OPEN ACCESS Research Paper

Clinico-Demographic Study of Genodermatoses in Patients Attending Baghdad Dermatology Center of Medical City

Zahraa Adil Dawood[1], Basman Medhat Fadheel [2]

ABSTRACT:

BACKGROUND:

Genodermatoses are inherited skin disorders with variable expressivity and high morbidity. Their epidemiology and expression show regional variation due to genetic and environmental influences.

OBJECTIVE:

To record genodermatoses' clinical and epidemiological characteristics among patients attending the Baghdad Dermatology Centre of Medical City in Baghdad. We also aim to shed light on regional factors, such as consanguinity and genetic trends, among the patients.

PATIENTS AND METHODS:

This cross-sectional study was conducted from April 2022 to August 2023. Clinical patients with a diagnosis of genodermatoses were evaluated, and demographic data, clinical characteristics, and pedigree analysis were performed on them.

RESULT:

Generalised skin lesions were most common (55.6%), followed by the extremities (30.9%), the scalp/face (24.7%), and the trunk (17.3%). There was a strong association between consanguinity (66.7%) and a positive family history (54.3%), respectively. Patterns of prevalence were observed by sex and age.

CONCLUSION:

These results highlight the importance of genodermatoses as a health burden in Iraq in consanguineous families. According to the study, genetic studies and management initiatives are critical for implementing early diagnosis and management strategies.

KEYWORDS: Consanguinity; Dermatology; Epidemiology; Genodermatoses; Iraq.

[1]FIBMS Center of Dermatology, Medical City Teaching Hospital, Baghdad, Iraq.
[2]FIBMS Department of Dermatology and Venereology College of Medicine, Baghdad University, Baghdad, Iraq.

Iraqi Postgraduate Medical Journal, 2026; Vol. 25 (1): 48-53
DOI: 10.52573/ipmj.2025.156613 Received: January 13, 2025 Accepted: July 30, 2025

INTRODUCTION:

Genodermatoses are rare and often challenging for non-specialists to diagnose and manage. The classification of these disorders is crucial as they often affect not only the skin but may have systemic implications. A significant categorisation includes disorders of keratinisation, epidermal and dermal blistering disorders, and neoplasia-associated genodermatoses. Among the most common classifications are epidermolysis bullosa (EB), neurofibromatosis, and disorders of cornification. [(1)] Managing the phenotypic manifestations of these disorders requires advanced clinical expertise. The epidemiological and therapeutic profiles of genodermatoses vary by region, so it's essential to be aware of them. [(2,3)] Studies like Al-Hamami et al. [(4)] have shown Iraqi genodermatosis prevalence. Demographic trends, disease severity, and familial inheritance patterns are poorly analyzed. In Iraq, these disorders are poorly understood, and clinico-demographic data on affected patients is needed. Plázár emphasized the rarity and variability of genodermatoses, underscoring the need for further phenotypic and biochemical studies to refine clinical and genetic diagnostic criteria. [(5)] The non-skin involvement of genodermatoses poses the need to study clinico-demographic profiles of genodermatoses in Iraq, given that some genodermatoses, such as Griscelli syndrome, can be deadly [(6)]. Age, gender, and location can help healthcare professionals identify vulnerable populations and implement early intervention strategies. These studies also reflect epidemiological patterns in other regions to improve global genodermatosis knowledge.[(7, 8)] Due to health issues and limited resources, Iraq needs genodermatosis research. These studies can improve patient prognosis by influencing public health policy and resource allocation. Parker et al. suggest that multidisciplinary care for genetic skin disorders can enhance patient management in Iraq by promoting collaboration between research and clinical practice. [(9)] The rationale of this study is

that clinical-demography profiling of genodermatoses in Iraq at the Baghdad Dermatology Centre is an academic endeavour essential for improving patient care. By addressing knowledge gaps in health conditions, healthcare professionals can improve outcomes and quality of life for Iraqi patients with these disorders.

PATIENTS AND METHODS:

This descriptive cross-sectional study was conducted at Baghdad Dermatology Centre/Medical City from April 2022 to August 2023. Data from patients with genodermatoses clinically diagnosed were reviewed. Verbal consent was obtained from all patients and their parents, in the case of children, for the use of their images and clinical information. The inclusion criteria for the study consisted of patients with confirmed genetic dermatological disorders who met the criteria for a genodermatosis diagnosis and provided verbal consent to participate in the study. Participants were excluded if their medical records lacked the necessary data for the study or if they had psychological conditions that would hinder data collection. The diagnosis was based on routine clinical criteria and confirmed by board-certified dermatologists using international standards (as outlined in the 4th edition of Dermatology by Bolognia, J.L., Schaffer, J.V., and Cerroni, L) [10]. An inter-rater agreement with at least two dermatologists was preserved to ensure adequate diagnostic reproducibility. However, patients with psychiatric illnesses that would interfere with clinical assessment were excluded, which could introduce bias. Demographics, age of onset, illness course, family history, and parental consanguinity were recorded. All participants underwent pedigree analysis. Clinical examinations included cutaneous and extracutaneous manifestations, as well as the distribution and severity of lesions. Histopathological analysis was conducted when necessary; however, genetic analysis was not performed. Molecular diagnostics should be included in future studies due to their genetic nature, allowing for more accurate data on genodermatoses. Each case underwent the indicated investigations, which included a Routine blood examination, including a complete blood count (CBC), erythrocyte sedimentation rate (ESR), liver function tests (LFT), renal function tests (RFT), and a urine routine and microscopy. Imaging (ultrasonography, computed tomography, and magnetic resonance imaging) was done wherever required. SPSS 28 software was used for statistical analysis. Chi-square and Fisher's exact tests were used to evaluate the relationship between consanguinity and the prevalence of genodermatoses, and to compare differences based on sex and age, using chi-square tests. A p-value of 0.05 and below was considered significant.

RESULTS:

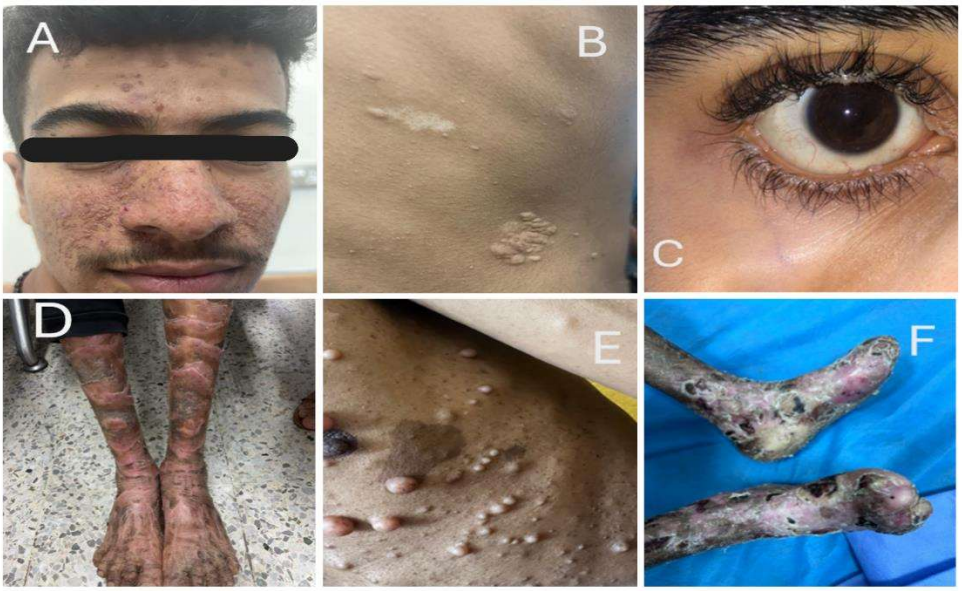
The study included 81 participants (mean age: 16.6 ± 13.5 years, range: 1–56 years). Men made up 50.6% and women 49.4%. Most were from Baghdad (97.5%), followed by Dyala and Wasit (1.2% each). A positive family history was found in 54.3% of cases, and consanguinity in 66.7%. Disease onset occurred at birth (38.3%), infancy (37.0%), childhood (14.8%), and adulthood (9.9%). Generalised skin lesions were most common (55.6%), followed by the extremities (30.9%), the scalp/face (24.7%), and the trunk (17.3%).

A highly significant association was found between categories of genodermatosis and the age of onset, as well as parental consanguinity ($p < 0.001$), suggesting a relationship between specific genetic skin disorders and their occurrence among consanguineous parents. Notably, we observed that genetic blistering diseases and disorders of cornification were overrepresented at early life stages, whereas the distribution of age groups was wider among disorders with malignant potential. Consanguinity rates were particularly high for genetic blistering diseases and disorders of cornification. In contrast, there was no statistically significant association between genodermatoses and gender ($p = 0.0503$) or family history ($p = 0.098$), suggesting that these factors may not be strong disease classifiers in this cohort. Such findings underscore the importance of being aware of the approaches to the clinical distribution of hereditary skin disorders by the age of onset and the relationship between affected siblings. (Table 1)

The figures (A - F) below show different genodermatosis subtypes found during the study.

Table 1: Distribution of Diseases according to sex, family history, consanguinity and age of onset.

	Gender		Age of Onset				Family Hx		Consanguinity	
	F	M	Birth	Infancy	Childhood	Adulthood	-ve	+ve	-ve	+ve
Acantholytic disorder	0 (0.0%)	5 (12.2%)	0 (0.0%)	0 (0.0%)	0 (0.0%	5 (62.5%)	4 (10.8%)	1 (2.3%)	5 (18.5%)	0 (0.0%)
Developmental abnormalities	1 (2.5%)	1 (2.4%)	2 (6.5%)	0 (0.0%)	0 (0.0%	0 (0.0%)	2 (5.4%)	0 (0.0%)	2 (7.4%)	0 (0.0%)
Disorder of cornification	8 (20.0%)	11 (26.8%)	8 (25.8%)	5 (16.7%)	6 (50.0%)	0 (0.0%)	9 (24.3%)	10 (22.7%)	8 (29.6%)	11 (20.4%)
Disorders of Connective Tissue	3 (7.5%)	2 (4.9%)	4 (12.9%)	0 (0.0%)	1 (8.3%)	0 (0.0%)	1 (2.7%)	4 (9.1%)	0 (0.0%)	5 (9.3%)
Disorders of Hair and Nails	2 (2.5%)	1 (2.4%)	0 (0.0%)	3 (10.0%)	0 (0.0%	0 (0.0%)	1 (2.7%)	2 (4.5%)	0 (0.0%)	3 (5.6%)
Disorders of Metabolism	0 (0.0%)	1 (2.4%)	0 (0.0%)	1 (3.3%)	0 (0.0%	0 (0.0%)	1 (2.7%)	0 (0.0%)	0 (0.0%)	1 (1.9%)
Disorders of Pigmentation	6 (15.0%)	6 (14.6%)	3 (9.7%)	5 (16.7%)	3 (25.0%)	1 (12.5%)	2 (5.4%)	10 (22.7%)	4 (14.8%)	8 (14.8%)
Disorders of Vascularization	3 (7.5%)	1 (2.4%)	3 (9.7%)	1 (3.3%)	0 (0.0%	0 (0.0%)	4 (10.8%)	0 (0.0%)	0 (0.0%)	4 (7.4%)
Disorders with Chromosome Abnormalities	1 (2.5%)	0 (0.0%)	1 (3.2%)	0 (0.0%)	0 (0.0%	0 (0.0%)	0 (0.0%)	1 (2.3%)	1 (3.7%)	0 (0.0%)
Disorders with Immunodeficiency	3 (7.5%)	2 (4.9%)	0 (0.0%)	5 (16.7%)	0 (0.0%	0 (0.0%)	2 (5.4%)	3 (6.8%)	0 (0.0%)	5 (9.3%)
Disorders with Malignant Potential	4 (10.0%)	5 (12.2%)	0 (0.0%)	6 (20.0%)	1 (8.3%)	2 (25.0%)	4 (10.8%)	5 (11.4%)	2 (7.4%)	7 (13.0%)
Disorders with Photosensitivity	1 (2.5%)	1 (2.4%)	0 (0.0%)	2 (6.7%)	0 (0.0%	0 (0.0%)	2 (5.4%)	0 (0.0%)	1 (3.7%)	1 (1.9%)
Disorders with Short Stature	3 (7.5%)	0 (0.0%)	3 (9.7%)	0 (0.0%)	0 (0.0%	0 (0.0%)	1 (2.7%)	2 (4.5%)	3 (11.1%)	0 (0.0%)
genetic blistering disease	5 (12.5%)	5 (12.2%)	7 (22.6%)	2 (6.7%)	1 (8.3%)	0 (0.0%)	4 (10.8%)	6 (13.6%)	1 (3.7%)	9 (16.7%)
Total	40 (100.0%)	41 (100.0%)	31 (100.0%)	30 (100.0%)	12 (100.0%)	8 (100.0%)	37 (100.0%)	44 (100.0%)	27 (100.0%)	54 (100.0%)



Figures A: angiofibromas in tuberous sclerosis.
B: Shagreen patch and ash leaf macules in tuberous sclerosis.
C: Beaded eyelid papules of lipoid proteinosis.
D: Plate-like scale in lamellar ichthyosis.
E: Café-au-lait macules, freckles & neurofibroma.
F: Mitten feet in a patient with autosomal recessive dystrophic epidermolysis bullosa

DISCUSSION:

Genodermatoses are a variety of genetic conditions that cause skin abnormalities and lower quality of life. The growing need for specialised dermatological care, especially in areas with limited access to dermatologists, underscores the importance of such studies.

A one-year study at our centre diagnosed 81 patients with genodermatoses. Al-Hamami et al.'s 2010 study, conducted at the exact centre, found that the frequency of genodermatoses among outpatient attendants was 83/ 20000 (0.42%). The fact that both studies were conducted over one year supports the incidence, suggesting that approximately 80 genodermatoses occur annually. The patients in our study averaged 16 years old, and 90% developed their condition during childhood or adolescence. This matches Al-Hamami et al.'s findings, where their ages at presentation ranged from 1 month to 60 years (median 10 years).[4] A separate study by Dalave et al. [6] in India aimed to identify the clinical-epidemiological features of genodermatoses in the pediatric age group and identified 35 clinically diagnosed cases within one centre over two years. Moreover, another study [11] conducted in India found that many cases occurred within the first decade of life. These findings collectively highlight the tendency for genodermatoses to manifest during childhood and adolescence, as observed in both our study and the study conducted in India. [11]

Our study's male-to-female ratio was almost equal, indicating no sex preference. We found a significant difference between these results and Al-Hamami et al.'s [4] two-to-one male-female ratio. Interestingly, Dalave et al. [6] found a one-to-one male-to-female ratio in pediatric patients. The patient population seeking consultation may affect sex differences in observed cases, so it must be considered when examining prevalence.

Given that genodermatoses are genetic disorders, the family history assessment is crucial to our study. In our study, approximately 54% of patients had a documented positive family history of genodermatoses. In contrast, the study by Al-Hamami et al. [4] at the exact centre revealed a lower percentage, with only 30% of their participants reporting a positive family history of the condition. This discrepancy in the rates of positive family history between the two studies underscores the variable hereditary nature of genodermatoses and the potential influence of genetic factors in different patient populations.

Our study's findings regarding the association between genodermatosis categories, age of onset, and parental consanguinity align with emerging literature on this subject. Specifically, our observation that genetic blistering diseases and cornification disorders are more prevalent at early life stages echoes trends documented in recent studies.[12-14] Cantile et al. clarify that genodermatoses exhibit a range of clinical manifestations associated with particular genetic mutations and their corresponding phenotypic expressions, emphasising the influence of hereditary factors, such as consanguinity, in these conditions. [12]

Moreover, the high consanguinity rates associated with genetic blistering disorders have been noted in recent studies, indicating an increased risk for these conditions in populations where close familial unions are common. This aligns with the findings of Ko et al. [13], who highlight the variability in clinical progression and age of onset for specific genodermatoses, emphasising the significance of genetic predisposition in the expression of these skin disorders. Similarly, a study by Frank et al. regarding the hereditary aspects of monogenic skin diseases reflects patterns of inheritance that may elucidate your findings on age and parental relations.[14]

In contrast to these associations, our results indicated no statistically significant correlations with gender or family history. This supports findings from other studies that highlight that while genetics plays a pivotal role in genodermatoses, environmental factors and other socio-demographic elements may also influence disease emergence and progression, albeit possibly to a lesser extent in specific populations. For example, Lehr et al. explore how genetic predisposition and the occurrence of autoimmune diseases can coexist in epidermolysis bullosa, suggesting multifactorial influences.[15]

Additionally, our study's indications of a wider distribution of age groups among disorders with malignant potential align with literature addressing the complexities involved in genetic risk factors for skin cancers within syndromic populations. Research by Vagher et al. discusses how hereditary syndromes can present significant variability in skin cancer manifestations, which parallels your observations regarding age distribution and potential malignancy risk.[16]

On another front, Subramanian's study discusses the increased prevalence of harmful homozygous variants in children of consanguineous parents, specifying that while some complex diseases show no significant association with consanguinity, the tendency for recessive

disorders remains evident.[17] This suggests a nuanced view where the type of genetic disorder can influence outcomes related to consanguinity, reflecting our findings that not all risk factors, such as family history or gender, show significant correlations in specific cohorts.

The authors were aware of some limitations, such as selection bias, because patients with severe or complex genodermatoses preferentially attend the Baghdad Dermatology Centre of Medical City. The study may not apply to all Iraqis, particularly those residing in rural or underserved areas. Patients with psychiatric illnesses that could interfere with clinical assessment were excluded to ensure accurate evaluations and data reliability. Cognitive or cooperation issues may bias symptom reporting and diagnosis. A lack of genetic testing limits the accuracy and specificity of diagnosis, as many of these disorders are genetic. Genetic testing was excluded due to cost and lab shortages. Future studies should utilise molecular diagnostics to enhance accuracy and gain a deeper understanding of the genetics underlying these disorders. In an epidemiological study, a sample size of 81 patients is considered small. Despite its limited generalizability, the sample provides meaningful statistical analysis for trends and associations.

CONCLUSION:

This study found that Genodermatoses are frequent in a specialised centre in Iraq and are linked to consanguinity and family history. These findings emphasise the significance of genetic studies in enhancing diagnostic accuracy and prompting treatment. Due to the single-centre study, multicenter studies are encouraged to represent a broader population. Policymakers may establish genetic counselling programs to promote safe genetics and reduce hereditary dermatological disorders in Iraq.

Authorship: All authors contributed equally to the study design, data collection, analysis, and manuscript preparation.

Funding: Nil.

Conflict of interest: Nil.

REFERENCES:

1. Spitz JL. Genodermatoses: A clinical guide to genetic skin disorders: Lippincott Williams & Wilkins; 2005.
2. Puttur N, Kapoor A, Lakhey K, Reddy A, Malik N, Deokar S. Investigating the Prevalence, Gender Predilection, and Inheritance Patterns of Genodermatoses: A Tertiary Hospital Study. Cureus. 2024;16(6):1-9.
3. Karabinta Y, Gassama M, Diakité B, Binta G, Keïta AS, Thiam H, et al. Epidemiological, Clinical and Therapeutic Profile of Genodermatoses in Mali. Journal of Dermatology Research and Therapy. 2020;6(2):1-6.
4. Al-Hamami HR, Noaimi AA, Al-Waiz MM, Al-Kabraty AS. Frequency of genodermatoses among Iraqi patients. Iraqi Postgraduate Medical Journal. 2010;9(1).
5. Plázár D, Meznerics FA, Pálla S, Anker P, Farkas K, Bánvölgyi A, et al. Dermoscopic Patterns of Genodermatoses: A Comprehensive Analysis. Biomedicines. 2023;11(10):2717.
6. Dalave K, Deora M, Sabhandasani S, Singh P, Mittal A, Shah B. Clinico-epidemiological study of genodermatoses in pediatric age group. International Journal of Research in Dermatology. 2019;6(1):34-42.
7. Ibrahim K, Owen C, Patel HP, May C, Baxter M, Sayer AA, et al. Can routine clinical data identify older patients at risk of poor healthcare outcomes on admission to hospital? BMC research notes. 2017;10:1-7.
8. Wiedermann CJ, Barbieri V, Engl A, Piccoliori G. Enhancing Healthcare Workforce Stability and Performance: Insights from a Cross-Sectional Relational Coordination Survey. Preprints. 2024;21:425.
9. Parker JC, Rangu S, Grand K, Bhoj E, Castelo-Soccio L, Sheppard SE. Genetic Skin Disorders: The Value of a Multidisciplinary Clinic. American Journal of Medical Genetics Part A. 2021;185(4):1159-67.
10. Bolognia J, Schaffer JV, Cerroni L. Dermatology. Fourth edition ed. Philadelphia: Elsevier; 2018.
11. Purkait R, Samanta T, Thakur S, Dhar S. Neurocutaneous syndrome: a prospective study. Indian J Dermatol. 2011;56(4):375-9.
12. Cantile T, Coppola N, Caponio VCA, Russo D, Bucci P, Spagnuolo G, et al. Oral Mucosa and Nails in Genodermatoses: A Diagnostic Challenge. Journal of Clinical Medicine. 2021;10(22):5404.
13. Ko CJ, Atzmony L, Lim YH, McNiff JM, Craiglow BG, Antaya RJ, et al. Review of Genodermatoses With Characteristic Histopathology and Potential Diagnostic Delay. Journal of Cutaneous Pathology. 2019;46(10):756-65.
14. Frank J, Has C, Betz RC. Selected Genodermatoses – Status Quo and Future Prospects. JDDG Journal Der Deutschen Dermatologischen Gesellschaft. 2023;21(4):337-41.

15. Lehr S, Felber F, Tantcheva-Poór I, Keßler C, Eming R, Nyström A, et al. Occurrence of Autoantibodies Against Skin Proteins in Patients With Hereditary Epidermolysis Bullosa Predisposes to Development of Autoimmune Blistering Disease. Frontiers in Immunology. 2022;13.
16. Vagher J, Gammon A, Kohlmann W, Jeter J. Non-Melanoma Skin Cancers and Other Cutaneous Manifestations in Bone Marrow Failure Syndromes and Rare DNA Repair Disorders. Frontiers in Oncology. 2022;12.
17. Subramanian S. The Abundance of Harmful Rare Homozygous Variants in Children of Consanguineous Parents. Biology. 2025;14(3):310.