

Clerking Example*

Ahmad is a 29 months old boy. He was referred to KKUH from another hospital. Informant is his mother.

P.C.: Up rolling of the eyes with abnormal upper limb movements for 1 minute

H.P.C:

Ahmad was in his usual state of health till early the morning of presentation at 7:00 am when his mother noticed that he was grunting while sleeping, with his eyes rolled upward and his arms stiffened and flexed. After one minute, the grunting and movements stopped and the patient vomited. Emergency medical services were called, and the patient was transported to the emergency department of another hospital. Vital signs, mental status, and findings on physical examination were reportedly normal. Laboratory evaluation was notable for a blood calcium level of 1.25 mmol/L (N. 2.12-2.62 mmol/L). IV calcium gluconate was administered. Ahmad was transferred to the emergency department of this hospital, arriving approximately 18 hours after a presumed seizure. No history of fever, rash, abnormal speech, movement, breathing, or bowel habits.

2 months ago, Ahmad had a febrile seizure associated with an acute viral illness. Two weeks prior to this presentation his mother started noticing "abnormal" gait that is getting worse.

Perinatal History: He was born at full term by normal vaginal delivery with no complications. Birth weight was 3.1 Kg.

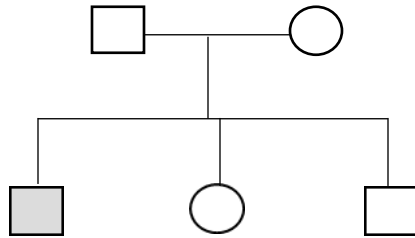
Nutrition: Ahmad was on exclusive breast milk till he was 5 months of age, when solid foods were gradually introduced into his diet and he remained on breast milk till 12 months of age. At that time Cow's milk-based formula was introduced, but he developed an allergic reaction in the form of generalized urticaria, lip swelling and shortness of breath and he stopped all dairy products since.

Vaccination: Up to date according to MOH schedule.

Development: He can run and jump, draw a line, speaks in 3-4 word sentences and understands 2 joined commands, and is dry by day and play with another child.

Past Medical History: He was diagnosed at 2 months of age to have atopic dermatitis. At 1 year of age he had an anaphylactic reaction to cow's milk and is strictly avoiding foods that contain dairy products.

Social history: Ahmad live with his parents and 2 siblings in north of Riyadh in a mid-size house. The family has good income. No smokers or pets at home.

Family History:

No parental consanguinity. Father has atopic dermatitis and mother is healthy. 4 years old sister is healthy and 7 years old brother has asthma.

Medications history: None

No known drug allergy.

Physical Examination:

Ahmad appeared well and was alert and interactive. Not pale or jaundiced. No edema. His ankles are widened bilaterally. There is mild bowing of the legs.

V/S: T. 36.9 C, Pulse 114/m, BP 95/63 mmHg, RR 24/m. O₂ sat 99% on RA

GP: Height 83.5 cm (3rd percentile for age), Weight 12.4 Kg (25th percentile for age), HC 49 cm (50th percentile for age)

CNS: Cranial nerves exam was normal. Muscle spasms occurred in the arms and legs with passive movement. Chvostek's sign (contraction of the facial muscles) was not elicited after tapping on the region of the facial nerve anterior to the external auditory canal; Trousseau's sign (carpal spasm) was not elicited after exertion of pressure from inflation of a sphygmomanometer on the upper arm. Muscle power, bulk, tone, and deep-tendon reflexes were normal. Sensation was grossly normal. Gait mild waddling.

HEENT: Normal head, TMs and throat. No dysmorphism.

CVS: Normal precordium and apex position. No thrill. Normal first and second heart sounds with no added sounds or murmurs.

Chest: Palpable costochondral junctions with depression on the lower chest. Resonant on percussion. Normal vesicular breath sounds with no added sounds.

Abdomen: Normal shape. Soft on superficial and deep palpation. No organomegaly. Normal bowel sounds.

Summary: 29 months old boy presented with a short episode of seizure. He was found to be hypocalcemic at the referring hospital. He is known to have cow's milk allergy. Examination revealed that his ankles are widened bilaterally. There is mild bowing of the legs. Muscle spasms were noted on passive movement of the extremities. His costochondral junctions are palpable with depression on the lower chest.

Differential Diagnosis:

1. Nutritional Rickets: Due to vitamin D deficiency, that may be secondary to poor intake or absorption. This is supported by hypocalcemia with seizure; the presence of typical physical signs of rickets including widened ankles with bilaterally bowing of the legs, palpable costochondral junctions (rachitic rosary) and depression on the lower chest (Harrison's sulcus); and the dairy dietary restriction. So far, nothing is against this possibility.
2. Genetic Rickets: Due to 25-OH or 1-alpha-OH vitamin D deficiency or deficiency of vitamin D receptor. They may present similar to nutritional rickets, but they are extremely rare and usually present earlier. In addition, our patient was healthy and there is no suggestive family history.
3. Other causes of hypocalcemia: These are generally less likely since they do not present with the same physical signs as our patient and most of them have special clinical features that are not present in our patient. Nevertheless, they should be kept in mind as remote possibilities. They can be grouped into 2 categories:
 - a. Hypocalcemia with low Parathyroid hormone (PTH):
 - i. Congenital: DiGeorge syndrome or familial.
 - ii. Acquired: HIV infection, autoimmune polyendocrinopathy syndrome 1 (APS1), iatrogenic.
 - b. Hypocalcemia with high PTH:
 - i. PTH end-organ resistance: Pseudohypoparathyroidism, hypomagnesemia, or mutations in PTH that affect its function.
 - ii. Ca loss: Due to renal disease, hyperphosphatemia, or osteoblastic metastasis.

Initial investigations:

- Ca: 1.4 mmol/l (L), ionized Ca 0.88 mmol/l (L)
- Ph: 1.37 mmol/l (borderline low)
- Mg: 0.78 mmol/l (N)
- Alk phos: 673 U/l (H)
- Urea, creatinine, electrolytes, glucose: normal
- CBC, diff: normal
- LFT: normal
- PTH, 25-OH Vit D, and 1,25-OH Vit D levels were ordered
- CXR and Knees x-ray were ordered.

Comments:

These initial results are consistent with nutritional rickets. Other differential diagnoses should still be kept in mind. However, some diagnoses can be readily ruled out such as hypomagnesemia, hyperphosphatemia and renal insufficiency.

Plan:

- Admit the patient.
- Continue Calcium gluconate IV 8.8 mmol/day over 24 hours until ionized Ca >1 mmol/l.
- Start Vitamin D3 at 50,000 IU on day 1 then 1000 IU per day

Progress Notes

Day 1

Subjective:

Ahmad's mother report that he had been fine. No seizures or other symptoms. Feeding well. Avoiding all dairy products.

Objective:

V/S within normal.

CNS and MSS exam: resolution of muscle spasm on passive movement of the limbs. No other abnormality.

- PTH: 177 pg/ml (N. 10-60)
- 25-OH Vit D: 4 ng/ml (N. 20-80)
- 1,25-OH Vit D: 16 pg/ml (N. 24-86)
- **Imaging studies:** Anteroposterior radiograph of the knees: The metaphyses throughout both knees show flaring and fraying and cupping. There is beaking of the medial aspect of the proximal tibial metaphysis. The radiolucent physes are widened. An anteroposterior radiograph of the chest shows subtle flaring and cupping of the right proximal humeral metaphysis and mild flaring of the anterior ends of the ribs.

Assessment:

The patient is clinically better and is in good condition. PTH is high which rules out causes of hypocalcemia with low PTH. 25-OH and 1,25-OH Vit D are both low. These results together with the results of the imaging studies are consistent with the diagnosis of nutritional rickets (vit D deficiency). The diagnosis of cow's milk protein allergy needs to be confirmed by testing for specific IgE.

Plan:

- Continue IV calcium and change the dose of vit D to 1000 IU per day starting from tomorrow.
- RAST (radioallergosorbant test) for cow's milk proteins.

Day 2

Subjective:

Ahmad has been fine. No new symptoms.

Objectives:

V/S: within normal

CNS and MSS Exam: unremarkable.

Assessment:

The patient continues to do well. It takes few days for Ca to reach a satisfactory level on IV supplementation before switching to oral.

Plan:

- Continue IV Calcium gluconate at 8.8 mmol/day and Vit D at 1000 IU/day.
- Will repeat Ca, ionized Ca, Ph, AlkPh, PTH, 25-OH and 1,25-OH vit D tomorrow am.

Day 3

Subjective:

No new complaints. Feeding well.

Objective:

V/S: within normal

General examination is unremarkable.

RAST for Cow's milk protein: 45 kU/L (negative is <0.35 kU/L)

Blood was obtained for Ca, ionized Ca, Ph, AlkPh, PTH, 25-OH and 1,25-OH vit D.

Assessment:

Stable and well. RAST is high which is consistent with cow's milk protein allergy.

Plan:

- Continue IV Calcium gluconate at 8.8 mmol/day and Vit D at 1000 IU/day.
- Will start extensively hydrolysed milk formula; peptamin junior as a dietary supplement 1-2 cans (250 mls/can) per day. This was discussed fully with the patient's mother.

Day 4:

Subjective:

No new complaints. Tolerating peptamine jr very well.

Objective:

V/S: within normal

General examination is unremarkable.

Ca: 2.25 mmol/L

Ionized ca: 1.2 mmol/L

Phosphorus: 1.47 mmol/L

AlkPh: 466 U/L

PTH: 135 pg/ml

25-OH vit D: 10 ng/ml

1,25-OH vit D: 316 pg/ml

Assessment:

The patient continues to do clinically well. Ca, ionized Ca, Ph have normalized. AlkPh and PTH are still high, but less than the initial readings. Also, 25-OH vit D is still low, but more than initial reading, while 1,25-HO vit D became high. These results indicate good response to treatment and confirms the diagnosis of nutritional rickets due to vit D deficiency. 1,25-OH vit D became high after being low. It has a short half-life and its level is variable as it depends on multiple factors including vit D3, Ca, Ph, and PTH levels. Therefore, we do not depend on it in determining vit D intake and activity. It is useful in ruling out genetic rickets in this context.

Plan:

- Stop IV Ca and start oral elemental Ca at a dose of 300 mg TID.
- Continue Vit D 1000 IU/day
- Discharge the patient on the above medications with follow up in the out-patient clinic after 2 weeks.
- Give the patient sufficient supply of peptamine Jr for the next 3 months to be re-prescribed as an out-patient.
- Repeat Ca, ionized Ca, Ph, AlkPh, PTH, and 25-OH 2 days before clinic visit.

*This material was prepared by Prof. Abdullah Alangari. Department of Pediatrics, KSU. It was modified from Case 39-2020. N Engl J Med. 383;25:2462-2470