Dyschromatosis universalis hereditaria with bilateral keratopathy- A rare co-existence

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Abstract
Dyschromatosis universalis hereditaria (DUH) is a pigmentary disorder of rare occurence comprising both hyperpigmented and hypopigmented macules forming an irregular pattern. Although ocular albinism, iris coloboma, aniridia, chorioretinal coloboma have been associated with it, we report a case of DUH with bilateral keratopathy which is a rare co-existence. This case represents the rarest association of DUH with decompensated cornea which was managed successfully with penetrating keratoplasty.

Keywords: Dyschromatosis, Keratopathy, Keratoplasty.

Introduction
Dyschromatosis, a group of pigmentary disorders, is characterized by the both hyperpigmented and hypopigmented pleomorphic macules. This condition was first described by Ichikawa and Hiraga in 1933.[1,2] It forms the part of wide spectrum of dyschromatosis disorders namely dyschromatosis universalis hereditaria (DUH), dyschromatosis symmetrica hereditaria (DSH), acropigmentation of Dohi, and a segmental form called unilateral dermatomal pigmentary dermatosis. DUH is commonly found in Japan some isolated cases have also been reported in India, Europe, Saudi Arabia and South America. We report a case of DUH with corneal decompensation managed successfully with penetrating keratoplasty.

Case Report
A 42 years old male presented in eye OPD with diminution of vision in his right eye for the last 20 years which was gradual, painless and progressive. His past history revealed development of brownish lesions all over the body from 6 years of age which were accompanied by itching and burning sensation on photoexposed areas and intermittent sloughing of skin which was more during summer season. He underwent penetrating keratoplasty in his left eye in 1997 for similar complaint with no postoperative complications. His family history revealed similar complaints of diminution of vision both eyes in his elder sister for which she underwent penetrating keratoplasty both eyes.

On cutaneous examination multiple generalized hyperpigmented macules of the size of 0.5mm to 5mm approximately interspersed with hypopigmented macules.(Fig. 1,2) The lesions were more distributed over the trunk (Fig. 3). There was involvement of oral mucosa, palms and soles. (Fig. 4,5,6) There were no lesions on hair, nails and teeth. His systemic examination showed no detected abnormality. Laboratory examination of blood profile i.e. complete hemogram, renal and liver function tests, within normal limits. Ultrasonogram of the kidney, urinary bladder and upper abdomen revealed no detectable abnormality. Skin biopsy of the hyperpimented macule showed increase in epidermal melanin in basal layers. (Fig. 7) Based on the clinical findings, a diagnosis of DUH was made.

Fig. 1, 2: Hyperpigmented macules over face and head.

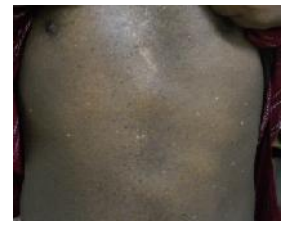

Fig. 3: numerous generalized hyperpigmented macules interspersed with spotty hypopigmented macules over trunk

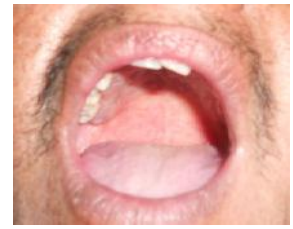

Fig. 4: hyperpigmented macules over hard and soft palate

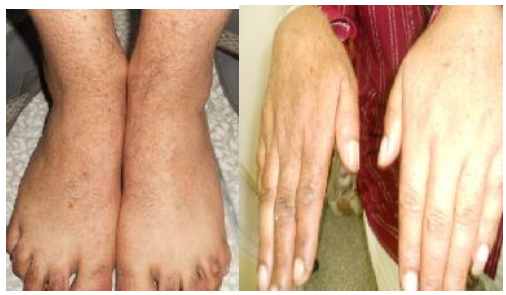

Fig. 5, 6: Involvement of feet and hands with hyperpigmented macules.

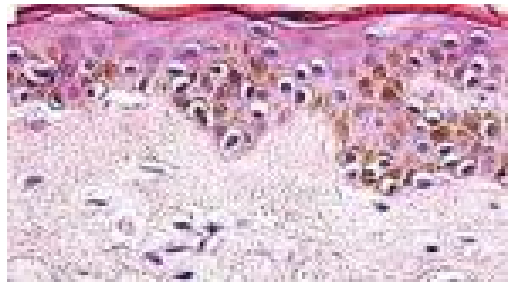

Fig. 7: Skin biopsy of hyperpigmented lesion showing increase in epidermal melanin in basal layers

On ocular examination vision in right eye was 3/60 and in left eye the visual acuity was 6/9. On slit lamp examination there was diffuse corneal odema in right eye (Fig. 8). The left eye showed clear corneal graft with no vascularisation, pigmentation or infiltration (Fig. 9). The intraocular pressure was 14 and 16 with applanation tonometer in right and left eye respectively. The patient was planned for penetrating keratoplasty right eye. The procedure was uneventful. The patient was put on topical tobramycin fluormetholone combination, cycloplegic homatropine and lubricant gels for 2 months. The patient was assessed for clarity of graft, epithelial defect, infiltration, and vascularisation everyday till discharge after 7 days (Fig. 10). He was followed up postoperatively weekly for 2weeks, 2 weekly for 2 months and, monthly thereafter for 3months with no complications. His visual acuity at 6 months postoperatively was 6/12 in right eye and 6/9 left eye.

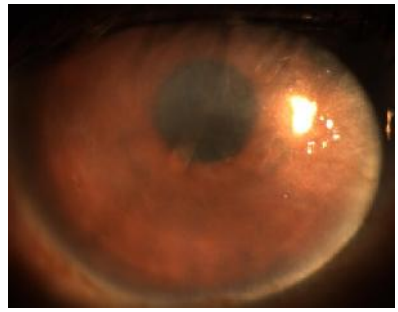

Fig. 8: Diffuse corneal oedema in right eye

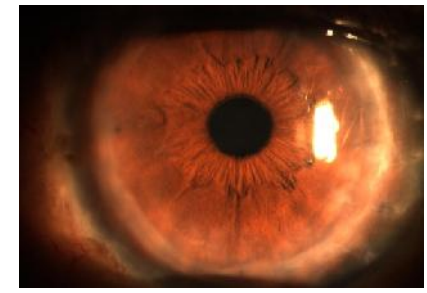

Fig. 9: Left eye showing clear corneal graft

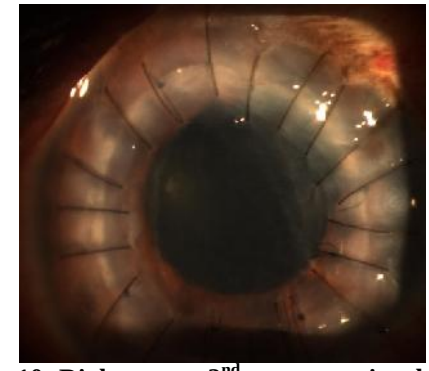

Fig. 10: Right eye on 2nd postoperative day of penetrating keratoplasty

Discussion

Dyschromatosis an autosomal dominant or autosomal recessive, is a rare pigmentary disorder. It is characterised by hyperpigmented and hypopigmented macules of variable shape and size.[3,4] The skin lesions develop in the first decade of life. The lesions mainly involve the trunk and extremities with the involvement of face in 50% of individuals.

The exact etiology of this DUH is not known. Although mutation in adenosine deaminase gene (ADAR1, DSRAD) had been reported in dyschromatosis symmetrica hereditaria involving acral areas but no such mutation is reported in DUH. Recently, studies have reported ABCB6 as the first pathogenic gene associated with DUH.[5,6] In genetically predisposed individuals, DUH is thought to occur due to alteration of the neural-melanocytic interaction in early embryonic life. According to recent studies mapping of the gene responsible for DUH has been done as 6q24.2-q25.2 (OMIM 127500). The diagnosis generally relies on the external clinical features as the biochemical origin of the gene defect is unknown.[7,8] On the histopathological examination of these lesionsthere is focal increase in melanin content of the epithelial basal layer in hyperpigmented lesions and decrease in melanin in hypopigmented lesion associated with pigmentary incontinence. According to recent electron microscopy, DUH has been reported to be a disorder of melanosome production in melanin units rather than a disorder of melanocyte number as it was considered earlier.[9]

DUH has systemic association with small stature, high-tone deafness, tuberous sclerosis. It may be

associated with central nervous system leading to mental retardation, epilepsy. Other associations include X-linked ocular albinism, insulin-dependent diabetes mellitus, erythrocyte, platelet and tryptophan metabolism abnormalities.[7,8,10]

Ocular manifestation of DUH include coloboma, coloboma in association with microphthalmia and aniridia. However this is the first case ever reported of DUH with keratopathy. Although rare, DUH forms an important differential diagnosis of xeroderma pigmentosum (XP) which is more frequent. In the present case there were no features of XP (e.g. poikiloderma, atrophy, or telangiectasia) and no occurrence of any skin malignancy.

Conclusion

This case to best of our knowledge represents the rarest association of DUH with decompensated cornea which was managed successfully with penetrating keratoplasty. Therefore this rare possibility of DUH with this rare association of corneal involvement should be considered while ruling out pigmentary disorders.

References

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