Dermolipoma in a case of split hand/foot malformation: A report of a novel ophthalmic presentation in a rare disease

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We report a novel ophthalmic presentation in the form of dermolipoma in a rare disease called split hand/foot malformation (SHFM), also known as ectrodactyly. This disease entity is characterized by a failure of development of the central ray of hands and/or feet, leading to a median "cleft" in one or more limbs. This often gives the patient a "lobster claw" appearance.

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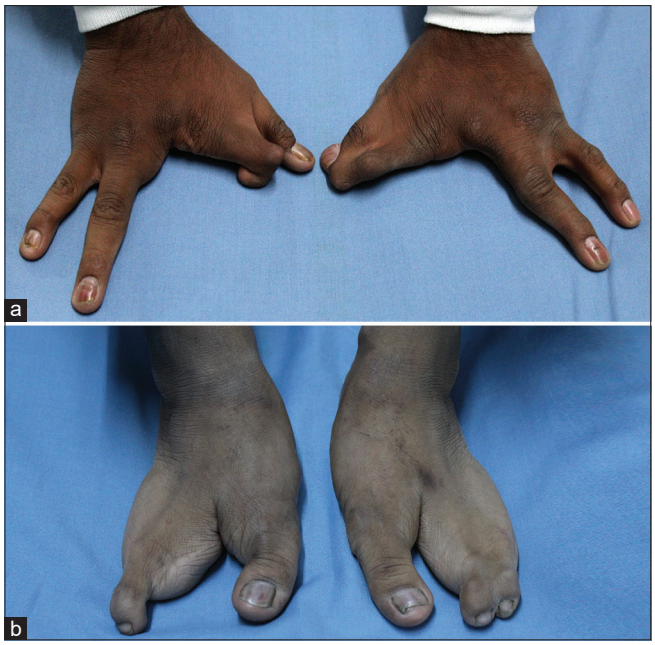

Figure 1: Both hands (a) and feet (b) showing ectrodactyly, with a characteristic median cleft, giving the limbs a "lobster claw" appearance. This cleft is believed to result from a central ray deficiency during development. Closer examination reveals ridges and discoloration of nails

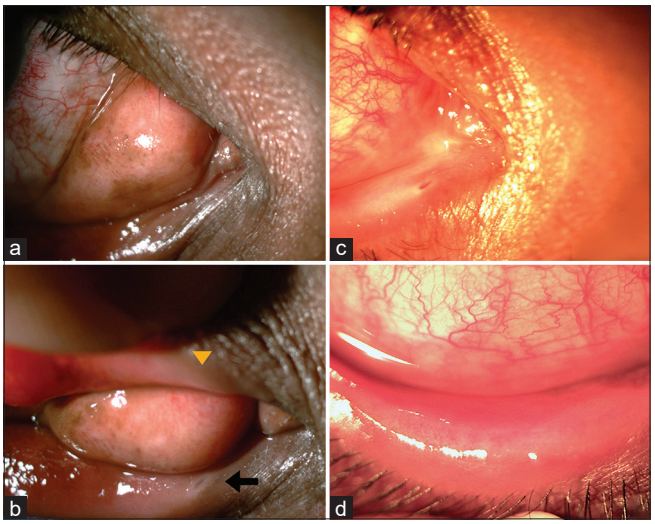

Figure 2: Slit-lamp photographs of right eye showing (a) an epibulbar mass in the inferonasal quadrant of the globe, abutting the caruncle and extending superiorly to just above the medial canthus, and inferiorly into the fornix. The mass had a pink-yellow tinge to it with a smooth surface and no hair follicles projecting through it; (b) upper punctal atresia (yellow arrowhead), with absence of a punctal papilla and a normal lower punctum (black arrow) with a slit-like configuration; (c) postoperative appearance of medial canthal region after mass excision; (d) absence of meibomian gland duct orifices in lower eyelid (taken post-operatively)

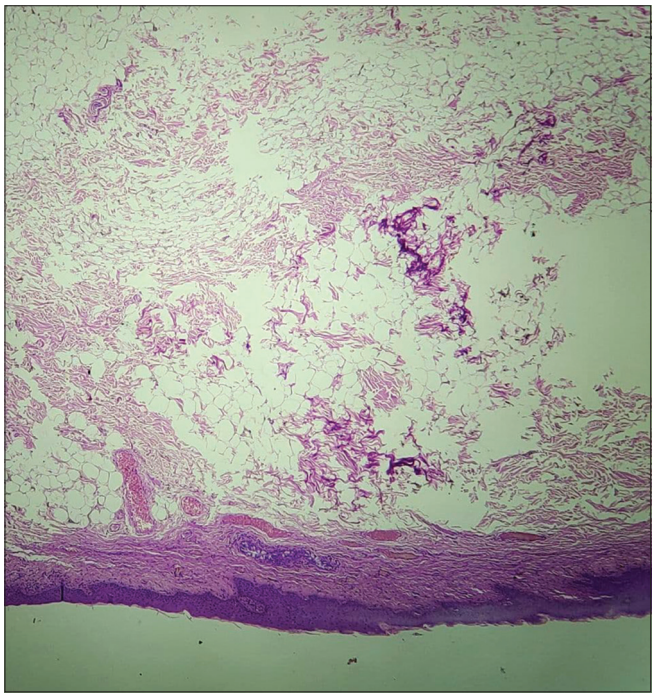

Figure 3: Histophotograph (H and E stain; original magnification 40×) showing conjunctival mucosa lined by cuboidal to columnar cells with goblet cells. Subepithelium shows lobules of adipose tissue, fibrosis, and absence of adnexal glands: features consistent with dermolipoma

Various ophthalmic features have been reported in association with SHFM. These most commonly include lacrimal system anomalies (punctal atresia, canalicular atresia, NLDO). Other findings such as congenital nystagmus, absent/dysplastic meibomian glands, corneal opacities, developmental glaucoma, and cataracts have been reported as well.

This case is unique as our patient presented with a mass lesion in his eye which was diagnosed as an epibulbar dermolipoma based on typical clinical appearance, later confirmed with histopathology. He also had upper punctal atresia and nasolacrimal duct obstruction on the same side. To the best of our knowledge, dermolipoma as a presenting ophthalmic feature in a case of SHFM has not been reported previously. The patient was managed with excision of the dermolipoma along with external dacryocystorhinostomy with monocanalicular intubation on his right side. He had good surgical outcome at six months follow-up, with resolution of symptoms of watering and no recurrence of the mass.

A 22-year-old male presented with a mass lesion in his right eye since birth, associated with watering since early childhood. There was no prior history of eye surgery/trauma. Birth and developmental history were unremarkable. Pedigree charting did not reveal any syndromic disorders in the family. General physical examination revealed ectrodactyly in all four limbs, with a median cleft running through both hands and feet [Fig. 1]. Nails showed ridging and discoloration. Oral cavity examination revealed oligodontia and conical lateral incisors. There were no skin/hair abnormalities.

Ophthalmic examination showed a best-corrected visual acuity of 6/6, N6 in both eyes. There was an epibulbar mass measuring 15 mm maximally in the inferonasal quadrant of the right eye [Fig. 2a]. The mass was pink-yellow in color; it was contiguous with overlying conjunctiva, and had a smooth surface with no hair follicles projecting through it. It was slightly mobile over the underlying sclera. We also noted an upper punctal

atresia [Fig. 2b] on the same side. Regurgitation of mucoid fluid was noted from the lower punctum on applying pressure over the lacrimal sac. There were no meibomian gland (MG) orifices visualized on the lower eyelid margins [Fig. 2d] in either eye, however upper eyelids had normal MG openings. Right-sided nasolacrimal duct obstruction (NLDO) with a patent lower canaliculus and upper-punctal atresia were confirmed on probing of the lacrimal system. Rest of the ophthalmic examination, including dry eye screening, was unremarkable in both eyes.

Based on the typical clinical appearance of the mass, a diagnosis of epibulbar dermolipoma was made, along with punctal atresia and NLDO on the right side, in association with split hand/foot malformation (SHFM).

Computed tomography of the orbits showed a soft-tissue density mass near the medial canthus of the right eye, separate from the nasolacrimal duct, not extending into the orbit.

The patient was offered genetic analysis for a comprehensive systemic diagnosis; however, he refused to undergo the same due to socio-economic factors.

He underwent excision of the mass combined with an external dacryocystorhinostomy with monocanalicular stent in the lower canaliculus under general anesthesia. The postoperative course was uneventful. He was given oral antibiotics with non-steroidal anti-inflammatory drugs, and topical steroid-antibiotic eye-drops along with a tear substitute.

Histopathology of the excised specimen confirmed the diagnosis of dermolipoma [Fig. 3].

The canalicular stent was removed at four months. The patient remained asymptomatic with respect to the watering and was satisfied with his cosmetic appearance. At the six months follow-up visit, there was no recurrence of the mass [Fig. 2c], and objective evaluation of the lacrimal drainage system revealed patency.

Discussion

Literature search of split hand/foot malformation, otherwise known as ectrodactyly, reveals an exhaustive list of systemic syndromes. However, ophthalmic features in SHFM have been sparingly reported. These range from lacrimal drainage system atresia[1-3] to congenital nystagmus,[4] congenital cataracts,[4] developmental glaucoma,[5] meibomian gland agenesis,[3] and resultant ocular surface disease and corneal perforations.[1,3]

To the best of our knowledge, an ophthalmic dermolipoma in association with SHFM has never been reported. This novel presentation makes our case unique. We hypothesize the origin of this dermolipoma to be a result of ectodermal dysplasia that otherwise manifests as skin/hair/teeth or lacrimal duct anomalies[1] in patients with syndromic SHFM.

It is recommended that such patients should have a long-term follow-up with ophthalmologists to monitor for potentially vision-threatening complications like glaucoma, and corneal perforation due to absent/dysplastic meibomian glands.[1,3,5]

Although our case report is limited by a lack of genetic testing, it would be worthwhile offering the same to such patients, to establish a complete diagnosis, and to assist in their systemic management.

Split hand/foot malformation or ectrodactyly presents with myriad ocular associations. Hence it is essential for ophthalmologists to look for anomalies in tissues with ectodermal origin. Such patients should be followed up long-term to avoid potentially blinding complications.

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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Nil.

Conflicts of interest

There are no conflicts of interest.

References

1. McNab AA, Potts MJ, Welham RA. The EEC syndrome and its ocular manifestations. Br J Ophthal-mol 1989;73:261-4.
2. Salmon MA, Wakefield MA. The nasolacrimal ducts and split-hand/split foot syndrome. Dev Med Child Neurol 1977;19:418-9.
3. Bonnar E, Logan P, Eustace P. Absent Meibomian glands: A marker for EEC syndrome. Eye 1996;10:355-61.
4. Pilarski RT, Pauli RM, Bresnick GH, Lebovitz RM. Karsch-Neugebauer syndrome: Split foot/split hand and congenital nystagmus. Clin Genet 1985;27:97-101.
5. Rewri P, Ali W, Grover A, Yadav V. Developmental glaucoma in a patient of ectrodactyly-ectodermal dysplasia. Indian J Ophthalmol 2020;68:910-1.