Case Report

Squamous Cell Carcinoma of the Lung with Metastasis to the GI Tract Associated with EGFR Exon 19 Deletion

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We describe three confirmed cases of squamous cell carcinoma (SCC) of the lung with metastasis to the gastrointestinal (GI) tract, with two having epidermal growth factor receptor (EGFR) exon 19 deletions in all available specimens. One of these patients received EGFR tyrosine kinase directed therapy for a brief period with some symptom relief. Consideration of EGFR exon 19 mutation testing in SCC of the lung, particularly for those with GI tract metastasis, may identify this potentially drug-targetable entity.

1. Introduction

Squamous cell carcinoma (SCC) of the lung metastasizing to the GI tract is an uncommon occurrence [1–4]. Epidermal growth factor receptor (EGFR) mutations have been identified in approximately 9% of metastatic SCC of the lung in one series [5]. We report three patients with metastatic SCC of the lung metastasizing to the gastrointestinal (GI) tract, two of whom had tumor with a confirmed EGFR exon 19 deletion.

2. Case Presentations

Case 1. A 39-year-old never-smoker man presented with cough, headaches, night sweats, 7-pound weight loss, and constipation for several weeks and was found to have multifocal metastases involving the lung, brain, and colon. A diagnostic colonoscopy was performed, and pathology confirmed metastatic SCC. Additional samples from the lung obtained by bronchoscopy demonstrated SCC consistent with a primary nonsmall cell lung cancer (NSCLC). He then underwent craniotomy and resection for a solitary cerebellar metastasis with pathology consistent with metastatic SCC of the lung. Subsequently, he was treated with cisplatin and gemcitabine, followed by vinorelbine and docetaxel for up to 3 cycles before developing extracranial disease progression. Tissues from the lung, brain, and colon underwent independent expert pathology review and confirmed metastatic SCC of the lung. Because of his never-smoker status, the brain metastasis sample was sent for EGFR mutation testing (exons 18–21) and found to have an EGFR exon 19 deletion. The patient was started on erlotinib and had a transient clinical response with resolution of night sweats and 4-pound weight gain. Three months later, progression by radiographic evidence of bone metastases was observed, and he passed away three weeks later. No postprogression sample was available for analysis.

Case 2. A 79-year-old man with a 20-pack-year smoking history who was diagnosed with a 6.2 cm stage IIA T2bN0M0 SCC lung cancer underwent surgical resection. He also had
Table 1: Analysis of tumor samples.

<table>
<thead>
<tr>
<th>Location of tumor sample</th>
<th>Presence of EGFR exon 19 deletion (Y/N)</th>
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<tbody>
<tr>
<td>Case 1 Lung, cerebellar, mediastinal lymph nodes from stations 2, 7, 10R, and 4R, and colon</td>
<td>Y in all 7 samples</td>
</tr>
<tr>
<td>Case 2 Lung and colon</td>
<td>Y in both samples</td>
</tr>
<tr>
<td>Case 3 Sigmoid colon</td>
<td>N</td>
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3. Discussion

Of the three confirmed SCC of the lung with metastasis to the GI tract, two had EGFR exon 19 deletions. We did not detect any discordance for the mutation findings, as both primary and metastatic tumor from the first two cases had the EGFR exon 19 deletions. This suggests that EGFR exon 19 deletions are present in the initial primary tumor clone that has metastatic potential. While Case 3 did not have a detected EGFR exon 19 deletion, it is unlikely that if we had available primary tumor tissue, this deletion would be detected. Because of limitations of tumor tissue sample for Cases 2 and 3, evaluation of EGFR mutation status was focused on exon 19 deletion.

One of two patients (Case 1) received EGFR tyrosine kinase directed therapy for a brief period with some symptom relief. One can only speculate that if the EGFR exon 19 mutation was identified earlier in his disease course, there may have been improved clinical benefit.

4. Conclusion

Consideration of EGFR exon 19 mutation testing in SCC of the lung, particularly for those with GI tract metastasis, may identify this potentially drug-targetable entity.

Conflict of Interests

Glen J. Weiss is on the speaker’s bureau for Genentech and Pfizer. He has received speaker’s fees from Medscape, Quintiles, Eli Lilly, Cephalon, Merrimack, and Roche/Ventana. The other authors have no conflict of interests to declare.

References
