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## **Tongue ulceration**

Dyskeratosis congenita (DC) is a rare and fatal disease, presenting with a classic triad of skin pigmentation, nail dystrophy, and oral leukoplakia. However, diagnosing DC is challenging based solely on the protean manifestations and multisystemic involvement. Therefore, it is urgent to identify an early feature facilitating the initial suspicion of DC.

Zhang et al. enrolled a cohort of six male children diagnosed with DC, all of whom exhibited erosions or ulcers on the tongue, while five of them did not display the complete classic triad. Strikingly, oral erosions or ulcers have never been included in any clinical diagnostic criteria for DC. Through a retrospective analysis, they further demonstrated that extensive and persistent tongue ulceration emerges as an early and practicable clinical marker, provoking suspicion of DC even in the absence of the classic triad.

The findings challenge prevailing diagnostic criteria and advocate for an expanded consideration of tongue ulceration as a primary and indicative manifestation of DC, thereby affording a strategic advantage for early detection and intervention of this lethal disease <sup>1)</sup>.

## **Differential diagnosis**

Tongue ulceration can arise from a wide range of conditions, from benign causes to serious systemic diseases or malignancies. Here's a structured differential diagnosis based on categories:
☐ Traumatic Causes Mechanical trauma (e.g., biting, sharp tooth, ill-fitting denture)
Thermal or chemical burns (e.g., hot food, acidic substances)
Frictional keratosis/ulceration (e.g., repeated irritation)
☐ Infectious Causes Viral: Herpes simplex virus (HSV) (primary or recurrent)
Varicella-zoster virus (shingles, esp. in older adults)
Coxsackie virus (e.g., hand-foot-mouth disease)
Bacterial: Secondary infection of traumatic ulcers
Syphilis (chancre, mucous patches)
Tuberculosis (rare)
Fungal: Candidiasis (particularly chronic hyperplastic candidiasis, often painful)
$\hfill \square$ Autoimmune and Immune-Mediated Conditions Aphthous ulcers (canker sores) – minor, major, or herpetiform
Behçet's disease
Lichen planus (erosive type)

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Lupus erythematosus	
Pemphigus vulgaris	
Mucous membrane pemphigoid	
☐ Neoplastic Causes Squamous cell card lateral tongue	cinoma – esp. in chronic, non-healing ulcers, indurated, often
Lymphoma – possible ulcerating mass	
Metastases (rare, but can occur)	
☐ Hematologic and Nutritional Iron-defic	ciency anemia
Vitamin B12 or folate deficiency	
Neutropenia / Agranulocytosis	
Leukemia - may cause ulceration from	thrombocytopenia or direct infiltration
☐ Drug-induced or latrogenic NSAIDs (a	ohthous-like ulcers)
Methotrexate	
Chemotherapy / radiotherapy (mucositi	s)
Immunosuppressants	
☐ Others Crohn's disease	
Celiac disease	
Sarcoidosis	
Stress-related ulcers	
Heavy metal toxicity (e.g., mercury, lea	d - rare)
☐ Key Workup Considerations Clinical hi	story (onset, duration, triggers, systemic symptoms)
Examination (size, location, base, borde	ers, induration)

Swab/culture or PCR if infection suspected

Biopsy for persistent or suspicious ulcers (esp. >2 weeks)

Blood tests (CBC, iron studies, B12, folate, autoimmune markers)

Zhang X, Dan H, Zhou Y, Sun W, Yang W, Zeng X. Extensive and persistent tongue ulceration is an early character of dyskeratosis congenita. Orphanet J Rare Dis. 2025 Apr 21;20(1):192. doi: 10.1186/s13023-025-03721-4. PMID: 40259308.

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