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Mesenchymal Neoplasm With *ACTB-GLI1* Fusion in an Individual With Neurofibromatosis Type 1

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A young man presented with an ulcerated tongue lesion (Figure 1). On the basis of the histologic features, numerous immunostains, and genetic studies, the tumor was diagnosed as a mesenchymal neoplasm with *ACTB-GLI1* fusion.

What statement is most accurate about a mesenchymal neoplasm with *ACTB-GLI1* fusion?

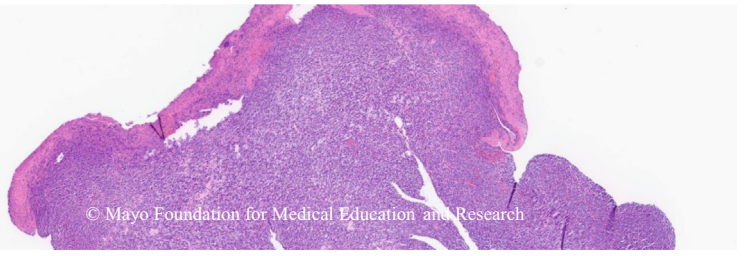

FIGURE 1. Low power shows a polypoid lesion with surface ulceration.

a. These tumors are uniformly fatal within a few months.
b. Thus far, these tumors have been reported only in the setting of neurofibromatosis type 1.
c. *ACTB-GLI1* fusions associated with these tumors are reported to result from a t(7;12) translocation.
d. These tumors most often occur in the skin of the distal lower extremities.

(see page 207 for answer)

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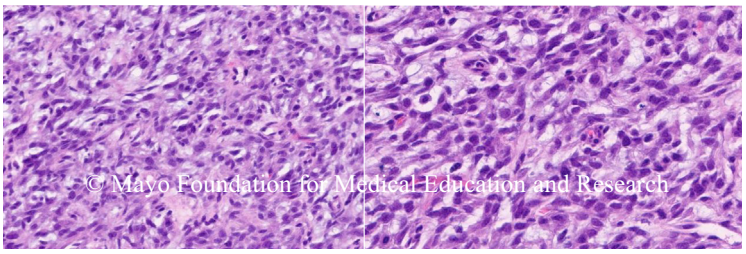

FIGURE 2. High power shows a neoplasm composed of round to ovoid to plump spindle cells arranged in cords and strands intervened by myxoid stroma with a delicate capillary-type vasculature. Scattered inflammatory cells are also present.

Answer: c. *ACTB-GLI1* fusions associated with these tumors are reported to result from a t(7;12) translocation.

Mesenchymal neoplasm with *ACTB-GLI1* fusion is recently described.[1,2] This case was particularly challenging as the individual was known to have neurofibromatosis type 1 (NF1). Thus, many entities were considered in the differential diagnosis. However, after extensive histologic evaluation, numerous immunohistochemical studies, and next-generation sequencing showing *ACTB-GLI1* fusion, the tumor was diagnosed as mesenchymal neoplasm with *ACTB-GLI1* fusion. This tumor is not, to our knowledge, specifically known to be associated with NF1. It may be coincidental that the patient has NF1, or it is possible that this could be a new association; but on the basis of a single case, one cannot draw any conclusions. *ACTB-GLI1* fusions resulting from a t(7;12) translocation have been described in a group of distinctive mesenchymal tumors falling into the histologic spectrum of pericytic tumors.[1,2] Most of the reported cases involved the tongue; however, other sites, such as stomach and bone, have been described. The tumor is composed of small monotonous-appearing epithelioid cells arranged in strands and cords separated by thin collagenous septa with a prominent capillary network (Figure 2). A broad panel of immunostains was negative for desmin, MyoD1, S100 protein, keratin AE1/AE3, and CD34. Whereas these tumors can resemble benign pericytomas morphologically and immunophenotypically, a subset of these tumors has been shown to have a propensity for distant metastases to lymph node or lung, which makes it important to recognize them.[2]

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2. Kerr DA, Pinto A, Subhawong TK, et al. Pericytoma with t(7;12) and *ACTB-GLI1* fusion: reevaluation of an unusual entity and its relationship to the spectrum of *GLI1* fusion-related neoplasms. *Am J Surg Pathol*. 2019;43(12):1682-1692.