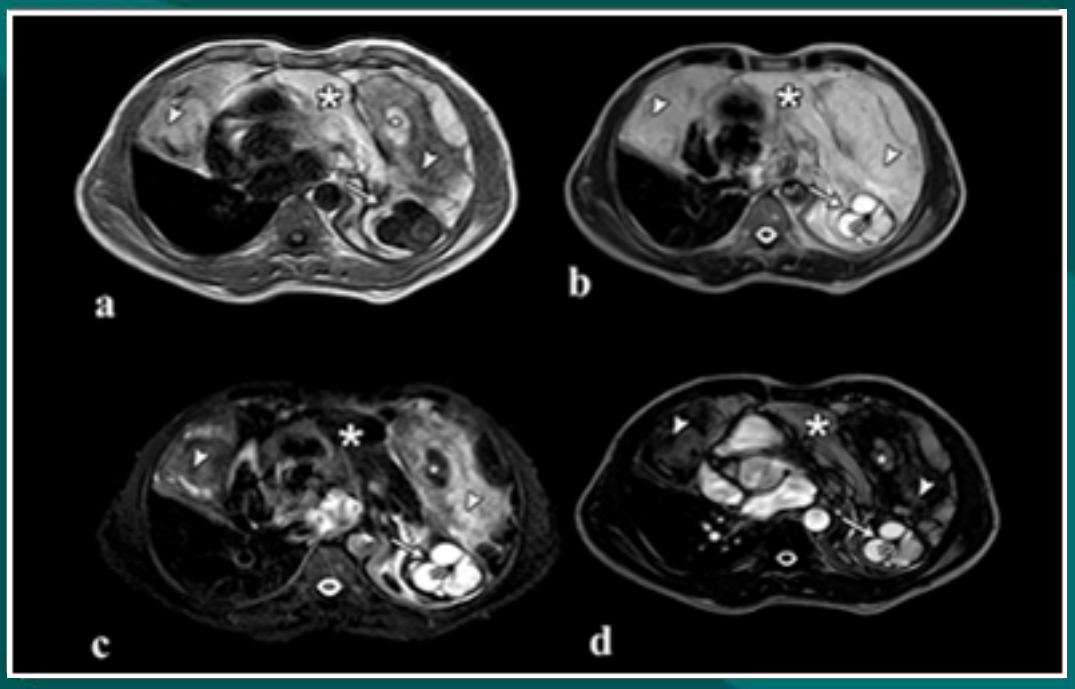
October 2015; Volume 6; Number 10

IJCRI

www.ijcasereportsandimages.com

International Journal of

CASE REPORTS AND IMAGES



Online submission at:
www.ijcasereportandimages.com/submit

ISSN: 0976-3198

International Journal of Case Reports and Images

Contact Details:

Editorial Office

Email: meditor@ijcasereportsandimages.com
Fax: +1-773-409-5040
Website: www.ijcasereportsandimages.com

Guidelines for Authors

Full instructions are available online at:
www.ijcasereportsandimages.com/submit/instructions-for-authors

Manuscript submission:
www.ijcasereportsandimages.com/submit

International Journal of Case Reports and Images

EDITORIAL BOARD

International Journal of Case Reports and Images (IJCRI)

Contact Details:

Editorial Office

Email: meditor@ijcasereportsandimages.com
Fax: +1-773-409-5040
Website: www.ijcasereportsandimages.com

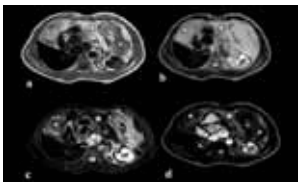
Guidelines for Authors

Full instructions are available online at:
www.ijcasereportsandimages.com/submit/instructions-for-authors

Manuscript submission:
www.ijcasereportsandimages.com/submit

International Journal of
Case Reports and Images

Cover Figure:



All Articles:

Open Access

Contents

Vol. 6, No. 10 (October 2015)

Cover Image

Case Report

Clinical Images

SUBMISSION INSTRUCTIONS
All manuscripts, including illustrations, should be submitted at: www.ijcasereportsandimages.com/submit or email to: submit@ijcasereportsandimages.com
Author Guidelines: www.ijcasereportsandimages.com/submit/instructions-for-authors.php
For any questions contact the Editorial Office via e-mail at: info@ijcasereportsandimages.com or Fax: 1-773-409-5040

CASE REPORT PEER REVIEWED | OPEN ACCESS

Giant solitary fibrous tumor from the lesser omentum causing a gastric outlet obstruction: A case report

Hirra Ali

ABSTRACT

Introduction: Solitary fibrous tumors (SFTs) are exceedingly rare tumors arising from the submesothelial mesenchymal layer. Case Report: A case of a 60-year-old male with paranoid schizophrenia presented with acute gastric obstruction secondary to a large, obstructive, mobile mass. Surgical exploration revealed a 17x15x10.5 cm extraluminal, pedunculated mass originating from the lesser omentum extrinsically compressing the gastric antrum and duodenum. The mass was surgically resected with immediate relief of patient symptoms. Pathology revealed a spindle cell sarcoma with a high mitotic index. Immunohistochemical analysis was positive for CD34 and a newly elucidated nuclear marker that is sensitive and specific for SFT, STAT 6, was used to confirm the diagnosis of SFT. Conclusion: To our knowledge, this is the largest described solitary fibrous tumor arising from the lesser omentum.

Keywords: Gastric outlet obstruction, Mesenchymal layer, Solitary fibrous tumor

Hirra Ali

Affiliations: MD, Montefiore Medical Center, Department of Surgery, The University Hospital for Albert Einstein College of Medicine, 182 East 210th Street, Lower Level, Bronx, NY, USA.

Corresponding Author: Hirra Ali, MD, Montefiore Medical Center, Department of Surgery, The University Hospital for Albert Einstein College of Medicine, 182 East 210th Street, Lower Level, Bronx, NY 10467, USA; Tel: 716-308-9327, Fax: 718-881-5074; Email: hirra.ali1984@gmail.com

Received: 03 May 2015
Accepted: 02 June 2015
Published: 01 October 2015

How to cite this article

Ali H. Giant solitary fibrous tumor from the lesser omentum causing a gastric outlet obstruction: A case report. Int J Case Rep Images 2015;6(10):606–609.

doi:10.5348/ijcri-201597-CR-10558

INTRODUCTION

Solitary fibrous tumors (SFTs) are rare and typically benign entities of mesenchymal origin accounting for less than 2% of all soft tissue tumors. They present as a slow growing mass in middle-aged adults in equal distribution amongst men and women. While they most commonly occur in the thorax, SFTs have been described in the extremities, head and neck, retroperitoneum, pelvis and peritoneum [1–3]. The infrequent presentation in the peritoneum and similarity to other soft tissue tumors makes the diagnosis difficult. We describe the case of a patient with paranoid schizophrenia presenting with acute gastric outlet obstruction secondary to a giant malignant SFT [4].

CASE REPORT

A 60-year-old male with disabling paranoid schizophrenia was sent from his group home to the emergency room after an unwitnessed fall and sustaining a left tibial-fibula comminuted fracture. The patient was admitted to the hospital for medical optimization of his poorly controlled diabetes and hypertension in anticipation of surgical repair. However prior to any intervention, he developed an acute onset of non-bilious emesis and abdominal distention. The patient reported previous such episodes which resolved without any intervention. He denied any abdominal surgery,

family history was unknown and his recent screening colonoscopy was normal. On exam, his abdomen was soft, markedly distended but there was no evidence of peritonitis. He was afebrile and hemodynamically stable. His laboratory values were all within normal limits, without evidence of a leukocytosis or acidosis. A bedside abdominal X-ray showed a distended stomach with a paucity of gas in the small bowel and large fecal burden in the colon (Figure 1). In the absence of any previous abdominal surgery, a computed tomography (CT) scan with only oral contrast was obtained. It revealed a 17-cm pedunculated mass compressing the gastric antrum causing marked dilation, however, there was no evidence of pneumatosis. A nasogastric tube was placed for decompression, and the patient obtained a CT scan with oral and intravenous contrast to ascertain the etiology of this mass. This subsequent CT scan revealed the known heterogeneously enhancing solid mass with eccentric calcification with pedicle inseparable from the duodenum/gastric pylorus. However, there was an interval migration of the mass from the right abdomen to the left with possible twisting on the mesentery (Figure 2). There was new gastric pneumatosis as well as new portal venous gas. The decision was made to emergently take the patient to the operating room for an exploratory laparotomy. In the operating room, a midline incision was made to enter the peritoneal cavity and immediately a large well circumscribed mass was visualized in the left lower quadrant. The mass was pedunculated and the broad base originated from the inferior aspect of the liver arising from the lesser omentum. This mobile base draped over the gastric antrum and first part of the duodenum causing the mechanical gastric outlet obstruction (Figure 3). On further exploration, there was no evidence of ascites, lymphadenopathy, or any distal metastasis. The anterior and posterior serosal surfaces of the stomach were carefully inspected and showed no signs of ischemia. The pedicle was suture ligated and divided at the origin and the specimen freely delivered from surrounding structures. This was sent to a regional tertiary referral oncology pathology unit. On gross report the tumor measured 17x15x10.5 cm and weighed 1.5 kg. It was described as having a rubbery to fleshy cut surface and focal cystic areas (Figure 4). Microscopically, the findings were that of a spindle cell sarcoma with large areas of necrosis as well as increased mitotic activity 8-10 MF/10 HPFs. Although the tumor was mostly composed of a monotonous proliferation of spindle cells separated by a prominent fibrous and sclerotic stroma, there were scattered pleomorphic cells throughout. The immunohistochemical stains show the tumor was diffusely positive for CD34, while negative for CD 117, S100, smooth muscle actin (SMA), CD31 and Factor VIII. Based on the clinical presentation, histologic appearance of a spindle cell lesion associated with abundant collagenous stroma and CD34 reactivity, one possibility included a malignant solitary fibrous tumor. In order to confirm this diagnosis further immunostaining for a recurrent gene fusion NAB2-STAT6 was found to be positive, confirming the diagnosis of SFT. The patient was observed and his postoperative course was uneventful. His NGT removed on postoperative day-1. The patient was discharged

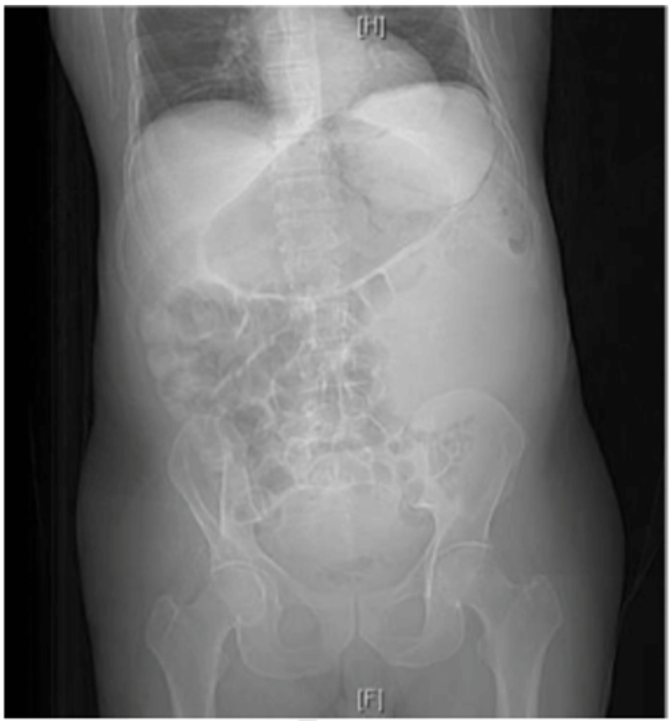


Figure 1: Abdominal X-ray showing markedly dilated stomach.

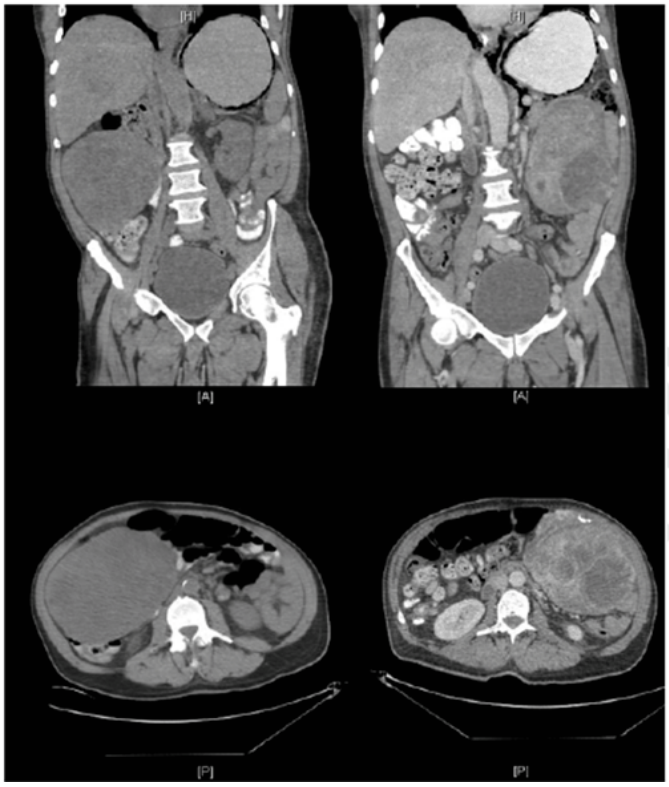


Figure 2: Computed tomography scan within 24 hours of each other depicting movement of obstructing mass from right abdomen to left.

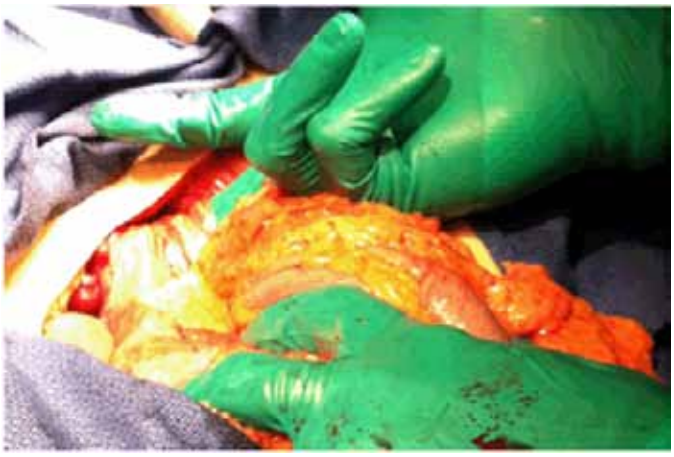


Figure 3: Large pedunculated mass from lesser omentum with surgeon finger placed posterior to stalk and anterior to pylorus at point of obstruction.

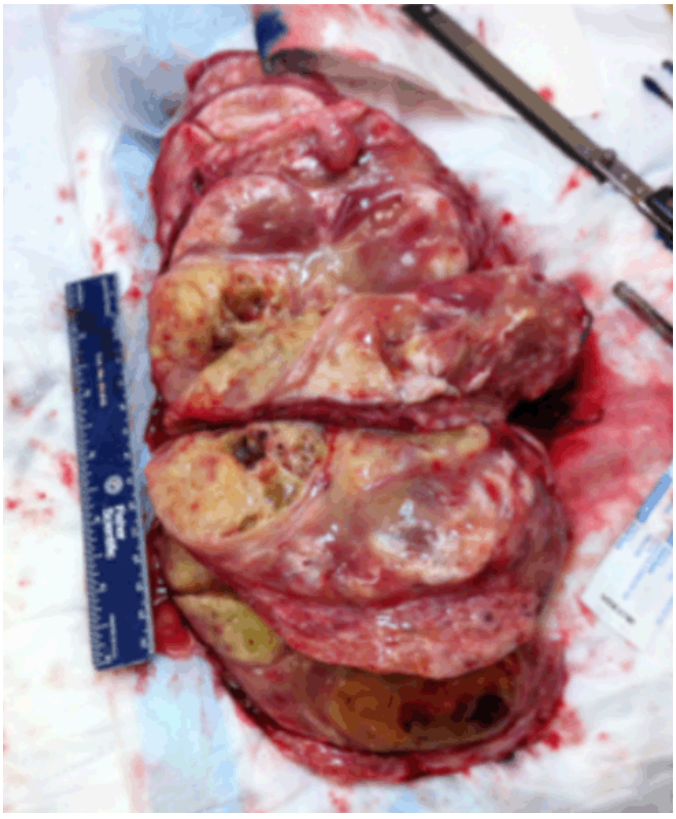


Figure 4: Gross pathology.

after he tolerated a regular diet with return of normal bowel function on postoperative day-2. On immediate postoperative and a three-month follow-up visits, he has been without any recurrence of symptoms.

DISCUSSION

Solitary fibrous tumors are exceedingly rare mesenchymal tumors. While originally described as tumors of the pleural cavity, extrapleural SFTs are now more common than pleural lesions [1–3]. This case of extrapleural SFT is unique not only in the location, size and presentation, but also in the new technology used to confirm diagnosis of SFT. These tumors can occur anywhere in the body, therefore, the clinical manifestations depend not only the site but also the size of the tumor. In 2012, Zong et al. reported that only five cases of SFT arising from the omentum have been described in literature [5]. Tumors larger than 10 cm are referred to as giant. Kudva et al. reported only 25 cases of giant SFTs in literature and only eight of these found in extrapleural sites, making our case unique in location, presentation and size [6]. Given our patient's psychiatric history and limited functional status, it is fortuitous his tumor was diagnosed while in the hospital for unrelated reasons. He endorses a history of chronic intermittent and self resolving obstructive symptoms, likely due the mobile pedunculated mass visualized on CT scan moving from right to left as visualized on CT scan obtained within 24 hours of each other. On CT scan, SFT typically appear as a smooth, well-defined mass with occasional calcifications. While they are hypervascular, there may be areas of non-enhancement corresponding to a cyst or necrosis [7–8]. This was the case in our patient and the mass was initially believed to be a gastrointestinal stromal tumor (GIST), the most common mesenchymal neoplasm in the gastrointestinal tract. Given the nonspecific radiographic characteristics, diagnosis is often made upon surgical resection. Microscopically, the tumor is composed of spindle cells with a patternless architecture composed of hyalinized collagen separating areas of hyper- and hypocellularity. Immunohistochemistry is the only way to confirm a diagnosis of SFT [9]. Our patient was positive for CD 34 reactivity, as is the case in 95% of SFT. However, two-thirds of GIST will also express CD 34 and nearly all express CD 117. The SFTs characteristically are negative for desmin, keratin, S100, CD 117, CD 31, as in our case. Therefore, the diagnosis of GIST was less likely given the negative stain for CD 117 [10]. A newly discovered immunohistochemical stain for nuclear STAT6, a surrogate for the NAB2-STAT6 gene fusion found in SFT, was positive and confirmed the diagnosis [4]. Most SFTs are benign but approximately 20% are described as malignant. The World Health Organization classification of soft tissue tumors describes malignant SFTs as having features including hypercellularity, at least focal moderate to marked cellular atypia, tumor necrosis, >4 mitoses/10 high-power fields, and infiltrative margins. However, there is not a clear consensus on what this prognosticates as histology is not a reliable indicator of the tumors course [1–3].

In a recent review of extrapleural SFT, 55% had malignant features as described above which corresponded to a 33% rate of recurrence and 38% rate of metastasis [11]. Our patient had a giant tumor with three of the four malignant features defined by the WHO classifications, making him high risk for recurrence and metastasis. As such while surgical resection is the

gold standard of treatment for SFT, annual long-term surveillance is of the utmost importance to determine recurrence, metastasis and ultimately malignant potential [12].

CONCLUSION

We describe our experience with a solitary fibrous tumor, a rare tumor presenting in a novel manner causing a gastric outlet obstruction. It is our hope that this contributes to the developing fund of knowledge on the ever enigmatic extrapleural solitary fibrous tumor.

****

Author Contributions

Hirra Ali – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

Authors declare no conflict of interest.

Copyright

REFERENCES

1. Guillou L, Fletcher JA, Fletcher CDM. Extrapleural solitary fibrous tumour and haemangiopericytoma. In: WHO classification of tumors Pathology and Genetics, Tumor of Soft Tissue and Bone. Lyon: IARC Press; 2002:86–90.
2. Gengler C, Guillou L. Solitary fibrous tumour and haemangiopericytoma: evolution of a concept. Histopathology 2006 Jan;48(1):63–74.
3. Huang HY, Sung MT, Eng HL, et al. Solitary fibrous tumor of the abdominal wall: a report of two cases immunohistochemical, flow cytometric, and ultrastructural studies and literature review. APMIS 2002 Mar;110(3):253–62.
4. Doyle LA, Vivero M, Fletcher CD, Mertens F, Hornick JL. Nuclear expression of STAT6 distinguishes solitary fibrous tumor from histologic mimics. Mod Pathol 2014 Mar;27(3):390–5.
5. Zong L, Chen P, Wang GY, Zhu QS. et al. Giant solitary fibrous tumor arising from greater omentum. World J Gastroenterol 2012 Nov 28;18(44):6515–20.
6. Kudva R, Monappa V, Rao A. Giant solitary fibrous tumor of the mesentery: a rare case. J Cancer Res Ther 2011 Jul-Sep;7(3):376–8.
7. Park SH, Kim MJ, Kwon J, et al. Solitary fibrous tumor arising from stomach: CT findings. Yonsei Med J 2007 Dec 31;48(6):1056–60.
8. Shanbhogue AK, Prasad SR, Takahashi N, Vikram R, Zaheer A, Sandrasegaran K. Somatic and visceral solitary fibrous tumors in the abdomen and pelvis: cross-sectional imaging spectrum. Radiographics 2011 Mar-Apr;31(2):393–408.
9. Vogels RJ, Vlenterie M, Versleijen-Jonkers YM, et al. Solitary fibrous tumor - clinicopathologic, immunohistochemical and molecular analysis of 28 cases. Diagn Pathol 2014 Nov 29;9:224.
10. Bishop JA, Rekhtman N, Chun J, Wakely PE Jr, Ali SZ. Malignant solitary fibrous tumor: cytopathologic findings and differential diagnosis. Cancer Cytopathol 2010 Apr 25;118(2):83–9.
11. Cranshaw IM, Gikas PD, Fisher C, Thway K, Thomas JM, Hayes AJ. Clinical outcomes of extra-thoracic solitary fibrous tumours. Eur J Surg Oncol 2009 Sep;35(9):994–8.
12. Tanaka M, Sawai H, Okada Y, et al. Malignant solitary fibrous tumor originating from the peritoneum and review of the literature. Med Sci Monit 2006 Oct;12(10):CS95–8.

CASE REPORT PEER REVIEWED | OPEN ACCESS

Miller–Dieker syndrome with hydronephrosis

Keya Lahiri, Fehmida Najmuddin, Rajesh Rai, Priya Patil Cholera

ABSTRACT

Introduction: Miller–Dieker syndrome (MDS) is a rare genetic disorder which is characterized by lissencephaly, facial dysmorphism and congenital malformations involving-multiple organs. Case Report: We, hereby describe a nine-month-old infant who presented to our tertiary care hospital with developmental delay, infantile spasm and bronchopneumonia. On examination, there was prominent forehead, bi-temporal hallowing, bilateral ptosis, upturned nares and low set ears. Other dysmorphic features were micrognathia, thickened upper lip, high arched palate, umbilical hernia and polydactyly was noted bilaterally in both the upper and lower limbs. The investigations, revealed Lissencephaly type 1 and left-sided hydronephrosis. The typical dysmorphic facies, neurological involvement and lissencephaly type 1 led to the diagnosis of Miller–Dieker syndrome. Conclusion: Miller–Dieker syndrome involving the genitourinary system anomaly in the form of hydronephrosis with pelvi-ureteric junction obstruction, has not been yet described in literature.

Keywords: Miller-dieker syndrome, Lissencephaly, Hydronephrosis

How to cite this article

Lahiri K, Najmuddin F, Rai R, Cholera PP. Miller–Dieker syndrome with hydronephrosis. Int J Case Rep Images 2015;6(10):610–613.

doi:10.5348/ijcri-201598-CR-10559

Keya Lahiri[1], Fehmida Najmuddin[2], Rajesh Rai[1], Priya Patil Cholera[3]

Affiliations: [1]Pediatrics, Professor, Department of Pediatrics, D Y Patil Medical College, Hospital & Research Centre, Maharashtra, India; [2]Pediatrics, Assistant Professor, Department of Pediatrics, D Y Patil Medical College, Hospital & Research Centre, Maharashtra, India; [3]Ophthalmology, Assistant Professor, Department of Ophthalmology, D.Y. Patil Medical College, Hospital & Research Centre, Maharashtra, India.

Corresponding Author: Dr. Fehmida Najmuddin, Department of paediatrics, D Y Patil Medical College, Hospital & Research Centre, Maharashtra, India; Ph: 0091-9920030533; Email: fehmidanc@hotmail.com

Received: 27 May 2015
Accepted: 22 June 2015
Published: 01 October 2015

INTRODUCTION

Miller–Dieker syndrome (MDS) results from a contiguous gene deletion involving 17p13.3 locus [1]. L1S1 gene is required for neurogenesis and neuronal migration and is located at 17p3. Miller–Dieker syndrome is usually associated with complete absence of this gene, which leads to classical Type I lissencephaly. Defect in the neuronal migration during embryonic development results in lissencephaly [2]. Apart from typical abnormal facies, MDS is also associated with intellectual disability, developmental delay, hypotonia and seizure disorder by first year of life.

CASE REPORT

A nine-month-old male infant presented to a tertiary care hospital with complaints of inability to hold neck since five months of age, convulsions since three months, cough for a period of ten days and fever for two days. Convulsions were characterized by rapid

forward bending of head and simultaneous movements of the arm suggestive of infantile spasms lasting for 2–3 seconds. Child was born of a non-consanguineous marriage with two well grown siblings. Mother did not attend the antenatal clinic. Infant was a full term normal vaginal delivery with birth weight of 3300 gms and was immunized till date. There was history of global developmental delay and the child belonged to a lower middle social class.

On examination the patient was conscious, moderately built and nourished with a heart rate 112/min, respiratory rate 54/min, temperature 98.6˚F and blood pressure 94/50 mmHg. Anthropometric measurements were normal for his age. On head to toe examination there was prominent forehead, bi-temporal hallowing, bilateral ptosis, upturned nares and low set ears. Polydactyly was noted bilaterally in both the upper and lower limbs (Figure 1).

Other dysmorphic features revealed micrognathia, thickened upper lip, high arched palate and umbilical hernia (Figure 2).

Cranial nerve involvement was in the form of bilateral ptosis with no response to sound and dysphagia with feeding difficulties. There was hypotonia with absent superficial reflexes and brisk deep tendon reflexes. There were no signs of meningeal irritation or cerebellar involvement. Other systems revealed bilateral crepitations along with hepatosplenomegaly.

Hemoglobin was 8.9 g/dl, total leucocyte count 30,000/mm^3 with neutrophils 64.3%, lymphocytes 30.2%, eosinophils 1.2%, monocytes 4.1% and platelets 6.01x10^5/mm^3. The peripheral smear was suggestive of microcytic hypochromic anemia with neutrophilic leukocytosis and blood culture report was negative.

Chest X-ray was suggestive of bronchopneumonia. Thyroid profile, serum electrolytes, calcium, alkaline phosphatase, liver and renal function tests were normal.

Magnetic resonance imaging (MRI) scan of brain showed smoothening of the cortical surface of bilateral cerebral hemispheres with hypoplastic sulci and broad flattened gyri. The cerebral cortex was thickened, smooth well defined grey-white matter differentiation noted along with bilateral Sylvian fissures appearing shallow. All these features were suggestive of lissencephaly type I (Figure 3) Cranial ultrasonography did not reveal any abnormality. A computed tomography (CT) scan of abdomen and pelvis showed left sided gross hydronephrosis without hydroureter and marked thinning of cortex which was suggestive of pelvi-ureteric junction (PUJ) obstruction.

An abnormal sleep EEG with right to left asymmetry and left occipital epileptogenesis was noted. Echocardiography and karyotyping were normal.

The patient was administered injectable ceftriaxone (75 mg/kg/day) for 10 days. Syrup phenobarbitone (5 mg/kg/day) was started due to repeated episodes of convulsions. A surgical intervention for hydronephrosis has been planned. Classical dysmorphic features and type 1 lissencephaly characterized him as MDS.

Figure 1: Showing polydactyly in the upper and lower limbs.

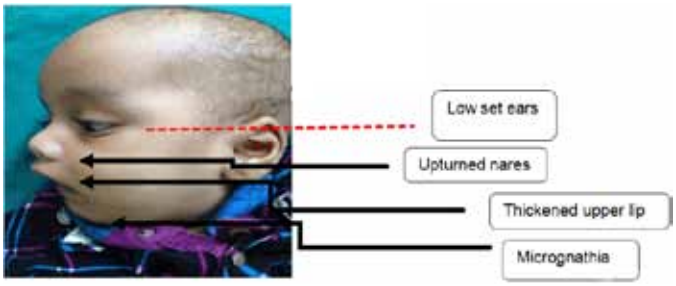


Figure 2: Showing various dysmorphic features.

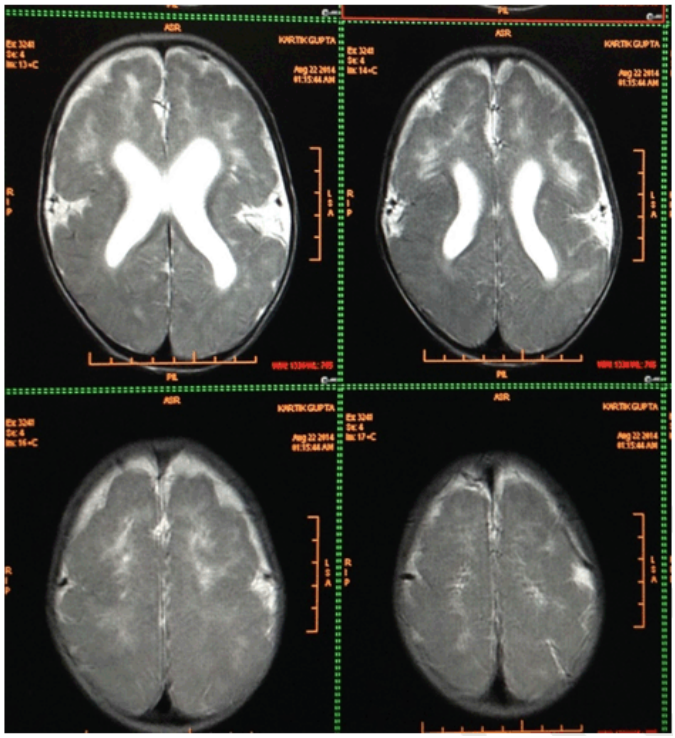


Figure 3: Showing features suggestive of lissencephaly type 1.

DISCUSSION

Three types of lissencephaly have been described viz. Type-1 being the classical form which is associated with MDS. Type -2, also known as cobblestone lissencephaly is associated with o-glycosylation enzyme defect and type-3 is a neurodegenerative process with abnormal apoptosis. Apart from MDS, type-1 lissencephaly is also associated with Norman-Roberts syndrome, but these patients have severe microcephaly [3]. Apart from the characteristic dysmorphic features and lissencephaly, there was renal

anomaly in the form of left-sided hydronephrosis with PUJ obstruction reported in our case.

These children have an increased life expectancy due to better seizure control, nasogastric feeding and improved nutrition [4]. The most common cause of mortality associated with this syndrome is aspiration and recurrent respiratory infections [5].

A detailed antenatal history and regular antenatal visits is essential. History of polyhydramnios, intrauterine growth retardation and reduced fetal movements are associated with MDS. Prenatal ultrasonography findings in MDS are smooth gyral pattern, ventriculomegaly, large subarachnoid space, congenital heart disease and omphalocele [6, 7]. Classical lissencephaly can be detected on imaging only after 28 weeks of gestation.

The recurrence risk for MDS is very low, as the chromosomal deletion is usually a de novo event. However, if it is associated with a familial reciprocal translocation, the recurrence risk for an abnormal live born can be as high as 33%. Normal results of fluorescent in situ hybridization (FISH) and chromosomal microarray tests cannot rule out partial and intragenic deletions or duplications of LIS1 gene [8].

The differential diagnosis of MDS includes Cornelia de Lange, Wolf-Hirschorn, Smith Lemli-Opitz and Zellweger syndrome as these syndromes too have facial dysmorphism, microcephaly, seizures and hypotonia but none of them are associated with lissencephaly [9].

The child has been gaining weight and has demonstrated reduced frequency of seizures on follow-up.

CONCLUSION

To conclude, any child presenting with neurological involvement, lissencephaly and typical dysmorphism should be investigated for the above syndrome.

****

Author Contributions

Keya Lahiri – Substantial contribution to conception and design, Acquisition of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Fehmida Najmuddin – Substantial contribution to conception and design, Acquisition of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Rajesh Rai – Substantial contribution to conception and design, Acquisition of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published.

Priya Patil Cholera – Substantial contribution to conception and design, Acquisition of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

Authors declare no conflict of interest.

Copyright

REFERENCES

1. Mahgoub L, Aziz K, Davies D, Leonard N. Leonard. Miller-dieker syndrome associated with congenital lobar emphysema. AJP Rep 2014 May;4(1):13–6.
2. Dobyns WB, Stratton RF, Parke JT, Greenberg F, Nussbaum RL, Ledbetter DH. Miller-Dieker syndrome: lissencephaly and monosomy 17p. J Pediatr 1983 Apr;102(4):552–8.
3. Verloes A, Elmaleh M, Gonzales M, Laquerrière A, Gressens P. [Genetic and clinical aspects of lissencephaly].
[Article in French]. Rev Neurol (Paris) 2007 May;163(5):533–47.
4. Pilz D. Miller-Dieker Syndrome. Orphanet encyclopedia. September 2003:1–3.
5. Spalice A, Parisi P, Nicita F, Pizzardi G, Del Balzo F, Iannetti P. Neuronal migration disorders: clinical, neuroradiologic and genetics aspects. Acta Paediatr 2009 Mar;98(3):421–33.
6. Chen CP, Chein SC. Prenatal Sonographic Features of Miller-Dieker Syndrome. J Med Ultrasound 2010;18(4):147–52.
7. Colavita L, De Vivo D, Pantaleo D, Pizzino MR, Sturiale M, Briuglia S. Miller-Dieker Syndrome. The Child A Journal Of Pediatrics 2014 Feb;1(1):1–2.
8. Dobyns WB, Das S. LIS1-Associated Lissencephaly/ Subcortical Band Heterotopia. In: Pagon RA, Adam MP, Ardinger HH, et al. editors. Seattle (WA): University of Washington, GeneReviews® [Internet]. Seattle; 1993-2015.
9. Rivas MV, Alvarez LA, Altman N. Pictoral review: Miller-Dieker syndrome. Int Pediat 1994;9:280–3.

CASE REPORT PEER REVIEWED | OPEN ACCESS

Anomalies origin of the right coronary artery from the left coronary sinus and coursing between the pulmonary artery and aorta associated with mitral stenosis

Uliks Ekmekçiu, Mimoza Lezha, Gjin Ndrepepa

ABSTRACT

Introduction: Coronary arteries anomalies are congenital. Usually, they are asymptomatic. They are found during the coronary angiography or computed tomography angiography. The most common coronary anomaly is separated origin of left anterior descending coronary artery and left circumflex artery coronary artery. Usually, it is a benign anomaly. Case Report: A 54-year-old male was admitted at the service of cardiology. Twenty-five years ago he was diagnosed with mitral stenosis and five years ago as having a thrombotic cerebrovascular accident. Electrocardiogram showed atrial fibrillation Trans-thoracic echocardiography showed calcified mitral stenosis, with an anatomical area of 1.1 cm². The patient was treated with oral anticoagulants (acenocumarol), beta-blockers (atenolol), and diuretics (hydrochlorothiazide plus spironolactone). The patient underwent coronary angiography which showed a 75% stenosis of the right coronary artery. The origin and course of right coronary artery was abnormal and thus a CT angiography was performed. The CT angiography confirmed that the origin of the right coronary artery was from the left coronary sinus and that the artery coursed between the aorta and the pulmonary artery. Under these circumstances, the patient was transferred to cardiac surgery where the mitral valve replacement and coronary artery bypass graft surgery were performed. The in-hospital course was uneventful. The patient was free shortness of breath and chest pain (angina). One month later CT angiography was repeated. The patient remained symptom-free and in good health status. Conclusion: The case highlights this anomaly and its potential association with mitral stenosis.

Keywords: Coronary artery anomaly, Mitral stenosis, Computed tomography angiography

How to cite this article

Ekmekçiu U, Lezha M, Ndrepepa G. Anomalies origin of the right coronary artery from the left coronary sinus and coursing between the pulmonary artery and aorta associated with mitral stenosis. Int J Case Rep Images 2015;6(10):614–617.

doi:10.5348/ijcri-201599-CR-10560

Uliks Ekmekçiu[1], Mimoza Lezha[2], Gjin Ndrepepa[3]

Affiliations: [1]Internist, Department of Cardiology, University Hospital Center "Mother Teresa" Tirana, Albania; [2]Cardiologist, Department of Cardiology, University Hospital Center "Mother Teresa" Tirana, Albania; [3]Cardiologist, Researcher, Deutches Herzzentrum, Technische Universität, Munich, Germany.

Corresponding Author: Mimoza Lezha, Tirana, Albania- 1000
Ph: +355682140304; E-mail: mlezha@hotmail.com

Received: 24 May 2015
Accepted: 01 July 2015
Published: 01 October 2015

INTRODUCTION

Coronary artery anomalies are present at birth. They are usually asymptomatic. The prevalence is less 1% [1, 2, 3]. They are found during coronary angiography or computed tomography angiography. The most common coronary anomaly is the separated origin of left anterior descending coronary artery and left circumflex artery. Usually, it is a benign anomaly. Anomalous origin of the right coronary artery from the left aortic sinus is found

in 0.03–0.9% of patients during coronary angiography. A higher incidence of coronary anomalies was observed in young victims of sudden death than in adults (4-15% versus 1%). A higher incidence of coronary artery anomalies in young versus old people with sudden cardiac death has also been reported. Most of coronary artery anomalies are usually asymptomatic and are recognised during autopsy. A single coronary artery and coronary artery fistulas are found in 0.024% and 0.2% of the autopsies, respectively. Coronary artery anomalies are the second most common case of sudden death in young athletes, after hypertrophic cardiomyopathy. No race or sex differences have been reported [1–5].

CASE REPORT

A 54-year-old male was admitted at the service of cardiology with the diagnosis of mitral stenosis. The patient complained of difficult breathing, chest pain and blue (cyanotic) fingers and lips during physical activities over the last 5–6 months. Twenty-five years ago he was diagnosed with mitral stenosis and five years ago as having a thrombotic cerebrovascular accident. The patient did not remember to have had any episode of rheumatic fever.

At physical examination he was right hand monoparesis, irregular cardiac rhythm and an accentuated second heart tone at the mitral valve area. Blood pressure was 120/80 mmHg. Heart rate was 100 beats per minute. Electrocardiogram showed atrial fibrillation trans-thoracic echocardiography showed calcified mitral stenosis, with an anatomical area of 1.1 cm^2, aortic regurgitation, left atrial dilatation and slight tricuspid valve regurgitation. Pulmonary artery systolic pressure was 30 mmHg (Figure 1).

Blood tests were as follows: leukocytes 8700 cells/mm^3, erythrocytes 5.610.000/mm^3, hematocrit 45%, platelets 233,000/mm^3, glucose 86 mg/dl, urea 30 mg/dl, creatinine 1.1 mg/dl, aspartate aminotransferase (AST) 29 UI/L, alanine-aminotransferase (ALT) 17 UI/L, total bilirubin 0.7 mg/dl, serum sodium 131 mmol/L, serum potassium 4.1 mmol/L, serum chlorine 97 mmol/L.

The patient was treated with oral anticoagulants (acenocumarol), beta-blockers (atenolol), and diuretics (hydrochlorothiazide plus spironolactone). Coronary angiography which showed a 75% stenosis of the right coronary artery (Figure 2).

The origin and course of right coronary artery was abnormal and thus a CT angiography was performed. The CT angiography confirmed that the origin of the right coronary artery was from the left coronary sinus and that the artery coursed between the aorta and the pulmonary artery (Figure 3 and Figure 4).

Under these circumstances, the patient was transferred to cardiac surgery where the mitral valve replacement and coronary artery bypass graft surgery (between aorta and right coronary artery) were performed.

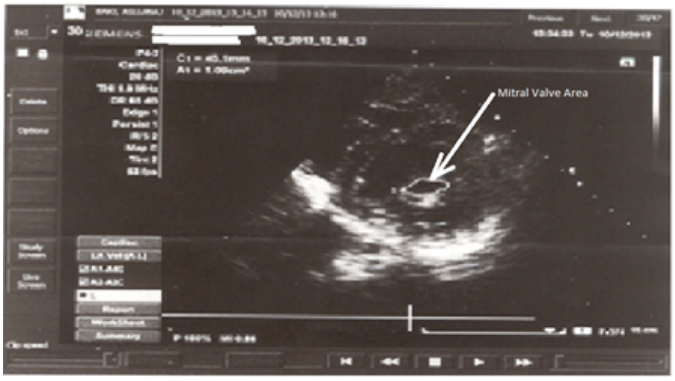


Figure 1: Transthoracic echocardiography, parasternal short axis view. Anatomical area of the mitral valve is depicted. Mitral valve (particularly the posterior cusp) is calcified. Maximal Gradient was 11.6 mmHg; mean gradient 5mmHg.

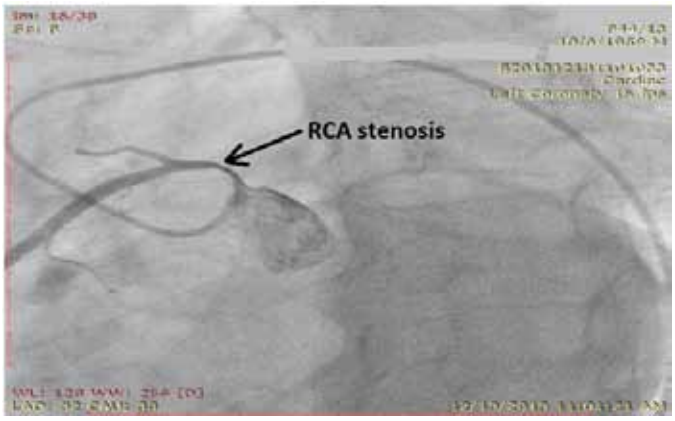


Figure 2: Coronary Angiography. Left anterior oblique caudal (spider) view. The image shows a 75% proximal stenosis of the right coronary artery. The stenosis in mechanical due to compression of the artery between aorta and pulmonary artery.

Figure 3: Computed tomography angiography. Course of right coronary artery is between Aorta and the pulmonary artery. Ao = Aorta; PA = Pulmonary artery.

The in-hospital course was uneventful. The patient was free shortness of breath and chest pain (angina). One month later CT angiography was repeated. The patient remained symptom-free and in good health status.

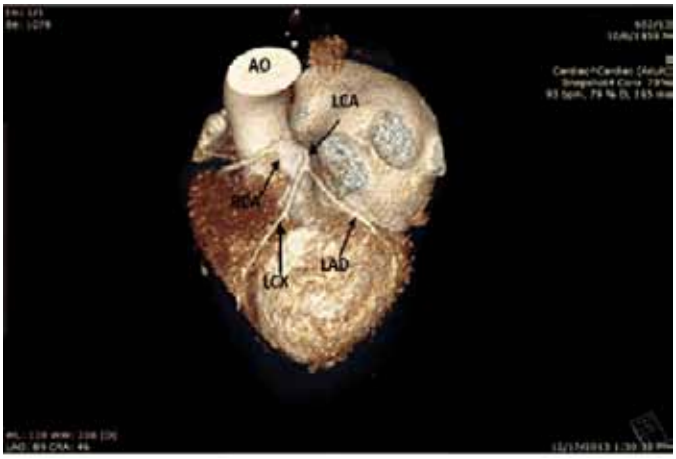


Figure 4: Computed tomography angiography reconstruction. Three dimensional view of coronary artery, origin. The origin of the right coronary artery is from the left coronary sinus. AO = Aorta; LAD = Left descending coronary artery; LCA = Left coronary artery; LCx = Left circumflex coronary artery. RCA = right coronary artery.

DISCUSSION

Herein, we presented a rare case of a coronary artery anomaly associated with mitral stenosis. However, it remains unclear whether mitral stenosis was congenital or rheumatic in origin or whether there is any causal relationship between the coronary artery anomaly and mitral stenosis. Moreover, we did not find any reported case in which anomalies of the origin of right coronary artery were associated with mitral stenosis. The patients had dyspnea and chest pain. However, it remains unknown whether these symptoms were due to mitral stenosis or right coronary narrowing and subsequent myocardial ischemia (due to compression of the right coronary artery from aorta and pulmonary artery, particularly during effort) [1, 2].

Anatomic anomalies of coronary arteries include anomalies of origin, course, termination or their combination. The most common anomaly is the independent origin of the left anterior descending and left circumflex coronary arteries. Second is the anomalous origin of the left circumflex from the right coronary artery. The rarest anomaly is origin of the right coronary artery from the left coronary sinus.

Coronary arteries are perpendicular to the aortic wall. Abnormal coronary arteries that get out ectopically usually course tangentially to the aortic wall and in close relationship to the aortic valve. Course between aorta and pulmonary artery may be associated with myocardial ischemia and sudden death. Although the reason for this variability is unknown, several mechanisms have been proposed:

1) compression of the right coronary artery between aorta and pulmonary artery during exercise
2) compression of the intramural segment of the proximal coronary artery by the aortic wall
3) spasm of coronary artery as a result of endothelial injury [1–4].

This anomaly is associated with sudden death in young athletes during exercise. It is not known whether this pathology mediates an increased risk for early development of coronary artery disease. In our patient, the anomalous origin of the right coronary artery was not associated with atherosclerotic stenosis. The right coronary artery stenosis was mechanical (compressive) [1–5].

CONCLUSION

We presented here a very rare case of anomalies right coronary artery origin from the left aortic sinus coursing between aorta and pulmonary artery associated this with severe mitral stenosis. Although, the patient showed a 75% narrowing of the right coronary artery, the stenosis was not of atherosclerotic origin. The case highlights this anomaly and its potential association with mitral stenosis and non-atherosclerotic (compressive) narrowing of the right coronary artery.

****

Author Contributions

Uliks Ekmekçiu – Substantial contributions to concept and design, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Mimoza Lezha – Substantial contributions to concept and design, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Gjin Ndrepepa – Substantial contributions to concept and design, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

Authors declare no conflict of interest.

REFERENCES

1. Vieira C, Nabais S, Salgado A, et al. Anomalous coronary origin: from suspicion to surgical revascularization. Rev Port Cardiol 2014 Jan;33(1):53. e1–5.
2. Iñiguez Romo A, Macaya Miquel C, Alfonso Monterola F, San Román Calvar JA, Goikolea Ruiz-Gómez J, Zarco Gutiérrez P. Congenital anomalies of coronary artery origin: a diagnostic challenge. Rev Esp Cardiol 1991 Mar;44(3):161–7.
3. Naruse Y, Sato A, Kakefuda Y, et al. Anomalous origin of the right coronary artery from the left sinus of valsalva in an elderly man: life-threatening worsening of angina after left lung lobectomy. Circulation 2012 Jun 19;125(24):e1021–4.
4. Jim MH, Siu CW, Ho HH, Miu R, Lee SW. Anomalous origin of the right coronary artery from the left coronary sinus is associated with early development of coronary artery disease. J Invasive Cardiol 2004 Sep;16(9):466–8.
5. Qin X, Xiong W, Guan E, Lu C. Coronary anomaly: anomalous right coronary artery originates from the left sinus of Valsalva and coursing between the pulmonary artery and aorta. Clin Interv Aging 2013;8:1217–20.

CASE REPORT PEER REVIEWED | OPEN ACCESS

A rare cause of rhabdomyolysis: Gitelman syndrome

Yaşar Yildirim, Ali Veysel Kara, Zulfikar Yilmaz, Erdal Bodakci, Vehbi Demircan, Ali Kemal Kadiroglu, Mehmet Emin Yilmaz

ABSTRACT

Introduction: Gitelman syndrome is a rare and autosomal recessive disorder characterized by hypokalemia, metabolic alkalosis, hypomagnesemia, hypocalciuria and hypertension. A careful history, physical examination and determination of urine chloride concentration are important for diagnosis. In this case report, we discuss a patient with hypokalemia and rhabdomyolysis which is diagnosed as Gitelman syndrome. Case Report: A patient with sudden loss of sensation in the arms and legs and difficulty in the moving admitted to the emergency service. She had intermittent weakness in the extremities and chronic fatigue complaints over the last ten years. In her laboratory examination, serum potassium level, serum creatinine level and creatine kinase level were found 1.4 mEq/L, 1.58 mg/dl and >4260, respectively. The patient was transferred to our clinic due to hypokalemia etiology. She was diagnosed as Gitelman syndrome after a detailed medical history, physical examination and laboratory evaluation. Central venous catheter was opened and vigorous potassium chloride replacement was made. Patient's symptoms and muscle enzymes were improved with potassium replacement. Oral potassium citrate, spironolactone, magnesium citrate was started in addition to intravenous potassium chloride after the patient was diagnosed as Gitelman syndrome. After that, clinical and laboratory findings of the patient were improved progressively and patient was discharged with normal laboratory findings. Conclusion: As a result; Gitelman syndrome should be thought in the differential diagnosis of hypokalemic rhabdomyolysis although it is a rare disease.

Keywords: Gitelman syndrome, Hypokalemia, Rhabdomyolysis, Sensation loss, Weakness

Yaşar Yildirim[1], Ali Veysel Kara[1], Zulfikar Yilmaz[1], Erdal Bodakci[2], Vehbi Demircan[2], Ali Kemal Kadiroglu[1], Mehmet Emin Yilmaz[1]

Affiliations: [1]Dicle University Faculty of Medicine, Department of Nephrology, Diyarbakır, Turkey; [2]Dicle University Faculty of Medicine, Department of Internal Medicine, Diyarbakır, Turkey.

Corresponding Author: Asistant Professor Dr. Yaşar Yildirim, Dicle University Faculty of Medicine, Department of Nephrology, Diyarbakır, Turkey; GSM: 0090 532 306 83 16

Fax: 0090 412 2488171; Email: yyil11@yahoo.com

Received: 10 June 2015
Accepted: 01 July 2015
Published: 01 October 2015

How to cite this article

Yildirim Y, Kara AV, Yilmaz Z, Bodakci E, Demircan V, Kadiroglu AK, Yilmaz ME. A rare cause of rhabdomyolysis: Gitelman syndrome. Int J Case Rep Images 2015;6(10):618–621.

doi:10.5348/ijcri-2015100-CR-10561

INTRODUCTION

Gitelman syndrome is a rare and autosomal recessive disorder characterized by hypokalemia, metabolic alkalosis, hypomagnesemia, hypocalciuria and hypertension [1, 2]. The prevalence of Gitelman syndrome was 1 in 40,000. It is one of the most frequent inherited renal tubular disorders [3]. Genetic mutations that responsible for disease affect the NaCl co-transporter in the distal tubule [4, 5]. Clinical manifestations such

as cramps of the arms and legs which are observed in nearly all patients and may be due to hypokalemia and hypomagnesemia , polyuria due to salt and water loss, fatigue due to renal salt wasting can be develop in the most of patients [1, 2]. Growth retardation may be seen rarely [6]. Other causes of hypokalemia and metabolic alkalosis especially vomiting and diuretic usage must be excluded before the diagnosis of Gitelman syndrome. A careful history, physical examination and determination of urine chloride concentration are important for diagnosis. Genetic testing is important for diagnosis but not widely used. In this case report, we discuss a patient with hypokalemia and rhabdomyolysis which is diagnosed as Gitelman syndrome.

CASE REPORT

A patient with sudden loss of sensation in the arms and legs and difficulty in the moving admitted to the emergency service. He had intermittent weakness in the extremities and chronic fatigue complaints over the last ten years. In her laboratory examination, serum potassium level, serum creatinine level and creatine kinase level were found 1.4 mmol/L (3.5–5.5 mmol/L) , 1.58 mg/dl 80.5–1.4 mg/dl) and >4260 U/l (40–165 U/l), respectively. She was hospitalized and intravenous and oral potassium replacement was made during 2 days. However, there was no increase in potassium levels. She was transferred to our clinic due to hypokalemia etiology. At the time of admission; she had no nausea, vomiting and diarrhea. There was no diuretic and laxative usage in the patient medical history. Physical examination, electrocardiography (normal QT interval), echocardiography and X-ray lung graphy of the patient revealed no abnormal finding. On abdominal ultrasonography; bilateral kidney size were at the upper limit and renal parenchymal echo were grade 1. Laboratory findings of the patient are given in Table 1. Central venous catheter was opened and vigorous potassium chloride replacement was made (50 mEq in 1000 ml normal saline, 3000 ml/day). There was no usage of drugs which can cause transcellular shift such as insulin, and b2 adrenergic agonists, and thyroid functions were normal. We detected urine potassium level to differentiate renal and extrarenal loses. Urine potassium were detected 30 mmol/l and 35 mmol/l (normally <15 mmol/l) in repeated tests. Transtubuler potassium gradient (TTPG) was 8 (>4 are significant). So, we thought increased distal potassium secretion. Causes which complicated with hypertension such as Cushing syndrome, Liddle syndrome, licorice ingestion, renal artery stenosis were ruled out due to patient was normotensive and she had central venous pressure of 4 cm H2O. Renal tubular acidosis was also ruled out due to metabolic alkalosis. Urine chloride level was 88 mmol/l. So, there were four main causes to explain hypokalemic metabolic alkalosis and elevated urine chloride level. These were thiazide and loop diuretic usage, Bartter syndrome and Gitelman syndrome. We ruled out Bartter syndrome because there were hypomagnesemia, hypocalciuria and urine calcium creatinine ratio <0.1. Hypomagnesemia, hypocalciuria, hypokalemia, metabolic alkalosis and high plasma renin activity (195.6 ng/ml/hour) was detected and patient was diagnosed as Gitelman syndrome. Patient‘s symptoms and muscle enzymes were improved with potassium replacement (120 mEq/day intravenous infusion in 3000 ml/day saline). Oral potassium citrate, spironolactone, magnesium citrate was started in addition to intravenous potassium chloride after the patient was diagnosed as Gitelman syndrome. After that, clinical and laboratory findings of the patient were improved progressively and patient was discharged with normal laboratory findings.

Table 1: Laboratory values of the patient at admission

Parameters	Patient (normal value)
Creatinine (mg/dl)	1.58 (0.5–1.4)
Urea (mg/dl)	64 (10–45)
Sodium (mEq/l)	139 (136–145)
Potassium (mmol/l)	1.8 (3.5–5.1)
Magnesium (mg/dl)	1.1 (1.5–2.6)
Calcium (mg/dl)	8.4 (8.4–10.2)
Daily urine calcium (mg)	34 (100–300)
Creatine kinase (U/l)	>4267 (40–165)
Lactate dehydrogenase (U/l)	896 (125–243)
Supine plasma renin (ng/mL/h)	195.6 (0.2–1.6)
Supine plasma aldosterone (pg/ml)	132.55 (30–160)
TSH (uUI/ml)	4.1 (0.27–4.2)
ACTH (pg/ml)	38 (10–48)
PTH (pg/ml)	83 (15–65)
25-OH-Vit D (ug/l)	20.69 (10–60)

DISCUSSION

Firstly described by Gitelman et al. in three adult patient who had intermittent episodes of muscle weakness and tetany with hypokalemia, hypomagnesemia and hypocalciuria. Gitelman syndrome is now a well-known inherited disorder of renal tubules. The primary pathology in this syndrome is in the renal tubular transport mechanism which caused by mutations in the gene coding for the thiazide-sensitive Na-Cl transporter in the distal tubule [7, 8]. Therefore, physical examination and laboratory findings in this syndrome such as

volume contraction, reduced blood pressure, increased renin activity, hypokalemia, hypomagnesemia and hypocalciuria are consistent with the persistent thiazide diuretic action and exclusion of diuretic usage and other conditions which cause hypokalemia and metabolic alkalosis with a normal or low blood pressure are very important for differential diagnosis. Bartter syndrome is the most important genetic disorder to consider in the differential diagnosis of Gitelman syndrome. Twenty-four hour urine calcium creatinine ratio is useful diagnostic marker to differentiate these two syndromes. Bettinelli et al. showed Bartter and Gitelman syndrome are easily distinguished on the basis of urinary calcium [9]. In Gitelman syndrome there is hypocalciuria, a low ratio of calcium to creatinine in 24 hour urine collection. Primary aldosteronism, which is another cause of hypokalemia and metabolic alkalosis, is usually not in the differential diagnosis since affected patients tend to be hypertensive and have a low plasma renin activity in contrast to elevated values in Bartter and Gitelman syndromes and with vomiting or diuretic use.

In contrast to Bartter syndrome, which is usually diagnosed in infancy or early childhood, Gitelman usually does not affect growth and typically presents in late childhood to adulthood although presentation in infancy has been described [10]. It commonly presents with cramps of the arms and legs, fatigue, ranging from mild to severe, seizures, polyuria and nocturia, chondrocalcinosis, growth retardation if presents younger, and surprisingly hypertension later in life which may be due to prolonged exposure to elevated renin and aldosterone levels [11]. Prolonged Qtc and arrhythmias with resultant palpitations and/or syncope has been estimated to occur in half of affected patients [12]. The prognosis of Gitelman syndrome is generally good, but a few patients with severe hypokalemia especially joined with severe hypomagnesemia and alkalosis may be at risk of developing life-threatening cardiac arrhythmias. Therefore, a detailed cardiac assessment is very important to identify patients at risk of malignant arrhythmias.

The tubular defects in Gitelman syndrome cannot be corrected. As a result, treatment, which must be life-long, is aimed at minimizing the effects of the secondary increases in renin, and aldosterone production and at correcting the volume deficit and electrolyte abnormalities. Potassium replacement, correction of hypomagnesemia, potassium sparing diuretics such as amiloride, triamterene and spironolactone can be used. Angiotensin converting enzyme inhibitors can also be used for hypokalemia control [12]. Correction of hypokalemia may need large amount of potassium especially in the form of potassium chloride due to better absorption (up to 500 mEq/d) [13]. Potassium-sparing diuretics amiloride (5–10 mg/day) and spironolactone (200–300 mg/day) can be used but in hypotensive patients, these drugs should be used cautiously [14]. Hypomagnesemia can contribute the urinary potassium loss. High doses, which are often difficult to tolerate (e.g., diarrhea with magnesium), are required to achieve a substantial elevation in serum potassium or magnesium. Non-steroidal anti-inflammatory drugs are not generally useful in Gitelman syndrome due to normal prostaglandin levels unlike Bartter syndrome.

In the light of above knowledge, patient was diagnosed as Gitelman syndrome. She had acute kidney injury due to hypokalemic rhabdomyolysis. All needed treatments were made as mentioned above and patient was discharged with improved symptoms and normal kidney functions

CONCLUSION

Gitelman syndrome should be thought in the differential diagnosis of hypokalemic rhabdomyolysis although it is a rare disease.

****

Author Contributions

Yasar Yildirim – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Ali Veysel Kara – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Zulfikar Yilmaz – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published
Erdal Bodakci – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published
Vehbi Demircan – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published
Ali Kemal Kadiroglu – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published
Mehmet Emin Yilmaz – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

Authors declare no conflict of interest.

Copyright

REFERENCES

1. Gitelman HJ, Graham JB, Welt LG. A new familial disorder characterized by hypokalemia and hypomagnesemia. Trans Assoc Am Physicians 1966;79:221–35.
2. Kurtz I. Molecular pathogenesis of Bartter's and Gitelman's syndromes. Kidney Int 1998 Oct;54(4):1396–410.
3. Ji W, Foo JN, O'Roak BJ, et al. Rare independent mutations in renal salt handling genes contribute to blood pressure variation. Nat Genet 2008 May;40(5):592–9.
4. Simon DB, Karet FE, Rodriguez-Soriano J, et al. Genetic heterogeneity of Bartter's syndrome revealed by mutations in the K+ channel, ROMK. Nat Genet 1996 Oct;14(2):152–6.
5. Simon DB, Nelson-Williams C, Bia MJ, et al. Gitelman's variant of Bartter's syndrome, inherited hypokalaemic alkalosis, is caused by mutations in the thiazide-sensitive Na-Cl cotransporter. Nat Genet 1996 Jan;12(1):24–30.
6. Riveira-Munoz E, Chang Q, Godefroid N, et al. Transcriptional and functional analyses of SLC12A3 mutations: new clues for the pathogenesis of Gitelman syndrome. J Am Soc Nephrol 2007 Apr;18(4):1271–83.
7. De Jong JC, Van Der Vliet WA, Van Den Heuvel LP, Willems PH, Knoers NV, Bindels RJ. et al. Functional expression of mutations in the human NaCl cotransporter: evidence for impaired routing mechanisms in Gitelman's syndrome. J Am Soc Nephrol 2002 Jun;13(6):1442–8.
8. Vargas-Poussou R, Dahan K, Kahila D, et al. Spectrum of mutations in Gitelman syndrome. J Am Soc Nephrol 2011 Apr;22(4):693–703.
9. Bettinelli A, Bianchetti MG, Girardin E, et al. Use of calcium excretion values to distinguish two forms of primary renal tubular hypokalemic alkalosis: Bartter and Gitelman syndromes. J Pediatr 1992 Jan;120(1):38–43.
10. Tammaro F, Bettinelli A, Cattarelli D, et al. Early appearance of hypokalemia in Gitelman syndrome. Pediatr Nephrol 2010 Oct;25(10):2179–82.
11. Berry MR, Robinson C, Karet Frankl FE. Unexpected clinical sequelae of Gitelman syndrome: hypertension in adulthood is common and females have higher potassium requirements. Nephrol Dial Transplant 2013 Jun;28(6):1533–42.
12. Foglia PE, Bettinelli A, Tosetto C, et al. Cardiac work up in primary renal hypokalaemia-hypomagnesaemia (Gitelman syndrome). Nephrol Dial Transplant 2004 Jun;19(6):1398–402.
13. Shaer AJ. Inherited primary renal tubular hypokalemic alkalosis: a review of Gitelman and Bartter syndromes. Am J Med Sci 2001 Dec;322(6):316–32.
14. Colussi G, Rombolà G, De Ferrari ME, Macaluso M, Minetti L. Correction of hypokalemia with antialdosterone therapy in Gitelman's syndrome. Am J Nephrol 1994;14(2):127–35.

CASE REPORT PEER REVIEWED | OPEN ACCESS

Giant pleomorphic liposarcoma of anterior mediastinum: A rare tumor at a rare site with atypical imaging features

Dandina Mahesh, Chamarthi Madhavi, Jaideep Kumar Trivedi, Vasamsetti Bhushan Rao, Raghava Kashyap, Malla Uma Maheswararao

ABSTRACT

Introduction: Liposarcomas in the mediastinum are very rare, with a reported incidence of less than 1% of all mediastinal tumors. Pleomorphic liposarcoma is the least frequent but most malignant histological subtype of liposarcoma, which has classical imaging features described in literature. Case Report: We report a rare case of mediastinal pleomorphic liposarcoma in an adult patient, presenting with atypical imaging features. Conclusion: The pathological and imaging features of various subtypes of liposarcoma are reviewed.

Keywords: Liposarcoma, Mediastinal tumors, Pleomorphic

Dandina Mahesh[1], Chamarthi Madhavi[1], Jaideep Kumar Trivedi[2], Vasamsetti Bhushan Rao[3], Raghava Kashyap[4], Malla Uma Maheswararao[5]

Affiliations: [1]MBBS, MD, Department of Radiodiagnosis, Maharajah's Institute of Medical Sciences, Vizianagaram, Andhra Pradesh, India; [2]MBBS, MS, Mch, Department of Cardiothoracic Surgery, Queens NRI Hospital, Visakhapatnam, Andhra Pradesh, India; [3]MBBS, MD, Department of Pathology, Maharajah's Institute of Medical Sciences, Vizianagaram, Andhra Pradesh, India; [4]MBBS, MD, Department of Nuclear Medicine and PET, Mahatma Gandhi Cancer Hospital and Research Institute, Visakhapatnam, Andhra Pradesh, India; [5]MBBS, DMRD,DNB, Department of Radiodiagnosis,Andhramedical college, Visakhapatnam, Andhra Pradesh, India.

Corresponding Author: Dr. Dandina Mahesh, Department of Radiodiagnosis, Maharajah's Institute of Medical Sciences, Vizianagaram, Andhra Pradesh 535217, India; Ph: 09177015555; Email: maheshdandina@gmail.com

Received: 23 January 2015
Accepted: 18 February 2015
Published: 01 October 2015

How to cite this article

Mahesh D, Madhavi C, Trivedi JK, Maheswararao MU, Rao VB, Kashyap R. Giant pleomorphic liposarcoma of anterior mediastinum: A rare tumor at a rare site with atypical imaging features. Int J Case Rep Images 2015;6(10):622–626.

doi:10.5348/ijcri-2015101-CR-10562

INTRODUCTION

Liposarcomas are malignant adiopocytic tumors of mesenchymal origin. They are the second most common type of soft tissue sarcomas after malignant fibrous histiocytoma, and constitute 15–20% cases of all soft tissue sarcomas which are commonly seen in extremities and in retroperitoneum. They are very rarely located in mediastinum, accounting for only less than 1% of all mediastinal tumors, the occurrence of pleomorphic variety of liposarcoma being even rarer. To the best of our knowledge, approximately 200 cases of liposarcomas are reported in mediastinum, in which only 20 to 25 cases categorized as pleomorphic variety. We report a case of giant Pleomorphic liposarcoma in anterior mediastinum extending into hemithorax on both the sides in an adult patient.

CASE REPORT

A 39-year-old male patient came to the outpatient department of Pulmonary Medicine, with chest discomfort and mild breathlessness of three months duration. There are no other cardiorespiratory symptoms like cough, fever, chest pain, palpitation or pedal edema.

The radiograph of chest showed large ill defined radiodense opacity, centered at mediastinum and extending into hemithorax on both sides (Figure 1a). On

lateral radiograph, there is obliteration of retrosternal clear space indicating the anterior mediastinal location of mass lesion (Figure 1b).

Plain and contrast computed tomography (CT) scan of chest showed a large heterogeneous mass in anterior mediastinum (Figure 2a). The mass lesion was seen extending into anterior half of right hemithorax, from apex to diaphragm with partial collapse of right lung (Figure 2c). On left side, the mass is occupying most of the hemithorax, causing collapse of lower lobe and lingular segments of left lung (Figure 2c).

The mass showed predominantly fatty component with interspersed soft tissue component and few cystic areas and a dense nodular calcifications (Figure 2c). There is mild heterogeneous enhancement of the soft tissue component on IV contrast administration (Figure 2b, 2d).

The MRI scan confirmed the same findings and clearly depicted the fatty, soft tissue and cystic components (Figure 3). Encasement of the vessels was seen. There was pericardial invasion on left side.

Preoperative CT scan guided biopsy was done from the soft tissue component, which was not conclusive. Surgical exploration was done by mid sternotomy and the mass was excised. Partial pericardectomy was done. The mass adherent to the left chest wall was released and left lung was inflated. The total dimensions of the mass measured 34x26x17 cm, weighing 4.2 kg (Figure 4). The patient was given adjuvant chemotherapy.

Pathological examination of the surgical specimen showed mature adipose tissue with multivacuolated lipoblast and spindle shaped cells arranged in fascicular pattern with multinucleated histoid giant cells (Figure 5). These findings are consistent with pleomorphic liposarcoma.

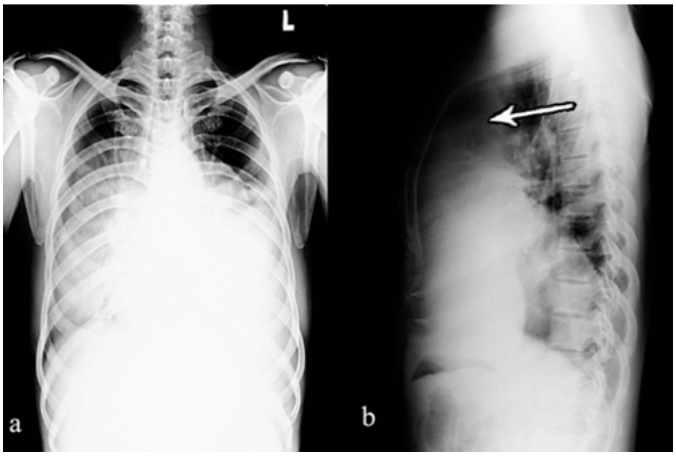


Figure 1: (a) Posteroanterior radiograph of chest showing large ill defined opacity involving mediastinum, extending into hemithorax on both sides, (b) Lateral radiograph of chest showing radiodense opacity filling the retrosternal space (arrow).

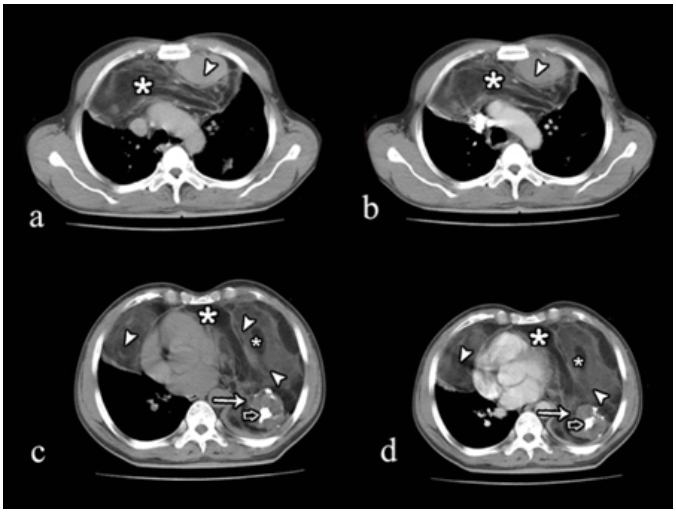


Figure 2: Axial computer tomography scan images of chest (a) plain and (b) contrast at the level of arch of aorta showing heterogeneous mass with fat (*) and soft tissue (arrow head) attenuation in anterior mediastinum with mild enhancement of solid component on contrast administration. Axial CT images of chest (c) plain and (d) contrast at the level of ventricles: showing heterogeneous mass with solid component (arrow head), fat (*), cystic areas (solid arrow) and nodular calcification (open arrow), occupying anterior half of right hemithorax and complete left hemithorax.

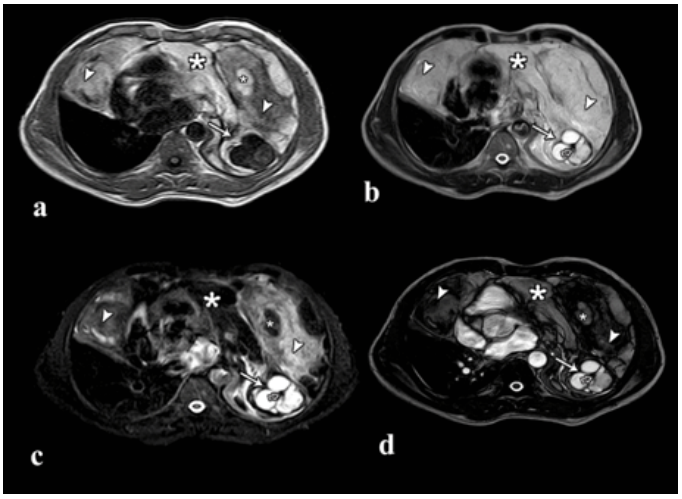


Figure 3: (a) Axial MRI T1W scan, (b) T2W scan, (c) Fat suppressed T2W and (d) BTFE images at the level of ventricles showing heterogeneous mass with solid component (arrow head), fat (*), cystic area (solid arrow) and nodular calcification (open arrow), occupying anterior half of right hemithorax and complete left hemithorax with pericardial invasion.

DISCUSSION

Liposarcomas are malignant mesenchymal neoplasm which usually occur in adults during 5th to 6th decade, Well differentiated type is the most common. Liposarcomas are rare in children. Few cases are reported in literature [1, 2]. The most common variant in children

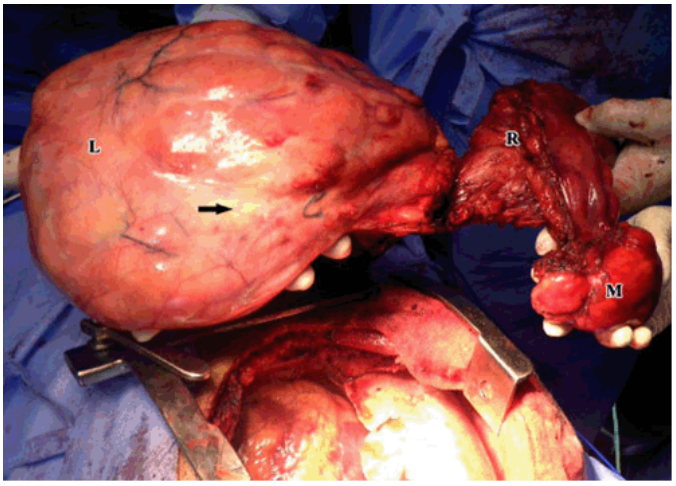


Figure 4: Intraoperative photograph showing large soft tissue mass interposed with adipose tissue (arrow); left side of the mass (L), right side of the mass (R) and mediastinal component (M).

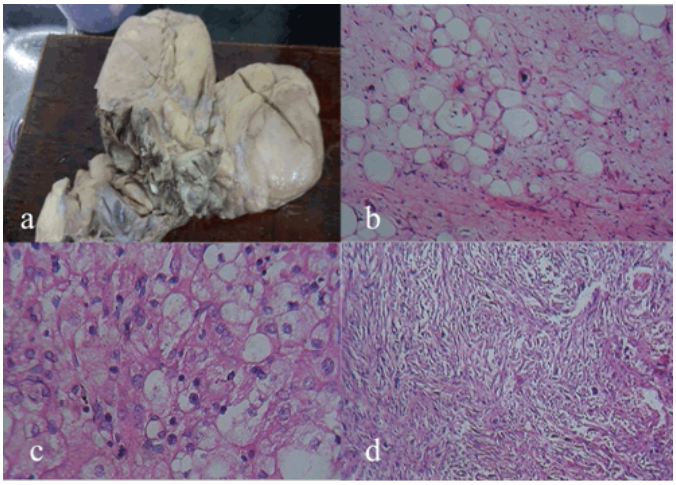


Figure 5: (a) Gross specimen showing adipose tissue, (b) Vacuolated pleomorphic lipoblast in tumor, (c) Pleomorphic round cells in tumor, and (d) MFH like area in pleomorphic liposarcoma.

is myxoid type [1]. Liposarcomas are classified by WHO in 2002, based on histological appearance into five sub groups (1) well differentiated, (2) dedifferentiated, (3) myxoid, (4) pleomorphic, and (5) mixed.

Clinically, patients are asymptomatic in initial stages, as the tumor grows in size, it results in nonspecific chest pain and breathlessness. Symptoms related to compression or invasion of adjacent organs may occur.

Chest radiography usually shows mediastinal widening or mass lesion. The findings of cross-sectional imaging depend on the histological type of liposarcoma.

Well differentiated liposarcoma is the least malignant variety of all liposarcomas, not usually associated with metastasis, and predominantly fat containing mass, constitutes more than 75% of tumor volume. The non fatty component is generally visualized as thick soft tissue with septations of more than 2 mm width, with occasional nodularity and calcifications. Well differentiated liposarcoma is also called atypical lipomatous tumor when it is superficial in location [3].

Dedifferentiated liposarcoma is a biomorphic neoplasm in which foci of high grade sarcoma arise within the well differentiated liposarcoma. So, it shares most of the imaging appearance of well differentiated liposarcoma, except that it displays a non-lipomatous component of at least 1 cm size, indicating the focus of dedifferentiation with a density similar to that of skeletal muscle. On contrast enhanced MRI scan, areas of dedifferentiation show heterogeneous or nodular enhancement, whereas the fat necrosis shows thin peripheral enhancement. Dedifferentiated liposarcoma is commonly located in retroperitoneum, mediastinum and inguinal regions, but uncommon in extremities [4].

Myxoid variety of liposarcoma shows predominantly gelatinous mass constituting fat less than 10% of tumor volume. The lesion may simulate a cyst on both CT and MRI scan, whereas ultrasonography is more useful in depicting the hypoechoic solid nature of tumor. The fatty component is seen as septations or small solid nodules within the lesion. The MRI scan is more sensitive in demonstrating the lipomatous component, with fat suppressed images being even more helpful. Extra-pulmonary metastases are more common in myxoid liposarcoma [5].

Pleomorphic liposarcoma is the most malignant variety, which has highest rate of recurrence and metastases [6]. It is seen as relatively well defined heterogeneous soft tissue mass with occasional areas of necrosis and hemorrhages. Being the most aggressive type of liposarcoma, it has the least amount of adipose tissue, when compared to other types of liposarcoma [4]. Due to relatively low quantity of adipose tissue, this tumor poses a difficulty in diagnosis on imaging and histopathology. Again MRI scan plays a major role in the detection of this minimal fatty component. Taking adequate tissue samples from both adipose and non adipose tissues is the key to accurate histopathological diagnosis [5].

Based on imaging manifestations of all these subtypes of liposarcoma, the unique feature to help in the diagnosis of all liposarcomas is the presence of fat. But the differentiation into subtypes depends on relative proportions and appearance of different constituents of the tumor. The other differentials of fat containing mediastinal masses are teratoma, thymolipoma, lipoma and pericardial fat pad.

In contrast to the classical imaging description of pleomorphic liposarcoma, the present case showed large amount of fatty tissue along with soft tissue component on both CT scan and MRI scan, and few cystic areas with dense nodular calcification. Mediastinal teratoma can be considered as a close differential diagnosis in present case, as it contains lipomatous, soft tissue, cystic components and calcifications.

Imaging plays a major role in determining the extent of tumor, involvement of adjacent organs, the evaluation of metastases and is of utmost help in guiding the

biopsy, to collect tissue from both lipomatous and non-lipomatous components, which is important for accurate pathological diagnosis and planning of management.

Treatment and prognosis of liposarcomas mainly depend on histological grading, the anatomical location and extent of tumor. Complete surgical removal is the main stay of treatment for liposarcomas. For deep-seated lesions where complete surgical removal is not possible, surgery combined with radiotherapy will reduce local recurrence [7]. Chemotherapy may be added as adjuvant to surgical excision in histologically aggressive tumor subtypes like pleomorphic liposarcoma, which helps improve the survival rate [8]. In present case after surgery patient underwent six cycles of chemotherapy with doxorubicin and ifosfamide and no recurrence was found in six months follow-up.

Usually, well differentiated liposarcomas have good prognosis with no metastatic potential. Myxoid liposarcomas prognosis depends on round cell component in the tumor. In pleomorphic liposarcoma, recurrence and metastases are more common with a five-year survival rate of 40–50% [7].

CONCLUSION

Mediastinal liposarcomas constitute a very rare variety of mediastinal tumors. They have a wide spectrum of imaging manifestations depending on the histological subtype. However, presence of a lipomatous component is the most important clue for the diagnosis. Our case discusses an atypical presentation of pleomorphic liposarcoma, which showed a large fatty component on imaging. This emphasizes the need of image guided biopsy and careful pathological examination of both lipomatous and non lipomatous components of tumor for proper pre-operative diagnosis.

****

Author Contributions

Dandina Mahesh – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Final approval of the version to be published

Chamarthi Madhavi – Substantial contributions to conception and design, Analysis and interpretation of data, Drafting the article, Final approval of the version to be published

Jaideep Kumar Trivedi – Acquisition of data, Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

Malla Uma Maheswararao – Acquisition of data, Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

Vasamsetti Bhushan Rao – Acquisition of data, Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

Raghava Kashyap – Substantial contributions to conception and design, Analysis and interpretation of data, Drafting the article, Final approval of the version to be published

Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

Authors declare no conflict of interest.

REFERENCES

1. Alaggio R, Coffin CM, Weiss SW, et al. Liposarcomas in young patients: a study of 82 cases occurring in patients younger than 22 years of age. Am J Surg Pathol 2009 May;33(5):645–58.
2. Saeed M, Plett S, Kim GE, Daldrup-Link H, Courtier J. Radiological-pathological correlation of pleomorphic liposarcoma of the anterior mediastinum in a 17-year-old girl. Pediatr Radiol 2010 Dec;40 Suppl 1:S68–70.
3. Fletcher CDM, Unni KK, Mertens F. Eds. Adipocytic tumors. In: WHO classification of tumors. Pathology and genetics: Tumors of soft tissue and bone. Lyon, France: IARC Press; 2002:19–46.
4. Murphey MD, Arcara LK, Fanburg-Smith J. From the archives of the AFIP: imaging of musculoskeletal liposarcoma with radiologic-pathologic correlation. Radiographics 2005 Sep-Oct;25(5):1371–95.
5. Pearlstone DB, Pisters PW, Bold RJ, et al. Patterns of recurrence in extremity liposarcoma: implications for staging and follow-up. Cancer 1999 Jan 1;85(1):85–92.
6. Downes KA, Goldblum JR, Montgomery EA, Fisher C. Pleomorphic liposarcoma: a clinicopathologic analysis of 19 cases. Mod Pathol 2001 Mar;14(3):179–84.
7. Oliveira AM, Nascimento AG. Pleomorphic liposarcoma. Semin Diagn Pathol 2001 Nov;18(4):274–85.
8. Eilber FC, Eilber FR, Eckardt J, et al. The impact of chemotherapy on the survival of patients with high-grade primary extremity liposarcoma. Ann Surg 2004 Oct;240(4):686–95; discussion 695–7.

CASE REPORT PEER REVIEWED | OPEN ACCESS

Adamantinoma of tibia: A case report

Rahadyan Magetsari, Yudha M. Sakti, Asa I. Z. Asikin, Zikrina A. Lanodiyu, Punto Dewo

ABSTRACT

Introduction: Adamantinoma is one of the rarest low-grade malignant bone tumors, representing 0.4% of them with only around 300 documented case of it. Case Report: We reported a rare case of primary adamantinoma of the tibia in a 35-year-old Indonesian mongoloid female which recurred after excision of the primary tumor seven years prior to admission. Conclusion: Adamantinoma is hard to metastasize except in the case of repeated and unsatisfying removal procedure. Wide local excision with a substantial margin of normal bone can be applied if early diagnosis is successfully made. If there has been more than one recurrence or in large tumor with extension to the surrounding soft tissues, radical resection or amputation is advisable. After radical treatment, there is a high percentage of healing. The main factor of recurrence is incomplete resection.

Keywords: Adamantinoma, Bone tumors, Primary adamantinoma, Tibia, Recurrence

Rahadyan Magetsari[1], Yudha M. Sakti[1], Asa I. Z. Asikin[1], Zikrina A. Lanodiyu[1], Punto Dewo[1]

Affiliations: [1]Department of Orthopaedics and Traumatology, Dr Sardjito General Hospital/ Faculty of Medicine, Universitas Gadjah Mada, Yogyakarta, Indonesia.

Corresponding Author: Zikrina A. Lanodiyu, MD, Department of Orthopaedics and Traumatology, Sardjito General Hospital / Universitas Gadjah Mada, Jalan Kesehatan no. 1, Yogyakarta 56281, Indonesia; Mob: +62 81 2277 2025; Ph: +62 274 515054; Fax: +62 274 553143; E-mail: zikrina.lanodiyu158@gmail.com

Received: 13 May 2015
Accepted: 06 August 2015
Published: 01 October 2015

How to cite this article

Magetsari R, Sakti YM, Asikin AIZ, Lanodiyu ZA, Dewo P. Adamantinoma of tibia: A case report. Int J Case Rep Images 2015;6(10):627–631.

doi:10.5348/ijcri-2015102-CR-10563

INTRODUCTION

Adamantinoma is one of the rarest low-grade malignant bone tumors, representing around 0.4% of them [1]. It was described initially by Fisher in 1913 and occurs most commonly between 10 and 50 years of age with slight male predominance. It was commonly found in the centerpart of long bone. However, some cases happened in other parts of the bone [2]. Around 90% of the cases there were reported previously was found in long tubular bones and 80% of it was found in the tibial mid shaft. Other long bones not uncommonly affected are the humerus, ulna, femur, fibula and radius. Early on it is confined to bone, furthermore there may be an extension inwards to the medullary canal or outwards beyond the periosteum. Pain is the most common clinical manifestation and local swelling is the common second clinical sign to appear. Distant metastases have been described to occur many years after the presentation of the primary lesion [2, 3] The incidence of recurrence is approximately 30% and those of metastases ranges between 10% and 20% [4]. We presented a case of adamantinoma of tibia with pneumothorax and suspected pulmonary metastasis with recurrence of the primary tumor after excision six years prior to admission.

CASE REPORT

A 35-year-old Indonesian mongoloid female patient presented with a painful lump in her left leg. She had history of lump in her left leg since seven years prior to admission. Initially, the lump was only 1 cm in diameter. There was no history of trauma. The patient was initially treated by general surgeon, and had undergone excision of the lump. After the surgery, recurrence occurred and the lump developed gradually. Within seven years, the patient did not routinely visit hospital to have the condition of the lump checked. One week prior to admission, the patient complained about difficulty in breathing. She came to a primary hospital and was found to have a pleural effusion in her right lung and chest tube was inserted. She was then referred to our hospital.

On the physical examination, a 10x10x5 cm hard tender mass was found in the left lower leg (Figure 1). Range of movement (ROM) in the left knee joint was full, but in the ankle joint, the ROM was limited. Laboratory results including tumor marker were unremarkable. Plain radiograph of the left leg showed lytic lesion in diaphysis and metaphysis of the left tibia with osteodestruction of tibia and extension to the surrounding soft tissue (Figure 2).

A chest radiograph was performed (Figure 3) which showed bullae in the superior lobe of the right lung, pneumothorax, fibroatelectasis in the middle lobe of the right lung, and right pleural effusion. A chest MSCT showed hydropneumothorax and multiple bullae of the right lung and solitary nodule in the left lung suspected to be a distant metastasis from the primary tumor site (Figure 4). The patient was assessed with hydropneumothorax of the right lung caused by possible malignancy.

A fine needle aspiration biopsy (FNAB) of the lesion in the left leg then performed with the result of clustered tumor cells within a group, with small cells

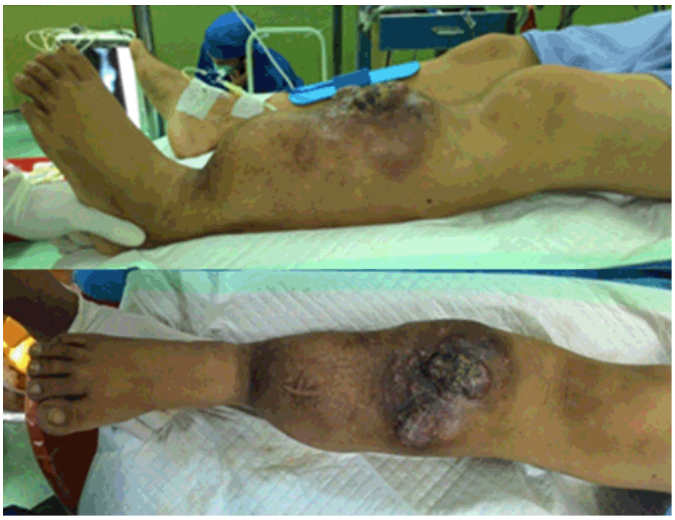


Figure 1: Clinical photograph of the lesion in the left leg showing a mass extending from middle third of the left leg to distal third of the left leg with swelling, redness, and skin defect.

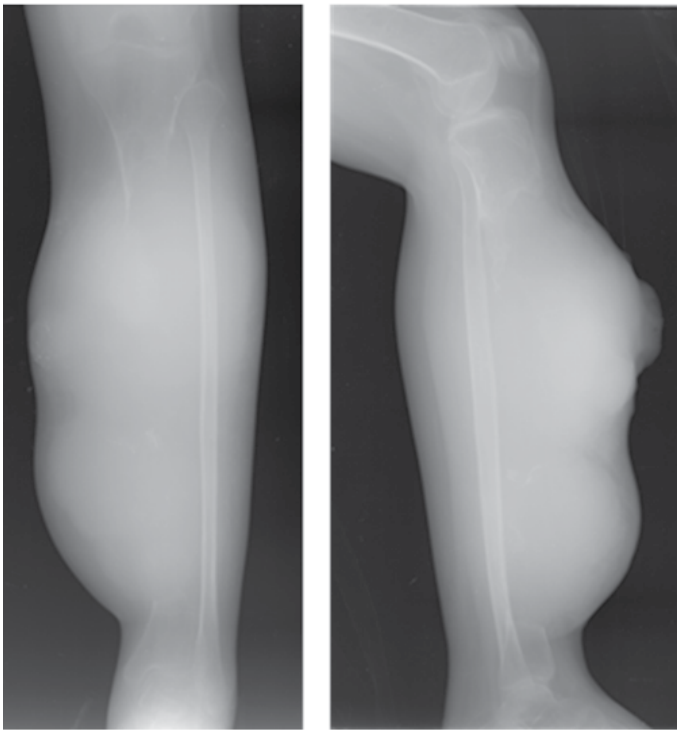


Figure 2 : Anterior and lateral X-ray of the left leg showing a lytic lesion in diaphysis and metaphysis of the left tibia with osteodestruction of tibia and extension to the surrounding soft tissue

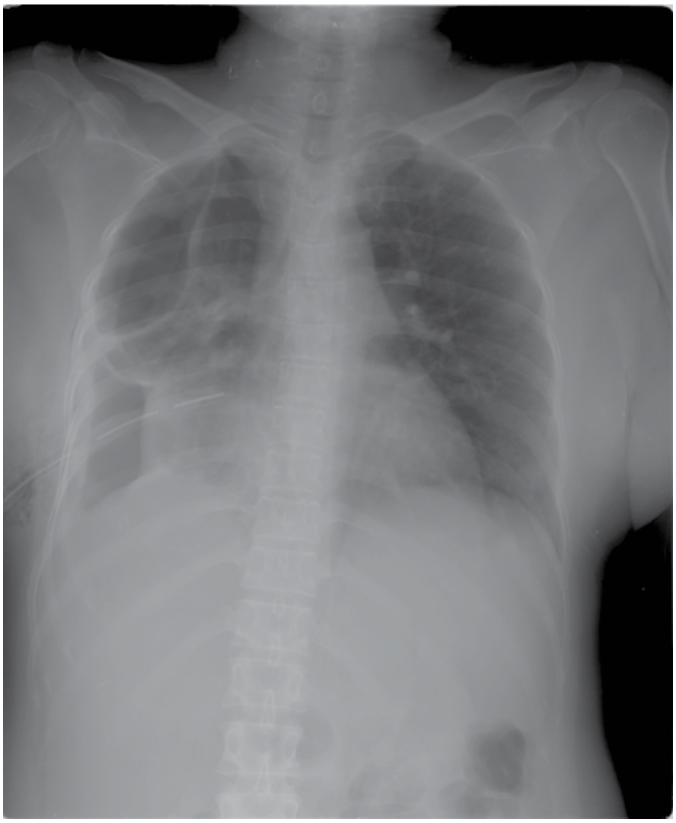


Figure 3 : Chest X-ray showing bullae in the superior lobe of the right lung, pneumothorax, fibro-atelectasis in the middle lobe of the right lung, and right pleural effusion.

with atypia, small amount of cytoplasm, spindle nucleus with hyperchromasia with background of lymphocyte and erythrocyte (Figure 5). A clinicopathological conference was then arranged with a recommendation of transfemoral amputation. A transfemoral amputation was then performed and the histopathological result showed a pattern of epithelial tumor with solid arrangement and infiltration to surrounding tissue, including to the upper dermis. The observation of the cells showed small to moderate cell sizes, small amount of cytoplasm, round to spindle nucleus with small amount of mitosis. The conclusion of the histopathological examination was islands of epithelial cells in a densely-populated stroma of spindle cells suggesting adamantinoma (Figure 6). The positive cytokeratin was found in the immunohistochemistry on the tumor cells and it was also relevant with the diagnosis of adamantinoma (Figure 7).

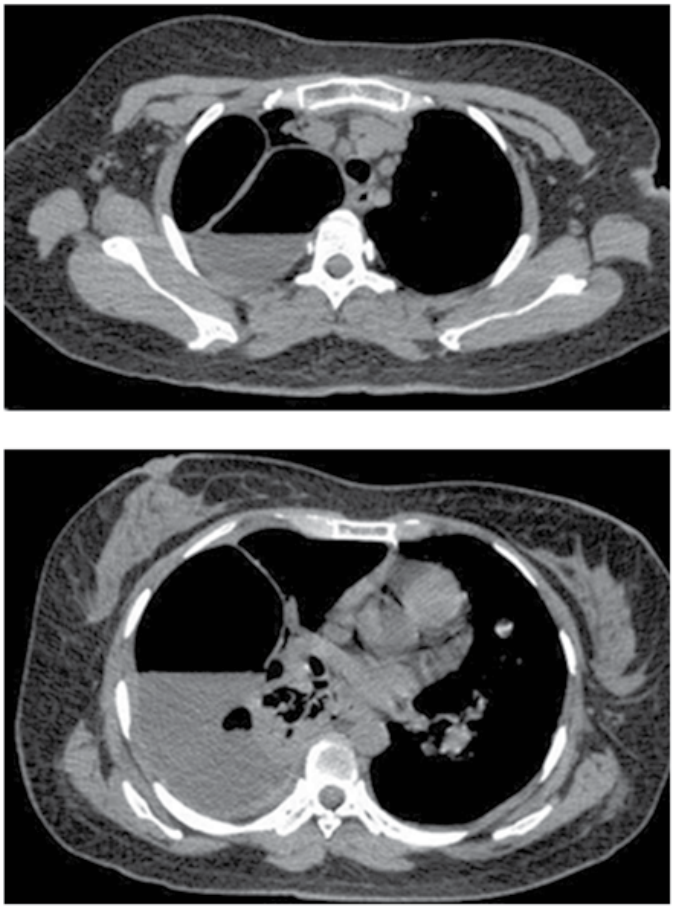


Figure 4 : Chest MSCT showing hydropneumothorax and multiple bullae of the right lung and solitary nodule in the left lung

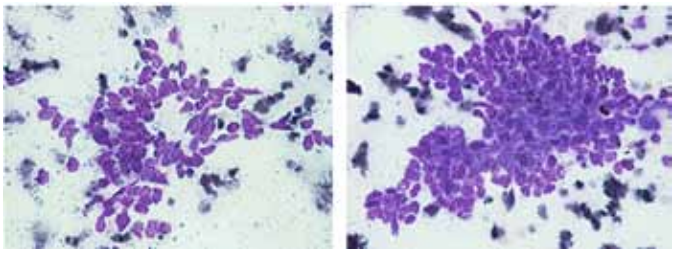


Figure 5 : FNAB of the lesion in the left leg showing clustered tumor cells within a group, with small cells with atypia, small amount of cytoplasm, spindle nucleus with hyperchromasia with background of lymphocyte and erythrocyte, and it showed malignant small round to spindle blue cell tumor suggesting soft tissue origin.

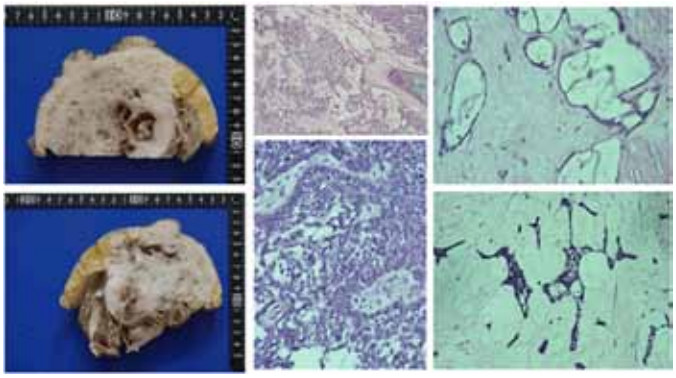


Figure 6 : Histopathological examination of the lesion showing a pattern of epithelial tumor with solid arrangement and infiltration to surrounding tissue, including to the upper dermis. The observation of the cells showed small to moderate cell sizes, small amount of cytoplasm, round to spindle nucleus with small amount of mitosis

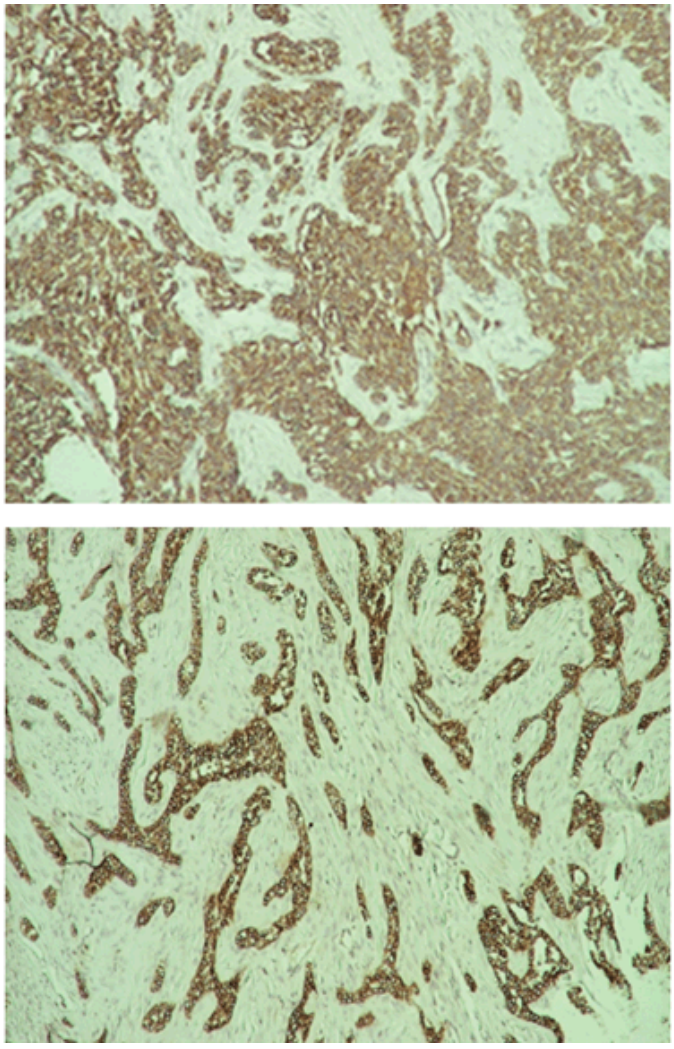


Figure 7 : Immunohistochemistry with cytokeratin was positive suggesting adamantinoma.

DISCUSSION

The symptoms initially present with nonspecific characteristics and its variety of the symptoms is related to location and extent of the disease. Pain is the most common clinical manifestation reported. It was in accordance to this case where the patient was presented with painful lump on her left lower leg. The onset of the clinical sign and symptom is insidious with slow and progressive characteristic. The patient usually tolerates symptoms for many years before seeking medical attention because of this characteristic of clinical manifestation as seen in this patient [5]. However, some of the patients present with swelling with or without pain as local swelling being the second most common symptom to appear. In addition, this case also showed that involvement of the anterior tibial surface can produce bowing of the tibia [6].

Based on radiographic assessment, adamantinoma is an osteolytic type with mono or multi-loculated lesions. In accordance to its benign characteristics, it is well circumscribed with septa and a peripheral condensation may appear. It is usually appear in the diaphysis or metaphysis of the anterior tibial. The lesion usually grows intracortical and it may spread longitudinally. However, it does not rule out the possibility of cortex destruction and marrow cavity invasion of the tumor [7]. The MSCT examination could reveal the characteristics of the lesion better than plain radiograph and can also detect another tumor site invisible on plain radiographs. The differential diagnosis based on plain radiograph examination including fibrous dysplasia and osteofibrous dysplasia [2].

The patient in this case had tumor in her left leg which develops progressively after the first excision seven years ago. This might be due to inadequate attempts of removal where incomplete resection may result in higher recurrence rate up to 30% in the period of 85 months. Therefore, adamantinoma is suggested to be treated with wide surgical excision and reconstruction or amputation since its likelihood of healing is high subsequent to radical treatment [2, 3]. If early diagnosis is successfully made, wide local excision with a substantial margin of normal bone is still acceptable [8]. However, if there has been more than one recurrence or in large tumor with extension to the surrounding soft tissues, radical resection or amputation is advisable as shown in this patient where knee amputation was performed [9].

Adamantinoma is a low-grade malignant tumor of epithelial origin which metastasizes late. This tumor is insensitive to radiation and has capabilities of metastasis, especially to the lung. [8]. Metastases including in lung or lymph nodes are rare and can occur in 10–20% of patients. Pulmonary metastases are more common that regional nodal metastases from this tumor. It is not uncommon to develop distant metastases even up to 10 years after detection of the primary [3, 5]. This was in accordance with this case, where the main reason to seek medical management was due to difficulty in breathing and it was correlated with repeated local recurrences that might be due to inadequate primary excision of the tumor. However, it is difficult to evaluate its true metastatic potential due to several reasons such as long term follow-up, low number of the case, and slow growing nature of the tumor [5]. Several reports in literature have described metastasectomy for pulmonary lesions in both curative and palliative settings with good results. Mean survival of patients with metastatic disease is reported to be 12 years. There appears to be no definitive role for radiotherapy or chemotherapy [10].

Due to its excellent prognosis, it is crucial to diagnose this rare bone tumor in the early stage. It can be achieved by histologic examination, where this tumor can be distinguished easily. However, not only this tumor is rare, the heterogeneity of the tumor presentation may lead to confusion in some cases. On the histologic examination with inadequate sample taking, epithelial component may be seen only focally in the differentiated adamantinoma case, therefore in some cases, extensive sampling of the tumor is mandatory [11]. The challenge in making diagnosis and preparing proper management emphasizes that clinicopathological conference among orthopedic surgeon, pathologist, and radiologist was very important to ensure the patient get the best treatment available in musculoskeletal tumor cases. At the moment where the diagnosis is certain, resection with wide surgical margins or amputation can be applied to the patient.

CONCLUSION

We reported a rare case of recurrent tibial adamantinoma of the left lower leg with pulmonary metastases managed by knee amputation. Appropriate diagnosis and treatment plan through clinicopathological conference is mandatory to ensure the patient receive the best management in musculoskeletal tumor cases.

****

Author Contributions

Rahadyan Magetsari – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

Yudha M. Sakti – Acquisition of data, Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

Asa IZ Asikin – Acquisition of data, Drafting the article, Final approval of the version to be published

Zikrina A. Lanodiyu – Substantial contributions to conception and design, Drafting the article, Final approval of the version to be published

Punto Dewo – Substantial contributions to conception and design, Analysis and interpretation of data, Drafting

the article, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor
The corresponding author is the guarantor of submission.

Conflict of Interest
Authors declare no conflict of interest.

Copyright

REFERENCES

1. Huvos AG. Adamantinoma of extragnathic bones. In: Bone Tumors. Diagnosis. Treatment and Prognosis, Huvos AG, ed. Philadelphia: WB. Saunders Company; 1991. p. 677–93.
2. Khémiri C, Mrabet D, Mizouni H, et al. Adamantinoma of the tibia and fibula with pulmonary metastasis: an unusual presentation. BMJ Case Rep 2011 Oct 16;2011. pii: bcr0620114318.
3. Ramesh R, Burrah R, Thambuchetty N, Shivakumar K, Ananthamurthy A, Manjunath S. Adamantinoma of the tibia: a case report. Indian J Surg Oncol 2012 Sep;3(3):239–41.
4. Filippou DK, Papadopoulos V, Kiparidou E, Demertzis NT. Adamantinoma of tibia: a case of late local recurrence along with lung metastases. J Postgrad Med 2003 Jan-Mar;49(1):75–7.
5. Moon NF, Mori H. Adamantinoma of the appendicular skeleton--updated. Clin Orthop Relat Res 1986 Mar;(204):215–37.
6. Qureshi AA, Shott S, Mallin BA, Gitelis S. Current trends in the management of adamantinoma of long bones. An international study. J Bone Joint Surg Am 2000 Aug;82-A(8):1122–31.
7. Szendroi M, Antal I, Arató G. Adamantinoma of long bones: a long-term follow-up study of 11 cases. Pathol Oncol Res 2009 Jun;15(2):209–16.
8. Solomon L, Warwick DJ, Nayagam S. Appley's System of Orthopaedics and Fractures, 8th ed. London: Arnold; New York: Co-published in USA by Oxford University Press Inc; 2001.
9. Hatori M, Watanabe M, Hosaka M, Sasano H, Narita M, Kokubun S. A classic adamantinoma arising from osteofibrous dysplasia-like adamantinoma in the lower leg: a case report and review of the literature. Tohoku J Exp Med 2006 May;209(1):53–9.
10. Gonçalves R, Saad Junior R, Dorgan Neto V, Botter M. A rare case of pneumothorax: metastatic adamantinoma. J Bras Pneumol 2008 Jun;34(6):425–9.
11. Jain D, Jain VK, Vasishta RK, Ranjan P, Kumar Y. Adamantinoma: a clinicopathological review and update. Diagn Pathol 2008 Feb 15;3:8.

CASE REPORT PEER REVIEWED | OPEN ACCESS

Acute coronary syndrome associated with phencyclidine use

Hakeem Ayinde, Robert Solomon, Maria-Elise Sanchez, James Diggs, Prafulla Mehrotra

ABSTRACT

Introduction: Many studies have documented the deleterious effects of psychoactive substances like cocaine and amphetamines on the coronary vasculature. However, the impact of phencyclidine (PCP) on the arteries of the heart has largely gone unrecorded. Case Report: We report a case of a 41-year-old female presented to our hospital with chest heaviness, shortness of breath, and nausea, which started at rest and lasted about 30 minutes. Her symptoms resolved on arrival to the emergency room. Electrocardiogram showed transient T wave inversions in V2 and V3 leads, and troponins peaked at 1.01 ng/ml 6 hours after arrival. She received standard therapy for non-ST elevation myocardial infarction. An urgent cardiac catheterization revealed severe vasospasm in 3 cm length of the proximal left anterior descending artery, and milder vasospasm in the mid-portion of the artery; spasm resolved after multiple doses of intracoronary nitroglycerin. We excluded the presence of common precipitants of coronary vasospasm. However, the patient admitted to phencyclidine use about two hours prior to the onset of symptoms, and a urine toxicology screen was positive only for the drug. Given the strong temporal relationship of symptoms to PCP use and absence of common precipitants of coronary vasospasm, we concluded that her coronary spasms were induced by PCP. Conclusion: We describe a case of acute coronary syndrome in a low risk patient thought to be induced by PCP. Our case illustrates the need for physicians to be aware of PCP ingestion as a possible cause of coronary artery spasm when presented with a young adult patient suffering from acute coronary syndrome.

Keywords: Chest pain, Coronary vasospasm, Phencyclidine

How to cite this article

Ayinde H, Solomon R, Sanchez ME, Diggs J, Mehrotra P. Acute coronary syndrome associated with phencyclidine use. Int J Case Rep Images 2015;6(10):632–635.

doi:10.5348/ijcri-2015103-CR-10564

Hakeem Ayinde[1], Robert Solomon[2], Maria-Elise Sanchez[2], James Diggs[3], Prafulla Mehrotra[4]

Affiliations: [1]Resident Physician, Department of Medicine, Howard University Hospital, Washington, DC; [2]Medical Student, Howard University College of Medicine, Washington DC; [3] Associate Professor, Division of Cardiology, Department of Internal Medicine, Howard University Hospital, Washington DC; [4]Professor and Interim Chair, Division of Cardiology, Department of Internal Medicine, Howard University Hospital, Washington DC.

Corresponding Author: Hakeem Ayinde 654 Girard Street NW apt 301 Washington, DC, USA, 20001; Ph: +1 347 746 1592; Fax: 2028657853; E-mail: Hayinde3@gmail.com

Received: 07 May 2015
Accepted: 12 June 2015
Published: 01 October 2015

INTRODUCTION

Phencyclidine (PCP), a synthetic compound originally intended to be used as an anesthetic drug, has been abused throughout the United States for several decades. Its prevalent use has especially been noted in Washington DC where in 2012, up to 12% of male arrestees tested positive for the drug, compared to a 1% rate in other major cities in the country [1]. While PCP is principally noted for its

hallucinogenic effects, it has also been reported to cause such cardiovascular effects as tachycardia, hypertension, and rarely cardiac arrest [2]. Even though animal studies have implicated PCP as a cause of coronary vasospasm, this has not been documented in humans to the best of our knowledge.

We discuss the case of a young adult woman at low risk for coronary artery disease, who presented with acute coronary syndrome due to coronary vasospasm in the setting of acute PCP ingestion.

CASE REPORT

A 41-year-old African American woman presented to the emergency department complaining of chest heaviness, palpitations, shortness of breath, and nausea at rest, which lasted about 30 minutes. She had two similar episodes in the previous month, and these resolved without therapy. Her only cardiovascular risk factor was smoking of a one-fourth pack of cigarettes per day. Her current medications included only vitamins and iron. Symptoms had improved on arrival to the emergency room, and vital signs were within normal limits with a blood pressure of 105/58 mmHg and pulse rate of 85 beats/minute. Physical examination was normal. Electrocardiogram showed transient T wave inversion in both leads V2 and V3 (Figure 1). Laboratory studies revealed rising troponin levels that peaked at 1.01 ng/ml 6 hours after arrival. She was diagnosed with non-ST elevation MI and given standard therapy of aspirin, clopidogrel, and enoxaparin, and she was subsequently prepared for urgent coronary angiography.

Coronary angiography revealed severe vasospasm in 3 cm length of the proximal left anterior descending artery, as well as a milder vasospasm in the mid-portion of the vessel (Figure 2A). Three doses of 200 μg intracoronary nitroglycerin were needed to relieve the vasospasm, after which there were no areas of flow-limiting stenosis identifiable in the coronary arteries (Figure 2B). Further history revealed PCP ingestion about 2 hours prior to the onset of symptoms, and a urine drug screen was positive only for phencyclidine. The patient was discharged on sublingual nitroglycerin as needed, and advised to avoid PCP and other drugs of abuse in the future.

DISCUSSION

To the best of our knowledge, this is the first reported association between PCP ingestion and acute coronary syndrome. Our patient was a young adult woman at low risk for coronary artery disease who presented with chest pain at rest and was found to have severe coronary spasm that was reversed by nitroglycerin during angiography. Urine toxicology screen was positive for PCP, but negative for other stimulants such as cocaine or amphetamine. We considered and excluded other common triggers for

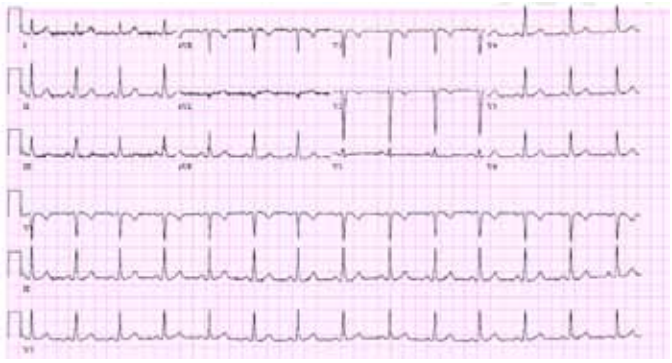


Figure 1: Initial electrocardiogram showing transient T wave inversions in V2 and V3

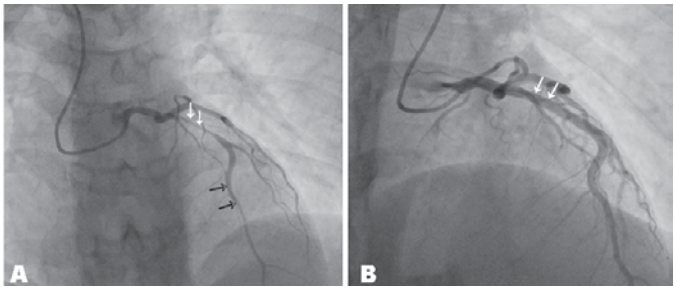


Figure 2: (A) Antero-posterior, cranial view of coronary angiogram showing severe spasm in proximal Left anterior descending (white arrows) and milder stenosis in the more distal portions artery (black arrows). (B) Resolution of spasm (arrows) after intracardiac nitroglycerin.

coronary spasm (Table 1) [3, 4] . She had a history of cigarette smoking but she did not smoke any cigarettes prior to onset of symptoms on the day she presented. We suggest that the coronary vasospasm may have been caused by PCP ingestion since her symptoms started only about 2 hours after smoking PCP.

PCP (1-(1-phenylcyclohyxyl) piperidine hydrochloride) is a noncompetitive NMDAR (N-methyl-D-aspartate receptor) antagonist initially introduced in the 1950s as a dissociative anesthetic agent but withdrawn because of its prolonged unmanageable side effects [5]. It is now a commonly abused street drug in major US cities, especially Washington DC [1].

The major cardiovascular effects in cases of PCP toxicity are hypertension and tachycardia [2], probably due to the inhibition of neuronal catecholamine reuptake or potentiation of noradrenaline release [6].

Although undocumented in humans, animal studies have demonstrated the effects of PCP on coronary vasculature [7–10]. In vitro, PCP appeared to have a vasoconstrictive effect on the coronary arteries in pigs and dogs. It also caused a reversible reduction of coronary blood flow in guinea pigs [7–10]. While PCP induced a paradoxical increase in coronary blood flow in dogs, a simultaneous ECG recording showed that ischemic changes were present [10]. The effects of PCP on the coronaries were inhibited by detromethorphan, a direct

Table 1: Associated factors and precipitants of coronary vasospasm

Associated factors and precipitants of coronary vasospasm	
Pharmacologic agents	**Physiologic factors**
Catecholamines (epinephrine, dobutamine, etc.)	Genetic predisposition
Hypercholinergic crisis	Stress (physical, mental)
Histamine (Kounis syndrome)	Exposure to cold
Ergot derivatives (e.g., ergonovine)	Valsalva maneuver
5-Hydroxytryptamine antagonists (Ondansetron, Dolasetron)	Hyperventilation
Smoking	Coronary intervention procedures (balloon angioplasty)
Beta-adrenergic blocking agents	
Withdrawal from chronic nitroglycerin use	Disease States
Cocaine	Thyrotoxicosis
Amphetamines	Pheochromocytoma
Ecstasy	Neurofibromatosis
Heavy alcohol use	Magnesium deficiency (particularly in alcoholics)
Postpartum bromocriptine use	
5-Hydroxytryptamine agonists (Sumatriptan)	
Butane	

inhibitor of the PCP receptor [7]. This suggests that PCP may act directly on receptors in animal coronary arteries.

Recent data on PCP effects in humans is not extensive most likely because it is not a widely prevalent drug of abuse [1], and because its clinical use has been discontinued for many years. Additionally, many street drug abusers ingest multiple drugs, and thus there may be confounding effects of these drugs if their activity and pharmacokinetics are not well known. For example, up to 34% of male arrestees in major US cities tested positive for multiple drugs in 2012 [1].

The closest drug to PCP that is in clinical use is Ketamine (2-(2-chlorophenyl)-2-(methylamino) cyclohexan-1-one). Ketamine is a structural analogue of PCP with similar effects but less toxicity, and it is used as a dissociative sedative for brief procedures, particularly in children. The side effects of ketamine and PCP are similar at toxic doses, and in addition, the former has been associated with chest pain when used for analgesia or as a drug of abuse.

Our patient developed chest pain two hours after smoking PCP. We acknowledge that the temporal relationship does not prove PCP as the culprit. However, evidence from animal studies and reports on ketamine (a structural analogue of PCP) use in humans support the suggestion of PCP as the precipitant of coronary spasm in our patient.

CONCLUSION

Although there is paucity of data in humans, phencyclidine (PCP) has been shown to cause vasoconstriction and reduction in coronary blood flow in animal models. Additional studies are needed to confirm the effects of the drug on the human coronary artery. Our case illustrates the need for physicians to be aware of PCP ingestion as a possible cause of coronary artery spasm in young adult patients presenting with acute coronary syndrome.

****

Acknowledgements
We acknowledge the suggestions of R. F. Gillum, MD, MS and Adedayo Ogunleye, DO.

Author Contributions
Hakeem Ayinde – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Robert Solomon – Acquisition of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Maria-Elise Sanchez – Acquisition of data, Analysis and interpretation of data, Drafting the article, Final approval of the version to be published
James Diggs – Acquisition of data, Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published
Prafulla Mehrotra – Substantial contributions to conception and design, Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor
The corresponding author is the guarantor of submission.

Conflict of Interest
Authors declare no conflict of interest.

REFERENCES

1. ADAM II 2012 annual report. [Available at: http://www.whitehouse.gov/sites/default/files/ondcp/policy-and-research/adam_ii_2012_annual_rpt_web.pdf. Accessed February 1, 2015]
2. McCarron MM, Schulze BW, Thompson GA, Conder MC, Goetz WA. Acute phencyclidine intoxication: incidence of clinical findings in 1,000 cases. Ann Emerg Med 1981 May;10(5):237–42.
3. Aniline O, Pitts FN Jr. Phencyclidine (PCP): a review and perspectives. Crit Rev Toxicol 1982 Apr;10(2):145–77.
4. Ilett KF, Jarrott B, O'Donnell SR, Wanstall JC. Mechanism of cardiovascular actions of 1-(1-phenylcyclohexyl)piperidine hydrochloride (phencyclidine). Br J Pharmacol Chemother 1966 Oct;28(1):73–83.
5. Shi GG, Xu SF. Phencyclidine receptors in porcine coronary artery. [Article in Chinese]. Zhongguo Yao Li Xue Bao 1993 Jan;14(1):39–41.
6. Shi GG, Xu SF. Effect of phencyclidine on contraction of porcine coronary vessel strips. Zhongguo Yao Li Xue Bao 1993 May;14(3):211–3.
7. Shi GG, Xu SF. Effect of phencyclidine on coronary artery flow in the isolated heart of the guinea-pig. Acta Acad Med Shanghai 1994;21:93–6.
8. Shi GG, Wei DS, Liu BJ, Xu SF. Effect of phencyclidine on dog coronary artery. Zhongguo Yao Li Xue Bao 1996 May;17(3):224–6.
9. Weiner AL, Vieira L, McKay CA, Bayer MJ. Ketamine abusers presenting to the emergency department: a case series. J Emerg Med 2000 May;18(4):447–51.
10. Ward J, Standage C. Angina pain precipitated by a continuous subcutaneous infusion of ketamine. J Pain Symptom Manage 2003 Jan;25(1):6–7.

CASE REPORT PEER REVIEWED | OPEN ACCESS

The appendix and the inguinal canal: Amyand's hernia a case report

Shariful Islam, Devin Hosein, Vinoo Bheem, Vijay Naraynsingh

ABSTRACT

Introduction: Inguinal hernia repairs are one of the most common operations performed by the general surgeon. Occasionally, an emergency repair must be performed in the setting of painful incarceration, strangulation or obstruction. It is, therefore, essential that sound knowledge of the anatomy of the inguinal canal and variants of the inguinal hernia are required. One such variant is an Amyand's hernia, a rare inguinal hernia defined by the presence of the appendix in the inguinal canal. Case Report: We report a 52-year-old male who presented to our institution with a painful incarcerated right inguinal hernia who subsequently underwent emergency repair. A Type 1 Amyand's hernia was discovered on operation, an appendectomy and a Lichtenstein hernia repair were then performed. Due to the rarity of this hernia there are no standard guidelines for repair with respect to performing appendectomy or the use of mesh post appendectomy. Conclusion: The aim of this report is to assist the surgeon in making an informed decision on treatment of this rarity by review of current literature.

Keywords: Inguinal hernia, Amyand's hernia, mesh repair

How to cite this article

Islam S, Hosein D, Bheem V, Naraynsingh V. The appendix and the inguinal canal: Amyand's hernia a case report. Int J Case Rep Images 2015;6(10):636–639.

doi:10.5348/ijcri-2015104-CR-10565

Shariful Islam[1], Devin Hosein[1], Vinoo Bheem[2], Vijay Naraynsingh[3]

Affiliations: [1]Senior House Officer, Surgery, San Fernando Teaching Hospital, Trinidad & Tobago; [2]Head of the Department of Surgery (Ag), San Fernando Teaching Hospital, Trinidad & Tobago; [3]Department of Clinical Surgical Sciences, University of the West Indies, St Augustine, Trinidad and Tobago.

Corresponding Author: Dr. Shariful Islam, MBBS, DM (Surgery), Registrar in General Surgery, San Fernando Teaching Hospital, Trinidad & Tobago; E-mail: shar_islam7@hotmail.com

Received: 08 July 2015
Accepted: 26 August 2015
Published: 01 October 2015

INTRODUCTION

Hernia repairs are one of the most common operations done by the general surgeon with inguinal hernias being the most common type of abdominal wall hernias [1]. Inguinal hernias often present as acute emergencies and require emergent repair, therefore extensive knowledge of the anatomy and the variants of hernias is essential. An Amyand's hernia is one such variant of an inguinal hernia. This rare type of hernia is characterized by the presence of the appendix in the inguinal canal. Amyand's hernia can be further classified by the presence of co-existing pathology involving the appendix, that is, whether the appendix is inflamed or if there is unrelated pathology associated with the hernia [2].

The first case of an appendix within the inguinal sac was described by Claudius Amyand and published in the Philosophical Transactions of the Royal Society of London in 1976 [3]. He is also credited with performing the first successful appendectomy [3].

It is, therefore, important that awareness of this hernia and appropriate treatment of this rarity be kept in mind by every surgeon performing a hernia repair.

CASE REPORT

A 52-year-old male presented to the emergency department with sudden onset right sided groin pain and swelling. He is a construction worker by occupation and has been diagnosed with a right sided inguinal hernia one year prior. He has missed his date for surgery on several occasions. The pain was localized to the hernia which was irreducible. He had one episode of vomiting with no other symptoms of intestinal obstruction. He has a 30-pack year smoking history with no chronic cough or any urinary symptoms.

On examination his abdomen was soft, with tenderness localized to right inguinal region and no signs of intestinal obstruction or peritonism. The hernia was tender, irreducible and no cough impulse was elicited. External genitals and rectal examinations were unremarkable.

Hematological investigations, chest and abdominal X-rays were unremarkable. The patient was scheduled for emergency hernia repair.

Findings on operation included sliding hernia with non-inflamed appendix in the inguinal canal (Figures 1 and 2) consistent with a Type 1 Amyand's hernia [4]. Appendectomy was performed and a macro-porous polypropylene mesh was placed via Lichtenstein method. The patient did well and was discharged day 1 post operation with prophylactic antibiotics. Histology revealed the appendix was not inflamed confirming the patient had a Type 1 Amyand's hernia [4]. The patient was followed-up in the surgical outpatient clinic and at sixth month follow-up he recovered fully.

DISCUSSION

Amyand's hernia is defined as the presence of the appendix in the inguinal canal. It accounts for less than 1% of all inguinal hernias [5]. Appendicitis occurring within the inguinal canal is even a rarer clinical entity [6, 7]. It almost always occurs on the right, however, there are a few cases of it occurring on the left [8, 9].

Amyand's hernia can be classified into different subtypes. The classification was put forward by Losanoff and Basson and incorporates the presence of co-existing pathology with the hernia. According to the classification, there are four different types [4].

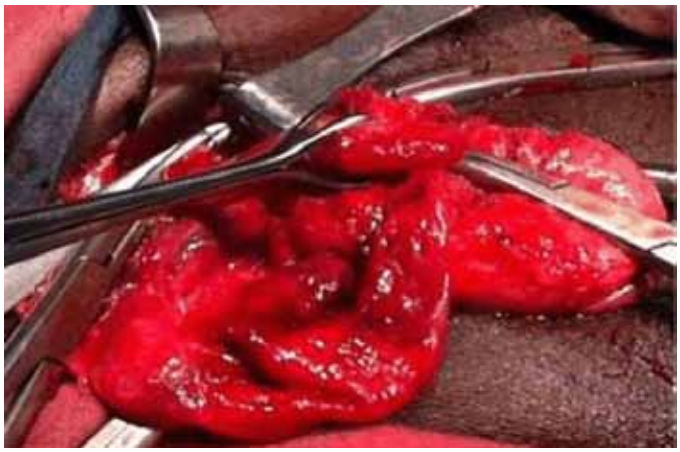


Figure 1: Appendix in the inguinal canal with Babcock placed on the appendix.

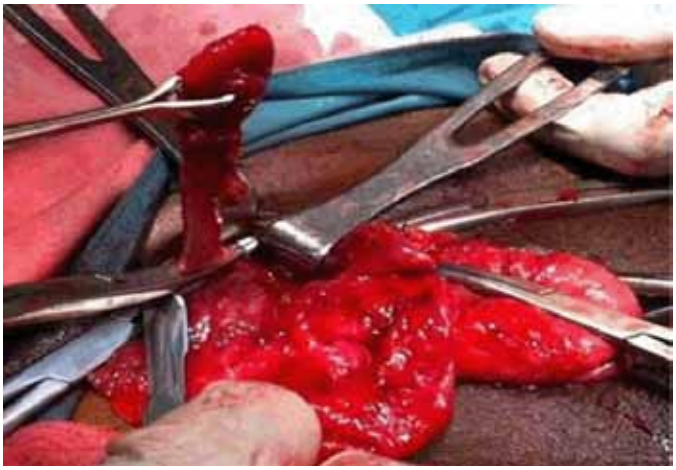


Figure 2: Appendectomy being performed through herniotomy.

Type 1 is the presence of a non-inflamed appendix in the inguinal canal.

Type 2 acute appendicitis in the inguinal canal without abdominal wall sepsis.

Type 3 acute appendicitis in the inguinal canal with abdominal wall sepsis.

Type 4 acute appendicitis in the inguinal canal and related or unrelated abdominal pathology.

Based on this classification, our patient had a Type 1 Amyand's hernia.

How can we diagnose the presence of an Amyand's hernia preoperatively? Amyand's hernia is diagnosed almost exclusively intraoperatively, however, there are a few cases where the diagnosis was made preoperatively. In these cases, this was done via the use of radiological investigations [10]. Having obtained the diagnosis of Amyand's hernia, how should one proceed? What is considered best management? Should the appendix be removed in a Type 1 hernia? Can we safely use mesh after performing an appendectomy? Amyand's hernia, being a rarity, presents a dilemma in terms of management as there is insufficient evidence to come to a definite conclusion.

In our case, the appendix was not inflamed and an appendectomy was performed. Some authors may choose not to remove the appendix [7, 11]. However, it is our belief that the appendix should be always removed in all Types of Amyand's hernia. If reduction of the appendix is performed, excessive manipulation may result in compromise of its blood supply therefore one may be reducing a compromised visceral structure. In addition, on reduction adhesions will develop between the appendix and the deep ring which will, in the event appendicitis develops, the presentation will be atypical. To further argue the point that appendectomy should be done in Type 1 Amyand's hernia, if one simply reduces the appendix and applies a mesh, the local inflammatory

response produced by the mesh can induce an appendicitis [12]. Conversely, an unrelated appendicitis can result in a severe mesh infection [13].

The issue of using a prosthetic mesh after appendectomy in an Amyand's hernia is a tenuous one. The benefits of performing a mesh repair of a hernia are well documented. Mesh repairs are associated with lower rates of recurrence compared to non-mesh repairs [14]. A review of 13 trials compared open hernia repairs with and without mesh found a significantly lower risk of recurrent hernia with mesh repairs [14]. In addition, the EU Hernia Trialists Collaboration reviewed 8221 patients and concluded a significantly higher recurrence rate for hernias repaired without mesh versus those performed with mesh using either open or laparoscopic techniques [15].

In our case, the patient is at increased risk for recurrence because of his occupation. Also, the appendix was noninflamed, therefore, mesh should be used. In type 2 and type 4 Amyand's hernias, appendectomy should be performed and mesh can be use depending on the amount of spillage at appendectomy and condition of surrounding tissues. In Type 3 Amyand's hernia, appendectomy should be performed however, mesh use, may not be indicated. There are cases where mesh has been used in infected tissue, however, we do not recommend this practice [16].

Finally, no discussion about Amyand's hernia is complete without brief mention of its femoral counterpart. A De Garengeot hernia is a rare subtype of femoral hernia in which the appendix lies within a femoral hernia [17]. Similarly to an Amyand's hernia, it presents its own diagnostic and therapeutic challenges that only a few surgeons had the honor of encountering.

CONCLUSION

Hernias repairs are routine operations done by the general surgeon, however, one must be aware of the rare variants. For the patient with the Amyand's hernia, the decision to perform an appendectomy and mesh repair should ultimately be individualized to each patient.

****

Acknowledgements
Informed consent was obtained from the patient to publish this case.

Author Contributions
Shariful Islam – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Devin Hosein – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published
Vinoo Bheem – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published
Vijay Naraynsingh – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor
The corresponding author is the guarantor of submission.

Conflict of Interest
Authors declare no conflict of interest.

REFERENCES

1. Dabbas N, Adams K, Pearson K, Royle G. Frequency of abdominal wall hernias: is classical teaching out of date? JRSM Short Rep 2011 Jan 19;2(1):5.
2. Holzheimer RG. Inguinal Hernia: classification, diagnosis and treatment--classic, traumatic and Sportsman's hernia. Eur J Med Res 2005 Mar 29;10(3):121–34.
3. Amyand C. Of an inguinal rupture, with a pin in the appendix caeci, incrusted with stone; and some observations on wounds in the guts. Philos Trans R Soc London 1736;39:329–36.
4. Losanoff JE, Basson MD. Amyand hernia: a classification to improve management. Hernia 2008 Jun;12(3):325–6.
5. Thomas WE, Vowles KD, Williamson RC. Appendicitis in external herniae. Ann R Coll Surg Engl 1982 Mar;64(2):121–2.
6. Salemis NS, Nisotakis K, Nazos K, Stavrinou P, Tsohataridis E. Perforated appendix and periappendicular abscess within an inguinal hernia. Hernia 2006 Dec;10(6):528–30.
7. D'Alia C, Lo Schiavo MG, Tonante A, et al. Amyand's hernia: case report and review of the literature. Hernia 2003 Jun;7(2):89–91.
8. Gupta S, Sharma R, Kaushik R. Left-sided Amyand's hernia. Singapore Med J 2005 Aug;46(8):424–5.
9. Weber RV, Hunt ZC, Kraal JG. Amyand's hernia: Etiologic and therapeutic implications of the two complications. Surg Rounds 1999;22:552–6.
10. Maizlin ZV, Mason AC, Brown C, Brown JA. CT findings of normal and inflamed appendix in groin hernia. Emerg Radiol 2007 Jun;14(2):97–100.
11. Sharma H, Gupta A, Shekhawat NS, Memon B, Memon MA. Amyand's hernia: a report of 18

consecutive patients over a 15-year period. Hernia 2007 Feb;11(1):31–5.
12. Jennings JM, Ng PCh. Laparoscopic appendicectomy for suspected mesh-induced appendicitis after laparoscopic transabdominal preperitoneal polypropylene mesh inguinal herniorraphy. J Minim Access Surg 2010 Jan;6(1):19–21.
13. Sebbag G, Ovnat A, Shaked G, Dukhno O, Levy I. Severe mesh graft infection consecutive to a perforated acute appendicitis 8 years after transabdominal laparoscopic right inguinal hernia repair: A review of the literature. J Pelvic Med Surg 2005;11:275–7.
14. Scott NW, McCormack K, Graham P, Go PM, Ross SJ, Grant AM. Open mesh versus non-mesh for repair of femoral and inguinal hernia. Cochrane Database Syst Rev 2002;(4):CD002197.
15. EU Hernia Trialists Collaboration. Mesh compared with non-mesh methods of open groin hernia repair: systematic review of randomized controlled trials. Br J Surg 2000 Jul;87(7):854–9.
16. Guzzo JL, Bochicchio GV, Henry S, Keller E, Scalea TM. Incarcerated inguinal hernia in the presence of Fournier's gangrene: a novel approach to a complex problem. Am Surg 2007 Jan;73(1):93–5.
17. De Garengeot RJC. Traite des operations de chirurgie. 2nd ed. Paris: Huart; 1731. p. 369–71.

CASE REPORT PEER REVIEWED | OPEN ACCESS

Neglected case of hydrocephalus in a five-year-old child

Moataz Hesham Abdelreheem, Marwa Mohammed Basyouni

ABSTRACT

Introduction: Hydrocephalus is abnormal accumulation of cerebrospinal fluid (CSF) in the ventricles of the brain. It is of two types: Congenital and acquired. Case Report: A child, five-year-old, presented to the emergency department with a severe neglected hydrocephalus which was not the complaint of the parents. Conclusion: Neglected hydrocephalus will leave an irreversible brain damage to the child. The best way to avoid that negligence is to spread health education in all areas with special focus in rural regions.

Keywords: Health education, Huge ventriculo-megaly, Hydrocephalus

How to cite this article

Abdelreheem MH, Basyouni MM. Neglected case of hydrocephalus in a five-year-old child. Int J Case Rep Images 2015;6(10):640–643.

doi:10.5348/ijcri-2015105-CR-10566

Moataz Hesham Abdelreheem[1], Marwa Mohammed Basyouni[1]

Affiliations: [1]House Officer, Faculty of Medicine, Alexandria University, Alexandria, Egypt.

Corresponding Author: Moataz Hesham Abdelreheem 34 El Sheikh Mohamed Abdo street, El Gomrok, Alexandria, Egypt, 21556; Ph: +2 01147431108; Email: moataz.abdelreheem@outlook.com

Received: 30 May 2015
Accepted: 22 June 2015
Published: 01 October 2015

INTRODUCTION

The term hydrocephalus refers to a condition in which the primary characteristic is excessive accumulation of cerebrospinal fluid (CSF) in the brain due to medical conditions that block its normal flow or absorption, resulting in an abnormal widening of ventricles, which in turn creates potentially harmful pressure on the tissues of the brain.

Hydrocephalus may be congenital (when a baby is born with the condition) or acquired (develops at the time of birth or at some point afterward). Congenital hydrocephalus may be of two types: communicating where the flow of CSF is blocked after it exits the ventricles. This form is called communicating because the CSF can still flow between the ventricles, which remain open. Or non-communicating (also called "obstructive" hydrocephalus) occurs when the flow of CSF is blocked along one or more of the narrow passages connecting the ventricles [1].

Anomalies of ventricular flow system would produce hydrocephalus directly. The most common abnormality of ventricular flow is atresia of the aqueduct of Sylvius [2]. Being the narrowest part of the cerebrospinal fluid (CSF) pathway (mean cross-sectional area is 0.5 mm^2 in children and 0.8 mm^2 (range 0.2–1.8 mm^2) in adults), the sylvian aqueduct is the most common site of intraventricular blockage of the CSF. This blocks the fluid flow system between the third and fourth ventricles causing dilatation of the lateral ventricles and the third ventricle. The infant may be born with complete atresia of the aqueduct of Sylvius or may develop full occlusion of the aqueduct over the first few days or weeks of life. There is also a genetic sex-linked recessive form of hydrocephalus secondary to atresia of the aqueduct of Sylvius.

Congenital hydrocephalus may be also caused by a congenital anomaly of the brain and its bony coverage, the most common cause of which is Arnold-Chiari malformation, a structural defect in the cerebellum [3]. When the indented bony space at the lower rear of the skull is smaller than normal, the cerebellum and brain stem can be pushed downward. The resulting pressure on

the cerebellum can block the flow of cerebrospinal fluid leading to hydrocephalus.

Congenital hydrocephalus can also occur in babies born prematurely (before 37 weeks of the pregnancy). Some premature babies have bleeding in the brain, which can block the flow of CSF and cause hydrocephalus. Other possible causes of congenital hydrocephalus include certain health conditions such as spina bifida and arachnoid cysts (fluid filled sacs located between the brain or spinal cord and the arachnoid membrane -one of the three membranes surrounding the brain and spinal cord).

Another form of hydrocephalus is identified, which does not fit exactly into the categories mentioned above and primarily affects adults: normal pressure hydrocephalus (NPH). The NPH is an abnormal increase of cerebrospinal fluid in the brain's ventricles that may result from a subarachnoid hemorrhage, head trauma, infection, tumor, or complications of surgery. However, many people develop NPH when none of these factors are present.

CASE REPORT

A five-year-old boy was presented to the emergency department with his parents with a history of falling from height. They lived in a rural area near Alexandria city. The father works as a farmer and the mother is a housewife, both of them were illiterate. The child was born term by normal labor. There was a history of an increased head size at birth, but the parents neglected this observation and they did not seek any medical advice although the head increased in size gradually over the time.

Once the child was examined, a significant large head size of the child was noticed, which was not the complaint of the parents. The child was underweight (11 kg) and his height was less normal in his age group (80 cm). At the beginning the child was not alert but after a short while he gained his consciousness. There were signs of mental retardation, drowsiness, impaired speech, short attention span and impairment in physical coordination. The parents reported problems in his learning abilities.

Upon examination of the head, its circumference was 66 cm. The scalp skin was shiny with apparent veins. Eye examination showed (setting sun sign).

Computed tomography (CT) scan of brain revealed enlarged frontal and posterior horns of the lateral ventricles and enlarged third and fourth ventricles. Such bilateral huge ventriculomegaly is accompanied by severe brain parenchymal loss as illustrated (Figures 1 and 2). It was noticed also that the enlargement of the ventricles is out of proportion with sulcal atrophy, i.e., relative normal sulcal size, which is an indication of normal intracranial pressure.

The images of the patient's brain were examined concerning the trauma that the child had after falling from height. The CT scan did not show any problem that

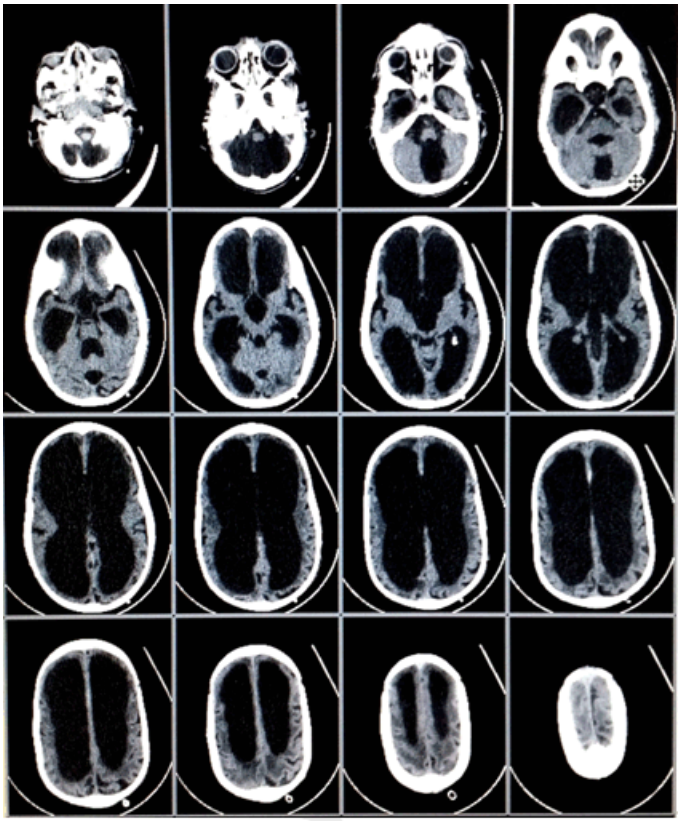


Figure 1: Axial non-enhanced computed tomography scan of the head of the patient demonstrating 16 sections of the brain at various levels. Note the extent of the dilatations of the ventricles with severe atrophy of the brain. Sulci look normal which give an idea about the intracranial pressure status.

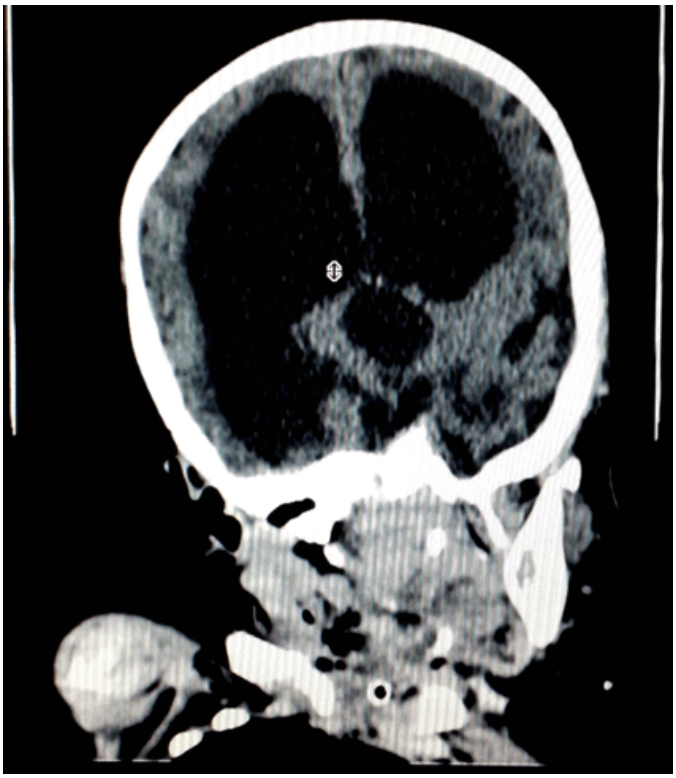


Figure 2: A coronal section of the computed tomography scan of the head of the patient showing another view of the hydrocephalic brain.

happened due to the fall or any kind of hemorrhage. The parents were assured that the trauma did not cause any problem and were informed about such neglected case of hydrocephalus and that the child will most probably have a normal life span. They were told, however, that there is no cure for such condition and the patient was booked for an appointment at the rehabilitation center which will help the child to cope with the activities of the daily living and improve his cognitive function. The child was discharged without any surgical intervention after a full system examination which cleared the child. Moreover, we told the parents that they must attend a follow-up visit every six months with the neurology department in the hospital so that the child's symptoms are monitored.

DISCUSSION

Hydrocephalus is a common pediatric neurosurgical condition. The prevalence of congenital and infantile hydrocephalus in the United States and Europe has been predicted to be 0.5 to 0.8 per 1000 live and still births [4–6]. If neglected, hydrocephalus is able to produce a devastating complications on the child's brain, for instance as a result of compression, and this will cause a severe form of mental retardation as observed in our case. In this case, the brain tissue is severely damaged that the tissue is minimal. It is very rare that a child can reach such stage of hydrocephalus. For such reason, hydrocephalus should be screened for and diagnosed as early as possible. It can be diagnosed among inside the uterus of the mother by CT scans and ultrasonography. Later in life, hydrocephalus is diagnosed through clinical neurological evaluation and by using cranial imaging techniques such as ultrasonography, CT scan, MRI scan, or pressure-monitoring techniques.

This case report raises two important questions. Is the health education in the rural areas sufficient enough? Such case of neglected hydrocephalus reflects poor health education among people living in the countryside and their unawareness of the importance of the follow-up and seeking medical advice as early as possible.

Secondly, will the traditional surgical shunt treatment of hydrocephalus be beneficial here? Those who survive without treatment have different amounts of irreversible intellectual, physical, and neurological disabilities.

CONCLUSION

To sum up, hydrocephalus can leave a child with a permanent irreversible damage. In order to prevent that from happening, the head of an infant or child should be protected from injury, Prompt treatment of infections and other disorders associated with hydrocephalus, and most important, improve the level of health education especially in rural areas.

****

Author Contributions

Moataz Hesham Abdelreheem – Substantial contributions to conception and design, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Marwa Mohammed Basyouni – Substantial contributions to conception and design, Acquisition of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

Authors declare no conflict of interest.

REFERENCES

1. Davson H. Dynamic aspects of cerebrospinal fluid. Dev Med Child Neurol Suppl 1972;27:1–16.
2. Dandy W. Diagnosis and treatment of strictures of the aqueduct of sylvius (causing hydrocephalus). Arch surg (Chicago) 1945;51(1):1–4.
3. Peach B. Arnold-Chiari Malformation: Morphogenesis. Arch Neurol 1965 May;12(5):527–35.
4. Fernell E, Hagberg G, Hagberg B. Infantile hydrocephalus epidemiology: an indicator of enhanced survival. Arch Dis Child Fetal Neonatal Ed 1994 Mar;70(2):F123–8.
5. Jeng S, Gupta N, Wrensch M, Zhao S, Wu YW. Prevalence of congenital hydrocephalus in California, 1991-2000. Pediatr Neurol 2011 Aug;45(2):67–71.
6. Garne E, Loane M, Addor MC, Boyd PA, Barisic I, Dolk H. Congenital hydrocephalus--prevalence, prenatal diagnosis and outcome of pregnancy in four European regions. Eur J Paediatr Neurol 2010 Mar;14(2):150–5.

CLINICAL IMAGES PEER REVIEWED | OPEN ACCESS

Bilateral megaureters secondary to neuropathic bladder in an adult

Tomoya Nishino, Yasue Watanabe, Keiji Sakurai, Seiji Morita

CASE REPORT

A 34-year-old female came to our hospital with complaints of anorexia and malaise that had persisted for approximately 10 days. She did not have abdominal or back tenderness, and her skin was dry. Her systolic blood pressure was 76 mmHg, and laboratory tests revealed an elevated creatinine level of 6.41 mg/dl, potassium level of 8.4 mEq/l, white blood cell count of 28,000/μl and C-reactive protein level of 17.98 mg/dl. She was admitted with diagnoses of urinary tract infection, acute kidney injury and hyperkalemia. At birth, she was diagnosed of schistorrhachis (Figure 1), and therefore, self-catheterization was performed. Abdominal computed tomography scan showed bilateral megaureters and hydronephrosis (Figure 2). Cystoscopy showed ends of both the ureters opening into the bladder. As her blood tests were abnormal (pH 7.360, serum HCO3 11.4 mmol/l), we performed hemodialysis once. Moreover, double J stents were placed in the ureters and antibiotic prophylaxis was administered. After treatment, her symptoms improved and blood tests became normal, and she was discharged. Based on this history, we diagnosed her as a case of neuropathic bladder secondary to schistorrhachis.

Tomoya Nishino[1], Yasue Watanabe[2], Keiji Sakurai[1], Seiji Morita[2]

Affiliations: [1]Department of Emergency and Critical Care Medicine, Tokai University School of Medicine; [2]Odawara Municipal Hospital, Department of Emergency and Critical Care Medicine

Corresponding Author: Tomoya Nishino, Department of Emergency and Critical Care Medicine, Tokai University School of Medicine, 259-1193 Simokasuya143, Isehara City, Kanagawa; Tel: 0463-93-1121, Fax: 0463-93-3701; Email: i10ve69@yahoo.co.jp

Received: 18 April 2015
Accepted: 23 May 2015
Published: 01 October 2015

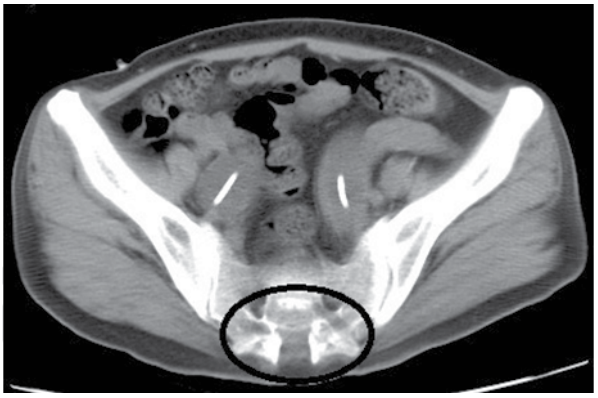


Figure 1: Abdominal computed tomography showing schistorrhachis.

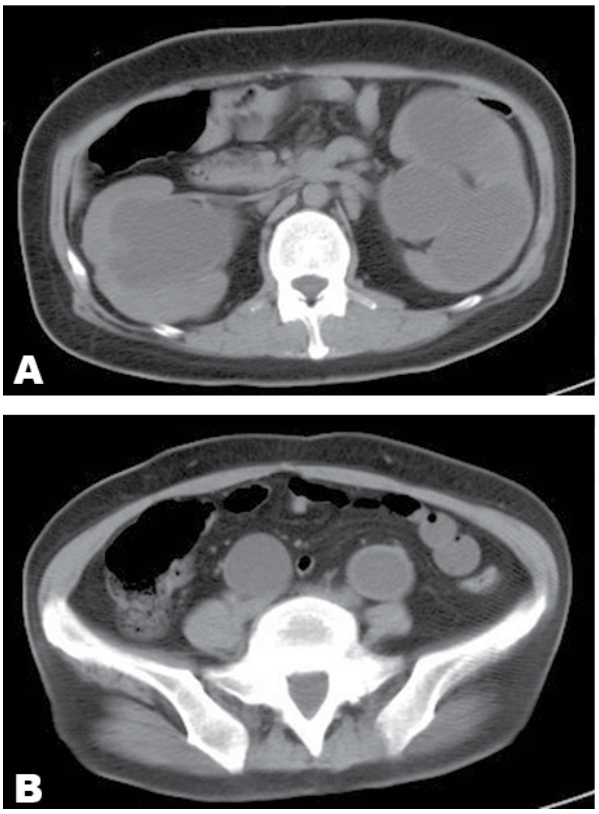


Figure 2: Abdominal computed tomography showing bilateral megaureters and hydronephrosis.

DISCUSSION

A dilated ureter is considered as a megaureter, and any diameter of the ureter >8 mm is considered abnormal. Although the occurrence of megaureter is common in children, it is rare in adults.

Megaureter can be classified as reflux, obstructed, and nonreflux-nonobstructed [1]. The causes of megaureter development are ureteropelvic junction obstruction, ureterovesical junction obstruction, neuropathic bladder, vesicoureterical reflux, high fetal urine output, and increased compliance of the fetal ureter. The presence of a megaureter could be considered when the patient has a urinary tract infection or hematuria. Moreover, ultrasonography scan, computed tomography, and magnetic resonance imaging scan can be used for its diagnosis. This was a case of reflux megaureter secondary to neuropathic bladder. Bilateral megaureters are rare in an adult.

Medical treatment may include antibiotic prophylaxis for infection, anticholinergic medication, and clean intermittent catheterization for elevated detrusor leak point pressure.

Surgical treatment may include ureteral plication or Infolding for moderately dilated ureters, and excisional tapering for massively dilated or thickened ureters.

Treatment of secondary megaureter is conservative. However, treatment of primary megaureter may involve surgery or may be conservative [2].

CONCLUSION

This case illustrates a neuropathic bladder secondary to schistorrhachis. Bilateral secondary megaureter in an adult is rare, and treatment of secondary megaureter is conservative.

Keywords: Adult, Bilateral megaureters, Medical treatment, Neuropathic bladder

****

How to cite this article

Nishino T, Watanabe Y, Sakurai K, Moritam S. Bilateral megaureters secondary to neuropathic bladder in an adult. Int J Case Rep Images 2015;6(10):644–646.

doi:10.5348/ijcri-201528-CL-10083

****

Author Contributions

Tomoya Nishino – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Yasue Watanabe – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

Keiji Sakurai – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

Seiji Morita – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

Authors declare no conflict of interest.

Copyright

REFERENCES

1. King LR. Megaloureter: definition, diagnosis and management. J Urol 1980 Feb;123(2):222–3.
2. Abraham GP, Das K, Ramaswami K, et al. Laparoscopic reconstruction for obstructive megaureter: single institution experience with short- and intermediate-term outcomes. J Endourol 2012 Sep;26(9):1187–91.

CLINICAL IMAGES PEER REVIEWED | OPEN ACCESS

Thyroid cancer diagnosed by a routine scan

Maria João Bugalho, Rafael Cabrera

CASE REPORT

A 64-year-old male underwent a Tc-99m thyroid scintigraphy, requested by his general practitioner, as part of the evaluation of an enlarged thyroid associated with subclinical hyperthyroidism (suppressed TSH and high normal values of thyroid hormones). Although rare [1, 2], the observation of abnormal bilateral uptake, outside of the thyroid bed, in a patient with thyroid (Figure 1; Panel A), was consistent with uptake by metastatic lymph nodes from a thyroid cancer. An ultrasound guided cytology was positive for papillary thyroid cancer (PTC).

Total thyroidectomy plus bilateral neck dissection were performed. The macroscopic description of the pathological report referred several poorly defined, whitish, nodules (< 1 cm) and the microscopic pathological report was of diffuse follicular variant of papillary thyroid carcinoma (DFVPTC) with extrathyroidal extension, vascular invasion and nodal metastases. This rare histological subtype is more common in young females and has been associated with an aggressive behavior [3].

Following surgery, the patient received two treatments with I-131 (total activity 304 mCi). Post first ^{131}I treatment (November/2010), whole body scan (WBS) showed uptake in the thyroid bed and in the lungs (TSH 100 µIU/ml, Thyroglobulin 42.8 ng/ml, Thyroglobulin antibodies negative) (Figure 1; Panel B). Thoracic computed tomography scan was negative. Post second treatment (April/2011), there was no abnormal uptake (TSH 87.5 µIU/ml, thyroglobulin 1.8 ng/ml, thyroglobulin antibodies were negative).

At last observation (April/2015), under levothyroxine suppression, the patient had no evidence of disease and both serum thyroglobulin and thyroglobulin antibodies were undetectable.

DISCUSSION

Lymph node metastases from a primary thyroid cancer usually do not take up either iodine or technetium. Therefore, radio isotopic detection of lymph node metastases secondary to thyroid cancer, in the presence of thyroid, is rare [1, 2]

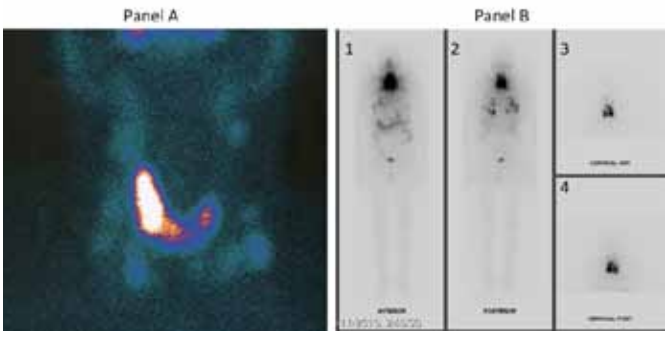


Figure 1: Panel A: Tc-99m thyroid scintigraphy showing diffuse and intense uptake in the thyroid gland and several foci of abnormal extrathyroidal accumulation of radiotracer; Panel B: Whole-body scan (1 anterior, 2 posterior) obtained on the third day after the first I-131 treatment showing intense radiotracer uptake in the thyroid bed and bilateral lungs. Neck spot views (3 anterior, 4 posterior) of the thyroid bed uptake. Corresponding biochemical evaluation (TSH 100 µIU/ml, thyroglobulin 42.8 ng/ml, thyroglobulin antibodies were negative).

Maria João Bugalho[1,2], Rafael Cabrera[3]

Affiliations: [1]Serviço de Endocrinologia, Instituto Português de Oncologia de Lisboa Francisco Gentil E.P.E., Lisboa, Portugal; [2]NOVA Medical School / Faculdade de Ciências Médicas, Universidade Nova de Lisboa, Portugal; [3]Serviço de Anatomia Patológica, Instituto Português de Oncologia de Lisboa Francisco Gentil E.P.E., Lisboa, Portugal.

Corresponding Author: Maria João Bugalho, Mailing address: Serviço de Endocrinologia, Instituto Português de Oncologia de Lisboa Francisco Gentil E.P.E., Rua Professor Lima Basto, 1099-023 Lisboa, Portugal; Ph: (+351) 21 7229818; E-mail: mjbugalho@ipolisboa.min-saude.pt; Fax: (+351) 21 7229844

Received: 05 June 2015
Accepted: 04 August 2015
Published: 01 October 2015

Among the different subtypes of follicular variant of PTC the DFVPTC is also rare, is considered to be more common in young females and has been associated with an aggressive behavior [3]. Contrasting with previous reports, the case presented herein had a favorable outcome. We hypothesized that, despite an aggressive histological pattern, the tumor might have high levels of sodium-iodide symporter (NIS) expression likely to explain the TC-99m uptake by lymph nodes, regardless the presence of thyroid, as well as the response to radioiodine treatment.

CONCLUSION

Normal thyroid tissue is more efficient at trapping radiotracer, either iodine or technetium, than tumor tissue. Thus, thyroid scan is generally useless to detect lymph node metastases in patients with the thyroid gland in place. However, extrathyroidal areas of uptake on a routine thyroid scan most probably correspond to lymph node metastases from a differentiated thyroid carcinoma.

Keywords: Cancer, Lymph node metastases, Tc-99m thyroid scintigraphy, Tissue, Thyroid

****

How to cite this article

Bugalho MJ, Cabrera R. Thyroid cancer diagnosed by a routine scan. Int J Case Rep Images 2015;6(10):647–648.

doi:10.5348/ijcri-201529-CL-10084

****

Author Contributions

Maria João Bugalho – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Rafael Cabrera – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

The authors declare that they have no conflict of interest. Ethical approval: This article does not contain any studies with human participants performed by any of the authors.

Copyright

REFERENCES

1. Kumaresan K, Sastry RA. Localization of Tc-99m pertechnetate in lymph node metastasis from occult thyroid carcinoma. Clin Nucl Med 1994 Dec;19(12):1112.
2. Boughattas S, Hassine H, Arifa N, Preoperative scintigraphic visualization of lymph nodes and pulmonary metastases from papillary thyroid carcinoma. Ann Nucl Med 2004 Feb;18(1):59–62.
3. Ivanova R, Soares P, Castro P, Sobrinho-Simões M. Diffuse (or multinodular) follicular variant of papillary thyroid carcinoma: a clinicopathologic and immunohistochemical analysis of ten cases of an aggressive form of differentiated thyroid carcinoma. Virchows Arch 2002 Apr;440(4):418–24.

CLINICAL IMAGES PEER REVIEWED | OPEN ACCESS

A rare cause of abdominal pain and gastrointestinal bleeding: Colonic lipoma causing intussusception

Daniela Ferreira, Marta Salgado, Isabel Pedroto

CASE REPORT

A 70-year-old male went to the emergency room with a 24-hour history of hematochezia preceded by pain in the right lower quadrant. The patient denied fever, weight loss, anorexia or vomiting. He was passing flatus and stools. His past medical history included ischemic heart disease, left renal neoplasia with nephrectomy two years previously and without evidence of recurrence, appendectomy in the childhood, right inguinal hernia repair, hyperuricemia, dyslipidemia, hypothyroidism treated with levothyroxine and colonic diverticula. On physical examination the patient was hemodynamically stable, afebrile and with pain on deep palpation of the right lower quadrant. Bright red blood was observed in the rectum. No other abnormal findings were noted. Blood tests demonstrated mild anemia (hemoglobin 11.2 g/dL) without leukocytosis or neutrophilia and elevation of C-reactive protein (88 mg/L). Renal and liver functions were normal. A colonoscopy was performed. Endoscopy revealed a violaceous mass with ulcerated/necrotic surface obstructing the lumen in the descending colon (Figure 1). After the colonoscopy the patient had an abdominal computed tomography scan that revealed a lipomatous ovoid tumor with 65 mm of major axis with origin in the ascending colon but with top ographic change, because of a complication: colic-colic intussusception with the typical *donut sign* (Figure 2). The patient underwent surgery that confirmed a lesion with 65 mm and origin in the colonic margin of the ileocecal valve that caused the intussusception. A right hemicolectomy was performed. The histopathology confirmed the diagnosis of giant lipoma with origin in the submucosa.

DISCUSSION

Colonic lipomas are common findings during routine colonoscopy. They are benign lesions usually located in the right colon with decreasing incidence from the cecum to the sigmoid colon. They usually arise from the submucosal layer, but can extend into the muscularis propria, while up to 10% are subserosal. Classically, endoscopic findings described for lipoma are three:

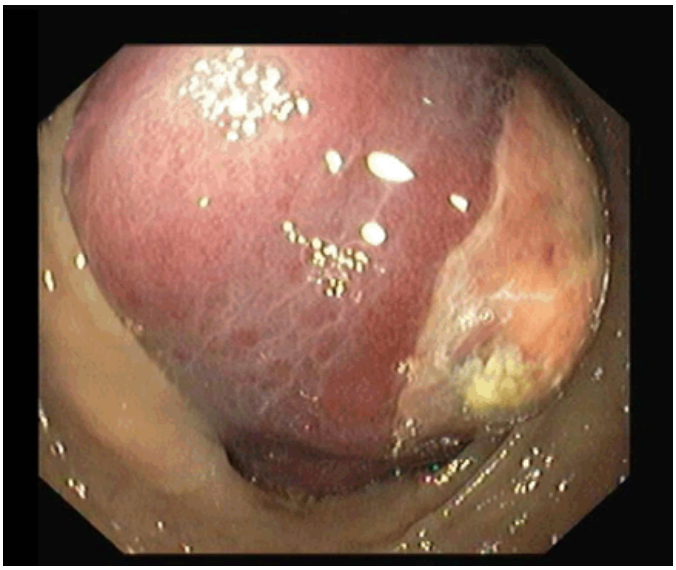


Figure 1: A violaceous mass with ulcerated/necrotic surface obstructing the lumen in the descending colon (Endoscopic image).

Daniela Ferreira[1], Marta Salgado[2], Isabel Pedroto[3]

Affiliations: [1]MD, Gastroenterology Resident, Gastroenterology Department, Hospital Santo António. Largo Professor Abel SalazarPorto. Portugal; [2]MD, Specialist in Gastroenterology, Gastroenterology Department, Hospital Santo António. Largo Professor Abel Salazar Porto. Portugal; [3]PhD, Specialist in Gastroenteroloy and Department Director, Hospital Santo António Largo Professor Abel Salazar Porto. Portugal.

Corresponding Author: Daniela Gonçalves Ferreira, Gastroenterology Department. Hospital Santo António. Largo Professor Abel Salazar 4099-001 Porto. Portugal; Tel: +351 222 077 500; Fax: +351 222 053 218; Email: danielagonferreira@gmail.com

Received: 18 December 2014
Accepted: 21 April 2015
Published: 01 October 2015

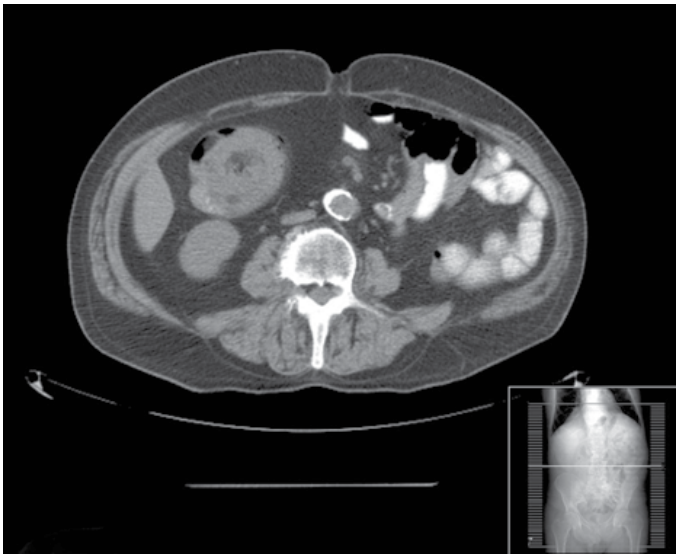


Figure 2: A lipomatous ovoid tumor complicated with a colic-colic intussusception with the typical donut sign.

the mucosa being elevated over the lipoma with the biopsy forceps (tent sign), indentation of the lipoma with the biopsy forceps (cushion sign) or the "naked fat sign" where the fat can be extruded after biopsy. When typical endoscopic findings are present and they are asymptomatic lipomas do not need additional study or follow up. However, when the typical endoscopic findings are absent additional study with endoscopic ultrasound and/or surgical resection should be considered. Surgical resection should also be considered when they become symptomatic. Lipomas larger than 4 cm in size (giant lipomas) can become symptomatic in 75% of patients. They can manifest as abdominal pain, alteration in bowel habits, gastrointestinal bleeding, perforation, obstruction or intussusceptions.

Intussusceptions are an uncommon complication of colonic lipomas and usually limited to one segment of the colon but can extend to more than one segment in some cases [1, 2]. Patients with this complication can present with abdominal pain and gastrointestinal bleeding. Abdominal computed tomography scan is the preferred diagnostic method, as the imaging characteristics of the tumors are typical for adipose tissue with the typical donut sign. Colonoscopy can show a mass with necrotic areas and ulcerated mucosa. Due to the nature of this lesion (subepithelial) biopsies are often inconclusive. The distinction between a colonic lipoma complicated with intussusception from other lesions (malignant epithelial or subepithelial) may be impossible based only on endoscopic aspects. Surgical resection is the treatment of choice when giant lipomas are complicated by intussusceptions.

CONCLUSION

The key learning element in this case is recognition of a rare and serious complication (intussusception) of a common endoscopic finding (colonic lipoma). In this case, the typical clinical presentation, endoscopic and radiologic appearance to this complication is shown and the approach discussed.

Keywords: Abdominal pain, Colonic lipoma, Gastrointestinal bleeding, Intussusceptions

****

How to cite this article

Ferreira D, Salgado M, Pedroto I. A rare cause of abdominal pain and gastrointestinal bleeding: Colonic lipoma causing intussusception. Int J Case Rep Images 2015;6(10):649–651.

doi:10.5348/ijcri-201530-CL-10085

****

Author Contributions

Daniela Ferreira – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Marta Salgado – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published
Isabel Pedroto – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

Authors declare no conflict of interest.

Copyright

REFERENCES

1. Zubaidi A, Al-Saif F, Silverman R. Adult intussusception: A retrospective review. Dis Colon Rectum 2006 Oct;49(10):1546–51.

2. Mouaqit O, Hasnai H, Chbani L, et al. Pedunculatedlipoma causing colo-colonic intussusception: A rare case report. BMC Surg 2013 Oct 30;13:51.

CLINICAL IMAGES PEER REVIEWED | OPEN ACCESS

Prominent tibial tubercles

Michael Flannery, Damien Knudson, Yumeng Zhang

CASE REPORT

Bilateral enlargement or hypertrophy of the tibial tuberosities (tubercles) is seen in our patient who previously had been diagnosed with sternoclavicular hyperostosis [1]. Our patient presented with these symptoms of approximately six months duration with no further increase in size. The tubercles (Figure 1) were moderately painful to touch with no warmth or erythema. The patient is a 49-year-old female with a history of hypertension and hypertriglyceridemia and osteoarthritic joint pain. Her medications included: tramadol 50 mg every six hours as needed, lisinopril 20 mg daily, hydrochlorothiazide 12.5 mg daily and fenofibrate 145 mg daily. Examination showed no fever or lymphadenopathy at any site. Both tubercles were enlarged right greater than left with generalized tenderness at the site of the tubercle but no pain at any other site around the knee. The patient had no history of Osgood-Schlatter disease as an adolescent. There were no recent infections and once enlarged there was no further growth of the lesions. Prior radiographic imaging of the sternoclavicular joint demonstrated no evidence of tumor or infection. Her laboratory studies were unremarkable [1]. The patient was treated with heat alternating with cold packs for 15 minutes four times daily along with meloxicam 15 mg daily. The patient could not afford a patellar band. The patient has ongoing diffuse idiopathic skeletal hyperostosis (DISH).

Michael Flannery[1], Damien Knudson[1], Yumeng Zhang[1]
Affiliations: [1]MD, FACP, University of South Florida, Morsani College of Medicine, 12901 Bruce B Downs Blvd. MDC Box 19 Room L1041, Tampa, Florida.
Corresponding Author: Michael T. Flannery, University of South Florida Morsani College of Medicine, 12901 Bruce B Downs Blvd. MDC Box 19 Room L1041, Tampa, Florida 33612; Ph: 813-974-6443; Fax: 813-905-9794; Email: mflanner@health.usf.edu

Received: 15 April 2015
Accepted: 30 June 2015
Published: 01 October 2015

DISCUSSION

Enlargement of the tibial tubercles in an adolescent would be classic for Osgood-Schlatter disease; however, our patient has no history of the disease, nor did she has evidence of the disease six months previously. Over use of the quadriceps muscle could result in an inflammatory process of the tibial tubercle. Our patient gave no specific history of a change in her activities that would suggest such inflammation. Of course the differential includes infection and tumor. However, she had no fever or leukocytosis. In addition, her involvement was bilateral and stable with no further growth making such diagnosis less likely. Complications include fracture of the ossicle and potential treatments may include

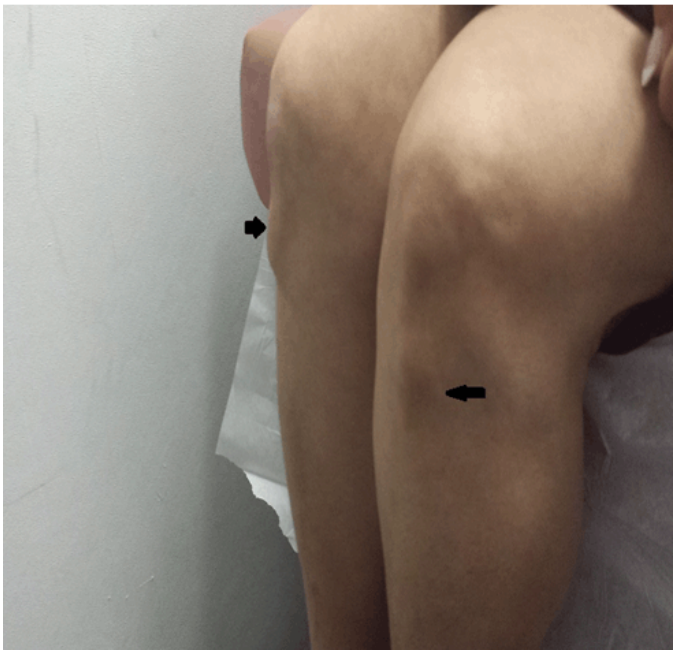


Figure 1: Enlarged tibial tubercles.

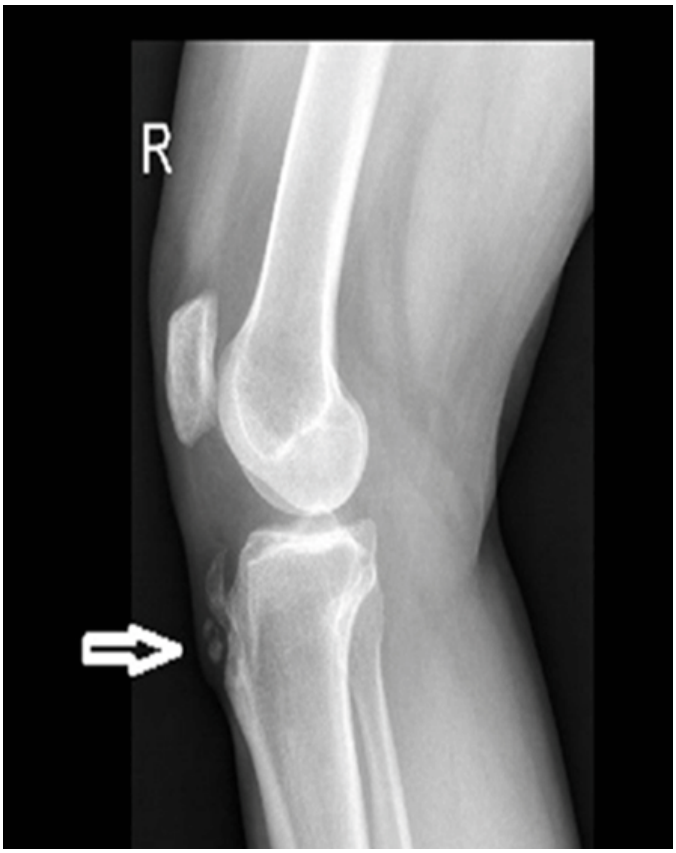


Figure 2: Extensive ossification of the tibial tuberosity of the right knee.

laparoscopic excision of an enlarged tubercle. Another possibility, given her prior diagnosis of sternoclavicular hyperostosis, would be an extraspinal manifestation of diffuse idiopathic skeletal hyperostosis (DISH) [2]. The hallmark of the disease is the ossification and calcification of the anterolateral aspect of the thoracic spine requiring involvement of at least four contiguous thoracic segments with preserved disc spaces and lack of apophyseal joint degeneration. Involvement may be asymptomatic in many individuals, while others suffer pain and decreased mobility. Peripheral involvement is usually distinguished by involvement of joints not usually involved by primary osteoarthritis, increased hypertrophic changes and calcification and ossification of enthesis (inflammation of connective tissue between tendon or ligament and bone) in sites other than joints. Some examples include hyperostosis of the ribs, sternoclavicular joint, olecranon and tibial tubercles. Interesting DISH is associated with a metabolic profile of hypertension, dyslipidemia and diabetes mellitus [2].

In our patient, non-steroidal anti-inflammatory drugs have offered relief along with tramadol. Her metabolic conditions have been treated appropriately and she needs to be monitored for diabetes. Radiography of her tibial tubercle on the right can be seen in Figure 2.

CONCLUSION

Our patient has two area of hyperostosis sternoclavicular and tibial tuberosity generating a theory that she may have extra-spinal feature of diffuse idiopathic skeletal hyperostosis (DISH).

Keywords: Enlarged tibial tubercles, Diffuse idiopathic skeletal hyperostosis, Osteoarthritic joint pain, Osgood-Schlatter disease, Osteoarthritis

****

How to cite this article

Flannery M, Knudson D, Zhang Y. Prominent tibial tubercles. Int J Case Rep Images 2015;6(10):652–654.

doi:10.5348/ijcri-201531-CL-10086

****

Author Contributions

Michael Flannery – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Damien Knudson – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published
Yumeng Zhang – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

Authors declare no conflict of interest.

Copyright

REFERENCES

1. Flannery MT, Kara F Villarreal AA. Sternoclavicular hyperostosis. Int J Case Rep Images 2014;5 (6):459–61.
2. Mader R, Sarzi-Puttini P, Atzeni F, Extraspinal manifestations of diffuse idiopathic skeletal hyperostosis. Rheumatology (Oxford) 2009 Dec;48(12):1478–81.

CLINICAL IMAGES PEER REVIEWED | OPEN ACCESS

Biphasic T-wave in patient with chest pain (Wellens' syndrome)

Ahmed Zedan, Osama Mukarram, Umer Malik, Raja Naidu

CASE REPORT

A 65-year-old Hispanic male with no prior medical history came to the emergency room for a new onset intermittent chest pain of 1 day in duration while working in his garage. The pain was described as chest tightness, mainly retrosternal, radiating to the left shoulder, associated with nausea and shortness of breath with no vomiting or diaphoresis.

The patient denied any prior history of chest pain, orthopnea or paroxysmal nocturnal dyspnea. His last episode of chest was 1 hour prior to his arrival to the emergency room. The patient used to smoke about 1 pack a day for more than 15 years.

Physical examination showed average built male in no distress, blood pressure of 135/80, pulse of 80 beat per minute, respiratory rate of 16, temperature of 98.8°C and sating 97% on room air. Cardiovascular examination was normal, as well as the remainder of the physical examination. On chest X-ray there was no cardiomegaly, infiltrates, or other abnormality. Initial EKG done in the emergency room while being pain free showed normal sinus rhythm, normal axis, narrow QRS, no Q-waves and isoelectric ST-segment. Initial laboratory work showed troponin of 0.09 (laboratory cut off 0.08) other laboratory examinations work were within normal including the lipid profile.

Ahmed Zedan[1], Osama Mukarram[1], Umer Malik[1], Raja Naidu[2]
Affiliations: [1]MD, Resident, Department ofInternal Medicine, Texas Tech University Health Since Center, Odessa, Texas, USA; [2]MD, Attending, Department ofInternal Medicine, Texas Tech University Health Since Center, Odessa, Texas, USA.
Corresponding Author: Ahmed Zedan, MD, Department of Internal Medicine, Texas Tech University Health Science Center, 800 W 4th St,Odessa, TX 79763, USA; Ph: (432) 335-2222; Email: ahmed.zedan@ttuhsc.edu, dr.ahmedzeidan@yahoo.com

Received: 09 April 2015
Accepted: 02 June 2015
Published: 01 October 2015

Patient was admitted for serial EKG and troponin follow-up, he was pain free during his hospital stay, and in one of the next EKG about 8 hours after admission biphasic T-waves in V1-V6 was seen (Figure 1) with minimal elevation in the troponin level to 0.10. The patient was taken for cardiac catheterization for further evaluation in light of these changes. Cardiac catheterization showed 99% occlusion of LAD (Figure 2), with grossly patent right coronary artery. He subsequently underwent balloon angioplasty and a drug-eluting stent placement in the proximal portion of the LAD. A follow-up left ventriculogram showed anterior apical wall hypokinesis with left ventricular ejection fraction (LVEF) of 45%. The patient continued to improve clinically and did not have any further episodes of chest pain or any shortness of breath during his hospital course. His follow-up EKG showed resolution of electrical changes from before. He was discharged a day later on aspirin, prasugrel, metoprolol and atorvastatin.

DISCUSSION

Wellens' syndrome is electrocardiographic (ECG) changes in the precordial T-wave segment, which are associated with critical stenosis of the proximal

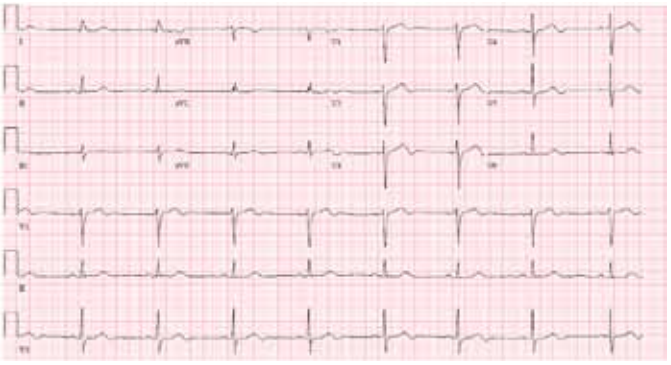


Figure 1: Electrocardiography performed eight hours after admission, showing biphasic T-wave inversion in leads V1–V6 (typical of type 1 Wellens' syndrome).

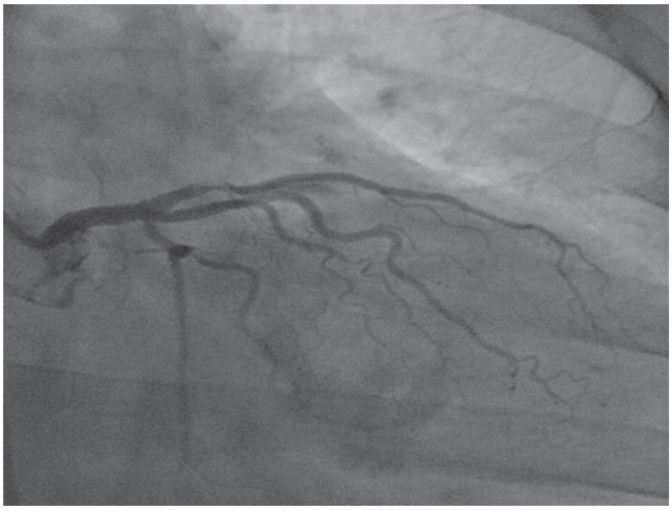


Figure 2: Coronary angiography performed next days after admission, showing a critical stenosis in the proximal part of the left anterior descending coronary artery.

left anterior descending (LAD) coronary artery. Electrocardiogram (EKG) is considered the key and gold standard investigation to diagnose acute coronary syndromes [1–3], at the same time it is important for predicting the site of the coronary stenosis. Based on this fact some patients admitted with unstable angina can be recognized as high risk considering the pattern of clinical findings and EKG changes associated with significant stenosis of the proximal (LAD) or as known as Wellens' syndrome.

Wellens' syndrome was first described in the early 1980s by de Zwaan et al. who identified a subset of patients with unstable angina with specific precordial T-wave changes and subsequently developed a large anterior wall myocardial infarction [4].Wellens' syndrome is a characteristic EKG findings described as classical T-wave changes in the precordial leads in pain free patient with history of intermittent chest pain [5]. These EKG changes in the absence of pathologic Q waves are predictive of a critical proximal LAD stenosis [2]. These EKG changes represent a re-perfusion of the LAD after a sudden occlusion causing a transient anterior ST elevation myocardial infarction (STEMI) that was unable to be captured on an EKG tracing.

Based on the EKG findings, Wellens' syndrome classified to 2 types [6], the most common is type 2 that occurs in about 75% of case with deep inversion of T-waves in V2-V3. Type 1 variant, characterized by biphasic T-waves in V2-V3 and occurs in about 25% of cases, more extensive T-wave inversion in pericardial leads can be seen with more proximal LAD lesion.

Criteria to diagnose Wellens' syndrome (Table 1) [7].

In one prospective study done in The Netherlands [8] 180 of 1,260 patients (14%) admitted to the hospital with unstable angina had the typical EKG pattern. 108 of these 180 patients (60%) had these EKG changes within the first 24 hours. All 180 patients had more than 50% stenosis of the left anterior descending artery (mean = 85% stenosis) with complete or near-complete occlusion in 59%. In Wellens' study [3], a group of 145 consecutive patients admitted for unstable angina, 26 (18%) had this EKG pattern, half on presentation and half within the next 24 hours. 75% of these patients went on to develop an anterior wall myocardial infarction despite relief of symptoms with initial medical therapy.

Table 1: Diagnostic criteria for Wellens' syndrome

History of intermittent chest pain
During episode of chest pain EKG is normal or mild elevation or depression of ST segment
Normal or mildly elevated cardiac enzymes
No pathological Q-waves in precordial leads or loss of R-waves progression
Deeply inverted or biphasic T-waves in V2 and V3, possibly V1, V4, V5 and/or V6 when pain free

Though T-wave inversion is a classical sign and well-known among physicians. The Wellens' syndrome type 1 is subtle and its significance is underappreciated. In this case report, we aim to increase awareness among Physicians in training and emergency room physicians about this ominous sign as 75% of these patients will develop acute anterior wall myocardial infarctions (MIs) within days unless intervention is undertaken appropriately.

In our patient, the history and EKG findings are classic for Wellens' syndrome with EKG showing the classic biphasic T-wave (Type 1) in the precordial leads. The patient had an episode of pain with subsequent relief just before presentation in the emergency room which was consistent with a reperfusion of the myocardium. His initial elevated troponin I confirmed that he had likely suffered some degree of myocardial infarction before presentation.

CONCLUSION

It is critical for emergency room physicians and medical residents to recognize and understand the Wellens' syndrome EKG finding. Early accurate identification of this pattern and early management of these patients by invasive revascularization can prevent progression to an extensive myocardial infarction and potential sudden cardiac death. It is also important to know that these patients should not undergo any form of cardiac stress test due to the danger of sudden cardiac death.

Keywords: Biphasic T-wave, Chest pain, Cardiac death Elevated cardiac enzymes, Wellens' syndrome

****

How to cite this article

Zedan A, Mukarram O, Malik U, Naidu R. Biphasic T-wave in patient with chest pain (Wellens' syndrome). Int J Case Rep Images 2015;6(10):655–657.

doi:10.5348/ijcri-201532-CL-10087

****

Author Contributions

Ahmed Zedan – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Osama Mukarram – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published
Umer Malik – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published
Raja Naidu – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

Authors declare no conflict of interest.

REFERENCES

1. Antman EM. Selwyn AP. Braunwald E. Ischemic heart disease. In: Harrison's. Principles of internal medicine. 17th ed. Saint Louis: Mc Graw Hill; 2008.
2. Smith SW, Whitwam W. Acute coronary syndromes. Emerg Med Clin North Am 2006 Feb;24(1):53–89, vi.
3. Strauss BH, Green M. Electrocardiographic prediction of ejection fraction and site of LAD occlusion in anterior myocardial infarction. Clin Cardiol 1993 Mar;16(3):213–7.
4. de Zwaan C, Bär FW, Wellens HJ. Characteristic electrocardiographic pattern indicating a critical stenosis high in left anterior descending coronary artery in patients admitted because of impending myocardial infarction. Am Heart J 1982 Apr;103(4 Pt 2):730–6.
5. Nisbet BC, Zlupko G. Repeat Wellens' syndrome: case report of critical proximal left anterior descending artery restenosis. J Emerg Med 2010 Sep;39(3):305–8.
6. Sobnosky S, Kohli R, Bleibel S. Wellens' Syndrome. Int J Cardiol 2006;3:1.
7. Tandy TK, Bottomy DP, Lewis JG. Wellens' syndrome. Ann Emerg Med 1999 Mar;33(3):347–51.
8. de Zwaan C, Bär FW, Janssen JH, et al: Angiographic and clinical characteristics of patients with unstable angina showing an ECG pattern indicating critical narrowing of the proximal LAD coronary artery. Am Heart J 1989 Mar;117(3):657–65.

CLINICAL IMAGES PEER REVIEWED | OPEN ACCESS

Child with mucopolysaccharidosis type IV: Morquio syndrome

Ali Akhtar, Sabina Manandhar, Eswat Ahmad

CASE REPORT

A five-year-old girl was presented at Armed Forces Institute of Radiology and Imaging for skeletal survey. On clinical examination she had corneal clouding, coarse facial features, short stature, and kyphotic deformity of spine. On radiography, skull was enlarged and J shaped sella, spine showed increased curvature and anterior beaking of vertebra. X-ray chest showed anterior widening of ribs (oar shaped ribs). Pelvis had widely flared iliac bones. Hand X-ray revealed proximal pointing of metacarpals of both hands. There was no mental retardation. She was diagnosed with mucopolysaccharidosis type IV: Morquio syndrome.

DISCUSSION

Mucopolysachharidoses (MPSs) are a family of rare, inherited (autosomal recessive) lysosomal storage disorders caused by deficiency of an enzyme involved in the degradation of glycosaminoglycans (GAGs): heparan sulfate, dermatan sulfate, keratan sulfate and chondroitin sulfate. They are classified as MPS I to MPSVII on the basis of clinical and biochemical studies [1].

Morquio and Brailsford reported cases of a disorder characterized by short neck, pectus carinatum, genu valga, pes planus, odontoid hypoplasia and normal intelligence independently. In 1960, this disorder was characterized as MPS caused by the lysosomal accumulation of GAGs and urinary excretion of the keratan sulfate [1].

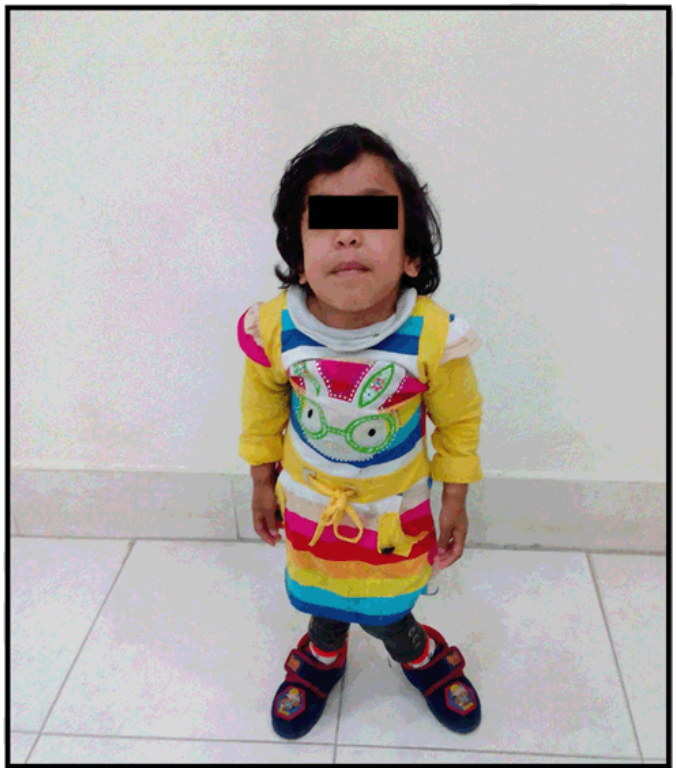


Figure 1: Child has short stature.

Matalon et al. discovered MPS IVA as caused by deficiency of galactosamine-6-sulfate. Arbisser et al. described a patient with normal N-acetylgalactosamine-6-sulfate but deficient lysosomal b-galactosidase known later to be MPSIVB, the milder one [1].

Patients with MPS IVA appear normal at birth with normal intellect and experience clinical onset of disease during early childhood. The most common presentation is skeletal deformity and growth retardation in the second or third decade of life [2, 3]. The child in our case was

Ali Akhtar[1], Sabina Manandhar[2], Eswat Ahmad[1]
Affiliations: [1]Army Medical College, Rawalpindi, Punjab, Pakistan; [2]Kathmandu University School of Medical Sciences, Nepal, manandharsabina01@gmail.com
Corresponding Author: Ali Akhtar, House No. 1007, Street No. 67, Sector G 9/4, Islamabad, City: Islamabad, State: Capital, Country: Pakistan, Postal Code: 44000; Ph: 0092 336 9303267; E-mail: aliakhtar126@gmail.com

Received: 13 June 2015
Accepted: 12 August 2015
Published: 01 October 2015

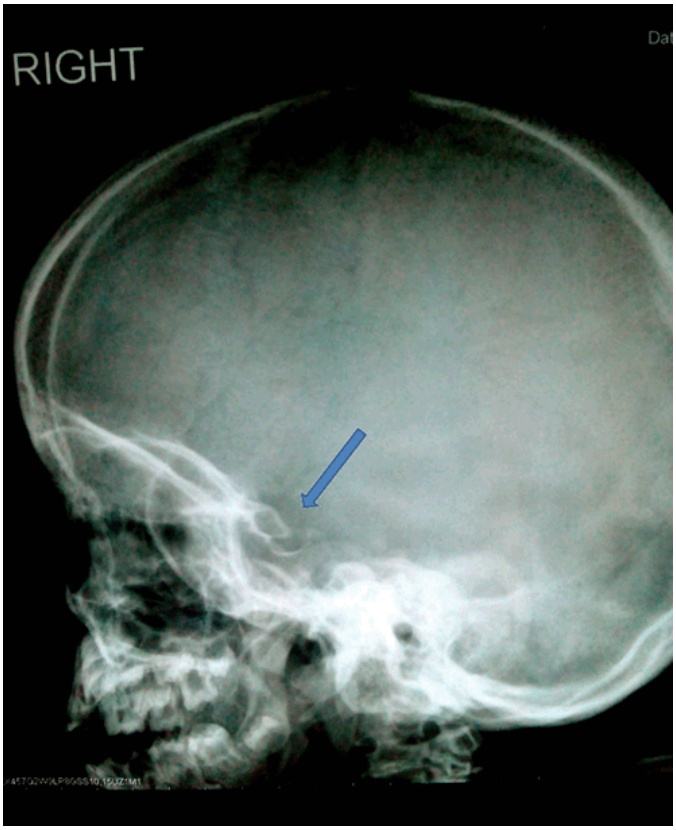


Figure 2: Arrow showing J-shaped Sella.

also normal at birth with normal intellect. Other clinical features of Morquio syndrome include [4–6]:

- Heart and valvular defects
- Joint hypermobility, knock-knee deformity, genu valgus and large fingers
- Widely spaced teeth with thin tooth enamel
- Bell shaped chest
- Pectus Carinatum
- Atlanto-axial instability, odontoid hypoplasia, myelopathy
- Scoliosis, compression of spinal cord
- Short neck and dwarfism
- Mild dysostotic multiplex, dysplastic hips, large unstable knees, large elbows and wrist and flat feet
- Progressive deafness
- Mild hepatosplenomegaly
- Mid face hypoplasia, mandibular protrusion
- Visual impairment
- Thin enamel with dentine visible, pitting and hypoplastic effects and sharp pointed cusps

No dental abnormalities were noted in our case.

In 2012, a familial tendency of unknown etiology has been described by Rekha et al. where three siblings in the same family were affected with the syndrome [7]. In 1952, Garn and Hurme described abnormalities of the teeth in nine siblings. Of them, three siblings showed thin enamel layer and in some places the dentine actually showed through. The surface was marked by numerous pits, and the enamel appeared to be structurally weak since it exhibited a tendency to fracture and flake off. In contrast, the six unaffected sibs had normal dentitions [8]. In our case, no other siblings were effected and history of consanguineous marriage was not elucidated.

There are two treatment options for patients with MPS – Hematopoietic stem cell transplantation and recombinant intravenous enzyme replacement therapy. Early diagnosis and treatment can improve patient outcome and prolong survival [9].

This report outlines the clinical and radiological findings found in a case of Morquio syndrome. An accurate diagnosis typically requires the recognition of specific clinical and/or radiographic signs and symptoms together with laboratory confirmation. The radiologist can play a critical role in ensuring that an accurate diagnosis is reached expeditiously by raising suspicion of an MPS disorder if dysostotic multiplex changes are evident.

CONCLUSION

Mucopolysaccharidosis (MPS) is a multisystem disorder and its diagnosis is based on clinical finding. Though it is a rare disease but its appropriate management and investigations are required to reach its diagnosis and treatment. Understanding the symptoms and progressive nature of MPS IV will provide a solid basis for evaluating the efficacy of treatment modalities.

Keywords: Glycosaminoglycans, Joint hypermobility, Morquio syndrome, Mucopolysachharidoses, Visual impairment

****

How to cite this article

Akhtar A, Manandhar S, Ahmad E. Child with mucopolysaccharidosis type IV: Morquio syndrome. Int J Case Rep Images 2015;6(10):658–660.

doi:10.5348/ijcri-201533-CL-10088

****

Acknowledgements
We are thankful to the patient and her parents. We also acknowledge Dr. Neelum for her assistance and time.

Author Contributions
Ali Akhtar – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for

important intellectual content, Final approval of the version to be published
Sabina Manandhar – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Eswat Ahmad – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor
The corresponding author is the guarantor of submission.

Conflict of Interest
Authors declare no conflict of interest.

Copyright

REFERENCES

1. Gorlin RJ, Cohen MM, Hennekam RCM. Syndromes of the Head and Neck. 4th edn. New york: Oxford University Press; 2001.
2. McIntosh N, Helms P, Smyth R. Forfar and Arneil's Textbook of Pediatrics. 6th Ed. Philadelphia: Churchill Livingstone Elsevier; 2003. p. 1241–42.
3. Tidy C. Morquio Syndrome. [Internet]. [Cited 2013 Apr14]. [Available at: http://www.patient.co.uk/doctor/Morquio's-Syndrome.htm]
4. Rimoin DL, Connor JM, Pyeritz RE, Korf BR. Emery and Rimoin's Principles and Practice of Medical Genetics, vol-3. 5th Ed. Philadelphia: Churchill Livingstone Elsevier; 2007. p. 2403–12.
5. Wheeless CR. Morquio's Syndrome. [Internet]. [Updated 2011 Sep 9; Cited 2013 Apr14]. [Available at: http://www.wheelessonline.com/ortho/morquios_syndrome.]
6. Lee NH, Cho SY, Maeng SH, et al. Clinical, radiologic, and genetic features of Korean patients with Mucopolysaccharidosis IVA. Korean J Pediatr 2012 Nov;55(11):430–7.
7. Rekka P, Rathna PV, Jagadeesh S, Seshadri S. Mucopolysaccharidoses type IV A (Morquio syndrome): a case series of three siblings. J Indian Soc Pedod Prev Dent 2012 Jan-Mar;30(1):66–9.
8. Garn SM, Hurme VO. Dental defects in three siblings afflicted with Morquio's disease. Br Dent J 1952;93:210–2.
9. Tomatsu S, Sawamoto K, Alméciga-Díaz CJ, et al. Impact of enzyme replacement therapy and hematopoietic stem cell transplantation in patients with Morquio A syndrome Drug Des Devel Ther. 2015 Apr 1;9:1937–53.