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## A STUDY ON CASE SERIES OF NON BULLOUS CONGENITAL ERYTHRODERMA

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### ABSTRACT

**Aim of the study:**-The objective of present study is to study three cases of congenital non bullous erythroderma. **Place of study:** From the out patient department of Dermatology at Velammal Medical College Hospital, Vellamal Village, Madurai. **Period of Study:** Three cases were studied during December 2012—January 2013 in the above Hospital. **Case Reports:**-Three cases were studied in detail. All three were boys of paediatric age group between 3 years to 7 years of age. In the first case, the boy was three years old presented with dry scaly lesion all over the body with itching, it was noticed few weeks after birth by his mother. In the second case also, it was boy aged 6 years who also had dry scaly lesion all over the face, limb and at flexors and third case was a boy aged 7 years who happens to be second son born to his non consanguineous parents. They were examined in detail along with proper antenatal history of mother like drug history, exposure to radiation, consanguineous history, and history of similar complaints.

**Discussion:**-All three cases were well compared and correlated with available literatures.

**Conclusion:**-These cases of congenital lesions have high rate of morbidity and they are of paramount clinical importance. Hence studied and reported.

**Key words:** Congenital erythroderma, nonbullous ichthyosis, consanguineous parents, lamellar ichthyosis, Autosomal Recessive Congenital Ichthyosis (ARCI), Congenital Ichthyosiform Erythroderma

### INTRODUCTION

Nonbullous Congenital Ichthyosiform Erythroderma is also known as Congenital Ichthyosiform Erythroderma<sup>1</sup> It is rare variety having an incidence of 1 in 200,000 to 300,000 population live births. Symptoms of Congenital Ichthyosiform Erythroderma are similar to Lamellar Ichthyosis. So they are grouped under Autosomal Recessive Congenital Ichthyosis (ARCI) since many scientists consider them as variant of the diseases<sup>2</sup>. Many of them will not fit into definition of Lamellar Ichthyosis (LI) or Congenital Ichthyosiform Erythroderma (CIE). They have features of both diseases. They are extremes of LI and CIE<sup>(3), and (4)</sup>. There are numerous genetic

defects that can cause Congenital Ichthyosis Erythroderma (CIE). The noted genes are Transglutaminase -1 gene (TGM-1), Lipoxigenase gene (ALOX12B), the lipoxigenase -3, (ALOXE3) and the ABHD5 gene<sup>5</sup>. Any genetic defect in ABHD5 gene can give rise to congenital Ichthyosiform Erythroderma (CIE) known as **Neutral lipid storage disease** (Chanarin -Dorfman Syndrome)<sup>6</sup>.

### Case Reports of Three Cases

**Case No-1:**-A boy aged 7 years born to his non consanguineous parents had lesion of non bullous erythroderma present all over the body in the

form of thick hyperkeratotic, hyperpigmented adherent scales present since birth. This condition was noticed by his mother since long time. No history of similar complaints in the family.

On examination, he had lesions over the face, trunk and extremities. Even creases were not spared. This condition was exaggerated during winter but less during summer. There was no other associated congenital anomalies, coloboma of eyes, heart defects, mental retardation.

He was second child born to his non consanguineous parents. His brother was normal. No family history of similar complaints. There was no history of collodion baby.

**Case No -2:** Second case was also male age 5 years born to non consanguineous parents. He had 90% of his body surface area was covered with lamellar scales. They were fine and coarse in nature distributed all over the body involving flexures. There was no contracture, ectropion, eclabium, no cracks and fissures over the body covered by dry scaly lesions. The area of distribution was on face, trunk, extremities and flexures. ..Seasonal variation was present that is worse during winter, and better during summer.

On Examination, he also had generalised hyperpigmented fine scales predominantly seen on the face and over the back. He was first child born to his non consanguineous parents. No other anomalies seen over the body

**Case No- 3:-** was also seen in boy aged three years. He had dry scaly lesion present all over the body noticed few weeks after birth persisting through the year., worsening during winter. There was itching all over the body. His nails and hairs were normal. This case was not running in the family.

**On examination,** he had generalised hyperkeratosis, hyper pigmented, adherent scales more seen on trunk and both extremities. There was no history of similar complaints in the family. He was first child born to non

consanguineous parents. He had no other congenital lesions.

## DISCUSSION

From 1980 onwards, on bullous autosomal Recessive has been classified into two different major Ichthyosis., namely Nonbullous congenital ichthyosiform erythroderma (NBCIE) and Lamellar Ichthyosis (LI). The distinguishing features between the two are nature of the scaling and erythrodermal scaling. There is also an intermediate phenotype between two ichthyosis. In Nonbullous congenital ichthyosiform erythroderma (NBCIE), histologically, there is parakeratosis followed by inflammatory cell infiltration. While in Lamellar Ichthyosis., stratum corneum is usually thicker, Nonbullous congenital ichthyosiform erythroderma (NBCIE) and Lamellar Ichthyosis (LI). OR LI are formed by mutations of any three genes i.e., TGM1, ALOXE3, OR ALOX12B. But specific genes have not yet been identified.<sup>7</sup> In patients with lamellar ichthyosis have large dark scales covering whole body surface. There is involvement of facial skin giving rise to eclabium and ectropion associated with nail deformity. These patients do not improve with age. While in namely Nonbullous congenital ichthyosiform erythroderma (NBCIE), generalised erythema with fine white scales associated with mild ectropion and eclabium. There is alopecia with hyperkeratosis of skin of palm and soles. Here skin improves with the age during childhood and puberty.<sup>7</sup> In two families suffering from NBCIE erythroderma, there were scaling along with longitudinal striations of nails without any manifestations of collodion phenotype<sup>8</sup>. Many patients with collodion phenotype found to harbour pathogenic mutations in TGM1 genes encoding epidermal enzyme Transglutaminase 1<sup>(9,10)</sup>

**PRESENT STUDY:-** This is study of Congenital Nonbullous Ichthyosiform Erythroderma (CNIE)

observed in three boys. The ages of the boys were between three to seven year all born to their non consanguineous parents. Their mothers did not have systemic diseases nor, history of drug intake, history of any difficulty in labour, nor bad obstetric history

On examination of these children, all three had dry, dark hyper pigmented hyperkeratosis, scales distributed in major parts of the body i.e., face, trunk, extremities involving creases of the joints, more densely seen in the creases of elbow joint There was seasonal variations, worsening during winter less in summer, but scales were present all throughout the year. Itching was present all over the body. But there was no covering of thin transparent membranes as seen in collodion babies. Nor their bodies showed deep cracks and fissures, eclabium ("O" shaped fish like mouth) nor ectropion (eversion of eye lid) as seen in Harlequin babies. There were no associated anomalies involving other systems of the body.

### CONCLUSION

Nonbullous congenital ichthyosiform erythroderma (NBICE) is a congenital disorder present since birth, resulting in high rate of morbidity. It may be familial and may be due to genetic disorder.

Hence karyotyping, genetic mapping and genetic counselling has to be done to all patients. Genetic study reveals the type of genes causing skin anomaly. Counselling has to be done to avoid inbreeding (consanguinity). If familial, advice has to be taken from obstetricians, geneticists, which are necessary for the parents. Hence these cases have been studied and reported.

**TAKE HOME MESSAGE:** Avoid consanguineous marriages and consult geneticists if there is a familial history of anomalies.

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### REFERENCES

1. Freedberg et al (2003): Fitzpatrick Dermatology in General Medicine (6<sup>th</sup> edition) Mc Graw Hill .ISBN 0-07.
2. Lamellar Ichthyosis/CIE –New concepts, new mutations (<http://www.scalyskin.org/content.cfm?> (accessed on Jan 21, 2013))
3. Congenital Ichthyosiform Erythroderma ([http://www.scalyskin.org/conent.cfm?content\\_ID\\_-92&COLUMNid=14](http://www.scalyskin.org/conent.cfm?content_ID_-92&COLUMNid=14)) Types Ichthyosis (accessed on Dec 17, 2012)
4. Congenital Ichthyosis ([http://malattierare.pediatric.unipd.it/publica\\_MR/mr\\_dx\\_asp?mr=187](http://malattierare.pediatric.unipd.it/publica_MR/mr_dx_asp?mr=187)), Rare Disease Registry, University of Padua Italy
5. James William, Berger, Timothy, Elston, Dirk, (2005): Andrews Diseases of skin, Clinical Dermatology (10<sup>th</sup> edition) Saunders. ISBN 0-7216-2921-0.
6. OMIM (Online Mendelian Inheritance in man) John Hopkins University, Ichthyosiform erythroderma, congenital, nonbullous, I (<http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=242100>)
7. Akiyama M, Sawamura D, Shimizu H: The clinical pattern of nonbullous congenital ichthyosiform erythroderma and Lamellar ichthyosis: Clin Exp Dermatol: 2003; May 28(3):235-240.
8. Lesueur F, Bouadjar B, Lefevre C et al Novel mutation in patients with autosomal recessive

congenital ichthyosis and evidence for genetic heterogeneity on chromosome 17p13.1. *Invest Dermatol* 2007 ;127(4) 829-834.

9. Rahgunath M Hennies HC Ahvazi B et al. Self healing collodion baby: a dynamic phenotype explained by particular

transglutaminase -1 mutation. *J. Invest Dermatol* :2003;12(2)224-228.

10. Hennies HC, Kuster W Wiebe V et al: Genotype /phenotype correlation in autosomal recessive lamellar ichthyosis. *Am J Hum Genet* 1998 :62(5)1952—1061.

**Figure1: Showing Presence of Dry Scaly Skin In A Three Years Old Boy**



**Fig-2: Showing Scaly Lesions Seen in 6 Years Old Boy Involving Chest, Back And Extremities.**



**FIG-3; Showing Scaly Lesions Seen in 7 years Old Boy Involving , Back And Extremities**

