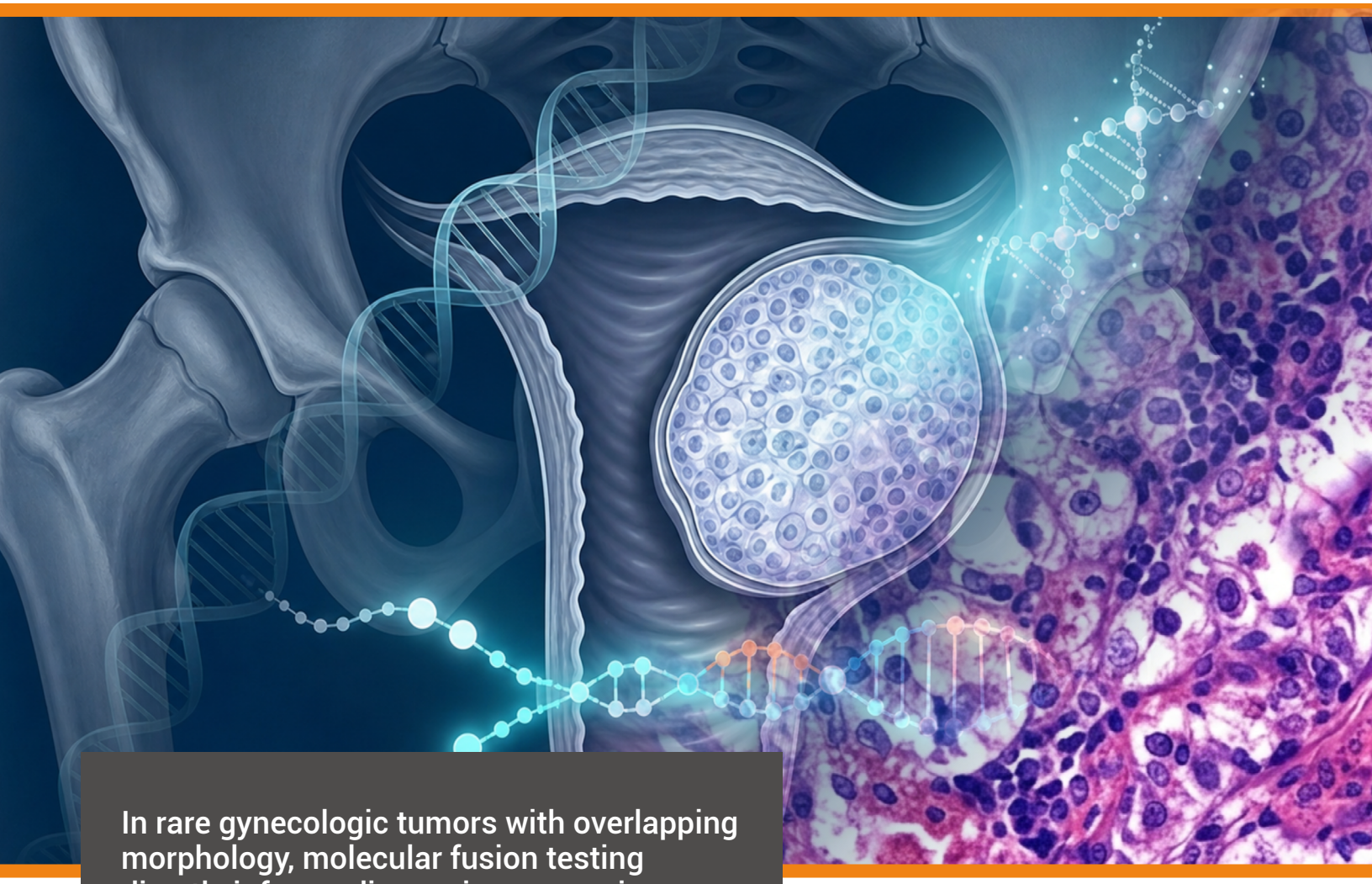
# Case Study

## TFE3-Rearranged Vaginal PEComa

### Molecular Confirmation in a Rare Gynecologic Tumor



In rare gynecologic tumors with overlapping morphology, molecular fusion testing directly informs diagnosis, prognosis, and treatment planning.

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## Diagnostic Challenge

Histology showed epithelioid cells with clear to eosinophilic cytoplasm. Immunohistochemistry demonstrated melanocytic marker expression with variable smooth muscle marker positivity, resulting in a **broad and clinically divergent differential diagnosis.**

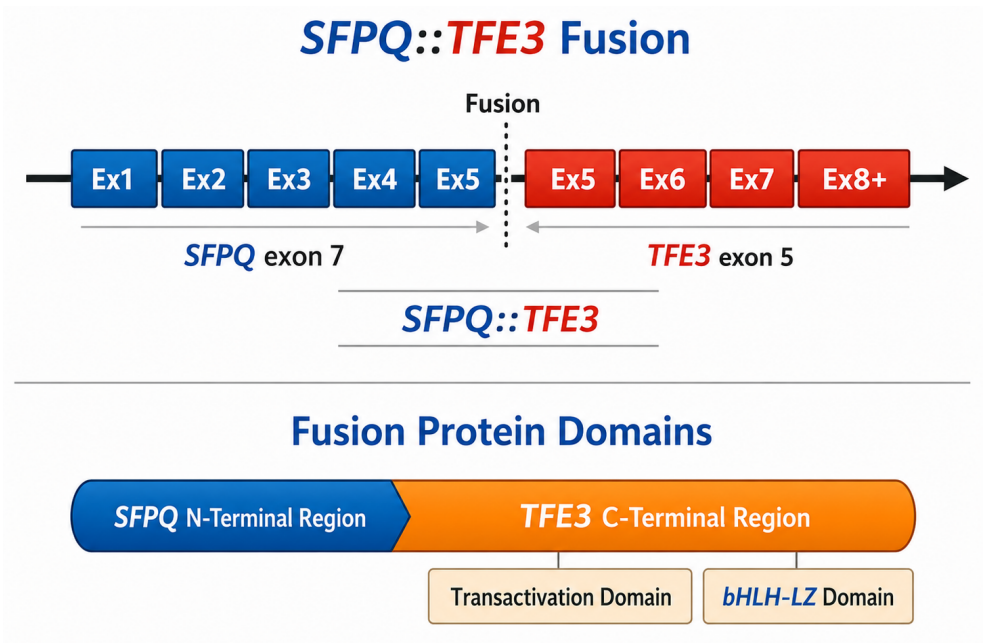


Figure 1
Dysregulated TFE3 Activity

## Clinical Scenario

A 46-year-old woman presented with a well-circumscribed vaginal mass. Imaging and morphology raised concern for a **clear cell carcinoma, melanoma, or mesenchymal neoplasm**, each with very different treatment pathways.

## Why This Matters for Oncologists

- Prevents misdiagnosis as carcinoma or melanoma, avoiding **unnecessary or inappropriate systemic therapy**
- Enables **accurate risk stratification** and tailored follow-up
- Supports consideration of **mTOR-pathway–based therapeutic strategies** when clinically indicated

## Molecular Finding That Clarified Diagnosist

Next-generation sequencing detected an **SFPQ::TFE3 gene fusion,** confirming **TFE3-translocation–associated PEComa**, a rare molecular subtype with distinct biologic behavior.

## Clinical Takeaway

In rare gynecologic tumors with overlapping morphology, **molecular fusion testing directly informs diagnosis, prognosis, and treatment planning.**

References

1. Argani P, Gross JM, Baraban E, Rooper LM, Chen S, Lin MT, Gocke C, Agaimy A, Lotan T, Suurmeijer AJH, Antonescu CR. TFE3-Rearranged PEComa/PEComa-like Neoplasms: Report of 25 New Cases Expanding the Clinicopathologic Spectrum and Highlighting its Association With Prior Exposure to Chemotherapy. Am J Surg Pathol, 2024; 48(7):777-789.

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