Case Report

A RARE UMBILICAL MYXOID LIPOSARCOMA IN A CHILD AND CUTANEOUS TOXIC EFFECT OF SELF TRADITIONAL HERBS USE: A CASE REPORT AND LITERATURE REVIEW

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Abstract

Background: Liposarcomas are very rare mesenchymal soft tissue tumours in children. Occurrence in many different body sites were reported, mostly on extremities but abdominal site is less common to which retroperitoneal sarcomas dominate and umbilical involvement unreported. Hence a foremost case reporting of umbilical Myxoid Liposarcoma (MLS) in English literature. Adherence to CAse REport (CARE) reporting guideline recommendation was attempted.

Case presentation: A 13-year-old boy with a recurrent `history of umbilical swelling of a year duration with no associated nausea, vomiting, diarrhea, jaundice or alternating bowel habit. Patient had traditional herbs application, resulting in crust, ulcerations and discharge of the umbilicus skin. Umbilical swelling of 8 x12 cm was found on examination, had wide excision and histology revealed pleomorphisim and hyperchromasia with capillary size vascular channel exhibiting 'chicken – wire' diagnosed as umbilical myxoid liposarcoma. Liver appeared normal on ultrasound. Surgery was uneventful and no recurrence reported.

Conclusion: Umbilical Myxoid Liposarcoma (MLS) is very rare and Paediatrics have excellent prognosis. Self-use of traditional herb medication can be detrimental to tissues without acheiving tumour cure. The scientific literature has paucity of umbilical MLS report, hence the essence of this paper.

Keywords: Umbilical, Rare, Myxoid liposarcoma, Child, Recurrence, Herbs, Traditional, Case report.

Cite this article: Bello US. A rare umbilical myxoid liposarcoma in a child and cutaneous toxic effect of self-traditional herbs use: a case report and literature review. Niger Delta J Med Med Res. 2026;5(2):48–52.

INTRODUCTION

Primary malignant umbilical tumours constitute 17% of cases and are rare, while metastatic umbilical tumours are more common, occurring in 83% of cases, although umbilical tumours are mostly seen in adults.[1] Liposarcomas (LS) are rare mesenchymal soft tissue sarcomas that account for about 13–20% of all soft tissue sarcomas, with sarcomas accounting for 1% and 15% of cancers in adults and paediatric patients respectively. Myxoid liposarcoma (MLS) accounts for 20–35% of all LS.[2] Liposarcoma occurs mainly in the 3rd to 7th decades of life,[3] although the average age at diagnosis is 50 years, with peak incidence between 30–50 years.[3] The extremities

are most commonly involved, accounting for 39–41% of cases, while the retroperitoneum accounts for 21–22%.[2] Truncal locations are less common,[3] and abdominal liposarcomas are rare, with the retroperitoneal type predominating.[5] Other sites include the head, neck, subcutaneous tissue, chest wall, axilla, pelvis, inguinal region, and mediastinum.[2,5,6] The recurrence rate ranges from 13–20%.[3]

The pattern of liposarcoma differs between paediatric and adult patients, with paediatric cases having a better prognosis, while detection of the characteristic chromosomal translocation remains the best diagnostic tool in doubtful cases.[4] Increased mitotic activity and tumour necrosis are associated with high-grade MLS and round-cell morphology, as tumours with a round-cell component behave aggressively. Local recurrence, metastasis, and survival are significantly influenced when the round-cell component exceeds 5%.[2] There is a widespread belief that medicinal herbs are harmless even when they provide no benefit, and they are often considered a cost-effective treatment option for tumours.[7] However, in this case, its use led to cutaneous irritation and necrosis without resolution of the tumour.

The English literature is silent on the umbilicus as a site for MLS, making this the first reported case, with the CAse REport (CARE) reporting guideline followed.[8]

CASE PRESENTATION

A 13-year-old boy presented with a recurrent history of umbilical swelling of 1 year duration, with no associated nausea, vomiting, diarrhoea, jaundice, or altered bowel habits. He experienced pain and itching, which led to the application of traditional herbs, resulting in discharge and ulceration (Figs. 1a & b). The patient had experienced a similar swelling four years earlier and again one year ago, for which surgeries were performed in a district hospital. There was no history of other body swellings or similar family history, although a prior stick injury was reported. There was no history of exposure to chemicals aside from the topical application of traditional herbs.

Examination revealed a young boy who was mildly pale, dehydrated, and tachypnoeic, with a respiratory rate of 21 cycles/min, pulse rate of 95 beats/min, blood pressure of 116/77 mmHg, temperature of 37.1°C, and SPO_2 of 97%. Status localis revealed a predominantly transverse healed scar over the umbilical region of the abdomen, with a protruding umbilical mass measuring 8 × 12 cm. The mass appeared septic, mildly tender, and had areas of necrotic tissue with crusts of traditional herbs present (Figs. 1a & b). The chest had good bilateral air entry, the abdomen showed no hepatomegaly, and other systemic examinations were essentially normal.

Figure 1: Umbilical swelling (a) septic with herbal crust (b) after antibiotics and daily dressing

Chest X-ray was normal. Plain abdominal X-ray revealed a huge radio-opaque shadow over the lower abdomen (Fig 2). Packed Cell Volume was 33%, White Blood Cell - 6.1 x 10^9/L; N-50%/E-2%/L-44%/M-4%. Abdomino-pelvic ultrasound scan: normal findings with normal liver size and echo pattern and no intrahepatic mass seen. Platelets- 270 x 10^9/L; ESR -68 mm/hr. Wound culture revealed *Staphylococcus aureus* and had erythromycin with daily dressing.

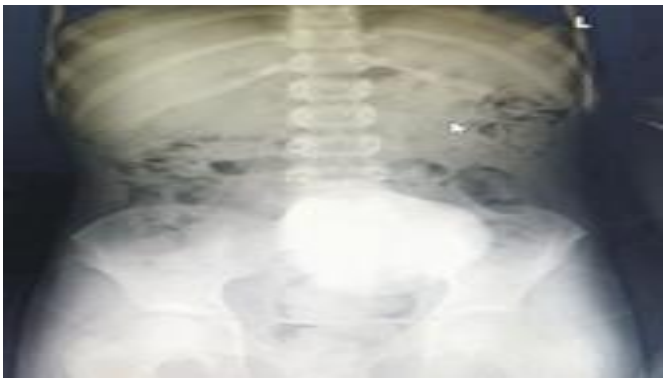

Figure 2: Plain abdominal X-ray showing radio-opaque oblong shadow of Umbilical MLS

Operative findings: General anaesthesia was administered with routine drape done and had a wide margin excision done over the peri umbilical region in excess of 12 x 8 cm mass showing a well granulated surface with a jelly-slimy feel (Figs 3 a & b). Histology revealed pleomorphisim and hyperchromasia with capillary size vascular channel exhibiting 'chicken – wire' in line with a diagnosis of umbilical myxoid liposarcoma. Post-operative recovery was uneventful. A cycle of Cyclophosphamide (400 mg), 5- fluorouracil (400 mg) and Doxurubucin (40 mg) was all that was afforded and administered on account of patient - related factor as father's financial constraint made for his preference. Surgical expense was assisted. No recurrence at 9 months visit and 18 months from contact.

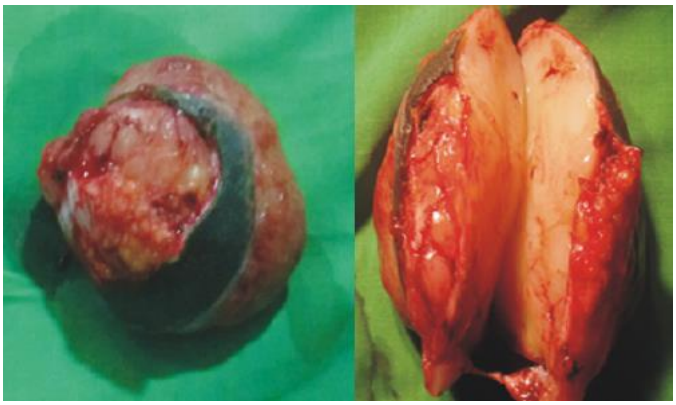

Figure 3: Excised tumour (a) with skin margin (b) Cut surface

DISCUSSION

Liposarcomas are mainly asymptomatic at onset until their size or location causes symptoms due to compression or direct invasion of adjacent tissues.[5] The present case initially manifested as an insidious, asymptomatic swelling, but itching and pain developed as the lesion enlarged over time. This is comparable to the asymptomatic case of an 11-year-old girl who presented early after 2 months, as reported by Matsunobu et al.[4] The histology in this case confirms the rarity of MLS occurring in the umbilicus. Limitations of this report include the lack of cytogenetic analysis, inadequate funding, and the absence of previously reported umbilical MLS cases in the medical literature for comparison, except for cases involving other abdominal locations. Karadayi[5] reported eight cases of abdominal liposarcoma in a case series, none of which involved the umbilicus.

The World Health Organization (WHO) classifies liposarcoma into five types, namely pleomorphic, round cell, myxoid, dedifferentiated, and well-differentiated types, with subtypes including sclerosing, adipocytic, and inflammatory variants. Studies have shown that well-differentiated and dedifferentiated liposarcomas are the commonest types, in descending order.[4,5,6] Genetic testing aids the diagnostic workup of liposarcoma subtypes, as a study involving 7,000 patients suggested potentially actionable mutations in about one-quarter of cases.[5]

Cytogenetic analysis of liposarcoma in a case studied by Miller et al. [5] showed a final karyotype of 46,XY,t(12;16)(q13;p11), which helps differentiate it from lipoblastoma, which has a karyotype of 46,XY,der(8)?t(8q;?),+mar. Karyotyping plays a significant role in differentiating these tumours, since myxoid liposarcoma can be microscopically indistinguishable from lipoblastoma.[5] In 20–35% of all LS cases, the myxoid type predominates, and more than 95% of MLS cases are characterised by t(12;16)(q13;p11) translocations, resulting in FUS-DDIT3 gene fusion, while about 5% of MLS tumours harbour a t(12;22)(q13;p12) translocation, resulting in DDIT3-EWSR1 gene fusion on 22q12.[2,9,10] Liposarcoma cells are positive for DDIT3 immunohistochemistry.[4] Özşen et al.[6] reported two female patients with liposarcoma measuring 10 cm and 4 cm, affecting the right thigh and left popliteal fossa in a 16-year-old and 12-year-old respectively. The 12-year-old developed recurrence two years later, and to prevent local recurrence, total excision with radiotherapy was recommended, while high-grade and poorly differentiated tumours carry a higher risk of distant metastases.[6] Cytogenetic analysis was unavailable in the present case.

Clinical guidelines recommend preoperative radiotherapy (50–50.4 Gy delivered in 1.8–2 Gy fractions) for soft tissue sarcomas of the extremities and trunk.[5] Of the eight abdominal cases reported by Karadayi,[5] none received radiotherapy, and the recurrence rate was 25%. Radiotherapy is commonly used for MLS tumours larger

than 5 cm, while chemotherapy is added for tumours larger than 10 cm, although it may also be administered for selected tumours measuring 5–10 cm.[9] Chemotherapy plays a role in high-grade tumours, and MLS is inherently responsive to chemotherapy, while chemotherapy remains the mainstay of treatment for metastatic disease.[9] Previously untreated patients are commonly treated with a combination of ifosamide and doxorubicin, with reported response rates ranging from 10–66%.[9] Adjuvant chemotherapy should be considered optional in high-grade tumours and in large localised tumours. According to the National Comprehensive Cancer Network (NCCN) guidelines, the efficacy of adjuvant chemotherapy in localised tumours is limited, and many studies have shown controversial outcomes.[10] Masunaga et al.,[10], in a study of 456 patients who underwent surgery alone compared with surgery plus adjuvant chemotherapy, found no statistically significant difference in local recurrence, metastasis, or disease-specific survival for localised MLS, including large soft tissue tumours greater than 10 cm and high-grade tumours. Similar findings were reported in the European Organisation for Research and Treatment of Cancer (EORTC) 62931 multicentre randomised controlled trial involving grade II or III resectable sarcomas of any site. However, when Pasquali et al. re-analysed the same trial in 2019 using a Sarculator nomogram, they found improved overall survival and prolonged disease-free survival after adjuvant chemotherapy in patients with extremity and trunk sarcomas who had a low predicted 10-year overall survival.[10]

Exposure to toxic plants, chemicals, or other environmental factors can lead to skin disorders such as rashes and irritation, termed dermatotoxic conditions.[7] This was observed in the present case following the use of traditional herbs, which resulted in cutaneous sepsis and necrosis. The history of a prior stick injury in this case may explain certain patterns of lipomatous tumours, as it has been suggested that blunt trauma can rupture fibrous septa connecting the skin to the deep fascia, thereby allowing adipose tissue proliferation, while others have postulated cytokines and growth factors as possible triggers.[11]

In the patient's own words at follow-up: "I feel well and the swelling is no more." The present case showed no recurrence at the 9-month follow-up visit, and the patient remained well 18 months after the last contact.

CONCLUSION

Cytogenetic facilities and analysis can be valuable adjuncts despite histological diagnosis after surgery, as they provide more detailed differentiation of tumours. Nevertheless, MLS has an excellent prognosis in children despite its rarity. The need to educate patients and communities about the toxic effects of traditional herbs is underscored. Financial constraints can further strain healthcare delivery when patients exhaust their resources on cheaper, unproven traditional medications that may lead to additional complications. This case highlights the extremely rare occurrence of MLS in the umbilicus as a first reported case and illustrates the challenges faced in resource-poor settings.

Funding: None declared

Conflict of Interest: None declared.

Informed Consent: Verbal and written informed consent was obtained from the father, who could not be reached further for an omission.

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