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Case study

USP6 gene rearrangement differentiates primary paranasal sinus solid aneurysmal bone cyst from other giant cell–rich lesions: report of a rare case ☆

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Summary Aneurysmal bone cysts (ABCs) mostly occur in the metaphysis of long bones. Primary paranasal ABCs are extremely rare, and most reported cases reveal typical histopathological features including cystic space with fibrous septa and hemorrhage. Solid-variant ABCs or solid ABCs lacking cyst formation may be histologically indistinguishable from giant cell reparative granulomas, giant cell tumor of bone, and brown tumor. Here we report the case of a 24-year-old woman with a paranasal mass diagnosed as *USP6*-rearranged solid ABC, mimicking giant cell reparative granuloma, giant cell tumor of bone, and brown tumor. For paranasal sinus bone or soft tissue tumors containing numerous giant cells, molecular analysis including the *USP6* gene may serve as a useful diagnostic tool to distinguish solid ABCs from other giant cell–rich lesions.

1. Introduction

Giant cell–rich lesions mostly occur in the bone and soft tissue of extremities, and the diagnosis is mostly based on distinct histopathological appearance and anatomic location. However, giant cell–rich lesions of the head and neck regions often present diagnostic challenges because of their rarity and numerous overlapping histopathological features. In the lesions of the head and neck origin, particularly of the craniofacial bone, giant cell reparative granuloma (GCRG) is considered as the most common giant cell–rich lesion. GCRG shares a similar histopathological appearance with brown tumor, giant cell tumor (GCT) of bone (GCTB), and solid aneurysmal bone cyst (ABC); brown tumor is relatively common and should be suspected in the cases of hyperparathyroidism, whereas the other 2 are rarely encountered in the head and neck regions, particularly in the paranasal sinus.

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An effective differentiation between them may rely on a molecular analysis including the *H3F3A/B* gene mutation and *USP6* gene rearrangement. Here, we report the rare case of a 24-year-old women with recurrent sinonasal mass diagnosed as solid-variant ABC.

2. Materials and methods

A 24-year-old woman presented with progressive right nasal obstruction and swelling in the right eye for 1 month. Blurred right eye vision was also noted. She was in her usual healthy state without known systemic diseases. She also denied any familial or hereditary disorder. No fever, purulent or blood-tinged discharge, epistaxis, or facial pain was also noted. Physical examination showed right nasal septum deviation and a protruding mass in the right ethmoid sinus. Blood laboratory examination revealed no abnormalities. Serum calcium, phosphorus, and parathormone levels were normal. Head and neck computed tomography showed a sinonasal tumor involving the right ethmoid sinus, maxillary sinus, orbital bone, and skull base (Fig. 1A-C). She underwent pansinusectomy with tumor removal. At the first surgery, the pathologist reported GCRG; however, recurrent sinonasal tumor was identified 3 months later. She then underwent pansinusectomy again, and solid-variant ABC was diagnosed. The tumor could not be completely resected. She then received adjuvant radiotherapy with 6000 cGy/30fr at the outpatient clinic and tolerated the radiotherapy well. A follow-up head and neck computed tomography 9 months after the second surgery showed mucosal thickening of the right frontal, maxillary, ethmoid, and sphenoid sinuses without tumor recurrence (Fig. 1D).

3. Results

3.1. Gross examination

The specimen comprised more than 10 soft tissue and bony fragments measuring up to 2.3 × 1.3 × 0.6 cm; they were reddish and elastic.

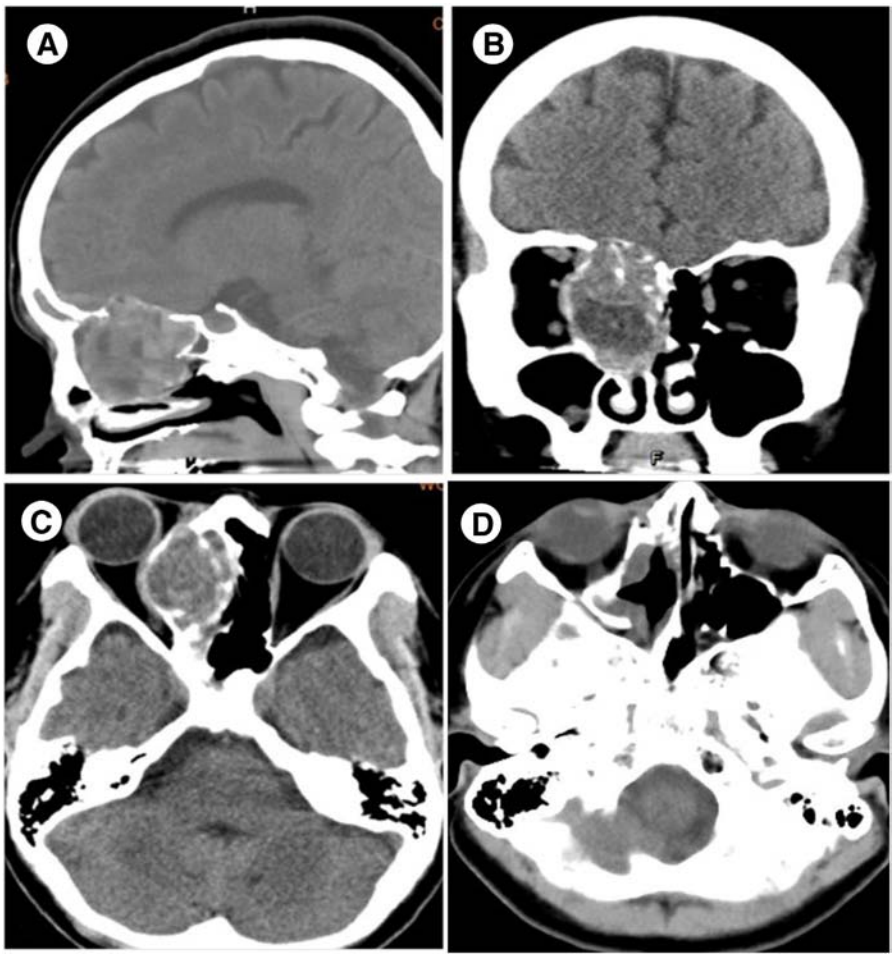

Fig. 1 Preoperative contrast-negative computed tomography demonstrated a heterogeneous solid lesion involving the right ethmoid sinus, maxillary sinus, orbit, and skull base. Mixed densities and bone remodeling were noted: sagittal (A), coronal (B), and axial (C). Postoperative contrast-negative computed tomography demonstrated mucosal thickening of the right frontal, maxillary, ethmoid, and sphenoid sinuses (D).