

## Resolving a differential diagnosis of metastatic malignant melanoma or malignant fibrous histiocytoma

*Monongalia General Hospital, Morgantown, WV*

**Provisional Diagnosis:** Monophasic synovial sarcoma or malignant fibrous histiocytoma

**Reference Laboratory Diagnosis:** Metastatic malignant melanoma

**Final Confirmed Diagnosis:** Malignant fibrous histiocytoma

### Initial Staining/Scan History:

- Immuno stain 1 (Monongalia): Positive for vimentin only with negative S100, cytokeratins, EMA, CD34, and desmin
- Immuno stain 2 (reference lab): Focally S100 and HMB-45 positive
- Immuno stain 3 (Monongalia): Vimentin positivity, focal EMA positivity in areas with epithelioid cell morphology and blush S100 staining ambiguous for interpretation. HMB-45 again negative, as was wide-spectrum Cytokeratin, CK7, CK20, CD34, smActin, Desmin, Factor VIII, NSE, CD68, and CD138
- Immuno stain 4 (reference lab): S100 positivity in epithelioid areas, HMB45 negative, and Melan-A negative

### Case Summary:

A 44-year-old male patient presented at Monongalia General Hospital with a large (20cm), left posterior chest lesion. An initial biopsy evaluated by the hospital's pathology lab rendered a provisional diagnosis of soft tissue neoplasm with monophasic synovial sarcoma or malignant fibrous histiocytoma in the differential.

The case was sent for consultation to an academic reference laboratory. This group provided a diagnosis of malignant melanocytic tumor consistent with metastatic malignant melanoma.



Three months later, the patient underwent definitive therapy with wide excision and axillary lymph node dissection. Monongalia forwarded the case to the reference laboratory again, which ran additional immuno stains, and found 18 lymph nodes negative for malignancy. The reference laboratory confirmed a diagnosis of residual malignant melanocytic tumor most consistent with metastatic malignant melanoma.

But Monongalia's chief pathologist was not convinced. *"I couldn't believe it. A melanoma 20cm in size with negative nodes and no evident metastatic disease would be highly unlikely."*

The pathologist then ordered a THEROS CancerTYPE ID test. The results of this molecular classification test statistically ruled out melanoma with a 95 percent confidence level and predicted malignant fibrous histiocytoma (which had been part of Monongalia's initial differential diagnosis). *"We were in a difficult position of needing to treat this patient appropriately and quickly, yet a world expert reference laboratory had rendered a diagnosis that we didn't agree with,"* said the chief pathologist.

Monongalia shared the THEROS CancerTYPE ID test findings with the reference laboratory; the laboratory subsequently changed their diagnosis of melanoma to malignant tumor consistent with poorly differentiated sarcoma focally producing melanin. The treatments for this sarcoma and melanoma are very different – the latter being significantly more toxic.

*"THEROS CancerTYPE ID allowed this patient to be categorized correctly and receive appropriate therapy,"* said Monongalia's chief pathologist. *"In addition, we now had a totally different prognosis for this man."*



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