

PERCUTANEOUS ULTRASOUND GUIDED FETAL SKIN SAMPLING A NEW APPROACH TO DIAGNOSE EPIDERMOLYSIS BULLOSA DYSTROPHICA LETHALIS.

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SUMMARY

An advanced and reliable technique of fetal skin sampling by the percutaneous Ultra-sound (U/S) guided technique has been developed at the All India Institute of Medical Sciences (AIIMS). This paper reports the first successful prenatal diagnosis of EPIDERMOLYSIS BULLOSA DYSTROPHICA (EBD) with Electron Microscopy of fetal skin obtained by this innovative method.

INTRODUCTION

Epidermolysis bullosa dystrophica (EBD) lethalis is a severe lethal skin disorder which is transmitted as an autosomal recessive condition. There is formation of subepidermal blisters which on Electron Microscopy shows separation at different levels in and around the basement membrane. Hypoplasia or absence of hemidesmosomes in basal layer in diagnostic.

No biochemical or molecular marker has yet been developed for prenatal diagnosis, which has been done world-wide by fetoscopic fetal skin sampling and electron microscopy.

This paper reports the first successful diagnosis of epidermiolysis bullosa lethalis by a more

sophisticated and safer technique : the percutaneous ultrasound (US) guided fetal skin biopsy, with Electron Microscopy of fetal skin.

Case Reports

1. Rekha - 20 yrs. G3 P2+0 with no living issue.
Obst. history :
 1. 1984 : FTND Pvt. Hosp. baby died after 2 months of E.B.D.
 2. 1986 : FTND Pvt. Hosp. baby died after 7 days of E.B.D.
 3. Present pregnancy

2. Sandhya :-
G2 P1+0 with no live issue Obst. History.
 1. 1980 LSCS for breech. Baby had EBD at birth, affecting whole body except face. Palms & soles were also involved. Toes of

(L) foot were self-mutilated, had syndactyly both hands. Baby died after 3 days.

2. Present pregnancy

TECHNIQUE

A new technique- the percutaneous ultrasound guided fetal skin biopsy has been developed at the AIIMS. This technique is essentially performed by the use of a fine 20 G cupped biopsy forceps 20 cms. long (0.8mm bore) passed through an 18 G guide needle with trocar 15 cms long (1.7mm bore) under ultrasound guidance. Fetal skin biopsies are then obtained from the limbs and trunk of about 17mm. diameter which are examined by Electron Microscopy (EM).

ELECTRON MICROSCOPY :

The specimen were processed routinely for electron microscopy. Ultra-thin sections were cut and studied in a Philips CM10 transmission electron microscope.

Both cases, on ultrastructural examination, showed intact basement membrane (Fig. 1) and intact anchoring fibrils (Fig. 2) attaching the basement membrane to the dermal collagen.

Follow Up

Case 1 She was lost to follow-up.

Case 2 She attended ANC regularly and delivered a healthy baby at term (Fig. 1)



Fig. 1 : Electron micrograph in a cord suspected of epidermolysis bullosa having intact basement membrane (arrow)

DISCUSSION

Several devastating and lethal skin disorders have no treatment as yet available, hence elimination of affected fetuses would be the earliest, safest, ethical and less psychologically trying alternative.

The method for diagnosing a lethal condition - epidermolysis bullosa dystrophica lethalis by fetal skin biopsy at 16-18 weeks pregnancy has been by the fetoscopic guided technique. This technique has an overall complication rate of 2-11% world-wide and 2.5% in the best centres.

EM examination of such minute fetal skin samples have already enabled antenatal diagnosis to be made of congenital bullous ichthyosiform erythroderma (epidermolytic hyperkeratosis) epidermolysis bullosa lethalis (Rodeck, 1980), epidermolysis bullosa dystrophica (Anton Lamprecht, 1981) and harlequin ichthyosis (Elias, 1980).

Epidermolysis bullosa dystrophica, is an autosomal recessively transmitted disease, the severest of the heterogenous group of congenital hereditary disorders of the skin, in which bullous lesions in the skin are induced by mechanical trauma and high temperature. Blisters occur over the peri-oral area, scalp, legs, diaper area and thorax, with sparing of the hands and feet except digits. There is growth retardation. Affected children may survive the second decade, but are

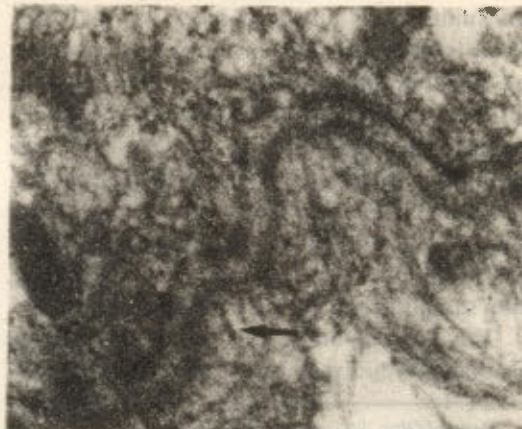


Fig. 2 : High powered view showing intact basement membrane and preserved anchoring fibrils (arrow)

seriously handicapped and incapacitated by adhesions, mutilations of hands and feet and mucus membrane lesions in mouth and esophagus.

The two cases studies at the AIIMS had 3 affected babies who died between 1 week to 2 months of life. Medical termination of pregnancy of affected, prenatally diagnosed fetuses is logically justified on medical, genetic and ethical grounds.

Percutaneous U/S guided Fetal Skin Biopsy is a safe and more advanced technique, added to the armamentarium of prenatal diagnosis.

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IMPACT OF ALTERED CONCEPTS IN THE MANAGEMENT OF INFERTILITY

B. Raman

SUMMARY

This article highlights the importance of updating and practicing the modern diagnostic technologies and the revised therapeutic measures. It could be realized that these modern concepts largely revolve around the fertility rate and permit the physician to be minimally invasive in diagnosis or therapeutic approaches.

part of female evaluation. (ii) Intercourse... (iii) Liberal application of sperm... (iv) In vitro fertilization... (v) In vitro fertilization with micromanipulation... (vi) In vitro fertilization with micromanipulation... (vii) In vitro fertilization with micromanipulation... (viii) In vitro fertilization with micromanipulation... (ix) In vitro fertilization with micromanipulation... (x) In vitro fertilization with micromanipulation...

INTRODUCTION
Since 1975, when we look back in the... (ii) In vitro fertilization with micromanipulation... (iii) In vitro fertilization with micromanipulation... (iv) In vitro fertilization with micromanipulation... (v) In vitro fertilization with micromanipulation... (vi) In vitro fertilization with micromanipulation... (vii) In vitro fertilization with micromanipulation... (viii) In vitro fertilization with micromanipulation... (ix) In vitro fertilization with micromanipulation... (x) In vitro fertilization with micromanipulation...