Case Reports

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Turner's Syndrome with Menstruation

In 1938 Turner described a syndrome characterized by gonadal dysgenesis, primary amenorrhea, short stature, and various somatic malformations. About 20 cases of Turner's syndrome with short menstrual periods have been published (Zarate et al, 1969; Kaufman et al, 1971) and it seemed useful to report a rare case of Turner's syndrome with regular menstruation and a 45,X karyotype.

Case Report

The family history was unremarkable. After finishing elementary school with satisfactory results the patient, Margot A., a maid born 5 December 1950, now lives with her mother whom she occasionally helps with the house work.

Case History. No complications were reported until 1954 when dysuria occurred. Radiology of both kidneys showed a slightly impaired function of the left kidney while the right kidney showed no function at all. X-rays of the retroperitoneal cavity showed the left kidney as a markedly enlarged shadow and the right kidney appeared very large and shapeless.

Blood urea and uric acid serum levels were within the normal limits, and the blood pressure was not elevated. A right nephrectomy was performed on 10 January 1964 and a double kidney with mega-ureters removed; there were no postoperative complications.

Gynaecological History. Menarche commenced at 14 years. The cycle was 28 to 30 days with medium heavy bleeding which lasted 5 to 6 days.

Clinical Studies. When seen in 1970 this 20-year-old girl was 143 cm in height and weighed 59-9 kg; she was moderately obese. The following anomalies were noted; epicanthus, gothic palate, micrognathia, a suggestion of cutis laxa, funnel chest, hyperelasticity of the joints, bilateral cubitus valgus, and hypoplastic (pseudo-
hypertrophic) skeletal muscles. The clinical examination of the internal organs failed to reveal any patho-

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logical findings. Radiology of the hands showed no pathological finding.

**Gynaecological Examination.** The nipples were normally situated but the areolae were reduced; the mammary glands were slightly hypoplastic but clearly demarcated; the amount and the distribution of the axillary and pubic hair were within normal limits; the vaginal and the urethral orifices were normal as were the adnexa and uterus.

**Hormone Levels.** FSH 0-1 mU, total 17-ketosteroids 15 mg/24 hr. These are normal values for urinary excretion.

**Cytogenetics**

An analysis of 600 cells from smears taken from both sides of the buccal cavity showed no sex chromatin bodies. No drumsticks were seen in 1000 polymorphonuclear leucocytes.

Blood cultures were carried out by a modification of the method of Arakaki and Sparkes (1963), and fibroblasts were cultured from a skin biopsy taken from the left lower arm. The distribution of chromosome counts is summarized in the Table. A karyotype analysis of 20

<table>
<thead>
<tr>
<th>Tissue</th>
<th>No. of Chromosomes per Cell</th>
<th>No. of Cells Examined</th>
<th>No. of Cells Karyotyped</th>
<th>Karyotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lymphocytes from peripheral blood</td>
<td>44</td>
<td>44 45 46 46</td>
<td>103</td>
<td>31</td>
</tr>
<tr>
<td>Fibroblasts from skin biopsy</td>
<td>2</td>
<td>45</td>
<td>85</td>
<td>27</td>
</tr>
</tbody>
</table>

modal metaphase traits from both skin and blood cultures showed only 15 members of the C group to be present. All cells with chromosome numbers different from 45 were analysed and showed random chromosome loss. No late replicating C group chromosome was seen after 3 hours continuous labelling with "H-thymidine just before harvesting the blood cultures.

**Discussion**

Zarate *et al* (1969) and van Campenhout *et al* (1969) argue that the Turner phenotype is most likely a mixture of several diseases rather than a specific syndrome. Bahner *et al* (1960a) reported a patient with Turner's syndrome (45,X in some 100 skin fibroblasts and sternal bone marrow cells) who gave birth to a son. The karyotype in 21 fibroblasts of the ovary of this 40-year-old woman (height 136 cm) was found to be 45,X as well (Bahner *et al*, 1960b) and they concluded that a 'single X chromosome is capable of guaranteeing a full generative fertility'.

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*Fig. 1a* and *1b.* The proposita at 20 years. Note the malformation, epicanthus, and funnel-chest.
Case Reports

London et al (1964) recorded the case of a 28-year-old woman who had menstrual periods for 5 years and then became amenorrhoeic. He assumed that the XX cells had to be present for the ovaries to function regularly as indicated by normal menstruation. If there is a lack of these XX cells primary amenorrhoea will occur and if the XX cells are 'used up' then the amenorrhoea will be secondary.

Summary

A case is reported of a 20-year-old woman with a 45, X complement, with dwarfism and numerous other signs of Turner's syndrome, who has been menstruating for the past five years.

The question remains whether a single X-chromosome is sufficient for fertility or whether a mosaic pattern has to be suspected in the ovaries of fertile females with Turner's syndrome.

We are indebted to John L. Hamerton DSc for expert advice and to Dr Immo Lawaczek who referred the patient. Cytogenetic studies were performed by Dr Heiner Cramer at the Institute of Human Genetics, University of Marburg.

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Deletion of the Long Arms of Chromosome 18 (46,XX,18q–) Associated with Absence of IgA and Hypothyroidism in an Adult

Case Report

The patient (Fig. 1) is a female aged 21. No details of her birth are known beyond the fact that her mother was aged 35 years and died 8 years later of lung cancer. There is no information about the father.

The patient was admitted to a mental deficiency hospital at age 8 years. She was thought to suffer from atypical Down's syndrome.

On examination at the age of 21 years her weight was 142 cm with a sole to pubis measurement of 69 cm and a span of 137 cm; head circumference was 51 cm. She weighed 62.14 kg. Her face showed midfacial hypoplasia and a carp mouth. There were bilateral epicantthic folds and an internal, alternating strabismus. Her hair, which was fair in colour, had normal female distribution although her head hair was receding slightly at the temples. Her fingers were short and all had ulnar loops. She had generalized muscular hypotonia. Specialist investigations showed the eyes to have normal refraction. She had 50% perceptive deafness. The external meati and petrous bones were normal. The patient was hypotensive (BP 100/50). A soft systolic murmur was present following exercise and was best heard at the apex. The heart was enlarged on x-ray and an ECG showed probable right ventricular hypertrophy. There were no physical signs of heart failure, and no goitre.

Her intelligence fell into the group of mild mental retardation (International Classification of Diseases).

Protein Studies

Serum total protein was 7.9 g/100 ml. Electrophoresis on cellulose acetate was normal with the exception of a moderately increased γ-globulin level (2.0 g/100 ml).

The serum immunoglobulins were examined on 4 occasions. At no time was IgA detectable by immunoelectrophoresis (Grabar and Burtin, 1964) on agar gel against specific antisera (Behringwerke AG, Marburg-Lahn, Germany). The IgG and IgM arcs appeared qualitatively normal. The serum concentrations of immunoglobulins were measured by single radial immunodiffusion (Mancini, Carbonara, and Heremans, 1965) in agar gel containing specific rabbit antisera against human immunoglobulins (Wellcome Reagents Ltd, Beckenham, England). The Wellcome antiserum to IgA appeared to be specific as it gave, on immunoelectrophoresis, a single arc with normal human serum.

The results of the immunodiffusion measurements are shown in the Table. The IgA concentration (measured

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