Hypophosphataemic Rickets due to lung Teratoma: a case report

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Introduction

Teratoma is a rare tumor that is derived from germ cells. This tumor is composed of one or more than one types of tissue, such as sebaceous glands, hair, and teeth. Teratoma is commonly benign and presents in gonadal organs such as the testicles and ovaries, mostly. But, this type of tumor can be seen in other organs especially in the thoracic cavity (1, 2).

Thoracic teratoma usually occurs in the mediastinum. In very rare cases, teratoma occurs in the lungs (3). Although pulmonary teratoma is very rare it was observed that this type of teratoma involves the upper lobes. The causes of the involvement are not defined until now (4).

Pulmonary teratoma may be presented with cough, chest pain, or hemoptysis (3, 4). Based on our knowledge, all studies about intrapulmonary teratoma are case reports. In this study, we presented a 57-year-old man who was a known case of hypophosphataemic rickets for 16 years and was finally diagnosed with pulmonary teratoma.

Case presentation

A 57-year-old man who was diagnosed with hypophosphataemic rickets about 16 years ago was referred to our clinic for more evaluations. About 16 years ago (2008), the patient was referred to another center due to mechanical back pain that was radiated to the hamstrings muscles. The patient woke up due to back pain at night. The pain was increasing in the lower extremities for months and the patient was unable to walk alone and he had a waddling gait. There was no history of the night sweating, weight loss, and using unpasteurized dairy. The laboratory studies were as follows: hemoglobin of 12.9 mg/dl, alkaline phosphatase (ALP) 964 U/L (normal:64-306 U/L), a calcium level of 10.1 mg/dl (normal:8.5-10.5 mg/dl), phosphor level 2.1 mg/dl (normal: 3-4.5 mg/dl), magnesium level 2.35 mg/dl (normal: 1.7-2.2 mg/dl), 25-hydroxy vitamin D 68 nmol/L (sufficient above 30 nmol/L), normal liver and kidney function tests, normal thyroid stimulating hormone (TSH) level, normal parathyroid hormone (PTH) level, and erythrocyte sedimentation rate (ESR) was 3 mm/hr. Wright, 2ME, 24-hour urine calcium, and phosphor were all within normal units. Electromyography (EMG) test and a Nerve Conduction Velocity (NCV) test were done and myopathy was found in EMG-NCV. Bone marrow aspiration was done and the findings were compatible with osteomalacia. Hypophosphataemc Rickets and myopathy were diagnosed for him based on clinical, laboratory, and EMG-NCV tests. Sulfasalazine and indomethacin were prescribed for him in that center for 6 months but the patient’s condition did not differ and after this time, the patient was referred to us.

We admitted him and started the evaluations. Findings of physical evaluations were as follows: proximal lower extremities muscle weakness (3/5), normal deep tendon reflex (DTR), positive Patrick test in left, vertebral tenderness, waddling gait. Rheumatologic tests including rheumatoid factor, anti-citrullinated peptide antibody, and anti-nuclear antibodies were within normal limits. A whole body CT scan was done and all were normal. Articular osteopenia and pathologic fractures in L2 and L3 vertebrae were found in X-ray radiography (Figure 1). A Celiac disease work-up was performed for him and anti-tissue transglutaminase
antibody and anti-endomysial antibody were within normal limits. Upper gastrointestinal endoscopy was done and it was normal. EMG-NCV test was performed again in our center and it was normal and there was no myopathy pattern. The patient was discharged with calcium carbonate, phosphate sandoz, and calcitriol capsules.

After two years (2010), the patient discontinued his medication spontaneously and he was referred to us with exacerbation of symptoms including muscle weakness of upper and lower extremities (4/5), atrophy of quadriceps muscles, and waddling gait. A decrease in the height of L1 and L2 was found in radiography. The patient was discharged with previous medications with higher doses.

The patient was referred for yearly follow-up in 2011 and he was admitted to assess tumor-induced osteomalacia but there were no radiologic findings about tumor occurrence and only the presentations of osteomalacia were found including a looser zone in the sacroiliac, cyst in the head of the humerus bone, evidence of previous fractures vertebral bones, gibbus deformity, and decreased vertebral height (L1 and L2) (Figure 1 & 2). An electromyogram test was done and it was normal. The Z-score of the forearm was -2.9. Brain, thorax, abdomen, pelvic, and paranasal sinuses CT scans were normal. The patient was discharged with previous medications.

The patient was followed up yearly. In the follow-up of 2018, he had lateral side pain in his right thigh. An X-ray radiography was requested and a right sub-trochanteric femoral fracture was observed (Figure 3). Laboratory study and BMD were requested. The Z-score of the spine was -2.3 and the Z-score of the neck of the femur was -2.8. Calcium was 9.9 mg/dl (normal: 8.5-10.5 mg/dl) and phosphor was 2 mg/dl (normal: 3-4.5 mg/dl). Parathyroid hormone was within normal limits. An orthopedic consult was requested and surgery, using a walking cane, and starting Teriparatide were recommended.

After one year he was referred for follow-up and based on laboratory findings (Calcium= 10.6 mg/dl, phosphor= 1.9 mg/dl, 25-hydroxy vitamin D= 43 nmol/L, PTH= 92 with normal range of up to 55), nuclear scan was requested and left parathyroid adenoma was diagnosed. PTH was evaluated in all previous work-ups and it was normal until the recent work-up. A surgery was done and the upper and lower parathyroid glands of the left side were resected. The patient was discharged with rocaltral, oral calcium carbonate, and phosphate sandoz, Teriparatide subcutaneously daily for three months, and then, Denosumab every 6 months.

Annual follow-ups were performed for him and a PET-CT scan of his lungs was done a large tumoral lesion was seen in the lower lobe of the left lung with increased metabolic activity, foci of calcifications, and fat densities that were compatible with teratoma (Figure 4). The patient underwent lung surgery and the lesion was resected. The pathology of the lesion was teratoma. The patient mentioned his symptoms (bone and muscle pains) decreased significantly and his medications were decreased. He was satisfied with the improvement in his general condition.

Discussion

Intrapulmonary teratoma (IPT) is a rare presentation of teratoma (3). Teratomas are classified as solid or cystic and mature or immature tumors. In this type of tumor, levels of serum tumor markers are negative or low and that are not diagnostic (5, 6). Teratoma is a type of germ cell tumor that can come from three layers of germ cells including endoderm, ectoderm, and mesoderm (7). Teratoma is common in the genital system and it is a common tumor of ovaries with the prevalence of 20%. The incidence rate of teratoma in the ovarian system was about 1.2-14.2 in 100,000 cases, in a study that was conducted on 460,000 women (8). Although, ovarian teratoma is not rare, but IPT is very rare and only there are some case reports about IPT. It is mentioned that IPT occurs mostly in the left upper lobe; however, in our case, it is presented in the left lower lobe (3, 9-11).

Teratoma is commonly benign and it grows slowly. This tumor can be present in any age but it was mentioned that IPT usually occurs in the first and second decades of life. Our patient was in the fifth decade of his life. So, our case showed that was an unusual case of IPT. IPT in our case had very slow growth and that was not visible in annual CT scanning. Malignant mediastinal tumors of germ cells are more prevalent in females.
but IPT incidence doesn’t differ between men and women. The IPT diagnosis is hard because it has several differential diagnoses (such as thyroid, or parathyroid adenoma, thymic carcinoma, and thymoma) and also it is slow growing with non-specific manifestation or test. In our case, the patient had parathyroid adenoma and the involved side of parathyroid was resected (12, 13). Our patient was diagnosed as hypophosphatemic rickets due to the low levels of serum phosphate and laboratory findings that could show this diagnosis. Hypophosphatemic rickets is a bone mineralization disorder that occurs due to acquired or inherited defects in the renal handling of phosphorus (14). This disorder causes osteomalacia but physicians should be aware about different causes that can mimic hypophosphatemic rickets such as tumors (15). Like our patient, firstly his disease was diagnosed as hypophosphatemic rickets but finally it was found that his osteomalacia was occurred due to tumor.

IPT’s manifestations can be different based on the location, the size of mass, and tissue type of teratoma. IPT may be complicated with pneumonia, abscess formation, and bronchiectasis but our patient didn’t have any pulmonary manifestation and his manifestations only were musculoskeletal (16, 17). Lack of the pulmonary manifestation and presentation were the main important point in our patient because he had no manifestation of lung involvement or mass presentation in radiologic assessments.

CT scan has a main role in diagnosis of IPT and can show the location of mass and also its invasion to surrounding structures such as pleura or chest wall (17). Teratoma in CT scan manifests as a well-defined mass with internal heterogeneous soft tissue, fluid, fat, or calcifications (16). Our case underwent CT scan but the lug CT scan was normal in until last follow up. PET-CT was done for him in the last follow up and the IPT was found. Although in the last follow up we observed a big lesion in the PET-CT scan but it could be seen in the CT scan, also. PET-CT scan has higher sensitivity and specificity in diagnosis of lung tumors. In some studies it was mentioned that PET-CT scan can diagnose mediastinal tumors very better than CT scan (18-20).

**Conclusion**

Intrapulmonary teratoma is a rare type of teratoma that can be presented in any site of the lungs. It can manifest as osteomalacia or hypophosphatemic rickets that don’t respond to common treatments such as calcium and phosphorus. As we found in our case, radiologic assessment is the main method for diagnosis of this disease and PET-CT is a good choice. If a patient with undiagnosed cause of hypophosphatemic rickets is referred to a physician, PET-CT should be performed as one of the good diagnostic methods.

**Ethics approval and consent to participate**

Approval was not needed by the local Clinical Research Ethics Committee for case reports.

**Consent for publication**

Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

**Conflict of interests**

The authors declare that they have no competing interests.

**Data availability statement**

Data is available if requested

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**Authors’ contributions**
FF conceived the idea to report the case. NB was responsible for data collection. FF and ME drafted the manuscript. NB commented on the manuscript. All authors read and approved the final manuscript.

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