

CASE REPORT

Klinefelter's Syndrome with Retroperitoneal Teratoma Masquerading as Adrenal Mass: A Very Rare Occurrence in Adults

Raya Almazrouei¹, Ali El-Houni¹, Yamina Houcinat², Ali Ghellai³

Departments of Endocrinology¹, Pathology² and Surgery³, Tawam Johns Hopkins Medicine Hospital, Al Ain, Abu Dhabi, United Arab Emirates.

Corresponding author: Dr. Ali El-Houni Email: elhouni@yahoo.co.uk

Published: 08 March 2014

Ibnosina J Med BS 2014;6(2):106-109

Received: 14 October 2013

Accepted: 11 November 2013

This article is available from: <http://www.ijmbs.org>

This is an Open Access article distributed under the terms of the Creative Commons Attribution 3.0 License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.

Abstract

The association of chromosomal disorders with various neoplasms is well established. We report a case of 36 years old man who was referred for preoperative evaluation of functional status of "presumed" right adrenal mass. The patient was found to have clinical features of hypogonadism and further investigations confirmed the diagnosis of Klinefelter's Syndrome. The patient proceeded to surgery and pathological examination of the removed mass revealed a mature retroperitoneal teratoma abutting the adrenal gland. This case demonstrates that patients with KS are at increased risk of extragonadal germ cell tumors unusual sites from the mediastinum and brain. Retroperitoneal teratoma in the adrenal gland region can be easily confused with adrenal related mass.

Key words: Klinefelter's syndrome, Adrenal incidentilomas, Hypogonadotrophic Hypogonadism, Retroperitoneal teratoma.

Introduction

Klinefelter's syndrome (KS) is the net result of sex chromosomes abnormality where in majority of cases there is an additional X chromosome in a male (47, XXY). In 10-20 % of Klinefelter's syndrome cases; other chromosomal abnormalities such as (46, XY/47, XXY mosaicism; 48, XXXY; 49, XXXXY) may be present (1,2). KS is often associated with various neoplasms, especially germ cell tumors (3,4). Mediastinum is the most favored site of extragonadal germ cell tumors with KS, which is somewhat different from those without KS (3,4). The retroperitoneal germ cell tumor is extremely rare in patients with KS. Therefore, establishing the correct diagnosis preoperatively requires careful assessment of the patient as a whole. We present here an illustrative case and a focused discuss the pertinent literature.

Case Presentation

A 36 years old south-east male was referred to the endocri-

nology clinic for preoperative evaluation of a “presumed” right adrenal mass. He originally presented with recurrent right upper quadrant abdominal pain. An abdominal ultrasound revealed a large heterogeneous mass in the right hepatorenal region. On direct questioning, the patient denied any history of constitutional symptoms such as fever, sweating or weight loss. There was no history of palpitations, headache or high blood pressure. Past history was notable for loss of libido, lack of secondary sexual characteristics. His social history revealed poor school performance and he is employed in construction works as a painter. He is married but the couple has been infertile. Physical examination revealed a normotensive tall young male. His height was 174 cm with long arm span and large hands. Also, he had soft high pitched voice. There was obvious lack of facial, axillary and pubic hair. He had marked prognathism (Figure 1). There were no cushinoid features. There was mild bilateral breast enlargement (Figure 1). Testicular examination showed bilateral small firm testicles of size 5 on orchidometry scale. Abdominal examination revealed a tender mass in the abdominal right upper quadrant of 12 X

13 cm and which was bimanually palpable. Cardiovascular, respiratory and neurological examinations were normal. Investigations showed undetectable serum total testosterone level of <0.90 nmol/L (Reference range: 6.0-27.0), undetectable serum estradiol <73 pmol/L (73-275), and very high gonadotrophin levels [Serum FSH was 31.7 IU/L (1.3-19.3) and serum LH was 10.8 IU/L (1.2-8.60)]. Karyotyping showed 47XXY genotype with no mosaicism. Screening for pheochromocytoma and subclinical Cushing’s syndrome as part of preoperative workup for an incidentally discovered adrenal mass was negative and the all the relevant results were within normal ranges. Their details are beyond the scope of the presentation. Abdominal CT scan showed a 12 x 13 x 12.7 cm sized mass. This was described as inhomogeneous complex mass lesion with cystic, solid, calcification, and fatty components conspicuously within the compartment of the right adrenal gland (Figure 2). The patient underwent right adrenalectomy and mass removal. The pathological examination of the surgical mass revealed mature teratoma with no immature component seen. The tumor itself measured 22x15cm. The tumor was surrounded



Figure 1. The physical features of the patient are shown including: long arm span (A), prognathism and lack of facial hair (B), bilateral breast enlargement with laparotomy scar (C) and large hands compared with right hand of a normal person (D).

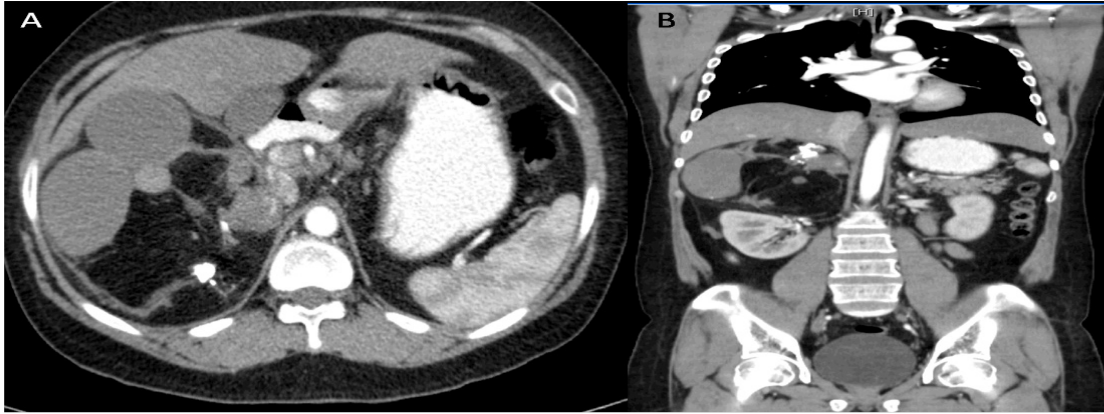


Figure 2. Abdominal CT scanning images show the 12 x 13 x 12.7 cm sized, inhomogeneous complex mass lesion with cystic, solid, calcified, and fatty components within the right adrenal gland region.

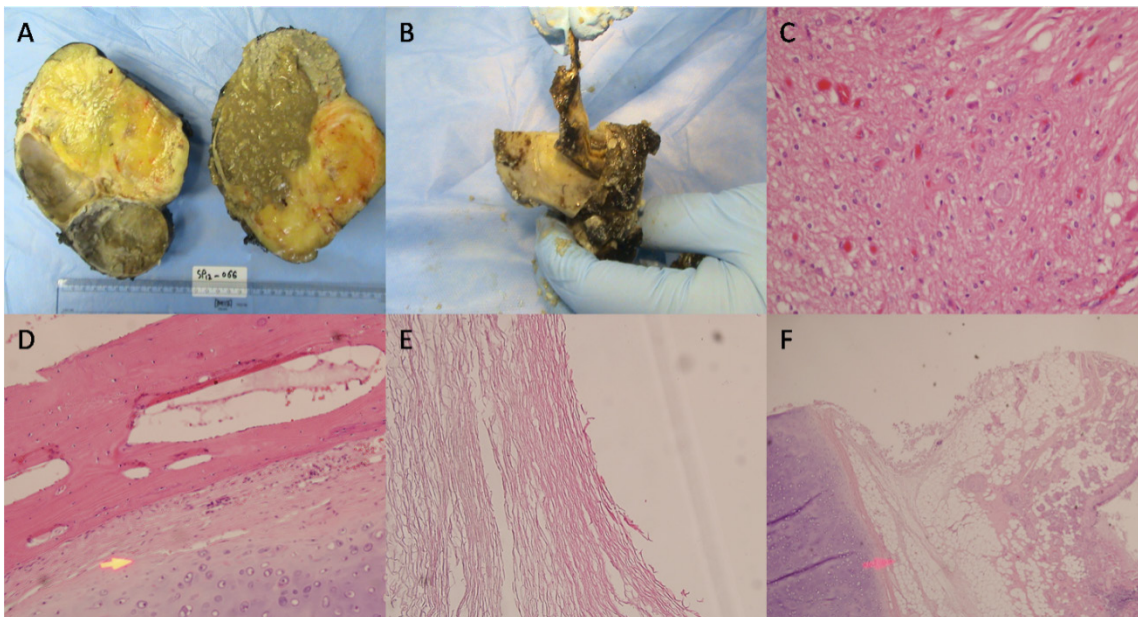


Figure 3. The postoperative pathological examination of the tumor mass illustrating a gross pathology of the whole mass cut open showing the pseudocapsule (A) and the compressed normal adrenal gland (B). The histological demonstration of various parts of the tumor demonstrates mature brain tissue (C), mature bone and cartilage tissue (D), Othokeratotic squamous tissue (E) and ciliated epithelium with glands, cartilage and fatty tissue (F).

by an intact “pseudocapsule”. No features of malignancy were seen within the mass. The compressed adrenal gland parenchyma could be identified at the periphery of the tumor (Figure 3). The patient was discharged home after the surgery to start hormonal replacement therapy and to be followed in the outpatient clinic.

Discussion

The classic clinical features of Klinefelter's syndrome are mainly due to hypergonadotrophic hypogonadism such

as infertility, small testes, decreased facial and body hair, gynecomastia, tall stature and eunuchoid features (1). KS is commonly missed diagnosis or diagnosed late in life. Only around 25% of patients are diagnosed (2). Wide variability of clinical features and lack of awareness among physicians are the most common contributors to delay or missed diagnosis (1). There are several associated conditions with KS that contribute to increased mortality among these patients (1,2,5). Such conditions may include metabolic syndrome, type 2 diabetes mellitus, osteoporosis,

venous ulceration, deep venous thrombosis, pulmonary embolism and propensity to autoimmune disorders as well as tumors (1,2,5). Different malignancies such as breast cancer, testicular tumors, leukemia, and lymphomas occur in 1% to 2% of the cases (5). KS is a well known risk factor for extragonadal germ cell tumors (3,4). Extragonadal germ cell tumors are usually located in the midline from the pineal gland to the coccyx (6). In adults, the most common sites, in order of frequency, are the anterior mediastinum, retroperitoneum, and the pineal and suprasellar regions. On the other hand, sacrococcygeal and intracranial germ cell tumors are most common sites in infants and young children (6). Owing to the fact that KS is a risk factor for extragonadal germ cell tumors, it has been proposed that cytogenetic analysis should always be performed in young patients with mediastinal teratoma (4). There are only few case reports about retroperitoneal extragonadal germ cell tumors in young adults with KS and most data comes from the younger patients (7-9). This case report demonstrates that the retroperitoneal location for germ cells tumors can extend to replace the adrenal gland and can be easily confused as adrenal gland-related mass. Recognizing that the patient has KS features and proceeding with this diagnosis can help the radiologist expand the scope of differential diagnosis when reporting such adrenal mass. This may help prevent unnecessary delay in surgery once the hormonal functionality of the mass has been assessed. Furthermore, this case clearly illustrates the importance of an early diagnosis of KS to improve patient's quality of life and enables better medical treatment (10). In our case, the patient's poor educational performance and the hypogonadism/infertility must have negatively impacted on his career achievements and social life.

References

1. Blevins CH, Wilson ME. Klinefelter's syndrome. *BMJ* 2012;345:e7558 .
2. Groth KA, Skakkebaek A, Høst C, Højbjerg Gravholt C, Bojesen A. Klinefelter syndrome-A clinical update. *J Clin Endocrinol Metab* 2013;98(1):20-30.
3. Aguirre D, Nieto K, Lazos M, Peña YR, Palma I, Kofman-Alfaro S, et al. Extragonadal germ cell tumors are often associated with Klinefelter syndrome. *Hum Pathol* 2006;37(4):477-80.
4. Sachdeva K, Makhoul I, Curti B, Jana BRP. Extragonadal germ cell tumors. [cited 2013 October 14]. available from: <http://emedicine.medscape.com/article/278174-overview/>.
5. Swerdlow AJ, Higgins CD, Schoemaker MJ, Wright

- AF, Jacobs PA, United Kingdom Clinical Cytogenetics Group. Mortality in patients with Klinefelter syndrome in Britain: A cohort study. *J Clin Endocrinol Metab* 2005;90(12):6516-22.
6. Gilligan TD, Kantoff PW. Extragonadal germ cell tumors involving the mediastinum and retroperitoneum. [cited 2013 October 14]. available from: <http://www.uptodate.com/>.
7. Hachimi-Idrissi S, Desmyttere S, Goossens A, Desprechins B, and Otten J. Retroperitoneal teratoma as first sign of Klinefelter's syndrome. *Arch Dis Child*. 1995;72(2):163-4.
8. Yong D, Lim JG, Choi JR, Park Q, Yang CH, Choi SH, Jeong HJ, Song KS. A case of Klinefelter syndrome with retroperitoneal teratoma. *Yonsei Med J*. 2000;41(1):136-9.
9. Czauderna P, Stoba C, Wysocka B, Iliszko M. Association of Klinefelter syndrome and abdominal teratoma: a case report. *J Pediatr Surg*. 1998;33(5):774-5.
10. Radicioni AF, De Marco E, Gianfrilli D, Granato S, Gandini L, Isidori AM et al. Strategies and advantages of early diagnosis in Klinefelter's syndrome. *Mol Hum Reprod* 2010;16(6):434-40.