GONADAL DYSGENESIS WITH CIRRHOSIS OF LIVER

(A Case Report)

by

MAYA C. LAKHANI

Introduction

Gonadal dysgenesis was first described by Morgagni in 1768. Later on several workers reviewed the literature and studied cases already reported and presented new cases. Here is reported one such case associated with cirrhosis of liver.

CASE REPORT

The patient, V. female aged 18 years presented with complaints of failure to menstruate, besides anorexia, vomiting and pain in abdomen since the age of 2 years. She was the eighth sibling. Her mother had died of cancer after a prolonged treatment when the patient was in her second year of life. She was prematurely born. Her father was an old man suffering from hypertensive heart disease.

The patient under report was a short statured, short necked, having a shield-shaped thorax cubitus valgus, and very short nails. Her arms' span was 57½" while her height was 56". Secondary sex characters were not developed, breasts being represented by 2 small nipples and the axillary and pubic hair being absent. Hypertrichosis was also there giving a hairy appearance. Her I.Q. was average. Liver was enlarged 2" below the right costal margin, firm, not tender. Pelvic examination revealed hypoplastic female external genitalia and small cervix and small uterus. Routine investigations gave normal findings. Karyopyknotic index seen in vaginal smear was 15, and cervical mucus failed to show fern pattern on drying. Endometrium was very scanty and showed proliferative phase.

From: Department of Obstetrics and Gynaecology, S.M.S. Medcal College (Zenana Hospital), Jaipur.

Accepted for publication on 23-4-80

Basal Body Temperature curve was uniphasic. Sex chromatin was positive for female. Dermal Ridge Counts were 169. Glucose tolerance test showed a hyperglycemic type of curve (fig. 1).

Glucose Tolerance Test of Case No Showing Hyperglycemic curve.

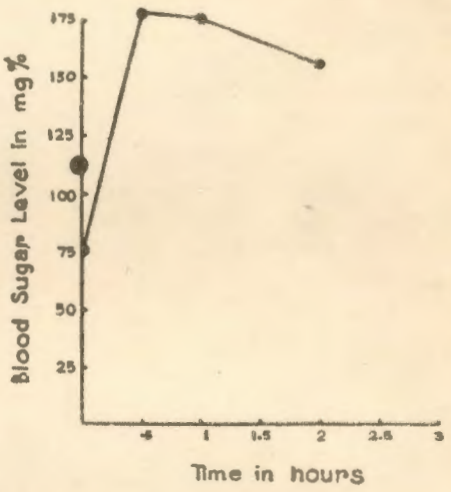

Fig. 1

The urinary 17-ketosteroids were 1.4 mg/24 hours, while the Iodine^{131} uptake study report was euthyroid. Liver functions were impaired in the form of raised alkaline phosphatase. Liver biopsy done on 19th December 1978 showed postnecrotic cirrhosis. X-Ray of skull showed normal sellaturcica.

Karyotyping and hormonal estimation of pituitary functions were not done because of lack of facilities.

Barium meal study revealed megalobulbus duodenum.

Comment

The term Gonadal Dysgenesis was imparted by Grumbach *et al*, in 1955, to a case with stunted growth, sexual infantilism, somatic abnormalities and streak ovaries. Various somatic and visceral abnormalities have been associated with the condition. But cirrhosis of liver associated with gonadal dysgenesis is unknown.

Initially, the cause was attributed to chromosomal pattern XO which was characteristic of Turner's syndrome (Ford and Jones, 1959). Ever since then however, many cases have been reported as gonadal dysgenesis with positive sex-chromatin bodies, which could be explained on the basis of mosaicism.

Dermal Ridge Count is a part of Dermatoglyphic and its clinical application has been suggested in 1966 by Valentine. In the present case it is 169 which is much above the normal counts (128) for a female. In the Valentine Series a case of Turner's Syndrome had 180 counts.

The mild oestrogenic effect and low 17-ketosteroid excretion in urine is suggestive of suppression of gonad and adrenal cortex by way of pituitary.

The presence of cirrhosis has worsened the prognosis of this case. Oestrogen could not be given to maintain normal vaginal mucosa and to induce menstruation because oestrogen is known to precipitate jaundice when given to women with chronic liver disease (Bearn *et al*, 1956 and Green and Rubin, 1959). She was discharged on good nutrition and anti-anemics only.

Summary

A case of gonadal dysgenesis with congenital cirrhosis of liver has been reported.

References

1. Bearn, A. G., Kunkel, H. G. and Slater, R. J.: Am. J. Med. 21: 3, 1956.
2. Ford, C. E. and Jones, K. W.: Lancet. 1: 711, 1959.
3. Green, P. and Rubin, L.: Am. J. Obstet. Gynec. 78: 141, 1959.
4. Grumbach, M. M., Van Wyk, J. J. and Wilkins, L.: Recent Progress Hormone Research. 14: 255, 1955.
5. Valentine, G. S.: Chromosome Disorders —An Introduction for Clinicians, 1st edition, William Hainemann Medical Books Ltd, London, 1966.