Ophthalmic manifestations in Rothmund–Thomson syndrome: Case report and review of literature

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A 24-year-old male patient presented to us with diminution of vision in both eyes with watering and photophobia for the past 8 years. General physical examination showed short stature and poikiloderma. Ocular findings include photophobia with reflex tearing, dry eye, cicatricial ectropion, symblepharon approaching pupillary area of cornea, and multiple superficial punctuate erosions on the cornea. Both eyelids showed scanty meibomian glands on infrared meibography. The rest of the anterior and posterior segment was normal. The patient was treated with topical lubricants which reduced photophobia and corneal erosions. He then underwent symblepharon release with buccal mucosal grafting, which improved ectropion. Patient improved symptomatically with reduction of photophobia and improvement in vision as well.

Key words: Dry eye, meibography, meibomian gland dysfunction, Rothmund–Thomson syndrome

Rothmund–Thomson syndrome is a genodermatosis presenting with characteristic facial rash (poikiloderma) associated with short stature, sparse scalp hair, eyelashes and eyebrows, juvenile cataracts, skeletal abnormalities, premature aging, and predisposition to cancer. Till date, only 300 cases have been reported.[1] Facial erythema spreading to extremities but sparing the trunk is the diagnostic hallmark. Ocular manifestations include juvenile cataracts, but secondary effects due to skin cicatrization and meibomian gland atrophy-like dry eye cannot be neglected. Prompt and timely management can help restore vision and also reduce disturbing symptoms such as photophobia, thus significantly improving the patient's quality of life.

Case Report

A 24-year-old male presented to the ophthalmology department with complaints of diminution of vision and photophobia.

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General physical examination showed short stature with a height of 148 cm, few hypopigmented and hyperpigmented atrophic skin patches, and intolerance to light [Fig. 1]. On ocular examination, best-corrected visual acuity in the right eye was 6/12 and left eye 6/9. Eyelid skin of both eyes showed hyperpigmented atrophy. Eyelid margin of right lower eyelid was everted due to the scarring of skin, leading to cicatricial ectropion. There was no loss of lashes. Conjunctiva of both eyes showed symblepharon [Fig. 2a]. On fluorescein staining of cornea, both eyes showed multiple staining superficial punctuate erosions. Schirmer's test showed reduced tears in both eyes with strips wetting 8 mm in the right eye and 6 mm in the left eye [Fig. 3]. Lacrimal syringing was patent in both sides. On imaging eyelids for meibomian photography, both eyes showed nearly absent meibomian glands [Fig. 4].

Patient was treated with topical lubricants. Right eye symblepharon was released, and buccal mucosal graft was sutured. Skin contracture leading to ectropion was released with full-thickness skin graft taken from arm. Two weeks postoperatively, the patient had relief from photophobia. Vision improved to 6/9 parts in both eyes. The residual diminution of vision was probably due to irregular astigmatism which could not be corrected with spectacles. Corneas in both eyes were clear. Ectropion was corrected, and puncta were better approximated to the globe to drain the tear film [Fig. 2b]. Postoperative Schirmer's test results were 16 mm in the right eye and 14 mm in the left eye. Tear breakup time was 9 s in the right eye and 8 s in the left eye.

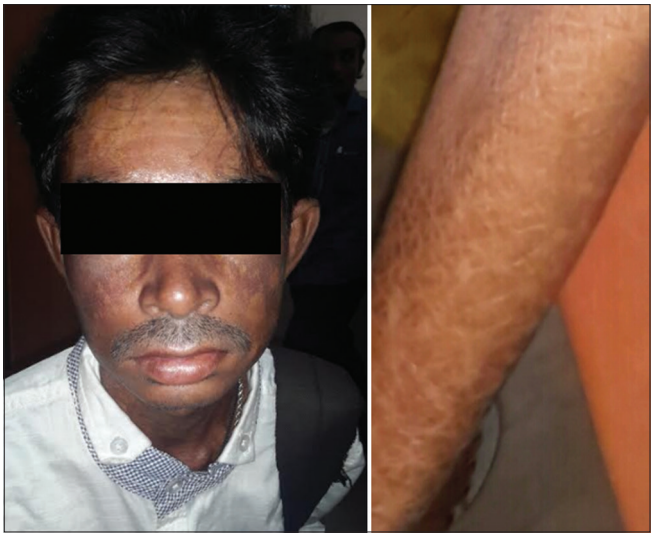

Figure 1: Hyperpigmented and hypopigmented skin lesions on patient's face and forearm

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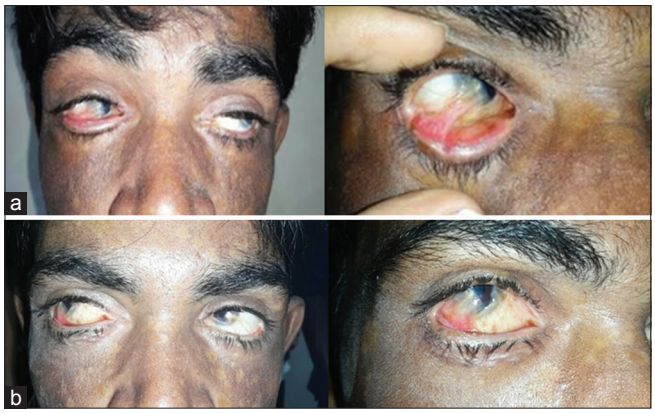

Figure 2: (a) Preoperative picture showing right eye lower eyelid ectropion and symblepharon. (b) Postoperative picture showing correction of ectropion and release of symblepharon

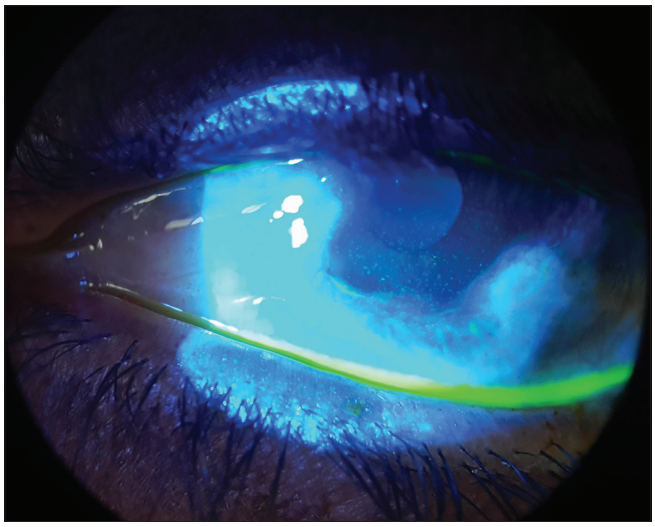

Figure 3: Fluorescein staining erosions on cornea of the left eye

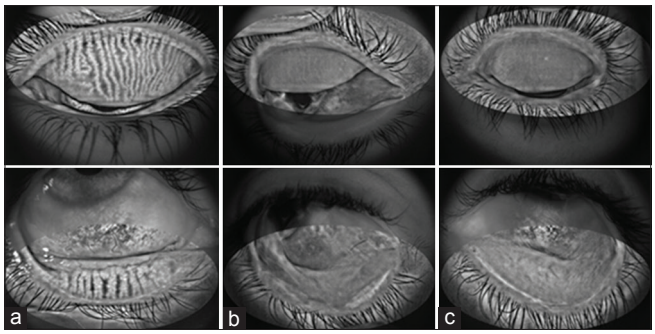

Figure 4: (a) Meibography of upper and lower eyelid of a normal patient showing meibomian glands. (b) Meibography of upper and lower eyelid of right eye of this patient showing absent meibomian glands. (c) Meibography of upper and lower eyelid of left eye of this patient showing absent meibomian glands

Discussion

Rothmund–Thomson syndrome is a very rare genetic disease with no reliable data on its prevalence. To date, approximately 300 patients have been recorded in the medical literature.[1] Inheritance pattern is variable. A few studies show autosomal recessive inheritance,[2] mutations in RECQL4 gene,[3,4] and DNA repair disorders.[5] Clinical spectrum[6,7] is highly variable, which brings together patients with shared and unique developmental defects; thus those who display an atypical or borderline clinical presentation may be overlooked. The diagnostic hallmark is facial erythema, which spreads to the extremities but spares the trunk, which then develops poikiloderma, a skin condition with hypopigmentation, hyperpigmentation, telangiectasias, and atrophy. Two clinical subforms have been defined: RTSI, which has poikiloderma, ectodermal dysplasia, and juvenile cataracts; and RTSII which is characterized by poikiloderma, congenital bone defects, and an increased risk of cancers such as osteosarcoma and skin cancer.[8] Skeletal abnormalities include frontal bossing, saddle nose, and congenital radial ray defects. Gastrointestinal, respiratory, and hematological disorders have also been reported in a few patients.

Ocular manifestations have not been established completely. A few cases commonly have shown juvenile cataracts. There are case reports showing bilateral ocular hypertension,[9] bilateral iris dysgenesis,[10] retinal and corneal atrophies, and corneal opacities.[9,10] Another case report shows the involvement of ocular surface such as inflammatory conjunctival disease with fornix shortening, symblepharon, with sparse eyelashes.[11] Here it is a case presenting with severe meibomian gland dysfunction (MGD) associated with this syndrome leading to dry eye. Infrared (IR) meibography helps in diagnosing the cause for dry eye and photophobia in these cases, apart from cicatricial ectropion due to pathology of skin.

MGD is a chronic, diffuse abnormality of the meibomian glands, commonly characterized by terminal duct obstruction or changes in the glandular secretion. This may result in evaporative dry eye due to alteration in tear film, symptoms of eye irritation with apparent inflammation, and ocular surface disease. MGD results in stasis of meibum inside the glands, dilatation of the ductal system, and loss of glandular tissue (gland dropout). Meibography[12] is an *in vivo* technique used to visualize meibomian glands morphology. The structure of the meibomian glands, including the ducts and acini, are observed in the images generated using meibography. There are two principles. One is the transillumination of the everted eyelid which uses IR light; the other is direct illumination using slit lamp, called noncontact meibography. On IR meibography,[13] normal meibomian glands are hypoilluminescent grape-like clusters while ducts and underlying tarsus are hyperilluminescent. Ductal obstruction can be visualized as hyperkeratinized plugs at the orifices, stasis, and dilatation of glands. Gland atrophy can be seen as dropout areas. Abnormal meibomian glands show features consistent with histopathologic studies where ducts appear dilated and glands become enlarged, tortuous, and eventually dropout. IR meibography thus remains the mainstay of meibomian gland imaging studies because of its reliability and feasibility in producing high-quality gross images of the meibomian glands and enabling easy clinical analysis of gland function.

Conclusion

Rothmund–Thomson syndrome is a rare genetic disorder presenting mainly with skin atrophic changes and systemic involvement. Ocular findings include juvenile cataract. Here is a rare case of syndrome showing MGD. Patient presented with

photophobia and diminution of vision along with other features such as poikiloderma and short stature. On thorough ocular evaluation, he was seen to have bilateral atrophy of meibomian glands, leading to dry eyes. Patient's symptoms improved on treatment with lubricants, further managed with symblepharon release and ectropion correction. Although rare, early diagnosis and prompt management of dry eye in this case significantly improved the patient's quality of life. A simple, noninvasive analysis of meibomian glands using IR meibography helped in diagnosing the cause for dry eye in this patient. Further management with buccal mucosal grafting after symblepharon release helped in maintaining tear film stability.

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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