A

Radiological finding

- Cupping of metaphysis
- Fraying of metaphysis
- Flaring of metaphysis
- Widening of epiphyseal plates
- Thining of cortex
- Bowing deformities of bilateral femur and tibia
- Osteopenia

*History taking and physical examination

History taking

- **Prenatal history** : GA(preterm)
- **Diet** : Calcium and vitamin D intake
- **Lifestyle** : Sun exposure
- **Underlying disease** : Liver, renal, GI malabsorption
- **Drug**
- **Family history** : Bone disease, short stature, history of fracture



Physical examination

- **Completed physical examination**
- **Skull** : Craniotabes, Frontal bossing, widening of fontanel
- **Chest** : Rachitic rosary, harrison groove, pigeon chest
- **Extremities** : Limb deformities, genu varus, genu vulgus, joint widening/swelling
- **Spine** : Spine deformities, kyphosis
- **Musculoskeletal** : Gait disturbance, bone pain, growth retardation, delayed dentition, enamel hypoplasia
- **Sign of hypocalcemia(Infant)**

Investigation

Serum

Bone panel

- Calcium, iCa
- Phosphate
- Magnesium
- Alkaline phosphatase
- PTH
- Albumin
- 25(OH) D
- 1, 25(OH)₂ D
- FGF 23

- Electrolyte
- BUN, creatinine

Urine

- Urinary analysis
- Calcium
- Phosphate
- Creatinine
- Tubular resorption for PO₄

$$TRP = \{1 - [(U_{PO_4}/P_{PO_4}) \times (P_{Cr}/U_{Cr})]\} \times 100$$

Normal range TPR 80-90%

Urine Ca/Cr

X-ray

- Wrist, knee, ankle
- Long bone
- Skeletal survey

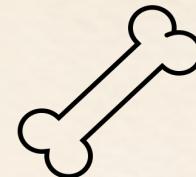


Normal values of serum calcium and phosphate

Phosphorus	(mg/dL)
0 - 9 day	4.5-9.0
10 days - 24 months	4.5-6.5
3 - 9 years	3.2-5.8
10 - 15 years	3.3-5.4
> 15 years	2.4-4.4

ALP	U/L
Infant	150-420
2-10 years	100-320
Adolescent boy	100-390
Adolescent girl	100-320
Adult	30-120

iPTH	pg/ml
Children and adults	10-65



Vitamin D25(OH)D	(ng/mL) _{ref.2}	(ng/mL) _{ref.3}
Sufficiency	30 - 100	> 20 - 100
Insufficiency	21 - 29	12 - 20
Deficiency	< 20	< 12

Calcium	(mg/dL)
Preterm	6.2 - 11
Term < 10 days	7.6 - 10.4
10 days- 24 months	9.0 - 11
2-12 years	8.8 - 10.8
12-18 years	8.4 - 10.2

1.Soghier L et al.AAP.Reference Range Values for Pediatric Care.

2.J Clin Endocrinol Metab 2011;96(7):1911-30

3.Thai society for pediatric endocrinology.ข้อแนะนำการป้องกันและรักษาภาวะพิร่องวิตามินดีในทารก เด็ก และ

Approach to Rickets

Table 64.2 Causes of Rickets

VITAMIN D DISORDERS

Nutritional vitamin D deficiency
Congenital vitamin D deficiency
Secondary vitamin D deficiency
Malabsorption
Increased degradation
Decreased liver 25-hydroxylase
Vitamin D-dependent rickets types 1A and 1B
Vitamin D-dependent rickets types 2A and 2B
Chronic kidney disease

CALCIUM DEFICIENCY

Low intake
Diet
Premature infants (rickets of prematurity)
Malabsorption
Primary disease
Dietary inhibitors of calcium absorption

PHOSPHORUS DEFICIENCY

Inadequate intake
Premature infants (rickets of prematurity)
Aluminum-containing antacids

RENAL LOSSES

X-linked hypophosphatemic rickets*
Autosomal dominant hypophosphatemic rickets*
Autosomal recessive hypophosphatemic rickets types 1 and 2*
Hereditary hypophosphatemic rickets with hypercalciuria
Overproduction of fibroblast growth factor-23
Tumor-induced rickets*
McCune-Albright syndrome*
Epidermal nevus syndrome*
Neurofibromatosis*
Fanconi syndrome
Dent disease
Distal renal tubular acidosis

*Disorders secondary to excess fibroblast growth factor-23.

Approach to Rickets

Calcipenic rickets

- Nutritional deficiency
- GI malabsorption
- Abnormal Vitamin D metabolism

Phosphopenic rickets

- Dietary phosphate deficiency
- GI malabsorption
- Increase renal phosphate loss

Calcipenic Rickets

1. Nutritional deficiency

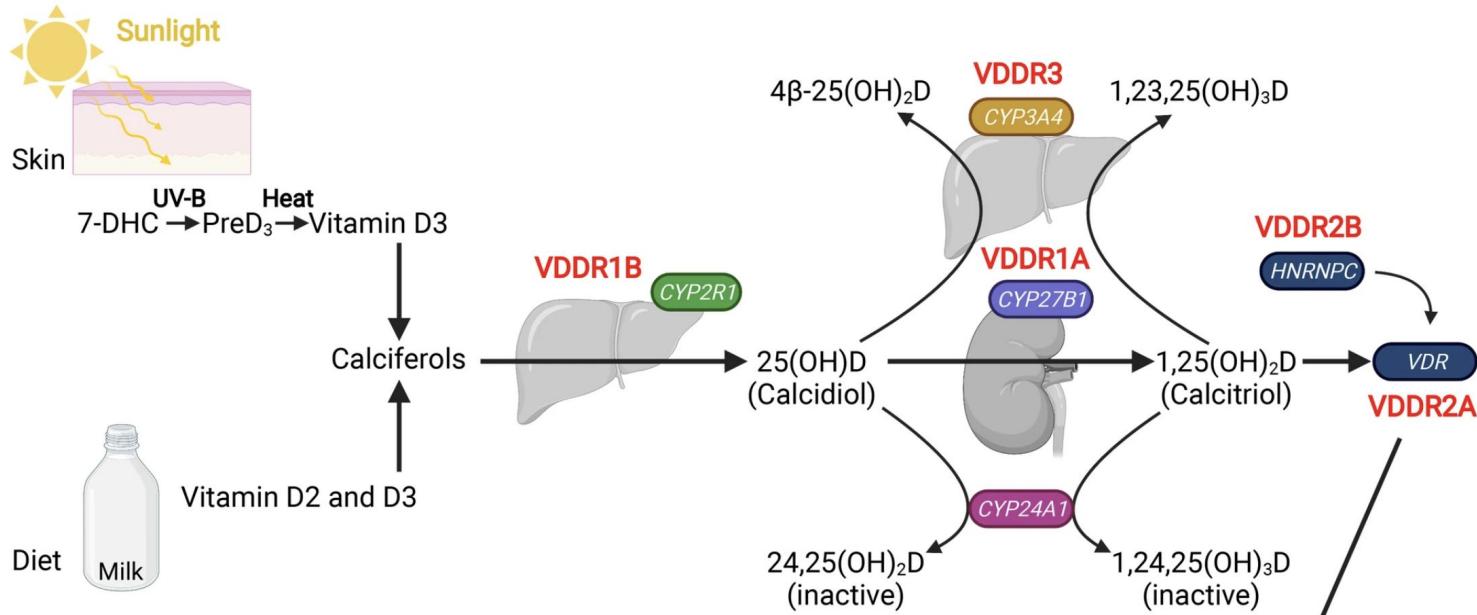
- Vitamin D deficiency (MC cause if nutritional deficiency rickets)
- Low calcium intake

2. Poor calcium absorption

- Malabsorption disease : celiac disease, cystic fibrosis

3. Genetic defect of vitamin D metabolism

Vitamin D metabolism



- ↑ Renal tubular calcium reabsorption
- ↑ Intestinal calcium absorption
- ↑ Osteoclastic bone reabsorption

Calcipenic Rickets

Disease	Pathogenesis	Gene	Ca	P	25(OH)D	1,25(OH) ₂ D
Nutritional rickets (Vitamin D deficiency)	Vitamin D deficiency	NA	↓ or N	↓ or N	N	↑ or N
Nutritional rickets (Ca deficiency)	Calcium deficiency	NA	↓ or N	↓ or N	↓↓	Varies
VDDR1A	Impaired synthesis of 1,25(OH) ₂ D	CYP27B1 (12q14.1)	↓	↓ or N	↑ or N	↓
VDDR1B	Impaired synthesis of 25(OH)D	CYP2R1 (11p15.2)	↓	↓ or N	↓↓	Varies
VDDR2A	Impaired signaling of the VDR	VDR (12q13.11)	↓	↓ or N	N	↑↑
VDDR2B	Impaired signaling of the VDR	HNRNPC	↓	↓ or N	N	↑↑
VDDR3	Inactivation of 1,25(OH) ₂ D	CYP3A4	↓	↓	↓	↓

Serum Ca↓, PTH↑, ALP↑, Urine Ca↓

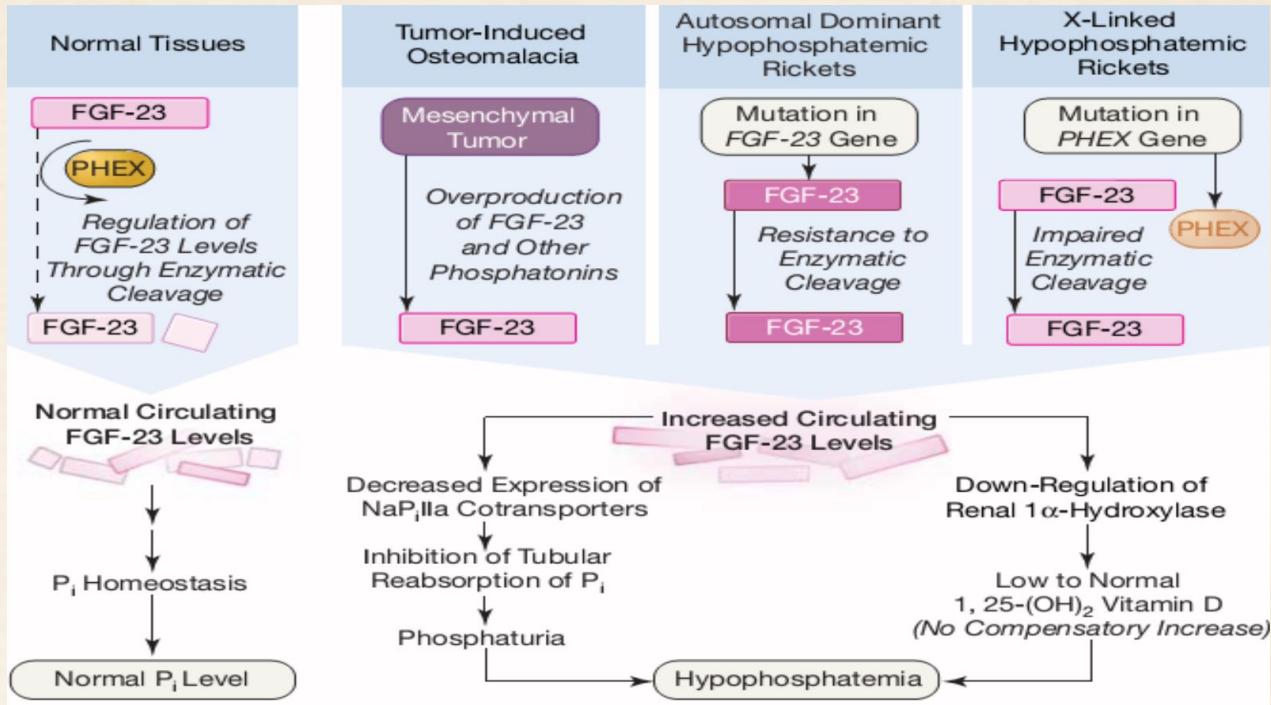
Phosphopenic Rickets

1. Dietary phosphate deficiency
2. Impaired intestinal absorption of Phosphate
3. Increase renal phosphate loss : Genetic defect, acquired caused



1. High FGF 23 : increase production or decrease degradation of FGF 23
2. Increase urinary phosphate loss due to mutation inactivation of sodium-dependent phosphate co-transporter at proximal tubule

Phosphopenic rickets



FGF-23

- Decreasing reabsorption of phosphate in the proximal tubule
- Decreases the activity of 1 α -OH \square Decreased production of 1,25(OH)₂D(calcitriol)

Phosphopenic Rickets

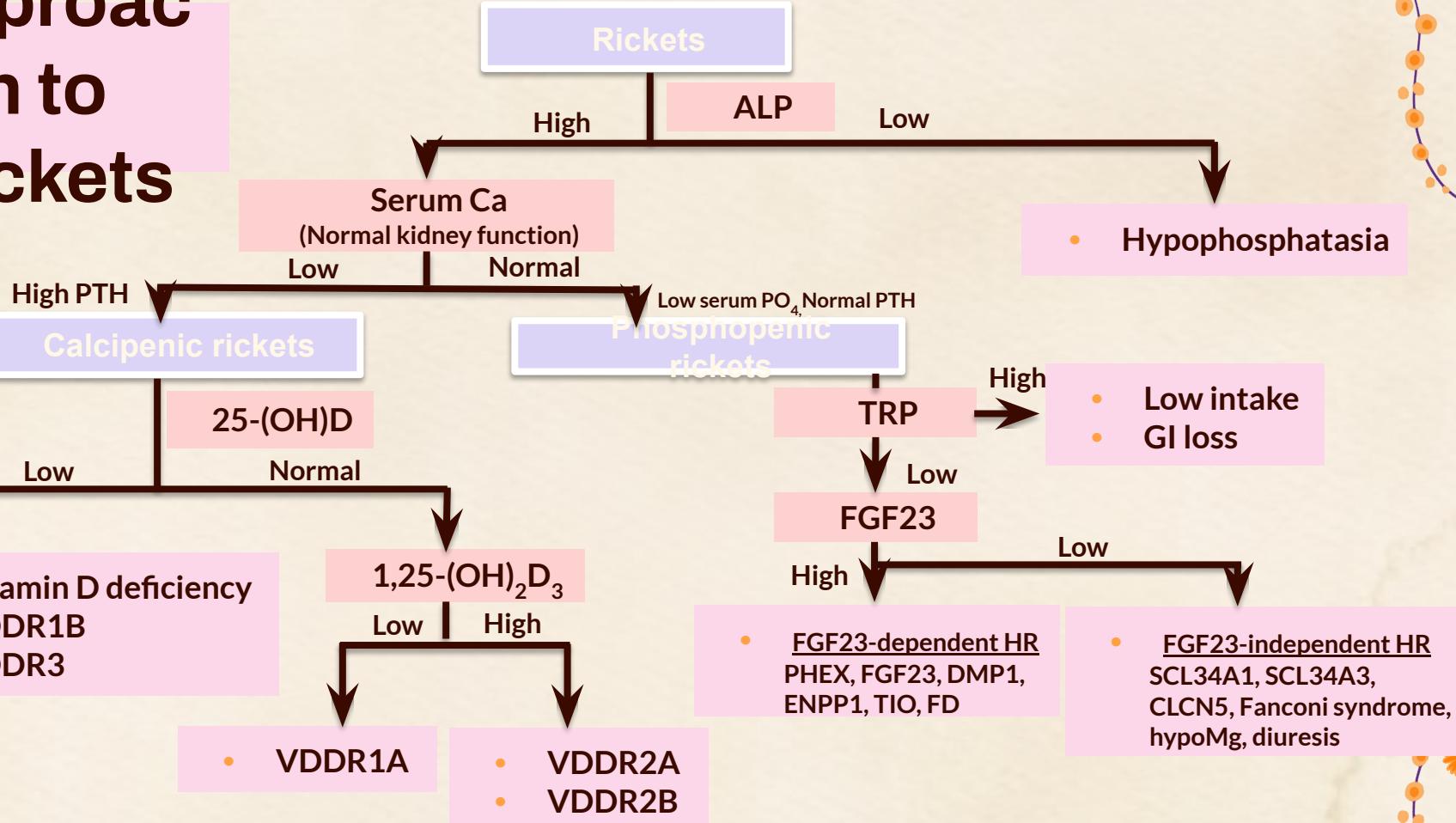
FGF23-dependent HR	Pathogenesis	Gene	Ca	P	ALP	PTH	TmP /GFR	Urine _{Ca/Cr}	FGF23
X-linked dominant HR	Increased FGF23 expression	PHEX	N	↓	↑	↓/N	↓	N	N/↑
Autosomal dominant HR	Resistance to proteolytic cleavage	FGF23	N	↓	↑	↓/N	↓	N	N/↑
Autosomal recessive HR	Increased FGF23 expression	DMP1	N	↓	↑	↓/N	↓	N	N/↑
McCune-Albright syndrome /polyostotic	Increased production of FGF23 by fibrous bone cells	GNAS1	N	↓/N	N/↑	↓/N	↓/N	N	N/↑
HR and hyperparathyroidism	Direct/Indirect effects of increased KLOTHO level	AlphaKL	N/↑	↓	↑	↓/N	↓	N	↑

Serum P low, TmP/GFR ↓, Urine Ca/Cr Normal, FGF23 N/↑

FGF23-independent HR	Pathogenesis	Gene	Ca	P	ALP	PTH	TmP /GFR	Urine _{Ca/Cr}	FGF23
Hereditary HR with hypercalciuria	Loss of function of NPTII	SLC34A3	N	↓	N/↑	N	↓	↑	↓/N
Hypophosphatemic rickets with nephrolithiasis&osteoporosis type 1	Loss of function of NaPi2a in the proximal tubule	SLC34A1	N/↑	↓	N/↑	↓/N	↓	↑	↓/N
Dent disease 1	Inactivate chloride channel	CLCN5	N	↓	↑	↓/N	↓	↑	↓/N

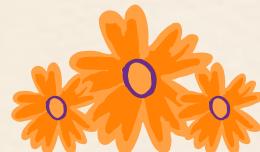
Serum P low, TmP/GFR ↓, Urine Ca/Cr ↑, FGF23 ↓/N

Approach to Rickets



04

Treatment and outcome



Treatment

Calcipenic rickets

Phosphopenic rickets

Calcium

Vitamin D

Phosphate

Treatment of Calcipenic Rickets

Vitamin D deficiency rickets

Age	Daily dose for 90 days, IU	Single dose, IU	Maintenance daily dose, IU
<3 months	2,000	NA	400
3–12 months	2,000	50,000	400
>12 months to 12 years	3,000–6,000	150,000	600
>12 years	6,000	300,000	600

NA = Not available. Reassess response to treatment after 3 months as further treatment may be required. Ensure a daily calcium intake of at least 500 mg. For conversion from IU to µg, divide by 40.

Maintain a blood level of 25(OH)D above 30 ng/mL

Treatment same as nutritional vitamin D deficiency

Source of vitamin D

Vitamin D₃(Cholecalciferol) : Cod liver oil, Salmon, Sardine, Mackerel, Tuna, Egg yolk, cheese

Vitamin D₂(Ergocalciferol) : Shiitake mushrooms

Vitamin D requirement

Aged < 1 yr : vitamin D 400 IU/day

Aged > 1 yr : vitamin D 600 IU/day

Calcium 30 – 75 mg/kg/days PO TID
For prevent hungry bone syndrome

Treatment of Calcipenic rickets

Genetic defect of vitamin D metabolism

- **VDDR1**

- Calcitriol 1 mcg/day or alphacalcidol 150 -200 ng/kg/day until normal bone x-ray then 25- 50 ng/kg/day
- Calcium supplement 50 – 75 mg/kg/day +-phosphate

- **VDDR2**

- High dose oral Calcitriol 1 – 6 mcg/kg/d, twice daily
- High dose iv/oral Calcium 1- 3 g/day

- **VDDR3**

- Viamin D₃ 20,000 – 50,000 IU daily

Goal

Low normal serum Ca
Normal serum P
High normal PTH

Avoid excessive of vitamin D

- Hypercalciuria, nephrocalcinosis
Keep Urine Ca < 4 mg/kg/day

Treatment of hypophosphatemic rickets

1. Elemental Phosphate

PO_4 Supplement 20 - 40 mg/kg/day divided in 3-5 times/day

Avoid dose > 80 mg/kg/day

2. Active vitamin D

Calcitriol 20 - 30 ng/kg/day divided in 1-3 doses

Alfacalcidol 30- 50 ng/kg/day devided in 1-2 doses

Goal

Normal ALP

Low normal P

Correct rickets

3. Burosumab(FGF23 Antibody) in XLH

Burosumab 0.4 mg/kg sc q 2 weeks

Avoid excessive of calcitriol

Hypercalciuria, nephrocalcinosis

Keep Urine Ca < 4 mg/kg/day

Follow-up

- Clinical : Growth, limb alignment, muscle strength, pain
- Ca, PO₄, ALP, PTH, 25OHD
- Urine Ca/Cr
- X-ray
- Renal ultrasound(for avoid nephrocalcinosis)

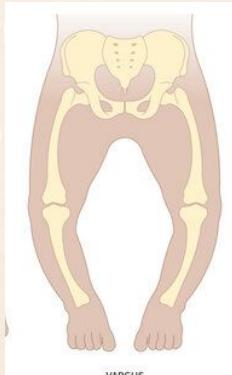
Prevention

- Bone deformities and functional problems

Bowlegs(Genu varum)

Physiologic bow legs

- Age < 2 years
- Bilateral and symmetry
- Normal gait
- Impaired growth



Pathologic bow legs

- Progressive/persistent
- Unilateral or asymmetry

Blount's disease

Rickets

Skeletal dysplasia

Renal insufficiency

Calcipenic

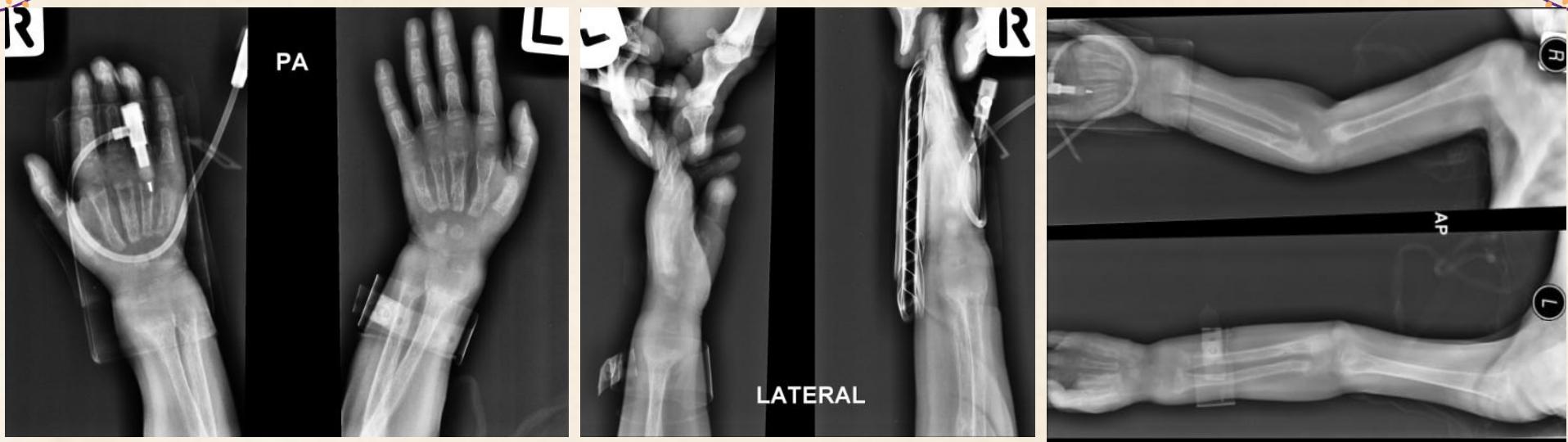
Phosphopenic

Others

Film X-ray



Film X-ray



Film X-ray



Laboratory

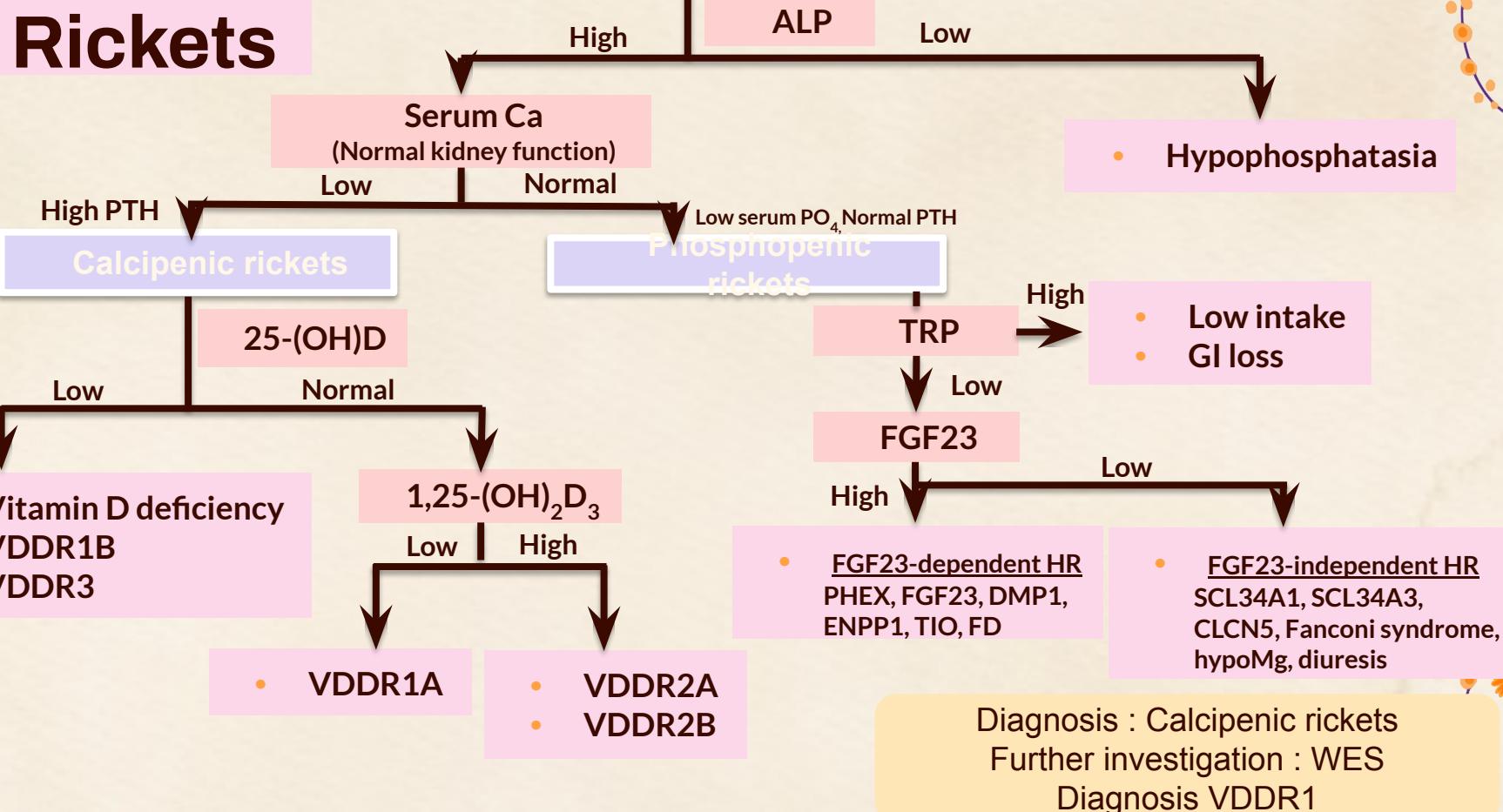
Bone panels

Ca	4.5 mg/dl
Mg	2.3 mg/dl
PO ₄	1.7 mg/dl
ALP	1,097 U/L
Vitamin D 25(OH)D	34.58 ng/ml
iPTH	348.3 pg/ml

Urine Cr	62.8 mg/dl
Urine Ca	3.9 mg/dl
Urine Ca/Cr	0.02
Urine P	181.1 mg/dl

Na	137 mEq/dl
K	3.54 mEq/dl
Cl	98.1 mEq/dl
CO2	20.1 mEq/dl
BUN	6.6 mg/dl
Cr	0.1 mg/dl
Total protein	7.7 g/dl
Albumin	4.6 g/dl

Approach to Rickets



Diagnosis and treatment

Calcipenic rickets

Management

- 10% Calcium gluconate
- CaCO_3 : 130 mg/kg/day
- Vitamin D_3 : 34 ng/kg/day
- Diet: add Milk 6 Oz x 4 feed

Ca	4.5 mg/dl
Mg	2.3 mg/dl
PO_4	1.7 mg/dl
Vitamin D 25(OH)D	34.58 ng/ml
PTH	348.3 pg/ml
ALP	1097 U/L

Next 10 days

Ca	8.1 mg/dl
Mg	2.1 mg/dl
PO_4	2.2 mg/dl
Vitamin D 25(OH)D	36.14 ng/ml
PTH	322.4 pg/ml

Next 4 years





05

Case presentation 2



Case 2 A 1 years old girl

Chief complaint :

Present illness

5 วันก่อนมาโรงพยาบาล เกร็งกระตุกแขนขารวมถึงลำตัวทั้ง 2 ข้าง 5 วินาที ตื่นรู้ตัวตลอด
หลังจากการตุก อาการปกติ ไม่ซึมลง ไม่มีไข้ ไม่มีผื่น ไม่มีแขนขาอ่อนแรง ไม่มีปัสสาวะอุจจาระ¹
 radix ขณะเป็นไม่ได้นอนหลับอยู่ ไม่มีไข้

1 วันก่อนมาโรงพยาบาล อาการดังกล่าวเป็นบ่อยมากขึ้น จึงมาโรงพยาบาล

Personal history

No underlying disease

History of UTI at age 7 month-old ผล ultrasound KUB normal

Feeding & nutrition กินแม่ผสานนมผสมถึงอายุ 4 เดือน หลังจากนั้น กินนมผสมสูตร 1 ตั้งแต่
อายุ 5 เดือน และกินอาหารบดตั้งแต่อายุ 6 เดือน ปัจจุบัน กินอาหาร 2 มื้อ; ข้าว $\frac{1}{2}$ ถึง 1 ทัพพี
ไก่ ปลา ไข่ 4-5 ช้อนโต๊ะ ไม่กินเนื้อแดงหรือเครื่องใน

Vaccination: ครบ EPI + Prevnar 2,4,6 month

คลอด Term AGA, no co-morbidity

Case 2 A 1 years old girl

- Development
 - GM เกาะ ยืน
 - FM ขึ้นเส้นยุ่งๆ ได้
 - PS ยิ้มหัวเราะ ตักข้าวกินเอง ได้แต่ยังหก
 - EL เริ่มมีคำโดดมีความหมาย
 - RL ทำตามคำสั่งง่ายๆ โดยซึ้งนิ่วบอก

Case 2 A 1 years old girl

Physical examination

- BT 36.8 c, PR 130min, RR 34/min, BP 86/55mmHg
- BW = 10.2 kg (P), Height = 74 cm (P)
- GA : Alert active
- HEENT : **Large forehead, alopecia**, fontanelle anterior 2x1 cm/ no bulging/depress, no separate suture no mass, no thyroid mass
- Heart : regular, normal S1S2, no murmur
- RS : clear, equal breath sound, no rachitic rosary, no Harrison's sulcus
- Abdomen :Soft, normoactive bowel sound, no mass, no hepatosplenomegaly
- Extremities : **Bowed legs, widening both wrist and ankle joint**, no redness or warm at all joint
- Others : WNL

Problem list

A 1 years old girl presented with bowlegs (Clinical rickets) and myoclonic seizure

* Film X-ray



Laboratory

Bone panels

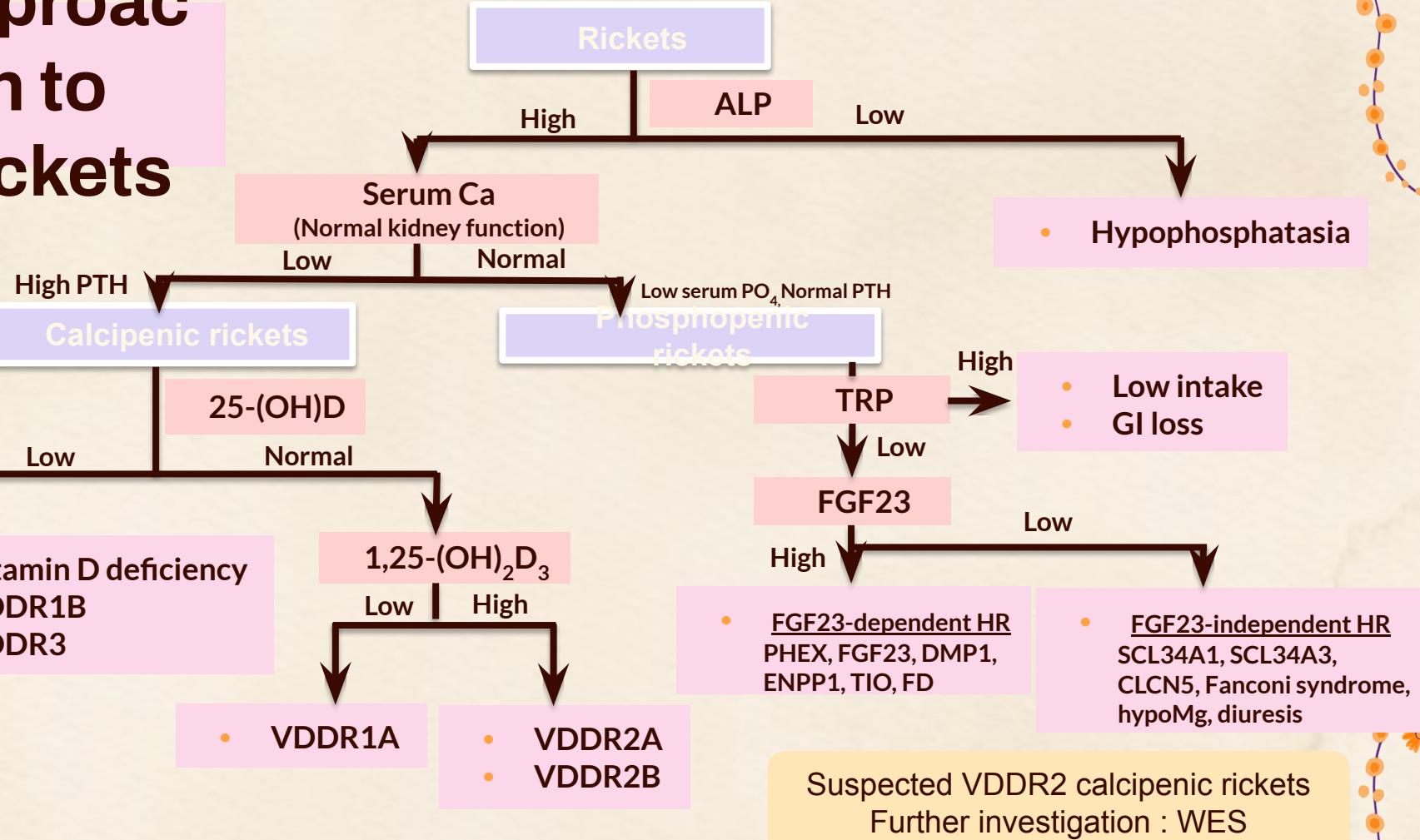
Ca	6.66 mg/dl
Mg	2.03 mg/dl
PO ₄	3.98 mg/dl
Vitamin D 25(OH)D	32.6 ng/ml
1,25(OH) ₂ D	> 600 pg/ml
PTH	565.5 pg/ml
ALP	1,677 U/L

Urine Cr	5.3 mg/dl
Urine Ca	0.5 mg/dl
Urine Ca/Cr	0.09

BUN	5.3 mg/dl
Cr	0.12 mg/dl



Approach to Rickets



Diagnosis and treatment

Calcipenic rickets : Vitamin D dependent rickets type 2

Management

10%Calcium Gluconate 10 ml IV x 5 doses then

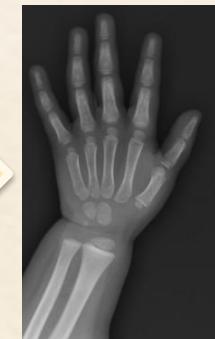
- Calcium carbonate (1.5gm) 2 tab po bid pc (244 mg/kg/day)
- Calcitriol (0.25mcg) 8 capsule po tid pc (612 mg/kg/day)
- Vitamin D₂ (20,000 IU) 1 cap po od weekly



Next 1 years
→



Next 1 years
→





05

Case presentation 3



Case 3 A 5 years 7 months old girl

Present illness

- อายุ 2 ปีเริ่มมีขาโก่งตัวเดี้ยตั้ง เวลาเดินแล้วปวดขา เข้ารับการตรวจที่ รพ. รัฐบาล แห่งหนึ่ง ส.ค. 59 ด้วยปัญหาตัวเดี้ยและมีความผิดปกติบริเวณข้อมีอะไรห้าบรวม ขาโก่ง 2 ข้าง ไม่มีร้อน ไม่มีไข้ ไม่มีผื่น ศีรษะบริเวณหน้าผากบวมโน สงสัยมารับการรักษาต่อ ที่ รพ.พระมงกุฎเกล้ารับมารักษาต่อ

Medication : Julie solution 4 ml x 3 feeds

Vitamin D3 (0.25)

Vitamin D2 (20,000 IU) 2 cap weekly

Family History: มารดา 31 ปี ไม่มีโรคประจำตัว, บิดาอายุ 32 ปี ไม่มีโรคประจำตัว

Paternal height = 160 cm, Maternal height = 130 cm [MPH = 151.5 cm]

Case 3 A 5 years 7 months old girl

Physical examination

- BT 36.5 c, PR 105min, RR 20/min, BP 91/62mmHg
- **BW 15.2 kg(<P3), Height 94 cm (<P3), U:L = 1.3 : 1**, Arm span = 98.5 cm
- GA : **Short stature**, alert active
- HEENT : **Frontal bossing**, fontanelle closed, no mass, no thyroid mass
- Heart : regular, normal S1S2, no murmur
- RS : clear, equal breath sound
- Abdomen : soft, normoactive bowel sound, no mass, no hepatosplenomegaly
- Extremities : **short stature with disproportion, bow legs, widening both wrist and ankle joints**, no redness or warm at all joint

Case 3 A 5 years 7 months old girl

Physical examination

- No spine deformity, no dimple, no mass
- Male genitalia, no ambiguous genitalia
- CNS : WNL

Problem list

A 5 years 7 months old girl presented with bowlegs
(Clinical rickets) and short stature with disproportionate(short limbs)

Film X-ray



Laboratory

Bone panels

Ca	9.3 mg/dl
Mg	2.0 mg/dl
PO ₄	2.1 mg/dl
ALP	589 U/L
Vitamin D 25(OH)D	24.02 ng/ml
iPTH	89.68 pg/ml

Urine Cr	31.7 mg/dl
Urine Ca	2 mg/dl
Urine Ca/Cr	0.06
Urine P	138.2 mg/dl
TRP	51%
TmP/GFR	1.07

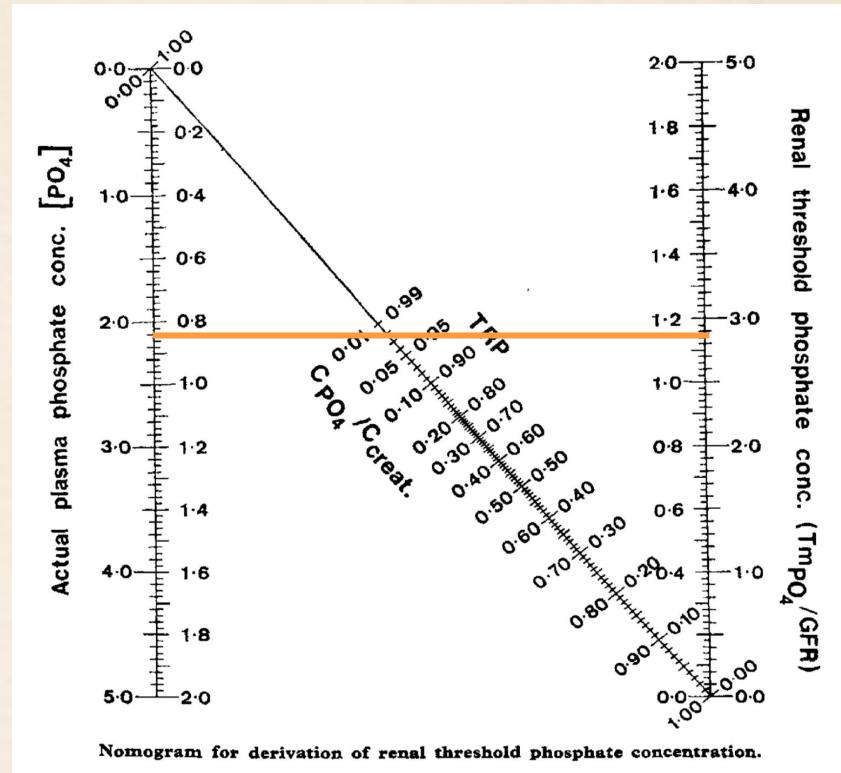
Na	139 mEq/dl
K	4.07 mEq/dl
Cl	107.3 mEq/dl
CO2	19.7 mEq/dl
BUN	12 mg/dl
Cr	0.24 mg/dl

Total protein	7.7 g/dl
Albumin	4.5 g/dl

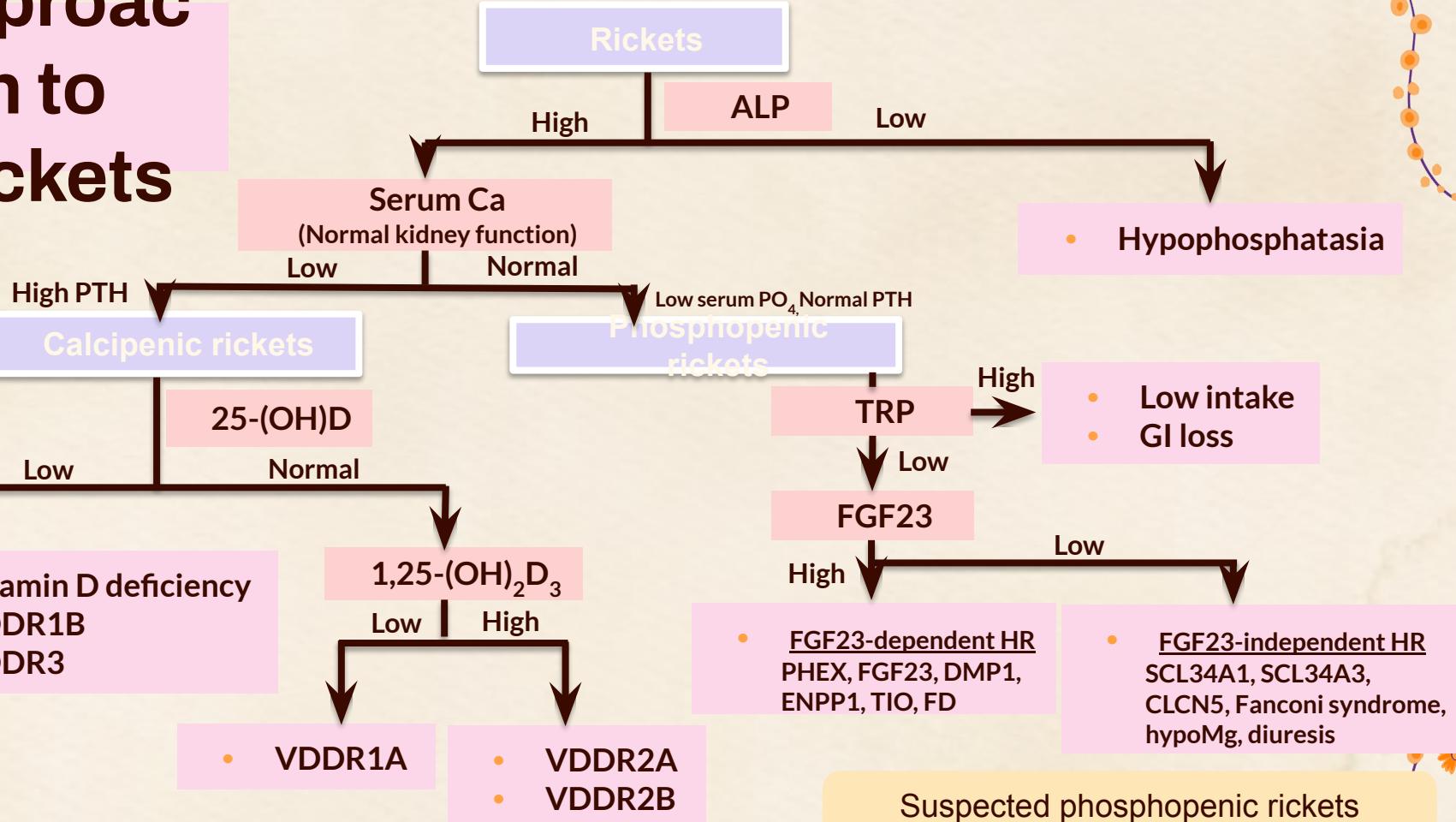
Laboratory

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Approach to Rickets



Suspected phosphopenic rickets
Further investigation : FGF23 & WES

Diagnosis and treatment

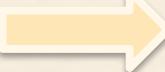
Phosphapenic rickets

Management

- Oral acidic phosphate solution (Joulie's solution) 5 ml po qid pc
- Vitamin D₃ (0.25) 1 tab po tid pc

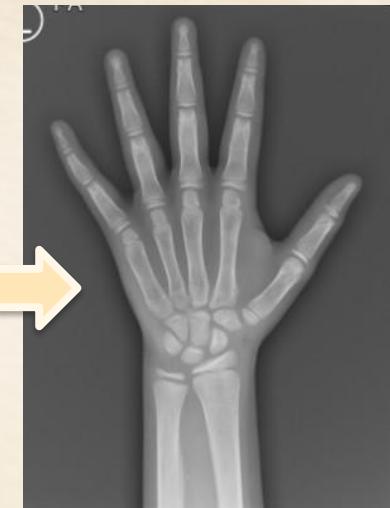
Ca	9.3mg/dl
Mg	2.0 mg/dl
PO ₄	2.1 mg/dl
Vitamin D 25(OH)D	24.02 ng/ml
PTH	89.68 pg/ml
ALP	569 U/L

Next 1 month



Ca	9.2 mg/dl
Mg	-
PO ₄	2.1 mg/dl
Vitamin D 25(OH)D	-
PTH	56.13 pg/ml
ALP	473 U/L

Follow up next 4 years medical treatment



Take home message

- Rickets is a common metabolic bone disorder.
- Rickets can classify in calcipenic rickets and phosphopenic rickets.
- Most common cause of rickets is vitamin D deficiency disorder.
- Adequate nutritional supplement is essential to prevent the rickets.
- Early detection and diagnosis not only prevent bone deformity, but also improve quality of life.

