

# Acetazolamide treatment in a patient with pseudohypoparathyroidism with venous calcification

P1-19



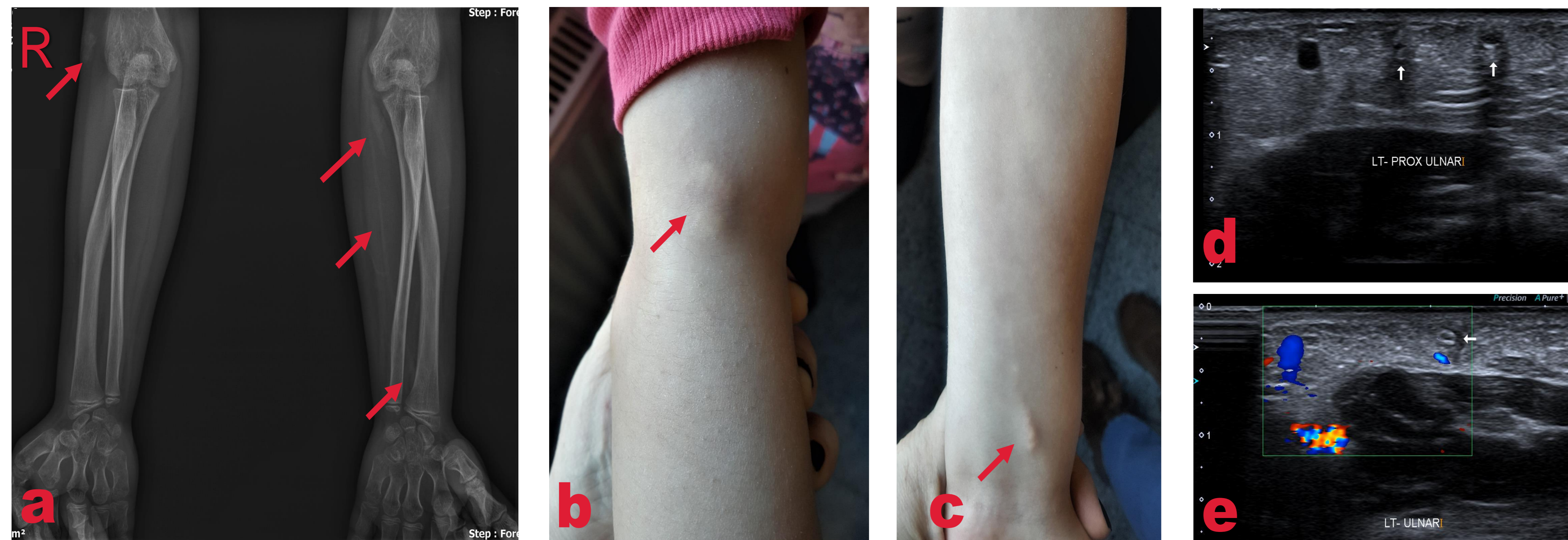
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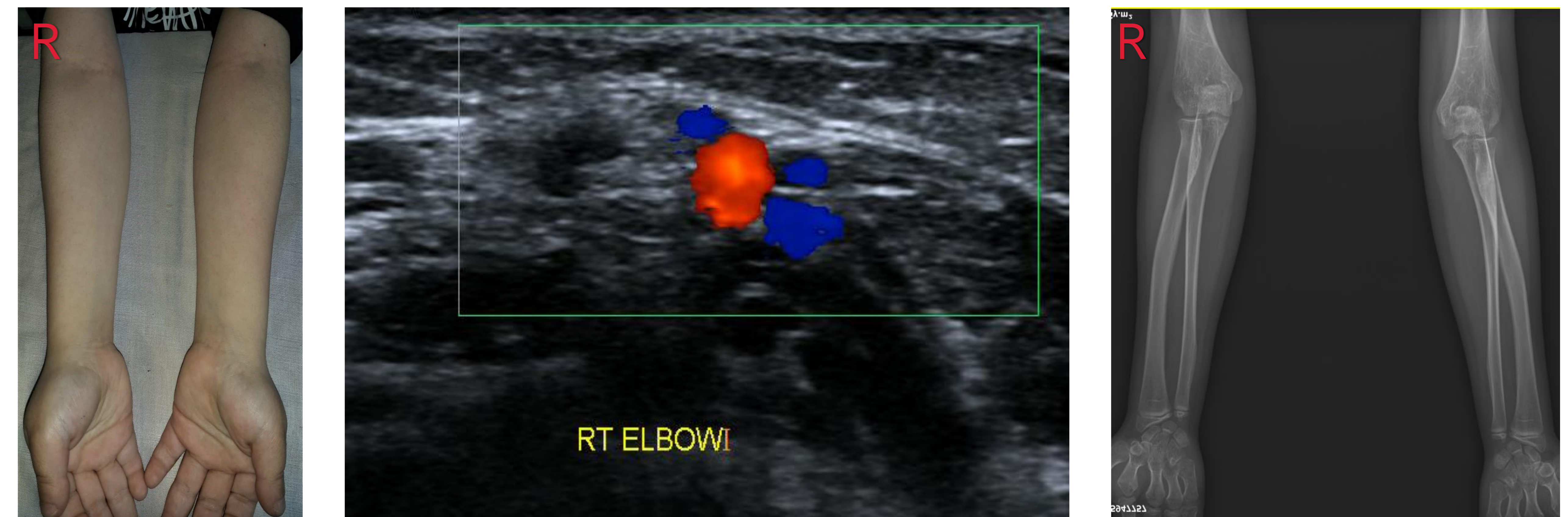
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- A 11<sup>7/12</sup> years old female patient presented with short stature, fatigue and exercise-induced carpopedal spasms.
- Diagnosis of PHP-1A was made based on hypocalcemia (5.4 mg/dl, N:8,8-10,8 mg/dL), hyperphosphatemia (2,2 mg/dl, N:3,3-5,4 mg/dL), elevated serum PTH (460 ng/L, N:15-65 ng/L) and AHO features and calcifications in the basal ganglia.
- Her height was 127.6 cm (-3.37 SDS)
- Weight was 30 kg (-1.81 SDS)
- The patient was treated with IV and oral calcium and calcitriol.
- During intravenous calcium therapy, extravasation of calcium occurred in left forearm and a small hematoma developed in the right antecubital area after phlebotomy.
- Three weeks later on follow-up examination, a palpable rigidity was detected along the extravasated vascular path on the left arm and a 2x1 cm solid nodule was palpable in the location of hematoma in the right antecubital area. X-ray of the arms demonstrated calcification in these areas.
- Venous doppler ultrasound examination was consistent with thrombophlebitis and intravascular calcifications in both arms.

- The patient was treated with antibiotics and low molecular weight heparin (LMWH) for thrombophlebitis.
- Acetazolamide (25 mg/kg/day) was started for calcifications which created mild metabolic acidosis (pH:7.30-7.35).
- LMWH was discontinued on 6 months and acetazolamide treatment was discontinued at 8 months of treatment when calcifications in both arms disappeared completely.
- No adverse effects were observed during treatment.
- Maternally inherited p.Arg999Thrfs\*47 heterozygous mutation was detected in the GNAS gene.



**Figure 1.** a. Subcutaneous calcification in right antecubital area and intravascular calcifications in left arm with X-ray, b. Subcutaneous calcification in right antecubital area, c. intravascular calcifications in left arm, d,e. Intravascular calcifications and venous thrombosis with doppler ultrasonography



**Figure 2** X-ray and doppler ultrasonography examinations after LMWH and acetazolamide treatment

## Learning points

- Acetazolamide, a carbonic anhydrase inhibitor, causes metabolic acidosis by increasing bicarbonate excretion from the proximal renal tubule thereby diminishing precipitation of calcium and phosphorus.
- Acetazolamide had been used for soft tissue calcifications in tumoral calcinosis cases, however not used in PHP-1A and vascular calcification previously.
- This case represents the first successful use of Acetazolamide for soft tissue and vascular calcifications in PHP and provides new insight for treatment of ectopic and vascular calcifications in PHP.

