Bilateral congenital lacrimal fistulas in an adult as part of ectrodactyly-ectodermal dysplasia-clefting syndrome: A rare anomaly

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Ectrodactyly-ectodermal dysplasia and clefting syndrome or "Lobster claw" deformity is a rare congenital anomaly that affects tissues of ectodermal and mesodermal origin. Nasolacrimal duct (NLD) obstruction with or without atresia of lacrimal passage is a common finding of such a syndrome. The authors report here even a rarer presentation of the syndrome which manifested as bilateral NLD obstruction and lacrimal fistula along with cleft lip and palate, syndactyly affecting all four limbs, mild mental retardation, otitis media, and sinusitis. Lacrimal duct obstruction and fistula were managed successfully with endoscopic dacryocystorhinostomy (DCR) which is a good alternative to lacrimal probing or open DCR in such a case.

Key words: Ectrodactyly-ectodermal dysplasia and clefting syndrome, endoscopic dacryocystorhinostomy, lacrimal fistula, nasolacrimal duct obstruction

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The ectrodactyly-ectodermal dysplasia-clefting (EEC) syndrome or "Lobster claw" deformity is a rare autosomal dominant familial disorder with variable penetrance. It involves both ectodermal and mesodermal tissues and consists of ectodermal dysplasia manifested as hypopigmentation of the skin and hair, scanty hair and eyebrows, absence of eyelashes, nail dystrophy, hypo- and microdontia, ectrodactyly, and cleft lip and palate.[1,2] Other associated clinical features include abnormalities of genitourinary system (i.e., renal agenesis, urethral atresia, and hydronephrosis), conductive or sensorineural hearing loss, choanal atresia, mammary gland or nipple hypoplasia, ophthalmological findings (i.e. nasolacrimal duct [NLD] obstruction, lower lid punctal stenosis, photophobia, corneal ulcerations, keratitis, blepharitis, and entropion), gland abnormalities (i.e. hypoplastic thymus, hypopituitarism, and growth hormone deficiency), and on exceptional circumstances presence of white sponge nevus, delayed developmental milestones, and malignant lymphoma but without any intellectual deficit.[2] Chromosome 19, within the region of D19S894 and D19S416, has been postulated as the locus for the abnormalities found in EEC syndrome. Recently, the p63 gene

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has been targeted in numerous studies.[3,4] Though the syndrome is rare, the exact prevalence of EEC syndrome is not known. Just more than 300 cases have been reported in literature so far.[5] Bilateral lacrimal fistulas may also be seen in patients with VACTERL, CHARGE, and Down's syndrome. Here, we present a case of EEC syndrome with bilateral congenital lacrimal fistulas, which is by far one of the rare combinations of this syndrome.

Case Report

A 17-year-old male student was admitted with epiphora and intermittent discharge of pus from an accessory openings situated close to the medial canthus of both eyes (more on the right side than left) since he was 4 years of age. He also presented with nasal regurgitation of fluid. There was clefting of the lip and palate, and the absence of digits in all four limbs since birth. There were no urinary symptoms. There was no family history of such a condition, but the patient was mildly mentally retarded since childhood. He underwent cleft lip repair surgery in childhood [Fig. 1].

The general examination was unremarkable except scar mark of previous cleft lip surgery over face and webbing of fingers of upper and lower limbs. On local examination, regurgitation of pus on pressure over lacrimal sac area test was positive bilaterally. Lacrimal syringing was performed, and there was regurgitation of fluid mixed with pus through the fistula and other punctum with hard stop bilaterally [Fig. 2].

Otoscopic examination showed purulent discharge with perforated tympanic membranes bilaterally. Ultrasonography of the lower abdomen and urinary tract were within normal limits. Paranasal sinus X-ray revealed little mucosal thickening of both maxillary sinuses with normal midline nasal septum and turbinates. Pure tone audiometry showed bilateral mild conductive hearing loss. High resolution computed tomography of temporal bone revealed only minimal soft tissue density lesions in middle ear bilaterally [Fig. 3].

Endoscopic dacryocystorhinostomy (DCR) operation was planned on both sides in two sittings under general anesthesia after diagnosis. The operation on the right side was done first as symptoms were more on that side. The surgery was performed with 0* 4 mm diameter rigid Hopkins rod telescope and the "mucosal flap" technique was used.[6] Nose is decongested with a mixture of 4% lignocaine and 1:1000 adrenaline solution in a ratio of 10:1 approximately 10 min before surgery. At the beginning of the surgery, 2 ml of readymade solution of 2% lignocaine containing 1:100000 adrenaline is injected just anterior to and above the middle turbinate. Now, an incision is taken starting 8 mm above the axilla of the middle turbinate and brought 8 mm anterior to it and from there vertically downward to the attachment of inferior turbinate and ultimately taken posteriorly to the insertion of uncinate process under the edge of middle turbinate. Thus, a posteriorly based mucosal flap is created which is raised to expose the suture line between the frontonasal process of maxilla and lacrimal bone. Now, a part of frontal process is removed using DCR punch forceps along the previously identified suture line to make a neo-ostium, and this is continued up to the fundus of the sac superiorly. Agger nasi cell forms a good guide to the fundus. Thus, the lacrimal sac is fully exposed. Now, the inferior is punctum is dilated, and the lacrimal sac is tented using Bowman's probe. Now, a vertical incision is made over the medial wall of the sac using DCR spear knife as posteriorly as possible. Free flow of normal saline was established inside the nose preoperatively while syringing [Fig. 4].

Two horizontal releasing incisions are given on superior and inferior aspects of the flap making it "I" shaped. The flaps are rolled out. Then, the center of the original nasal mucosal flap is trimmed to make it "C" shaped to cover the raw bony edges. Normally, anterior lacrimal flap is covered by mucosal flap. Usually, there is no need to pack the nose at the end of the procedure. All patients receive broad spectrum antibiotic therapy for 5 days and antibiotic-steroid eye drop for 2 weeks. Saline nasal spray was administered immediately after the surgery and for 4 weeks postoperatively to aid in removal of blood clots from the nasal cavity. The tearing was relieved postoperatively on the right side. Lacrimal syringing performed on the seventh, fourteenth, and twenty-first postoperative day along with nasal endoscopy showed normal passage of saline in the nose and throat. The patient was followed up to 1 year. Spontaneous resolution of lacrimal fistula observed on successful treatment of concurrent NLD obstruction even without excision of the tract. We performed endoscopic DCR on the left side after 3 months and obtained similar results. Sinusitis and chronic otitis media were treated conservatively and responded. Cleft lip was repaired in the childhood by a plastic surgeon.

Discussion

The acronym EEC was coined by Rudiger in 1970 and recognised first as a distinct clinical entity by Cockayne, who drew attention to "dacryocystitis" which commonly affects these patients and its association with "atresia of the lacrimal ducts," in its first published report. Since that description in 1936, several other reports have appeared in the ophthalmic literature detailing its ocular manifestations.[5] There are very few studies which tell about its prevalence in general population. According to one study, EEC is relatively common than other similar syndromes (i.e. Acro-dermato-ungual-lacrimal-tooth syndrome (ADULT), Limb-mammary syndrome (LMS), etc.) with over 200 published cases.[3] Another study observes a large Dutch family with autosomal dominant limb mammary syndrome (LMS) with at least 27 affected members. Lacrimal duct atresia was found in 7 out of 15 such patients with LMS. In all these syndromes, NLD atresia is a common feature.[4] According to another case report, NLD obstruction was present in the most of the affected members of a family of ADULT syndrome and was resistant to therapy.[7] Mcnab *et al*. studied some families with EEC syndrome and showed lacrimal canalicular atresia or absent lacrimal puncta along with NLD obstruction with cleft lip and palate are common in such families. Moreover, limb deformities and lacrimal drainage anomalies though common are not unique to it.[5] We found a similar combination of findings in our case. Wojcicki *et al*. operated 10 such patients with NLD atresia by either duct dilatation or open DCR and observed both procedure effectively improved patients' quality of life.[8] According to one case series, obstructed lacrimal canaliculi or NLD may be associated with lacrimal gland dysfunction. In such patients with dry eye and obstructed NLD surgery may be required. They operated successfully three cases without any technical difficulties. The only postoperative complication was cheese-wiring of the lid margins from the inserted O'donoghue tubes in one case. This was abetted by

Figure 1: Gross appearance of the patient

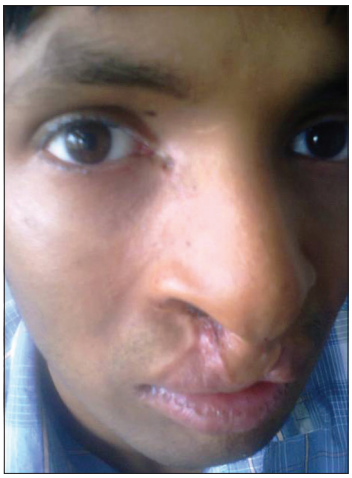

Figure 2: Right sided lacrimal fistula and repaired cleft lip of the same patient

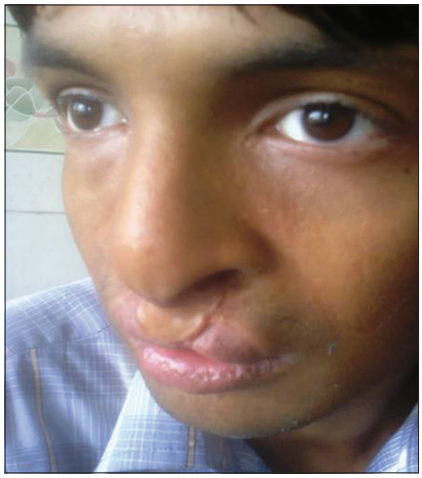

Figure 3: Smaller left lacrimal fistula of the patient

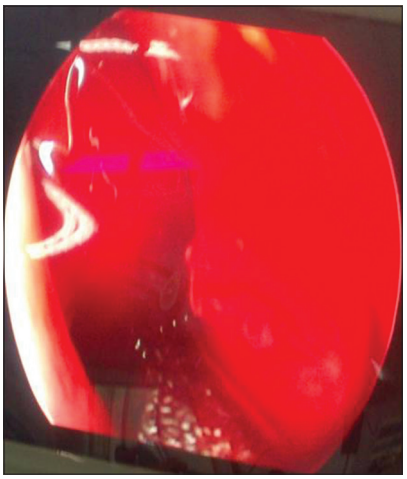

Figure 4: Peroperative picture during endo-DCR showing free passage of water on syringing after excision of sac wall

the abnormally thin lid margins overlying the canaliculi. Moreover, they have not found it necessary to resort to dacryocystectomy or Lester Jones bypass tubes.[5] Tien and Tien described a case of EEC syndrome with bilateral lacrimal fistulae where they excised the fistulas and intubated the NLDs with improvement in symptoms.[7] Elmann *et al*. reported a case of EEC syndrome with left lacrimal fistula and other bilateral congenital lacrimal anomalies who underwent left conjunctivo DCR with the insertion of a Jones tube with a resolution of lacrimation postoperatively. According to the authors that is the only second report detailing management of congenital lacrimal anomalies in EEC syndrome, and the first describing management of punctal atresia with conjunctivo DCR and Jones tube placement.[9] In that sense, ours is the second case to report bilateral lacrimal fistulas in a patient of EEC syndrome and first only to manage it by endoscopic endonasal DCR surgery. We performed endoscopic DCR alone because our patient was an adult one and making a big neo-ostium endonasally, we wanted to allow the fistula tract heal spontaneously thus avoiding an extra surgery like fistulectomy, and our postoperative result was very encouraging. Endo DCR also avoids external scar mark on the skin and do not hamper lacrimal endothelial pump mechanism. We did not find any particular technical problem preoperatively. Al-Salem *et al*. studied fifteen patients of congenital lacrimal fistula and found the spontaneous resolution of discharge through fistula in 27% cases on successful treatment of concurrent NLD obstruction with lacrimal sac massage, so as our case.[10] Buss *et al*. reported clinical presentation and management of 24 cases of EEC syndrome observed conductive deafness and genitourinary problems in some cases required surgical intervention.[11] We were able to manage otitis media and sinusitis conservatively.

We would like to conclude that EEC syndrome presenting with bilateral lacrimal fistulas is a rare and crippling condition, it needs multidisciplinary team approach consisting of an orthopedic surgeon, psychiatrists, ophthalmic and plastic surgeon, urologist, and ENT surgeon to manage such a case. A patient of EEC syndrome presenting with NLD obstruction and fistula, endoscopic endonasal DCR can be a better alternative to duct dilatation and even open DCR.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients

understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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