International Case Reports Journal
Case Report (ISSN: 2770-9647)

# Soft Tissue Rhabdoid Tumour in a 19 Year Old African Male: A Case Report

**Thulo Molefi[1*], Christa Solomon[2]**

[1]Department of Medical Oncology, Faculty of Health Sciences, University of Pretoria, Pretoria, South Africa

[2]Department of Anatomical Pathology, National Health Laboratory Services, Faculty of Health Sciences, University of Pretoria, Pretoria, South Africa

## ABSTRACT

Rhabdoid Tumours (RT) is rare, rapidly progressive neoplasms that typically occur in childhood; are often metastatic at presentation and are known for their high mortality. These malignancies are even rarer in adults, occur at a variety of anatomic locations and are classified into three categories. A 19 year old male with no pre-existing illnesses or family history of malignancy, presented with metastatic malignant extrarenal, extracranial rhabdoid tumour (MERT) and after a short period of investigation and therapy demised within a year of his diagnosis. A definitive chemotherapy regimen is yet to be identified for this malignancy and despite the identification of a candidate drug target; the management of rhabdoid tumours remains a therapeutic challenge. Further study is required to underpin the molecular biology of this malignancy and with better understanding, targeted therapy will be discovered and applied; particularly for irresectable and metastatic disease.

**Keywords:** MERT, Rhabdoid Tumour, SMARCB1

**Citation:** *Thulo Molefi, Christa Solomon. Soft Tissue Rhabdoid Tumour in a 19 Year Old African Male: A Case Report. Int Case Rep Jour. 2022;2(4):1-5. Doi: https://doi.org/10.5281/zenodo.6382149*

**Received Date:** 02 March, 2022; **Accepted Date:** 15 March, 2022; **Published Date:** 19 March, 2022

***Corresponding author:** Thulo Molefi. Department of Medical Oncology, Faculty of Health Sciences, University of Pretoria, Pretoria, South Africa



## INTRODUCTION

Rhabdoid Tumours (RT) are rare, rapidly progressive neoplasms that typically occur in childhood.[1] They are often metastatic at presentation and are known for their high mortality.[2]

These malignancies are even rarer in adults.[1] RT's occur at a variety of anatomic locations and are classified into three categories: malignant rhabdoid tumour of the kidney (most common), Atypical Teratoid/Rhabdoid Tumour (AT/RT) of the brain (second most common) and malignant extrarenal, extracranial rhabdoid tumour (MERT).[3]

Only two South African paper's reporting paediatric cases of AT/RT and cutaneous rhabdoid tumours have been published[4,5] with no adult case reports from Africa found in literature. This case report describes a young adult male with MERT seen at Steve Biko Academic Hospital's (SBAH) department of medical oncology.

## CASE REPORT

A 19 year old male with no pre-existing illnesses or family history of malignancy, was referred to SBAH's department of medical oncology after incomplete resection of a right thigh mass 9 months earlier.
At presentation he had a ECOG performance state (PS) of 2 with a body mass index of 17.1 kg/$m^2$. He had rib and pelvic bone pain, local recurrence in his right upper thigh (9x11cm mass), a mass on his upper back (6x6cm), a perianal mass (8x8cm) and localised rib tenderness at his lower left hemithorax.

A Positron Emission Tomography-Computed Tomography (PET/CT) 2 months prior to presentation documented tumour recurrence at his right thigh, increased uptake in multiple lymph node regions (inguinal, iliac, paraaortic, paratracheal, precarinal and hilar) with additional metastases to his bones ($7^{th}$ left rib, L4 vertebrae) and pleura.
The resection specimen consisted of a polypoid, tan-white mass measuring 8x7x2.9cm surfaced by skin. The specimen weighed 96g. Areas of haemorrhage could be seen.

Light microscopy yielded a fairly circumscribed, nonencapsulated and multinodular tumour involving the dermis and extending into the deep subcutaneous tissue and peripheral excision margins.

The tumour was composed of a solid proliferation of polygonal cells with prominent rhabdoid features. The vesicular nuclei were eccentric and nucleoli prominent. The abundant cytoplasm was brightly eosinophilic and numerous atypical mitoses could be identified. Multinucleated giant cells as well as evidence fresh and old hemorrhage were noted.

No clear line of differentiation could be established as the tumour proved positive for EMA, CK7 (patchy), FLI1 and CD99 only.

A pancytokeratin (AE1/AE3), TLE1, desmin, myogenin, myoD1, S100, HMB45, melanA, SOX10, CD34, CD31, factor VIII, CD138, CD56, MUM1 and SMA proved negative.

The tumor cells showed loss of expression of the SMARCB1 protein (INI1).

The cytomorphology and immunohistochemical profile were in keeping with a Malignant Extrarenal Rhabdoid Tumour (MERT).

Two weeks after his initial visit, he was started on combination chemotherapy with adriamycin (30g/$m^2$, day 1) and ifosfamide (3750mg/$m^2$) with mesna renoprotection on a day 1 and 2 schedule per cycle, and zolendronate to reduce the risk of skeletal related events.

Three weeks later when presenting for his 2nd cycle, his PS was 3 with a 3 day history of lower limb weakness (power 3/5), with bowel and bladder incontinence and 3x3 cm, grade 2 decubitus ulcer. A magnetic resonance imaging (MRI) scan confirmed spinal cord compression by a new mass at T6/7 vertebrae with early infiltration of the thoracic aorta, left hemidiaphragm and spleen, new lung, pleural and diffuse bone metastases. He was started on steroids and referred to the radiation oncology department for palliative radiotherapy.

Four weeks later his PS had now diminished to 4, his sacral ulcer was necrotic and was then down referred to his local hospital for supportive care and he infelicitously demised 5 days later.

## DISCUSSION

MERT's are rare and aggressive tumours, as confirmed in this case report. Because of their rarity they present a management challenge. Multiple agents have been used for this malignancy with sombre results[1,2], however a definitive chemotherapy regimen is yet to be identified, despite the inclusion of adriamycin being recognised as an important measure for survival.[1]

Monosomies, translocations and deletions involving chromosome 22 were the first genetic abnormalities identified with later studies recognising SMARCB1 aberrations, on the long arm of this chromosome, as the main oncogenic driving protein. Multiple therapies targeting this molecule, directly or indirectly, have been investigated with varying non-practice changing results.[2] A subtype of these tumours has also been found to be SMARCB1 negative[1] and this occurrence and its implication requires further evaluation.

Rhabdoid tumours at different sites and age groups have been shown to behave disparately[1,2] and further study is required to underpin this divergence. Optimistically with more cases being reported globally, this cancer's molecular biology will be better understood and targeted therapy will be discovered; particularly for irresectable and metastatic disease.(Figures 1-5)

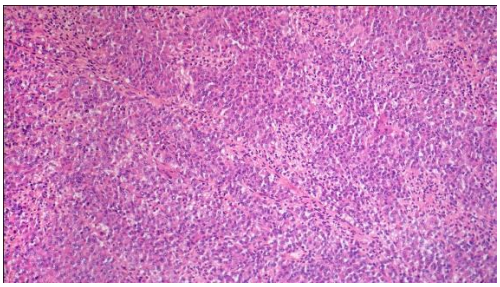

**Figure 1:** Hematoxylin and eosin preparation of the thigh mass.
Sheets of uniform epithelioid cells with prominent nucleoli and abundant eosinophilic cytoplasm (100 X).

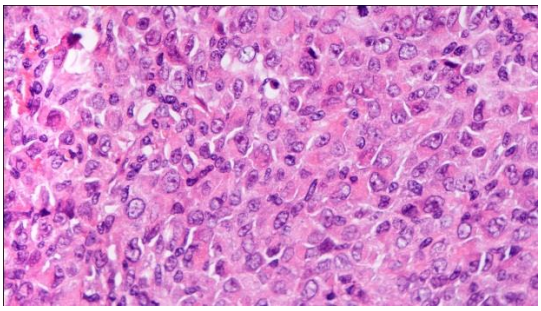

Figure 2: Hematoxylin and eosin preparation of the thigh mass.

Tumour with prominent rhabdoid features (400 X).

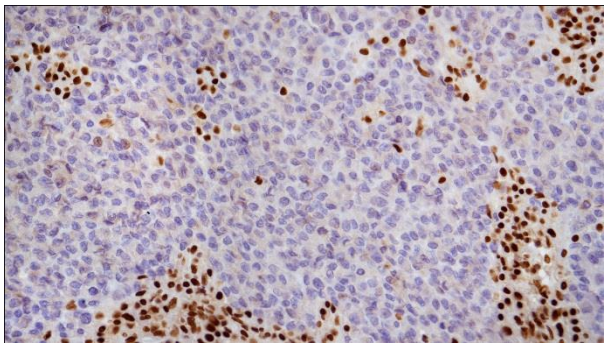

**Figure 3:** Tumour cells showing loss of expression of the SMARCB1/INI1 protein (INI1 immunohistochemical stain - 200X).

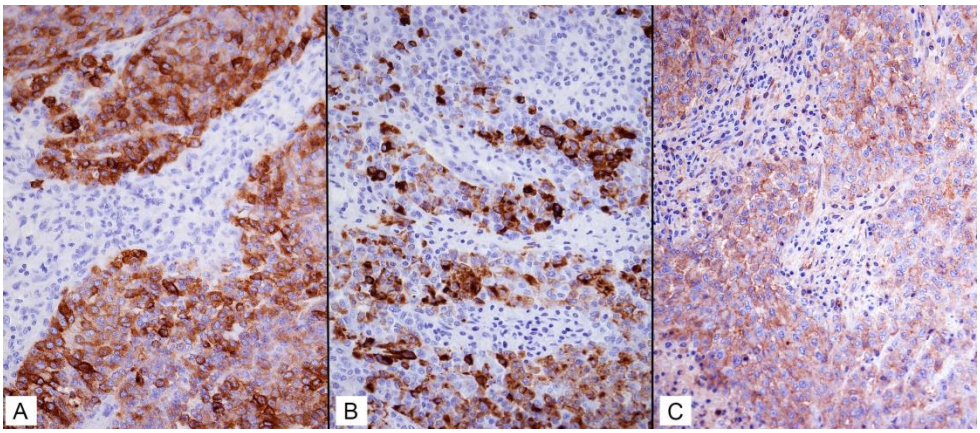


**Figure 4:** The tumour showed expression of EMA, CK7 and CD99.

**Figure 4A:** EMA immunohistochemical stain (200X)

**Figure 4B:** CK7 immunohistochemical stain (200X)

**Figure 4C:** CD99 immunohistochemical stain (200X).

**References**

1. Brennan B.M.D, Stiller C, Bourdeaut F. Extracranial Rhabdoid Tumours: What We Have Learned so far and Future Directions. Lancet Oncol. 2013;14:329-336.

2. J C Fanburg-Smith, M Hengge, U R Hengge, J S Smith Jr, M Miettinen. Extrarenal Rhabdoid Tumours of the Soft Tissue: A Clinicopathologic and Immunohistochemical Study of 18 Cases. A Ann Diagn Pathol. 1998;2(6):351-62.
3. Molina N, Leis A. Extrarenal Rhabdoid Tumor of the Brachial Plexus in A Five-Year-Old Female: A Case Report and Review of the Literature. J Pediatr Surg Case Rep. 2016;15:5-9.
4. N Mahomed, J Naidoo, S Dlangamandla, S Andronikou, S Pather, K Pillay. Atypical Teratoid/Rhabdoid Tumour in A Supratentorial Location: A Report of Two Cases. South African Journal of Child Health. 2015;8(4):159-161.
5. Davies MRQ, Mogilner JG. Congenital Malignant Rhabdoid Tumour of the Skin. Pediatr Surg Int.1991;6: 230-232.