Cryosurgery (N₂O) Application to Remove Lip Lesions of Lipoid Proteinosis Syndrome: A Case Report

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Abstract

Lipoid proteinosis is a rare hereditary metabolic disorder transmitted as an autosomal recessive trait. It is characterized by the deposition of an amorphous hyaline-like material (glycoprotein) in the mucous membranes, skin and various internal organs. Cryosurgery (N₂O) was applied to remove and reshape the lip lesions in a case of this rare syndrome.

The patient was a 24-year-old female. Cryosurgery (−63°C N₂O for one minute) was performed to reshape some areas of the upper lip. The lips were softer and had better esthetics after treatment. The use of cryosurgery offers advantages over surgery in reshaping of the lip lesions in this syndrome, since suturing is not feasible in rigid mucosa of these patients.

Key words: Cryosurgery, lipoid proteinosis syndrome, N₂O.

Introduction

Lipoid proteinosis, also known as hyalinosis cutis et mucosae (OMIM 247100), was first described by Urbach and Wiethe in 1929. Since then, over 250 cases of this autosomal recessive disorder have been described. It is characterized by the deposition of an amorphous hyaline substance (glycoprotein) in the mucous membrane and skin. Hoarseness and thickening of vocal cords are the most characteristic symptoms present from infancy or early childhood. Another classic and most easily recognizable sign is the beaded eyelid papules. The other cutaneous changes may include waxy, yellow papules and nodules with generalized skin thickening. The mucosa of the pharynx, tongue, soft palate, tonsils and lips is also infiltrated. Other features may include epilepsy and calcification in the temporal lobes or hippocampi.

Since suturing is difficult because of rigid mucosa in these patients, the use of cryosurgery seems to offer advantages for removing oral nodular lesions. Therefore, cryosurgery (N₂O) was performed to remove and reshape the lip lesions in a case of this rare syndrome.

Case report

The patient was a 24-year-old female from Shahrekord, Central Iran, who sought medical care at the Department of Oral Medicine, Isfahan University of Medical Sciences in 2003. The patient was completely edentulous and showed the classic signs of lipoid proteinosis (Figure 1). During oral examination, the oral mucosa was rigid and labial mucosa had become nodular and thickened. The patient’s tongue was rigid and severely restricted in movement.

The patient had six sisters and one brother, and two of her sisters (25 and 11 years old) suffered from the same syndrome. Other members of her family showed no signs of this disease. The results of complete blood analysis and routine urinalysis were all within the normal limits. Lateral cephalometric radiography of the skull revealed calcifications in the suprasellar region. The biopsy of lip lesions was performed with a scalpel under lo-
cal anesthesthesia and examined by a pathologist to confirm the diagnosis. Suturing after biopsy was difficult because the mucosa was rigid and the sutures cut into it.

The patient suffered from some nodules on upper lip. After applying local anesthetic spray on the site, cryosurgery (N₂O, C 502T, EMD Co; Tehran, Iran) was performed (−63°C for one minute), as this treatment has been used for similar superficial lesions. It was applied on a continuous area of the upper lip. The surgery was done in two sessions; half of the lip was involved in each (Figure 2a,b). Ibuprofen 400 mg (qid) was prescribed for pain control.

The superficial layers were fallen by day 3. After the operation, the patient had pain for about a week with a visual analogue scale of up to five. Healing was good after approximately one month. The upper lip was softer than before treatment.

In one year follow up, no new nodules were formed; however, some fissures were created on her lip (Figure 2c). After the treatments, the appearance and the emotional state of this young female had improved.

Discussion

Lipoid proteinosis is a rare autosomal recessive disorder accompanied by hoarseness as well as infiltration and thickening of the skin and certain mucous membranes. Recently, lipoid proteinosis was mapped to 1q21 and pathogenetic loss-of-function mutations were identified in the extracellular matrix protein 1 (ECM1) gene. Some treatments for this disorder have already been reported. A case of lipoid proteinosis was successfully treated with dimethyl sulfoxide, however, in three other cases, it showed no beneficial effect. Retinoids have shown encouraging results in cutaneous lesions in previous studies. D-Penicillamine had a favorable effect in a patient with this syndrome. Microlaryngoscopy and dissection of the vocal cords, dermabrasion, chemical skin peeling and blepharoplasty were performed in some cases.

Lasers are used in many fields of medicine and dentistry. Carbon dioxide laser surgery of thickened vocal cords and beaded eyelid papules has proved to be helpful in some studies. Lasers were applied to remove oral lesions in one case of this syndrome.

Cryosurgery is the use of very low temperatures to destroy tissue. Immediately following cryosurgery, tissues are virtually indistinguishable from the normal, but latent damage is produced which progresses to severe destruction of the tissue. It can be used for removing many oral lesions like benign or malignant tumors. It is a safe, effective method

Figure 1. Lip and tongue involvement in the 24-year-old patient with lipoid proteinosis (a); eyelid papules (b) and skin lesions (c) in the same patient.

Figure 2. The use of cryogun for reshaping the upper lip (a); the ice ball observation at the site of cryosurgery immediately after removing cryogen (b); lip appearance in one-year follow up (c).
of treatment for many benign soft tissue lesions in the oral cavity. As suturing was difficult in this patient and mucosa was rigid, the use of cryosurgery seemed to offer some advantages in removing oral lesions in this case. The firm tissues were removed without any bleeding and deformity of the lip. It seems that surgical methods like cryosurgery and laser\textsuperscript{16,17} have some advantages over conventional surgery for removing oral lesions of lipid proteinosis syndrome as there is no bleeding and no need to suture, and it is more comfortable for the patient.

The described approach is considered an early report of N\textsubscript{2}O cryosurgery being used in removal and reshaping of lip lesions of this rare syndrome. This treatment seemed to be better than the conventional surgery.

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