

White Plaques on the Cheeks

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Online Case: www.dentalcare.com/en-us/professional-education/case-challenges/case-challenge-075



The following Case Challenge is provided in conjunction with the UT Health San Antonio School of Dentistry faculty.

Amy is a 9-year-old-female who was referred by her pediatrician for evaluation of asymptomatic white lesions on her buccal mucosa.

After you have finished reviewing the available diagnostic information, make the diagnosis.

Diagnostic Information

History of Present Illness

Amy is a 9-year-old-female who was referred by her pediatrician for evaluation of asymptomatic white lesions on her buccal mucosa. Her parents have been aware of Amy's condition since early childhood. Her father and younger sibling have similar lesions. No specific diagnosis has ever been established. Amy has no complaints related to the involved areas. She is healthy, has shown normal growth and development, and has done well in school. An incisional biopsy was performed to establish a definitive diagnosis.

Medical History

- Pertinent medical history: no significant medical history; routine pediatric care; vaccinations are current
- Medications: daily children's vitamins
- Adverse drug affects: none
- Pertinent family history: maternal: obesity, social drinker; paternal: smoker, social drinker; sibling: healthy younger brother
- Social history: lives with biological mother, father, and brother

Clinical Findings

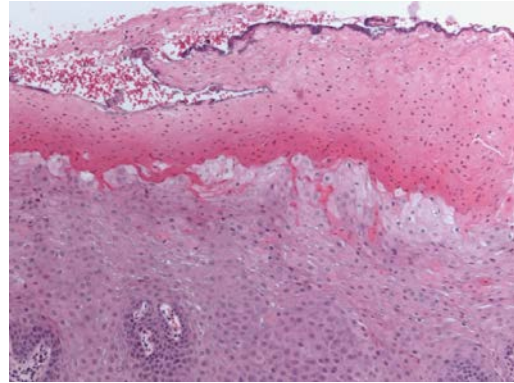
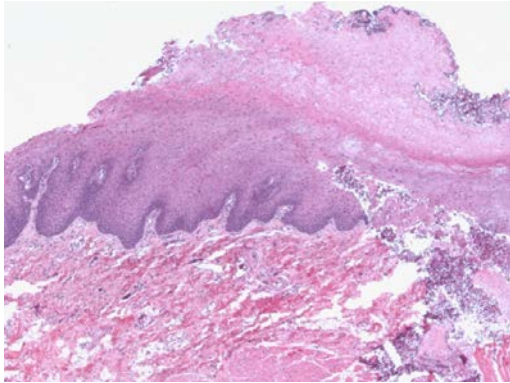
Examination reveals a healthy, alert, well-nourished 9-year-old-female. Extra-oral examination shows normal facial symmetry. No skin, hair, or nail changes are seen. The conjunctivae appear normal. Cranial nerve function is intact. No lymphadenopathy or cervical masses are noted on palpation of the neck. Intra-oral examination shows bilateral ragged thick fissured white plaques involving the buccal mucosa bilaterally. No areas of ulceration are seen. No other mucosal lesions are seen. The dentition is in good repair. Occlusal development is in the mixed dentition stage and appears appropriate for her age. Temporomandibular joint function is normal. The patient has a lip licking habit with slight exfoliative cheilitis. An incisional biopsy of the left buccal mucosa was performed under local anesthesia.



Figures 1 & 2. Clinical examination reveals roughened thick fissured areas of leukoplakia on the right and left buccal mucosa.

Histopathologic Findings

Microscopic examination of the incisional biopsy shows hyperplastic surface mucosal epithelium with elongated rete ridges, acanthosis, and thick irregular hyperparakeratosis (Figures 3 and 4). The epithelial cells in the spinous layer display cytoplasmic clearing and perinuclear eosinophilic cytoplasmic condensation (Figure 5). Surface bacterial overgrowth is present.



Figures 3 & 4. Low and medium power microscopic images showing hyperplastic squamous epithelium with elongated rete ridges, acanthosis, cytoplasmic clearing, thick ragged hyperparakeratosis, and surface bacterial colonization.

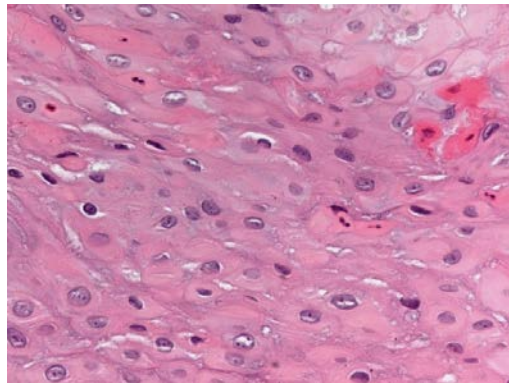


Figure 5. High-power microscopic image showing epithelial cells with pink to eosinophilic perinuclear cytoplasmic condensation and cytoplasmic clearing.

Select Diagnosis

Can you make the correct diagnosis?

9 year-old-female with bilateral white plaques on the buccal mucosa.



Select the Correct Diagnosis

- A. Leukoedema
- B. Dyskeratosis congenita
- C. White sponge nevus
- D. Pachyonychia congenita

Leukoedema

Choice A. Sorry, this is not the correct diagnosis.

Leukoedema is a very common oral mucosal condition that may be considered a variation of normal.^{1,4} It is seen in 70%-90% of black adults and 50% of black children.¹ It is much less common in whites. There is an increased incidence in smokers. The etiology of leukoedema is uncertain but it appears to be related to epithelial edema set against a background of mucosal pigmentation. Leukoedema usually presents bilaterally as a diffuse folded milky grey-white area on the buccal mucosa. Occasionally, the leukoedema will extend to the labial mucosa, floor of mouth, and palatal areas. Similar findings have also been reported on the laryngeal and genital mucosa. Characteristically, the area of leukoedema will greatly diminish or disappear with stretching of the mucosa. This finding is very helpful in recognizing this condition and will expose any other lesions underlying the edematous mucosa. Histologic findings include nonspecific acanthosis, intracellular edema and parakeratosis. Leukoedema is a benign condition and does not require treatment. The prognosis is excellent. The histopathologic findings do not support the diagnosis for this case.

Please re-evaluate the information about this case.

Dyskeratosis congenita

Choice B. Sorry, this is not the correct diagnosis.

Dyskeratosis congenita (DC) is a rare inherited X-linked recessive genetic disorder related to mutations in the DKC1 gene.^{1,5-8} A mutation of the DKC1 gene disrupts normal telomerase function. Telomerase plays an important role in determining cellular longevity. Because this is an X-linked condition, there is a strong male predilection. DC is usually first recognized during childhood when patients develop skin hyperpigmentation involving the face, neck, and chest. Dysplastic nail changes are also present. Oral lesions consist of bullae, erosions, and areas of leukoplakia involving the tongue and buccal mucosa. Rapidly progressive periodontal disease has also been reported. The leukoplakic oral lesions have a significant risk of evolving into epithelial dysplasia, and eventually invasive squamous cell carcinoma. Hematologic disorders also develop including thrombocytopenia, anemia, and pancytopenia (aplastic anemia). A biopsy of the oral leukoplakic lesions will show epithelial atrophy and hyperkeratosis. There is often progression to epithelial dysplasia and then invasive carcinoma. Patients with DC require continued close follow-up for the development of these life threatening complications. The oral manifestations of DC can be managed with supportive/palliative care. Anabolic steroids may be helpful in managing the hematologic complications and a few patients may benefit from allogeneic bone marrow transplant. The prognosis for DC is poor. The histopathologic findings do not support the diagnosis for this case.

Please re-evaluate the information about this case.

White sponge nevus

Choice C. Congratulations! You are correct.

White sponge nevus is an uncommon autosomal dominant inherited condition involving mutations in the genes for keratin 4 or keratin 13.^{1,9-11} These mutations result in abnormal keratinization of mucosal surfaces. The disease is usually diagnosed at birth or in early childhood. The most common presentation is the presence of bilateral thick white roughened fissured plaques on the buccal mucosa. Other oral mucosal surfaces including the labial mucosa, ventral tongue, soft palate, alveolar mucosa, and floor of mouth may also be involved. Occasionally, lesions may be seen on the mucosa of the nose, esophagus, larynx, and anogenital region. Most often, the lesions are asymptomatic. A biopsy will show epithelial hyperplasia, acanthosis, and thick hyperparakeratosis. The epithelial cells in the superficial spinous layer will characteristically show clearing of the cytoplasm and eosinophilic perinuclear cytoplasmic condensation representing accumulated keratin filaments. These features can be readily seen with a Papanicolau stained cytologic smear preparation. White sponge nevus is an innocuous condition and does not require treatment. The prognosis is good.

Pachyonychia congenita

Choice D. Sorry, this is not the correct diagnosis.

Pachyonychia congenita (PC) is a rare inherited autosomal dominant genetic disorder related to mutations of keratin genes.^{1,12-14} PC results in the abnormal accumulation of keratin and primarily affects the nails and skin. It is often first recognized at birth or in infancy. Patients present with thickened deformed tubular toe nails and finger nails. Palmar and plantar hyperkeratosis occurs and results in markedly thickened callus-like lesions. Hyperhidrosis of the involved areas is common. Keratin may also build-up in hair follicles producing multiple cutaneous papules. Patients may have severe disabling pain on walking due to fissuring/cracking of the soles of the feet and blister formation under the thick calluses. Nail loss may also occur. Oral involvement consists of thickened white plaques of the dorsal and lateral tongue. Other areas of oral mucosa that are subjected to trauma, including the palate, buccal mucosa, and alveolar mucosa may also develop leukoplakic lesions. Laryngeal involvement can produce hoarseness and dyspnea. Neonatal teeth may be seen in some patients. Microscopic examination will show epithelial acanthosis, hyperkeratosis, and perinuclear clearing. Treatment consists of continued removal of the thick keratin build-up. Oral retinoid medications may be helpful. Severely deformed nails may need to be surgically removed. There is no apparent malignant potential. However, the disease can cause significant pain and suffering. The histopathologic findings do not support the diagnosis for this case.

Please re-evaluate the information about this case.

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