Respiratory Bronchiolitis With Christ-Siemens-Taurine Syndrome

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ABSTRACT

We the present the case of Respiratory Bronchiolitis in whom Christ Siemens Taurine syndrome was also diagnosed after stepwise approach, such kind of rare entity found in this part of the world should be an eye opener for every one.

Key words: Breathlesness,Congenital, Chromosomal ,Ectodermal Dysplasia,Bronchiolitis, Macules.

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Case report is Original: YES

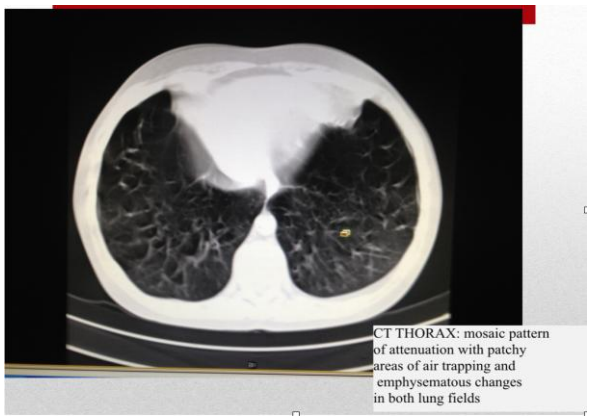
Whether case report publishes any where? NO

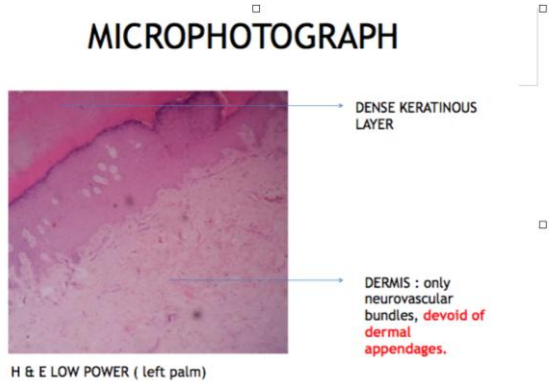
INTRODUCTION

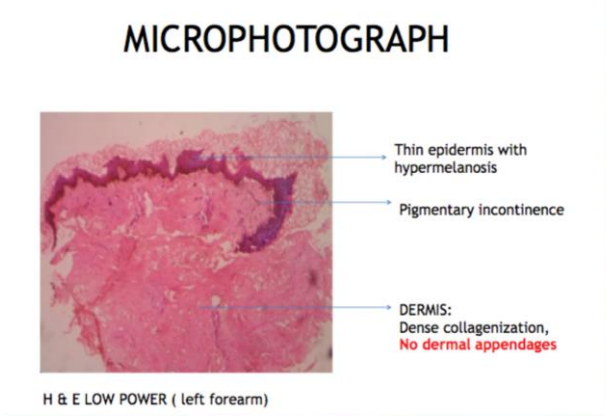
30 Year old male daily labour from East Godavari district came with a complaint of cough,breathlessness since 3 days.He had similar kind of repeated exacerbations since child-hood with hypo pigmented macules all over the body,Along with family history of similar illness in other siblings.His clinical examination and radiological picture was very nearly suggestive of respiratory bronchiolitis along with cutaneous involvement {anhydrotic ectodermal dysplasia} which we do not commonly see in this part of the world and should be an eye opener for every one.

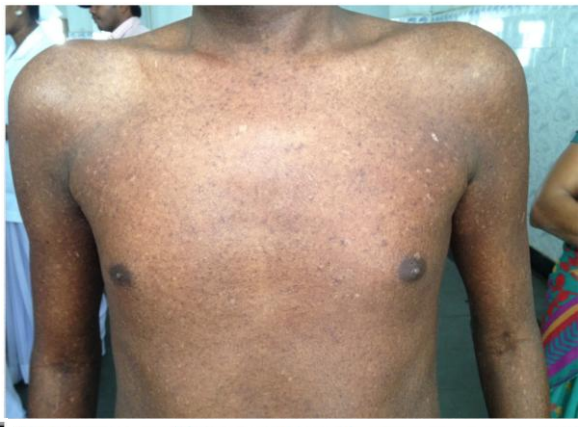
CASE REPORT

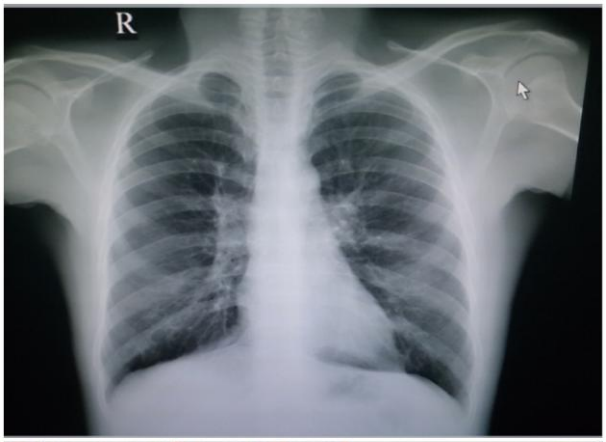
30 Year male came with a presenting complaint of breathlessness and dry cough,where the latter was insidious and progressive in nature,he had similar exacerbations atlas 5-6 times every year.Born to consanguinity and his sibling died at age of 5 with similar complaints,he had diffuse hypo/hyper pigmented macule over the body with dry skins described in fig 1 (a)(b)(c),clinical examination was normal,chest X-ray was within normal limits as shown in fig 2(b),CT showed mosaic attenuation of both lung fields with patchy areas of air trapping and emphysematous changes as described below in 3(a), 2d echo revealed mildly dilated rv ,alpha 1 AT levels were within normal ,pft showed mixed pattern.skin biopsy revealed ectodermal dysplasia 4(a)(b) and he had no chromosomal abnormalities.


CT THORAX: mosaic pattern of attenuation with patchy areas of air trapping and emphysematous changes in both lung fields


MICROPHOTOGRAPH
DENSE KERATINOUS LAYER
DERMIS : only neurovascular bundles, devoid of dermal appendages.
H & E LOW POWER (left palm)


MICROPHOTOGRAPH
Thin epidermis with hypermelanosis
Pigmentary incontinence
DERMIS:
Dense collagenization,
No dermal appendages
H & E LOW POWER (left forearm)

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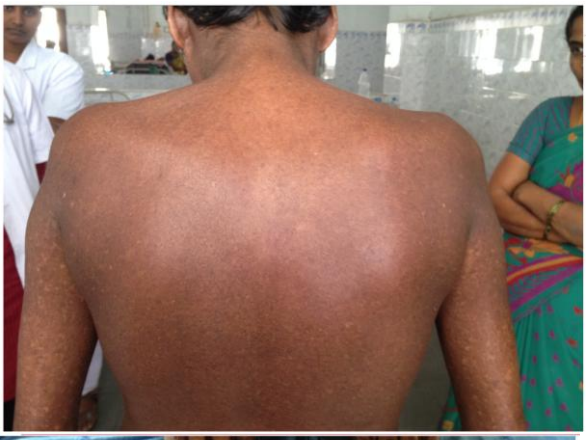

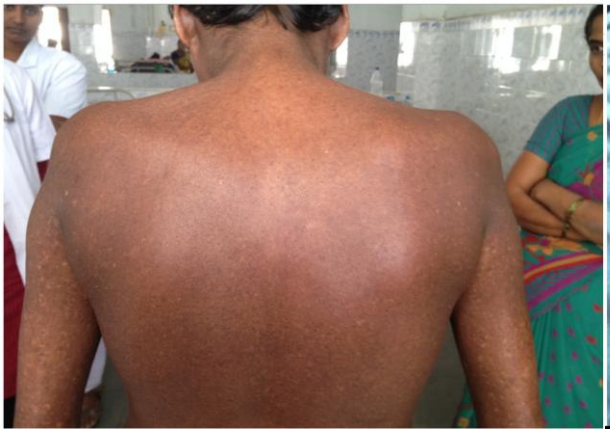

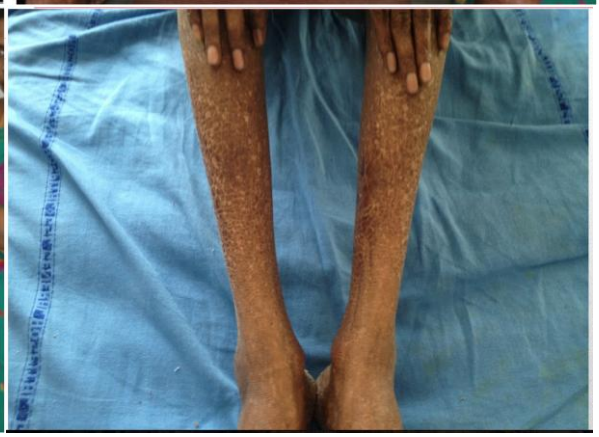

DISCUSSION

Bronchiolitis is pathologically defined as any inflammatory process that involves air-conducting passages measuring less than 2 mm in diameter. Clinical and pathologic manifestations of bronchiolitis are diverse. Depending on the nature of the underlying disease, it may be acute or chronic and present with obstructive or restrictive physiology
Ectodermal dysplasia (ED) is a genetic disorder that has congenital birth defects of two or more ectodermal structures. Ectodermal dysplasia is divided into two types hidrotic ectodermal dysplasia (Clouston syndrome) and hypohidrotic (anhidrotic) ectodermal dysplasia (Christ-Siemens-Touraine syndrome)

Hypohidrotic ectodermal dysplasia (HED) is an X-linked condition and is the most common form of ED. In this type of syndrome, there are no sweat glands or they are significantly decreased has characteristic (hypotrichosis), (asteatosis), (anhidrosis) [2].

CONCLUSION

Patients presenting with above clinical features should be thoroughly investigated for early & effective diagnosis

So this paper should act as an eye opener when faced with such rare cases, optimal treatment for these patients should require the multidisciplinary collaborative efforts of health professionals. .

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