

# Misdiagnosed High-Grade Undifferentiated Sarcoma Mimicking Infantile Myofibroma: A Case Report

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## Abstract

Accurate prenatal diagnosis of fetal masses is challenging due to their varied presentation and the limitations of current imaging modalities, which are typically restricted to obstetric US and non-contrast fetal MRI. We present a case involving a right forearm mass first identified on prenatal US and MRI, which initially appeared benign and was diagnosed as infantile myofibroma following excisional biopsy. However, due to rapid growth and persistent symptoms, the patient ultimately underwent transhumeral amputation, with final pathology revealing a high-grade undifferentiated sarcoma. This case highlights the diagnostic complexity of fetal soft tissue tumors and underscores the importance of maintaining a broad differential, including malignancy, even when imaging and initial pathology suggest a benign process.

**Keywords:** prenatal imaging, fetal tumor, infantile myofibroma, undifferentiated sarcoma, fetal MRI, diagnostic error, pediatric oncology, prenatal diagnosis, biopsy misdiagnosis, soft tissue tumor

## Case Summary

A pregnant adult patient was referred at 20 weeks' gestation following abnormal anatomy US that revealed an enlarged right arm with clenched fingers. Fetal MRI and US confirmed right forearm enlargement, thickening of the radius and ulna, and dystrophic soft tissue calcifications. The remainder of the exam was unremarkable. At birth via cesarean section, a firm, nodular mass was noted on the right forearm. MRI 4 days

after delivery revealed bone remodeling and a hyperintense, heterogeneous soft tissue mass with calcifications and patchy enhancement. An excisional biopsy performed at 6 weeks yielded a diagnosis of infantile myofibroma, and the patient was observed over the next few months. By 4 months, the mass had enlarged and was causing pain and loss of function, necessitating transhumeral amputation. Final pathology demonstrated high-grade undifferentiated sarcoma. At 5 months, the patient developed a recurrence of the mass

requiring shoulder disarticulation and is currently undergoing chemotherapy.

## Imaging Findings

Fetal MRI (Figure 1) and US (Figure 2) at 22 weeks' gestation showed enlargement of the right forearm with thickened radius and ulna. The mass appeared hyperintense on T2-weighted sequences. Postnatal MRI on day 4 (Figure 3) demonstrated deformity of the forearm bones and a hyperintense, heterogeneously enhancing mass with calcifications on T2-weighted sequences. At 5 months, MRI (Figure 4) revealed a lobulated, enhancing soft tissue mass surrounding the midhumerus without obvious proximal bony involvement.

## Diagnosis

High-grade undifferentiated sarcoma.  
Differential diagnoses: venous malformation with phleboliths,

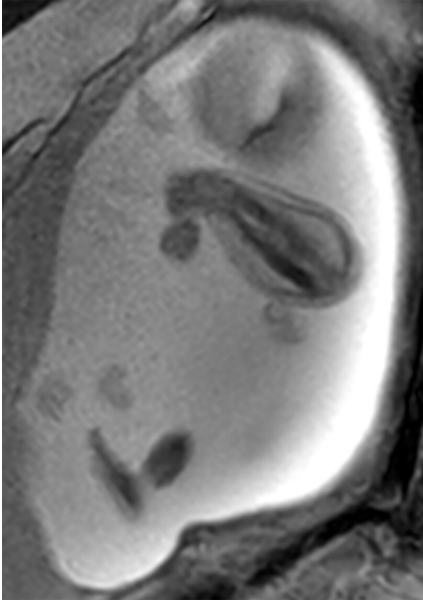
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**Informed Consent:** Patient confidentiality was maintained according to institutional policy. Written informed consent was not obtained but is encouraged in future similar cases.

**Figure 1.** Sagittal T2-weighted fetal MRI demonstrates enlargement of the right forearm soft tissues with thickening of the radius and ulna.



**Figure 2.** Coronal US confirms soft tissue enlargement of the right forearm and bony enlargement without associated fluid collection.

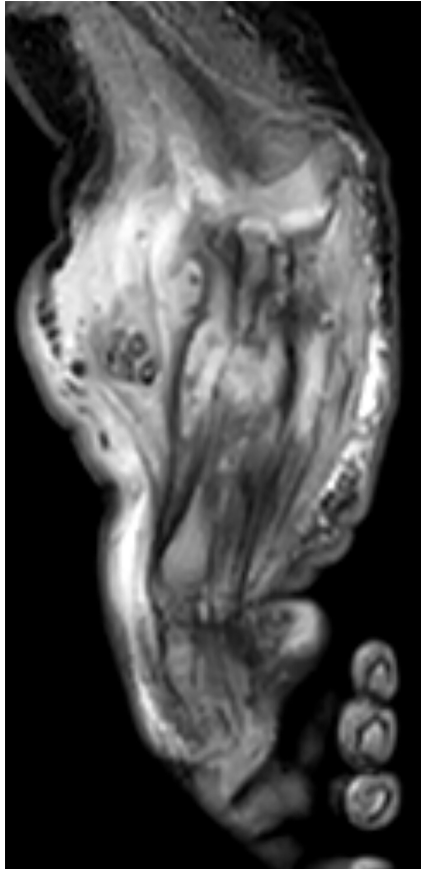


macrodyostrophia lipomatosa, constriction ring from amniotic band sequence, kaposiform hemangioendothelioma, myofibroma, fibrosarcoma, rhabdomyosarcoma, and osteogenesis imperfecta type V.

## Discussion

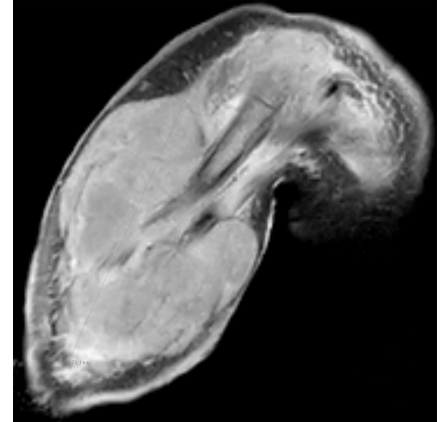
Diagnosis of prenatal masses presents significant challenges due to variability in presentation, limited imaging modalities (US and non-contrast MRI), and differences between fetal and pediatric tumors.<sup>1</sup> Prenatal imaging can guide postnatal management but cannot provide

**Figure 3.** Sagittal T2-weighted MRI demonstrates further enlargement of the forearm, irregular contouring of the radius and ulna, and dystrophic soft tissue calcifications 4 days after birth.



a definitive diagnosis, as fetal tumors differ from pediatric tumors in their biology, growth kinetics, and prognosis.<sup>1,2</sup> In this case, prenatal US first identified the abnormality, and fetal MRI was instrumental in further characterizing the lesion, narrowing the differential to entities such as infantile myofibroma, fibrosarcoma, and vascular malformations. Prenatal imaging also guided delivery planning, prompting transfer to a specialized hospital for postnatal MRI and further evaluation of the lesion. Sarcomas, rare in the prenatal period, are malignant connective tissue tumors accounting for about 5% of childhood tumors.<sup>3</sup> Undifferentiated sarcoma—a poorly differentiated subtype—is a diagnosis of exclusion and

**Figure 4.** Coronal T2-weighted MRI demonstrates a lobulated, circumferential, enhancing soft tissue mass surrounding the mid humerus without obvious bony involvement of the proximal humerus at 5 months. Additionally, a fluid collection of the distal arm near the amputation stump is present.



commonly occurs in individuals over 50 and in the lower extremities.<sup>4</sup> Prenatal and initial postnatal findings suggested a benign mass due to a lack of invasive features, the presence of dystrophic calcifications, and slow growth.<sup>5,6</sup> Initial biopsy findings were consistent with infantile myofibroma, possibly due to overlapping histopathological features with sarcomas such as hypercellularity and infiltrative growth.<sup>7,8</sup> Sampling error may also have contributed to the initial misdiagnosis due to the well-documented phenomenon of intratumoral heterogeneity and the dynamic nature of pediatric tissues, which are characterized by ongoing growth and high cellular turnover.<sup>9,10</sup> Postnatal progression, including rapid growth and pain, suggested malignancy.

Venous malformation with phleboliths may also present with soft tissue calcifications but typically demonstrates well-delineated phleboliths without associated skeletal anomalies, which were not observed in this case.<sup>6</sup> The absence of additional lesions and the localized nature of the mass also lowered suspicion for multifocal or syndromic pathology. Ultimately, the worsening clinical course—marked

by increasing pain, functional limitation, and mass progression—necessitated transhumeral amputation.

The diagnosis was confirmed after resection and recurrence, illustrating the limitations of both imaging and single-site biopsies in heterogeneous tumors. Early consideration of malignancy could have prompted earlier definitive treatment and potentially reduced the extent of surgical morbidity.

This case highlights the importance of maintaining malignancy in the differential diagnosis for fetal soft tissue masses, even when imaging and initial pathology appear benign. Improved outcomes rely on advanced imaging techniques, such as diffusion-weighted MRI, magnetic resonance spectroscopy, and total-body MRI, alongside careful multidisciplinary evaluation and clinicopathologic correlation, which together enhance lesion characterization, detect multifocal disease, and guide biopsy to the most diagnostically relevant areas.<sup>11-13</sup>

## Conclusion

This case demonstrates the challenges in diagnosing fetal soft tissue masses and

emphasizes the potential for misdiagnosis, particularly when biopsy and imaging findings appear benign. Maintaining a broad differential and monitoring for clinical progression are crucial. Advances in imaging and multidisciplinary collaboration may improve early and accurate diagnosis.

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