IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 20, Issue 11 Ser.9 (November. 2021), PP 16-23
www.iosrjournals.org

A Clinico-Epidemiological Profile of Hereditary Palmoplantar Keratodermas: An Observational study

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Abstract:
Background :*Hereditary palmoplantar keratodermas (HPPKD) are a heterogenous group of keratinizing disorders classified based upon their mode of inheritance, age of onset, morphology and distribution of palmoplantar thickening.Clinically classified into four major types :1.Diffuse2.Focal3.Striate and 4.Punctate.They are more prone for secondary infections like tinea pedis, tinea manuum, pitted keratolysis and maceration of skin.As they are rare disorders, literature reports are based on individual family reports.Only few studies are available about the prevalence of palmoplantar keratoderma in the Indian population till date.*
Aim : *To study the clinical and epidemiological trend of hereditary palmoplantar keratoderma.*
Objective : *To analyze the age and sex distribution. To analyze various clinical presentations .*
Materials and methods *: A prospective observational study was carried out from June 2019 to June 2021 in patients with hereditary palmoplantar keratoderma attending Out Patient Department(OPD) of Dermatology , Siddhartha medical college, a tertiary care hospital, .*
Result:*In this study out of 48,740 out patientcases, total number of cases with HPPKD found to be 35 with prevalence range of 0.07% .Out of which HPPKD syndromes constituted 0.045% and HPPKD associated with othergenodermatoses found to be 0.027%.Most commonly observed in second decade with a male to female ratio of 1.18:1.History of consanguinity is noted in 60% of patients with positive family history in 30%.Most are autosomal dominant inherited. Diffuse transgrediens type is the common pattern observed in which Greither's was most common.*
Conclusion: *There is a wide genetic and phenotypic heterogeneity, due to which reaching an accurate diagnosis only on the basis of clinical features is challenging, hence a thorough history taking including family history is essential to take therapeutic decisions and offer genetic counselling.*
Key word: *Hereditary, Palmoplantar keratoderma , Inherited , Genodermatoses, Syndrome*

Date of Submission: 14-11-2021 Date of Acceptance: 29-11-2021

I. Introduction

Palmoplantar keratodermas(PPKD) represents a diverse group of hereditary and acquired disorders in which there is hyperkeratosis of the palms and soles[1,2]. To date, the diagnosis of the different hereditary PPKDs(HPPKDs) is based mainly on inheritance pattern, age at onset, morphology, distribution and severity of hyperkeratosis, pattern of additional dermatologic and systemic manifestations, histopathological findings and relative treatment resistance[3,4]. Sporadic (spontaneous) mutations need to be considered in those without a family history or late onset disease[5].

The HPPKDs have been reviewed extensively in the literature over the past years and in 1996, Stevens et al proposed a classification scheme[6]. Later on, in 2005, Itin and Fistrol classified the HPPKDs according to inheritance pattern and molecular basis[6].

In a broad sense, HPPKDs may be divided, on the basis of the phenotypic consequences of the underlying genetic defect into syndromic and non-syndromic or on the basis of distinctive palmoplantar involvement into Diffuse, Focal, Punctate and Striate[7] .

The present study is taken up due to paucity of literature about epidemiology of HPPKDs and to appraise the different clinical patterns of HPPKDs.

II. Material and Methods

Study design: A prospective observational study
Study period: June 2019- June 2021 – 2years
Data collection: Based on the proforma, detailed analysis was done which include the history taking, with particular emphasis on the age of onset of the disease, progression of the disease, involvement beyond the palms and soles, discoloration of the keratotic surface, recurrent skin infections, recurrent blisters, constricting bands over digits, autoamputation of digits. Detailed family history, history of consanguinity noted. History of appendageal involvement, eyes and ears and involvement of other systems were taken into account.
Sample size: 35
Inclusion criteria:
Patients with PPKD present since birth, infancy, early childhood & adolescence
Exclusion criteria:
Patients of PPKD with adult onset associated with systemic disorders, infections and inflammatory dermatoses
Statistical analysis :
Simple proportions and percentages for comparing different variables like age, sex, etc. were used. Comparison was tested for statistical significance using Chi-square test & t tests wherever applicable.

III. Results :

Out of 48,740 patients who attended our OPD total number of patients with hereditary palmoplantar keratoderma(HPPKD) were 35, corresponding to 0.07% of prevalence. Males(19 – 54.29%) outnumbered females (16=-45.71%) with a male to female ratio of 1.18:1. (Table-1 &2)

TABLE -1 : Prevalence of HereditaryPalmoplantar Keratoderma (HPPKD)

Total Out-Patients	HPPKD	Prevalence
48, 740	35	0.07%

Table -2: Sex distribution of HPPKDs

Total HPPKD	35
Males	19(54.29%)
Females	16(45.71%)

Most number of patients presented to hospital with symptoms and complications of PPKDS during their second decade of life i.e., 11-20 years of life (45.7% of cases) followed by first decade (20%). Whereas the patients gave history of the onset of PPKDS most commonly in first decade **17(48.6%)** followed by second decade**10(28.6%)**. (Tables 3 &4)

Table 3: Age wise presentation of hereditary palmoplantar keratoderma (HPPKD)

AGE IN DECADES	**HPPKD**
1-10	**7(20%)**
11-20	**16(45.7%)**
21-30	5 (14.3%)
31-40	4(11.4%)
41-50	2 (5.7%)
51-60	1(2.9%)

Table 4 : Age of onset of hereditary palmoplantar keratoderma(HPPKD)

AGE IN DECADES	**H PPKD**
1-10	**17(48.6%)**
11-20	**10(28.6%)**
21-30	7(20%)
31-40	1(2.9%)
41-50	0(0%)
51-60	0(0%)

The most common pattern of hereditary palmoplantar keratoderma(HPKKD) in both males and females is diffuse transgrediens 15 (42.8%) of which Greither–transgrediens&Progrediens[8](22.8%) being the commonest. There is no statistical difference between incidence of different patterns of HPPKDs among males and females with P value of 0.36 calculated by t-test. (tables 5,6&7)

Table 5 : Clinical patterns Of Hereditary Palmoplantar Keratoderma(HPPKD)

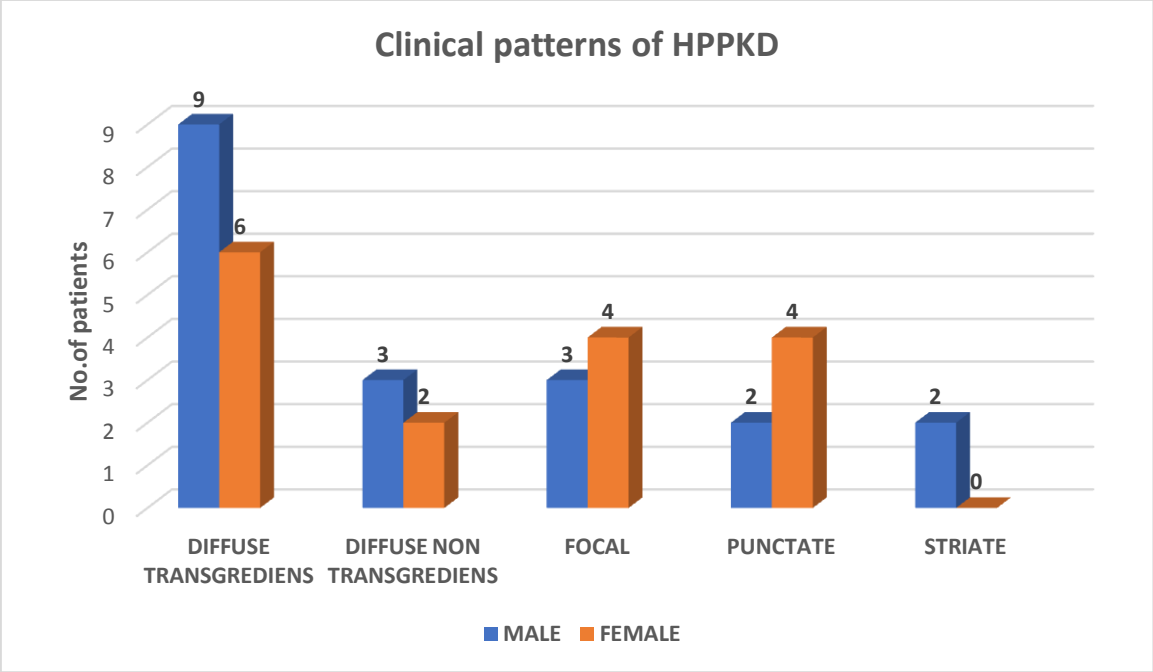


TABLE 6 : Clinical types of Hereditary Palmoplantar Keratoderma (HPPKD)

PPK SYNDROMES	NO. OF PATIENTS
GREITHER'S SYNDROME	**8(22.8%)**
VOHWINKEL SYNDROME	4(11.4%)
MAL DE MELEDA SYNDROME	3(8.5%)
PAPILLON LEFEVRE SYNDROME	3(8.5%)
UNNA THOST SYNDROME	2(5.7%)
OLMSTED SYNDROME	1(2.8%)
JADASSOHN LEWANDOWSKY SYNDROME	1(2.8%)

GENODERMATOSES AND PPK	NO. OF PATIENTS
ICHTHYOSISVULGARIS	**5**(14.2%)
DARIERS DISEASE	3(8.5%)
FAMILIAL PITYRIASIS RUBRA PILARIS	2(5.7%)
NON-BULLOUS ICHTHYOSIFORM ERYTHRODERMA	1(2.8%)
LAMELLAR ICHTHYOSIS	1(2.8%)
CLOUSTON SYNDROME	1(2.8%)

The disorder with the largest number of family members affected in a pedigree was Greither PPK with 5 members which was also a common finding in study done by Gulati.S, Thappa et al[8].
Out of 35 patients with HPPKD , 21(60%) patients had positive family history of similar illness. Third degree consanguinity with positive family history 15 (42.8%) was the most common finding among the patients (table 7)

TABLE 7: Family history and consanguinity in relation to hereditary palmoplantar keratoderma(HPPKD)

CONSANGUINITY	POSITIVE FAMILY HISTORY	NEGATIVE FAMILY HISTORY
Non consanguinity	1	8
2^0 consanguinity	5	4
3^0 consanguinity	**15**	2

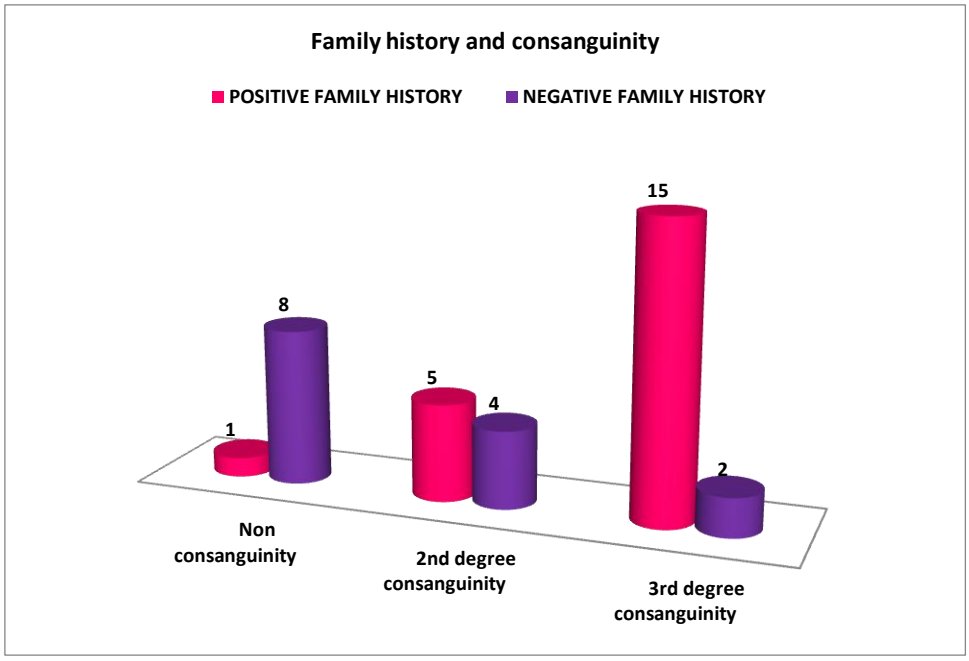
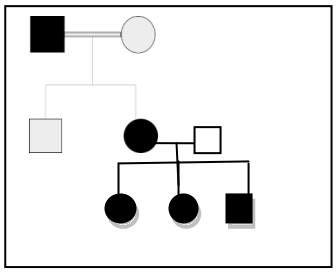


Figure 1: Greither's syndrome in a family &Pedigree chart



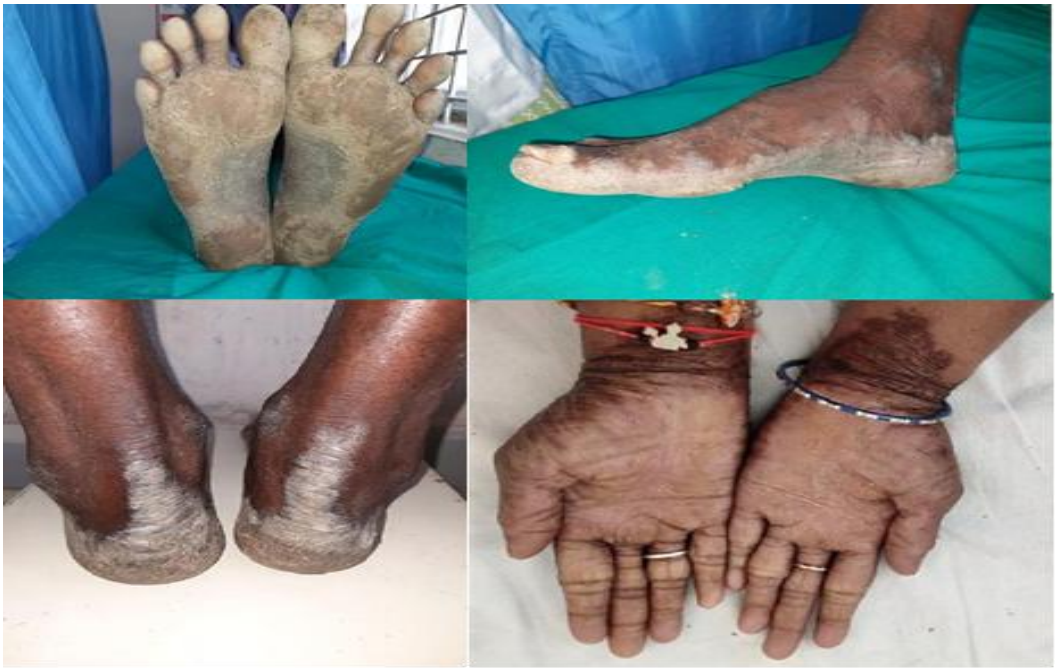
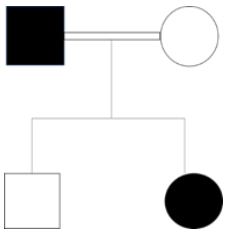
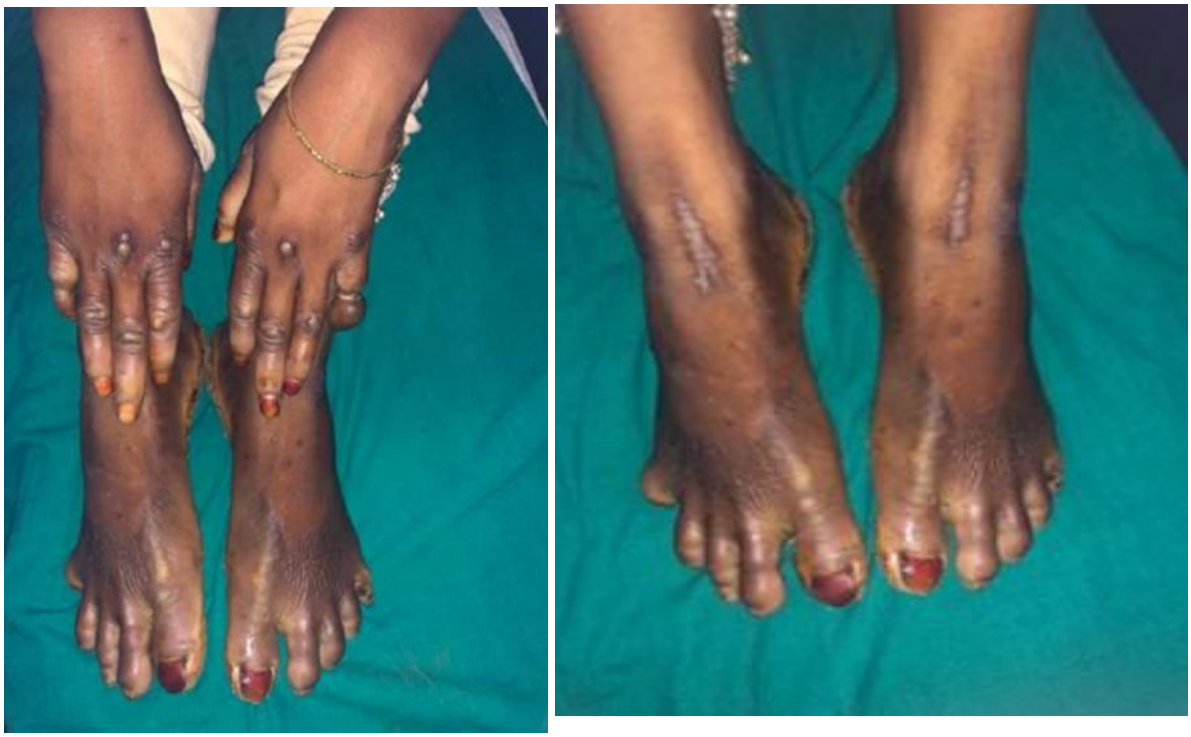
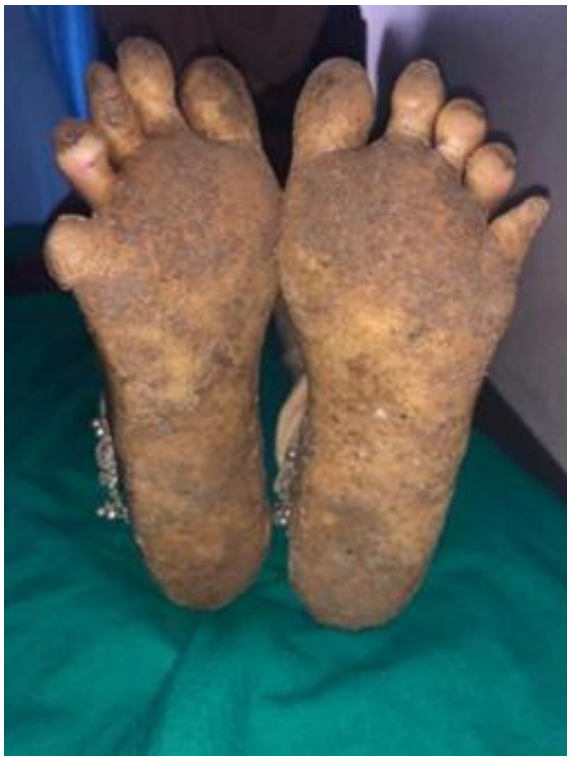
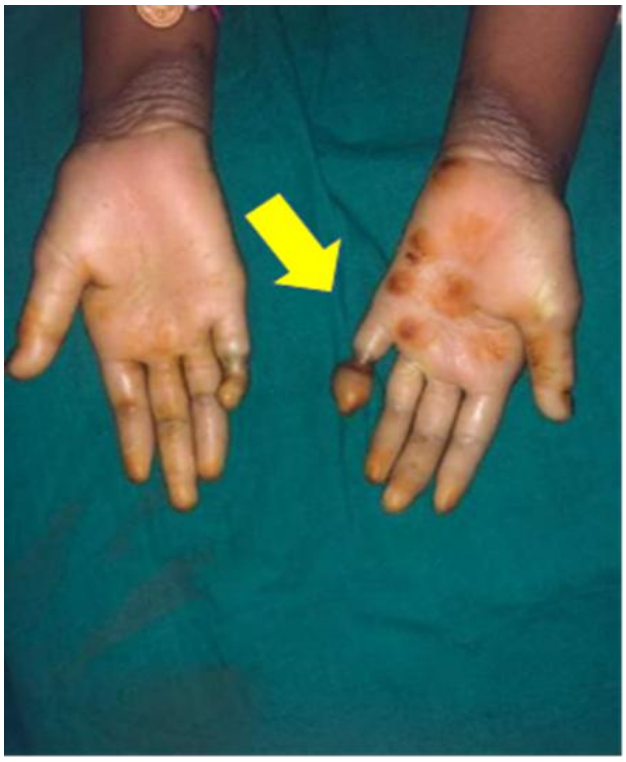


Figure 2: Vohwinkel syndrome& Pedigree chart

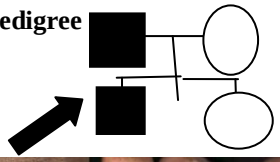


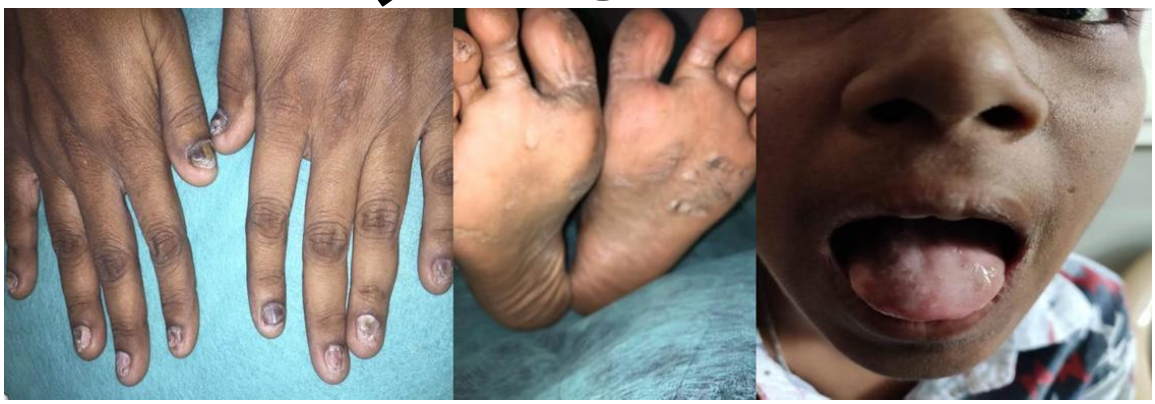




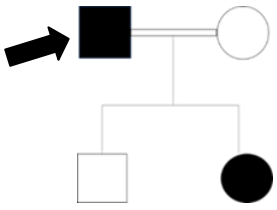


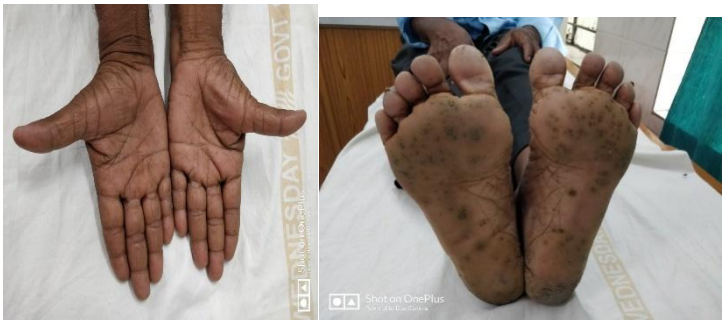
Pachyonychia congenita type 1&pedigree



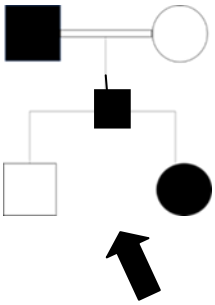


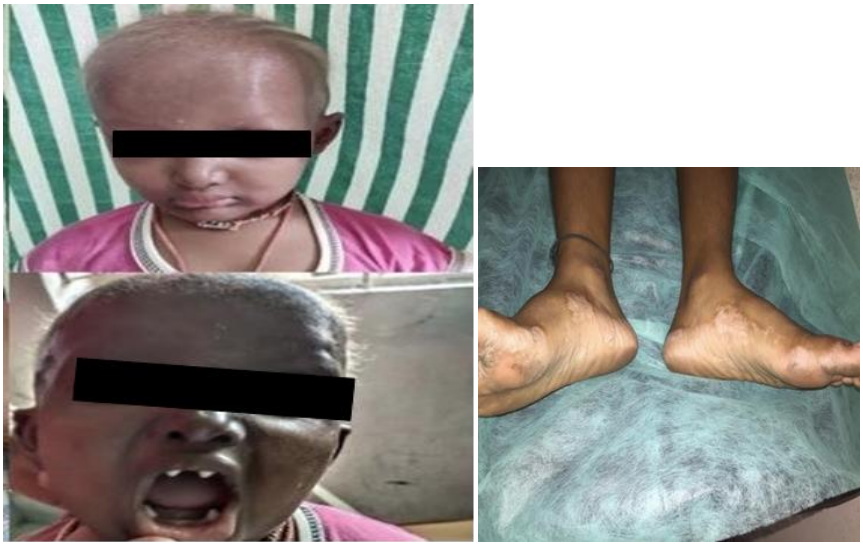
Punctate keratoderma&pedigree



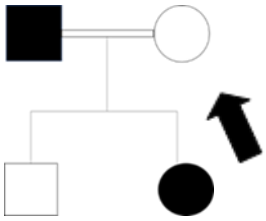


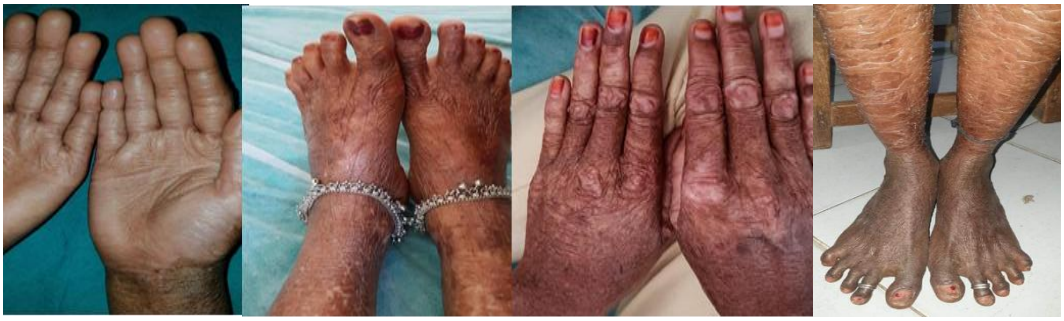
Clouston syndrome&pedigree





Ichthyosis vulgaris&pedigree





IV. Discussion

In the present study conducted at one of the states from South India , Andhra Pradesh , the prevalence and prevalence rate of hereditary palmoplantar keratoderma (HPPKD) (total -35) in our hospital was found to be 0.07% (7.1 /10,000) which is higher than Gulati.S, Thappa et al[8] prevalence of HPPKD as 5.2 /10,000 from Pondicherry and S.C. Murthy et al[9] prevalence of HPPKD 5.3 /10000 from Karnataka. This shows varied prevalence among South Indian states.

The prevalence of HPPKDs is almost same in the present study with a male to female ratio of 1.18:1(M:F) is comparable to 1.28:1 (M:F) S.C. Murthy et al[9] done in 2008but lower than another South Indian study done in 1997 Gulati.S, Thappa et al[8] ratio was 4.2: 1 (M:F). This can be attributed to social stigma among women population to approach a hospital during 1990s when compared to the gender equality achieved during the present era.

The age of presentation to the hospital that is prevalence was high among the age group 11-20 years 16 (45.7%) which is concurrent with findings made by Gulati.S, Thappa et al[8] and S.C. Murthy et al[9] where the age of presentation was between 0-10 years of life 67.7% of cases and 68.8% of cases respectively.
The age of onset in the present study was 0-10 years similar to above studies[5,6]. The discrepancy of late presentation to the hospital can be attributed to the following 3 reasons:
1. As complications start to occur in second decade,
2.Lockdown restrictions in COVID pandemic since last 2 years in India and
3. Increased number of tertiary care medical hospitals nearby , as the pool of patients gets distributed among these hospitals and patients receive primary care .

The most common pattern of HPPKD in the present study is diffuse transgrediens 15 (42.8%)of which Greither–transgrediens&Progrediens[8](22.8%)most prevalent which is not comparable to the similar studies done by Gulati.S, Thappa et al[8] and S.C. Murthy et al[9] where the Unna-Thost PPK was the most common PPK with 38.7% and 37.5% prevalence respectively.

HPPKDs associated with genodermatoses most common was ichthyosis vulgaris 5(14.2%) which was comparable to studies of Gulati.S, Thappa et al[8] and S.C. Murthy et al[9] with 2(6.45%) and 3(18.75%), but the prevalence is not the same.

We have seen only 2 cases of striate clinical pattern of HPPKD. Other less common clinical variants in this study are among PPK syndromes are Olmsted syndrome and pachyonychia congenita , among Genodermatoses associated PPKDs are Non BullousIchthyosiform Erythroderma, Lamellar Ichthyosis and Clouston Syndrome.

Analysis of HPPKDs revealed that most of the HPPKDs are Autosomal Dominant(AD) inherited . There is positive family history and offsprings of consanguineous marriage 20 (57.14%) had positive family history 21(60%) of similar illness which signifies the need of early genetic counselling, prenatal screening and pre-marital counseling discouraging consanguineous marriages.

V. Conclusion

The Hereditary palmoplantar keratodermas (HPPKD) are a heterogeneous group of conditions with a biologically fascinating diversity of genetic mutations and phenotypic presentations which make diagnosis difficult. Modern sequencing techniques have aided our ability to re-classify these conditions. In hospitals where facilities are not available for genetic analysis clinical diagnosis plays a crucial role in diagnosing and planning the suitable management protocol which is mainly symptomatic and prevents further complications such as mutilations and deformities as these conditions are incurable.

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