"A CROSS SECTIONAL AND OBSERVATIONAL STUDY OF DERMATOLOGY LIFE QUALITY INDEX SCORE IN GENODERMATOSES PATIENTS AT A TERTIARY CARE HOSPITAL."

By

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In

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DEPARTMENT OF DERMATOLOGY, VENEREOLOGY AND LEPROSY

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BELAGAVI-590010

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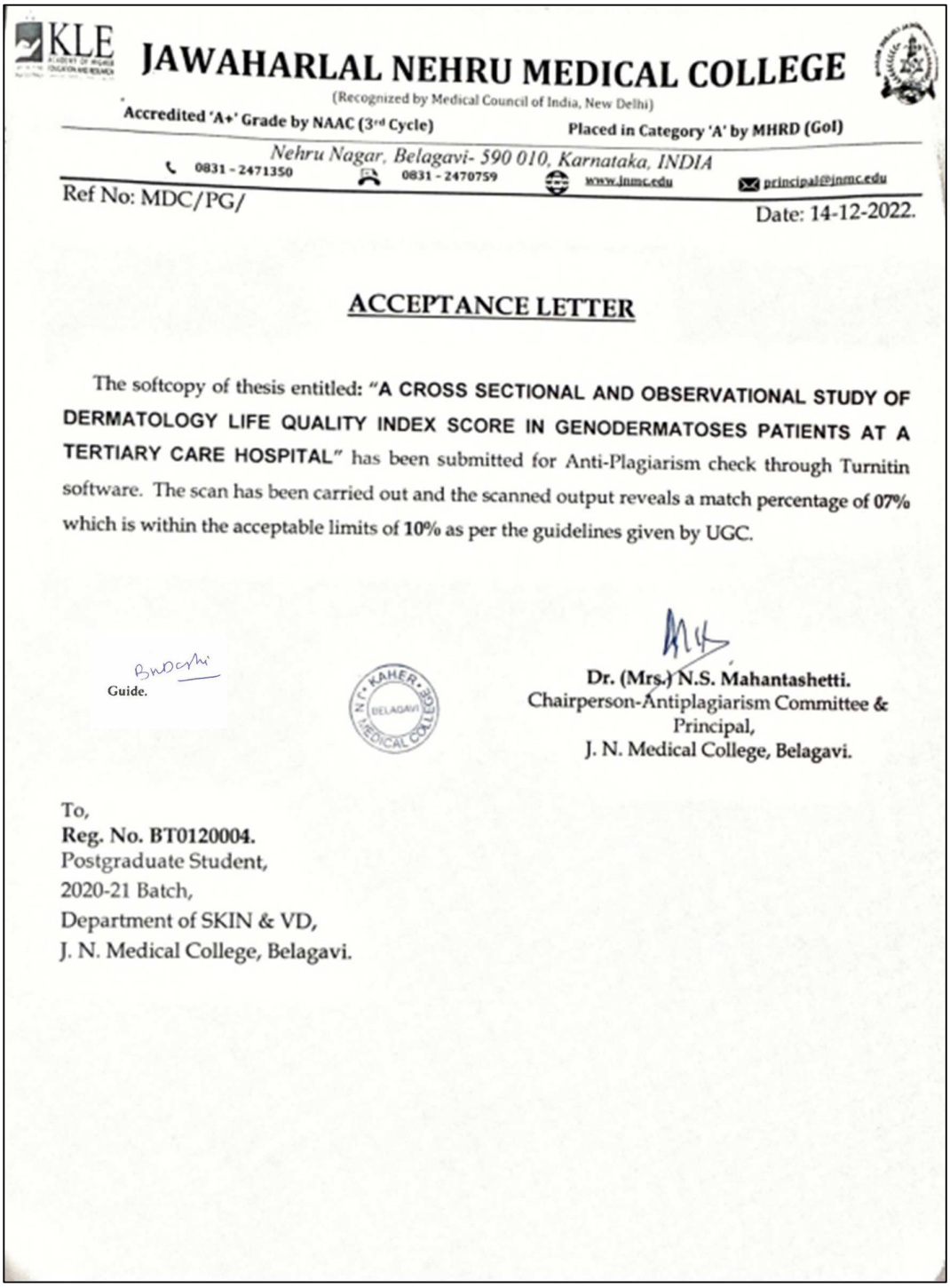

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LIST OF ABBREVIATIONS USED

Sl. No.	Abbreviation	Expansion
1	DLQI	Dermatology Life Quality Index
2	RXLI	Non syndromic recessive X-linked ichthyosis
3	IV	Ichthyosis vulgaris
4	ARCI	Autosomal recessive congenital ichthyosis
5	HI	Harlequin ichthyosis
6	LI	Lamellar ichthyosis
7	CIE	Congenital ichthyosiform erythroderma
8	SHCB	Self-healing collodion baby
9	BSI	Bathing suit ichthyosis
10	KPI	Keratinopathic ichthyosis
11	EI	Epidermolytic ichthyosis
12	ICM	Ichthyosis Curth–Macklin
13	CRIE	Congenital reticular ichthyosiform erythroderma

14	AEI	Annular epidermolytic ichthyosis
15	SEI	Superficial epidermolytic ichthyosis
16	AREI	Autosomal recessive epidermolytic ichthyosis
17	GJB2	Gap junction protein beta 2
18	Cx26	Connexin-26
19	SCC	Squamous Cell Carcinoma
20	PPK	Palmoplantar keratodermas
21	PC	Pachyonychia congenita
22	PLS	Papillon Lefèvre syndrome
23	NF	Neurofibromatosis
24	TSC	Tuberous sclerosis complex
25	EB	Epidermolysis bullosa
26	XP	Xeroderma pigmentosum
27	NER	Nucleotide excision repair
28	QOL	Quality of life

ABSTRACT

Although genodermatoses accounts for a small number of patients in dermatology practice, their diagnosis is of immense significance as most of these are life-long, multi-system disorders with limited treatment options. Some of them risk malignancy and account for premature deaths[1].

Common genodermatoses which we come across in practice belongs to the ichthyosis group, followed subsequently by epidermolysis bullosa, ectodermal dysplasias, albinism, cutis laxa, xeroderma pigmentosum and dyskeratosis congenita[3].As these can cause cosmetic disfigurement, patients present to the dermatologist for their unusual appearance[2].

The rarity of the diseases and scant awareness causes major roadblocks in the management and research on patients with genodermatoses.

Finlay and Khan have designed DLQI (Dermatology Life Quality Index) to assess the social and psychological effect of dermatological conditions on the quality of life in adults[3].

As most genodermatoses lead to disfigurement and cause psychological distress, assessing the DLQI in them may provide some insight into patient's psyche, foster patient-physician relationship thereby leading to better compliance and treatment out comes.

Results: In our study, out of 55 cases, 31(59%) were males in which 21 (67.74%) cases had very large DLQI grading and 24(41%) were females out of which 19 (79.1%) cases had very large effect according to DLQI grading.

14 different types of genodermatoses were noted, where predominantly neurofibromatosis accounted for 17(30.9%) cases, followed by 11(20%) icthyosis vulgaris cases.

Most patients belonged to the age group of 21-40 years which constituted 37 (67.3%) cases. Mean duration of disease onset was 11.03 years. Associated systemic disorders like diabetes and hypertension seen in 2 (3.6%) cases each, 1 (1.8%) case had epilepsy

Out of 55 cases that are included in the study 12 (21.8%) had a positive family history. 33/55 (60%) cases are unmarried out of which 29 (87.87%) had a very large DLQI grading. In this study history of consanguineous marriage in parents was present in 22 /55(40%) cases.

2 (3.6%) cases out of 55 cases went to psychiatrist for counselling regarding the disease and had a very large DLQI grading. Counselling regarding the disease done only in 2 (3.6%) cases out of 55 cases.

In this study 40/55 (72.7%) cases had very large DLQI grading, out of 40 cases with very large DLQI grading 31 (77.5%) belonged to the age group of 21-40 years.

Conclusion: To the best of our knowledge our study is the first to study the affect on quality of life across various factors wherein we observed that female sex, 21-40 years age group and u- married subjects had a very large affect as per DLQI score which lead to feeling of embarrassment, disturbed social activities and relationship with friends and family.

Patients with genodermatoses as well as their parents require to be counselled regarding the nature of their disease, chances of familial involvement, the associated possible systemic involvement , need for regular hospital visits and educate them regarding the available treatment options as these impact their quality of life.

Social help and vocational rehabilitation too need to be developed to help integrate these patients with the mainstream.

Limitation:

1. Small sample size. A study with larger sample is needed to validate our studies findings.

2. Along with assessment of patient's quality of life, a study to assess the patient's parents/guardians quality of life might help in understanding the overall impact on inter- personal relationship and family life.

Key words: Genodermatoses, DLQI

TABLE OF CONTENTS

LIST OF TABLES

LIST OF CHARTS

LIST OF FIGURES

INTRODUCTION

Although genodermatoses accounts for a small number of patients in dermatology practice, their diagnosis is of immense significance as most of these are life-long, multi-system disorders with limited treatment options. Some of them risk malignancy and account for premature deaths[1].

Common genodermatoses which we come across in practice belongs to the ichthyosis group, followed subsequently by epidermolysis bullosa, ectodermal dysplasias, albinism, cutis laxa, xeroderma pigmentosum and dyskeratosis congenita[3].As these can cause cosmetic disfigurement, patients present to the dermatologist for their unusual appearance[2].

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OBJECTIVE

- To evaluate the Quality of life in patients of genodermatoses using Dermatology Quality of Life Index

REVIEW OF LITERATURE

Genodermatoses belong to a group of inherited disorders which present with skin and systemic involvement. They are caused by the lethal mutations arising from mosaicism or may be inherited through the Mendelian modes of inheritance. The structures involved in the development of these include the keratinocyte, the collagen, the toenail, the hair, and the teeth.

DISORDERS OF CORNIFICATION

ICHTHYOSIS

‘‘Ichthyosis’’ belongs to a group of keratinizing disorders which can be either inherited or acquired. This term is derived from the Greek word‘‘ichthyosis,’’ which means ‘‘fish’’, given that the skin of affected patients often resembles fish scales[4].

CLASSIFICATION OF ICHTHYOSIS

NON SYNDROMIC FORMS :

Common ichthyoses

1. Non syndromic recessive X-linked ichthyosis (RXLI)
2. Ichthyosis vulgaris (IV)

Autosomal recessive congenital ichthyosis (ARCI)

1. Harlequin ichthyosis (HI)
2. Lamellar ichthyosis (LI)
3. Congenital ichthyosiform erythroderma (CIE)
4. Self-healing collodion baby (SHCB)
5. Acral self-healing collodion baby
6. Bathing suit ichthyosis (BSI)

Keratinopathic ichthyosis (KPI)

1. Epidermolytic ichthyosis (EI)
2. Ichthyosis Curth–Macklin (ICM)
3. Congenital reticular ichthyosiform erythroderma (CRIE)
4. Annular epidermolytic ichthyosis (AEI)
5. Superficial epidermolytic ichthyosis (SEI)

Autosomal recessive epidermolytic ichthyosis (AREI)

1. Epidermolytic naevi

Syndromic forms:

1. X linked ichthyosis syndromes
2. Autosomal ichthyosis syndromes with prominent hair abnormalities:
3. Netherton syndrome.
4. Autosomal ichthyosis syndromes with prominent neurological signs
5. Autosomal ichthyosis syndromes with deafness: KID syndrome.
6. Autosomal ichthyosis syndromes with transient neonatal respiratory distress[5].

Various types of congenital ichthyosis present with different clinical features ranging from mild to severe affection. Additionally, these diseases have significant implication of transmission to their offspring[6].

These disorders clinically present with varied morphology of scales, xerosis, atopy, cryptorchidism, hair shaft disorders etc. Investigations like amniocentesis, chorionic villous biopsy, histopathology, electron microscopy, FISH are useful for diagnosis, however genetic analysis is essential for confirmation. Emollients, keratolytics, vitamin A & D supplementation and retinoids form the crux of treatment.

ICHTHYOSIS VULGARIS

It is the commonest inherited type of ichthyosis[7]. It has an autosomal dominant pattern of inheritance with variable phenotypic expression and penetrance,

Pathogenesis

An absence or reduction of filaggrin as well its precursor, profilaggrin is observed in the epidermis of patients on electron microscopic examination[8]. As mRNA expression is decreased, there is selective impairment of the post transcriptional control of profilaggrin synthesis or the profilaggrin gene[9,10].

Scaling results from loss of water retaining aminoacids derived from filaggrinfcatabolism leading to hyperkeratosis aptly described as retention keratosis secondary to increased adhesiveness of stratum corneum.

Clinical features

Scales in ichythosis are visible from the first few months of age. Symptoms and severity varies seasonally with improvement during the summer and when there is an increase in humidity. It is known to worsen in dry, cold environment.

Scaling in patients can be seen at the age of three months[12], with scales appearing most pronounced on the extensor aspects of extremities.

Usually, face is spared. Small, flaky partially adherent scales with turned up edges are seen. Palms and soles show accentuated skin markings.

COLLODION MEMBRANE

Introduction: "collodion baby" refers to an infant who is born encased in a tight shiny membrane at birth resembling a plastic wrap. It is one of the first cutaneous manifestation of some types of ichthyosis[12].

Pathophysiology:

Mutation in TGM1 gene is seen which is responsible for transfer of lipids to upper layers of skin during cornification[13].

Signs & Symptoms:

Over the next 2-3 weeks of birth, collodion membrane is seen to crack and peel off. Tightness of the membrane is responsible for the eyelids to out-turn to form ectropion. Eclabium occurs due to out-turning of lips due to membrane tightness

Once this membrane is fully shed, the underlying skin of the infant might display one of several ichthyosis skin types. Congenital icthyosiform erythroderma (CIE) and ARCI-lamellar ichthyosis are the common variants of ichthyosis associated with collodion membrane.

Rarely, it may be seen in Netherton syndrome, some rare forms of ichthyosis, and nearly always in harlequin ichthyosis. About 10% of infants do not show any skin involvement post shedding;- "self-healing collodion baby."

Complications:

Increased risk of infection from microorganisms, trans-epidermal fluid loss, electrolyte imbalance, unstable body temperature and pneumonia after peeling of the membrane can be seen in some infants.

HARLEQUIN FETUS

Introduction:

Characterized by defective desquamation and diffuset epidermal hyperkeratinization, is a relatively less common but severe form occuring due to mutations of a keratinocyte lipid transporter.

Clinical features:

At birth, thick plate-like scales with fissures are seen along with severe ectropion and eclabium.

In later months of life, the thick plate like covering gets shed off leaving a diffuse erythematous, scaly skin underneath which persists for life. Majority of the affected infants succumb in the perinatal period secondary to the compromised epidermis[14].

LAMELLAR ICHTHYOSIS

Introduction:

Occurs as an autosomal recessive disorder secondary to mutation in TGM1 gene. Most common clinical manifestation in infants affected by lamellar ichthyosis is a collodion membrane.

Other cutaneous manifestations include red and scaly skin , ectropion, eclabium, hair loss, palmoplantar keratoderma, nail anomalies, dehydration and respiratory issues[15].

Management:

Management is supportive largely based on presenting signs and symptoms . For infants, providing a moist environment in an incubator and preventing secondary infection is of prime importance.

Petrolatum-based emollients are advocated to keep the skin soft, supple, and hydrated. As affected children age, treatment to promote peeling and thinning of the stratum corneum is recommended.

Humidification with long baths, lubricating agents, and keratolytics such as alpha-hydroxy acid or urea based formulations are prescribed routinely. For ectropion, corneal lubrication with artificial tears is recommended. Topical or oral retinoid therapy considered with laboratory monitoring and due precautions[16].

KID SYNDROME

Introduction:

It is a rare autosomal dominant multi-system disorder, characterized by keratitis, erythrokeratoderma and sensorineural deafness.

Pathophysiology:

Occurs secondary to mutation in gap junction protein beta 2 (GJB2) located on the long arm of human chromosome 13 which encodes the structural protein 'connexin-26'(Cx26)[17] responsible for formation of gap junction channels which connects neighbouring cells and permits exchange of small molecules and ions.

Prognosis:

Early diagnosis is important for timely use of hearing aids and speech therapy. Follow-up off patients is essential as it is known to be associated with follicular occlusion triad, follicular tumors and SCCs[18].

Management

Use of emollients after bathing in the form of lotions containing alpha-hydroxy acids, ceramides are required lifelong.

PALMOPLANTAR KERATODERMAS

Introduction:

Hereditary palmoplantar keratodermas (PPK) belong to a group of disorders of keratinisation clinically manifesting as thick hyperkeratotic palms and soles occurring due to genetic mutations in keratin, desmosomes, loricrin, cathepsin C, gap junction protein genes.

Most present in infancy either as an isolated case or with abnormalities of nails, teeth, or other organs. Autosomal dominant or autosomal recessive pattern of inheritance is seen[18].

KERATOSIS PALMOPLANTARIS TRANSGREDIENS OF SIEMENS

Clinical presentation:

It is an autosomal recessive type of thick palmoplantar keratoderma with erythematous border, trangrediens, and progrediens. Association with hyperhidrosis and secondary infection leads to malodour.

PPK with honeycomb pattern:

Pachyonychia congenita (PC):

Pachyonychia congenita (PC) is a rare genodermatosis with approximately 450 cases reported till date[19].

Pathophysiology:

An autosomal dominant disorder,arising as a result of mutations in one of the four genes encoding for keratin. On the basis of the keratin mutation, it is further classified into PC6a, PC 6b, PC16, PC17, and PC unknown[20].

Clinical features:

Skin, nails, oral mucosa, and teeth are affected in association with characteristic plantar keratoderma and thickened toenails

Management:

Daily moisturization is the mainstay of therapy in combination with prophylactic antibacterial and antifungals topically.

Molecular studies are expensive but essential for accurate classification, diagnosis, genetic counselling, and treatment[21].

Vohwinkel syndrome:

The classical honey comb appearance with keratotic plaques in the shape of stair fish is seen over the knuckles, wrists, elbows, and knees. A mutation in GJB2 gene encoding connexin[22] with autosomal dominant pattern of inheritance occurs.

Papillon Lefèvre syndrome (PLS):

Inherited as an autosomal recessive condition which manifests as diffuse symmetrical keratoderma with trans-gradiens, severe periodontitis at an early age causing primary and permanent dentitions to be lost along with increase in susceptibility to skin and systemic infections[23].

NEUROFIBROMATOSIS

Neurofibromatosis (NF) constitutes a group of inherited disorders that lead to the formation of neural tumours and other distinctive features.

The two main types are:

1. Type -1 NF (85%)

2. Type-2 NF (10%)

NEUROFIBROMATOSIS - 1

Definition

Neurofibromatoses-1 is an autosomal dominant condition, presenting with multiple cafe-au-lait macules, axillary and palmar freckling, neurofibromas along with Lisch nodules in the iris. Neurofibromatoses -2 is presents with bilateral acoustic neuromas , menigiomas and other CNS tumors.

Etiology

The gene for NF1 is located on chromosome 17 which encodes for neurofibromin expressed on neuronal cells, schwann cells, oligodendrocytes, melanocytes and leucocytes. Neurofibromin is a protein which is shown to be capable of downregulating the Ras activity.

CLINICAL FEATURES

Diagnosis of Neurofibromatosis requires two or more of the following[24].

1. Presence of Six or more cafe-au-lait macules > 5mm in diameter in prepubertal age and > 15mm in post pubertal age.

2. Two or more neurofibromas or one plexiform NF.

3. Axillary or inguinal freckling.

4. Optic glioma

5. Two or more Lisch nodules

6. sphenoid dysplasia or thinning of long bone cortex with or without pseudoarthrosis

7. A first degree relative with NF-1 by the above criteria

TUBEROUS SCLEROSIS

Definition

Tuberous sclerosis complexk(TSC) also known as EPILOIA is inherited as an autosomal dominant condition clinically manifesting as hamartomas of the skin, brain, heart, eyes and kidneys.

EPIDERMOLYSIS BULLOSA (EB)

Introduction:

EB belongs to a heterogeneous group of inherited disorders of blistering of the skin and mucosae secondary to mechanical trauma.

Pathophysiology:

Defect in the genes encoding for proteins like collagen and keratin, which mediates the adhesion of the epidermis with the underlying dermis is seen[25].

Classification is based depending upon the level of split at the basement membrane (BM) zone

EB simplex- intraepidermal split- AD .

Junctional- at the level of lamina lucida.- AR.

Dystrophic- sublamina densa-AD/AR

Kindler syndrome- mixed cleavage-AD/AR

Clinical features:

Table 2: Clinical features Epidermolysis bullosa[26]

Eb simplex	Localized blisters, limited mucosal involvement, palmoplantar hyperkeratosis, nail dystrophy
Dystrophic Eb	Haemorrhagic blisters,nscarring, milia formation, pseudosyndactyly, severe mucosal involvement, significant morbidity.
Junctional Eb	Widespread blistering, scarring, significant granulation tissue, severe mucosal involvement, dental pitting, alopecia, nail dystrophy
Kindler syndrome	acral skin blistering, photosensitivity, progressive poikiloderma, and diffusen cutaneous atrophy. Involvement of the oral mucosa, gingiva.

DARIERS DISEASE / KERATOSIS FOLLICULARIS

Introduction:

It is an autosomal dominant genodermatoses showing greasy hyperkeratotic papules in seborrheic areas, nail abnormalities and mucosal changes.

Clinical features:

Greasy papules, wartyf plaques seen predominantly in seborrheic distribution along with hypertrophic and malodorous and painful fissures in flexures. Cobble stone appearance can be seen over palatal and alveolar mucosa in oral cavity.

Fragility of nails, red and white longitudinal streaks and V-shaped notches at the free margin are characteristic. Sun, heatfand sweating cause exacerbation of the disease[27].

XERODERMA PIGMENTOSUM

Introduction:

Xeroderma pigmentosum (XP) is an autosoml recessive disorder of DNA repair . It is characterized by cutaneous and ocular photosensitivity. In patients of XP, there is an increased potential of neoplasms like basal cell carcinoma, squamous cell carcinoma and melanoma[28].

Pathophysiology:

The basic defect arises as a result of a mutation leading to an absence or deficiency of nucleotide excision repair (NER) enzymes important for the repair of

deoxyribonucleic acid damaged by ultraviolet radiation, resulting in the development of various cutaneous malignancies.

XP patients below 20 years of age have more than 1000-fold risk of developing cutaneous malignancies. Mutations occuring in any of the nine corresponding genes of the nine complementation groups i.e XPA, ERCC3 (XPB), XPC, ERCC2, DDB2, ERCC4, ERCC5, ERCC1 and POLH1 may lead to XP.

Management:

Patients are advised to remain indoors or cover themselves in case one has to go outdoors so as to avoid exposure to sunlight.

Prognosis:

Timely identification is essential for proper counselling of the patient and parents and to advise regarding protective measures to be taken in order to avoid cutaneous malignancies.

Simultaneous screening of the siblings for early detection and formation of XP groups to spread awareness among the general population are some of the steps that need to be undertaken[30].

LIPOID PROTEINOSIS/URBACH WIETHE DISEASE

Introduction:

Lipoid proteinosis is a rare progressive autosomal recessive disease characterized by deposition of hyaline in the skin, upper aero-digestive tract, and internal organs.

Clinical features:

Infants present with a shrill cry at birth and hoarseness of voice due to laryngeal infiltration, followed by blistering leading to pox like and acneiform scars, thickening of the skin, and certain mucous membranes. Moniliform blepharosis associated with waxy, yellow papules and nodules over lid margins are a classic presentation.

Hyperkeratosis or verrucous changes may appear over frictional sites. Extracutaneous features may include epilepsy and neuropsychiatric abnormalities.

Management:

As there is a lack of definitive therapy, the following have been tried with variable success.

Topical and oral corticosteroids

Oral dimethyl sulfoxide.

D-penicillamine

laser/dermabrasion of papules.

Acitretin for voice hoarseness

Quality of life (QOL)

The extent to which the quality of life (QOL) is impacted in a person varies depending on the individual; natural history of the disease; the subject's demographics; personality and character; traditional values and habits; the subject's life situation; and the people surrounding them and their attitudes in the society.

Every dermatologist uses his/her perception regarding how a patient’s condition affects his/her life in order to assist him/her so as to decide upon therapy in clinical practice.

Given that the dermatologists may not be able to accurately estimate this parameter, it would be of immense help to have a formal measure for QOL impairment in this situation.

Dermatology Life quality Index (DLQI)

The main dermatology specific quality of life tool, DQLT, was developed in 1994. Since then it has been used in over 80 countries, and is available in over 100 languages. The purpose of the Dermatology Life Quality of Index questionnaire is that it acts as a template for assessing the impact of skin disease on patients' lives.

This questionnaire being self explanatory, makes it possible to be administered to the respondents without providing a detailed explanation.

The DLQI questionnaire is consists of 10 questions, each one of them having four possible answers which is given a score of 0 (not at all) to 3 (very much).
The DLQI score can range between 0 and 30; the higher the score meaning that the greater is the impairment of quality of life.

DLQI score Interpretation

0-1 —No effect at all

2-5 —Small effect

6-10 —Moderate effect

11-20 —Very large effect

21-30 —Extremely large effect

MATERIAL AND METHODS

Study location:

Department of Dermatology, Venereology and leprosy at a tertiary care hospital

Study duration:

One year January 2021 to December 2021

Study design:

Cross sectional study

Sample size:

55 patients were enrolled in the study

Sample size was calculated as per the formula

$$n = \frac{p(100-p)Z^2}{E^2}$$

Where ***n*** is the sample size required, **p** is the percentage occurrence of a state or condition (proportion or prevalence), **E** is the percentage maximum error required, **Z** is the value corresponding to level of confidence required.

With the assumption that 15% of the subjects would have low quality of life and percentage of maximum error as 10% at 95% confidence level, the sample size was obtained as follows,

$$n = \frac{15 \times 85 \times (1.96)^2}{10^2}$$

$$n = 48.97 \approx 49$$

Minimum sample size required was 49

Purposive sampling method was employed for recruiting patients.

Study population:

This study was conducted on patients of genodermatoses in the age group 16-60 years, irrespective of their sex, attending the out patient department of dermatology at KLE's DR. PRABHAKAR KORE HOSPITAL.

The patient was sensitised about the subject of the study and informed regarding the personal nature of the questionnaire. All those who gave informed written consent were given the DLQI questionnaire (English, Hindi, Marathi, Kannada) to fill.

The DLQI was computed by adding the scores of each question which resulted in a maximum score of 30 and a minimum of 0.

Higher the score, the more quality of life was impaired. Responses to DLQI was recorded and scoring was done in accordance to the guidelines of Finlay and Khan.

Inclusion criteria:

Subjects of either sex of age group 16-60 years, with a diagnosis of genodermatoses as confirmed by a dermatologist.

Subjects willing to give informed consent.

Exclusion criteria:

Children age <16 years

Illiterate patients

Data analysis:

Data was analyzed using SPSS v21. Categorical data was represented as frequencies and percentages. Continuous data was represented as mean and standard deviation. Chi square test was used as test of significance for categorical data. ANOVA teat was used as test of significance for continuous data(more than 2 groups). P value less than 0.05 was considered as statistically significant. Bar charts and pie charts were used for pictorial representation of data.

RESULTS AND OBSERVATIONS

A total of 55 patients of genodermatoses presenting to our dermatology department were enrolled over a period of one year from 1st January to 31st December 2021

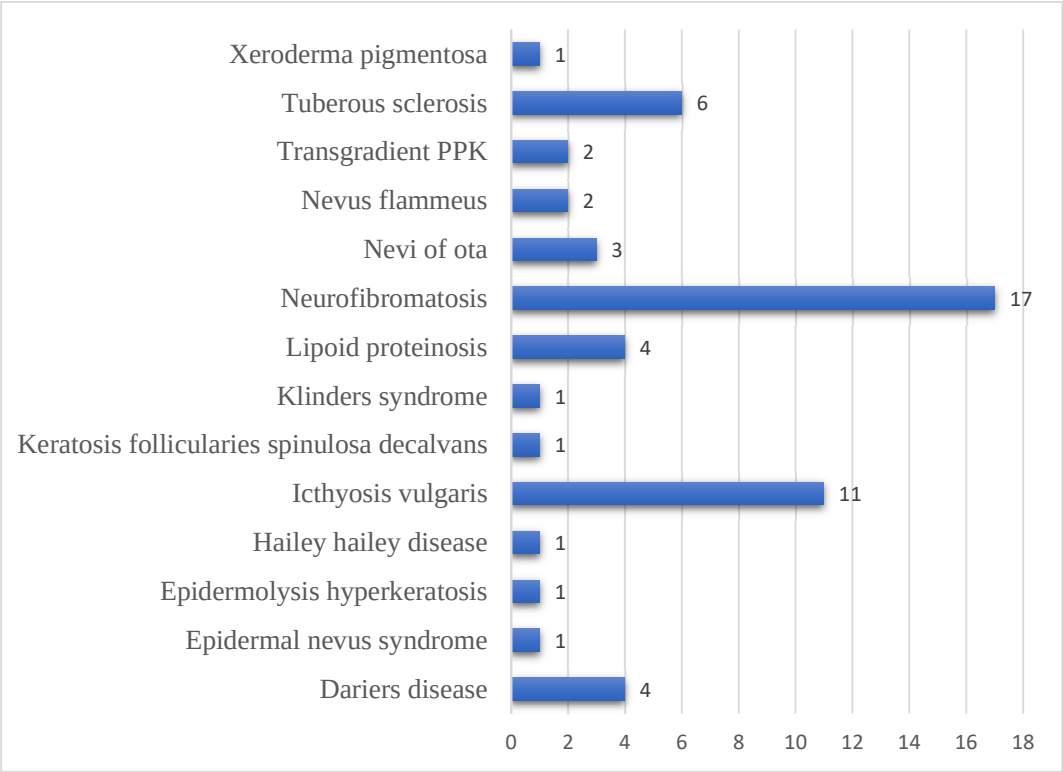
1. Genodermatoses distribution

Out of 55 cases included in the study, 17 (30.9%) were of neurofibromatosis, 11 (20%) were of icthyosis vulgaris, 6 (10.9%) had tuberous sclerosis, 4 (7.3%) were of Dariers disease and lipoid proteinosis each, 3 (5.5%) were of nevus of Ota, 2 (3.6%) of nevus flammeus and transgradient PPK and 1 (1.8%) of epidermal nevus syndrome, epidermolysis hyperkeratosis, Hailey Hailey disease, Keratosis follicularis spinulosa decalvans, Kindlers syndrome and xeroderma pigmentosa each.

Table 1 - Genodermatoses distribution

Genodermatoses	Frequency	Percent
Darier's disease	4	7.3
Epidermal nevus syndrome	1	1.8
Epidermolysis hyperkeratosis	1	1.8
Hailey hailey disease	1	1.8
Icthyosis vulgaris	11	20.0
Keratosis follicularies spinulosa decalvans	1	1.8
Klinders syndrome	1	1.8
Lipoid proteinosis	4	7.3
Neurofibromatosis	17	30.9
Nevi of ota	3	5.5
Nevus flammeus	2	3.6
Transgradient PPK	2	3.6
Tuberous sclerosis	6	10.9
Xeroderma pigmentosa	1	1.8
Total	55	100.0

Chart 1: Genodermatoses distribution



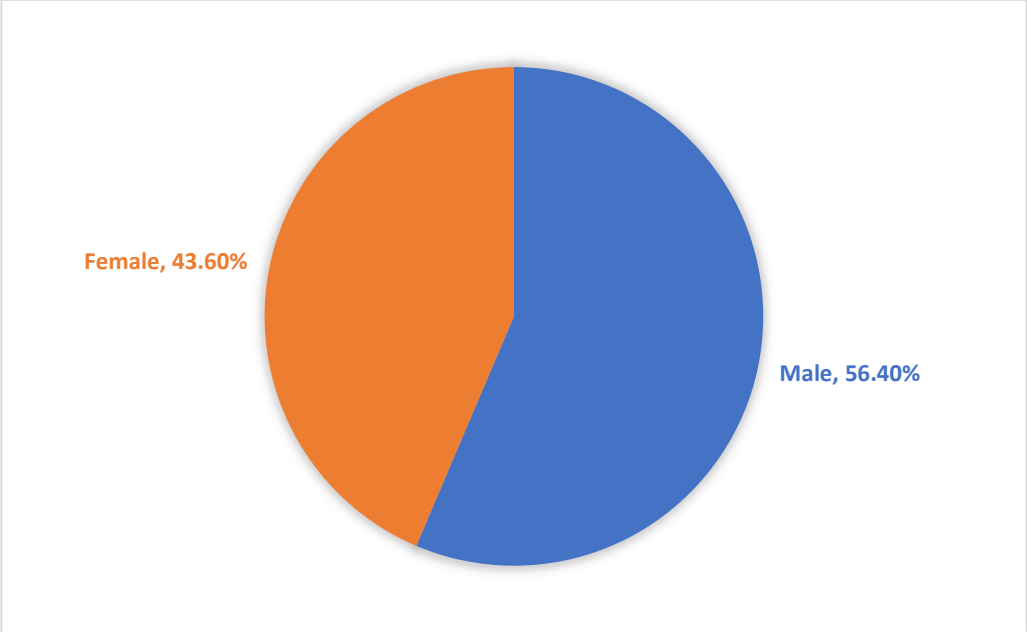
2. Gender distribution

Out of 55 cases 31 (56.4%) were male and 24 (43.6%) were female, indicating a male predominance.

Table 2 - Gender distribution

Gender	Frequency	Percent
Male	31	56.4
Female	24	43.6
Total	55	100.0

Chart 2: Gender distribution



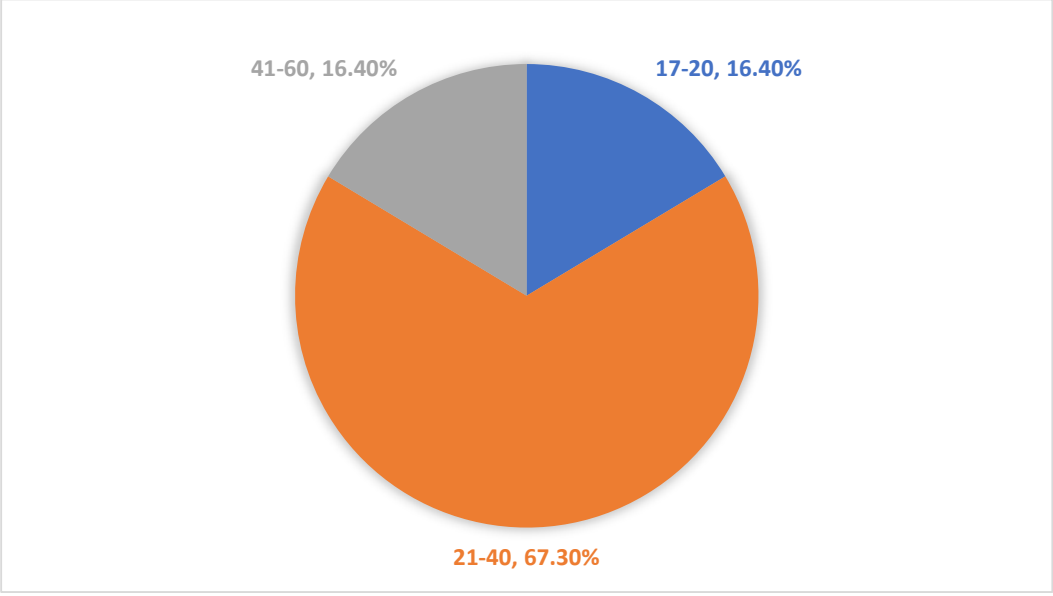
3. Age distribution

Most patients belonged to the age group of 21-40 years which constituted 37 (67.3%) cases. Cases in 17-20 years age group were 9 (16.4%), 41-60 years age group were 9 (16.4%). The average age of patients enrolled in this study was 29.09 years.

Table 3: Age distribution

Age group	Frequency	Percent
17-20 years	9	16.4
21-40 years	37	67.3
41-60 years	9	16.4
Total	55	100.0

Chart 3: Age distribution



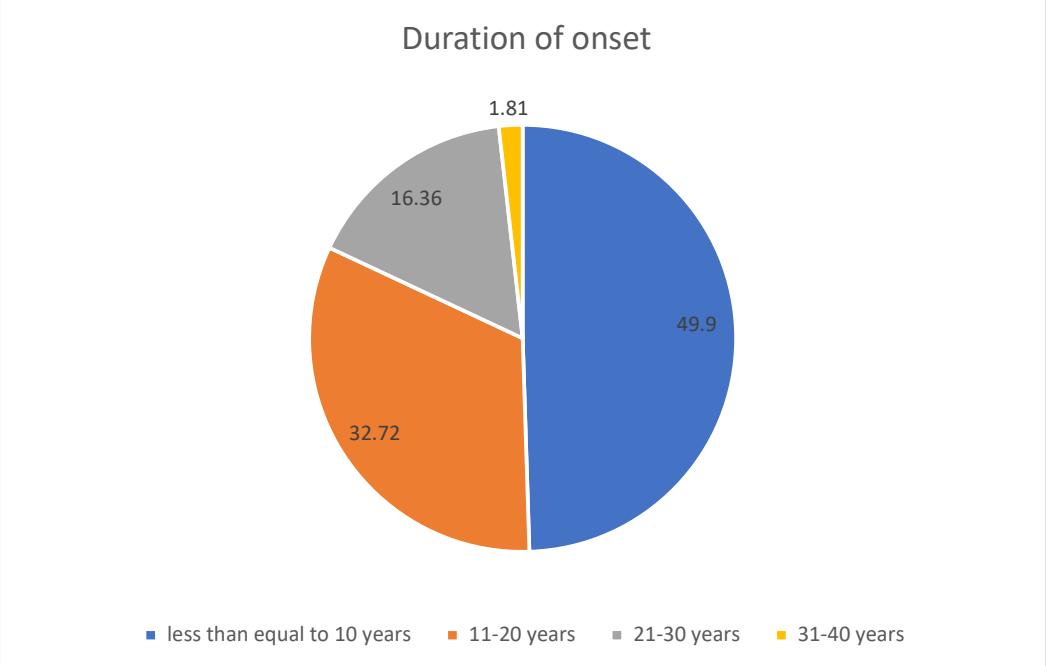
4. Duration of onset

The duration of onset of skin manifestations in the cases studied ranged from one year to 35 years. Mean duration of disease onset was 11.03 years. Most patients in the study i.e. 27 (49.09%) had disease onset of less than 10 years, followed by 18 (32.72%) patients who had disease onset between 11-20 years.

Table 4: Duration of disease onset

Duration of onset	Frequency	Percent
<=10 years	27	49.09
11-20 years	18	32.72
21-30 years	9	16.36
31-40 years	1	1.81

Chart 4: Duration of onset



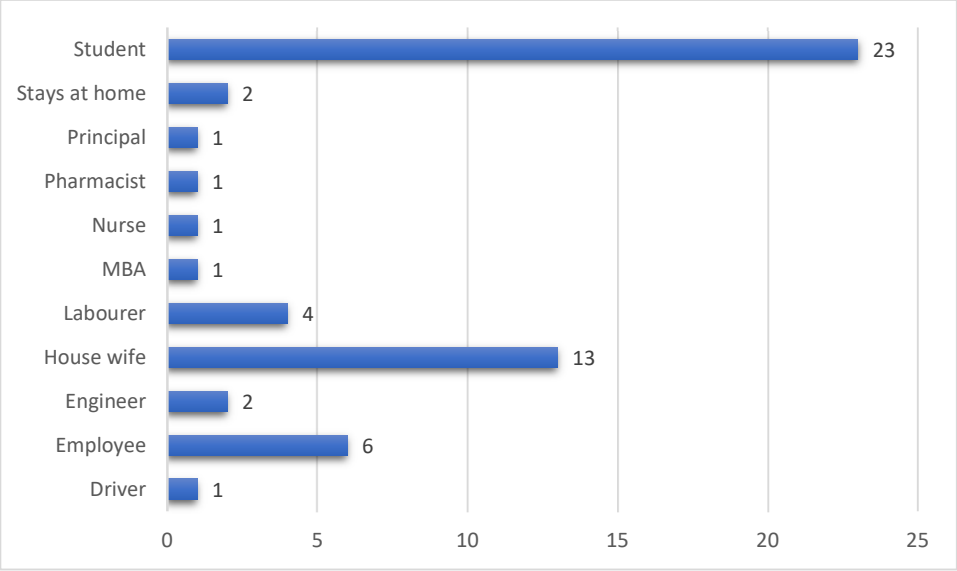
5. Occupation

23/55 (41.8%) cases were students followed by 13/55 (23.6%) who were house wives, 6/55 (10.9%) were employees, 4/55 (7.3%) were laborers and 2/55 (3.6%) were engineers and non-working.

Table 5: Occupation of the patients

Occupation	Frequency	Percent
Driver	1	1.8
Employee	6	10.9
Engineer	2	3.6
House wife	13	23.6
Labourer	4	7.3
MBA	1	1.8
Nurse	1	1.8
Pharmacist	1	1.8
Principal	1	1.8
stays at home / non-working	2	3.6
Student	23	41.8
Total	55	100.0

Chart 5: Occupation of the lesions



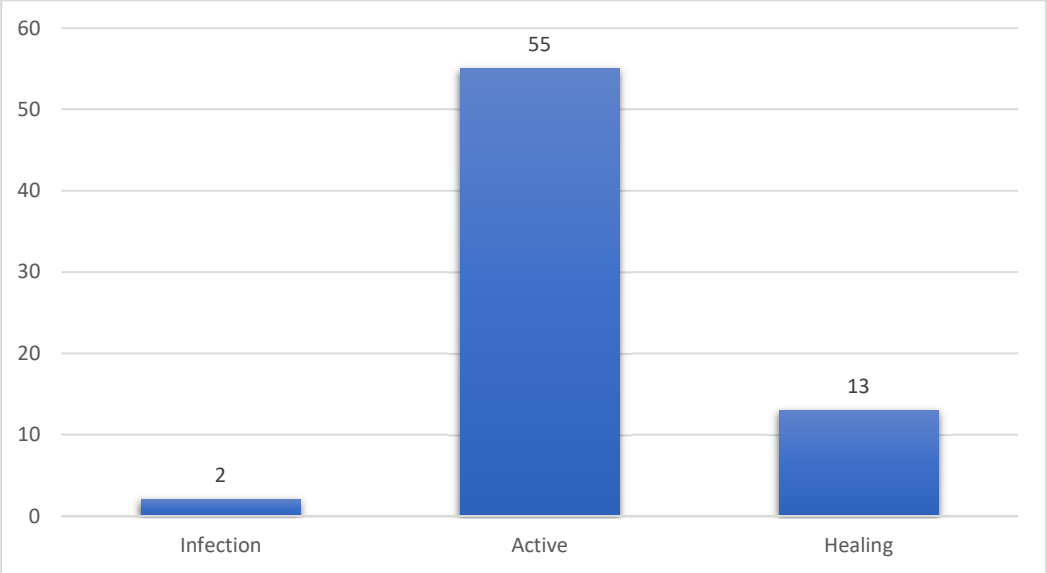
6. Status of lesions

All 55 cases had active lesions in context to the underlying genodermatoses followed by 13/55 (23.63%) cases who had both active as well as healing lesions, 2/55 (3.63%) cases had infected and active lesions.

Table 6: Status of lesions

Conditions of the lesion	Frequency	Percent
Infection	2	3.63
Active	55	100.0
Healing	13	23.63

Chart 6: Status of the lesion



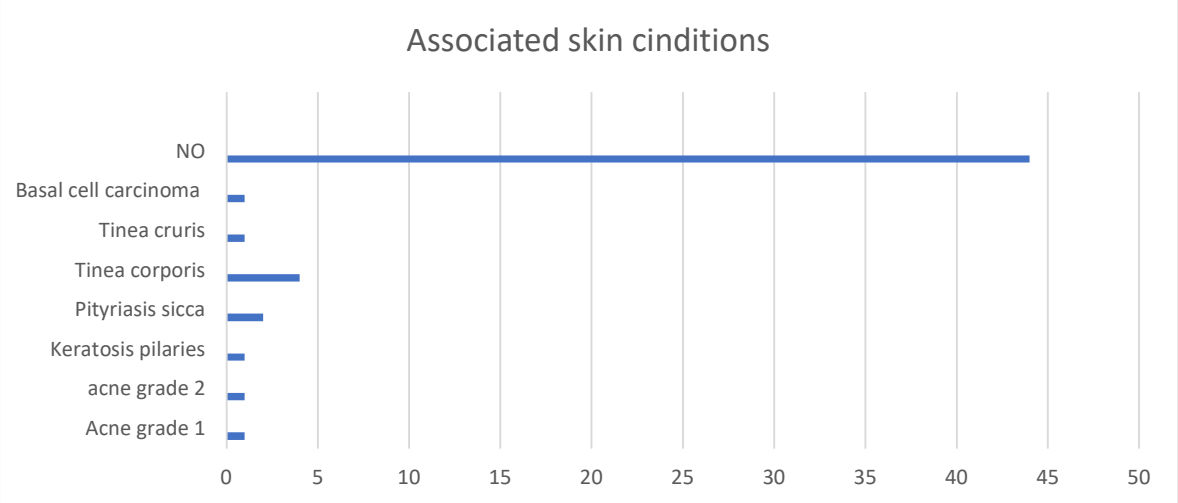
7. Other associated skin conditions

45/55 (81.8%) cases had no associated skin conditions followed by 4/55 (7.3%) cases who had tinea corporis and 2 (3.6%) cases presented with pityriasis sicca.

Table 7: Associated skin conditions

Associated skin lesions	Frequency	Percent
Acne	2	3.6
Keratosis pilaries	1	1.8
Pityriasis sicca	2	3.6
Tinea corporis	4	7.3
Tinea cruris	1	1.8
Basal cell carcinoma	1	1.8
No	44	80
Total	55	100.0

Chart 7: Other associated skin conditions



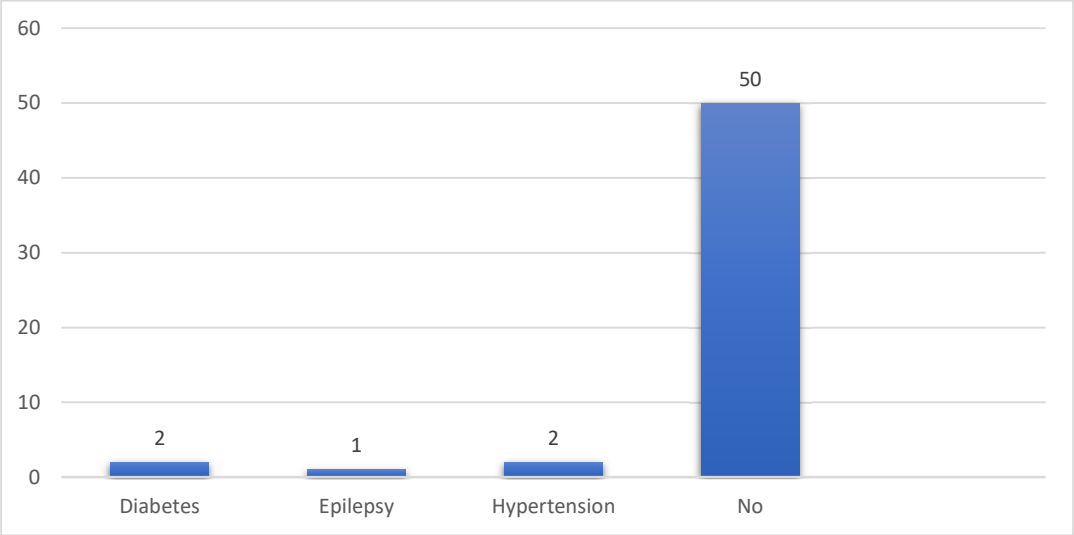
8. Associated systemic disorders

Associated systemic disorders like diabetes and hypertension seen in 2 (3.6%) cases each, 1 (1.8%) case had epilepsy

Table 8: Associated systemic disorders

Associated systemic disorders	Frequency	Percent
Diabetes	2	3.6
Epilepsy	1	1.8
Hypertension	2	3.6
No	50	90.9
Total	55	100.0

Chart 8: Associated systemic disorders



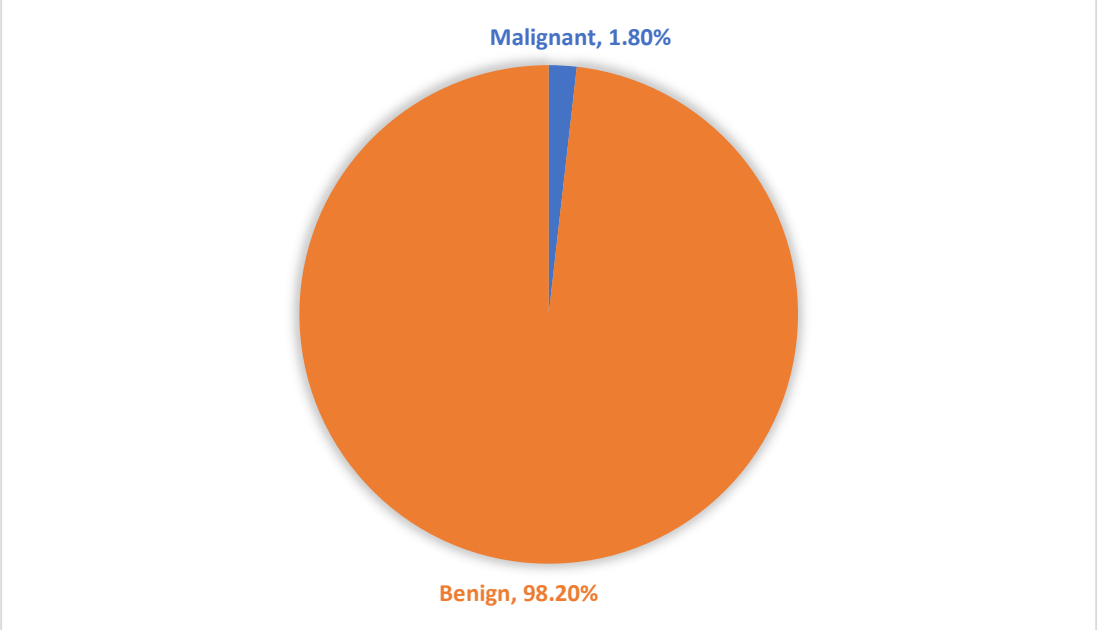
9. Malignant and benign lesions

Only 1 (1.8%) case out of 55 cases had a malignant skin lesion.

Table 9: Malignant and benign lesions

	Frequency	Percent
Malignant	1	1.8
Benign	54	98.2
Total	55	100.0

Chart 9: Malignant and Benign lesions



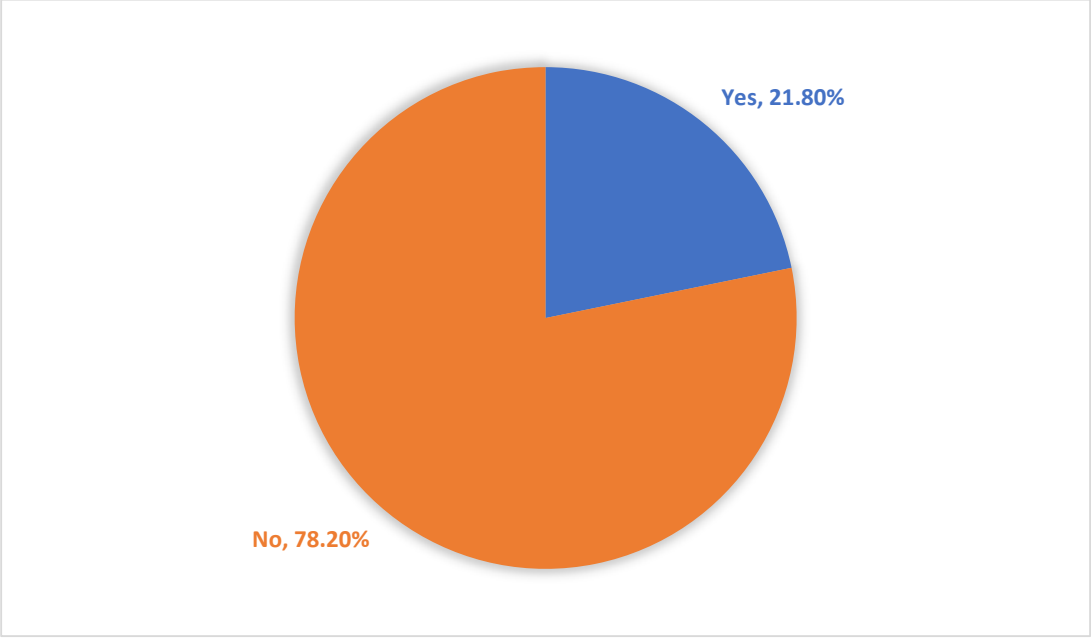
10. Family history

Out of 55 cases that are included in the study 12 (21.8%) had a positive family history and 43/55 (78.2%) cases had no family history.

Table 10: Family history

Family history	Frequency	Percent
Yes	12	21.8
No	43	78.2
Total	55	100.0

Chart 10: Family history



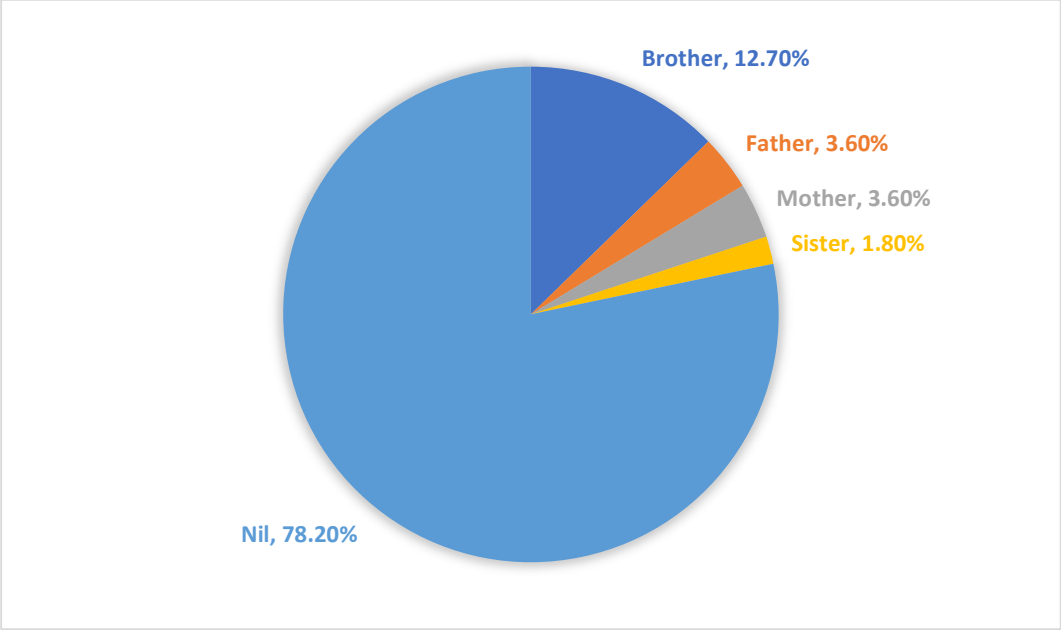
11. Genodermatoses in relations

Out of 12 (21.8%) positive family history cases, 7 (12.7%) cases had family history in brother, 2 (3.6%) cases in father and mother.

Table 11: Genodermatoses in relations

Genodermatoses in relations	Frequency	Percent
Brother	7	12.7
Father	2	3.6
Mother	2	3.6
Sister	1	1.8
Nil	43	78.2
Total	55	100.0

Chart 11: Genodermatoses in relations



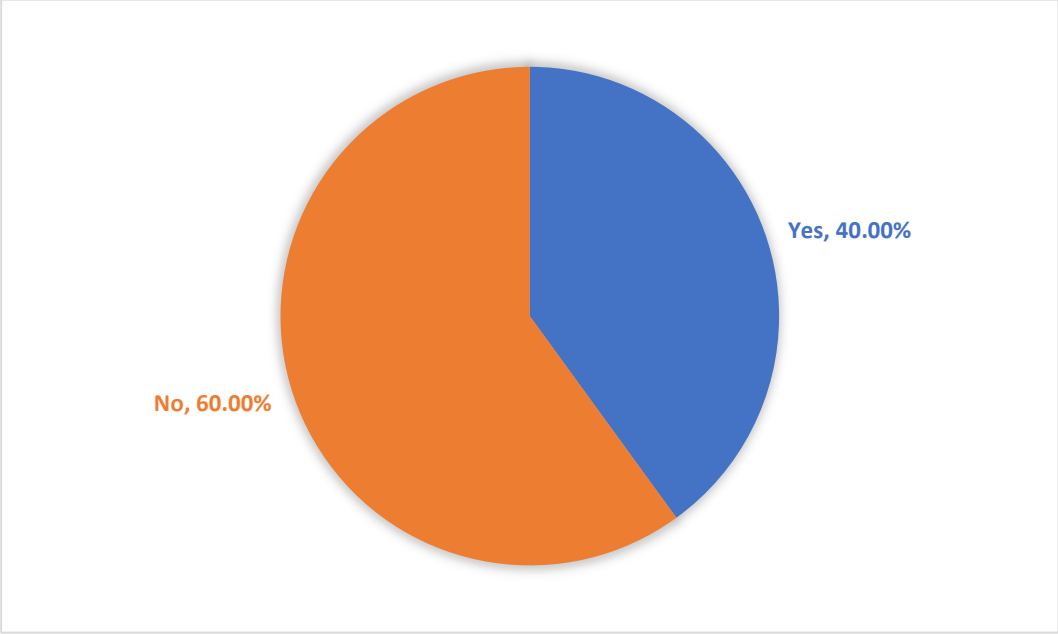
12. Marital history

Out of 55 cases, 33 (60%) cases are unmarried and 22 (40%) cases are married.

Table 12: Marital history

Marital history	Frequency	Percent
Married	22	40.0
Unmarried	33	60.0
Total	55	100.0

Chart 12: Marital history



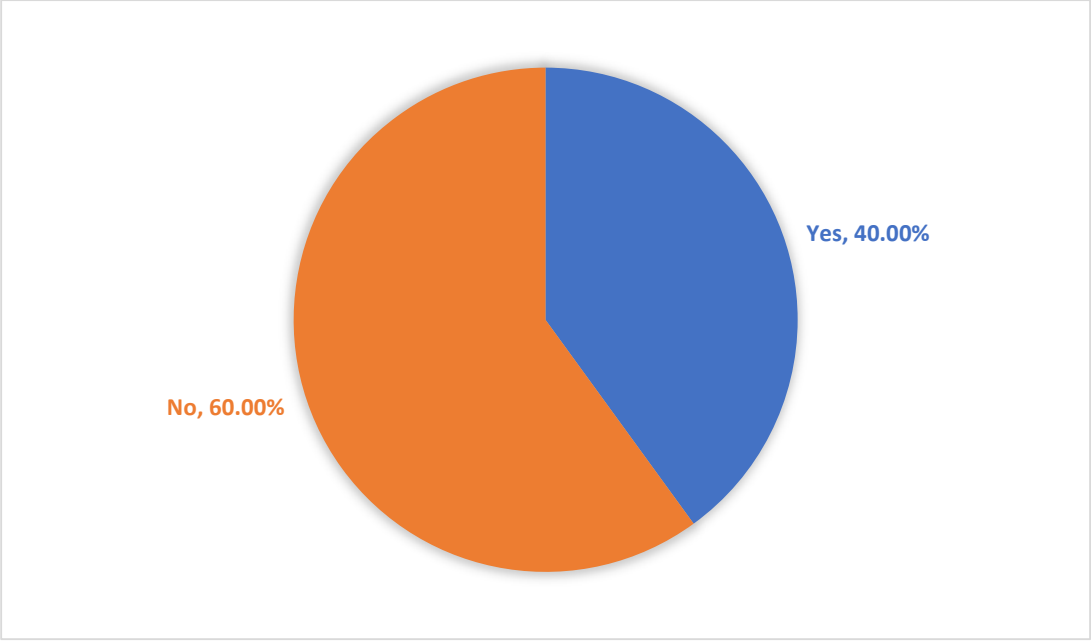
13. Consanguinity in parents

In this study history of consanguineous marriage in parents was present in 22 (40%) cases out of 55 cases.

Table 13: Consanguinity in parents

Consanguinity in parents	Frequency	Percent
Yes	22	40.0
No	33	60.0
Total	55	100.0

Chart 13: Consanguinity in parents



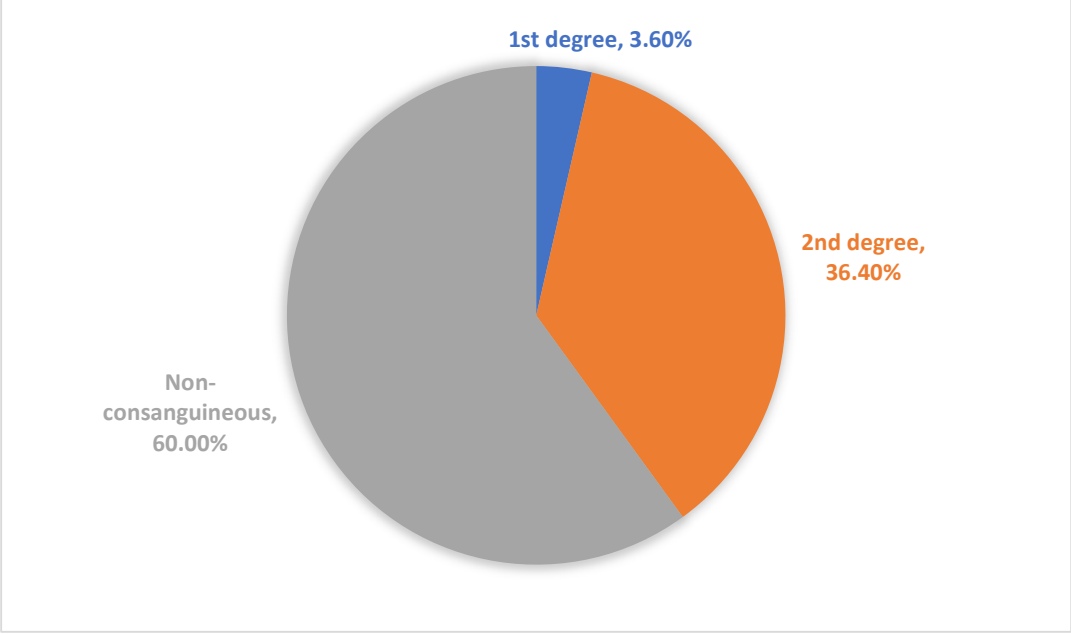
14. Degree of consanguineous marriage in parents

20 (36.4%) cases had a history of 2nd degree consanguineous marriage in parent and 2 (3.6%) cases had 1st degree consanguineous marriage in parents.

Table 14: Degree of consanguineous marriage in parents

Degree of consanguineous	Frequency	Percent
1st degree	2	3.6
2nd degree	20	36.4
Non-consanguineous	33	60.0
Total	55	100.0

Chart 14: Degree of consanguineous marriage in parents



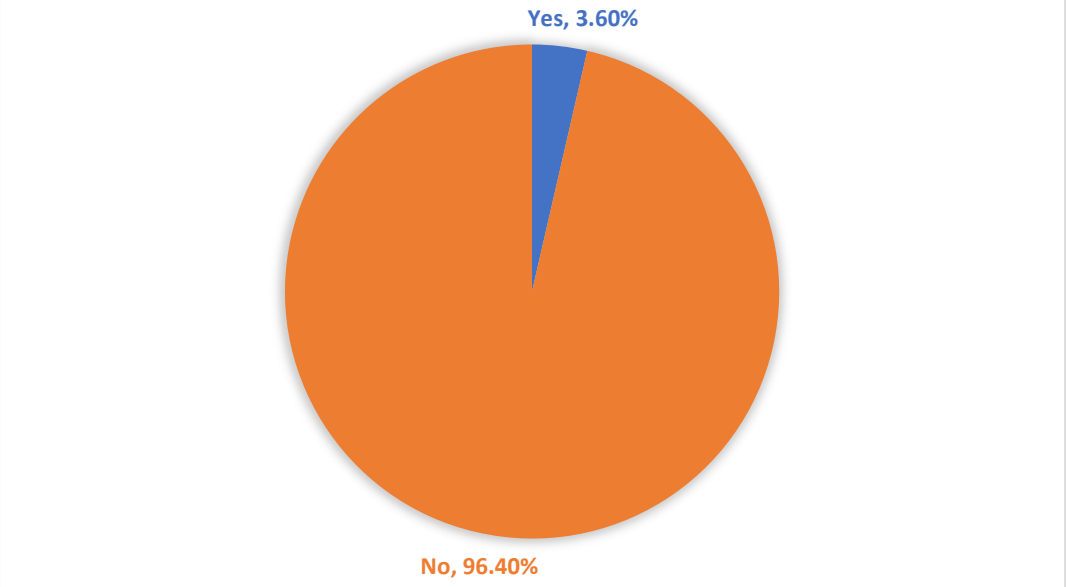
15. Psychiatric visit

Out of 55 cases that are included in the study 2 (3.6%) cases had visited a psychiatrist for counselling in context to their underlying skin conditions.

Table 15: Psychiatric visit

Psychiatric visit	Frequency	Percent
Yes	2	3.6
No	53	96.4
Total	55	100.0

Chart 15: Psychiatric visit



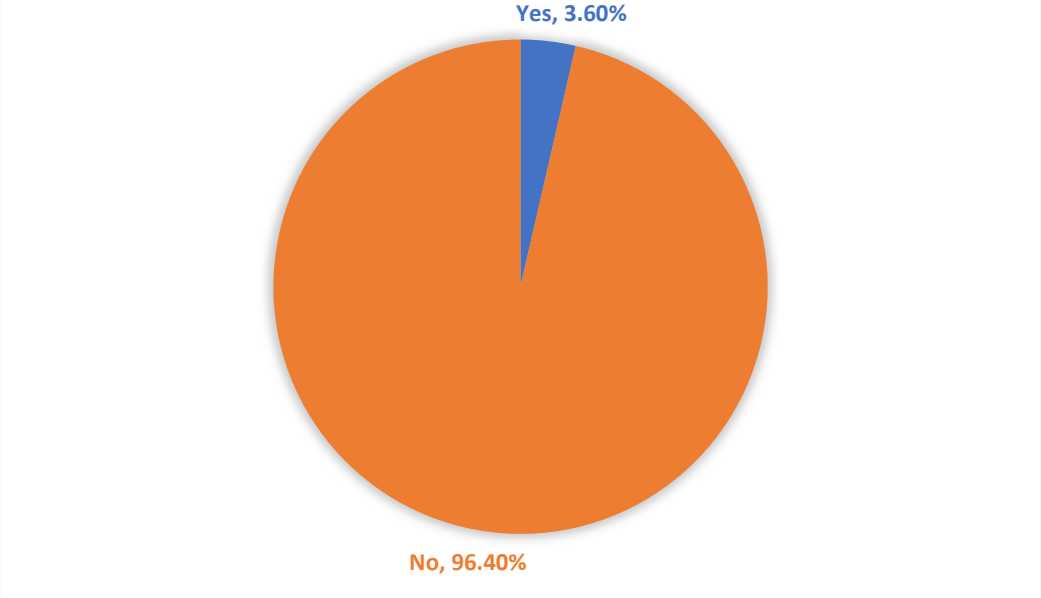
16. Counselling regarding the disease

Counselling regarding the disease done only in 2 (3.6%) cases out of 55 cases i.e. in xeroderma pigmentosum and nevus flammeus cases.

Table 16: Counselling regarding the disease

Counselling regarding the disease	Frequency	Percent
Yes	2	3.6
No	53	96.4
Total	55	100.0

Chart 16: Counselling regarding the disease



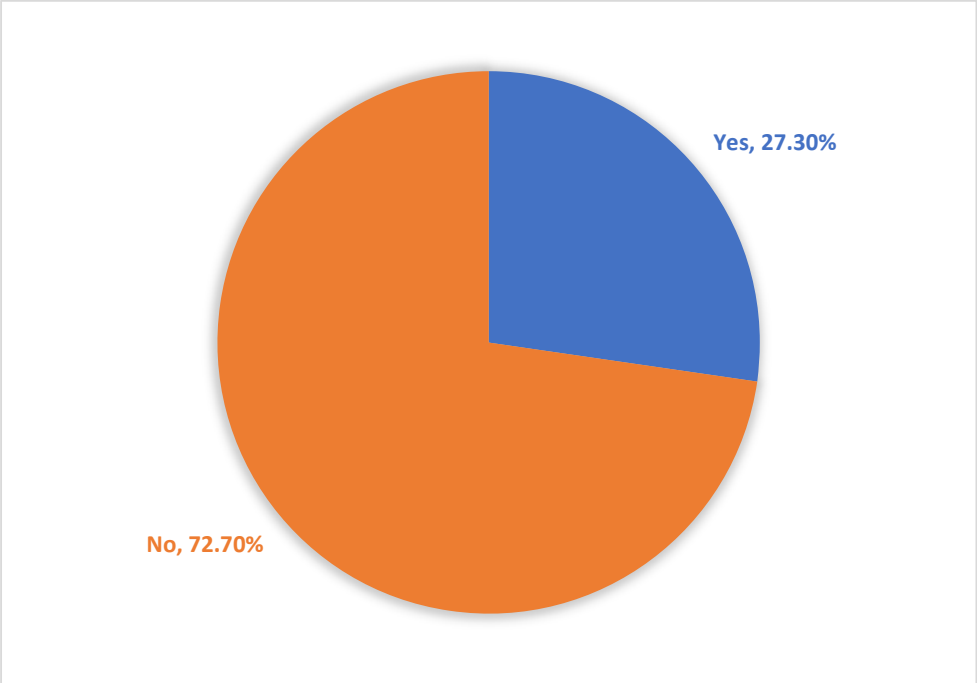
17. On treatment for disease

Out of 55 cases that are included in the study 40 (72.7%) cases were not on any treatment for the underlying disease and 15 (27.3%) cases were seeking treatment.

Table 17: On treatment for disease

On treatment	Frequency	Percent
Yes	15	27.3
No	40	72.7
Total	55	100.0

Chart 17: On treatment for disease



18. DLQI Grading

Out of 55 cases that are included in the study 40 (72.7%) cases had very large DLQI grading followed by moderate DLQI grading in 10 (18.2%) cases. 3 (5.5%) cases had extremely large DLQI grading and 2 (3.6%) had small DLQI grading.

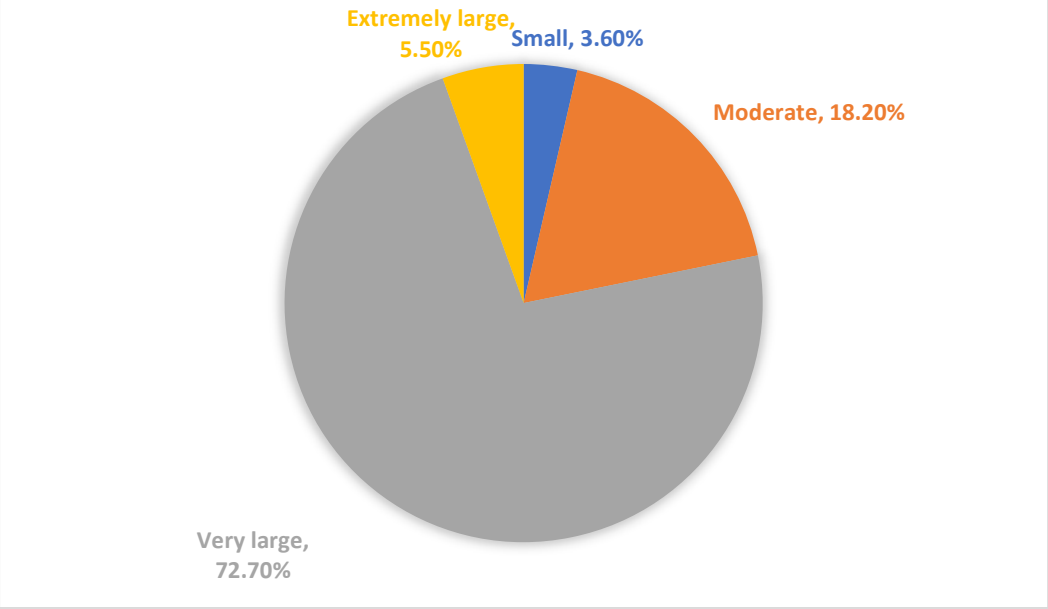
Table 18a: Interpretation of DLQI scores

DLQI score	Interpretation
0-1	No effect
2-5	Small effect
6-10	Moderate effect
11-20	Very large effect
21-30	Extremely large effect

Table 18b: DLQI grading

DLQI Grading	Frequency	Percent
Small	2	3.6
Moderate	10	18.2
Very large	40	72.7
Extremely large	3	5.5
Total	55	100.0

Chart 18: DLQI Grading



19. Association between age group and DLQI grading

Out of 40 cases with very large DLQI grading 31 (77.5%) belonged to the age group of 21-40 years, 8 (20%) belonged to the age group of 17-20 years.

Moderate DLQI grading seen in 10 (18.2%) out of 55 cases. In which 6 (60%) belonged to the age group between 41-60 years, followed by 4 (40%) in age group between 21-40 years.

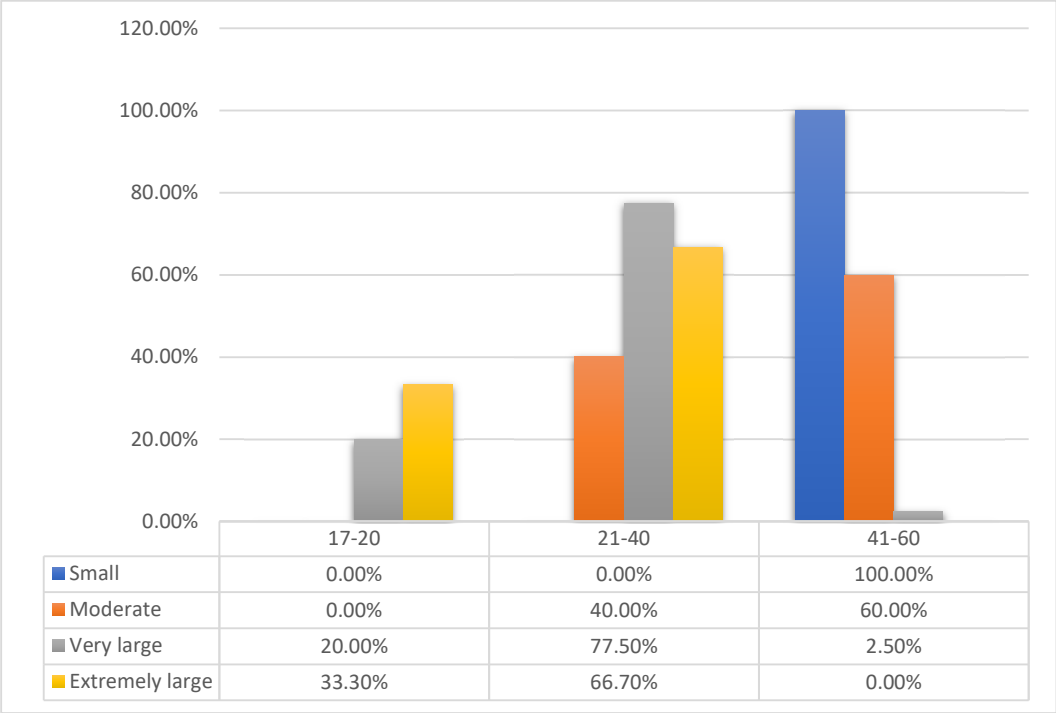
3 (5.5%) out of 55 cases had extremely large DLQI grading in which 2 (66.7%) were in the age group between 21-40 years followed by 1 (33.3%) in age group between 17-20 years.

Table 19: Association of age group to DLQI grading

			DLQI				Total
			Small	**Moderate**	**Very large**	**Extremely large**	
AGE GROUP	**17-20**	Count	0	0	8	1	9
		%	0.0%	0.0%	20.0%	33.3%	16.4%
	21-40	Count	0	4	31	2	37
		%	0.0%	40.0%	77.5%	66.7%	67.3%
	41-60	Count	2	6	1	0	9
		%	100.0%	60.0%	2.5%	0.0%	16.4%
Total		Count	2	10	40	3	55
		%	100.0%	100.0%	100.0%	100.0%	100.0%

CHI SQUARE = 31.263, P VALUE = 0.001 (S)

Chart 19: Association of age group to DLQI grading



	17-20	21-40	41-60
Small	0.00%	0.00%	100.00%
Moderate	0.00%	40.00%	60.00%
Very large	20.00%	77.50%	2.50%
Extremely large	33.30%	66.70%	0.00%

20. Association of gender with DLQI grading

In this study 31 cases were male out of which 21 (67.74%) cases had very large DLQI grading followed by moderate effect in 6 (19.35%) cases, very large DLQI grading noted in 3 (9.67%) cases.

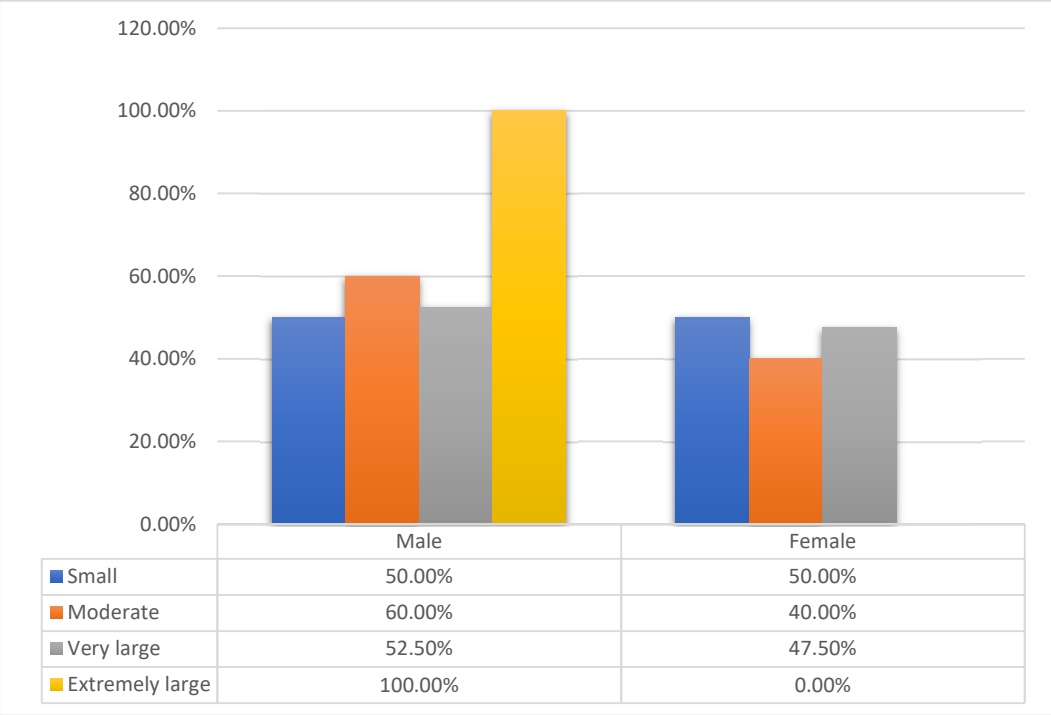
Out of 24 females that were included in the study 19 (79.1%) cases had very large effect according to DLQI grading followed by moderate effect in 4 (16.66%) cases.

Table 20: Association of gender with DLQI grading

			DLQI				Total
			Small	**Moderate**	**Very large**	**Extremely large**	
Gender	**Male**	Count	1	6	21	3	31
		%	50.0%	60.0%	52.5%	100.0%	56.4%
	Female	Count	1	4	19	0	24
		%	50.0%	40.0%	47.5%	0.0%	43.6%
Total		Count	2	10	40	3	55
		%	100.0 %	100.0%	100.0%	100.0%	100.0%

CHI SQUARE = 2.652, P VALUE = 0.448 (NS)

Chart 20: Association of gender with DLQI grading



	Male	Female
Small	50.00%	50.00%
Moderate	60.00%	40.00%
Very large	52.50%	47.50%
Extremely large	100.00%	0.00%

21. Association of malignant and benign lesions with DLQI grading

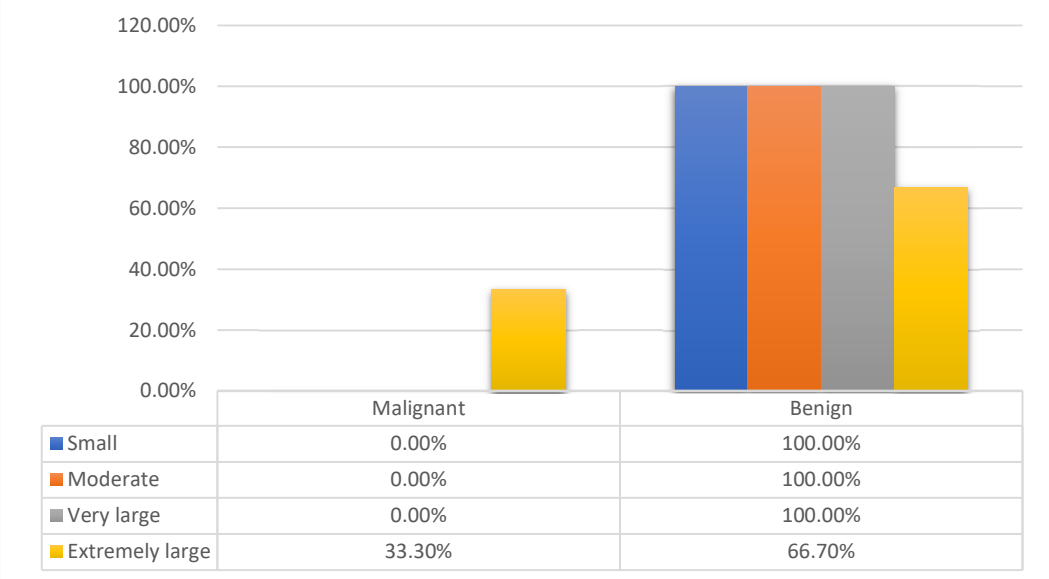
Out of 55 cases, 54 (88.2%) cases had benign lesions and 1 (1.8%) case had malignant lesion with a extremely large DLQI grading.

Table 21: Association of malignant and benign lesions to DLQI grading

			DLQI				Total
			Small	**Moderate**	**Very large**	**Extremely large**	
Malignant or Benign lesions	**Malignant**	Count	0	0	0	1	1
		%	0.0%	0.0%	0.0%	33.3%	1.8%
	Benign	Count	2	10	40	2	54
		%	100.0 %	100.0%	100.0%	66.7%	98.2%
Total		Count	2	10	40	3	55
		%	100.0 %	100.0%	100.0%	100.0%	100.0%

CHI SQUARE = 17.654, P VALUE = 0.001 (S)

Chart 21: Association of malignant and benign lesions to DLQI grading



22. Association of Marital history with DLQI grading

33 (60%) cases out of 55 were un-married out of which 29 (87.87%) had a very large DLQI grading, followed by extremely large DLQI grading in 3 (9.09%) cases.

In this study 22 (40%) cases were married out of 55 cases. Very large DLQI grading seen in 11 (50%) cases and moderate effect seen in 9 (40.90%) cases.

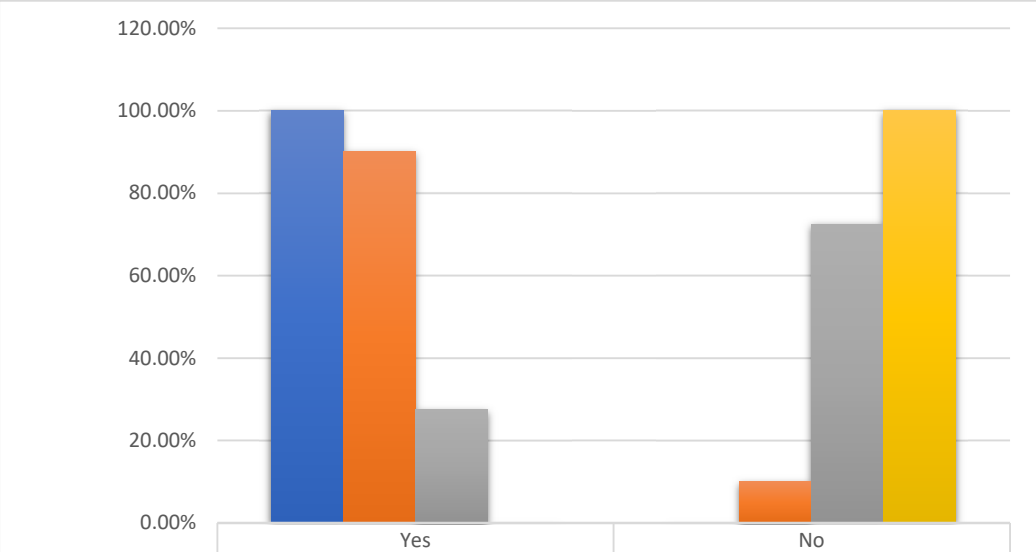
In this study there is a very large effect on the un-married population than a married population.

Table 22: Association of Marital history to DLQI grading

			DLQI				Total
			Small	**Moderate**	**Very large**	**Extremely large**	
Marital history	**Yes**	Count	2	9	11	0	22
		%	100.0%	90.0%	27.5%	0.0%	40.0%
	No	Count	0	1	29	3	33
		%	0.0%	10.0%	72.5%	100.0%	60.0%
Total		Count	2	10	40	3	55
		%	100.0%	100.0%	100.0%	100.0%	100.0%

CHI SQUARE = 18.021, P VALUE = 0.001 (S)

Chart 22: Association of Marital history with DLQI grading



	Yes	No
Small	100.00%	0.00%
Moderate	90.00%	10.00%
Very large	27.50%	72.50%
Extremely large	0.00%	100.00%

23. Association of Psychiatric visit with DLQI Grading

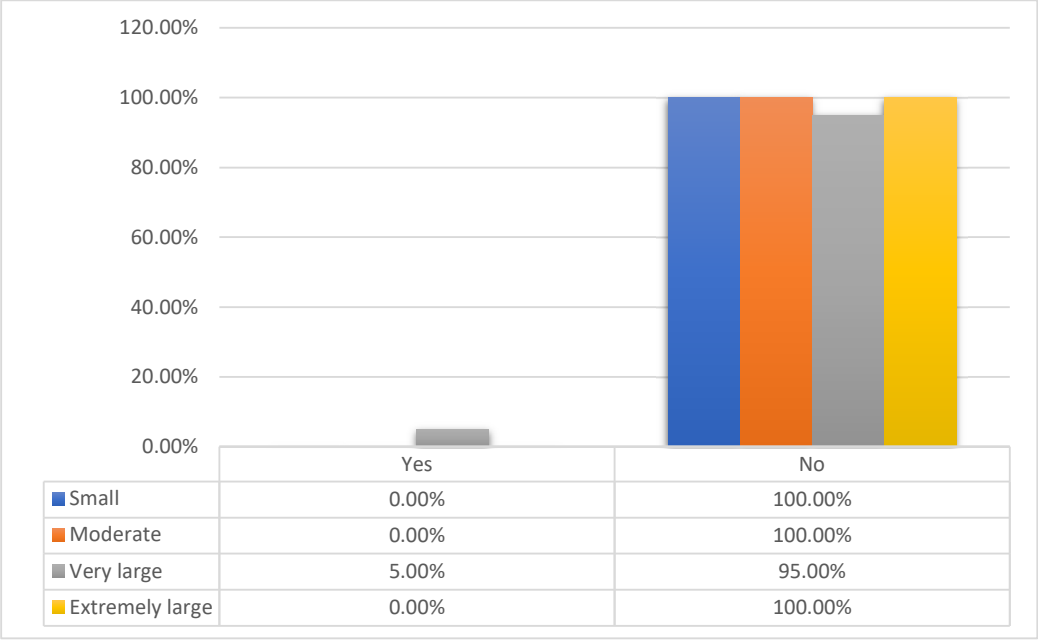
2 (3.6%) cases out of 55 cases went to psychiatrist for counselling regarding the disease and had a very large DLQI grading

Table 23: Association of Psychiatric visit with DLQI grading

			DLQI				Total
			Small	**Moderate**	**Very large**	**Extremely large**	
Psychiatric visit	**Yes**	Count	0	0	2	0	2
		%	0.0%	0.0%	5.0%	0.0%	3.6%
	No	Count	2	10	38	3	53
		%	100.0%	100.0%	95.0%	100.0%	96.4%
Total		Count	2	10	40	3	55
		%	100.0%	100.0%	100.0%	100.0%	100.0%

CHI SQUARE = 0.778, P VALUE = 0.043 (S)

Chart 23: Association of psychiatric visit with DLQI grading



24. Association of Counselling regarding disease to DLQI grading

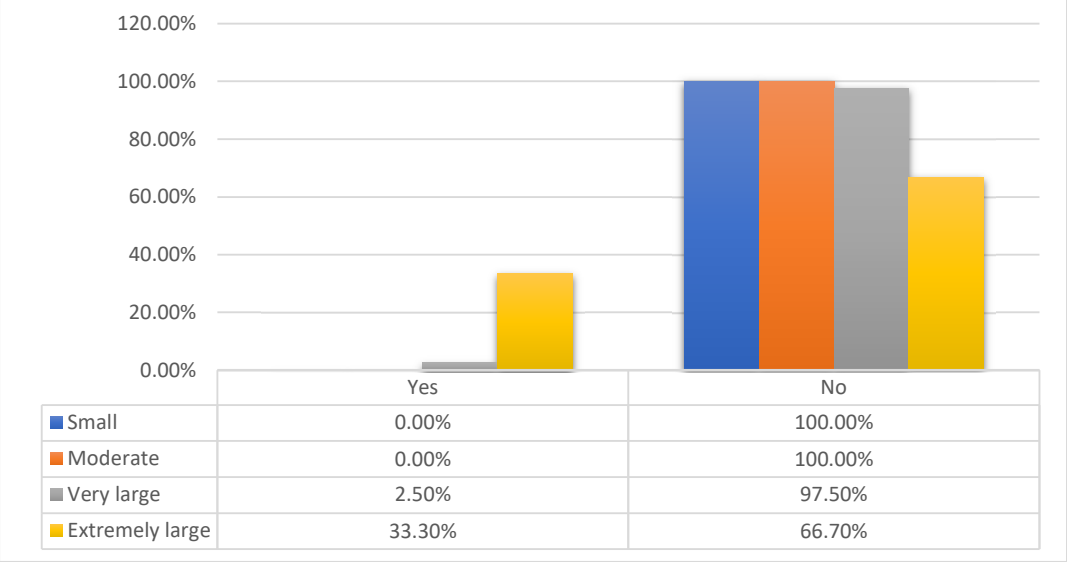
Only 2 (3.6%) cases out of 55 cases were counselled regarding the disease and had very large and extremely large DLQI scores.

Table 24: Association of counselling regarding the disease with DLQI grading

			DLQI				Total
			Small	**Moderate**	**Very large**	**Extremely large**	
Counselling regarding disease	**Yes**	Count	0	0	1	1	2
		%	0.0%	0.0%	2.5%	33.3%	3.6%
	No	Count	2	10	39	2	53
		%	100.0%	100.0%	97.5%	66.7%	96.4%
Total		Count	2	10	40	3	55
		%	100.0%	100.0%	100.0%	100.0%	100.0 %

CHI SQUARE = 8.151, P VALUE = 0.043 (S)

CHART 24: Association of counselling regarding the disease with DLQI grading



25. Association with treatment for disease and DLQI grading

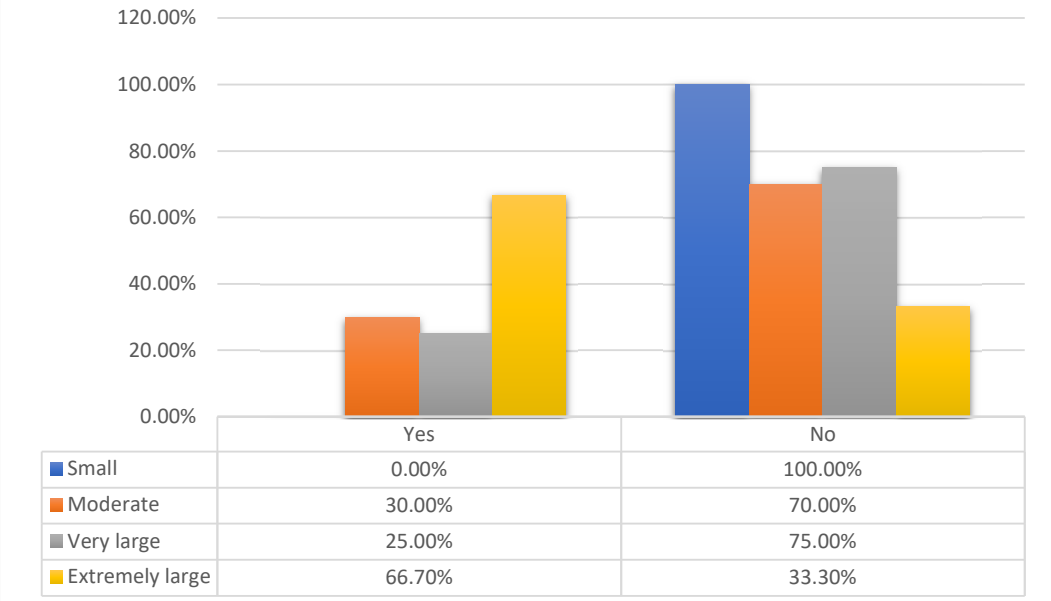
15 (27.3%) cases out of 55 cases were on treatment regarding the disease Out of which 10 (25%) cases had a very large DLQI scores. 40 cases were not on any treatment regarding the disease, out of which 30 (70%) cases had a very large DLQI score

Table 25: Association with treatment for disease and DLQI grading

			DLQI				Total
			Small	**Moderate**	**Very large**	**Extremely large**	
On treatment for disease	**Yes**	Count	0	3	10	2	15
		%	0.0%	30.0%	25.0%	66.7%	27.3%
	No	Count	2	7	30	1	40
		%	100.0%	70.0%	75.0%	33.3%	72.7%
Total		Count	2	10	40	3	55
		%	100.0%	100.0%	100.0%	100.0%	100.0%

CHI SQUARE = 3.239, P VALUE = 0.356 (NS)

Chart 25: Association with treatment for disease and DLQI grading



	Yes	No
Small	0.00%	100.00%
Moderate	30.00%	70.00%
Very large	25.00%	75.00%
Extremely large	66.70%	33.30%

DISCUSSION

Previous studies on genodermatoses so far have been of use for the purpose of prenatal diagnosis and genetic counselling as they are mainly disease specific. Currently, there is dearth of literature on genodermatoses as a whole in context to its spectrum, cutaneous affections and systemic involvement with only a few such studies done in India.

Literature review reveals very few global studies on patterns of genodermatoses, such as the study by Federica Dassoni et al. in paediatric population where 24 different genodermatoses in 122 affected individuals (0.4% of the total case load) was studied and another similar study involving adult patients by Regina C. Betz et al.[31,32] In India, studies on genodermatoses by Sunil kumar et al in paediatric patients and Farah Sameem et al[33,34]. in paediatric as well as adult patients have been done previously.

However, none of the above have assessed the quality of life in genodermatoses patients except for a study from southern India on genodermatoses by Karthik Raja et al.[36] However, in this study, individual factors affecting DLQI score were not studied. Hence we did a study for assessing the effect of skin involvement in genodermatoses on quality of life and study the individual factors affecting the DLQI scores.

AGE DISTRIBUTION OF GENODERMATOSES:

Of the 49 patients in study done by Karthik Raja et al,7 belonged to (1-20 years) age group, 24 were in (21-40years) group, 15 in (41-60 years) group, and 3 were > 60 years of age. Where as in our study, 9 were in age group of (17-20 years),

37 in age group of (21-40 years) and 9 in age group of (41-60 years). Both studies showed that maximum patients belonged to 21-40 years age group.

GENDER DISTRIBUTION OF GENODERMATOSES

In a study by Karthik Raja et al, 59% were males and 41% were females which is comparable with our study where in affected males and females were 56.4% and 43.6% respectively.

PREVALANCE

In our study, 14 different genodermatoses affecting 55 individuals (0.46% of total cases) were noted whereas Karthik Raja et al[36] observed a total of 20 different genodermatoses in 49 individuals (0.32% of total cases). Federica Dassoni et al. too noted prevalence of 0.4% similar to ours. However, Farah Sameem et al and Sunil Kumar et al in their studies observed a relatively higher prevalence of 0.72% and 0.62% respectively.

POSITIVE FAMILY HISTORY AND CONSANGUNITY:

Out of the total 55 cases in our study, 12 (21.8%) gave history of similar illness in family. This is in contrast with the findings of Karthik Raja et al[36] in which 34.6% had a positive family history. Farah Sammem et al and Sunil Kumar et.al observed a positive family history in 33.5% and 32.3 % respectively. This discrepancy must have been due to the variation in types of genodermatoses studied as inheritance pattern differs accordingly in those with history of consanguinity in parents. In our study 40% were born out of consanguinity in parents contradictory to finding of Farah Sameem et al,where consaguinity was observed in 57.8 %.

PATTERNS OF GENODERMATOSES IN THE STUDY POPULATION

Ichthyosis and neurofibromatosis were the most common cases observed in our study.

Ichthyosis

Ichthyosis was seen in 20% of the cases in our study whereas it was observed in 33.59% by Farah Sammem et al. In their study. However, Sunil Kumar et al[33] reported a very high incidence of 55.8% in their study.

In those affected with ichthyosis vulgaris, 27.2% had no history of consanguinity in parents in contrast to the observations by Sivayadevi et al[36] wherein history of consanguinity was noted in 76%.

Neurofibromatosis

In our study, neurofibromatosis accounted for 30.9% of the cases. However, in the study by Karthik Raja et al [36] it was noted in 10.2% cases.

EFFECT OF DERMATOSES ON QUALITY OF LIFE:

Literature review shows no such study which has compared the DLQI scores with occupational status, disease activity, percentage of body involvement, age of onset, marital history, or those on treatment for disease.

DLQI SCORE:

In our study, 31 were male of which 21 (67.74%) had a very large DLQI grade. Of the 24 females in our study 19 (79.1%) had a very large effect according to

DLQI grading. This shows that females seemed to have more effect on quality of life than males.

40/55 cases in our study had very large DLQI grading out of which 31 (77.5%) cases belonged to 21-40 years age group and 8 (20%) cases from 17-20 years age group. This indicates that age group of 21-40 years had very large effect. However, in the study by Karthik Raja et al,[36] the same age group was seen to have a moderate effect in 35.6% patients.

33 (60%) /55 were unmarried, of which 29 (87.87%) had a very large DLQI grading, followed by extremely large DLQI grading in 3 (9.09%) cases. This showed that singles had more effect on quality of life.

1 (1.8%) case had malignant lesion with a extremely large DLQI grading, indicating that malignant lesion had more effect on DLQI score than benign lesions.

In our study it was found that 15/55 (27.3%) were seeking treatment for the disease. Out of which 10 (66.6%) cases had a very large DLQI scores.

40 cases were not on any treatment regarding the disease, of which 30 (70%) had a very large DLQI score. Hence it was observed in our study that there was no significant correlation between DLQI and patients seeking treatment.

DOMAINWISE EFFECT ON DLQI:

Students, house wives and employees showed to have a common affection in the domain of “symptoms and feelings” where as in study by Karthik raja et al., only students were seen to have more effect in the “symptoms and feelings” domain.

In our study" personal relationship" domain was seen to be affected in 52.7% in contradiction to the study by Karthik Raja et. al where only 35% were affected.

Domain wise comparison of our study with that of Karthik Raja et al.[36] showed similar results with respective to the domains involving leisure, daily activities and no affection

Table 26: Domain wise effect of disease on DLQI

Heading	Our study (n=55)	Karthik Raja et al (n=49)
Symptoms and feelings	38 (69%)	30(61%)
Daily activities	32 (58.1%)	26(53%)
Leisure	25 (45.4%)	21(43%)
Work and school	14 (25.4%)	5 (10%)
Personal relationship	29 (52.7%)	17(35%)
Treatment	30 (54.5%)	15(30%)
No affection	3 (5.4%)	6(12%)

The mean age of subjects in this study was 29.9 years and mean percentage of body involvement was 23.67% by using rule of 9.

Average age of onset of the lesions was around 11.04 years and the mean DLQI score in our study was 14.73 with a minimum score of 5 and maximum score of 27.

Table:27 Denoting the age, percentage body involvement, age of onset of the disease and DLQI scores

	Age in years	percentage of body involvement	Age of onset in years	DLQI score
Mean	29.09	23.67	11.04	14.73
Minimum	17	2	1	5
Maximum	60	90	35	27

33(82.5%)/55 cases had new lesions/progression of skin involvement and they were seen to have a very large DLQI score.

CONCLUSION

To the best of our knowledge our study is the first to study the affect on quality of life across various factors wherein we observed that female sex, 21-40 years age group and u- married subjects had a very large affect as per DLQI score which lead to feeling of embarrassment, disturbed social activities and relationship with friends and family.

Patients with genodermatoses as well as their parents require to be counselled regarding the nature of their disease, chances of familial involvement, the associated possible systemic involvement, need for regular hospital visits and educate them regarding the available treatment options as these impact their quality of life.

Social help and vocational rehabilitation too need to be developed to help integrate these patients with the mainstream.

LIMITATIONS

1. Small sample size. A study with larger sample is needed to validate our studies findings.

2. Along with assessment of patient's quality of life, a study to assess the patient's parents/guardians quality of life might help in understanding the overall impact on inter- personal relationship and family life.

SUMMARY

Genodermatoses patients constitutes a small but significant number in dermatological practice. Most of the diseases are life-long, multi-system disorders with limited treatment options.

The present study was conducted on all patients in the age group 16-60 years attending the dermatology department of KLE's DR. PRABHAKAR KORE HOSPITAL having genodermatoses. All consenting patients were administered the DLQI questionnaire (English, Hindi, Marathi, Kannada).

Our study included 55 patients out of which 56.4% were males and 43.6 % were females. The study included subjects from 16-60, the mean age group was 29.09 +/- 10 years.

In our study subjects as housewives and students were more common. All patients had active disease. Out of 55 included subjects, 2 subjects had diabetes, 2 had hypertensions, 1 had epilepsy.

There was only a single case with malignancy in the study.

Family history was positive in 21.8% cases, with second degree consanguinity in 40% cases.

Only 2 out of 55 had undergone / received counselling done regarding the disease when diagnosed.

Positive correlation of DLQI score was seen in younger age group, females, single individuals, having disease activity and with those who had undergone

counselling for the disease. However there was no significant correlation seen with DLQI score in those seeking treatment for the disease.

This study shows that there is affect on the quality of life of patients with genodermatoses. Therefore such patients and their parents need to be counselled about the disease, transmission patterns, chances of systemic involvement and various treatment options.

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ANNEXURE I – CONSENT FORM

Title of the study: "A CROSS SECTIONAL AND OBSERVATIONAL STUDY OF DERMATOLOGY LIFE QUALITY INDEX SCORE IN GENODERMATOSES PATIENTS AT A TERTIARY CARE HOSPITAL"

The study is conducted by Dr. S PAL KISHAN REDDY, post graduate(M.D) student in dermatology under the guidance of Dr. BHAVANA R.DOSHI MD,DVD,FIDD professor and HOD, department of Dermatology, Venereology and Leprosy, JNMC, Belagavi.

Respected Siri/Madam,

We invite you in participate in our study as you are eligible for the same. During the study you will be asked some questions in details regarding your present complaints.

Purpose of the study:

Genodermatoses cause cosmetic disfigurement and patient present to the dermatologist for their unusual appearance. Hence with the DLQI questionnaire intends to observe the quality of the patient. You are being requested to participate in this study because you have been diagnosed to have genodermatoses.

Procedure:

You will be asked to give a detailed history of your disease, undergo a physical examination, followed by the questionnaire along with the clinical pictures of the lesions.

Risk and Benefits:

The result of you taking part in this research would help health care providers towards a better understanding the quality of life, thus we will able to provide improved patient care.

Alternatives:

If you decide not to participate in this study, you will still be receiving the usual standard care for your disease.

Privacy and confidentiality:

Your privacy will be respected and all information collected about you during the course of study will be kept confidential. Your identity will be remain undisclosed.

Relations with institutional policy:

The Jawaharlal Nehru medical college will provide, within the limitations of the laws of the state of Karnataka, facilities and medical attention to patients who suffer injuries as a result of participating in this project.

Financial incentives:

You shall not be receiving any payment or any financial incentives for participating in this study.

Authorization to publish results:

The result of this study may be published for scientific purposes or presented to a scientific to scientific group. Your identity, however, will be maintained confidential at all times.

Voluntary participation:

Your participation in this study is voluntary. Your decision whether or not to participate will neither affect the care of your current disease, nor your future relations with the doctor or the hospital. In the event if you suffer any physical injury as the result of your participation in this study, you may contact Dr. S PAL KISHAN REDDY, telephone No: 9164084683 or Dr. BHAVANA R.DOSHI MD, DVD, FIDD telephone No: 9422306523. In the event of an emergency, you should contact KLE's Dr. Prabhakar kore hospital and medical research centre on telephone No: 08312473777.

In case you need further information regarding your rights as a study participant you may please contact Dr. HARSHA HEGDE, chairperson, JNMC, IEC and scientist D, ICMR, National Institute of Traditional medicine, Belagavi-9480422500.

ANNEXURE II – PROFORMA

PROFORMA

Name:

Age:

IP No:

Sex:

Occupation:

Address:

Phone No:

Diagnosis:

Part A: To be filled by practitioner

1) Type of lesion observed

   Macule……

   Papules……...

   Patches……

   Plaques……

   Scars……

   Erosions……

   Other lesions……

2) Condition of the skin lesion.

Infective lesions □

Active lesions □

Healing □

3) Any other lesion associated along with genodermatoses

Yes □

No □

If yes, then mention the other lesions observed along with genodermatoses

.....

Part B: To be filled by the patient

1) Are the lesions associated with other systemic disorders?

Yes □

No □

If, yes then mention the name of the associated disease.

.....

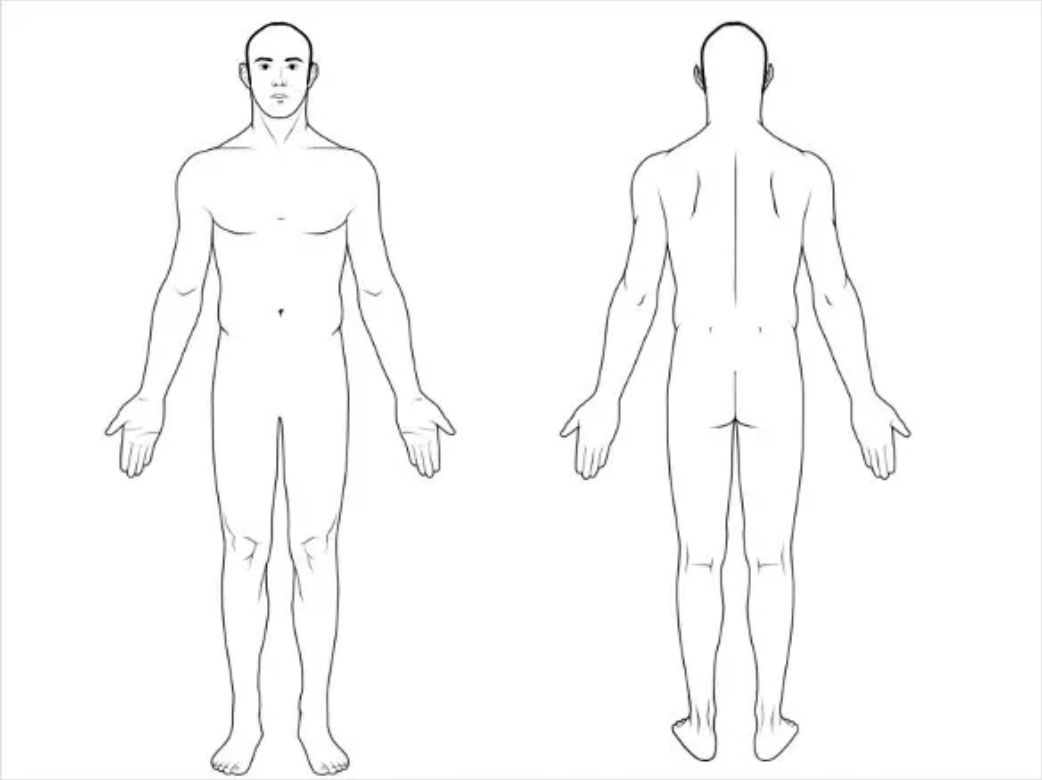
2) Are the visible lesions malignant or benign?

Yes □

No □

If yes, then mention the duration.

3) site and portion of skin present over the body?



4) Are there any similar lesions present in your family?

Yes □

No □

5) age of onset of skin lesions.....

6) Marital history

I) marital status

Married □

Un married □

7) Consanguineous marriage in parents

Yes

No

If yes, consanguineous marriage

1st degree □

2nd degree □

8) Are the lesions over skin are exposed out of the daily dressing style?

Yes □

No □

9) Have you ever visited psychiatrist for counselling regarding your lesions on your skin?

Yes □

No □

10) Have you been counselled regarding your skin disease/aware of the skin disease and its association?

Yes

No

DERMATOLOGY LIFE QUALITY INDEX (DLQI)

Hospital No: Name:

Address:

Date: Score: Diagnosis:

The aim of this questionnaire is to measure how much your skin problem has affected your life OVER THE LAST WEEK. Please tick ( ) one box for each question.

1) Over the last week, how **itchy, sore, painful or stinging** has your skin been?

Very much □

A lot □

A little □

Not at all □

2) Over the last week, how **embarrassed or self conscious** have you been because of your skin?

very much □

A lot □

A little □

Not at all □

3) Over the last week, how much has your **skin interfered** with you going **shopping** or looking after your home or garden?

Very much □

A lot □

A little □

Not at all □ Not relevant □

4) Over the last week, how much has your **skin influenced the clothes you wear**?

Very much □

A lot □

A little □

Not at all □ Not relevant □

5) Over the last week, how much has your **skin affected any social** or leisure activities?

Very much □

A lot □

A little □

Not at all □ Not relevant □

6) Over the last week, how much has your **skin made it difficult** for you to do any **sport**?

Very much □

A lot □

A little □

Not at all □ Not relevant □

7) Over the last week, has your **skin prevented you from working or studying**?

Yes □

No □

If "No", over the last week how much has your **skin been a problem at work or studying?**

Very much □

A lot □

A little □

Not at all □ Not relavent □

8) Over the last week, how much has your **skin created problems** with your **partner or any of your close friends or relatives**?

Very much □

A lot □

A little □

Not at all □ Not relevant □

9) Over the last week, how much has your **skin caused any sexual difficulties**?

Very much □

A lot □

A little □

Not at all □ Not relevant □

10) Over the last week, how much of a problem has the **treatment for your skin** been, for example by making your home messy, or by **taking up time**?

Very much □

A lot □

A little □

Not at all □ Not relevant □

ANNEXURE III – PHOTOGRAPHS

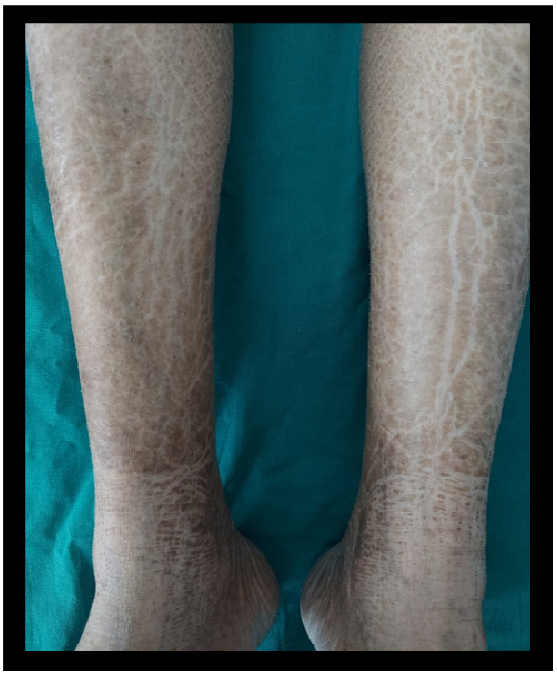


Figure: 1 Clinical image of Ichthyosis Vulgaris over lower limb

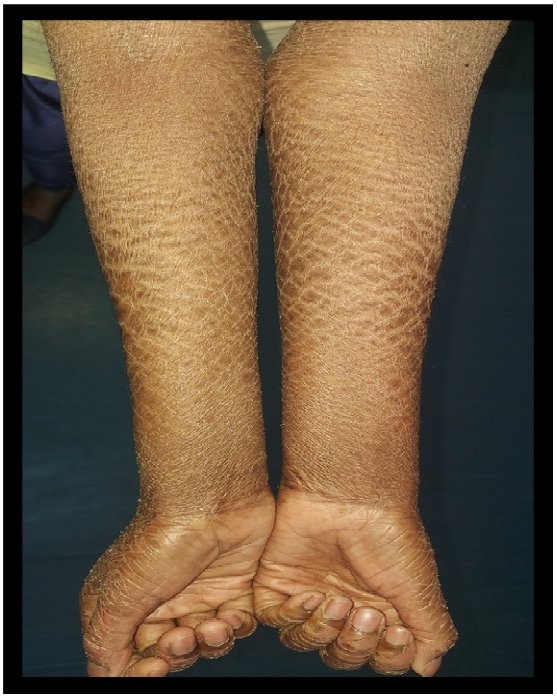


Figure: 2 Clinical image of Ichthyosis Vulgaris over upper limbs

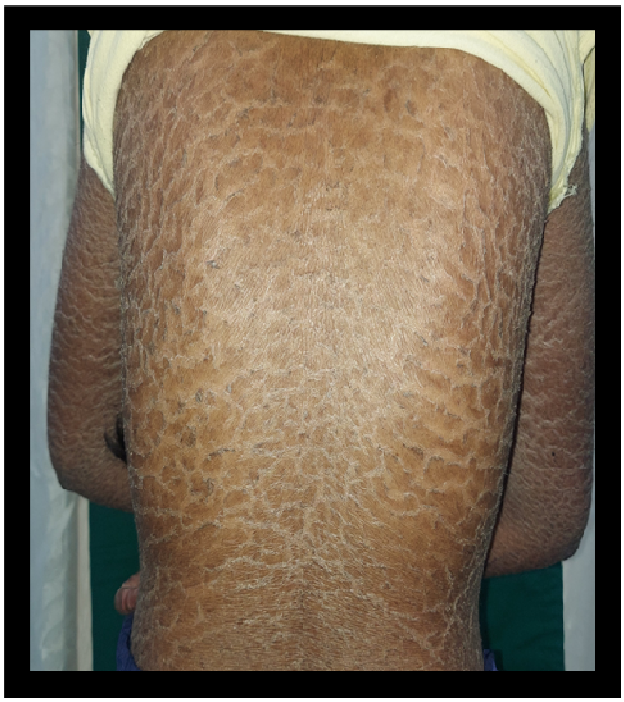


Figure: 3 Clinical image of Ichthyosis Vulgaris over trunk

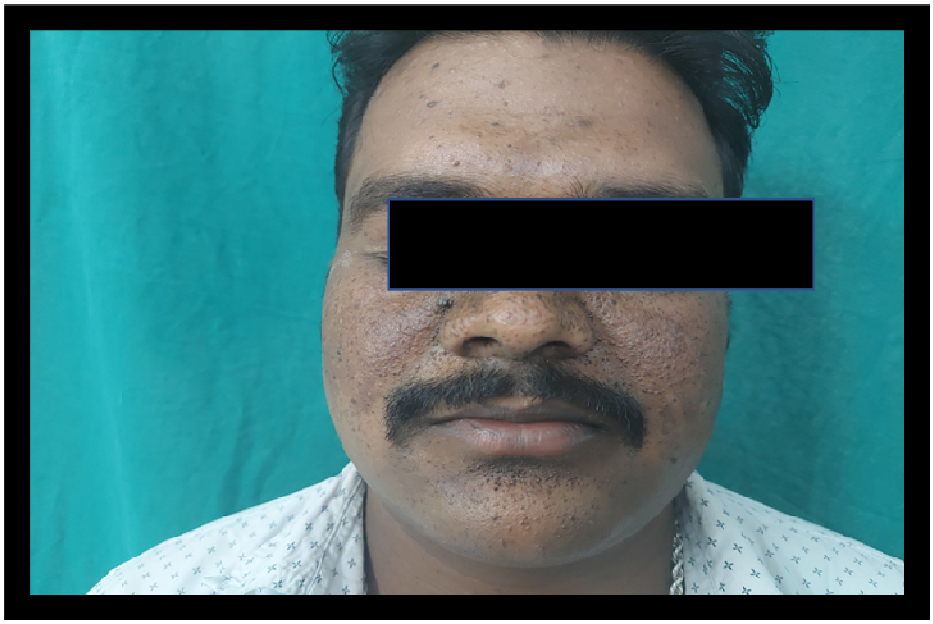


Figure: 4 Clinical image of Tuberous sclerosis

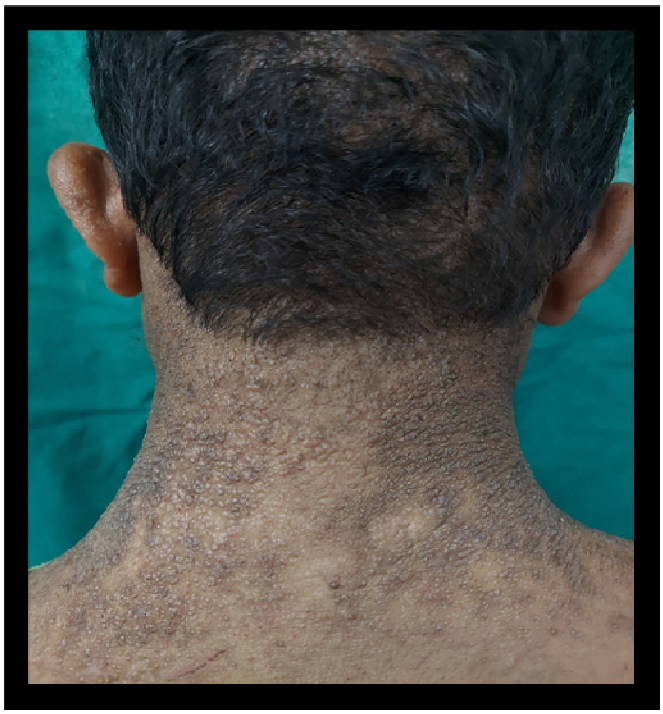


Figure: 5 Clinical image of Lipoid proteinosis over neck

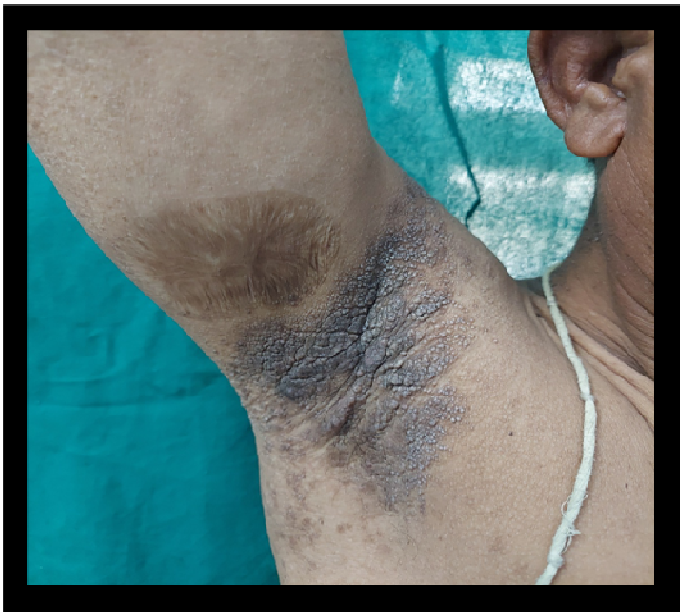


Figure: 6 Clinical image of Lipoid proteinosis over axilla

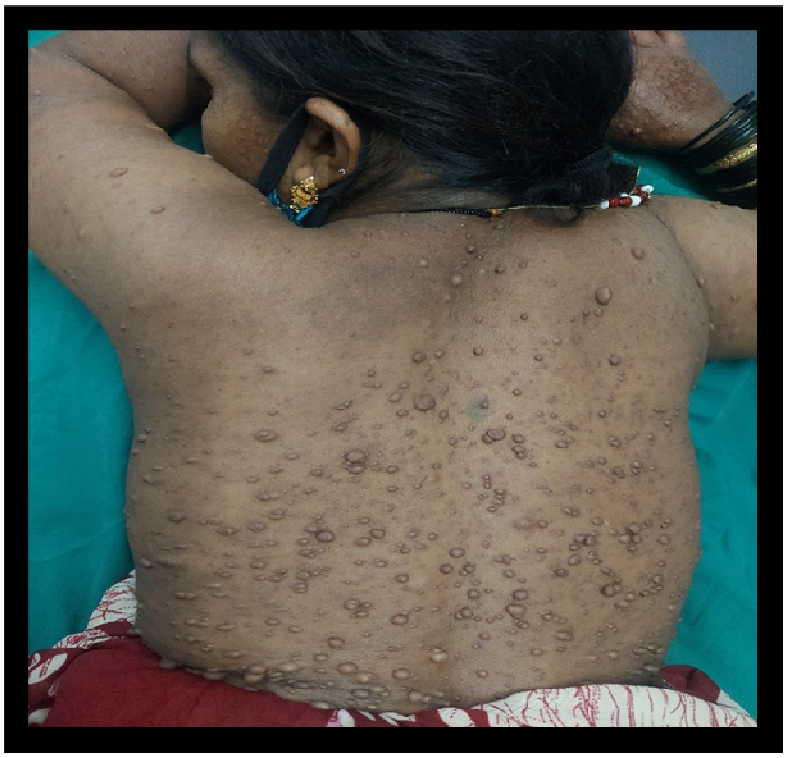


Figure: 7 Clinical image of Neurofibromatosis over trunk

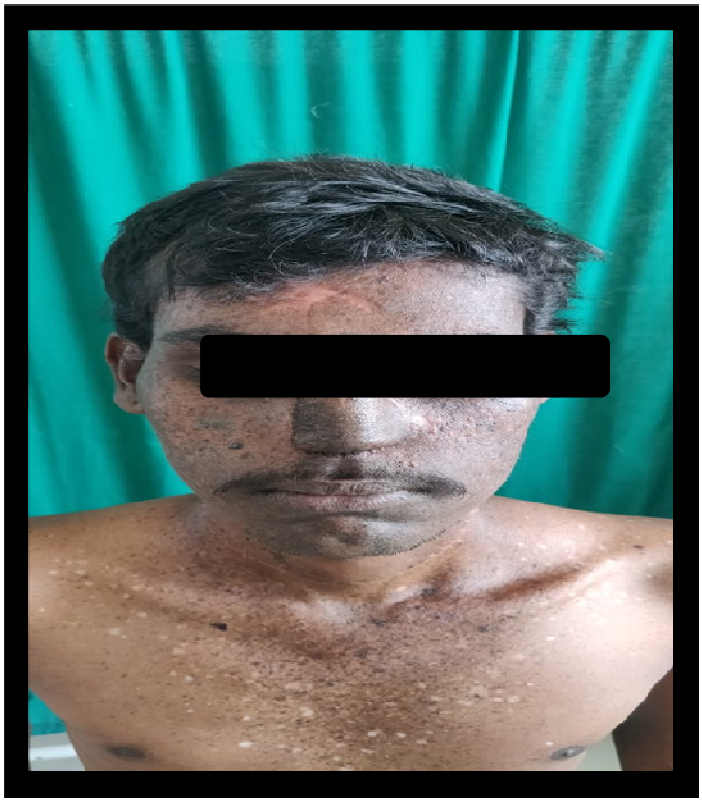


Figure: 8 Clinical image of Xeroderma pigmentosa

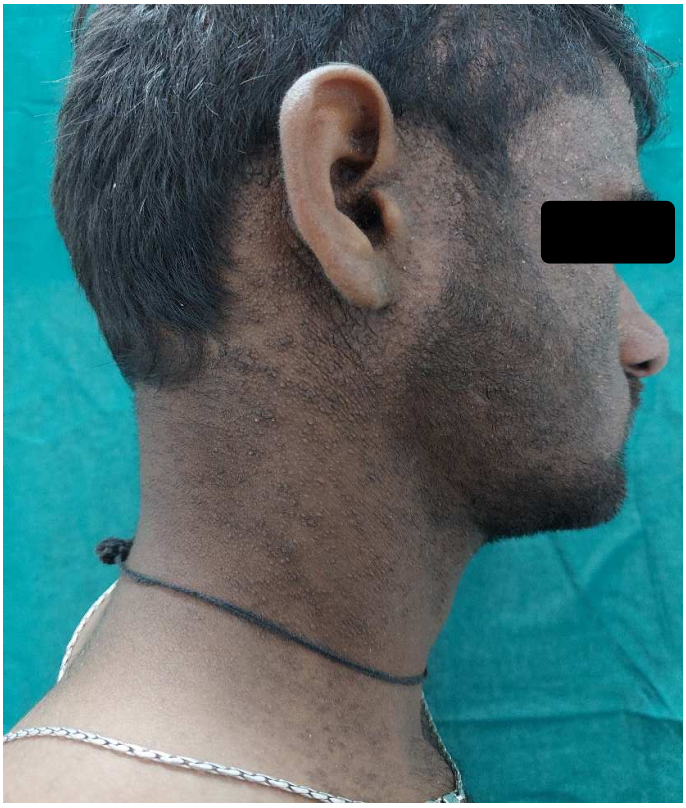


Figure: 8 Clinical image of Dariers disease

ANNEXURE IV - KEY TO MASTER CHART

Gender:

- Male: 1
- Female: 2

Condition of lesions:

- Infected: 1
- Active: 2
- Healing: 3

Associated skin lesions, systemic disorders, benign or malignant lesions, family history, family history with relations, marital history: parents consanguineous, degree of consanguineous, lesions expose out of dressing style, psychiatric visits, counseling regarding disease and on treatment for disease:

- Yes: 1
- No: 2