An External Molecular Testing Case: Unexpected Diagnosis with a Crucial FISH

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Patient History

70 year-old Female

Symptom: cough
  ◦ another hospital.

CT of the chest:
  ◦ «a mass in subcaval location, next to esophagus and atrium.»
Patient History

Video thoroscopical biopsy of the mass
- FFPE blocks

External diagnosis: “malignant tumor”

No access to the imaging of the tumor.

Previous EBUS hilar lymph node sampling
- Nonsmall Cell Carcinoma
- No access to slides
On request form

Gazi University Hospital Pathology Department, Ankara

Molecular tests for lung carcinoma

- EGFR PCR
- ALK FISH
- ROS1 FISH

- NGS, «selected cases»
Morphological Features

- Dense cellular sheets
- Fascicular growth pattern
  - Focal palisading
- Ovoid-Spindle cells
  - Bland morphology
Morphological Features

Stromal hyalinisation
Morphological Features

Minimal Myxoid Stromal Changes
Differential Diagnosis

Small cell lung carcinoma
Synovial Sarcoma
MPNST
Ewing Sarcoma
Malignant Mesothelioma
Immunohistochemistry
Bcl-2
Immunohistochemistry
CD99
Immunohistochemistry
Cytokeratin 7
## Others

<table>
<thead>
<tr>
<th>Immunohistochemistry</th>
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<tbody>
<tr>
<td>Calretinin</td>
<td>Positive (focal)</td>
</tr>
<tr>
<td>TTF-1</td>
<td>Negative</td>
</tr>
<tr>
<td>Synaptophysin</td>
<td>Negative</td>
</tr>
<tr>
<td>FLI-1</td>
<td>Preserved</td>
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<table>
<thead>
<tr>
<th>FISH</th>
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<tbody>
<tr>
<td>EWSR</td>
<td>Negative</td>
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FISH
SS18-SSX
Current Diagnosis

Mediastinal located monophasic fibrous synovial sarcoma

- with variant FISH t(x,18) pattern
Synovial Sarcoma

Young adults (mostly)

5-10% of all soft tissue sarcoma

Deep soft tissues of extremities (mostly)
  ◦ Lung, Heart, Mediastinum rare locations

Slowly growing mass

Metastasis 50% cases
  ◦ Lung most common site
SS18-SSX

t(x;18) (p11.2;q11.2)
  ◦ %95 of the SS cases

FISH break-apart
  ◦ SS18
  ◦ centromeric 3’ and telomeric 5’ positions, with green and red signal respectively
  ◦ 1F+1R+1G

1F+1R
  ◦ Similar pattern ALK, ROS1

SS18-SSX
\( t(X;18)(p11.2;q11.2) \)
SS18 Interpretation
SS18
Uncommon patterns

1F + 1R

≥ 1F + ≥ 2R

2F + 1R

≥ 2F + ≥ 2R

Conclusion

Diagnosis of external molecular cases must be reevaluated/studied

Synovial sarcoma sample may be received from Thoracic surgery department

The molecular pathologist should be aware of uncommon FISH patterns
Special Thanks

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