

PARA-RECTAL ALVEOLAR RHABDOMYOSARCOMA IN A YOUNG ADULT REVEALED BY ANASARCA: A MISLEADING PRESENTATION WITH FATAL OUTCOME IN A RESOURCE-LIMITED SETTING

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ABSTRACT

Alveolar rhabdomyosarcoma (ARMS) is a rare and highly aggressive malignant soft-tissue tumour in adults, in whom the 5-year overall survival does not exceed 20–30% in recent cohorts. Its clinical presentation may be misleading and mimic infectious or haematological disease, particularly in tuberculosis-endemic, resource-limited settings. We report a 26-year-old man admitted for massive anasarca and ascites evolving over three months, with severe general condition impairment (Eastern Cooperative Oncology Group [ECOG] performance status 3) and cachexia masked by fluid overload. Digital rectal examination revealed a hard, fixed, extraluminal anterior rectal mass about 5 cm from the anal verge. Investigations showed microcytic anaemia, hypoalbuminaemia (26 g/L), a negative GeneXpert assay and normal germ-cell markers. Chest radiography demonstrated bilateral pleural effusion with a normal cardiac silhouette, and computed tomography confirmed a heterogeneous pelvic mass compressing the rectum and encasing the posterior bladder wall, without bone lysis. An ultrasound-guided percutaneous endorectal biopsy was performed after a multidisciplinary decision. The patient died 11 days after admission, before results were available, in a setting of multi-organ failure. Post-mortem histopathology showed small round cells arranged in fibrous-septa-separated lobules with extensive central necrosis, an appearance compatible with alveolar rhabdomyosarcoma; confirmatory immunohistochemistry (myogenin, MyoD1, desmin) was unavailable. Adult ARMS may present as anasarca, a rare but documented scenario. Early percutaneous biopsy remains essential to avoid diagnostic delay and loss of therapeutic opportunity, even where advanced diagnostic facilities are lacking.

Keywords: Case Report, Alveolar Rhabdomyosarcoma, Para-Rectal Mass, Anasarca, Delayed Diagnosis, Resource-Limited Settings, Niger

INTRODUCTION

Rhabdomyosarcoma is the most common soft-tissue tumour in children and adolescents, but it is rare and carries a poorer prognosis in adults, in whom it accounts for less than 1% of malignancies and an annual incidence of about 0.4 cases per million population (Amer *et al.*, 2019). Among its histological subtypes, the alveolar form (ARMS) is distinguished by marked biological aggressiveness: 5-year overall survival does not exceed 20–30% in the most recent adult cohorts (Amer *et al.*, 2019; Mäkinen *et al.*, 2021), owing to early metastatic potential and the frequent PAX3–FOXO1 and PAX7–FOXO1 chromosomal rearrangements, recognised as independent adverse prognostic factors (Shern *et al.*, 2021).

The rarity of adult ARMS and the paucity of dedicated series mean that its clinical semiology remains poorly codified, particularly for deep pelvic locations. Para-rectal forms are exceptionally reported, and their revelation by a generalised oedemato-ascitic syndrome — anasarca — is described only in isolated observations. This gap is especially concerning in resource-limited, tuberculosis-endemic regions, where

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an abdomino-pelvic mass associated with ascites is readily and wrongly attributed to peritoneal tuberculosis or a haematological malignancy, at the cost of diagnostic delay and loss of therapeutic opportunity (Adebusoye *et al.*, 2024; Susilo *et al.*, 2025).

We report an observation of para-rectal ARMS in a young adult revealed by anasarca, managed at Zinder National Hospital (Niger). The primary objective was to describe this misleading clinical presentation and the diagnostic pathway undertaken in a technically constrained environment; the secondary objectives were to analyse the clinical course and to identify the obstacles to early histological diagnosis. This observation is a reminder that, in line with international recommendations, any suspicious deep mass should undergo biopsy before any therapeutic decision, including before antituberculous trial therapy (Dangoor *et al.*, 2016).

CASE

Patient and History

A 26-year-old male farmer, resident in the Zinder region (Niger), with no relevant medical or surgical history and no known tuberculosis contact, presented with progressive abdominal distension, abdominal pain and lower-limb oedema. The condition had been evolving for three months, marked by increasing abdominal girth, oedema extending to the root of the limbs, and profound deterioration of general condition with undernutrition and dehydration.

Clinical Examination

Vital signs were: heart rate 132/min (regular tachycardia); blood pressure 125/64 mmHg; respiratory rate 12/min; peripheral oxygen saturation 94% on room air; weight 74 kg and height 1.85 m (weight increased by massive fluid retention). The abdomen was markedly distended with large-volume ascites and periumbilical collateral venous circulation. Bilateral, tender, mobile inguinal lymphadenopathy was present. Digital rectal examination revealed a hard, extraluminal anterior rectal mass fixed to the deep plane, about 5 cm from the anal verge, non-stenosing, with palpable micronodules. The lower limbs showed bilateral, soft, pitting oedema extending to the root of the limbs (Figure 1). Eastern Cooperative Oncology Group (ECOG) performance status was 3, with marked mucocutaneous pallor.



Figure 1: Clinical appearance on admission (A, B).

Differential Diagnosis

Given the combination of a pelvic mass, ascites, anasarca and impaired general condition in a young adult, the hypotheses raised at the departmental meeting were: (1) non-Hodgkin lymphoma; (2) germ-cell tumour (seminomatous or non-seminomatous); (3) extraosseous pelvic Ewing sarcoma; (4) desmoplastic small round cell tumour, which should be systematically considered before an abdomino-pelvic small round cell mass in young men (Wang *et al.*, 2022); and (5) pseudotumoral peritoneal tuberculosis.

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Investigations

Laboratory. White cells 8,100/ μ L; haemoglobin 9.3 g/dL (hypochromic microcytic anaemia); platelets 514,000/ μ L (reactive thrombocytosis). GeneXpert negative. Blood glucose 4.99 mmol/L; creatinine 46.9 μ mol/L; urea 2.42 mmol/L (preserved renal function). Albumin 26 g/L (hypoalbuminaemia). Lactate dehydrogenase (LDH), beta-human chorionic gonadotropin (β -hCG) and alpha-fetoprotein (AFP) were normal, making a germ-cell tumour unlikely.

Chest radiography (Figure 2). Bilateral pleural effusion of moderate abundance, predominantly left-sided; normal cardiothoracic index (excluding a cardiac cause of anasarca); no excavated parenchymal lesion suggestive of tuberculosis.

Abdomino-pelvic ultrasound. Heterogeneous subvesical tissue mass with large-volume ascites.

Abdomino-pelvic computed tomography (Figure 3). Heterogeneous pelvic soft-tissue mass compressing the rectum and encasing the posterior bladder wall, without visible sacral bone lysis. Pelvic magnetic resonance imaging (MRI), the reference examination for tissue characterisation of pelvic sarcomas (Dangoor *et al.*, 2016), was not available at our institution.



Figure 2: Chest radiograph — bilateral pleural effusion.

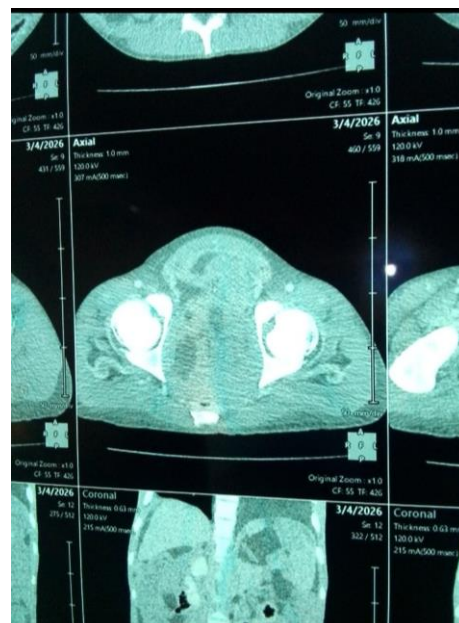


Figure 3: Axial pelvic computed tomography — pelvic mass.

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Multidisciplinary Decision and Diagnosis

At the multidisciplinary team (MDT) meeting, an ultrasound-guided percutaneous endorectal biopsy of the mass with histopathological examination was decided, together with symptomatic treatment (ascites tapping, analgesics, rehydration). The histopathology report, received the day after the patient's death, described a proliferation of small round cells arranged in lobules separated by fibrous septa, with extensive central necrosis — an appearance compatible with alveolar rhabdomyosarcoma. Immunohistochemistry (myogenin, MyoD1, desmin) — markers whose sensitivity for rhabdomyosarcoma reaches approximately 97% (Morotti *et al.*, 2006) — could not be performed owing to unavailable antibodies; the diagnosis therefore remains morphological and not formally confirmed immunohistochemically.

Outcome

The course was marked by rapid, irreversible deterioration of general, respiratory and haemodynamic status. The patient died 11 days after admission, in a setting of multi-organ failure.

DISCUSSION

This case, reported in accordance with the CARE (Consensus-based Clinical Case Reporting) guidelines (Riley *et al.*, 2017), describes an adult para-rectal alveolar rhabdomyosarcoma with a misleading presentation and analyses the obstacles to diagnosis in a resource-limited setting.

Epidemiology and Prognosis of Adult ARMS

ARMS is the most aggressive rhabdomyosarcoma subtype. In adults it constitutes an entity of more severe prognosis and lower chemosensitivity than its paediatric counterpart (Amer *et al.*, 2019; Mäkinen *et al.*, 2021). In a contemporary analysis of the Surveillance, Epidemiology and End Results (SEER) database involving 592 patients with ARMS, the independent adverse prognostic factors were distant metastasis, regional nodal involvement and age ≥ 20 years, whereas surgery and radiotherapy were associated with improved cancer-specific survival (about 43% at 5 years) (Obeidat *et al.*, 2025). Our patient combined several of these unfavourable factors (metastatic pleural disease, adult age, no feasible multimodal treatment).

PAX3/7–FOXO1 Rearrangement

Molecularly, ARMS is characterised in most cases by a PAX3–FOXO1 or PAX7–FOXO1 fusion, whose status is a major prognostic determinant associated with early metastatic potential (Shern *et al.*, 2021). In the absence of molecular biology in our setting, this status could not be determined; this limitation is frequent in the management of sarcomas in sub-Saharan Africa (Adebusoye *et al.*, 2024).

Rapidity of the Clinical Course

The striking feature of this observation is death 11 days after admission, only three months after symptom onset. In the retrospective cohort of Mäkinen *et al.* (2021), which included 50 adult patients with rhabdomyosarcoma in Denmark, median overall survival was 2.3 years and 5-year survival 30% (15% in metastatic disease), underscoring the severity of this entity even in high-resource countries.

Mechanisms of Anasarca

Presentation as anasarca, rare but documented for pelvic masses, likely results from three combined mechanisms: (1) ilio-caval venous compression by the pelvic mass; (2) lymphostasis from tumour infiltration of the inguino-pelvic lymph node chains; and (3) hypoalbuminaemia related to undernutrition and chronic inflammation, consistent with the measured albumin of 26 g/L.

Cognitive Bias and Diagnostic Delay in Endemic Areas

This case illustrates a common cognitive bias in infection-endemic areas: attributing a pelvic mass with ascites to tuberculosis or lymphoma, thereby delaying the diagnosis of sarcoma. Tuberculosis and abdomino-pelvic malignancy closely mimic one another clinically, radiologically and on tumour markers, a source of diagnostic wandering in high tuberculosis-burden regions (Susilo *et al.*, 2025). In sub-Saharan Africa, soft-tissue sarcomas, although uncommon, are often diagnosed at an advanced stage owing to limited technical facilities and specialised pathways (Adebusoye *et al.*, 2024). Desmoplastic small round cell tumour, which preferentially affects young men and presents as an abdomino-pelvic small round cell mass, is an essential differential diagnosis in this setting (Wang *et al.*, 2022). In line with recommendations,

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any suspicious deep mass should undergo percutaneous biopsy before any therapeutic decision, including before antituberculous trial therapy (Dangoor *et al.*, 2016).

Study Limitations

The main limitations of this observation are: (1) the absence of immunohistochemistry (myogenin, MyoD1, desmin), which would have allowed formal confirmation of the diagnosis and exclusion of small round cell differentials (Morotti *et al.*, 2006; Wang *et al.*, 2022); (2) the absence of pelvic MRI, the reference examination for characterising pelvic sarcomas (Dangoor *et al.*, 2016); and (3) the absence of molecular biology (PAX-FOXO1 status) (Shern *et al.*, 2021). The diagnosis therefore remains morphologically compatible but unconfirmed. These limitations constitute the educational value of the observation: a shortage of technical resources must not delay the histological work-up.

CONCLUSION

Adult alveolar rhabdomyosarcoma is a rare but formidably aggressive tumour that may mimic infectious or haematological disease in endemic areas. Presentation as anasarca, although unusual, should prompt consideration of a pelvic sarcoma alongside tuberculosis and lymphoma. Early percutaneous biopsy remains the only means of obtaining histological certainty and avoiding loss of therapeutic opportunity, particularly in settings with limited diagnostic resources. The lack of advanced technical facilities cannot justify a diagnostic delay.

ETHICAL CONSIDERATIONS

This case report describes a retrospective clinical observation. As the patient had died, oral consent for publication was obtained from the first-degree family, in accordance with international recommendations for case reports published after a patient's death. Patient anonymity was preserved: all identifying data were removed, including nominative identifiers appearing on imaging documents. A non-objection statement was obtained from the head of Department of Surgery B, Zinder National Hospital.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

AUTHOR CONTRIBUTIONS

Amadou Magagi I: conception, data collection, manuscript drafting, interpretation of imaging data, manuscript revision; Awandjinou BD: conception, data collection, manuscript drafting; Mamoudou AN: coordination of care, manuscript revision. Adamou H: clinical supervision, critical revision, final validation; All authors participated in the MDT meeting of 11 March 2026 and approved the final version.

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EXPLANATION TO THE FIGURES

Figure 1: Clinical appearance on admission. (A) Marked abdominal distension with periumbilical collateral venous circulation; (B) bilateral pitting oedema of the lower limbs. Original images.

Figure 2: Postero-anterior chest radiograph showing bilateral pleural effusion of moderate abundance, predominantly left-sided, with a normal cardiac silhouette. Original image; patient identifiers were masked.

Figure 3: Axial pelvic computed tomography showing a heterogeneous soft-tissue mass compressing the rectum and abutting the posterior bladder wall, without bone lysis. Original image. (Abbrev. CT-computed tomography)

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