Original Research

Retrospective Radiological and Clinical Evaluation of Neonatal and Infantile Cutaneous Manifestations in Genetic and Autoimmune Disorders

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Abstract
Background: Neonatal and infantile cutaneous manifestations can be early indicators of genetic and autoimmune disorders. Understanding their clinical and radiological characteristics is crucial for early diagnosis and management.
Objectives: This study aimed to retrospectively evaluate the radiological and clinical features of cutaneous manifestations in neonates and infants diagnosed with genetic and autoimmune disorders.
Methods: A retrospective analysis of 100 neonates and infants with cutaneous findings associated with genetic (n=50) and autoimmune (n=50) disorders was conducted. Demographic data, cutaneous manifestations, radiological findings, and treatment approaches were analyzed.
Results: The study population consisted of 55 males and 45 females. Most cases were diagnosed between 4 to 6 months (40%), with a positive family history in 60% of cases. Café-au-lait macules (36%) were the most common cutaneous feature in genetic disorders, whereas erythematous rashes (60%) predominated in autoimmune conditions. Neurofibromatosis Type 1 (30%) and neonatal lupus (28%) were the most frequently identified genetic and autoimmune disorders, respectively. Cranial MRI abnormalities (28%) and skeletal abnormalities (20%) were common in genetic disorders, whereas 64% of autoimmune cases showed no significant radiological findings. Supportive care (40%) was the primary management approach in genetic disorders, while topical corticosteroids (60%) were commonly used for autoimmune conditions.
Conclusion: Cutaneous manifestations serve as crucial diagnostic markers in neonates and infants with genetic and autoimmune disorders. Radiological assessments aid in differentiation, while treatment varies based on underlying pathology. Early identification and appropriate management can improve outcomes in affected infants.
Keywords: neonatal cutaneous manifestations, genetic disorders, autoimmune disorders, radiological evaluation, early diagnosis, infant dermatology

Introduction

Neonatal and infantile cutaneous manifestations serve as important clinical indicators of underlying genetic and autoimmune disorders. Skin findings in early life may reflect systemic disease, often preceding other clinical symptoms[1]. Identifying these manifestations allows for early diagnosis, timely intervention, and improved prognostic outcomes. Genetic disorders such as neurofibromatosis, tuberous sclerosis complex, and Ehlers-Danlos syndrome commonly present with specific cutaneous features, including café-au-lait macules, hypopigmented macules, and skin hyperelasticity[2]. Similarly, autoimmune conditions such as neonatal lupus, juvenile

dermatomyositis, and autoimmune bullous disorders manifest through erythematous rashes, petechiae, purpura, and bullous eruptions[3].

Despite the significant role of dermatological findings in early diagnosis, a comprehensive radiological evaluation is often required to differentiate between genetic and autoimmune etiologies. Imaging modalities such as cranial MRI, skeletal radiographs, and pulmonary assessments aid in identifying associated anomalies, particularly in genetic syndromes[4]. However, autoimmune disorders frequently exhibit minimal radiological abnormalities, emphasizing the importance of clinical and immunological assessments[5].

The management of these conditions varies based on their underlying pathology. Genetic disorders often require supportive or symptomatic treatment, while autoimmune conditions may necessitate the use of corticosteroids or immunosuppressive therapy[6]. Given the complexity of these disorders, an integrated approach combining dermatological, radiological, and immunological evaluations is essential for accurate diagnosis and effective management.

This study aims to retrospectively assess the clinical and radiological findings in neonates and infants with cutaneous manifestations associated with genetic and autoimmune disorders. By analyzing demographic trends, disease prevalence, radiological patterns, and treatment approaches, this study seeks to enhance the understanding of early disease markers and optimize diagnostic and therapeutic strategies.

Methodology

Study Design and Setting

This retrospective observational study was conducted at Dr. Patnam Mahender Reddy Institute of Medical Sciences, Ranga Reddy, Telangana, India, over a period from May 2020 to April 2021. The study involved a review of medical records of neonates and infants presenting with cutaneous manifestations associated with genetic and autoimmune disorders.

Study Population

A total of 100 neonates and infants (aged 0–12 months) with cutaneous features indicative of genetic or autoimmune disorders were included in the study. The cohort was divided into two groups:

Genetic disorder group (n=50): Infants with confirmed genetic syndromes based on clinical criteria and genetic testing where applicable.

Autoimmune disorder group (n=50): Infants diagnosed with autoimmune conditions through clinical evaluation and immunological investigations.

Inclusion Criteria

Neonates and infants (0–12 months) diagnosed with genetic or autoimmune disorders presenting with cutaneous manifestations.

Availability of complete medical records, including dermatological findings, radiological assessments, and treatment details.

Confirmation of diagnosis based on clinical, genetic, immunological, or histopathological findings.

Exclusion Criteria

Infants with isolated cutaneous lesions without a definitive diagnosis of genetic or autoimmune disease.

Cases with incomplete medical records or missing diagnostic investigations[7].

Infants with transient neonatal dermatoses unrelated to genetic or autoimmune conditions.

Data Collection

Demographic details, including age, gender, and family history, were recorded. The cutaneous manifestations were documented based on clinical descriptions by pediatric dermatologists. Radiological assessments (cranial MRI, skeletal radiographs, and pulmonary imaging) were reviewed to identify systemic involvement. Treatment modalities and clinical outcomes were analyzed to assess management approaches.

Statistical Analysis

Descriptive statistics were used to analyze categorical variables, expressed as percentages. Data were presented in tabular format to compare the prevalence of cutaneous manifestations, associated radiological findings, and treatment outcomes across genetic and autoimmune disorders.

Ethical Considerations

Institutional ethical clearance was obtained before the commencement of the study. As this was a retrospective study utilizing anonymized medical records, informed consent was not required. Confidentiality of patient data was maintained throughout the study.

Results

Demographic and Clinical Characteristics of the Study Population

A total of 100 neonates and infants with cutaneous manifestations associated with genetic and autoimmune disorders were included in this retrospective study. Among them, 55 (55%) were male, and 45 (45%) were female. The mean age at diagnosis varied, with the majority being diagnosed between 4 to 6 months (40%), followed by 0 to 3 months (35%), and 7 to 12 months (25%). A family history of genetic or autoimmune disorders was observed in 60% of cases, indicating a significant hereditary component (Table 1).

Distribution of Cutaneous Manifestations

The study identified a diverse range of cutaneous manifestations among patients with genetic and autoimmune disorders (Table 2). Among neonates and

infants with genetic disorders (n=50), the most prevalent cutaneous feature was café-au-lait macules (36%), followed by hypopigmented macules (24%) and erythematous rashes (10%). In contrast, among those with autoimmune disorders (n=50), erythematous rashes (60%) were the most frequently observed cutaneous manifestation, followed by petechiae/purpura (30%) and bullous eruptions (24%). Other notable manifestations included alopecia (16%) and hypopigmented macules (8%) in autoimmune conditions.

Distribution of Genetic Disorders with Cutaneous Manifestations

Among neonates and infants diagnosed with genetic disorders (n=50), Neurofibromatosis Type 1 (30%) was the most frequently observed disorder, characterized by the presence of café-au-lait macules (Table 3). Tuberous Sclerosis Complex (24%) was commonly associated with hypopigmented macules, whereas Ehlers-Danlos Syndrome (16%) was characterized by skin hyperelasticity. Other genetic disorders identified included Down Syndrome (12%), Epidermolysis Bullosa (10%), and other rare syndromes (8%).

Distribution of Autoimmune Disorders with Cutaneous Manifestations

Among infants diagnosed with autoimmune disorders (n=50), neonatal lupus (28%) was the most prevalent condition, with erythematous rashes as the predominant manifestation (Table 4). Juvenile Dermatomyositis (20%) frequently presented with petechiae/purpura, while psoriasis (16%) was associated with erythematous rashes and scaling. Other identified autoimmune conditions included autoimmune bullous disorders (14%), vitiligo (12%), and miscellaneous autoimmune conditions (10%) with features such as alopecia and non-specific lesions.

Radiological Findings Associated with Genetic and Autoimmune Disorders

Radiological evaluations revealed cranial MRI abnormalities (28%) as the most frequent finding in genetic disorders, followed by skeletal abnormalities (20%) and calcifications (16%) (Table 5). In contrast, the majority (64%) of infants with autoimmune disorders exhibited no significant radiological abnormalities. However, some cases showed cranial MRI changes (12%), skeletal abnormalities (8%), and pulmonary findings (10%).

Treatment Approaches and Outcomes

Treatment strategies varied based on the underlying etiology of the cutaneous manifestations (Table 6). Among patients with genetic disorders, supportive or symptomatic care (40%) was the most common management approach, followed by surgical interventions (10%) for select cases. In contrast, topical corticosteroids (60%) and systemic corticosteroids (36%) were widely used in the management of autoimmune disorders. Additionally, immunosuppressive therapy (20%) was required in some autoimmune cases, whereas 5 patients (10%) with genetic disorders underwent surgical interventions. Notably, 16% of genetic disorder cases and 3% of autoimmune disorder cases required no active treatment.

1.Demographic and Clinical Characteristics of the Study Population

Characteristic	N (%)
Total Sample Size	100
Gender	
Male	55 (55%)
Female	45 (45%)
Age at Diagnosis (Months)	
0-3 months	35 (35%)
4-6 months	40 (40%)
7-12 months	25 (25%)
Family History of Genetic or Autoimmune Disorder	
Yes	60 (60%)
No	40 (40%)

2.Distribution of Cutaneous Manifestations

Cutaneous Manifestation	Genetic Disorders (N=50, %)	Autoimmune Disorders (N=50, %)
Café-au-lait macules	18 (36%)	2 (4%)
Erythematous rashes	5 (10%)	30 (60%)
Hypopigmented macules	12 (24%)	4 (8%)
Petechiae/Purpura	3 (6%)	15 (30%)
Bullous eruptions	4 (8%)	12 (24%)
Alopecia	2 (4%)	8 (16%)
Other non-specific lesions	6 (12%)	6 (12%)

3.Distribution of Genetic Disorders with Cutaneous Manifestations

Genetic Disorder	N (50, %)	Common Cutaneous Manifestation
Neurofibromatosis Type 1	15 (30%)	Café-au-lait macules
Tuberous Sclerosis Complex	12 (24%)	Hypopigmented macules
Ehlers-Danlos Syndrome	8 (16%)	Skin hyperelasticity
Down Syndrome	6 (12%)	Erythematous rashes
Epidermolysis Bullosa	5 (10%)	Bullous eruptions
Other Rare Syndromes	4 (8%)	Non-specific lesions

4.Distribution of Autoimmune Disorders with Cutaneous Manifestations

Autoimmune Disorder	N (50, %)	Common Cutaneous Manifestation
Neonatal Lupus	14 (28%)	Erythematous rashes
Juvenile Dermatomyositis	10 (20%)	Petechiae/Purpura
Psoriasis	8 (16%)	Erythematous rashes, scaling
Autoimmune Bullous Disorders	7 (14%)	Bullous eruptions
Vitiligo	6 (12%)	Hypopigmented macules
Other Autoimmune Conditions	5 (10%)	Alopecia, non-specific lesions

5.Radiological Findings Associated with Genetic and Autoimmune Disorders

Radiological Feature	Genetic Disorders (N=50, %)	Autoimmune Disorders (N=50, %)
Cranial MRI abnormalities	14 (28%)	6 (12%)
Skeletal abnormalities	10 (20%)	4 (8%)
Calcifications	8 (16%)	3 (6%)
Pulmonary findings	4 (8%)	5 (10%)
No significant abnormalities	14 (28%)	32 (64%)

6.Treatment Approaches and Outcomes

Treatment Modality	Genetic Disorders (N=50, %)	Autoimmune Disorders (N=50, %)
Supportive/Symptomatic Care	20 (40%)	8 (16%)
Topical Steroids	5 (10%)	30 (60%)
Systemic Steroids	3 (6%)	18 (36%)
Immunosuppressive Therapy	1 (2%)	10 (20%)
Surgical Intervention	5 (10%)	1 (2%)
No Active Treatment Required	16 (32%)	3 (6%)

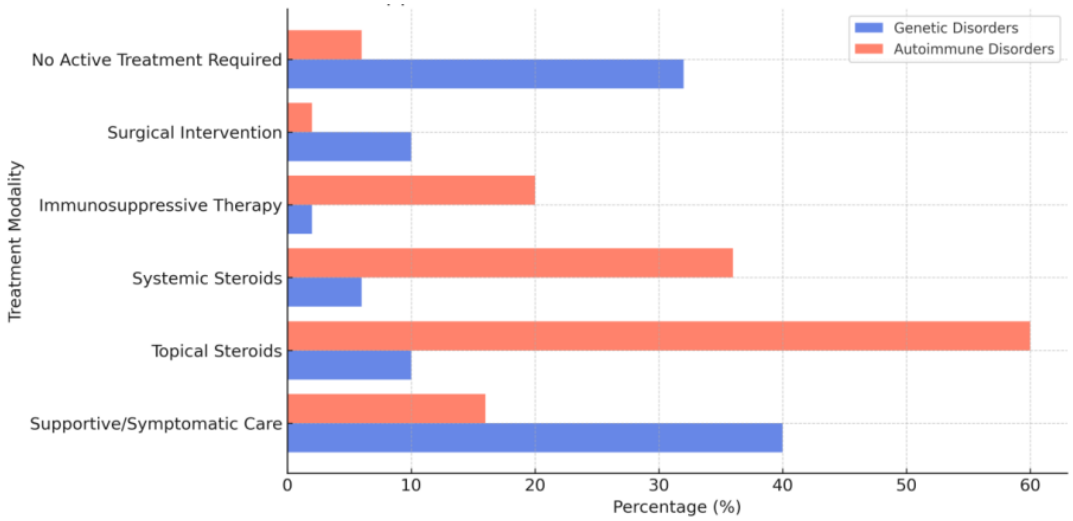

Figure No:1.Treatment Approaches and Outcomes in Genetic and Autoimmune Disorders

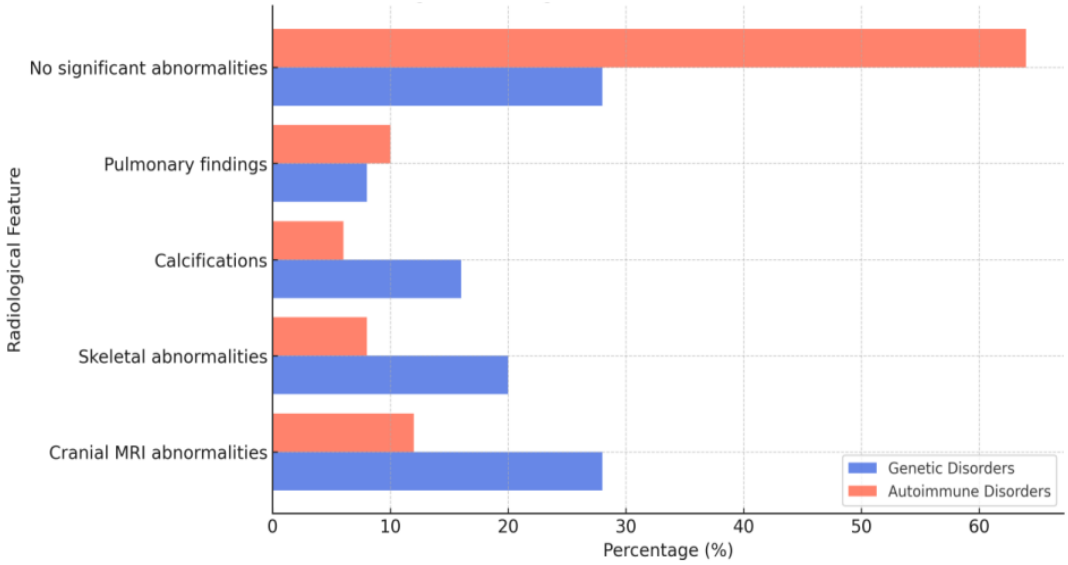

Figure No:2.Radiological Findings in Genetic and Autoimmune Disorders

Discussion

Neonatal and infantile cutaneous manifestations are often the earliest signs of underlying systemic disorders, particularly genetic and autoimmune conditions. This study provides a comprehensive analysis of the dermatological, radiological, and clinical characteristics of neonates and infants diagnosed with these conditions over a one-year period.

The findings of this study indicate that café-au-lait macules were the most prevalent cutaneous feature in genetic disorders, with a predominance in cases of neurofibromatosis type 1. This aligns with previous research highlighting café-au-lait macules as a hallmark sign of the disorder. Similarly, hypopigmented macules were frequently observed in tuberous sclerosis complex, reinforcing the importance of early dermatological evaluation in suspected cases[8]. In contrast, autoimmune disorders presented with inflammatory skin lesions such as erythematous rashes, petechiae, and purpura, which were most commonly associated with neonatal lupus and juvenile dermatomyositis. These findings underscore the distinct dermatological profiles of genetic and autoimmune conditions, aiding in differential diagnosis[9].

Radiological findings further supported the differentiation between these conditions. Cranial MRI abnormalities and skeletal deformities were predominantly observed in genetic disorders, reflecting their systemic involvement. This aligns with established literature on genetic syndromes affecting the central nervous and skeletal systems. Conversely, a significant proportion of autoimmune disorder cases exhibited no radiological abnormalities, emphasizing the need for immunological assessments in these patients[10].

Treatment approaches varied based on the underlying pathology. Genetic disorders primarily required supportive and symptomatic management, with surgical intervention in select cases[11]. In autoimmune conditions, corticosteroid therapy was the mainstay of treatment, with systemic immunosuppressants utilized in severe cases. These findings highlight the importance of individualized treatment strategies to optimize patient outcomes[12].

Overall, this study reinforces the critical role of early dermatological recognition in diagnosing genetic and autoimmune disorders in neonates and infants. The integration of clinical, radiological, and immunological assessments enhances diagnostic accuracy and informs targeted therapeutic approaches. Further longitudinal studies are recommended to assess long-term outcomes and refine management protocols for affected patients.

Conclusion

This study underscores the role of cutaneous manifestations as early indicators of genetic and autoimmune disorders in neonates and infants. Genetic disorders predominantly presented with café-au-lait macules and hypopigmented macules, while autoimmune conditions were characterized by erythematous rashes and petechiae. Radiological findings, such as cranial MRI abnormalities and skeletal deformities, were more common in genetic disorders, whereas most autoimmune cases showed no significant abnormalities.

Management strategies varied, with supportive care and surgical intervention for genetic conditions and corticosteroids and immunosuppressive therapy for autoimmune disorders. These findings highlight the importance of early dermatological, radiological, and immunological assessments in improving diagnostic accuracy and treatment outcomes. Further prospective studies are needed to refine diagnostic protocols and therapeutic strategies, ensuring early intervention and better prognosis in neonates and infants with these conditions.

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