



# DIAGNOSTIC CHALLENGE

## Answers

### Case 1

**1. Diagnosis:**

Venous Malformation.

**Differential Diagnosis:**

Condition	Distinguishing Clinical Features
Hemangioma	Appears shortly after birth, shows rapid growth phase followed by involution
Arteriovenous Malformation (AVM)	High-flow lesion, pulsatile with palpable thrill
Mucocele	Usually seen on the lower lip; non-compressible and lacks color change with Valsalva
Kaposi's Sarcoma	Firm, non-compressible, often multiple purplish nodules; may be associated with immunocompromised states

**Discussion:**

Venous malformations are congenital, low-flow vascular anomalies formed by dilated venous channels due to developmental errors in embryonic venous formation. Present at birth, they may become noticeable later depending on their depth and location.

Mutations in genes such as TIE2 (TEK) and PIK3CA are associated with these malformations, leading to abnormal endothelial cell signaling and poorly organized venous networks. They can occur anywhere in the body but are most frequent in the head and neck region, particularly on the lips, tongue, and buccal mucosa.

Clinically, venous malformations present as soft, compressible, bluish swellings that enlarge with dependency, crying, or the Valsalva maneuver. They are non-pulsatile, low-flow, and cool to touch, with no thrill or bruit. On aspiration, dark venous blood may be obtained, which stops easily on applying pressure. The lesion may cause pain or swelling, especially if thrombosis occurs, and can result in functional or cosmetic problems depending on its location.

Investigations include Doppler ultrasound, which confirms a low-flow, compressible lesion, and MRI, which is the best imaging method for assessing the extent and depth. Computerized Tomography(CT) scans may show calcified phleboliths. Aspiration and occasionally venography can further support the diagnosis.

Histologically, venous malformations consist of dilated, thin-walled vascular channels lined by normal endothelium without cellular proliferation. Thrombi or phleboliths may be present due to slow blood flow.

Treatment depends on size and symptoms. Small, asymptomatic lesions may be observed, while sclerotherapy is the preferred treatment for most cases. Surgical excision is considered for localized lesions or residual masses after sclerotherapy. Laser therapy can be used for superficial mucosal lesions, and compression therapy is helpful for extremity involvement.

**Our Patient:**

Our patient was managed by sclerotherapy using sodium tetradecyl sulphate.. A total of three sclerotherapy sessions were performed at regular intervals, producing significant cosmetic and functional improvement. The patient was kept on regular follow-up to monitor healing, assess for recurrence, and ensure complete resolution of the lesion.

# Answers

## Case 2

### 1. Diagnosis

Squamous cell carcinoma

### 2. Investigation

Incisional Biopsy

### Discussion:

Oral squamous cell carcinoma is a malignant epithelial tumor arising from the stratified squamous epithelium of the oral cavity. It is the most common type of oral cancer, accounting for more than 90% of cases. The disease is characterized by locally invasive growth and potential metastasis to regional lymph nodes.

Major causative factors include tobacco use, alcohol consumption, and chronic irritation from sharp teeth or dentures. HPV types 16 and 18, poor oral hygiene, nutritional deficiencies, p53 gene mutations, and sun exposure (for lip lesions) also contribute.

OSCC can occur at various sites, most commonly the lateral border and ventral surface of the tongue, floor of the mouth, buccal mucosa, gingiva, and retromolar region.

Clinically, it often presents as a non-healing ulcer or exophytic growth that is indurated, irregular, and may bleed on touch. The lesion may be painful or tender, and in advanced stages can cause difficulty in chewing, swallowing, or speech. The

surrounding mucosa may show leukoplakic or erythroplakic changes, and regional lymph nodes are often enlarged and firm due to metastasis.

Histologically, the tumor shows invasive malignant squamous cells with keratin pearls, nuclear pleomorphism, and increased mitotic activity.

Diagnosis is confirmed by an incisional biopsy, while CT or MRI helps assess the extent of the lesion and bone involvement. Fine Needle Aspiration Cytology (FNAC) is used for evaluating lymph nodes, and chest X-ray or PET scan may be done to detect distant metastasis.

Treatment depends on the stage of the disease. Early lesions are treated by surgical excision or radiotherapy, while advanced cases often require combined surgery, radiotherapy, and chemotherapy. Neck dissection is done if nodes are involved, followed by reconstructive surgery, if required.

### Our Patient:

A segmental resection of the mandible was performed from the left lower canine to the molar region, followed by reconstruction with a free fibula flap. A left modified radical neck dissection (levels I–IV) was also carried out, and postoperative radiotherapy was planned depending on nodal status and margin involvement.

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