Rhabdomyoma

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Introduction and Classification

Rhabdomyomas are rare benign tumors arising from striated muscle. They are divided topographically into cardiac and extracardiac. Cardiac rhabdomyomas, the most common type, are believed to be hamartomas and represent 90% of cardiac tumors of infancy. Extracardiac rhabdomyomas, which are neoplastic, are further classified into three subtypes—adult, fetal, and genital type, based on their distinct clinical and histological presentations (Table 38.1). The head and neck region is the principle site of involvement in 95% of cases of extracardiac rhabdomyomas [1]. They are extremely rare tumors accounting for less than 2% of all neoplasms showing striated muscle differentiation—their malignant counterpart, rhabdomyosarcomas, are far more common, representing 98% of all skeletal muscle tumors. Rhabdomyoma and rhabdomyosarcoma are considered independent entities [2].

Etiology

No racial or geographic predilection for rhabdomyoma has been identified to date. Unlike the cardiac type, which is typically seen in patients with tuberous sclerosis, extracardiac rhabdomyomas have no association with tuberous sclerosis. Due to the rarity of rhabdomyomas, an incidence cannot be estimated.

Clinical Presentation

Fetal rhabdomyomas: These are the least common of all rhabdomyoma types. Since their first description by Dehner in 1972, case reports and small series have attempted to characterize them [3]. It is hypothesized that fetal rhabdomyomas may in fact arise from fetal rests. They are seen primarily in the head and neck region of male infants usually less than 3 years of age, and typically present as a solitary mass, with symptoms related to their specific site—they may present with hoarseness, dysphasia, dysphagia, or respiratory distress [4–6].

Two variants, *classic* and *intermediate*, have been described:

- The *classic or myxoid* subtype is the most common and usually presents in the head and neck region as a well-demarcated subcutaneous mass, particularly affecting the preauricular and postauricular regions. It has been described only in infants during the first year of life.
- The *intermediate or cellular* form occurs more often in soft tissue and mucosal sites of the head and neck region, typically the tongue, nasopharynx, larynx, orbit, and neck. It has been described in case reports in the infratemporal fossa and cricopharyngeus [7, 8].

Adult rhabdomyomas These are thought to arise from the branchial musculature of third and fourth branchial arches. They occur most often in the upper aerodigestive tract and neck musculature of male adults over the age of 40. The most common sites include the mucosa of the larynx, oropharynx, floor of the mouth, and lip. They usually present as slow-growing masses with symptoms often related to aerodigestive tract obstruction, dyspnoea, hoarseness, dysphagia, and new-onset sleep apnea. They are usually solitary but there have been reports of multifocality [9, 10].

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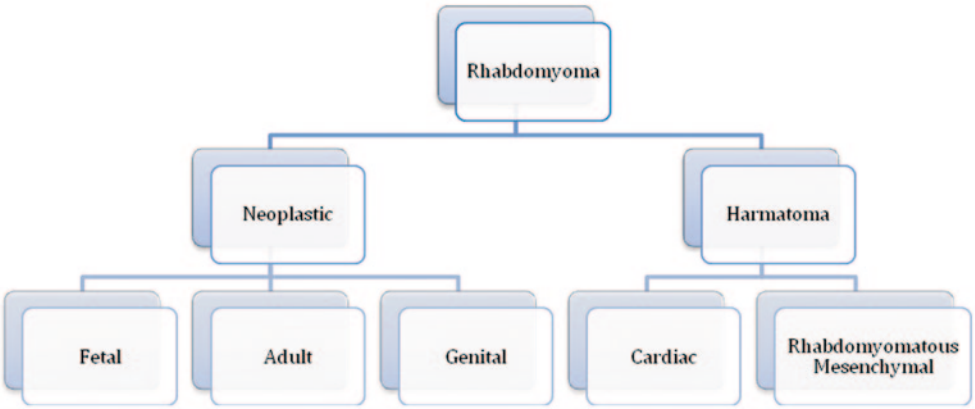
Table 38.1 Classification of rhabdomyoma



Histology

Macroscopically, rhabdomyomas are well circumscribed and encapsulated, of soft consistency, tan to grayish in color, and homogenous and mucoid when cut. Fetal and adult rhabdomyomas can be differentiated by histologic criteria and immunohistochemistry (Fig. 38.1).

Fetal rhabdomyoma

- The *classic* immature form, is identified by the presence of a mixture of bland primitive spindle cells with elongated muscle cells containing indistinct cytoplasm and muscle fibers. These spindle cells are haphazardly arranged in a fibromyxoid stroma and resemble myoblasts at 6–10 weeks of embryonic development. Myoblasts may be seen at different stages of differentiation.
- The *intermediate* form shows a greater degree and a greater number of cells with skeletal muscle differentiation, "rhabdomyoblastic maturation". Overlapping features can also be seen between the two forms. Immunohistochemically fetal rhabdomyoma typically expresses desmin, muscle specific actin and myoglobin. Primitive mesenchymal cells unpredictably express S-100 protein, glial fibrillary acidic protein, smooth muscle actin, and vimentin.

Adult rhabdomyoma This is characterized by the presence of sheets of well-differentiated large cells that resemble striated muscle cells. The cells are deeply eosinophilic polygonal cells, with small peripherally placed nuclei and occasional intracellular vacuoles. Cross-striations are a hallmark of identification and minimal or no mitotic activity is present. Muscle specific actin, desmin, and myoglobin are expressed to a higher degree than fetal rhabdomyomas, but vimentin is not expressed in the adult form.

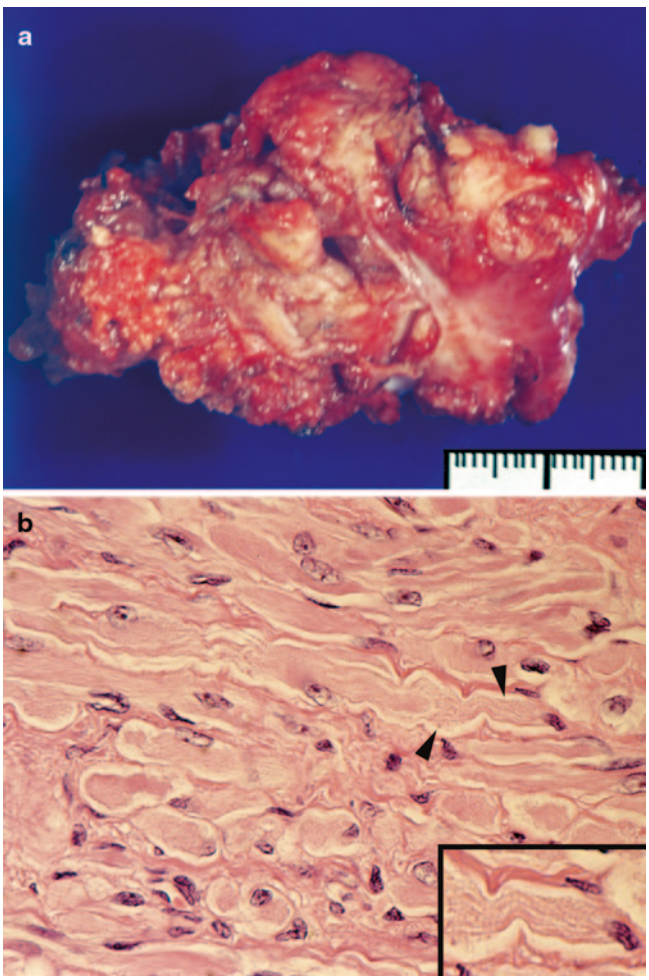


Fig. 38.1 Rhabdomyoma of neck. **a** Cut surface of lobulated soft tissue mass with a meaty appearance and a *red* and *white-gray* color. The mass had a soft rubbery consistency and absence of necrosis. **b** Tumor cells have the appearance of well-differentiated rhabdomyoblasts, some with visible striations (seen between *arrowheads* and in inset)

Table 38.2 Histological characteristics of tumors in the differential diagnosis

Tumor	Histology	Immunohistochemistry
Rhabdomyoma		
Fetal	Primitive spindle cells, myoblasts, no mitotic figures	Express muscle specific actin, desmin, and myoglobin. Weak S-100, vimentin expression
Adult	Well-differentiated skeletal muscle cells, cross striations	Strong actin, desmin and myoglobin expression. Do not express vimentin
Rhabdomyosarcoma	Cellular pleomorphism, nuclear atypia, mitotic figures, necrosis, invasion	
Granular cell tumor	Smaller cells	Strong staining for S-100
Hibernoma	Mixture of granulated and smaller vacuolated cells, lipocytes (resembles fetal fat)	May express S-100, Do not express muscle immunostains

Differential Diagnosis

The diagnosis of fetal rhabdomyoma is complicated due to the paucity of cases and the similarities between *rhabdomyosarcoma*. Distinction from the spindle cell variant of embryonal rhabdomyosarcoma can be notoriously difficult. Unlike fetal rhabdomyomas, which are well circumscribed and do not invade and destroy adjacent soft tissue, rhabdomyosarcomas, have infiltrative margins and invade normal tissues. Histologically, rhabdomyosarcomas can be differentiated by the presence of cellular atypia, increased mitotic activity, lack of differentiation, and similarity to other sarcomas. Foci of necrosis and hemorrhage are often also present in rhabdomyosarcoma.

Other tumors in the differential diagnosis include benign hamartomatous lesions, such as neuromuscular hamartomas and rhabdomyomatous mesenchymal hamartomas of the skin, teratoma, vascular malformation, neurofibroma, schwannoma, granular cell tumor, hibernoma, paraganglioma, and malignant tumors with skeletal muscle differentiation. Immunohistochemical stains including S-100, desmin, and myoglobulin may also be helpful in making exclusive diagnosis (Table 38.2). For example, granular cell tumors express S-100 protein but skeletal muscle markers usually are absent.

Evaluation

Clinical diagnosis of fetal rhabdomyoma can be challenging due to the absence of distinctive clinical characteristics.

Biopsy Diagnosis is made histologically via trucut biopsy (under radiographic guidance), open biopsy or excision biopsy of the lesion. Fine needle aspiration may also be a helpful tool in the work-up of rhabdomyomas. Cytological features suggestive of rhabdomyomas include cohesive clusters of spindle cells and rhabdomyoblasts with abundant eosinophilic granular cytoplasm, often peripherally located nuclei, cross-striations, elongated intracytoplasmic inclusions, and absence of mitotic figures. It is important to differentiate findings from a rhabdomyosarcoma, which will typically show pleomorphic nuclei and cellular atypical. There have been number of reports of solitary rhabdomyomas, which were correctly diagnosed with fine needle aspiration cytology preoperatively [11, 12].

Radiology The radiographic appearances of extracardiac rhabdomyomas have not been well defined. Although, imaging alone may not clearly differentiate rhabdomyomas from other benign neoplasms, the submucosal location of rhabdomyomas and the absence of invasion into surrounding tissues may help to distinguish them from malignant lesions. Both computed tomography (CT) and magnetic resonance imaging (MRI) are helpful in determining tumor characteristics, including the size, extent of local involvement, necrosis, nature of the tumor, including its occasional multilobar feature, and multifocality. Imaging is recommended prior to biopsy or excision. Fetal ultrasound and MRI can identify rhabdomyoma in utero at approximately 12–16 weeks gestation.

CT Imaging Rhabdomyoma appears as a well-defined, often multilobed, mass with the same density as surrounding muscle on unenhanced CT. With administration of contrast media, the tumor shows mild homogeneous enhancement with regular margins (Fig. 38.2).

MRI Imaging On T1-weighted images rhabdomyoma appears the same density as surrounding muscle. Intensity is heightened on T2-weighted images. Enhancement with gadolinium demonstrates a mild diffusely homogenous mass with regular margins.

Management

Complete excision of a rhabdomyoma with negative margins is usually curative. No cases of aggressive local tumor growth or metastasis have been documented. In cases where

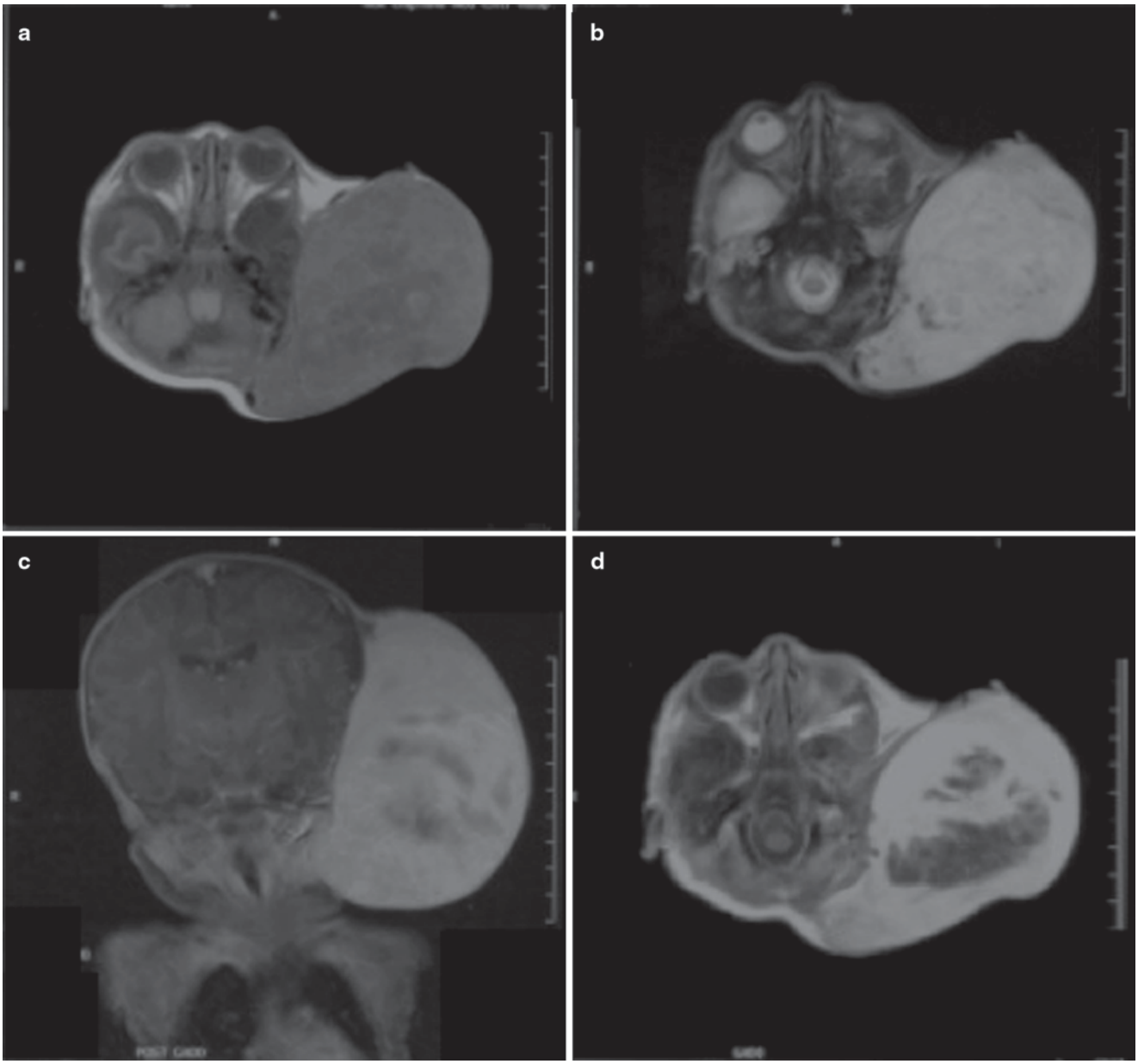


Fig. 38.2 An axial CT scan showing a left cervical fetal rhabdomyoma in a male newborn infant. T1 weighted-images (**a**), the tumor is diffusely homogenous to muscle with regular margins; T2 weighted-images (**b**), the intensity of the tumor is increased. Coronal (**c**) and axial (**d**) CT images with gadolinium (**c**) show hyperintensity of the tumor to surrounding muscle with areas of central necrosis

the tumor is extensive, craniofacial resection may be necessary with reconstruction (Figs. 38.3 and 38.4). Rare local tumor recurrences have been reported, possibly attributed to incomplete excision [13]. A complete workup to exclude rhabdomyosarcoma is essential in all cases of recurrence. Recurrence is treated with further excision. Malignant transformation of rhabdomyomas is very rare although it has been reported [2]. Chemotherapy and radiotherapy do not have a role to play.

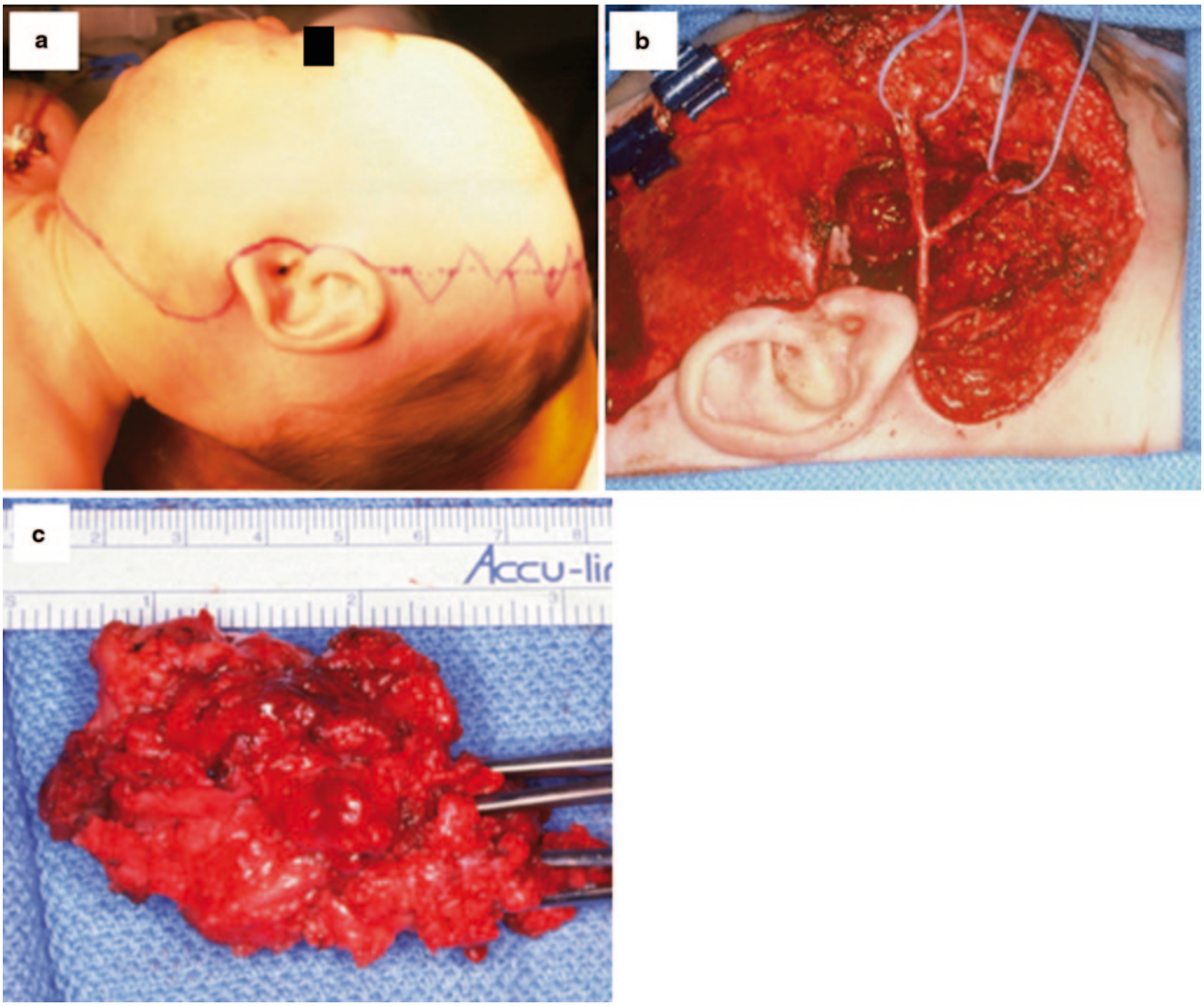


Fig. 38.3 A newborn male infant with a fetal rhabdomyoma involving the left parotid gland and extending into the left mandible and maxillary bone. A left modified Blair parotidectomy incision was used to approach the tumor (**a**). The left facial nerve was dissected and preserved (**b**). The resected tumor was deep to the facial nerve, measuring approximately 7 × 6 cm in size (**c**).

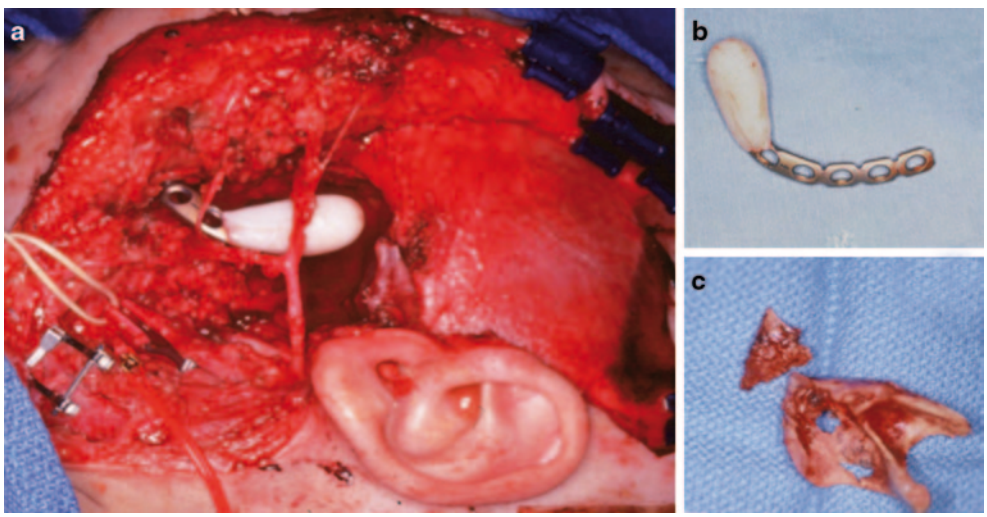


Fig. 38.4 Intra-operative images of the patient in Fig. 38.2. A titanium prosthesis was used to reconstruct the mandible. The prosthesis is seen in situ deep to main trunk and upper branches of the facial nerve (**a**). The titanium prosthesis (**b**). The resected sphenoid bone (**c**).

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