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# Chilblain Lupus: An Overview

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**Abstract:** CHLE is a form of chronic CLE that is associated with disease progression to SLE. While both CLE and SLE are subtypes of lupus, SLE has a significantly higher rate of both morbidity and mortality. This means that although CHLE is considered a rare form of CLE, it is important for clinicians to be able to recognize this disease and differentiate it with other similar diagnoses, such as regular chilblain. This can be aided by using the Mayo Clinic Criteria, which is a diagnostic criteria for CHLE.

**Keywords:** Chilblain lupus, CHLE, SLE.

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## 1. Introduction

Lupus is divided into 4 subtypes, which are SLE (Systemic lupus erythematosus), CLE (Cutaneous lupus erythematosus), neonatal lupus, and DIL (Drug-induced lupus). Out of all these subtypes, SLE is the most common subtype, with CLE following behind (National Research Center on Lupus, 2013). However, the morbidity and mortality rate of SLE are far higher than CLE. Moreover, some forms of CLE are also associated with progression to SLE. Although the most common type of CLE that usually progresses to SLE is acute CLE, some forms of chronic CLE such as generalized DLE (Discoid lupus erythematosus) and CHLE (Chilblain lupus erythematosus) are also associated with disease progression to the systemic form of lupus (Elman et al., 2019; Gkogkolou et al., 2014).

## 2. Method

This article employs a narrative review method by synthesizing relevant scientific literature on chilblain lupus erythematosus (CHLE). Sources were identified through searches in databases such as PubMed, Google Scholar, and ScienceDirect using keywords including "chilblain lupus," "CHLE," "cutaneous lupus erythematosus," and "systemic lupus erythematosus." Eligible materials included research articles, case reports, and review papers discussing the epidemiology, etiology, diagnosis,

treatment, and clinical significance of CHLE. All selected literature was analyzed qualitatively to provide a comprehensive and structured overview of current knowledge regarding this condition.

## 3. Result dan Discussion

CHLE is a form of chronic CLE that is also known as 'Hutchinson lupus' or 'Perniotic lupus'. Patients with CHLE usually present with a chief complaint of itching and/or mildly painful sensation on a reddish skin lesion (Ge Vivin Vinister et al., 2023) that worsens upon cold stimulation (Patel & Faddy Hardo, 2013). Physical examination will reveal red-violaceous plaques over the acral surfaces of the body, with the most common sites being distal parts of the extremity (Gkogkolou et al., 2014). Ge Vivin Vinister et al. (2023) also stated that nose and ears can be involved, although less commonly.

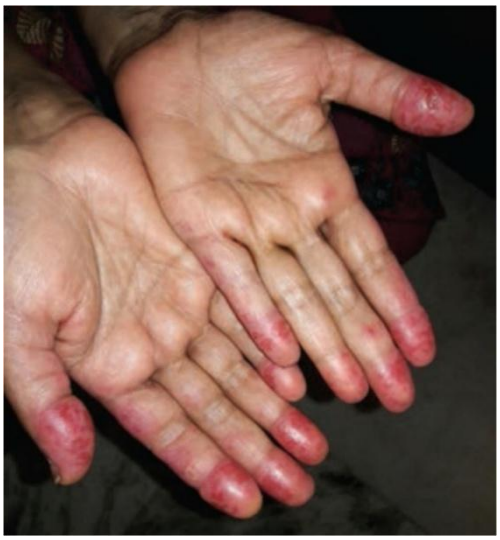

Figure 1. Clinical Picture of CHLE (Ge Vivin Vinister et al., 2023)

### 3.1. Epidemiology

CHLE is considered a rare form of CLE, with estimates of only 3 – 11% cases of all CLE cases (Tamayo et al., 2022). Furthermore, a study in 2008 noted that there were only up to 70 cases documented in the world (Hedrich et al., 2008).

### 3.2. Etiology

In general, CHLE can be divided into familial and sporadic forms. Familial CHLE was found to be caused by three mutations in the TREX1 (Prime Repair Exonuclease 1) gene. It is the only known monogenic form of CLE (Hazlianda & Diamanda, 2023) Familial CHLE is inherited in an autosomal dominant pattern (Miyagawa, 2023). On the

contrary, the pathogenesis of sporadic CHLE is still poorly understood. Some studies link immunological factors such as hypergammaglobulinemia and the presence of autoantibodies, especially rheumatic factors, to have a role in sporadic CHLE. These autoantibodies are said to increase blood viscosity and reduce blood flow, thus resulting in stasis. Vasoconstriction and microvascular injury, which are provoked by cold stimuli, are also stimulating factors (Hedrich et al., 2008).

### 3.2. Diagnostic Criteria

The most widely used diagnostic criteria for CHLE is the Mayo Clinic Criteria, which was proposed in 1994. However, there still are still no formally published sensitivity nor specificity data on this diagnostic criteria, which could be on account of the small number of CHLE cases.

Table 1. Mayo Clinic Criteria for CHLE (1994) (Ge Vivin Vinister et al., 2023)

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|---|
| **Major Criteria** |
| 1. Localized erythema and swelling involving acral sites and persistent for >24 h |
| **Minor Criteria** |
| 1. Onset and/or worsening in cooler months (between November and March)<br>2. Histopathologic findings of skin biopsy consistent with pernio (eg, dermal oedema with superficial and deep perivascular lymphocytic infiltrate) and without findings of lupus erythematosus<br>3. Response to conservative treatments (ie, warming and drying of affected areas) |
| **Diagnosis requires major criterion and at least one of the minor criteria to be fulfilled** |

The histopathology findings of CHLE could include vacuolar interface dermatitis, perivascular lymphocytic infiltration, and/or papillary oedema. Further immunofluorescence staining would also show deposition of IgM, IgA, C3, and fibrinogen on the dermoepidermal junction (Patel & Faddy Hardo, 2013).

### 3.2. Treatment

Most patients with CHLE respond well to symptomatic treatment only, with upmost importance being cold avoidance. If there are infected necrotic areas, patient should also be given anti-microbial therapy. If symptomatic treatment and cold avoidance does not provide relief, about half of CHLE patients benefit from topical steroids (Hedrich et al., 2008). Another study also stated topical steroids to be used as a first line treatment for CHLE, with other alternatives being topical calcineurin inhibitors (Patel & Faddy Hardo, 2013) and calcium channel blockers (Ge Vivin Vinister et al., 2023).

However, the use of calcium channel blockers is also found to increase the risk of developing DIL (Patel & Faddy Hardo, 2013). Only a few numbers of patient need to be managed with systemic steroids (Hedrich et al., 2008.

### 3.2. Clinical Importance

Some studies have found an association between CHLE and SLE. One study found that 18% patients with CHLE progressed to SLE (Dubey et al., 2022), while another study found the number to be as high as 20% (Hedrich et al., 2008) and 24% (Gkogkolou et al., 2014). This underlines the importance of correctly diagnosing CHLE as opposed to regular chilblains, since regular chilblains are not associated with progression to SLE. Differentiating between the two can be aided by using the Mayo Clinic Criteria for CHLE.

## 4. Conclusion

CHLE are associated with the possibility of progressing to SLE, while regular chilblains are not associated with progression to SLE. Differentiating between the two can be aided by using the Mayo Clinic Criteria for CHLE.

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