PERCUTANEOUS ULTRASOUND GUIDED FETAL SKIN SAMPLING

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

KAMAL BUCKSHEE . D. DEKA . MANOJ SINGH . ABHAY DHOND

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

Dept. of Obst. & Gynec. A.I.I.M.S., New Delhi Accepted for Publication on 26/8/91 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 dicd 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

203

JOURNAL OF OBSTETRICS AND GYNAECOLOGY

(L) foot were self-multilated, 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 CMIO transmission electron microscope.

Both cases, on ultrastructural examination, showed intact basement membrane (Fig. 1) and intact enchoring fibrils (Fig. 2) attaching the basement membrance 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 epidemolysis 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 emabled 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 hereditory 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 bissement membrane and preserved anchoring fibrils (arrow)

204

PERCUTANEOUS ULTRASOUND GUIDED FETAL SKIN SAMPLING

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.

REFERENCE

- 1. Anton Lamperecht I, Rauskolb R, Javonovic V, Kern B, Arnold ML, Schenck W, (Lancat 2: 1077, 1981)
- Elias S, Mazur M, Sabbagha R, Esterdy NB, Simpson L, 1980. Clinical genetics 17:275,1980
 - 3. Rodeck CH, Eady RAJ, Gosden CM, Lancet 1:949,1980

SUMMARY

This actuck highlight the importance of merging and practicing the modern dispositive technologies and the reduct therepeatic measures. It could be realised that these fundarm concepts forque machine ing the tertility gets, and permit the physician to be minimally for airs in diagnostic or therapeutic approaches.

INTRODUCTION

Since 1975, when we nook interest in the super speciality' of infertility, there have been infinitedom charges in and newer additions to diagrastic and therapeutic orwanic maximum. Ever mass this period we have been continuously everying our management protocoll in par with the modern technological developments. These evolutionary charges in the investigation and economics of infiniticity over a period of 17 years, conting with early 1989 lists been summarised in our varber communications (Rajen, 1989, and 1983).

Over the hart 2 years, ending with March, 1991, we have had the following changed converts in the management of infertility, which were not practical in my coeffer series. They include (DEndovaeinal concentrative as an inte-

the state of species space products and the species of the species

provide the second evaluation (10) transmitter manifering of the ideal its attacts actualitie within evolution of the ideal its attacts actualitie within bettin mutics even for subjects with slightest mentional affections, and affect histogizet much mentional affections, and affect histogizet much evites, and inner with "unceptioned infinities". (10) to minimize the quarity "diagramble lipervise open evited approach to improve open with an idea in a maximize the quarity "diagramble inprove open and transconist the operative inprove open in a transmitter particular with unlike intermole transconist the operative inprove open in call transconist the operative inprove open in a statement of insubjects with unlikering attach of randomic the parameter of the antiperior (v) particular (ciller endomicrices) at PID (v) particular (ciller endomicrices) at PID (v) of randomic for antiperior (viii) preference for endomic of randomic for antiperior (viii) endomice the overrow follice is patientian and duritering (or the overrow follice patientian and duritering (or the overrow follice patientian and duritering (or the overrow follice is patientian and duritering (or the overrow follice patientian and duritering (or the overrow follice is patientian and duritering (or the sonography is patienter of atomical patientian overrow following of atomogen (if) (if) produce opping overrow following (or or the atomogen (or (if) (or produce opping overrow following (or or the atomical and (or (or or or or overrow following) (or or other atomogen (or (if) (or other overrow following) (or other or other overrow following) (or other or other overrow following) (or other overrow over (or overr