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# TURNER'S SYNDROME IN THE MALE

BY

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AND

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Turner's syndrome in the female (Turner, 1938 ; Wilkins and Fleischmann, 1944 ; Lisser *et al.*, 1947 ; Jackson and Sougin-Mibashan, 1953) is characterized by lack of development of the ovaries, short stature, webbing of the neck and other skeletal abnormalities. Ovarian agenesis means lack of Graafian follicles and hence an absence of both ova and female sex hormones (germinal and endocrinal deficiency). The affected person therefore is not fertile, does not menstruate, and lacks breast development, while her pituitary gland produces large amounts of gonadotropic hormone in an attempt to stimulate a non-existent end-organ.

## The Male Homologue

It is not so easy to define the homologous condition in the male, because in the testis the germinal tissue and the endocrine tissue (believed to be situated in the Leydig cells) are separate, so that a lack of development of either one or both might be claimed to represent the sexual side of "male Turner's syndrome." Engle (1950) suggested that an absence of tubular germinal epithelium should be the basic analogous condition. Sohval (1951), apparently agreeing with this, points out that there may be variable impairment of testicular secretory function in addition. There may thus be a *variability* in the presence and degree of hormonal disturbance in the "male Turner." The theoretical effects of pure germinal and pure endocrinal agenesis are shown in Table I.

TABLE I.—*Theoretical Features of Pure Germinal Agenesis, Pure Testicular Endocrinal Agenesis, and a Mixture of Both*

| | Germinal | Endocrinal | Mixed |
|---|---|---|---|
| Testes | Small | Normal | Small |
| Sperm | Azoospermia | Present (biopsy) | Absent |
| Penis | Normal | Small | Small |
| Potency | Potent | Impotent | Impotent |
| Body hair | Normal | Diminished | Diminished |
| Voice | Normal | High | High |
| Gynaecomastia | No* | May occur | May occur |
| Span | Normal | Large | Large |
| Osteoporosis | No | Yes | Yes |
| F.S.H. | Normal* | High | High |
| 17-Ketosteroids | Normal | Reduced | Reduced |

* These are doubtful because of the possibility that the germinal epithelium may also form a specific hormone (Sohval, 1951).

This consideration explains why the five cases of male Turner's syndrome so far reported (Flavell, 1943 ; McCullagh, 1948 ; Greenblatt and Nieburgs, 1948 ; Reforzo-Membrives *et al.*, 1949 ; Sohval, 1951)* are less homogenous than the female group, since they show varying degrees of endocrinal deficiency and will not, for instance, be expected necessarily to have high urinary gonadotropins. The K.R.A. syndrome (Klinefelter, Reifenstein, and Albright, 1942) is somewhat comparable, comprising azoospermia, small testicles, high urinary F.S.H., and gynaecomastia. The basic lesion is different—a hyalinization and sclerosis of tubules rather than a lack of development. This syndrome has been expanded by Heller and Nelson (1945) and Howard *et al.* (1950) to contain cases without gynaecomastia and with a variable degree of androgen insufficiency, while retaining the basic pathological change. As with the "male Turner," therefore, the K.R.A. syndrome becomes a resultant of mixed germinal and endocrinal deficiencies. The special features of Turner's syndrome in the male, as in the female, include shortness and stockiness in stature and the various skeletal anomalies of which webbing of the neck is most outstanding, while the gonadal lesion is one of lack of development.

Table II shows the nature of the "mixture" of germinal and endocrinal defects in reported cases of male Turner's syndrome and compares this with the K.R.A. condition. This table includes the following case, which we believe to be an example of male Turner's syndrome.

* Rossi and Caflisch (1951) mention other cases from the Continental literature, but we lack detailed information concerning these.

TABLE II.—*Features of the "Male Turner" and the Klinefelter–Reifenstein–Albright (K.R.A.) Syndrome*

| | Classical K.R.A. | Modified K.R.A. | Flavell's Case | McCullagh's Case | Greenblatt's Case | Reforzo-Membrives's Case* | Sohval's Case | Present Case |
|---|---|---|---|---|---|---|---|---|
| Testes | Small | Small | Small | Small | Small | Small | Small | Small |
| Sperm | Azoospermia | Azoospermia | — | — | Azoospermia | — | No semen | No semen |
| Penis | Normal | Variable | Normal | — | Large | Normal | Small | Normal |
| Potency | ,, | ,, | ,, | — | — | — | No ejaculation | Totally impotent |
| Body hair | Masculine | ,, | Sparse and female | — | — | — } Aged 10 | Sparse and female | Very hairy |
| Voice | ,, | ,, | High | High | — | — | Masculine | High-ish |
| Gynaecomastia | Yes | ,, | No | — | — | — | Yes | No |
| Stature | Normal | Normal | Short | Short | Short | Short | Short | Short |
| Span | Wide | Wide | — | — | — | — | — | Wide |
| Congenital anomalies | Nil | Nil | Webbed neck, cubitus valgus, cervical spina bifida | Webbed neck, cubitus valgus, microphthalmos | Webbed neck, cubitus valgus | Webbed neck, cubitus valgus, epicanthic folds | Cubitus valgus, cervical vertebral anomalies | Webbed neck, no valgus |
| Osteoporosis | No | No | — | — | — | Yes | No | Yes |
| 17-Ketosteroids | Normal | Low | — | Low | Low normal | Low | Low normal | Low |
| F.S.H. | High | High | — | Normal | Normal | ,, | High | High |
| Testicular biopsy | Tubular sclerosis and hyalinization; clumps of Leydig cells | | — | Hypoplasia, no Leydig cells | Tubular hypoplasia, Leydig cells present | As in pituitary dwarfism | Tubular aplasia, Leydig cells present | — |

The sign — denotes lack of information. * Two similar cases in early childhood (aged 7 and 3 respectively) have been reported by Cunningham and Harley (1951) and Dorff *et al.* (1948) without testicular biopsies.

### Case Report

A coloured houseboy aged 30, of simple, facile, cheerful, and deceitful character, came to hospital complaining of a vague respiratory illness. He had always been of small stature : height, 58½ in. (149 cm.) ; span, 60½ in. (154 cm.) ; lower segment, 29 in. (74 cm.) ; upper segment, 29½ in. (75 cm.). His neck was markedly webbed (Fig. 2). There was no cubitus valgus. His muscles were very well developed—a midget "Mr. Universe." Sexually he also appeared fully equipped (Fig. 1), yet he denied having ever experienced sexual feelings of any kind and stated he had never had an erection or an emission. (Curiously, one of the few reported male cases had an enlarged phallus (Greenblatt and Nieburgs, 1948) as a possible additional congenital abnormality.) We were unable to obtain a specimen of semen. There was some disagreement regarding the size and consistency of the testes — possibly smaller and firmer than usual. The prostate was small. The blood pressure was 145/75 mm. Hg.

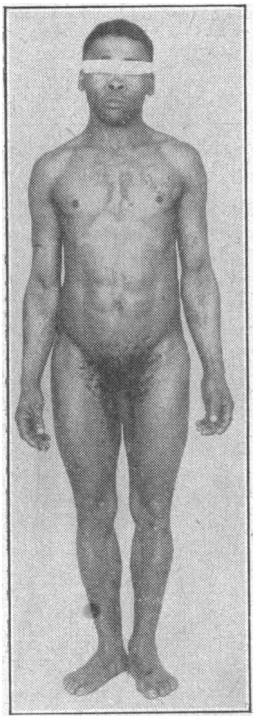

FIG. 1.—Short and muscular, with masculine stature and well-developed phallus.

Radiographs showed mild generalized osteoporosis. The 17-ketosteroids were 5.9 mg. per 24 hours (single estimation—method of Holtorff and Koch, 1940). The Wassermann reaction was negative. Urinary gonadotropins: 96 units per 24 hours (method of Klinefelter, Albright, and Griswold, 1943).

Unfortunately the crucial test —a testicular biopsy — was refused, and the patient left not only hospital but also the district with his employer's belongings before we could get urine to repeat our estimations. However, the high gonadotropins certainly confirm the presence of primary gonadal deficiency.

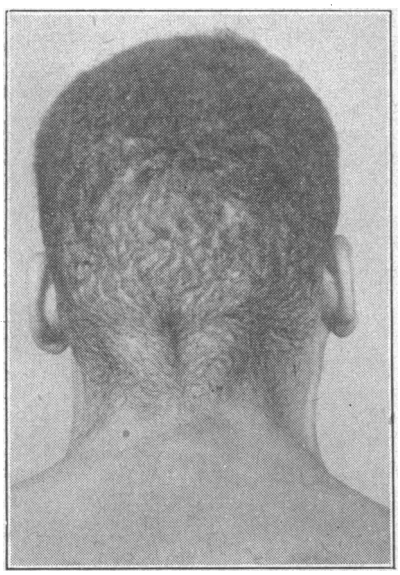

FIG. 2.—Note webbing of neck.

### Summary

We have observed a case representing Turner's syndrome in the male — the homologue of ovarian agenesis with short stature and congenital anomalies. We have made this an excuse for a consideration of what the male homologue should be and for a comparison of this with the Klinefelter–Reifenstein–Albright syndrome.

We conclude that the absence of homogeneity in the various reports of these conditions is due to the separation of endocrinal and germinal tissue in the male gonad, whereas in the female gonad one single tissue—the Graafian follicle—serves both endocrinal and germinal functions.

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# ELECTROLYTE IMBALANCE AFTER OPERATION FOR PITUITARY TUMOUR

BY

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A proportion of patients who have a pituitary adenoma or cyst present clinical evidence of hypopituitarism. Further light has been shed on this syndrome by Sheehan (1939) and Sheehan and Summers (1949), and by the development of biochemical tests of endocrine function.

In patients undergoing surgical treatment a hypopituitary crisis may be precipitated by the stress of the operation and by disturbance of function of the remaining pituitary tissue. This may prove fatal. Grant (1948), in his series of 71 cases, reported three in which a relatively simple operation was followed by stupor, coma, and death on the fifth, ninth, and twelfth days. At necropsy on two of these atrophy of the gonads and adrenal and thyroid glands was demonstrated.

The crisis may present with a variety of endocrine and biochemical disturbances, but is commonly characterized by the gradual onset of drowsiness progressing to stupor and coma, usually over a number of days. It is sometimes preceded by convulsions (Sheehan and Summers, 1952).

The management of hypopituitarism has advanced considerably in recent years. A combination of testosterone, thyroid, and deoxycortone acetate (D.C.A.) as substitution therapy is usually employed (Robertson and Kirkpatrick, 1951a), but cortisone and adrenocorticotrophic hormone (A.C.T.H.) have been used with more striking effect (Robertson and Kirkpatrick, 1951b ; Summers and Sheehan, 1951 ; Rolland and Matthews, 1952).

Treatment of the emergency of hypopituitary coma has been discussed by Sheehan and Summers (1952) :