CLINICAL CONFERENCE

GONADAL DYSGENESIS DIAGNOSED IN INFANCY

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ONE CAN hardly pick up a medical journal these days without finding one or more articles concerned with chromosomal abnormalities. The case presented here is that of a patient with gonadal dysgenesis (Bonnivie-Ullrich-Turner syndrome), which represents one interesting type of abnormality of the chromosomes.

CASE REPORT

The patient was a 11½-month-old white female infant, whose mother was gravida III, para III. The pregnancy was entirely normal, with no virus infections or irradiation during the first trimester. The patient had edema of the dorsa of the hands and feet at birth. When she was first seen at 23 months the edema of the hands had subsided and the skin was very lax. The dorsa of the feet still exhibited pitting edema. In addition, she had a short neck, low set ears, cutis laxa in the neck region, a shield-like chest with widely separated, hypoplastic nipples, a high arched palate, telangiectasia of the cheek and forehead, and hypoplasia of fingernails and toenails. The blood pressure was normal and femoral pulses were palpable. The external genitalia were normal.

Although these patients usually develop increased urinary gonadotropins at puberty, an investigator has reported1 three patients with gonadal dysgenesis who excreted increased amounts of gonadotropins in urine before puberty, the youngest being 2 years 8 months old.

Between 2 and 4 months of age the patient developed an infection of the urinary tract that indicated the possibility of a renal anomaly. Intravenous and retrograde urograms revealed normal findings, except for a bifid renal pelvis. A chromosomal count revealed five cells, with a diploid number of 45. Buccal smears from the parents were normal.

COMMENT

Turner2 described a syndrome of sexual infantilism, cubitus valgus and webbed neck in post-pubertal females. They had amenorrhea, no breast development, short stature and markedly elevated values for gonadotropins in urine. Diagnostic pneumoperitoneum or laparotomy in patients with this syndrome usually reveals an infantile uterus and tubes, and no ovaries. Various studies demonstrated that fibrous strands along the tubes contained ovarian stroma but no germinal epithelium or follicles. Congenital lymphangiectatic edema of the hands and feet, with cutis laxa and numerous other anomalies (Bonnevie-Ullrich syndrome) was considered a separate clinical entity until a few years ago. When it was found that about 80% of patients in both groups had a negative sex chromatin pattern,3 the two syndromes were combined as Bonnevie-Ullrich-Turner syndrome or gonadal dysgenesis.4,5 (See Table I for associated

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TABLE I
SOME RECENT DEVELOPMENTS INVOLVING ANOMALIES OF THE SEX CHROMOSOMES

<table>
<thead>
<tr>
<th>Clinical State</th>
<th>Chromatin Pattern</th>
<th>Sex Chromosomes</th>
<th>Autosomes (no.)</th>
<th>Diploid Number</th>
<th>References</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normal female</td>
<td>Pos (40%, cells)</td>
<td>XX</td>
<td>44</td>
<td>46</td>
<td>Tjio: Hereditas, 42: 1, 1956</td>
</tr>
<tr>
<td>Gonadal Dysgenesis (Bonnevie-Ullrich-Turner syndrome)*</td>
<td>Neg (80% pts)</td>
<td>XO</td>
<td>44</td>
<td>45</td>
<td>Ford: Lancet, 1: 711, 1959</td>
</tr>
<tr>
<td>Klinefelter's syndrome†</td>
<td>Pos</td>
<td>XXY</td>
<td>44</td>
<td>47</td>
<td>Jacobs &amp; Strong: Nature, 189: 304, 1959</td>
</tr>
<tr>
<td>With mongolism</td>
<td>Pos</td>
<td>XXY</td>
<td>45</td>
<td>48</td>
<td>Ford: Lancet, 1: 709, 1959</td>
</tr>
<tr>
<td>Primary amenity and micro-orchidism</td>
<td>Pos (40% double chromatin)</td>
<td>XXXY</td>
<td>44</td>
<td>48</td>
<td>Ferguson-Smith: Lancet, 2: 184, 1959</td>
</tr>
<tr>
<td>Double male (father XYY)</td>
<td>Pos</td>
<td>XYYY</td>
<td>44</td>
<td>48</td>
<td>Muldahl &amp; Ockey: Lancet, 2: 492, 1960</td>
</tr>
<tr>
<td>Triploidy‡</td>
<td>Pos</td>
<td>XXY</td>
<td>66</td>
<td>69</td>
<td>Book: Lancet, 1: 838, 1960</td>
</tr>
<tr>
<td>Superfemale or metafemale§</td>
<td>Pos in 71% cells: double chromatin in 13% cells</td>
<td>XXX</td>
<td>44</td>
<td>47</td>
<td>Jacobs et al.: Lancet, 2: 425, 1959</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Frazer et al.: Lancet, 2: 626, 1960</td>
</tr>
<tr>
<td>Testicular feminization,‖ or hereditary male pseudo-hermaphroditism¶</td>
<td>Neg</td>
<td>XY</td>
<td>44</td>
<td>46</td>
<td>Jacobs: Lancet, 2: 591, 1959</td>
</tr>
<tr>
<td>True hermaphroditism</td>
<td>Neg</td>
<td>XY/OX</td>
<td>44</td>
<td>45-46</td>
<td>Hirshhorn: Lancet, 2: 319, 1960</td>
</tr>
</tbody>
</table>

* Short stature, shield-like chest, overweight, cubitus valgus, abnormal facies, webbed neck, abnormal nails, lymphedema, cutis laxa, mental retardation, aortic coarctation, renal anomalies, telangiectasia and sexual infantilism
† Eunuchoid, small testes, mental deficiency.
‡ 1-year-old boy with lipomatosis, micrognathia, syndactyly of hands and feet, ataxia, retardation and porencephaly.
§ May have sexual infantilism and mental deficiency, ovaries postmenopausal. One mentally defective woman had a normal boy.
‖ Testicular feminization is included for comparison with gonadal dysgenesis.
¶ Female habitus, ectopic testes, inguinal hernia, primary amenorrhea, color blindness and mental deficiency.
congenital anomalies. It was thought that these patients were male pseudohermaphrodites. Recently, chromosomal counts revealed an XO pattern of the sex chromosomes, and this is now interpreted to mean that the patients are incomplete females, lacking the second X chromosome. Recent studies indicate that the sex chromatin body found in female cell-nuclei is probably due to a single X that is heterochromatic, i.e., stains differently from the other X chromosome and the autosomes. Perhaps it is this X that is missing in these patients, accounting for a negative chromatin pattern. However a 12-year-old patient with gonadal dysgenesis, chromatin-positive nuclei and an XO chromosome constitution has been reported. In addition, a female with XO sex chromosome constitution, no stigmata of Turner's syndrome except short stature and late menarche, and who gave birth to a normal male child has been described.

A summary of these variants and other abnormalities of the sex chromosomes is presented in Table I in an attempt to summarize some of the interesting new developments in the understanding of sexual anomalies.

REFERENCES


CLINICAL CONFERENCE: GONADAL DYSGENESIS DIAGNOSED IN INFANCY
Herbert I. Lerner
Pediatrics 1961;28:508

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