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Case Report
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Oral mass revealing Chédiak–Higashi syndrome

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Abstract. This case report describes common oral inflammatory findings leading to the identification of Chédiak–Higashi syndrome (CHS). A 15-year-old girl presented with an enlarging and painful mass on the upper lip. Two weeks after the initial visit, the mass showed further protrusion in the absence of fever. Magnetic resonance imaging revealed a well-circumscribed cystic lesion with a thick capsule, and suggested an abscess derived from the mucous cyst in the upper lip. Inflammation indices were not elevated; however neutrophils were significantly lower than the normal level. Giant cytoplasmic granules in neutrophils, eosinophils, and lymphocytes, which are pathognomonic of CHS, were noted. The patient displayed brownish-red hair with some grey hair, and partial oculocutaneous albinism. Hepatosplenomegaly was evident on ultrasonography. The final diagnosis was of an oral infection facilitated by the adolescent form of CHS (gene *CHS1/LYST* at 1q42.1-2). This report offers a reminder that lip swelling may represent the initial manifestation of the inflammatory response in a patient with loss of immunocompetence due to pathologies such as CHS, and may rarely present as the patient's main complaint.

Key words: Oral mass; abscess; Chédiak–Higashi syndrome.

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Chédiak–Higashi syndrome (CHS) is a rare, genetically autosomal-recessive disease (gene *CHS1/LYST* at 1q42.1-2), manifesting as partial oculocutaneous albinism, tendency towards haemorrhage, and increased susceptibility to infection[1]. This syndrome was first reported by Beguez Cesar in 1943. The haematological features of the disease were emphasized by Chédiak in 1952, and then by Higashi in 1954. Most CHS patients die young due to lymphoproliferative histiocytosis in the accelerated phase, unless they undergo allogeneic hematopoietic stem cell transplantation. Systemic and oral manifestations in animals with CHS have been investigated, and include early-onset periodontitis, ulcerative lesions of the gingiva, a haemorrhagic inflammatory response of the marginal gingiva, and tooth exfoliation[2]. However, there are few reports in the literature on the features of the oral manifestations in patients with CHS[3,4]. Advanced periodontitis, including early and severe periodontal destruction, has most often been mentioned.

The case of a mass on the upper lip leading to the identification of CHS is described herein. Furthermore, the importance of abscesses occurring at an uncommon site such as the upper lip is discussed.

Case report

In November 2015, a 15-year-old girl was referred to Saiseikai Matsusaka General Hospital complaining of an acute enlarging mass on the upper lip, with spontaneous pain and swelling. The patient had often experienced febrile convulsions and pneumonia following common colds since infancy, but had not been diagnosed with any congenital disorder

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and had no history of surgical procedures or trauma. A hard, firm mass, measuring 15 × 30 mm, was palpable in the lining of the right upper lip and appeared as a nodule with a normal mucosal colouration. Ultrasonography (US) of the mass showed a relatively homogeneous hypoechoic appearance in comparison with the normal parenchyma of the lip. The mass measured 32.2 × 9.5 × 5.7 mm and had a rich blood supply and distinct border (Fig. 1A). It was initially suspected that the mass represented not an abscess, but a mucous cyst or benign tumour, such as schwannoma, haemangioma, or adenoma, accompanied by slight inflammation. Poor oral hygiene was reflected by gingival swelling, redness, multiple dental caries, and tooth mobility, indicating juvenile periodontitis. Panoramic radiography revealed severe alveolar resorption around all of the patient's teeth (Fig. 2A). At this time, the patient was encouraged to gargle after tooth brushing; antibiotics were not prescribed.

Two weeks after the initial visit, the mass showed further protrusion (Fig. 1B) in the absence of any episodes of fever or chills. On magnetic resonance imaging (MRI), T2-weighted images revealed a well-circumscribed cystic lesion surrounded by halos of signal hyperintensity (Fig. 1C). Contrast intensification around three cystic lesions (diameters of 6 mm, 6 mm, and 11 mm) on T1-weighted images reflected the thick capsule (Fig. 1D), and high signal intensity on diffusion-weighted images suggested an abscess derived from the mucous cyst in the upper lip. No findings suggestive of abscess in the cervical lymph nodes were seen.

It was suspected that a bacterial infection might have caused both the periodontitis and the formation of the mass. Laboratory analyses were undertaken to evaluate the degree of inflammation and systemic conditions. Although no elevations in inflammation indices were evident, neutrophils were significantly lower than the normal level (white blood cell count $7 \times 10^3/\mu l$, platelet count $17.9 \times 10^4/\mu l$, C-reactive protein 0.1 mg/dl, neutrophils 11.6%, lymphocytes 81.1%, monocytes 6%, eosinophils 0.7%, basophils 0.6%). Blood smears exhibited giant cytoplasmic granules in eosinophils (Fig. 2B), neutrophils (Fig. 2C), and lymphocytes, which are features pathognomonic of CHS[2].

It was hypothesized that the patient might be under a state of immune compromise due to a condition such as CHS, and that an opportunistic infection might have accelerated the formation of the abscess in the lip. Inflammatory markers might thus have failed to increase despite the existence of the obvious inflammation on clinical examination. Systemic antibiotics were therefore administered (amoxicillin 750 mg/day for 7 days). The size of the mass, which correlated with the degree of spontaneous pain and redness, decreased dramatically following the initiation of antibiotics (day 21, 8 × 8 mm,

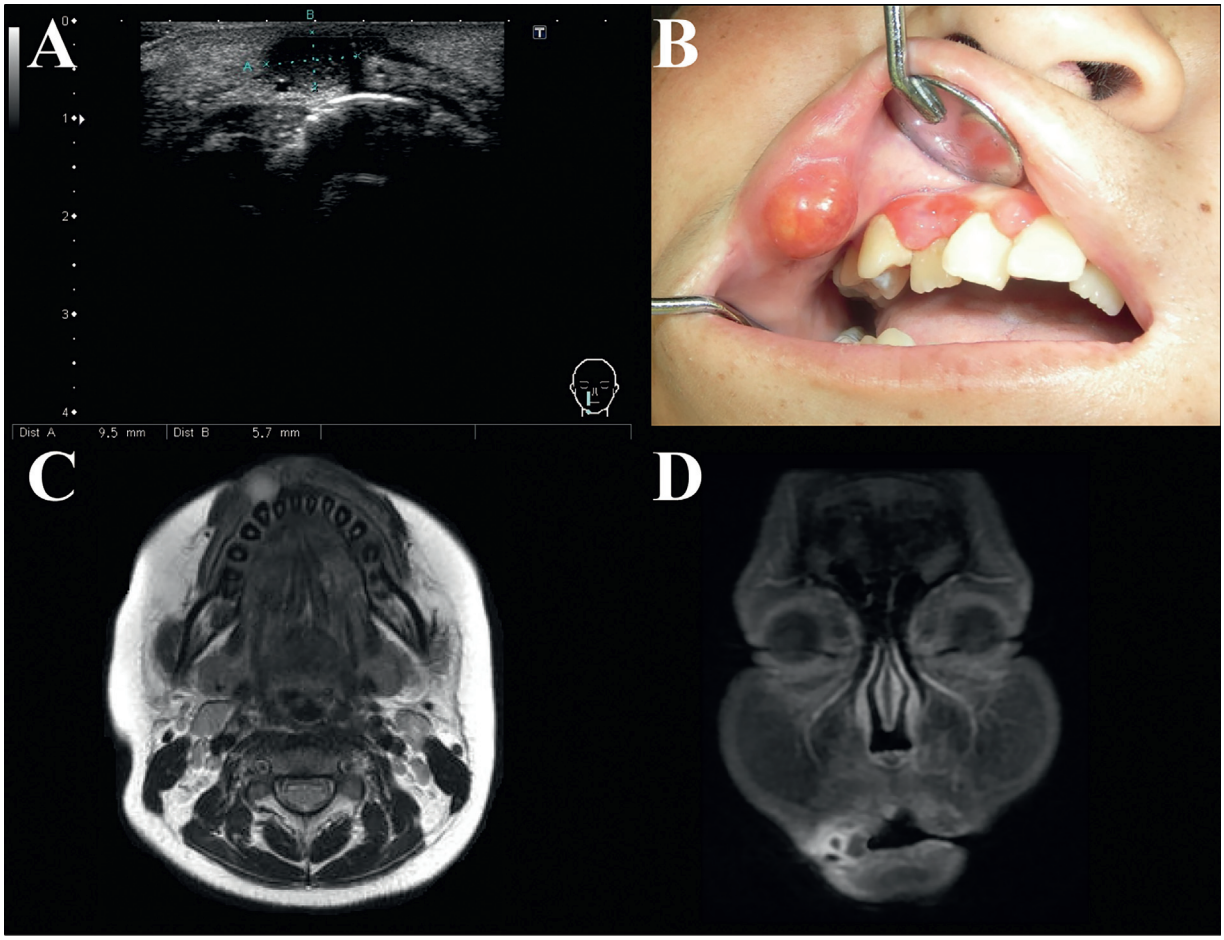

Fig. 1. Oral manifestations in a patient with Chédiak–Higashi syndrome. At the initial visit, ultrasonography of the mass on the lip showed a relatively homogeneous hypoechoic appearance with a distinct border (A). Two weeks later, gingival swelling had reduced, but the lip swelling had progressed to an abscess (B). On magnetic resonance imaging, T2-weighted images showed a well-circumscribed cystic lesion surrounded by hyperintense signal halos (C); T1-weighted images showed contrast intensification around three cystic lesions reflecting the thick capsule (D).

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