

Obstructive Shock Due to a Mediastinal Mass in a Young Male with Klinefelter Syndrome

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Abstract

A 21-year-old male presented with precordial chest pain, shortness of breath, and weight loss. A large anterior mediastinal mass was found on radiographic studies, associated with a left pleural effusion, and a pericardial effusion with tamponade physiology. Pericardiocentesis was performed that led to the compression of the right pulmonary artery by the massive tumor resulting in acute cor pulmonale and refractory shock state. The patient consequently underwent an emergent near-complete tumor resection that resolved the obstructive shock. Histopathology identified a mixed germ cell tumor with components of teratoma and an aggressive yolk sac tumor. Residual tumor was treated with chemotherapy.

Keywords

Mediastinal tumor; Klinefelter syndrome; Germ cell tumor.

Back Ground

Primary anterior mediastinal neoplasms comprise a diverse group of tumors and account for 50% of all mediastinal masses. A variety of mediastinal germ cell tumors — teratomas, seminomas, nonseminomatous germ cell tumors — account for 10% to 15% of mediastinal tumors in adults, and 25%, in children [1]. A mass in the anterior mediastinal compartment is more likely to be malignant than those found in the other compartments [2]. Mixed tumors containing a combination of mature teratoma and other germ-cell elements are considered as nonseminomatous germ-cell-tumors (GCTs). Mixed tumors may contain more than one cell type such as yolk sac tumor, choriocarcinoma, or embryonal carcinoma. In contrast to gonadal nonseminomatous GCTs, mediastinal nonseminomatous GCTs commonly include yolk sac tumors (either pure or mixed with other types) [3,4], while they are infrequently associated with embryonal carcinoma.

We hereby, discuss a rare presentation of a massive malignant non-seminomatous germ cell tumor in a young male with Klinefelter syndrome.

Case Presentation

A 21-year-old man, with a history of Klinefelter syndrome, presented to his primary care physician for left sided chest discomfort ongoing for a week. He noticed a dull aching precordial chest pain after trivial blunt trauma that failed to resolve with over-the-counter analgesics. He had also noticed fatigue, exertional shortness of breath and a 40 pound weight loss over the past 6 weeks prior to presentation. Past medical history was notable for Klinefelter syndrome and asthma for which he used testosterone pump, beclomethasone inhaler respectively. His father was diagnosed with a pheochromocytoma at age 15 and two of his maternal uncles had hematologic malignancies. He denied smoking, alcohol or recreational drug use. A chest radiograph showed an enlarged cardiac silhouette with a left pleural effusion (figure 1). A computed tomogram (CT) of the chest revealed a large heterogeneously enhancing anterior mass measuring 18 x 11 x 19 cm, compressing left main bronchus, and displacing other mediastinal structures, as well as a pericardial effusion and a large left pleural effusion with compressive atelectasis and enlarged lymph nodes (figure 2, 3, 4, 5). The patient was referred to our hospital for further management.

On examination, he was alert, oriented, and in no acute distress. Vital signs on presentation were as follows: temperature 98.7° F, pulse 112 beats per minute, respiratory rate 18 breaths per minute, blood pressure 112/58 mm Hg, oxygen saturation 97% on room air. Pulsus paradoxus was noted. He had jugular venous distension. Breath sounds were diminished over his left hemithorax. Cardiac auscultation revealed muffled heart sounds without any appreciable murmur or friction rub. Marked atrophy of both testicles was noted. The rest of the physical exam was unremarkable.

Investigations

Complete blood count, serum electrolytes, creatinine, and coagulation profile were normal. Hepatic function panel revealed; alanine transaminase 836 IU/L (normal range 7-56 IU/L), aspartate transaminase 832 IU/L (normal range 5-40 IU/L), total bilirubin 6.1 mg/dL (normal range 0.3-1.9 mg/dL), direct bilirubin 2.8 mg/dL (normal range 0-0.3 mg/dL), and albumin 3.3 g/dL (normal range 3.4-5.4 g/dL). Serum lactate dehydrogenase was mildly elevated at 247 U/L (140-280 U/L).

Treatment

A bedside echocardiogram was performed in the emergency department that showed a large pericardial effusion with features of early tamponade (Figure 6). Within a few hours of presentation to the emergency department the patient was found to be hypotensive and tachycardic. Electrocardiogram revealed low voltage (figure 7). Serum alpha fetoprotein (AFP) levels were 29,464 ng/mL (normal <10 ng/mL). An urgent pericardiocentesis was performed in the cardiac catheterization laboratory and about 1400 mL of straw-colored pericardial fluid was drained. A 14 Fr chest tube was placed that drained 800 mL of straw colored pleural fluid from the left side. Over next several hours, the patient continued to be hypotensive and in shock state that was refractory to vasopressors. A chest CT scan was repeated that showed marked compression and narrowing of right main pulmonary artery by the large tumor with acute right heart strain. Compression of the trachea and proximal right and left bronchi were also noted. After multi-disciplinary team consultation, the patient underwent surgical biopsy on the next day of presentation. Histopathological examination of the frozen section suggested a benign teratoma.

One day after the biopsy, patient was subjected to surgical resection. It was noted during the surgery that his blood pressure improved in the left lateral decubitus position. To relieve the compressive effect of the tumor on the vessels and cardiac chambers, he was maintained in the left lateral decubitus position through his operative and post-operative course. Intra-operatively, the tumor was found to be well encapsulated and easily freed from the surrounding mediastinal structures (Figure 8). The tumor was completely excised except for a small portion that remained behind the innominate vein where it was adherent and inseparable. Serum AFP decreased to 4607 ng/mL postoperatively. Final histopathology showed a malignant germ cell tumor with areas of teratoma and an aggressive yolk sac tumor. Pathology reports and elevated AFP thus confirmed a malignant mediastinal non-seminomatous GCTs. Metastatic workup, including CT of the brain, abdomen and pelvis were all negative. Shock state resolved one day after the surgical resection of the tumor that allowed our patient to be weaned off vasopressor support.

Outcome & Follow up

One week following the tumor resection, the patient developed pulmonary embolism. His course was also complicated with the formation of mediastinal phlegmon 10 days after the initial resection that required surgical drainage and broad spectrum antibiotics. After recovery from post-operative complications, he was started on systemic chemotherapy with bleomycin, etoposide and cisplatin for residual tumor. Serum AFP on follow up at 6 months was 82.9 ng/mL and he continued to be symptom free.

Discussion

Anterior mediastinal nonseminomatous GCTs are aggressive tumors that are often metastatic at presentation. Mediastinal nonseminomatous GCTs are rare, are diagnosed late [3,4] and have a worse prognosis with a five year overall survival of less than fifty percent [5,6]. They are more common in men and usually are diagnosed in individuals younger than 40 years [3,4]. Independent negative prognostic factors include age ≥ 24 years and size of primary mediastinal tumor ≥ 19 cm. Median overall survival (OS) and mean 2-year OS is 15 months and 40% respectively for patients with poor prognosis [7]. Most patients present with non-specific symptoms such as fever, weight loss, chest pain, or dyspnea. Few patients have symptoms of superior vena cava syndrome due to the compression of mediastinal structures. Mediastinal tumors that produce bioactive substances may be associated with gynecomastia and precocious puberty in children.

Whereas a higher tumor risk in this condition is controversial, Klinefelter syndrome has a relative risk of 66.7% for malignant mediastinal germ cell tumor. Approximately 20% of nonseminomatous malignant mediastinal germ cell tumors are associated with Klinefelter syndrome — 50 times the expected frequency [8].

Diagnosis may be confused with thymoma, thymic carcinoma or Hodgkin's disease. A careful clinical history and serum tumor marker level such as AFP may be helpful in making accurate diagnosis. Takeda S et al. reported elevated serum tumor markers in approximately 90% of their patients with nonseminomatous GCT [9]. Liu Yet al. noted in their case series, all 34 patients with non-seminomatous GCT diagnosed on histology had an elevated β -Human Chorionic Gonadotrophin (β -hCG) and/or AFP [10]. An elevated AFP, imaging findings, and a high index of suspicion can lead through the differential diagnoses for these tumors.

A multimodality approach combining chemotherapy with surgery is generally warranted. In our patient since the initial impression on histology was that of a benign tumor and due to the obstructive shock, surgery was performed first followed by chemotherapy. Currently the standard chemotherapy regimen for primary mediastinal malignant germ cell tumors is with bleomycin, etoposide, and cisplatin [11]. Alternative regimens that include carboplatin or paclitaxel may be associated with better outcome in this patient subgroup [7]. These tumors are relatively radio-resistant. In patients with large mediastinal non-seminomatous GCT, aggressive cisplatin-based chemotherapy followed by resection of the residual tumor has been reported as the best approach to improve the survival of these patients [10].

Serial monitoring of serum AFP and β -hCG is helpful in monitoring the response to therapy and detecting early recurrence.

Figures

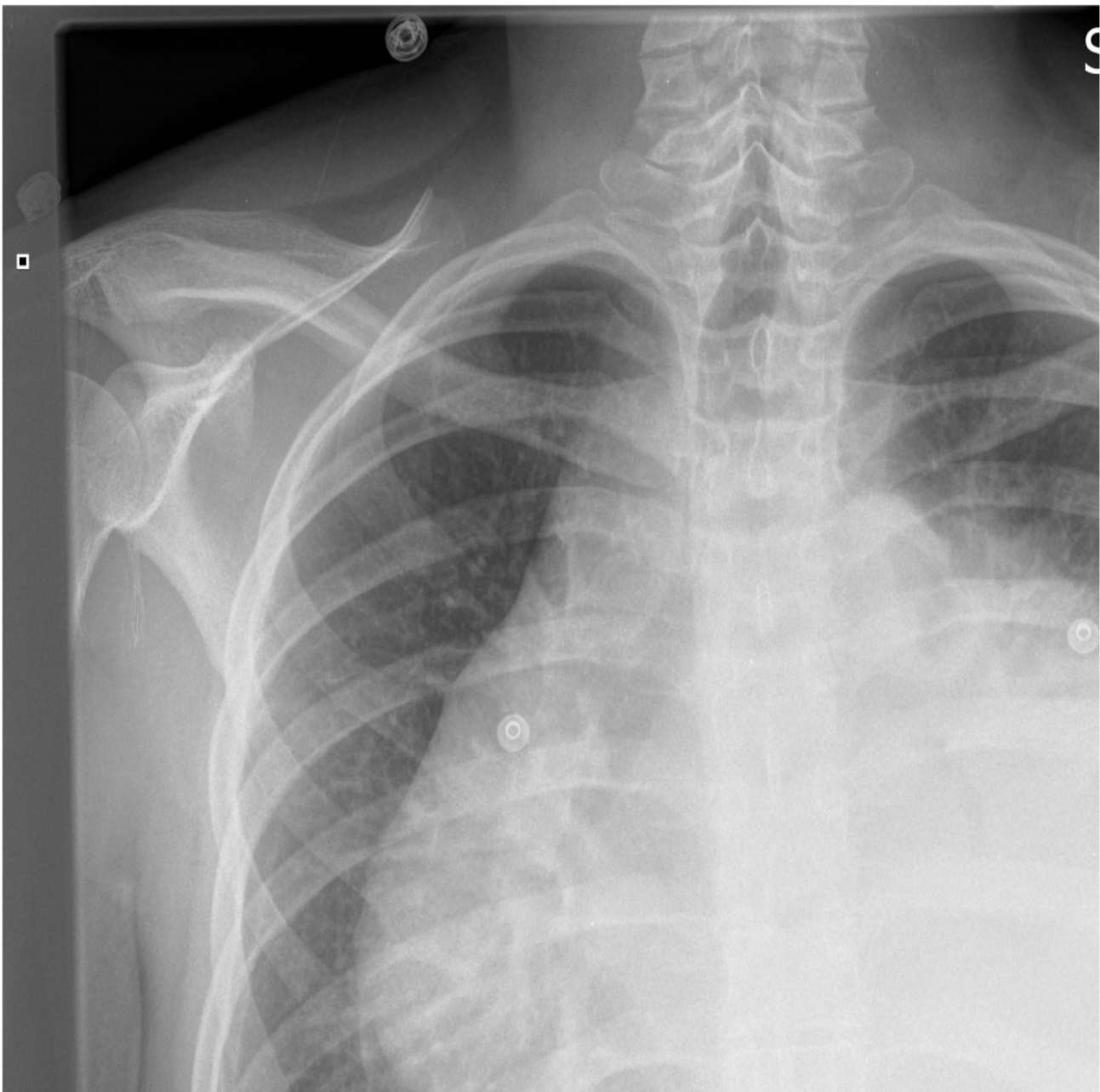


Figure 1: Enlarged cardiac silhouette with left pleural effusion

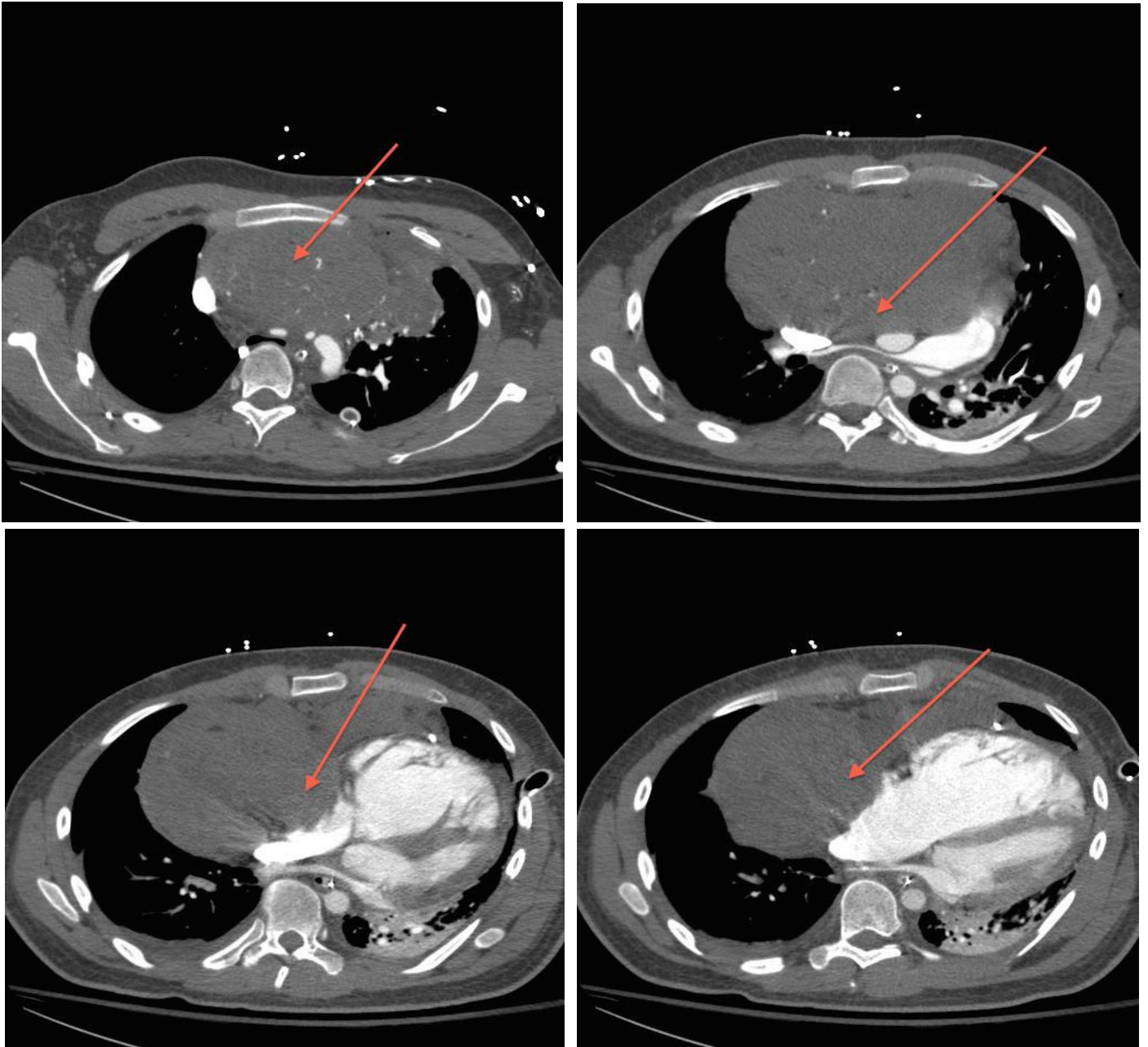


Figure 2,3,4,5: A large heterogenous mass (arrow) compressing mediastinal structures

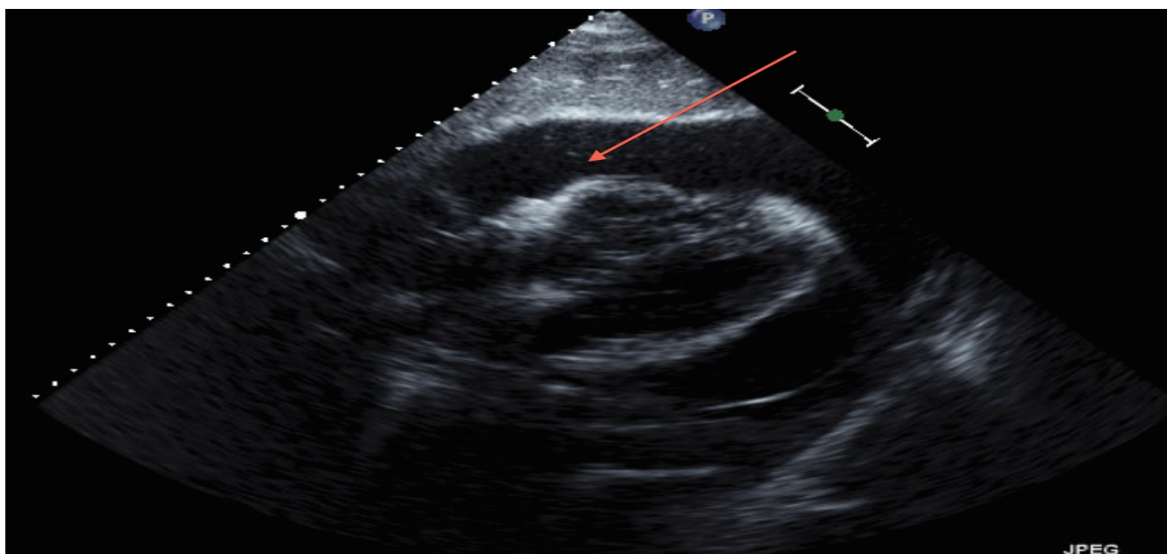


Figure 6: Bedside echocardiogram showing a large pericardial effusion (arrow)

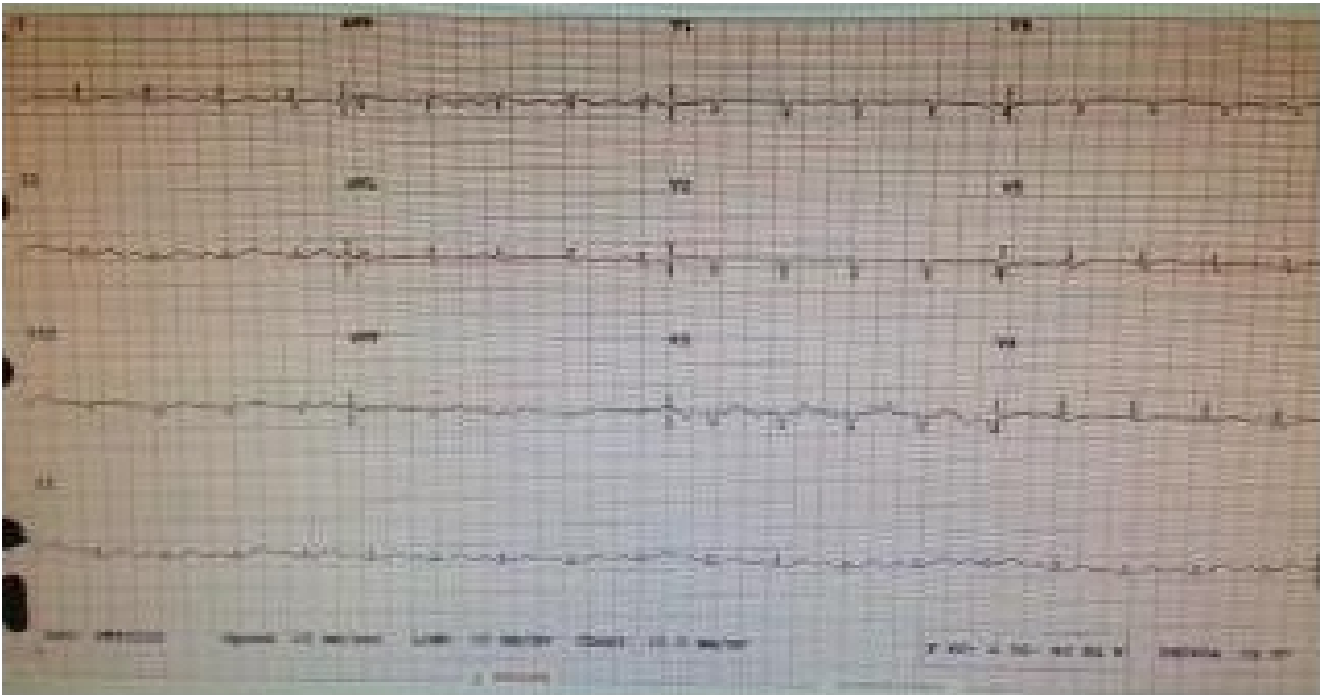


Figure 7: Electrocardiogram showing low voltage

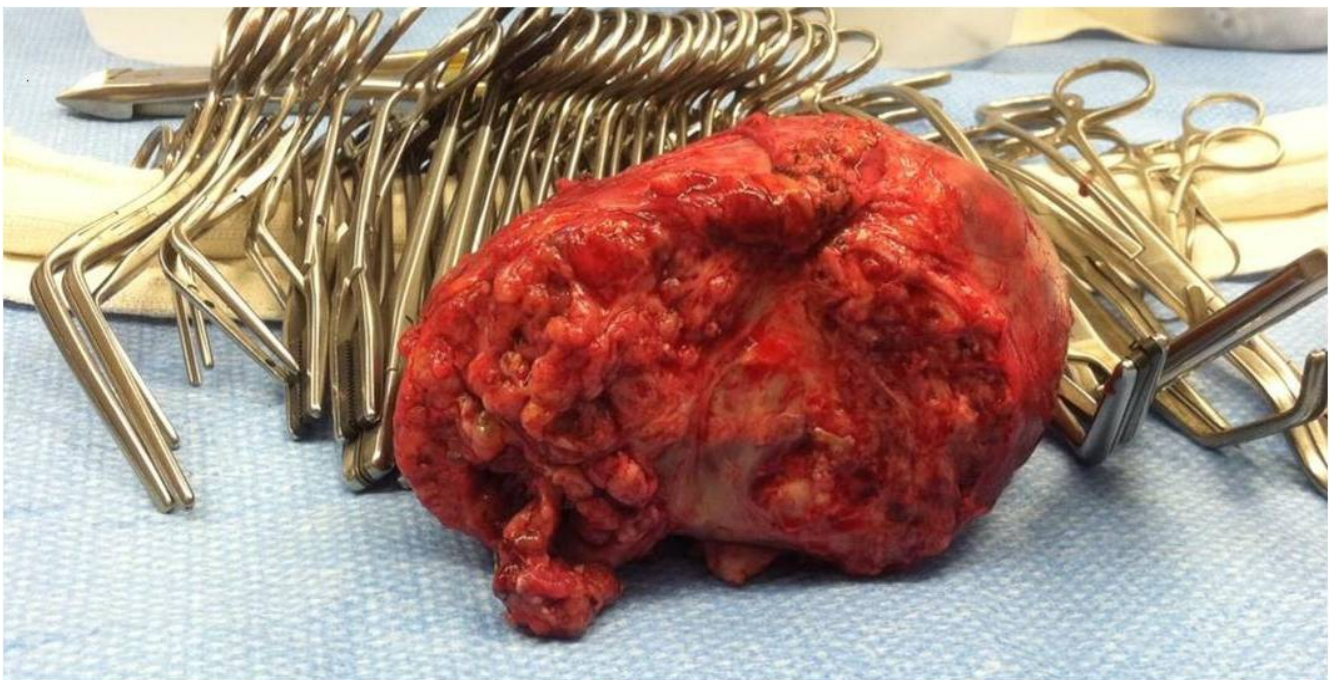


Figure 8: Tumor after excision

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