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THE SPECTRUM OF GENODERMATOSES IN EARLY NEONATES OF WESTERN RAJASTHAN

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Abstract

Background & Aims - Genodermatoses are inherited skin disorders, presenting with multisystem involvement leading to increased morbidity and mortatlity. Many of these disorders are rare. This study is aimed to document the prevalence and diverse clinical presentations of various genetic skin disorders among early neonates from Western Rajasthan, India.

Methods – Five thousand early neonates delivered at tertiary care teaching hospital during March 2016 to Feb 2017 were included and detailed cutaneous and demographic features were studied.

Results – Total 54 neonates presented with genetic skin disorders out of 5000 under study. Prevalence of genodermatoses was found to be 1.08. The most common disorder was collodion baby in 47 neonates followed by harlequin baby in 5 neonates. History of consanguinity was positive in 26 (72.22%) cases, majority of them being from the Muslim community.

Limitation - Lack of genetic testing is the major pitfall of this study.

Conclusion – This is the first of its kind from this part of country, showing prevalence and pattern of genodermatoses in early neonates. Pre-marital genetic counseling can be beneficial in such cases. Prenatal diagnosis would be the first step for early detection of these genodermatoses.

Keywords – Genodermatoses, Collodion baby, Harlequin baby, Goltz syndrome, Incontinentia pigmenti

Introduction

Neonatal skin disorders are quite stressing, physiologically for the newborn and psychologically for the parents. The newborn skin can present with a vast range of conditions, from benign diseases to life-threatening ones. Genodermatoses are group of inherited disorders with cutaneous and systemic involvement causing increased morbidity and mortality.[1] Although, rarely seen, they are quite distressing for the paediatricians who may not be so familiar with skin changes.

The exact prevalence and burden of these diseases is still undefined in India. Only few case reports and series are reported in literature.

Hence, this prompted us to undertake a study to know the incidence and diverse clinical presentations of genodermatoses in early neonates in our area. To the best of our knowledge, this is a study, first of its kind in Western Rajasthan.

Material and method

A hospital-based prospective study of 5000 early neonates was conducted in the period of March 2016 to February 2017. All neonates delivered in tertiary care teaching hospital irrespective of gestational age, sex and mode of delivery were included in the study. Babies born outside the hospital and mothers unwilling to give consent for examination for their words were excluded.

After taking an informed consent from the guardians of the neonate, neonatal cutaneous findings were assessed within 7 days of birth. Detailed demographic data including parental consanguinity, family history and pedigree were recorded. Diagnosis was made on clinical grounds and when needed, relevant investigations were done. The observations pertaining to cutaneous parameters were expressed in percentage. The relationship between skin lesions and various maternal-neonatal aspects was calculated using Z-test, with $p \leq 0.05$ considered statistically significant.

Results

In our study 5000 early neonates were examined, out of them, 54 neonates (1.08%) had genodermatoses. There were more males (33/54) than females (21/54) presenting with genodermatoses. Out of 54 neonates, 32 (59.25%) were full term birth and 22 (40.74%) were preterm newborns. Thirty six (66.67%) mothers were Muslim and remaining 18 (33.34%) were Hindu. Twenty nine (53.70%) were of primiparous mothers whereas the remaining were of multiparous. The most common mode of delivery was normal vaginal delivery in 38 (70.37%) women, followed by Caesarean delivery in 16 (29.62%).

Most common genodermatoses was collodion baby in 47 (0.94%) neonates followed by harlequin baby in 5 (0.1%), Goltz syndrome in 1 (0.02%) and incontinentia pigmenti (IP) in 1 (0.02%) neonate. Out of 47 collodion baby 65.96% (31/47) were male. Amongst 5 harlequin baby 60% (3/5) were female neonates whereas single cases of Goltz syndrome and IP were females. (Figure 1-6)

In genodermatoses all changes were more common in low birth weight neonates except IP which was in normal weight neonate. Collodion baby, harlequin baby and Goltz syndrome were seen more in preterm neonates whereas IP nenonate was a full term baby. All genodermatoses were noted more common in multipara mothers, except harlequin baby which was noted in primipara mothers (80%).

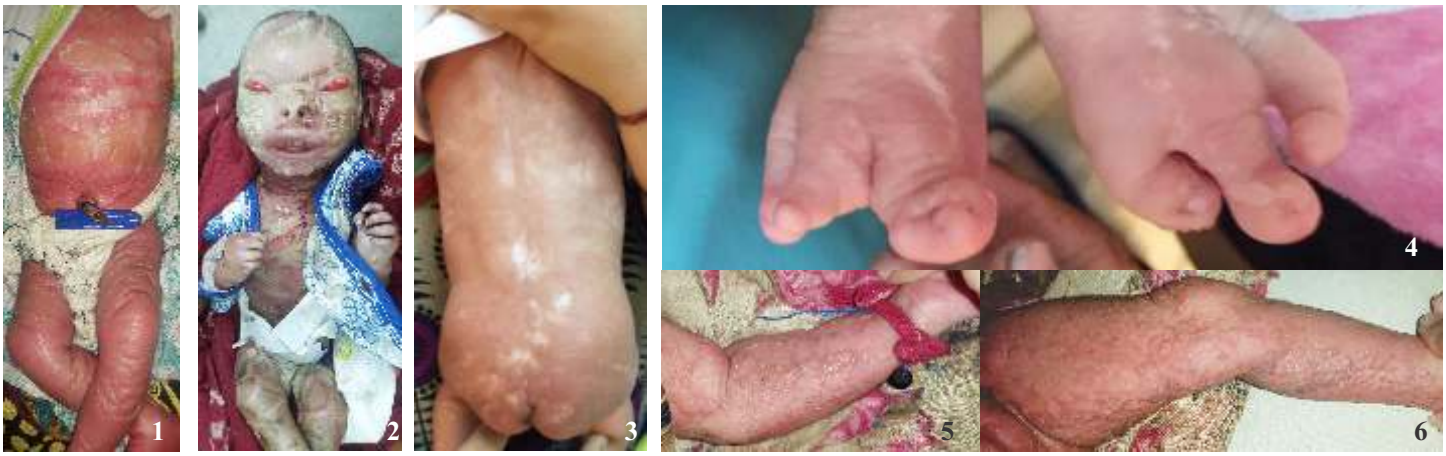

Figure 1: Collodion baby, **Figure 2:** Newborn showing extensive areas of diamond-like skin plates and fissuring characteristic of harlequin baby, **Figure 3:** Case of Goltz syndrome showing asymmetrical linear streaks of hypopigmented atrophic plaques which follows Blaschko's line. **Figure 4:** Case of Goltz syndrome showing ectrodactyly and syndactyly of fingers, **Figure 5 & 6:** Showing multiple linear eruptions of blisters on upper and lower limb along the Blaschko's lines in a newborn, typical of incontinentia pigmenti.

A positive history of the same disorder in the family was observed in 10/54 (18.51%) cases. Out of 36 Muslim neonates, parental consanguinity was noted in 26 (72.22%) neonates of genetic skin disorders. In collodion baby and harlequin baby consanguinous marriage history was statistically significant ($p<0.05$).

Table 1: Prevalence of genodermatoses in neonates

Pathological skin changes	Number of neonates
Collodion baby	47 (0.94%)
Harlequin baby	5 (0.1%)
Goltz syndrome	1 (0.02%)
Incontinentia pigmenti	1 (0.02%)

Discussion

The neonatal life is a phase of rapid adaptation in which the skin plays an important role. The neonatal integument may present with physiological skin changes, transient skin conditions; pathological changes like developmental malformation, genodermatoses, dermatitis, infections and iatrogenic disorders.

Genetic disorders may be grouped into three categories- Chromosomal (numerical-trisomy/monosomy or structural-translocations, deletions, and duplications), Mendelian (autosomal dominant, autosomal recessive or X-linked recessive genes) and Multifactorial.[2]

In our study, 5000 early neonates (the first 7 days of life) were thoroughly examined for genodermatoses related cutaneous changes with analysis of any association between neonatal and maternal factors.

Total 54 neonates with genetic skin disorders were seen among 5000 examined early neonates giving the prevalence of about 1.08 in our population. The exact incidence of these disorders has not been reported in the literature but it is thought that at least 1% of all live births had disorders inherited in a simple Mendelian fashion.[3] The study conducted by Kumar[4] et al showed genodermatoses with a prevalence of 0.62%. The commonest group of disorders was of ichthyosis vulgaris in Kumar[4] et al study whereas collodion baby in 47 neonates followed by harlequin baby in 5 neonates were the most prevalent in our study. Family history was positive (18.51%) in our study similar to other studies.[1,5] The high prevalence of inherited dermatoses amongst family members may be due to traditions that encourage the marriage of relatives. In our study, 72.22% of Muslim parents were married to the first or second-degree relatives which was also noted by Sameem[1] et al study. In this study, there was a male preponderance amongst genodermatoses sufferers similar to other studies.[5,6] Single case of Goltz syndrome and IP each, were noted in our study, both being X-linked dominant; lethal in male. IP neonate was term normal weight baby by a normal vaginal delivery similar to other case reports.[7,8]

All cases were diagnosed clinically and confirmation was done by histopathological examination if required. Therefore, obtaining the family history, consanguinity between the parents, and the presence of other skin disorders in offspring would be very helpful for early diagnosis of the genetic skin diseases. Mutation screening and genetic counseling of family members would be important, especially in families with a consanguinity.

Lack of genetic testing is the major pitfall of this study due to unavailability and prohibited cost in our Institutional setup.

Conclusion

The exact magnitude of genetic skin disorders are unknown because these are rare with increased morbidity and mortality. In this study, prevalence of genodermatoses was about 1.08 in our population. Ichthyosis group of disorders were most common among early neonates. Consanguineous marriage history was positive in 72.22% Muslim mothers. Prenatal diagnosis and genetic counseling are important tool for preventing these disorders.

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