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GONADAL DYSGENESIS (TURNER’S SYNDROME) WITH ASSOCIATED LIVER DISEASE AND BLEEDING ESOPHAGEAL VARICES

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In 1938, Turner1 described a triad of infantilism with webbing of the neck and deformity of the elbow (cubitus valgus) in seven female patients. Their breasts, vaginae and uteri were infantile and menstruation had not occurred. In 1942, Varney, Kenyon and Koch2 described four young women with short stature and retarded sexual development who had high titers of urinary gonadotropin. This latter finding obviated hypopituitarism as the cause of the observed sexual infantilism. In the same year Albright et al3 reported on “primary ovarian insufficiency and decreased stature.” The frequency of associated congenital anomalies, especially webbing of the neck, cubitus valgus, coarctation of the aorta, late union of the epiphyses, osteoporosis, precocious senility and hypertension have been noted by many authors. This report concerns a case of gonadal dysgenesis with associated idiopathic liver disease and bleeding esophageal varices.

CASE REPORT

(V. M., Case number 198641). The patient is a 55-year-old unmarried, white female who presented herself to the Division of Gastroenterology in November of 1958. She had felt well until 5 days previously when she developed malaise, nausea and raised “coffee ground” emesis. Her stools had become darker than usual. She denied anorexia, pain and weight loss. There had been no change in her bowel habits. Interestingly, she related that she had been amenorrheic all her life.

On physical examination she appeared extremely pale and small in stature. (Fig. 1) She appeared older than her chronological age. Her height was 59 inches. She weighed 92½ pounds. Blood pressure was 120 systolic and 80 diastolic. Her pulse was 88 per minute. Temperature was 98.6 degrees F. The patient exhibited a kyphoscoliosis and deformity of both wrists caused by previous Colles’ fractures. Her skin was atrophic with several small scattered ecchymoses present. There was a trace of axillary hair and about one-third the normally expected adult pubic hair. Her body proportions appeared to be grossly normal. Breast development was infantile with poor areolar and nipple differentiation. Pelvic examination revealed an infantile vagina with a very small introitus. The ovaries and uterus could not be palpated rectally. Black stool was in the rectum. There was no webbing of the neck or valgus cubitus. Examination of the heart, lungs and abdomen was normal. Arterial spiders and palmar erythema were absent.

The patient was admitted to the hospital as a problem of gastrointestinal bleeding. The initial hemoglobin was 7.7 gm. per 100 ml. The leukocyte count was 10,000 per

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Figure 1
54 year old patient with gonadal dysgenesis. Height 59 inches. Note sparse pubic hair and premature senility.

Figure 2
Roentgenogram of distal esophagus showing esophageal varices in patient in Fig. 1.

cu. mm. The blood urea nitrogen and fasting blood sugar were normal. Gastric analysis showed normal gastric acidity. There was 12 per cent retention of brom-sulphalein dye after 45 minutes. The total serum cholesterol was 200 mg. per 100 ml. with an ester fraction of 55 per cent. An electrophoretic pattern of the serum proteins revealed a total protein of 5.8 gm. per 100 ml., albumin 3.49 gm., alpha-1 globulin 0.28 gm., alpha-2 globulin 0.49 gm., beta globulin 0.94 gm., and gamma globulin 0.59 gm. The alkaline phosphatase was 7.8 Bodanski units per 100 ml. Thymol turbidity was 2 units and the cephalin cholesterol flocculation was 0. The serum transaminase (SGOT) was 19 units. The serum calcium and phosphorus were normal. A 24 hour urine for follicle stimulating hormone (FSH) was positive at the level of 64 mouse uterine units. A male sex chromatin pattern was present on a buccal smear. Roentgen examination of the gastrointestinal tract showed esophageal varices. (Fig. 2) Marked osteoporosis and kyphoscoliosis of the dorsal and lumbar spine were evident on bone x-rays. Histological study of liver tissue obtained through percutaneous needle biopsy was interpreted as showing periportal fibrosis. (Fig. 3, 4)

The etiology of the patient’s liver disease and esophageal varices was an enigma. She did not use alcohol and her diet had always been excellent. She was employed as a clerical worker in a business office. There was no known exposure to recognized hepatic poisons. The patient was one of 8 offspring and was the shortest sibling. Her parents were of average height. Her birth weight was 5 pounds and she reported
that she had always been small and did not recall any period when growth ceased. She appeared to be of average intelligence and completed high school.

The patient had been seen at this hospital 25 years previously when she was age 30. She had caught her right hand in a washing-machine wringer. Fractures were absent by roentgen studies but it was noted that there was delayed union of the epiphyses of the distal radius and ulna. A short time later the patient reappeared in the Orthopedic clinic seeking examination for low back pain which had been present for about 3 years. Spine x-rays at that time were interpreted as showing marked osteoporosis and scoliosis. The fifth lumbar vertebra was slightly forward with respect to the body of the sacrum and was felt to represent prespondylolisthesis. The patient was not seen again until the above episode of bleeding from esophageal varices.

The patient received a total of 2000 ml. of whole blood during her hospitalization and her hemoglobin at time of discharge was 12.4 gm. per 100 ml. A therapeutic program consisting of diet, vitamins, oral estrogen and testosterone is anticipated.
Gonadal Dysgenesis

DISCUSSION

It has been surprising and interesting that an individual with the female or male chromosomal constitution (XX or XY) may still develop into either a male or female. Animal experimentation has demonstrated that early removal or destruction of the fetal gonads leads invariably to female development of the ducts and genitalia. This observation emphasized the importance of embryonic testes in counteracting the inherent tendency of the fetus to feminize. Individuals with "ovarian agenesis" had always been considered to be genetic females. In 1950 Wilkins recognizing the clinical significance of these animal experiments predicted that some patients with this disorder should be chromosomal males. The demonstration of a sex chromatin dimorphism by Barr and its relative easy determination through buccal smears has established that about 80 per cent of "girls" with Turner's Syndrome are genetic males. Objection has been raised to the term ovarian agenesis inasmuch as rudimentary mesonephric elements of the gonad are present in the genital ridge. It has been suggested that gonadal dysgenesis is more descriptive than ovarian agenesis.

The separation of this syndrome from pituitary dwarfism was greatly abetted by the discovery of high urinary gonadotropin levels. Other differentiating features are: (a) The individual are short rather than dwarfs (b) The bone ages are only slightly retarded than markedly so (c) These patients have a reduced amount of axillary and pubic hair rather than none at all (d) Estrogen therapy in these individuals leads to marked increase in axillary and pubic hair whereas it has no such effect in panhypopituitarism (e) These patients are quite strong and well nourished rather than weak and undernourished (f) These patients are prone to have other congenital anomalies (g) In an insulin tolerance test these patients exhibit normal hypoglycemia as opposed to hypoglycemia unresponsiveness.

Mellinger has written that the disorder is not familial and it is believed to result from an acquired defect of the fertilized ovum, with multiple developmental anomalies, including the failure of gonadal tissue differentiation. Experimental work with amphibians has shown that fertilized overripe eggs develop abnormally. Even if only mildly affected the germ plasm proves highly susceptible to the damage. Witchi et al believe that germinal degradation (blastophthoria) may be the primary cause of gonadal dysgenesis. Gonadal insufficiency may be but one of a series of congenital abnormalities occurring in the same individual.

The patient in this report is interesting because of the manifest absence of the usual etiologies for her liver disease. It is possible that her liver disease is present on a congenital basis as are her other anomalies. If this were the case however, one might have expected an earlier clinical appearance of her liver disease. The liver's failure to keep circulating estrogens at a normal level has been a popular hypothesis explaining the appearance of spiders. If this notion is correct one would not expect the appearance of spiders in this patient since the major source of her estrogens is congenitally absent.

SUMMARY

A case report of a patient with gonadal dysgenesis (Turner's Syndrome) and associated idiopathic liver disease and bleeding esophageal varices is presented.
REFERENCES


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