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OCULAR MANIFESTATIONS IN GENODERMATOSES.

Ophthalmology

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ABSTRACT

PURPOSE: To evaluate the Ocular manifestations in patients with genodermatoses. **METHODS:** 49 cases in age group 16-60 years, with a diagnosis of genodermatoses were included in the study. All the patients underwent a complete ophthalmic examination, the findings were noted. **RESULTS:** The most common condition noted was NF 1(23 cases) and NF 2 (18 cases). Other diseases seen were Dariers disease (1 case) Lamellar ichthyosis (7 cases) and Epidermal nevus syndrome (1 case). 10 cases out of 23(43.47%) having NF1 showed Lisch nodules, 13(56.52%) showed eyelid neurofibromas and 1(4%) showed optic nerve glioma. In patients with NF2, 2 cases (11.11%) showed cataracts. Among patients with lamellar ichthyosis, 1 case (14.28%) had ectropion. In the patient with Epidermal nevus syndrome retinal pigment epithelium changes were seen. No ocular finding was seen in the patient with Dariers disease. **CONCLUSION:** Genodermatoses have significant ocular findings. These conditions are rare and hence it is essential to have a complete assessment of these patients with respect to ophthalmological manifestations for better diagnosis and earlier management.

KEYWORDS

Skin Diseases, Genetic, Eye Manifestations, Eye Abnormalities

INTRODUCTION

Skin and eyes have a common embryological origin from the embryonic surface ectoderm. Ocular manifestations in dermatological diseases are common and have diagnostic value. Genodermatoses or genetic diseases of the skin are a group of inherited disorders with cutaneous and systemic signs and symptoms; Deepika Pandhi [1] Genetic skin diseases often have extracutaneous manifestations. Ocular findings are one of the most common associations and can be the initial presenting sign of dermatological diseases. These manifestations may have significant clinical implications like visual disturbance and ocular discomfort , Jen M et al [2] Manifestations like corneal opacities seen in X-linked ichthyosis are asymptomatic but characteristic of a particular genodermatoses.

The rarity of the conditions and the lack of awareness are the cause of major hurdles in the management and the planning of research in this specialty, Deepika Pandhi [1] Early recognition of the ocular involvement of dermatological diseases is helpful in reaching the diagnosis and managing the patient properly for a better outcome; Lamia S et al [3]

At present there are very few studies that emphasize the ocular manifestations of these dermatological diseases. Hence this study aims at understanding the occurrence and severity of ocular manifestations in genetic dermatological diseases.

MATERIALS AND METHODS

This is a cross sectional study. The study was conducted on patients diagnosed with genodermatoses at a tertiary care hospital in Southern India. The study was conducted on patients in the age group of 16-60 years who came to the Dermatology OPD, were diagnosed with genodermatoses and referred to the Ophthalmology OPD for further evaluation.

All the patients diagnosed with genodermatoses, underwent a complete ophthalmic examination, that is, assessment of visual acuity, Anterior segment examination, intraocular pressure assessment and posterior segment examination and the findings were noted.

The research was approved by the institutional review board (IRB) and informed consent was obtained from the subjects after explanation of the nature and possible consequences of the study.

RESULTS

The most common condition noted was NF 1(23 cases) and NF 2 (18 cases). Other diseases seen were Dariers disease (1 case) Lamellar ichthyosis (Fig 4) (7 cases) and Epidermal nevus syndrome (1 case).

10 cases out of 23(43.47%) having NF1 showed eyelid neurofibroma, 13(56.52%) showed Lisch nodules and 1(4%) showed optic nerve glioma (Fig 2,3,5)

In patients with NF2, 2 cases (11.11%) showed cataracts (Fig 6). Among patients with lamellar ichthyosis, 1 case (14.28%) had xerophthalmia and 4 cases (57.14%) had conjunctival congestion. In the patient with Epidermal nevus syndrome, rod – cone dystrophy was seen with pigment epithelium changes (Fig 1). No ocular finding was seen in the patient with Dariers disease.

DISCUSSION

Genodermatoses have significant ocular findings. In our study we came across patients with Neurofibromatosis 1 and 2, Lamellar ichthyosis, Darier's disease and Epidermal nevus syndrome. These diseases are rarely found and so are the ocular findings seen in these patients.

Neurofibromatosis is an autosomal dominant disorder secondary to mutation in the neurofibromin gene. NF 1 is characterized by cutaneous, CNS and ocular manifestations; Lamia S et al [3]. The most commonly seen ocular finding in NF 1 is the presence of Lisch nodules which was seen in 56.52% patients diagnosed with NF1. NF 2 is less common than NF 1 and the most common ocular finding seen in NF 2 is cataract.

Lamellar ichthyosis is an autosomal recessive disorder characterised

by large plate-like scales, Lamia S et al. [3]. The most common ocular manifestation is cicatricial ectropion but it also manifests with changes such as xerophthalmia, exposure keratopathy, and conjunctival congestion.

Darier disease (DD) is a rare autosomal dominant inherited skin disease characterized by hyperkeratotic papules and plaques, prominent nails, and mucous membrane involvement; Helwe Hammad et al [4]. In a few studies patients with Darier's disease showed blepharitis and dry eye symptoms. A study by Helwe Hammad et al noted similar findings of blepharitis but quoted that there was no correlation between the ophthalmologic findings and disease severity or body surface area involved; Helwe Hammad et al [4] .In our study we saw only 1 patient with Darier disease and no ophthalmic manifestations were noted.

The epidermal nevus syndromes (ENS) are a group of congenital syndromes comprising epidermal nevi in conjunction with central nervous system (CNS), ocular, musculoskeletal, and other organ anomalies; Justin J et al [5] Various ocular findings are a part of ENS which consist of eyelid nevi, coloboma, nystagmus and retinal anomalies; Justin J et al [5]. In our study we found nystagmus, rod- cone dystrophy and retinal piment epithelium changes in the patient of ENS. The rarity of these conditions and lack of awareness are major obstacles in their evaluation and treatment. Therefore it is essential to have a complete assessment of these patients with respect to ophthalmological manifestations. It is important for the clinician to be alert to the possibility that the patient may be presenting for the first time with one or more features of a genetic disease so that appropriate investigation and counselling can take place.

Tables : Ocular Manifestations Of Genodermatosis

TABLE 1: Total patients with genodermatosis

Sr no	Genetic skin disease	Number of patients
1	Neurofibromatosis 1	23
2	Neurofibromatosis 2	18
3	Lamellar ichthyosis	7
4	Darier's disease	1
5	Epidermal nevus syndrome	1
TOTAL		50

TABLE 2 : Ocular signs in Genodermatosis

Sr no	Genetic skin disease	Ocular signs	No. of Patients with ocular signs	Percentage
1	Neurofibromatosis 1	Eyelid neurofibroma	10	43.47%
		Lisch nodules	13	56.52%
		Optic nerve glioma	1	4%
2	Neurofibromatosis 2	Cataract	2	11.11%
3	Lamellar ichthyosis	Dry eye	1	14.28%
		Conjunctival congestion	4	57.14%
4	Darier's disease	Nil	0	0
5	Epidermal nevus syndrome	Rod cone dystrophy	1	100%
		Pigmentary epithelial changes	1	100%
		Nystagmus	1	100%

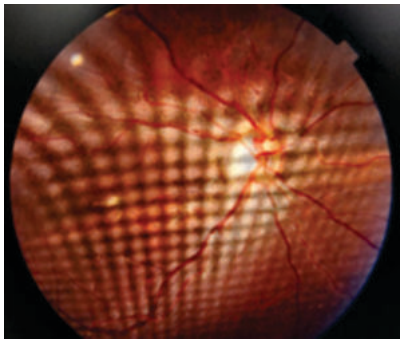

Fig 1. Fundus changes in Epidermal Nevus Syndrome

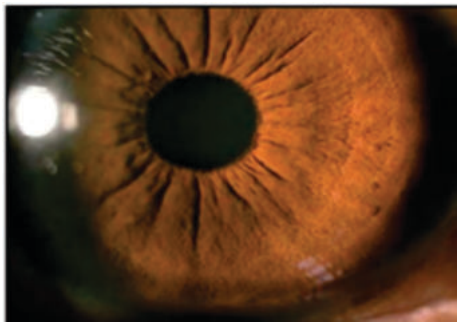

Fig 2. Lisch nodules in Neurofibromatosis 1

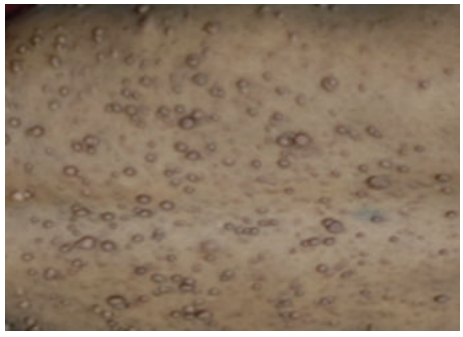

Fig 3. Lesions in Neurofibromatosis 1

Fig 4. Lamellar Ichthyosis

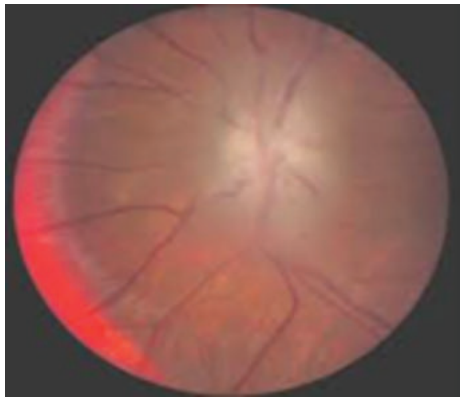

Fig 5. Optic nerve glioma

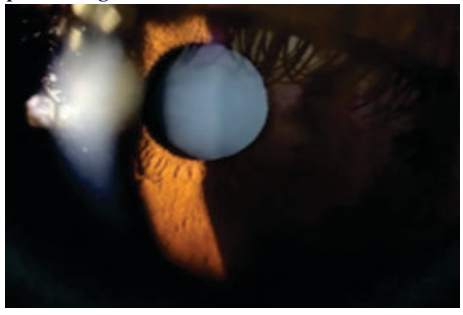

Fig 6. Immature cataract in Neurofibromatosis 2

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