InnoVationInfo Journal of Pediatrics and Infants

ISSN: 2581-7493
Volume 1: 1
J Pediat Infants 2018

Management Dilemma of Anterior Mediastinal Desmoid Tumour in a Three-Year-Old Boy

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Abstract

Desmoid tumour is a rare, benign, local invasive and fibroblastic proliferative tumour of deep soft tissues. It affects more females than males with a ratio of 2:1. The reported incidence is 2 to 4 per million per year in the population. The tumour is linked to familial Adenomatous Polyposis (FAP) and some of the risk factors associated with it include pregnancy and previous trauma. Our case is of a three year old boy who initially presented at age of 6 months, asymptomatic. The radiological investigations included chest X-ray (CXR), Computed Tomography (CT) and Magnetic Resonance Imaging (MRI) which were typical of desmoid tumour. The en-bloc resection of the tumour was undertaken via partial upper median sternotomy and the chest was closed primarily.

Histology diagnosis confirmed a desmoid tumour and a patient was discharged home five days later without complications. He was not offered radiotherapy/chemotherapy. The patient came for a two week follow-up with no signs of residual tumour of both physical examination and CXR. The patient presented two years later after defaulting for subsequent follow-ups with recurrent large, invasive tumour which posed management dilemma. The dilemma was that a tumour required mutilate resection and extensive reconstructions, which could comprise respiration mechanics and his general functional state. The multidisciplinary team including plastic surgeons, thoracic surgeons, medical oncologist and radiologist opted for medical treatment.

Keywords: Desmoid tumour, Desmoid –type fibromatosis, Anterior mediastinal tumour.

Introduction

Desmoids –type tumour (DF) is a benign, fibroblastic proliferative tumour of deep soft tissues, which is characterized by local invasion, but does not metastasize. The reported incidence is 2-4 individuals per million per year [1]. they account to 3.5% of fibrous tumours and 0.03% of all neoplasms [2]. They are more common in females than males with a ratio of 2:1. They commonly affect individuals aged 15-60. Two thirds of these tumours are intra-abdominal [3]. Intrathoracic DF are very rare. Most tumours are sporadic. About 7.5% of these tumours are linked to Familial adenomatous Polyposis (FAP) [3]. Hormonal changes during pregnancy trauma and/prior surgery are known to be risk factors for development of DF. Surgery with wide tumour resection remains a gold standard for treatment. However, the recurrence rate remains higher ranging between 29-54% despite leaving negative surgical resection margins.

Case Report

Our case is of a three-year-old boy who had previously been referred by his mother to our unit for a mass that was growing on thoracic inlet

Article Information

Article Type: Case Report
Article Number: JPI108
Received Date: 06 April, 2018
Accepted Date: 22 May, 2018
Published Date: 28 May, 2018

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Citation: Sekgololo JM, Chauke RF (2018) Management Dilemma of Anterior Mediastinal Desmoid Tumour in a Three-Year-Old Boy. J Pediat Infants Vol: 1, Issu: 1 (31-36).

and extending to anterior chest wall while he was still six months old. A mass was reported to be noticed one month after birth and was about 0.5 × 0.5 × 1 CM (about 10 cents coin size). It was painless, firm with no signs of inflammation. It grew rapidly and quintupled in size at 6 months.

There was no history of trauma/surgery and familial Gardner's syndrome. The child was asymptomatic on presentation. He was not coughing, not having pain, no shortness breath and arm weakness were evident. He had systolic blood pressure (SBP) of 80mmHg, heart rate: 90 bpm, respiratory rate: 24 breaths/minute and all were within normal limits for his age and body size. Growth curve did not indicate evidence of failure to thrive. The chest examination showed a multilobulated mass, fixed on thoracic inlet and extending to anterior chest wall, with no signs of previous trauma/surgery. It was firm, non-tender and measured about 5 × 6 × 5 CM.

As part of work-up Beta human Choriogonadotropin, alpha fetoprotein and urine catecholamines were done and were all within normal limits. A plain chest X-ray with anteroposterior and lateral view showed opacity obliterating retrosternal area and extending to anterior chest wall Figure 1A.

Chest CT-scan showed a well circumscribed non-enhancing mass extending from anterior mediastinal wall to the anterior surface of a sternum and parasternal areas. The manubrium and upper part of sternal body were invaded (Figure 2A-D). MRI showed heterogeneous contrast enhanced mass displacing the vascular structures with areas of decreased enhancement suggesting presence of necrosis. The sagittal cuts showed tumour extension from anterior mediastinum to anterior chest wall.

A multidisciplinary team of plastic surgeons, thoracic surgeons, oncologist, psychologist, social workers and radiologist met to discuss on best management for the child. The team agreed on operating the child as a matter of urgency because of possible airway compromise due to rapid growing nature of the tumour. The operation was undertaken three days later by the team of plastic and thoracic surgeons. The operative approach was through partial upper median sternotomy and a tumour was dissected from surrounding tissues using both blunt and sharp dissection Figure 3. The en-bloc resection of a mass was undertaken. The tumour measured 10 × 6 × 7 CM, was lobulated and circumscribed. There was no resection of bones and chest wall muscles involved, only periosteum of posterior and anterior surface of manubrium and upper sternal body were stripped off. The primary closure of the sternum was undertaken after the procedure.

The child was discharged 5 days after operation on non-steroidal anti-inflammatory drugs without complications. No radiation and or chemotherapy were offered to the child. At two weeks follow-up the child was asymptomatic and thriving well. The chest X-ray did not show any sign of residual tumour Figure 1B and histology confirmed desmoids tumour.

The histology showed proliferative spindle shaped cells, which were arranged in fascicles compressing small blood vessels. The S 100 marker was negative, which ruled out neural tumour in origin. The beta-catenin marker was positive which is in keeping with desmoid tumour (Figure 4A-D).

The patient defaulted follow-up which was scheduled after three months. The patient was brought back to hospital two years later (at the age of three) with recurrent large invasive tumour of the same size as the resected one. The radiological investigation showed that it was the same size as the previously resected one Figure. The sternum was

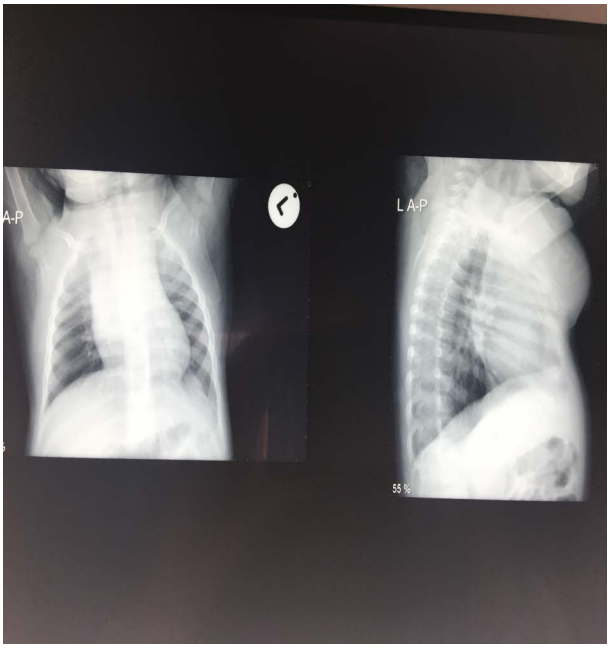


Figure 1A: CXR of anterior mediastinal DT extending to anterior chest wall anteroposterior view depicts enlarged superior mediastinum and lateral view shows opacity on the anterior chest wall.

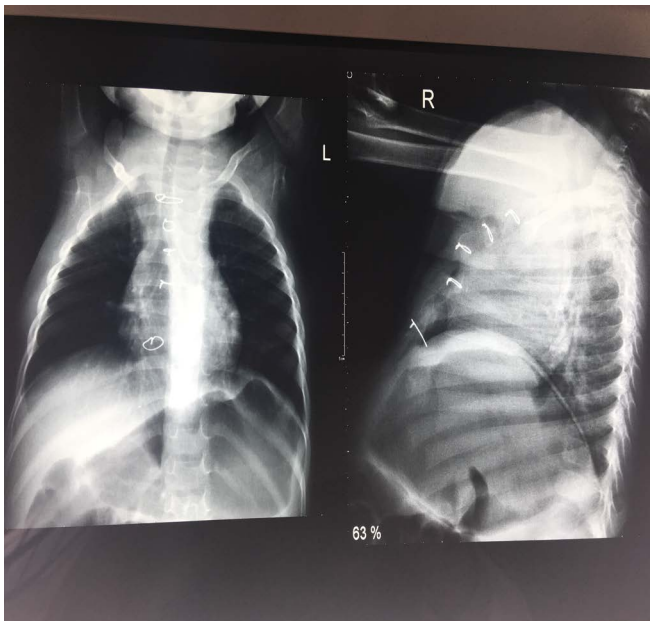


Figure 1B: CXR of anterior mediastinal DT extending to anterior chest wall Post- operative CXR anterior posterior view shows relatively normal sized mediastinum and lateral view shows the absence of anterior opacity as seen on pre-operative films.

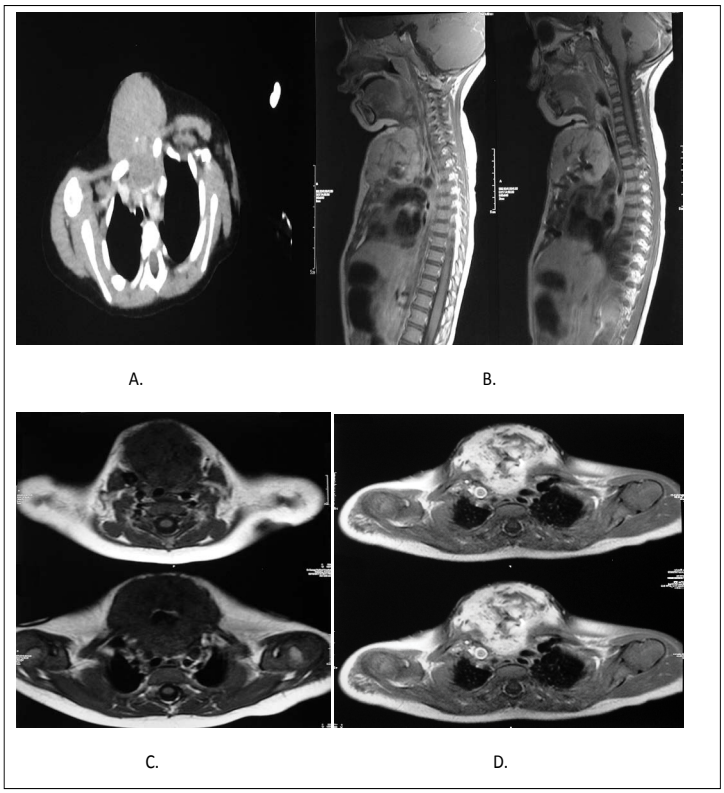


Figure 2: Desmoid tumour of the anterior mediastinum extending to the anterior chest wall. **A**: Axial cut of contrasted CT-scan depicting a well circumscribed non-enhancing tumour extending from anterior mediastinum to the anterior chest wall, with invasion to sternum; **B**: Coronal cuts of MRI without contrast showing tumour extension from anterior mediastinum to anterior chest wall; **C**: shows a tumour displacing vessels on an axial section of T1-weighted MRI without contrast; **D**: Axial section of T1- weighted MRI with contrast showing heterogeneous enhancement with displacement of vessels, and focal areas of decreased enhancement suggesting necrosis.

destroyed except the lower third part and vessels were encased by the tumour (Figure 5A,B). The child was still asymptomatic except that there was evidence of failure to thrive on a growth curve.

The multidisciplinary team decided on treating the child non-operatively with a trial of chemotherapy. The child was started on Vinblastine and Methotrexate once weekly for six months. On the fourth month of treatment, the tumour did not regress, instead it increased in size on examination. The team stood on its decision not to operate because of possible mutilation and extensive reconstructions, which could compromise the child's respiratory mechanics and general function.

The mother requested that the child be transferred to their local hospital for convenience, and transfers were arranged. The child could not be traced back after that.

Discussion

Desmoid tumours (DTs) are a group of rare benign soft tissue tumours which result from monoclonal proliferation of well-differentiated fibroblast [4]. Desmoid-type fibromatosis (DF) is also known as desmoid tumour or musculoaponeurotic fibromatosis. It is classified by WHO as intermediate grade because of its invasive growth and absence of metastatic potential [5]. It has shown the aggressive and invasive features also in our case.

They account for 0.03% of all neoplasia with estimated incidence ranging from 2-4 per individual per million per year. The women are more likely affected than males with a ratio of 2:1. The incidence of DF has been reported to be 1000-fold higher in patients with Familial Adenomatous Polyposis (FAP), in which Adenomatous Polyposis Coli (APC) gene is mutated [6]. The risk factors associated with DT are pregnancy (increased oestrogen), physical trauma and Gardner 's syndrome [7]. In our case none of the above risk factors were found.

Morphology

The DT is yellowish-white on cross-section with some fasciculations. The DTs are variable in size and are often poorly circumscribed. The histology is characterized by fibroblastic proliferative and myofibroblast type of spindle cells [2]. In our case, the cells were proliferative, spindle shaped arranged in fascicles and compressing on small blood vessels. Beta catenin is positive in 80% of the neoplasm as it was in our case [1]. The S 100 marker is usually negative in DT, which helps to rule out neural tumours in origin. In our case, S100 was negative, confirming diagnosis of DT.

The DT can present into two forms, as slow or rapid growing tumour. It can be diagnosed accidentally when doing an X-ray for unrelated conditions or noticed as an asymptomatic growing mass as in our case. It becomes symptomatic when it compresses the proximal structures.

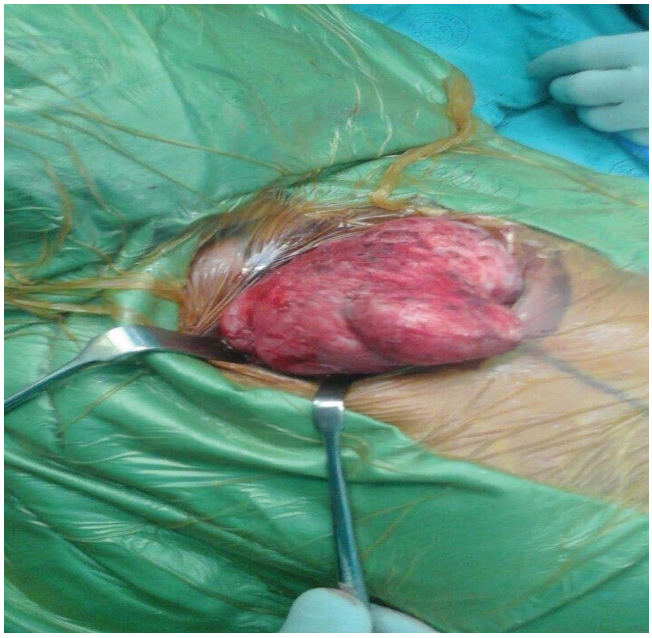


Figure 3: Desmoid tumour is extrioriorized through partial upper median sternotomy.

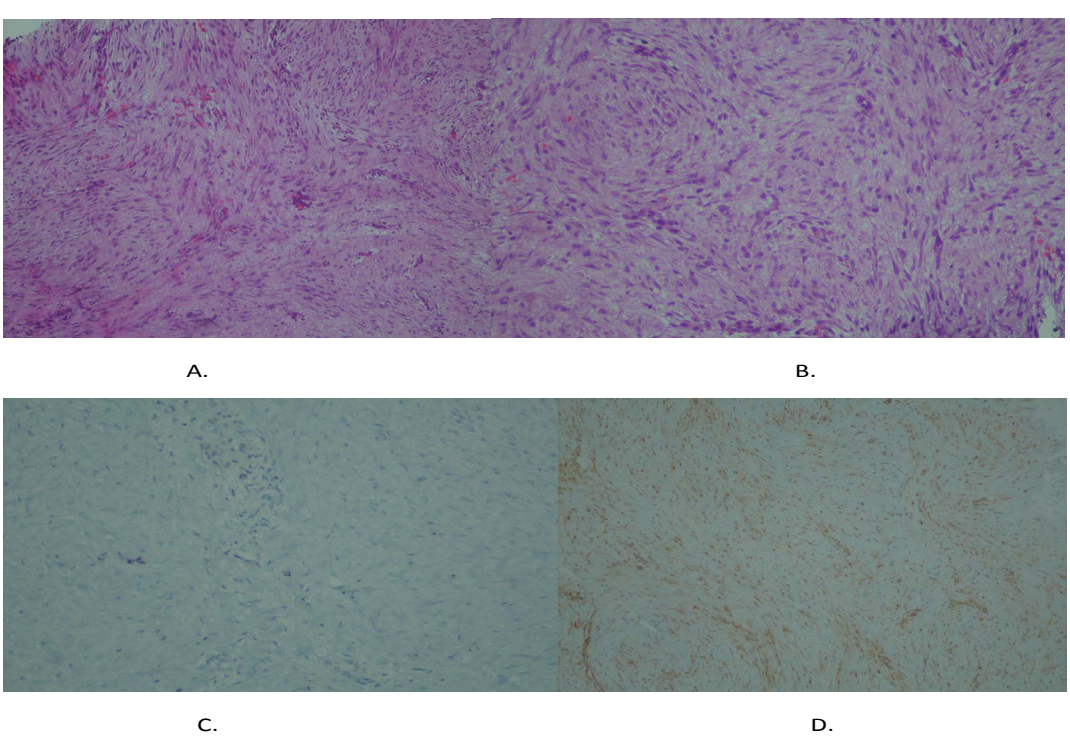
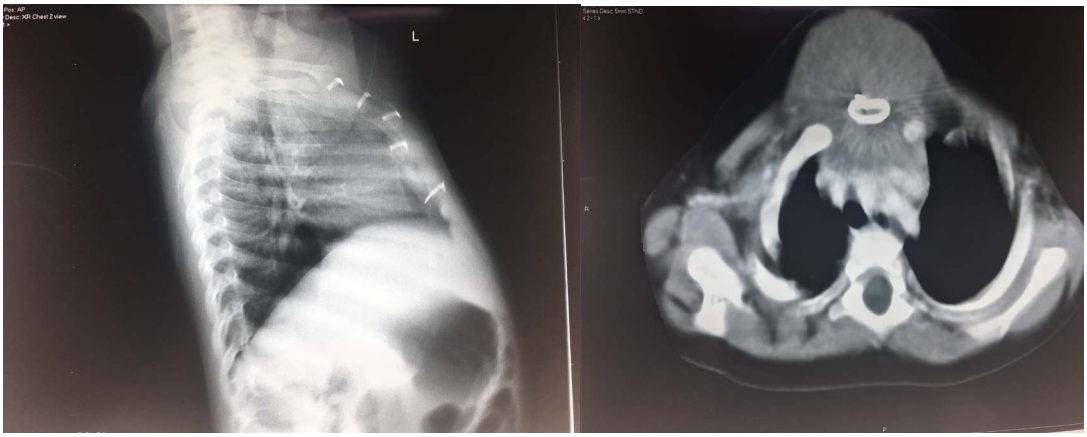


Figure 4: Desmoid tumour histology. **A** and **B**: low magnification haematoxylin and Eosin 10 X and 20 X depicting proliferative spindle shaped cells arrange in fascicles compressing small vessels; **C**: Low magnification 10 X depicting negative S100 marker; **D**: Low magnification 10 X depicting positive Beta-catenin marker.

Compression symptoms of neurovascular structures include pain and paraesthesia. The respiratory symptoms include cough, chest pain, dyspnoea and recurrent chest infections. The compression of cardiovascular symptoms by intrathoracic DT include dyspnoea due to decrease cardiac output following a decreased preload. In our case, the child was asymptomatic before initial operation and after recurrence.

A chest X-ray (CXR) is a first line radiological investigation. It will show homogenous opacity, which is sometimes ovoid in shape, as in our case. In our case CXR on anteroposterior view showed enlarged superior mediastinum and lateral view showed opacity obliterating retrosternal space with an extension to anterior chest wall. CT-scan defines the exact position of the tumour, its proximal relations and helps in planning surgical treatment. The DT tumours on



A. B.

Figure 5: Recurrent desmoid tumour. **A**: Lateral CXR showing opacity on anterior chest wall extending from manubrium to upper third of the sternal body; **B**: CT-scan with contrast depicting non-enhacing tumour with a stainless wire that is isodense to the bones.

CT-scan are usually isodense to the skeletal muscles, which was also seen in our case. MRI is a mainstay investigation in DTs. It can be used for primary diagnosis, local surgical staging and monitoring of the tumour [1]. It also shows the relationship of the tumour to the proximal structures such as blood vessels, nerves and bones. T1-weighted in our case may showed heterogeneous signal intensity in keeping with variable quantities and distribution of myofibroblast. These are features confirming DT.

Histology gives a definitive diagnosis. Features confirming DT on histology are proliferative spindle shaped cells arranged in fascicles and compressing small blood vessels. The eosinophilic background of collagen is typical. These features are elicited of histology of our patient. The molecular biological studies can be added to confirm diagnosis in case of inconclusive histology results. A Beta-catenin positive marker strongly suggests DT. It is seen in more than 80% cases of DT and was also positive in our case.

Surgical treatment

Surgery is the mainstay of treatment. The approach depends on location of the tumour. Resection should be with wide margins aiming at achieving negative resection margins, which can be difficult due to proximity of vital structures and its local invasive nature. Regular follow up is mandatory due to high recurrence rate of 29-54% [8]. In our case patient defaulted follow-up and presented with recurrent locally advance tumour which was not amenable to surgery according to our team of experts. The adjuvant radiation has shown to decrease recurrence in case of a residual tumour/positive microscopic margins.

Medical treatment

It has been attempted in cases were patient is inoperable/ surgery is unsuccessful as in our case after recurrence. Medical treatment using non-cytotoxic drugs include hormonal therapy (anti-oestrogen), non-steroidal anti-inflammatory and interferon alpha. Cytotoxic drugs include Vinblastine (VBL) and Methotrexate MTX), which were used in our case. The combination of VBL and MTX is favoured in children because it is tolerable.

Conclusion

Desmoids tumours are rare, benign, fibroblastic proliferative tumours characterized by local invasiveness and they are non-metastatic. Gender ratio is usually 2:1 (F: M), and they commonly affect age group 15-60 years. They are often asymptomatic or can present with signs and symptoms of compression. The radiological investigations for DT are typical. The histology is a definitive mode of diagnosis, but in case on inconclusive results molecular biological markers such as Beta-catenin and S100 can be used. The gold standard treatment is surgical resection of the tumour with wide margins. The follow-up is mandatory because of high recurrence rate.

Acknowledgements

The authors thank:

- Dr T Kekana, Dr Bapela and team (anatomical pathologists) for their assistance with histopathology analysis
- Dr E Seleka (language editor)
- Team of thoracic and plastic surgeons involved
- The department of Radiology (Dr George Mukhari Academic Hospital)

Additional Information

Patient Consent

Consent to publish this case was not obtained because, the report does not contain any personal information that could lead to the identification of the patient.

Authorship

As authors, we attest to meet the current ICMJE criteria for Authorship.

Conflict of interest

The authors (Dr. Sekgololo Joseph Motshedi and Prof. Chauke Risenga Frank) have no financial disclosures.

Supplementary Document

Choose The Best Answear From The Following

1. The following are histological features of desmoid tumour except...

A. proliferative fibroblasts

B. proliferative myofibroblasts

C. high cellularity

D. spindle shaped cells

2. A gold standard investigation for desmoid tumour is...

A. Chest x-ray

B. CT-scan and MRI

C. Histology

D. Molecular biology

3. The best treatment of desmoid includes...

A. Surgery only

B. Surgery and chemotherapy

C. Chemotherapy and radiation only

D. Radiotherapy and surgery

4. Which of the following is a risk factor for desmoid tumour?

A. Gender

B. age

C. Familial adenomatous polyposis (FAP)

D. smoking

Answers for questionnaires are as follows: 1.c 2.c. 3.d. 4.c

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