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Case Report

Treatment of bilateral punctal stenosis and nasolacrimal duct obstruction in an 11-year-old case with dyskeratosis congenita

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ABSTRACT

The purpose of this study was to report the external dacryocystorhinostomy (ext-DCR) using silicone intubation in a child with dyskeratosis congenita (DC) who had presented by bilaterally acquired nasolacrimal duct obstruction (NLDO), punctal stenosis, and mucocutaneous changes. We report the 13-month results of an 11-year-old boy with DC who underwent bilateral ext-DCR with silicone stenting under general anesthesia. Ophthalmic examination revealed bilateral punctal stenosis and NLDO with normal fundus examination. He was referred to pediatrics and was diagnosed as DC, based on classic reticular skin pigmentation, nail dystrophy, and oral leukoplakia, without any other systemic involvement. Treatment consisted of bilateral ext-DCR using silicone stenting by 6 months. Management of surgery and post-operative 13-month follow up results was observed. DC is a rare heterogeneous multisystem disorder of telomere maintenance, which may present with ophthalmologic features. Although the lacrimal system abnormalities have been reported most frequently ocular findings in DC, this is the first case to present the management and long-term results of DCR using silicone intubation. This case aims to raise awareness of the various systemic and ocular manifestations and possible complications of DC and to present long-term results of ext-DC in a patient with DC.

Keywords: Dyskeratosis congenita, Punctal stenosis, Nasolacrimal duct obstruction, External dacryocystorhinostomy, Stent intubation

INTRODUCTION

Dyskeratosis congenita (DC) is a rare hereditary disease caused by abnormally short telomeres and mutations in telomere-associated genes, with a prevalence of about one in 1 million at birth.[1] It can be inherited in X-linked, autosomal dominant (OD), or autosomal recessive (OR) forms. DC occurs mostly in males and clinically is diagnosed between 5 and 15 age of years.[2,3] Classically, DC presents with a triad of mucocutaneous abnormalities-reticular skin pigmentation, nails and/or finger dystrophy, oral leukoplakia, and bone marrow failure (BMF). Multiple other manifestations have been associated with the disease, including ophthalmologic, gastrointestinal, urogenital, pulmonary, and neurological disease.[2-4]

Ocular manifestations have been reported in 40–78% of patients with DC,[4-8] and the most common ocular findings have been observed as lacrimal abnormalities, blepharitis, cicatricial entropion, and trichiasis in the literature.[1,4,5,8] Nasolacrimal duct obstruction

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(NLDO), punctal stenosis, or punctal atresia have been identified as the primary cause of lacrimal abnormalities in DC.[4,5,8] However, there is a lack of case studies regarding the management of lacrimal system abnormalities. A publication reporting endoscopic or external dacryocystorhinostomy (ext-DCR) and its results have not been found in a patient with DC. Herein, we presented an 11-year-old patient with DC has bilateral punctal stenosis and acquired NLDO and discuss the long-term results of ext-DCR and stenting applied to the patient for the first in the literature.

CASE REPORT

An 11-year-old boy presented with bilateral persistent epiphora, mucopurulent discharge, nail dystrophy, oral leukoplakia, and skin pigmentation. He had a 4-year history of mucocutaneous and nail abnormalities, and 2 year-history of bilateral constant epiphora, and discharge. On physical examination, he presented with general dry skin, reticular pigmentation on his neck [Figure 1a] and ear [Figure 1b], periorbital hyperpigmentation [Figure 1c], cracked and atrophied nails [Figure 1d], and oral leukoplakia [Figure 1e]. He complained of worsening of existing skin problems and a socially embarrassing from persistent wiping and crying appearance of epiphora which increased during downward gaze and reading. He was referred to pediatrics and DC was diagnosed based on developmental delay and classical mucocutaneous changes. His medical records did not include BMF, anemia, and other systemic findings. The patient had an 8-year-old brother with similar skin pigmentation. This sibling was undergone complete ophthalmic examination and referred to pediatrics in terms of other systemic findings of DC.

Based on the ophthalmologic evaluation, visual acuity and fundus examination were normal and anterior segment examination showed bilateral conjunctival hyperemia, punctal stenosis, and discharge increasing by pressure over the lacrimal sac. Bilateral NLDO with a hard stop was observed in lacrimal irrigation. The patient was recommended ext-DCR with silicone stenting and was referred to an otolaryngologist for further evaluation. Bilateral ext-DCR surgery with silicone stenting was performed at 2-week intervals under hypotensive general anesthesia. During the surgery, due to the larynx is relatively higher in the neck, tracheal intubation showed difficulties. To prevent bleeding, the nose was filled with cotton scales dipped in lidocaine. Local anesthetic (% 2 lidocaine with adrenaline 1:10,000) was injected over the incision site and into the nasal mucosa during surgery. The surgery was performed with 8 mm-skin incision, and bony opening was measured as 10 × 10 mm and 11 × 11 mm, right and left side, respectively. In both surgeries, equal-sized anterior and posterior mucosal flaps were created, followed by the lower and upper punctum, which were carefully dilated, and a bicanalicular silicone stent was placed. The posterior mucosal flaps and then anterior flaps were sutured together with 6/0 Vicryl, followed by skin incision which was sutured with 6/0 Prolene. There was no observed any pre-operative

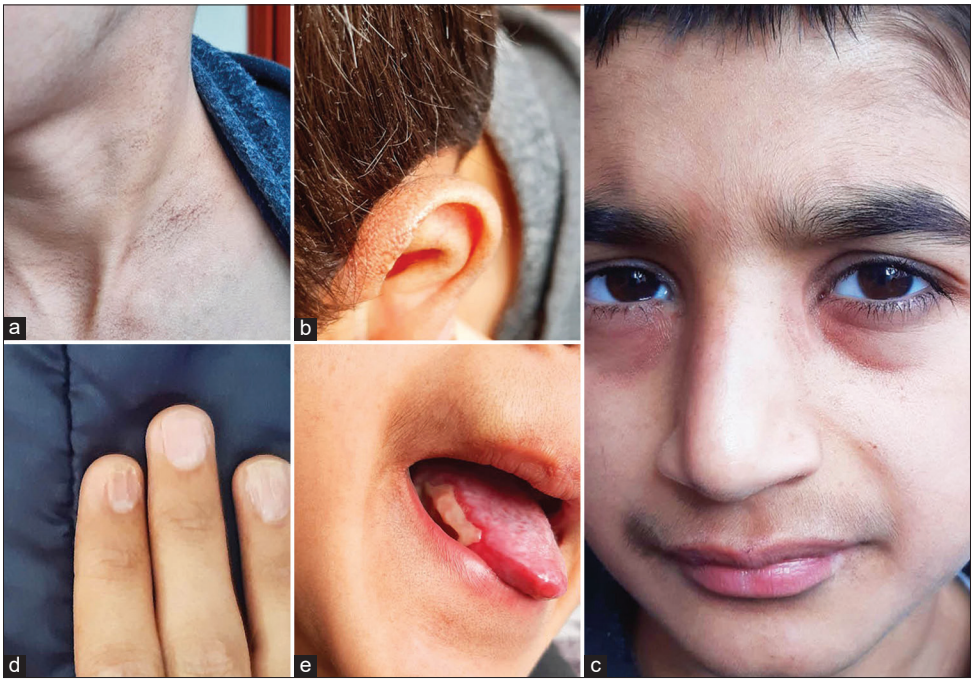

Figure 1: The diagnosis of DC was established with mucocutaneous findings. The reticular hyperpigmentation on his neck (a), ear (b), periocular hyperpigmentation (c), fingernail dystrophy showing longitudinal white lines and ridding (d), and oral leukoplakia (e).

or intraoperative complications. Postoperatively, the patient was treated with topical and systemic antibiotic therapy for 10 days. Follow-up examination was performed weekly during the 1st month, monthly over the next 6 months, and then every 2 months. The tubes were removed 6 months after the surgery was performed, and no recurrence both eyes were observed during the approximately 13-month follow-up.

DISCUSSION

The majority of the mutations known to cause DC result in defects in the length and maintenance of telomeres, a major determinant of stem cell maintenance.[1-3] Shortened and unstable telomeres mainly impact rapidly dividing cells caused by accelerated shortenings of DNA, such as bone marrow and mucocutaneous tissues.[2,3] Due to the etiology, patients are more susceptible to the malignant transformation, including aplastic anemia, squamous cell carcinoma, and leukemia. BMF, which frequently develops by 20 years of age and affects up to 80% of patients by the age of 30, is the main cause of early mortality.[1-3] DC is a heterogeneous disease with multiple and variable clinical manifestations based on the affected gene, and there is considerable clinical variability between patients.[3,5]

The current patient has manifested with developmental delay by early childhood, and fingernail atrophy, reticular skin hyperpigmentation, and oral leukoplakia at the age of seven. Classical mucocutaneous changes are the most common features, usually appearing under the age of ten, of up to 90% of the patients with DC.[2] Due to the possibility of malignant transformation from leukoplakia and hyperpigmentation areas, they need to be monitored periodically and warned about predisposing factors such as ultraviolet exposure and smoking.[8]

Epiphora has been reported as the most common ophthalmological symptom associated with DC, up to 78%.[4,5,8] It has been reported to be mostly associated with punctal stenosis or punctal atresia secondary to epithelial abnormalities in the mucous membranes, rather than NLDO. However, there is no knowledge of its frequency. Moreover, NLDO incidence may be higher than thought due to difficulties in performing lacrimal irrigation due to punctal stenosis or atresia in patients with DC. Although few publications are reporting punctal stenosis that has been treated with dilatation and conservative medical treatments, there are insufficient data on long-term follow-up on treatment strategies.[4,5,8] However, any surgical intervention to the punctum may result in recurrence due to mucous membrane abnormalities. Since our case had bilateral punctal stenosis accompanied by NLDO, ext-DCR with silicone stenting was planned without considering any additional surgical intervention except for dilatation to the punctum.

Pediatric DCR has challenges due to anatomical factors and healing-related issues.[9,10] Causes of failure in pediatric DCR are mostly associated with aggressive healing responses, leading to closure of ostium and granuloma formation.[9] Patients with DC may have more excessive healing response due to mucous membrane abnormalities, which may reduce surgical success. Besides, excessive bleeding, which is a major concern during pediatric DCR,[9,10] may be a particularly serious problem due to BMF, which is likely to occur in DC patients. In the current case, surgical success has been achieved by bleeding control during the surgery, the creation of a large ostium (at least 10 mm in diameter), both anterior and posterior mucosal flap anastomosis, and insertion of the silicone tube for 6 months. During the surgery, removal of bone was relatively easy and no difficulty or difference in the whole procedure was found in comparison with any other pediatric DCRs. There is some controversy about stent intubation in pediatric DCR, but most studies suggest considering stenting for all pediatric DCRs.[10] It has been reported that silicone tubes can be left in place for 6 weeks to 6 months in the literature.[9,10] In our case, the silicone stent was left in place for 6 months without any complications related to stenting. Due to the susceptibility to canalicular and punctal stenosis in DC patients, we recommend silicone intubation as long as possible without causing any iatrogenic damage to the punctum and canaliculus.

A small number of patients with DC may show retinal vascular changes, retinal neovascularization, exudative retinopathy, retinal detachment, and corneal neovascularization related to limbal stem cell deficiency.[6,7] Besides, systemic treatment of DC with radiation or systemic steroids will increase the risk of glaucoma and cataract formation.[4] A baseline and lifetime periodic ophthalmic examination should be recommended for every patient with DC.

Patients can apply to an ophthalmologist with a complaint of epiphora and mucopurulent discharge without a known diagnosis of DC. It is important to perceive the classical and more subtle manifestations of the disorder and refer to the investigation of multisystem involvement. This is the first case report that discusses the management of DCR surgery in a child with DC in the literature. Persistent epiphora may affect academic success and social life in school-age children. Thus, when the patient's systemic condition is suitable for surgery, if there is no sign of acute dacryocystitis, the surgery should be decided as soon as possible. Surgical success can be achieved with pre-operative preparation, and appropriate intraoperative and post-operative management.

CONCLUSION

Before the DCR performing, an ophthalmologist may be aware of mucocutaneous abnormalities and its predisposition to recurrence. It is important to recognize various presentations

of DC and its possible complications, including BMI, aplastic anemia, malignant transformation, and other serious ophthalmic features. Patients should be recalled for regularly follow-ups, where the physician plays an important role.

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Adhered to the ethical principles outlined in the Declaration of Helsinki as amended in 2013.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no conflicts of interest.

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