



# Undifferentiated Round Cell Sarcoma in a Forequarter Amputation

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## Abstract

The case presented offers an example of an aggressive undifferentiated round cell sarcoma (URCS) tumor in a 55-year-old female who underwent a unique procedure which produced a seldom-seen specimen: forequarter amputation. The initial biopsy of the mass appeared to be an Ewing sarcoma (ES) or a primitive neuroectodermal tumor. Immunohistochemical staining and tumor profiling was essential to determining the final diagnosis of URCS. Since prior chemotherapy and radiation therapy were performed prior to amputation, the pathologists' assistant (PA) must be aware of CAP protocol for soft tissue tumors, which indicates that a gross impression of the percentage of necrotic tumor should be reported from a cross-section through the longest axis of the tumor.<sup>1</sup> The PA must also be aware of the extensive testing necessary to clinch the diagnosis, since URCS is a diagnosis of exclusion reached by eliminating many histological look-alikes. The possibility of tissue exhaustion due to extensive ancillary testing of biopsies should be considered by the PA, and measures taken to preserve the initial biopsy for further studies so that molecular confirmation of the tumor type can be obtained.

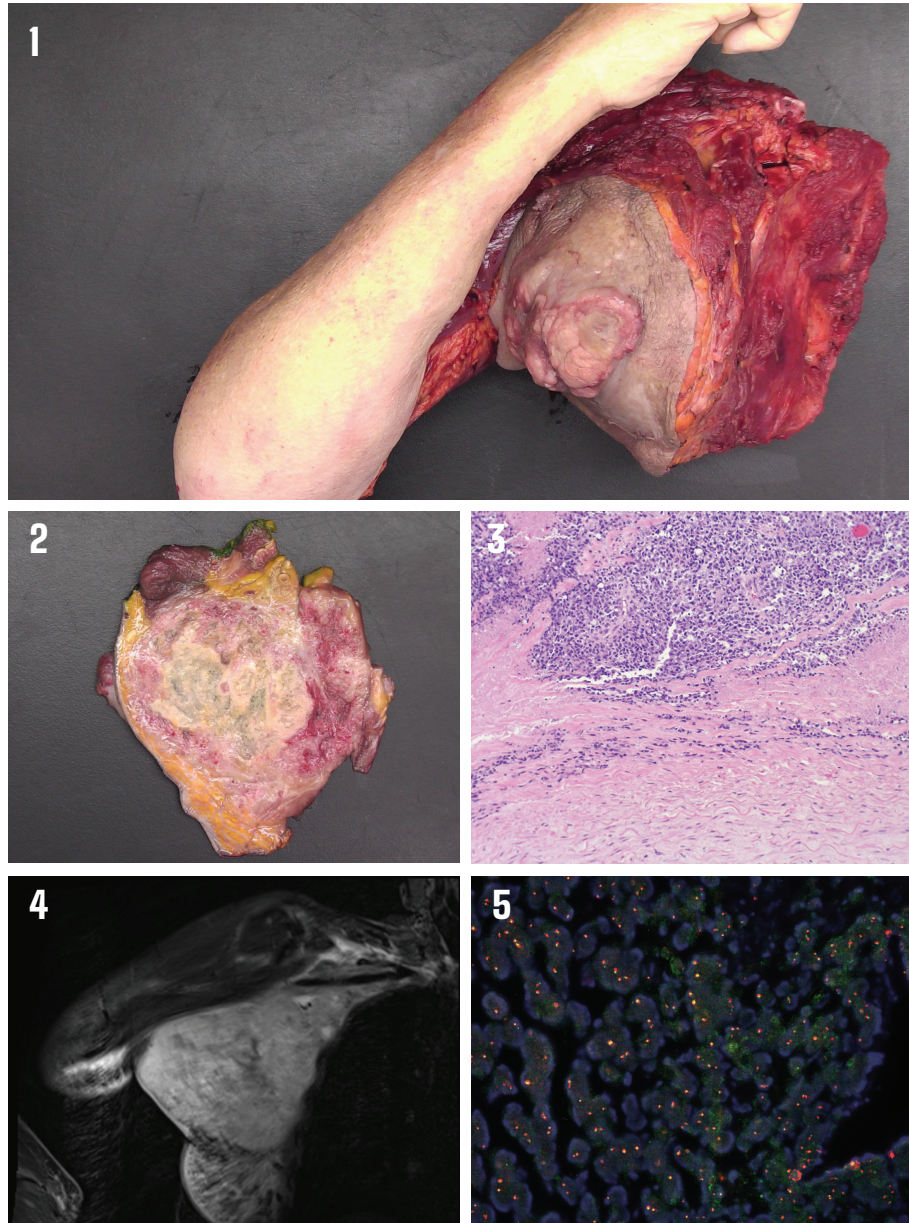
## Background

A 55-year-old female with a family history of breast cancer and a 40 pack/year history of smoking presented with a painful mass in the right axilla. Multiple tissue cores of the larger axillary mass were sent for Cancer Type ID molecular testing, which reported a likely ES/primitive neuroectodermal tumor. The tumor was deemed unresectable due to the fixed nature and neurovascular involvement which was noted by imaging (Figure 4).

Upon presentation to our institution, pathology was consulted and immunohistochemical (IHC) studies of the initial biopsy revealed URCS, instead of ES as previously diagnosed. A PET scan showed that the mass had increased in size, despite several rounds of chemotherapy and radiation therapy. Several pulmonary nodules were identified. A palliative surgery was performed after the patient was admitted to the emergency department for intense pain. At this point the mass exhibited fungating growth and yellow, watery seepage which had turned bloody that morning.

## References

- 1 Laurini JA, Cooper K, Fletcher CDM. PDF. February 2020.
- 2 Goldblum JR, Folpe AL, Weiss SW, Enzinger FM, Weiss SW. *Enzinger and Weiss's Soft Tissue Tumors*. Philadelphia, PA: Saunders/Elsevier; 2014.
- 3 Alholle A, Karanian M, Brini AT, et al. Genetic analyses of undifferentiated small round cell sarcoma identifies a novel sarcoma subtype with a recurrent CRTC1-5518 gene fusion. *The Journal of Pathology*. 2018;245(2):186-196. doi:10.1002/path.5071.
- 4 Calonje E, Damaskou V, Lazar AJ, McKee Pathology of the Skin. Philadelphia: Elsevier; 2019.



## Results

The specimen included an 8.0 x 1.5 x 1.0 cm portion of clavicle, 14.0 x 14.0 x 1.0 cm scapula, 30.0 x 6.5 cm humerus, and 25.0 x 7.0 cm forearm with attached hand (Figure 1). The 13.5 x 12.5 x 11.5 cm tumor appeared well-encapsulated, with a 6.0 x 5.5 cm focal exophytic portion involving the anterior skin. The neurovascular bundle was grossly involved. The tumor abutted but did not grossly involve the scapula. The remaining soft tissue, skin, and bone margins were grossly uninvolved. No gross lesions were identified in the vasculature of the upper arm. Sectioning of the mass revealed heterogenous cut surfaces with approximately 50% necrosis. Gross hemorrhage was absent.

Because the patient had undergone prior chemotherapy and radiation therapy, a full-thickness representative cross-section of the tumor was submitted to evaluate treatment response histologically (Figure 2). A gross estimation of tumor necrosis was also provided as per CAP guidelines.<sup>1</sup>

IHC and Fluorescence in situ hybridization (FISH) studies revealed no EWSR1 or CIC gene rearrangements (Figure 5). EWSR1 rearrangements are a defining characteristic of ES<sup>2</sup>, and CIC rearrangements are common in Ewing-like sarcomas.<sup>3,4</sup> The tumor was thus diagnosed as an undifferentiated round cell sarcoma.

## Figures:

1. Forequarter amputation specimen with exophytic tumor.
2. Cross section of tumor demonstrating necrosis.
3. 10x magnification of tumor involving axillary chest wall soft tissue. Small, round tumor cells with a high nuclear to cytoplasmic ratio are shown infiltrating layers of muscle.
4. Coronal STIR MRI of axillary mass.
5. FISH break-apart probe showing lack of an EWSR1 gene rearrangement.

## Conclusion

- Since URCS is a diagnosis of exclusion, the PA should be aware of the many ancillary tests required leading to the possibility of tissue exhaustion and take steps to preserve the maximum amount of biopsy tissue.
- In sarcomas that have received prior chemotherapy or radiation therapy, the PA should submit a full-thickness cross section and gross estimation of necrosis so that treatment effect can be evaluated as per CAP protocol guidelines.