Severe Hypophosphatemic Rickets Due to Tumor-Induced Osteomalacia

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Aim:

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- ✓ Hypophosphatemic rickets is usually due to genetic causes but in rare cases it can develop secondary to MAS, Fanconi syndrome, or oncogenic causes.
- Induced ✓ Oncogenic osteomalacia (Tumor Osteomalacia - TIO) is a rare acquired paraneoplastic syndrome
- ✓ It develops as a result of excessive phosphate loss from renal tubules by FGF23 released from tumour tissue.

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Ca (mg/dL)	8.7
P (mg/dL)	0.6
ALP (U/L)	615
PTH (pg/mL)	119.1
25(OH) vitD (ng/mL)	13.3
1-25(OH) vitD (ng/L)	36
TRP (%)	86
TmP/GFR (mg/dL)	0.51
Intact FGF23 (pg/mL)	1329



Table: The laboratory findings at diagnosis

Figure 1: The cyst in the right tibia in 2018

Case:

- ✓ A 16-year-old male boy who had been completely healthy until 13 years of age underwent surgery for leg pain and limping that began 3 years ago and was found to have a cyst in the proximal right tibia (Figure 1). He developed severe deformities due to recurrent fractures after surgery and applied to us for further examination (Figure 2).
- painful deformities, tachypnea, tachycardia on physical examination. The results of the laboratory tests are summarized in Table 1.

- ✓ Due to severe hypophosphatemia;
- oral phosphorus (up to 55 mg/kg/d),
- calcitriol (up to 60 ng/kg/d)
- calcidiol (1200 U/day)
- ✓ MRI of the right knee was taken to determine the possible location of the tumour (Figure 3). GA -68-DOTATATE-PET was performed due to the suspicious appearance of the mass in right knee and metastasis.
- ✓ An 8.5x7.5 cm heterogeneous mass with minimal involvement was detected.
- ✓ In biopsy, a phosphaturic mesenchymal tumour with osteoblastoma-like appearance was detected. Surgical resection was planned.



Figure 2: The patient's appearance at the presentation

Figure 3: The mass on MRI of the right knee

Conclusion:

- ✓ TIO is usually diagnosed late and difficult to localize because of their slow progression and small size.
- ✓ Extensive resection of the tumour is very critical for both the chances of cure and prevention of recurrence.
- ✓ This case presented due to its severity and large mass.

