Chromosome Studies in 150 Sexually Abnormal Patients (A Summarized Report)\textsuperscript{1,2}

Part II

Sajiro Makino

Zoological Institute, Hokkaido University, Sapporo, Japan

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Part II describes cytogenetical and clinical features of 150 sexually abnormal patients data of which were collected during a period from September 1959 to July 1965 in the writer’s laboratory in collaboration with his co-workers and clinicians of various medical fields. General remarks and conclusion were given in Part I, together with some representative karyotypes and related references (Makino, S. 1967: Chromosome studies in 150 sexually abnormal patients (a summarized report), Part I. Cytologia 31: 309-323).

DESCRIPTION OF 150 CASES

Phenotypic males

Klinefelter’s syndrome with an XXXXY sex chromosome complement (1 case)

Case #1 Male, 12 years


Sex chromatin: One-positive 20.0\%, two-positive 33.6\%, three-positive 37.4\%, four-positive 2.1\%, and negative 8.0\%.

Chromosome counts:

\[
\begin{array}{ccccc}
<48 & 48 & 49 & 50 & >50 \\
\text{Blood} & - & 2 & 100 & 3 & - & 105 \\
\end{array}
\]


Klinefelter’s syndrome with an XXXY sex chromosome complement (1 case)

Case #2 Male, 16 years


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\textsuperscript{1} Contribution No. 770 from the Zoological Institute, Faculty of Science, Hokkaido University, Sapporo, Japan.

\textsuperscript{2} With the cooperation of Dr. Motomichi Sasaki in organizing data.

Sex chromatin: One-positive 34.6%, two-positive 42.6%, three-positive 1.6%, and negative 21.6%.

Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;47</th>
<th>47</th>
<th>48</th>
<th>&gt;49</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>1</td>
<td>96</td>
<td>3</td>
<td>100</td>
</tr>
</tbody>
</table>

Karyotype: 48-XXXY. Reference: Makino et al. 1964b

Klinefelter's syndrome with an XXYY sex chromosome complement (1 case)

Case #3 Male, 14 years
Clinical features: Height 162 cm. Weight 44 kg. Upper measurement 75 cm. Symphysis to sole 87 cm. IQ 63. Maternal age 38 years. Paternal age 39 years. Both parents and two siblings normal and healthy. Epicanthic folds noted in both eyes. Clinodactyly on the fifth fingers. No gynecomastia. Normal-sized penis and scrotum. Extremely small testes; right 1.7 x 1.1 x 1.0 cm, left 1.9 x 1.1 x 1.1 cm. Prostate not palpable. No pubic hair. Normal bone age. Testicular biopsy revealed a few hyalinized tubules showing thickening of the lamina propria, and a few tubules containing many Sertoli cells and a few spermatogonia. Leydig cells formed large clumps. 17-KS 2.27-4.28 mg/day. Gonadotropin 48 muu/day. 17-OHCS 0.96-3.54 mg/day.

Sex chromatin: One-positive 65.0%, two-positive 0.5%, and negative 34.5%.

Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;47</th>
<th>47</th>
<th>48</th>
<th>&gt;48</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>3</td>
<td>37</td>
<td>—</td>
<td>40</td>
</tr>
</tbody>
</table>


Klinefelter's syndrome with an XX/XXY sex chromosome complement (1 case)

Case #4 Male, 17 years

Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;45</th>
<th>45</th>
<th>46</th>
<th>47</th>
<th>&gt;47</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>3</td>
<td>13</td>
<td>20</td>
<td>—</td>
<td>36</td>
</tr>
</tbody>
</table>

Skin — 1 4 5 1 11


Klinefelter's syndrome with an XXY sex chromosome complement (29 cases)

Case #5 Male, 44 years
Clinical features: Testicular biopsy revealed small atrophic hyalinized tubules with atrophic spermatogenesis. 17-KS 11.23 mg/day. Gonadotropin 48 muu/day. Sex chromatin positive.

Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;46</th>
<th>46</th>
<th>47</th>
<th>&gt;47</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>1</td>
<td>16</td>
<td>—</td>
<td>17</td>
</tr>
</tbody>
</table>

Bone marrow — 1 16 — 17

Case #6 Male, 30 years
Clinical features: Underdeveloped external genitalia. Testes: right 1.8×0.9×1.2 cm, left 1.6×1.2×1.1 cm. Impotent. No child during four years married life. IQ 92. 17-KS 3.86 mg/day. Gonadotropin 80 muu/day.

Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;45</th>
<th>45</th>
<th>46</th>
<th>47</th>
<th>48</th>
<th>&gt;48</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>1</td>
<td>2</td>
<td>15</td>
<td>1</td>
<td></td>
<td>19</td>
</tr>
</tbody>
</table>


Case #7 Male, 25 years
Clinical features: Gynecomastia present. Penis of a normal size. Atrophic testes, 2.2×2.0×2.0 cm. Hypoplastic prostate. Pubic hair of a normal male type.

Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;46</th>
<th>46</th>
<th>47</th>
<th>48</th>
<th>&gt;48</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>1</td>
<td>46</td>
<td>1</td>
<td></td>
<td>48</td>
</tr>
</tbody>
</table>


Case #8 Male, 31 years

Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;46</th>
<th>46</th>
<th>47</th>
<th>48</th>
<th>&gt;48</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>3</td>
<td>25</td>
<td>2</td>
<td></td>
<td>30</td>
</tr>
</tbody>
</table>


Case #9 Male, 20 years

Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;45</th>
<th>45</th>
<th>46</th>
<th>47</th>
<th>48</th>
<th>&gt;48</th>
<th>total</th>
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<tbody>
<tr>
<td></td>
<td>1</td>
<td>1</td>
<td>31</td>
<td>1</td>
<td></td>
<td>34</td>
</tr>
</tbody>
</table>


Case #10 Male, 29 years

Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;47</th>
<th>47</th>
<th>48</th>
<th>&gt;48</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>33</td>
<td>2</td>
<td></td>
<td>35</td>
</tr>
</tbody>
</table>


Case #11 Male, 20 years

Chromosome counts:
Case #12 Male, 13 years
Clinical features: Height 153 cm. Weight 37 kg. Pubis to crown 68 cm. Pubis to sole 85 cm. IQ 68. Maternal age 32 years. Paternal age 40 years. Penis and scrotum normal size. No gynecomastia. Testes; right 2.9×1.8×2.0 cm, left 2.8×1.9×1.5 cm. Well-formed prepuberal seminiferous tubules with thickening of lamina propria and hyalinization. A few Leydig cells. 17-KS 5.6-6.07 mg/day. Gonadotropin 24-48 mIU/day. 17-OHCS 1.33-2.88 mg/day.

Sex chromatin: One-positive 57.5%, two-positive 4.5%, and negative 38.0%.

Chromosome counts:

<table>
<thead>
<tr>
<th>Blood</th>
<th>&lt;45</th>
<th>45</th>
<th>46</th>
<th>47</th>
<th>48</th>
<th>&gt;48</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>1</td>
<td>35</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>36</td>
</tr>
</tbody>
</table>


Case #13 Male, 13 years
Clinical features: Height 154.5 cm. Weight 37 kg. Upper measurement 73.5 cm. Symphysis to sole 81 cm. IQ 76. Maternal age 19 years. Paternal age 28 years. Penis and scrotum normal in size. No gynecomastia. Right testis 2.4×1.7×1.6 cm. Left testis, 2.5×1.8×1.5 cm. Well-formed prepuberal seminiferous tubules with thickening of lamina propria and hyalinization. A few Leydig cells. 17-KS 2.16-4.52 mg/day. Gonadotropin 24-48 mIU/day. 17-OHCS 1.24-1.55 mg/day.

Sex chromatin: One-positive 66.0%, two-positive 1.5%, and negative 32.5%.

Chromosome counts:

<table>
<thead>
<tr>
<th>Blood</th>
<th>&lt;45</th>
<th>45</th>
<th>46</th>
<th>47</th>
<th>48</th>
<th>&gt;48</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>3</td>
<td>36</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>40</td>
</tr>
</tbody>
</table>


Case #14 Male, 18 years

Chromosome counts:

<table>
<thead>
<tr>
<th>Blood</th>
<th>&lt;46</th>
<th>46</th>
<th>47</th>
<th>48</th>
<th>&gt;48</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>2</td>
<td>60</td>
<td></td>
<td></td>
<td></td>
<td>63</td>
</tr>
</tbody>
</table>


Case #15 Male, 22 years
Clinical features: Small penis, 4.0 cm long. Small testes; right 1.8×1.3×1.0 cm, left 1.7×1.2×1.0 cm. Atrophy of seminiferous tubules. Clumps of Leydig cells. Hypoplastic prostate. High excretion of gonadotropin. Cleft palate with hare-lip. Sex chromatin positive (63%). IQ 59.

Chromosome counts:

<table>
<thead>
<tr>
<th>Blood</th>
<th>&lt;46</th>
<th>46</th>
<th>47</th>
<th>&gt;47</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>1</td>
<td>31</td>
<td></td>
<td></td>
<td>32</td>
</tr>
</tbody>
</table>

Case #16 Male, 20 years
Clinical features: Small penis, 5.8 cm long. Small testes; right 1.5\times1.0\times0.8 cm, left 1.6\times1.0\times0.8 cm. Gynecomastia present. Hypoplastic prostate. Slight epicanthus. IQ 90. Sex chromatin positive (78%).

Chromosome counts:
\[
\begin{array}{cccc}
<47 & 47 & 48 & >48 \\
\hline
\text{Blood} & 34 & 1 & 35 \\
\text{Karyotype:} & 47-XXY. & \text{Reference:} & \text{Makino et al. 1964c.}
\end{array}
\]

Case #17 Male, 29 years
Clinical features: Small penis. Small testes; right 2.0\times1.5\times1.4 cm, left 1.7\times0.8\times0.8 cm. Left testes undescended. Atrophic seminiferous tubules. Hypoplastic prostate. Scanty pubic hair. Sex chromatin positive (47.5%). IQ 59.

Chromosome counts:
\[
\begin{array}{cccc}
<46 & 46 & 47 & >47 \\
\hline
\text{Blood} & 1 & 49 & 50 \\
\text{Karyotype:} & 47-XXY. & \text{Reference:} & \text{Makino et al. 1964c.}
\end{array}
\]

Case #18 Male, 23 years

Chromosome counts:
\[
\begin{array}{cccc}
<45 & 45 & 46 & 47 >47 \\
\hline
\text{Blood} & 1 & 1 & 28 & 30 \\
\text{Karyotype:} & 47-XXY. & \text{Reference:} & \text{Makino et al. 1964c.}
\end{array}
\]

Case #19 Male, 14 years

Chromosome counts:
\[
\begin{array}{cccc}
<45 & 45 & 46 & 47 >47 \\
\hline
\text{Bone marrow} & 1 & 2 & 12 & 15 \\
\text{Karyotype:} & 47-XXY. & \text{Reference:} & \text{Makino et al. 1960.}
\end{array}
\]

Case #20 Male, 53 years

Chromosome counts:
\[
\begin{array}{cccc}
<45 & 45 & 46 & 47 >47 \\
\hline
\text{Bone marrow} & 1 & 1 & 5 & 6 \\
\text{Karyotype:} & 47-XXY. & \text{Reference:} & \text{Makino et al. 1960.}
\end{array}
\]

Case #21 Male, 11 years
Clinical features: Normal external genitalia. Normal testes; right 2.4\times1.7\times1.5 cm, left 2.5\times1.6\times1.3 cm. Histologically normal for his age. High excretion of estrogen and gonadotropin. Hypoplastic prostate. Sex chromatin positive (65%). IQ 77.

Chromosome counts:
\[
\begin{array}{cccc}
<46 & 46 & 47 & >47 \\
\hline
\text{Blood} & 2 & 24 & 26 \\
\text{Karyotype:} & 47-XXY. & \text{Reference:} & \text{Takagi et al. 1966.}
\end{array}
\]
Case # 22  Male, 12 years
Clinical features: Normal external genitalia. Normal testes, 2.3×1.6×1.3 cm, with normal histology for his age. High urinary excretion of 17-KS, estrogen and gonadotropin. Sex chromatin positive (62%). IQ 69.

Chromosome counts:

\[
\begin{array}{cccc}
<46 & 46 & 47 & >47 \\
\text{Blood} & 8 & 61 & 69 \\
\end{array}
\]


Case # 23  Male, 14 years
Clinical features: Normal external genitalia with scanty pubic hair. Right testis 2.5×1.8×1.3 cm. Left testis 2.5×1.8×1.3 cm. Hyaline-degeneration of seminiferous tubules with increase of Leydig cells. Small prostate. High excretion of estrogen and gonadotropin. Sex chromatin positive (44%). IQ 78.

Chromosome counts:

\[
\begin{array}{cccc}
<46 & 46 & 47 & >47 \\
\text{Blood} & 2 & 39 & 41 \\
\end{array}
\]


Case # 24  Male, 15 years
Clinical features: Normal external genitalia without pubic hair. Right testis 2.0×1.5×1.3 cm. Left testis 2.0×1.6×1.0 cm. Hyalinized seminiferous tubules with numerous Leydig cells. Hypoplastic prostate. High excretion of estrogen and gonadotropin. Sex chromatin positive (61%). IQ 74. Clinodactyly of the fifth fingers.

Chromosome counts:

\[
\begin{array}{cccc}
<47 & 47 & >47 \\
\text{Blood} & 28 & 28 \\
\end{array}
\]


Case # 25  Male, 15 years
Clinical features: Normal external genitalia. Right testis 2.2×1.4×1.1 cm. Left testis 2.0×1.5×1.2 cm. Atrophic seminiferous tubules with many Leydig cells. High excretion of 17-KS, 17-OHCS, estrogen and gonadotropin. Sex chromatin positive (55%). IQ 76.

Chromosome counts:

\[
\begin{array}{cccc}
<46 & 46 & 47 & >47 \\
\text{Blood} & 3 & 40 & 43 \\
\end{array}
\]


Case # 26  Male, 28 years
Clinical features: Normal external genitalia. Pubic hair normal. Small prostate. Small testes; right 2.0×1.5×1.2 cm, containing scanty degenerated seminiferous tubules and clumps of Leydig cells. Sex chromatin positive (64.5%). Normal intelligence.

Chromosome counts:

\[
\begin{array}{cccc}
<46 & 46 & 47 & >47 \\
\text{Blood} & 2 & 18 & 20 \\
\end{array}
\]


Case # 27  Male, 29 years
Clinical features: Diagnosed as Klinefelter's syndrome. Clinical details not available.

Chromosome counts:
<46 46 47 >47 total
Blood — 2 40 — 42

Case #28 Male, 37 years
Clinical features: Diagnosed as Klinefelter's syndrome. Clinical details not available.
Chromosome counts:
<44 44 45 46 47 >47 total
Blood — 1 1 — 29 — 31

Case #29 Male, 25 years
Clinical features: No clinical data available.
Chromosome counts:
<47 47 48 >48 total
Blood — 13 1(?) — 14

Case #30 Male, 21 years
Clinical features: Small testes, 1.8×1.5×1.2 cm in both. Scanty pubic hair. Sex chromatin positive.
Chromosome counts:
<47 47 >47 total
Blood — 9 — 9

Case #31 Male, 25 years
Clinical features: Diagnosed as Klinefelter's syndrome. Details not available.
Chromosome counts: Mode at 47 in 20 cells counted.

Case #32 Male, 34 years
Clinical features: Diagnosed as Klinefelter's syndrome. Details not available.
Chromosome counts: Mode at 47 in 60 cells counted.

Case #33 Male, 37 years
Clinical features: Diagnosed as Klinefelter's syndrome. Clinical details not available.
Chromosome counts: Mode at 47 in 53 cells counted.

Suspected Klinefelter's syndrome with an XY sex mechanism (3 cases)

Case #34 Male, 32 years
Chromosome counts:
<46 46 47 >47 total
Blood — 9 1 — 10

Case #35 Male, 10 years
Clinical features: Underdeveloped external genitalia. Small testes. 17-KS 3.6 mg/day.
Gonadotropin 4 mu/day.

Chromosome counts:

<table>
<thead>
<tr>
<th></th>
<th>&lt;45</th>
<th>45</th>
<th>46</th>
<th>47</th>
<th>&gt;47</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood</td>
<td>4</td>
<td>24</td>
<td>1</td>
<td>1</td>
<td></td>
<td>29</td>
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</tbody>
</table>


Case #36 Male, 26 years
Clinical features: Data not available.
Chromosome counts: Mode at 46 in 10 cells counted.

Patients with hypospadias (9 cases)

Case #37 Male, 22 years
Clinical features: Hypospadias penoscrotalis. Urethral groove failed to close completely and opened in the peno-scrotal angle with chordee.
Chromosome counts:

<table>
<thead>
<tr>
<th></th>
<th>&lt;46</th>
<th>46</th>
<th>&gt;46</th>
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<tbody>
<tr>
<td>Blood</td>
<td>7</td>
<td></td>
<td></td>
<td>7</td>
</tr>
</tbody>
</table>


Case #38 Male, 14 years
Clinical features: Hypospadias. Palpable testes in hypoplastic scrotum. Small crooked penis, about 2.5 cm long. Prostate not palpated digitally.
Chromosome counts:

<table>
<thead>
<tr>
<th></th>
<th>&lt;44</th>
<th>44</th>
<th>45</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood</td>
<td>1</td>
<td>2</td>
<td>11</td>
<td></td>
<td></td>
<td>14</td>
</tr>
</tbody>
</table>


Case #39 Male, 5 years
Clinical features: Hypospadias penis. Abnormal urethral opening at the base of the crooked penis.
Chromosome counts:

<table>
<thead>
<tr>
<th></th>
<th>&lt;46</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
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<tr>
<td>Blood</td>
<td>31</td>
<td></td>
<td></td>
<td>31</td>
</tr>
</tbody>
</table>


Case #40 Male, 15 years
Clinical features: Hypospadias penis. Urethral groove failed to close completely and opened on the ventral surface of the penis with chordee.
Chromosome counts:

<table>
<thead>
<tr>
<th></th>
<th>&lt;44</th>
<th>44</th>
<th>45</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood</td>
<td>1</td>
<td>1</td>
<td>23</td>
<td></td>
<td></td>
<td>25</td>
</tr>
</tbody>
</table>


Case #41 Male, 6 years
Chromosome counts:

<table>
<thead>
<tr>
<th></th>
<th>&lt;45</th>
<th>45</th>
<th>46</th>
<th>&gt;46</th>
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</thead>
<tbody>
<tr>
<td>Blood</td>
<td>1</td>
<td>24</td>
<td></td>
<td></td>
<td>25</td>
</tr>
</tbody>
</table>

Case #42 Male, 15 years
Chromosome counts:
- Blood
  - <45: 1
  - 45: 49
  - 46: 50


Case #43 Male, 5 years
Clinical features: Hypospadia penis. Urethral opening at the base of the penis.
Chromosome counts:
- Blood
  - <45: 1
  - 45: 48
  - 46: 1
  - >47: 50


Case #44 Male, 30 years
Clinical features: Hypospadia.
Chromosome counts:
- Blood
  - <46: 22
  - 46: 1
  - >47: 23


Case #45 Male, 18 years
Clinical features: Hypospadia penis. Testes and scrotum normal in size. No other abnormality. Sex Chromatin negative.
Chromosome counts:
- Blood
  - <46: 7
  - 46: 1
  - >47: 8


Patients with cryptorchism (25 cases)

Case #46 Male, 18 months
Chromosome counts:
- Blood
  - <45: 1
  - 45: 50
  - 46: 1
  - >47: 52

Karyotype: 46-XY. One of no. 15 chromosomes had an unusually enlarged short arm. The same abnormality was found in the patient's father, paternal grandfather and two paternal aunts. Reference: Kato et al. 1965, Makino et al. 1966.

Case #47 Male, 6 years
Clinical features: Bilateral inguinal testes. Other external genitalia, normal as a male.
Chromosome counts:
- Blood
  - <46: 14
  - 46: 14
  - >46: 14

Case #48 Male, 5 years
Chromosome counts:

\[
\begin{array}{cccc}
<45 & 45 & 46 & >46 \\
\text{Blood} & - & 3 & 27 & - & 30
\end{array}
\]

Case #49 Male, 14 years
Clinical features: Bilateral undescended testes.
Chromosome counts:

\[
\begin{array}{cccc}
<45 & 45 & 46 & >46 \\
\text{Blood} & - & 3 & 27 & - & 30
\end{array}
\]

Case #50 Male, 2 years
Clinical features: Bilateral inguinal testes.
Chromosome counts:

\[
\begin{array}{cccc}
<45 & 45 & 46 & >46 \\
\text{Blood} & - & 2 & 64 & - & 66
\end{array}
\]

Case #51 Male, 5 years
Clinical features: Bilateral inguinal testes.
Chromosome counts:

\[
\begin{array}{cccc}
<46 & 46 & >46 \\
\text{Blood} & - & 32 & - & 32
\end{array}
\]

Case #52 Male, 7 years
Clinical features: Bilateral undescended testes. Further data not available.
Chromosome counts:

\[
\begin{array}{cccc}
<45 & 45 & 46 & >46 \\
\text{Blood} & - & 2 & 43 & - & 45
\end{array}
\]

Case #53 Male, 3 years
Clinical features: Bilateral undescended testes. Further data not available.
Chromosome counts:

\[
\begin{array}{cccc}
<45 & 45 & 46 & >46 \\
\text{Blood} & - & 1 & 18 & - & 19
\end{array}
\]

Case #54 Male, 5 years
Clinical features: Bilateral inguinal testes. Further details not available.
Chromosome counts:

\[
\begin{array}{cccc}
<46 & 46 & >46 \\
\text{Blood} & - & 25 & - & 25
\end{array}
\]
Case #55 Male, 33 years

Chromosome counts:

\[
\begin{array}{cccccc}
& <45 & 45 & 46 & 47 & >47 \\
Blood & 1 & 12 & 1 & - & 14 \\
\end{array}
\]

Karyotype: 46-XY.

Case #56 Male, 18 years
Clinical features: Left undescended testes, atrophic histologically. 17-KS 7 mg/day.

Chromosome counts:

\[
\begin{array}{cccccc}
& <46 & 46 & >46 \\
Bone marrow & - & 25 & - & 25 \\
\end{array}
\]


Case #57 Male, 6 years
Clinical features: Left undescended testis. Other external genitalia normal as a male.

Chromosome counts:

\[
\begin{array}{cccccc}
& <45 & 45 & 46 & >46 \\
Blood & 3 & 23 & - & 26 \\
\end{array}
\]


Case #58 Male, 5 years
Clinical features: Left undescended testis. General body weakness, slightly underdeveloped for his age. External genitalia normal.

Chromosome counts:

\[
\begin{array}{cccccc}
& <43 & 43 & 44 & 45 & 46 & 47 & >47 \\
Blood & - & 1 & - & 1 & 57 & 3 & - & 62 \\
\end{array}
\]


Case #59 Male, 22 years
Clinical features: Left undescended testis. Body hair growth normal as a male. Histologically the left testis infantile. Sex chromatin negative.

Chromosome counts:

\[
\begin{array}{cccccc}
& <46 & 47 & 47 & >47 \\
Blood & - & 26 & 1 & - & 27 \\
\end{array}
\]


Case #60 Male, 5 years
Clinical features: Left undescended testis.

Chromosome counts:

\[
\begin{array}{cccccc}
& <46 & 46 & >46 \\
Blood & - & 25 & - & 25 \\
\end{array}
\]


Case #61 Male, 11 years
Clinical features: Right abdominal testis.

Chromosome counts:

\[
\begin{array}{cccccc}
& <46 & 46 & >46 \\
Blood & - & 22 & - & 22 \\
\end{array}
\]

Case #62 Male, 15 years
Clinical features: Right inguinal testis. Details not available.
Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;44</th>
<th>44</th>
<th>45</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood</td>
<td>1</td>
<td>5</td>
<td>37</td>
<td>—</td>
<td>43</td>
</tr>
</tbody>
</table>
Karyotype: 46-XY. Reference: Makino et al. 1964d.

Case #63 Male, 15 years
Clinical features: Right inguinal testes. Further data not available.
Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;45</th>
<th>45</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood</td>
<td>5</td>
<td>52</td>
<td>—</td>
<td>57</td>
</tr>
</tbody>
</table>

Case #64 Male, 12 years
Clinical features: Right inguinal testis. Further details not available.
Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;46</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood</td>
<td>—</td>
<td>35</td>
<td>—</td>
</tr>
</tbody>
</table>

Case #65 Male, age unknown
Clinical features: Undescended testes. Further details not available.
Chromosome counts: Mode at 46 in 25 cells counted.

Case #66 Male, 10 years
Clinical features: Undescended testes. Further data not available.
Chromosome counts: Mode at 46 in 8 cells counted.

Case #67 Male, 11 years
Clinical features: Undescended testes. Further data not available.
Chromosome counts: Mode at 46 in 25 cells counted.

Case #68 Male, 7 years
Clinical features: Undescended testes. Further data not available.
Chromosome counts: Mode at 46 in 62 cells counted.

Case #69 Male, 52 years
Clinical features: Left undescended testis. Absence of right testis. Other external genitalia, normal as a male. 17-KS 3.85 mg/day.
Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;45</th>
<th>45</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood</td>
<td>2</td>
<td>19</td>
<td>—</td>
<td>21</td>
</tr>
</tbody>
</table>

Case #70 Male, 5 years
Clinical features: Left testis undescended. Absence of right testis.
Chromosome counts:

<45  45  46  >46  total
Blood -  1  23  -  24


Patients with gynecomastia (4 cases)

Case #71  Male, 13 years
Clinical features: Gynecomastia. Mental retardation. No symptom of Klinefelter's syndrome.

Chromosome counts:

<46  46  >46  total
Bone marrow -  12  -  12

Case #72  Male, 10 years
Clinical features: Unilateral gynecomastia, on the right breast, with marked fibrosis and hyaline degeneration. Normal external genitalia. Histologically normal testes. Drumstick positive. 17-KS 3.0mg/day.

Chromosome counts:

<45  45  46  47  >47  total
Bone marrow -  1  14  2 (? ) -  17

Case #73  Male, 21 years

Chromosome counts:

<45  45  46  >46  total
Blood -  1  22  -  23

Case #74  Male, 14 years
Clinical features: Gynecomastia. Atrophic left testis showing hypoplastic seminiferous tubules without sperm.

Chromosome counts:

<46  46  >46  total
Blood -  12  -  12

Patients with an XO/XY sex chromosome complement (3 cases)

Case #75  Male, 56 years
Clinical features: Apparently normal man with recently developed gynecomastia on both sides. Testis slightly larger than the normal and soft. He has fathered seven apparently normal children by two wives. Testicular biopsy showed an oligospermic picture. Sex chromatin negative.

Chromosome counts:

<44  44  45  46  47  >47  total
Blood -  3  11  45  -  1  60
Skin -  4  9  18  2  -  33
Case #76  Male, 28 years  
Clinical features: Height 180 cm. Weight 75 kg. Infantile external genitalia. No facial hair growth. Pelvis, shoulder and chest of a female type. Bone age of 16.5 years. Testicular biopsy revealed infantile tissue. One or two sections from both testes showed no evidence of the ovarian tissue. Sex chromatin negative.

Chromosome counts:  
\[ <44 \quad 44 \quad 45 \quad 46 \quad 47 \quad >47 \quad \text{total} \]
\[ \text{Blood} \quad 1 \quad 7 \quad 41 \quad 1 \quad 50 \]

Case #77  Male, 6 years  

Chromosome counts:  
\[ <44 \quad 44 \quad 45 \quad 46 \quad >46 \quad \text{total} \]
\[ \text{Blood} \quad 3 \quad 35 \quad 145 \quad 183 \]
Karyotype: 45-XO/46-XY. Reference: Makino et al. 1964c.

Miscellaneous sex abnormalities (19 cases)

Case #78  Male, 4 years  

Chromosome counts:  
\[ <46 \quad 46 \quad >46 \quad \text{total} \]
\[ \text{Blood} \quad 2 \quad 9 \quad 9 \]
Karyotype: 46-XY. Reference: Makino et al. 1962d.

Case #79  Male, 6 years  
Clinical features: Normal penis with underdeveloped scrotum. Right abdominal ectopic testis (1.4 x 0.8 x 0.6 cm). Left perineal ectopic testis (1.4 x 0.7 x 0.6 cm). Normal excretion of urinary hormones.

Chromosome counts:  
\[ <45 \quad 45 \quad 46 \quad >46 \quad \text{total} \]
\[ \text{Blood} \quad 2 \quad 15 \quad 17 \]

Case #80  Male, 17 years  
Clinical features: Eunuchoidism. Slight gynecomastia. Small penis, 1.5 cm long. Small testes; right 1.0 x 1.3 x 1.0 cm, left 1.5 x 1.0 x 0.5 cm, with hyaline-degenerated seminiferous tubules containing spermatogonia, clumps of Leydig cells and no sperm. Normal excretion of gonadotropin and estrogen but high 17-KS excretion.

Chromosome counts:  
\[ <46 \quad 46 \quad >46 \quad \text{total} \]
\[ \text{Blood} \quad 30 \quad 30 \]

Case #81  Male, 19 years  
Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;44</th>
<th>44</th>
<th>45</th>
<th>46</th>
<th>46</th>
<th>&gt;47</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood</td>
<td>—</td>
<td>2</td>
<td>3</td>
<td>25</td>
<td>1</td>
<td>—</td>
</tr>
</tbody>
</table>


Case #82 Male, 25 years

Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;44</th>
<th>44</th>
<th>45</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood</td>
<td>—</td>
<td>2</td>
<td>2</td>
<td>17</td>
<td>—</td>
</tr>
</tbody>
</table>


Case #83 Male, 6 years

Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;45</th>
<th>45</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood</td>
<td>—</td>
<td>1</td>
<td>15</td>
<td>—</td>
</tr>
</tbody>
</table>


Case #84 Male, 28 years

Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;46</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood</td>
<td>—</td>
<td>20</td>
<td>—</td>
</tr>
</tbody>
</table>

Karyotype: 46-XY. Reference: Makino et al. 1962d.

Case #85 Male, 5 years

Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;46</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bone marrow and blood</td>
<td>—</td>
<td>10</td>
<td>—</td>
</tr>
</tbody>
</table>


Case #86 Male, 28 years

Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;46</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bone marrow</td>
<td>—</td>
<td>21</td>
<td>—</td>
</tr>
</tbody>
</table>


Case #87 Male, 18 years
Chromosome counts:

<table>
<thead>
<tr>
<th>Case #</th>
<th>Diagnosis</th>
<th>Clinical features</th>
</tr>
</thead>
<tbody>
<tr>
<td>#88</td>
<td>Hypergonadotropic hypogonadism</td>
<td>Hypoplastic scrotum and penis. Eunuchoid proportions. Obesity. No pubic hair. 17-KS 4.1 mg/day. Gonadotropin 64 muu/day. 17-OHCS 6.1 mg/day. Diagnosed as hypergonadotropic hypogonadism.</td>
</tr>
<tr>
<td>#89</td>
<td>Hypogonadotropic hypogonadism</td>
<td>Eunuchoid proportions. Small penis, 4 x 1.5 cm. Gynecomastia. 17-KS 2.5-3.5 mg/day. Gonadotropin 12 muu/day. Sex chromatin negative. Diagnosed as hypogonadotropic hypogonadism.</td>
</tr>
<tr>
<td>#90</td>
<td>Hypogonadism</td>
<td>Undescended right testis. Underdeveloped penis and scrotum. Normal mentality.</td>
</tr>
<tr>
<td>#91</td>
<td>Hypogonadism</td>
<td>Infantile genitalia and small penis. Bone age of 14 years. Testicular biopsy revealed hyalinization of tubules, stroma filled with fibrous cells. Spermatogonia present but no spermatogenesis. Sex chromatin negative.</td>
</tr>
<tr>
<td>#92</td>
<td>Hypogonadism</td>
<td>Infantile genitalia and small penis. Bone age of 13.5 years. Testicular biopsy revealed infatile tissue, a few Leydig cells, no spermatogenesis. Sex chromatin negative.</td>
</tr>
<tr>
<td>#93</td>
<td>Hypogonadism</td>
<td>External genitalia apparently normal. Pubic hair of a female type.</td>
</tr>
</tbody>
</table>
Body and facial hair growth scanty. Sperm counts normal but sperms mostly non-motile. Sex chromatin negative.

Chromosome counts:
\[
\begin{array}{ccc}
<46 & 46 & >46 \\
Blood & 30 & 30 \\
\end{array}
\]

Case #94 Male, 18 years
Clinical features: Small testes. Pubic hair and axillary hair scanty.

Chromosome counts:
\[
\begin{array}{ccc}
<45 & 46 & >46 \\
Blood & 2 & 28 \\
\end{array}
\]

Case #95 Male, 1 year

Chromosome counts:
\[
\begin{array}{ccc}
<46 & 46 & >46 \\
Blood & 29 & 29 \\
\end{array}
\]

Case #96 Male, 6 months
Clinical features: Diagnosed as male pseudohermaphroditism. Clinical data not available.

Chromosome counts: Mode at 46 in 22 cells counted in leucocytes.


Patients with Azoospermia or oligospermia (17 cases)

Case #97 Male, 30 years

Chromosome counts:
\[
\begin{array}{ccc}
<45 & 45 & 46 & >46 \\
Blood & 3 & 37 & 39 \\
\end{array}
\]

Case #98 Male, 29 years
Clinical features: Azoospermia with a normal volume of semen. Penis and scrotum normally developed.

Chromosome counts:
\[
\begin{array}{cccc}
<45 & 45 & 46 & 47 & >47 \\
Blood & 3 & 18 & 2 & 23 \\
\end{array}
\]

Case #99 Male, 30 years
Clinical features: Azoospermia. No child during three years of married life.

Chromosome counts:
\[
\begin{array}{cccc}
<44 & 44 & 45 & 46 & >46 \\
Blood & 1 & 6 & 49 & 56 \\
\end{array}
\]
Karyotype: 46-XY. Reference: Makino et al. 1964d.

Case #100 Male, 39 years
Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;44</th>
<th>44</th>
<th>45</th>
<th>46</th>
<th>47</th>
<th>&gt;47</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>1</td>
<td>39</td>
<td>1</td>
<td></td>
<td></td>
<td>42</td>
</tr>
</tbody>
</table>

Karyotype: 46-XY. Reference: Makino et al. 1964d.

Case #101 Male, 28 years
Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;44</th>
<th>44</th>
<th>45</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>4</td>
<td>58</td>
<td>1</td>
<td></td>
<td>63</td>
<td></td>
</tr>
</tbody>
</table>


Case #102 Male, 25 years
Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;44</th>
<th>44</th>
<th>45</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>2</td>
<td>1</td>
<td>26</td>
<td>1</td>
<td></td>
<td>30</td>
</tr>
</tbody>
</table>


Case #103 Male, 38 years
Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;45</th>
<th>45</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>43</td>
<td></td>
<td>44</td>
<td></td>
</tr>
</tbody>
</table>


Case #104 Male, 31 years
Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;46</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>29</td>
<td></td>
<td></td>
<td>29</td>
</tr>
</tbody>
</table>


Case #105 Male, 38 years
Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;46</th>
<th>46</th>
<th>47</th>
<th>&gt;47</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>15</td>
<td>1</td>
<td></td>
<td>16</td>
<td></td>
</tr>
</tbody>
</table>


Case #106 Male, 31 years
Clinical features: Azoospermia. Sterility.
Chromosome counts:

<45  45  46  >46  total
Blood —  3   18  —  21

Case #107  Male, 28 years
Clinical features: Azoospermia. Sterility. Small penis and testes. showing atrophic seminiferous tubules.
Chromosome counts:

<45  45  46  >46  total
Blood —  2   34  —  36

Case #108  Male, 28 years
Chromosome counts:

<46  46  >46  total
Blood —  19  —  19

Case #109  Male, 34 years
Clinical features: Azoospermia. Sterility.
Chromosome counts:

<46  46  >46  total
Blood —  25  —  25

Case #110  Male, 18 years
Clinical features: Azoospermia. Further data not available.
Chromosome counts:

<45  45  46  >46  total
Blood —  3   61  —  64

Case #111  Male, 32 years
Chromosome counts:

<45  45  46  47  >47  total
Blood —  8   50   2   —  60

Case #112  Male, 27 years
Clinical features: Azoospermia. Further data not available.
Chromosome counts:

<46  46  >46  total
Blood —  12  —  12

Case #113  Male, 58 years
Clinical features: Oligospermia. Further data not available.
Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;45</th>
<th>45</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>3</td>
<td>83</td>
<td></td>
<td>86</td>
</tr>
</tbody>
</table>

Karyotype: 46-XY. Reference: Makino et al. 1964d.

**Phenotypic females**

*Turner's syndrome* (3 cases)

Case #114 Female, 20 years
Clinical features: Short stature. Primary amenorrhea. No neck webbing. Sex chromatin negative. 17-KS 3.96 mg/day.

Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;45</th>
<th>45</th>
<th>&gt;45</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>17</td>
<td></td>
<td>17</td>
</tr>
</tbody>
</table>


Case #115 Female, 18 years

Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;45</th>
<th>45</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>13</td>
<td>1</td>
<td></td>
<td>14</td>
</tr>
</tbody>
</table>

Karyotype: 45-XO. Reference: Makino et al. 1962b.

Case #116 Female, 1 year

Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;44</th>
<th>44</th>
<th>45</th>
<th>&gt;45</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>2</td>
<td>28</td>
<td></td>
<td>30</td>
</tr>
</tbody>
</table>


*Patients with an enlarged clitoris* (13 cases)

Case #117 Female, brought up as a male, 5 years
Clinical features: Enlarged clitoris with penile configuration. Mis-diagnosed as crooked penis with hypospadia. Uterus and ovary present.

Chromosome counts:

<table>
<thead>
<tr>
<th>&lt;44</th>
<th>44</th>
<th>45</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>1</td>
<td>66</td>
<td></td>
<td></td>
<td>67</td>
</tr>
</tbody>
</table>

Skin — 3 5 41 — 49
Case #118 Female, 1 month
Clinical features: Enlarged clitoris, 1.5×1.0 cm, with penile configuration. Enlarged major labia. Normal ovary, 3×1.5 cm. Normal oviducts. Drumstick positive. 17-KS 0.08 mg/day.

Chromosome counts:

<table>
<thead>
<tr>
<th></th>
<th>&lt;45</th>
<th>45</th>
<th>46</th>
<th>47</th>
<th>&gt;47</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bone marrow</td>
<td>1 (?)</td>
<td>3</td>
<td>1 (?)</td>
<td>5</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>


Case #119 Female, 4 years
Clinical features: Enlarged clitoris. Sex chromatin positive.

Chromosome counts:

<table>
<thead>
<tr>
<th></th>
<th>&lt;45</th>
<th>45</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood</td>
<td>2</td>
<td>15</td>
<td></td>
<td>17</td>
<td></td>
</tr>
</tbody>
</table>


Case #120 Female, 29 years
Clinical features: Enlarged clitoris. No child during eight years of married life. Menstruation irregular. Sex chromatin positive (20%).

Chromosome counts:

<table>
<thead>
<tr>
<th></th>
<th>&lt;46</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood</td>
<td>15</td>
<td>15</td>
<td></td>
<td>15</td>
</tr>
</tbody>
</table>


Case #121 Female, 4 months
Clinical features: Enlarged clitoris. Further data not available.


Case #122 Female, 12 years
Clinical features: Enlarged clitoris. Further details not available.


Case #123 Female, new born
Clinical features: Enlarged clitoris. Further data not available.


Case #124 Female, 5 years
Clinical features: Enlarged clitoris. Further data not available.


Case #125 Female, 18 years
Clinical features: Enlarged clitoris. Further data not available.


Case #126 Female, 4.5 years
Chromosome counts:

<43 43 44 45 46 46 total

Blood — 1 — 2 27 — 30


Case #127 Female, 1 month
Her mother was treated with lutenic hormone at the 5th month of gestation.

Chromosome counts:

<45 45 46 46 total

Blood — 3 45 — 48


Case #128 Female, 1 month
Unable to suck. Suspicion of some abnormalities in internal organs.

Chromosome counts:

<45 45 46 46 total

Blood — 2 31 — 33


Case #129 Female, 1 year
Clinical features: Enlarged clitoris. Further data not available.

Chromosome counts:

<46 46 46 total

Bone marrow — 10 — 10


Patients without vagina (7 cases).

Case #130 Female, 2 years
Clinical features: Absence of vagina. Abnormal external genitalia. Her mother received progesterone therapy during pregnancy. 17-KS 0.44 mg/day.

Chromosome counts:

<46 46 47 47 total

Bone marrow — 25 1 — 26


Case #131 Female, 24 years

Chromosome counts:

<45 45 46 46 total

Bone marrow — 2 12 — 14


Case #132 Female, 23 years
Clinical features: Height 153.4 cm. Weight 51.0 kg. Primary Amenorrhea. Absence of vagina. Underdeveloped uterus. Small-sized ovary. Sex chromatin positive (31%).

Chromosome counts:
1966 Chromosome Studies in 150 Sexually Abnormal Patients, II

<table>
<thead>
<tr>
<th>Chromosome counts:</th>
<th>&lt;44</th>
<th>44</th>
<th>45</th>
<th>46</th>
<th>47</th>
<th>&gt;47</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood</td>
<td>---</td>
<td>1</td>
<td>1</td>
<td>21</td>
<td>1</td>
<td>---</td>
<td>24</td>
</tr>
</tbody>
</table>


Case #133 Female, 20 years
Clinical features: Height 157.2 cm. Weight 56.0 kg. Absence of vagina. Primary amenorrhea. Normal secondary sexual characters. Laparotomy revealed small uterus, hard string like structures in place of fallopian tubes and small ovaries on both sides. Sex chromatin positive (39%).

Chromosome counts:  

<table>
<thead>
<tr>
<th>Chromosome counts:</th>
<th>&lt;44</th>
<th>44</th>
<th>45</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood</td>
<td>---</td>
<td>---</td>
<td>---</td>
<td>11</td>
<td>---</td>
<td>11</td>
</tr>
<tr>
<td>Skin</td>
<td>1</td>
<td>5</td>
<td>---</td>
<td>---</td>
<td>---</td>
<td>6</td>
</tr>
<tr>
<td>Bone marrow</td>
<td>---</td>
<td>---</td>
<td>---</td>
<td>1</td>
<td>---</td>
<td>1</td>
</tr>
</tbody>
</table>


Case #134 Female, 20 years
Clinical features: Absence of vagina and uterus. Primary amenorrhea. Full development of secondary sexual characters. Normal ovaries, right 3.0 x 2.0 x 1.5 cm, left 3.5 x 2.0 x 1.5 cm. Sex chromatin positive (32%).

Chromosome counts:  

<table>
<thead>
<tr>
<th>Chromosome counts:</th>
<th>&lt;46</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood</td>
<td>---</td>
<td>27</td>
<td>---</td>
<td>27</td>
</tr>
</tbody>
</table>


Case #135 Female, 25 years
Clinical features: Height 149 cm. Weight 50 kg. Absence of vagina. Primary amenorrhea. Small uterus. Ovary not palpable. Sex chromatin positive (38%).

Chromosome counts:  

<table>
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<tr>
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<th>46</th>
<th>&gt;47</th>
<th>total</th>
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</thead>
<tbody>
<tr>
<td>Blood</td>
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<td>16</td>
<td>1</td>
<td>---</td>
<td>17</td>
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</tbody>
</table>


Case #136 Female, 23 years

Chromosome counts:  

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<tr>
<th>Chromosome counts:</th>
<th>&lt;46</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood</td>
<td>---</td>
<td>12</td>
<td>---</td>
<td>12</td>
</tr>
</tbody>
</table>


Adrenogenital syndrome (6 cases)

Case #137 Female, 6 years
Clinical features: Unusually enlarged clitoris. 17-KS 15-20 mg/day. Pregnanetriol 2.4 mg/day. Diagnosed as adrenogenital syndrome.

Chromosome counts:  

<table>
<thead>
<tr>
<th>Chromosome counts:</th>
<th>&lt;46</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bone marrow</td>
<td>---</td>
<td>20</td>
<td>---</td>
<td>20</td>
</tr>
</tbody>
</table>

Cytologia 31, 1966 25
Case #138 Female, 4 years
Karyotype counts:

\[
\begin{array}{cccc}
<46 & 46 & >46 & \text{total} \\
\end{array}
\]

Bone marrow — 35 — 35


Case #139 Female, 22 years

Chromosome counts:

\[
\begin{array}{ccccccc}
<44 & 45 & 46 & 47 & >47 & \text{total} \\
1 & 2 & 36 & 2 & — & 41 \\
\end{array}
\]


Case #140 Female brought up as a male, 18 years
Clinical features: Enlarged clitoris. Uterus and ovary present. Sex chromatin positive. Diagnosed as adrenogenital syndrome.

Chromosome counts:

\[
\begin{array}{ccccccc}
<45 & 45 & 46 & 47 & >47 & \text{total} \\
1 & 111 & 1 & — & 113 \\
\end{array}
\]


Case #141 Female, 12 years
Clinical features: Male type external genitalia. Enlarged clitoris. Pseudopubertas praecox. Her sister shows similar clinical features. Diagnosed as adrenogenital syndrome.

Chromosome counts:

\[
\begin{array}{ccccccc}
<45 & 45 & 46 & 47 & >47 & \text{total} \\
1 & 111 & 1 & — & 113 \\
\end{array}
\]


Case #142 Female, 5 years
Clinical features: Diagnosed as adrenogenital syndrome. Further data not available.

Chromosome counts:

\[
\begin{array}{ccccccc}
<44 & 45 & 46 & >46 & \text{total} \\
2 & 2 & 24 & — & 28 \\
\end{array}
\]


Miscellaneous sex abnormalities 3 (cases)

Case #143 Female, 17 years

Chromosome counts:

\[
\begin{array}{ccccccc}
<46 & 46 & 47 & >47 & \text{total} \\
3 & 56 & — & 59 \\
\end{array}
\]

Case #144  Female, 22 years

Clinical features: Hypoplastic uterus. Primary amenorrhea.

Chromosome counts:

\[ \begin{array}{cccccc}
<44 & 44 & 45 & 46 & 47 & 48 & >48 & \text{total} \\
\text{Blood} & - & 2 & 6 & 122 & - & 3 & - & 133 \\
\text{Karyotype: 46-XX. Reference: Makino et al. 1964d.}
\end{array} \]

Case #145  Female, 2 years

Clinical features: Epispadia. Further data not available.

Chromosome counts:

\[ \begin{array}{cccc}
<46 & 46 & >46 & \text{total} \\
\text{Blood} & - & 30 & - & 30 \\
\text{Karyotype: 46-XX. Reference: Takagi et al. 1966.}
\end{array} \]

Testicular feminization (3 cases)

Case #146  Female, 28 years

Clinical features: Female type external genitalia including vagina. Scanty pubic and axillary hair. Primary amenorrhea. Absence of uterus. Inguinal tumor (testis?).

Chromosome counts:

\[ \begin{array}{cccc}
<46 & 46 & >46 & \text{total} \\
\text{Bone marrow} & - & 20 & - & 20 \\
\text{Karyotype: 46-XY. Reference: Makino et al. 1963a.}
\end{array} \]

Case #147  Female, 21 years


Chromosome counts:

\[ \begin{array}{ccccccc}
<44 & 44 & 45 & 46 & >46 & \text{total} \\
\text{Bone marrow} & - & 1 & - & 7 & - & 8 \\
\text{Karyotype: 46-XY. Reference: Makino et al. 1962b.}
\end{array} \]

Case #148  Female, 17 years

Clinical features: Female type external genitalia. Well-developed breast. No pubic and axillary hair. Internal organs of a male type except for the remnant of vagina. Ovaries, oviducts and uterus not detected. Undescended testes. 17-KS 3.85 mg/day.

Chromosome counts:

\[ \begin{array}{ccccccc}
<45 & 45 & 46 & 47 & >47 & \text{total} \\
\text{Blood} & - & 5 & 28 & 3 & - & 36 \\
\text{Karyotype: 46-XY. Reference: Makino et al. 1963b.}
\end{array} \]

True hermaphroditism and agonadism

True hermaphroditism (1 case)

Case #149  Brought up as a female, 25 years

Clinical features: Large clitoris with a penile configuration, about 5 cm long. Rudimentary vagina. Female type urethral opening. Unusually thickened major labia. Poorly developed breast. Man-like voice. Bipartite uterus. Ovo-testis on the right side only, which contained atrophic follicles, follicular cysts and ovarian stroma. The ovarian stroma showed structures resembling testicular tissue including atrophic seminiferous tubules and interstitial tissues. Germ cells showed no gametogenic activity.
Chromosome counts:

<table>
<thead>
<tr>
<th></th>
<th>&lt;44</th>
<th>44</th>
<th>45</th>
<th>46</th>
<th>47</th>
<th>&gt;47</th>
<th>total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bone marrow</td>
<td>1</td>
<td></td>
<td></td>
<td>23</td>
<td></td>
<td>1</td>
<td>27</td>
</tr>
</tbody>
</table>

Agonadism (1 case)

Case #150 Ambiguous sex, 3 years
Clinical features: Tiny clitoris-like protrusion. No gonad and internal genital organs except for bilateral small tubular structures (Wolffian remnants). Sex chromatin negative. Drumstick negative. Diagnosed as agonadism.

Chromosome counts:

<table>
<thead>
<tr>
<th></th>
<th>&lt;46</th>
<th>46</th>
<th>&gt;46</th>
<th>total</th>
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</thead>
<tbody>
<tr>
<td>Blood</td>
<td></td>
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</tr>
</tbody>
</table>

For the literature, refer to Part I.