Clinical evaluation of newly developed scoring system for DSD (DSD-SS)
: Association of DSD-SS with assigned gender in 45,X/46,XY mosaicism

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Background and Aim
✓ Clinical features of DSD are highly variable and sometimes difficult to be evaluated based on the conventional scoring system.
✓ We developed a new scoring system to comprehensively evaluate the clinical features of DSD (Poster #1-416).
✓ Clinical evaluation of this system has never been performed.

Method
✓ The clinical characteristics of 46 patients with 45,X/46,XY mosaicism was retrospectively evaluated based on the medical records and scored based on DSD-SS.
✓ Patients with the diagnosis of Turner syndrome were excluded.
✓ Turner females are excluded from the subsequent study.

45,X/46,XY mosaicism
✓ A rare sex chromosomal DSD
✓ A broad spectrum of clinical features ranging from normal male to almost female phenotype
✓ Left-right difference in clinical features is often observed.
✓ Gender is often difficult to be assigned.

Summary of 46 subjects

<table>
<thead>
<tr>
<th>Gender</th>
<th>Number</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Male</td>
<td>26</td>
<td>26.1%</td>
</tr>
<tr>
<td>Female</td>
<td>18</td>
<td>18.3%</td>
</tr>
<tr>
<td>Turner Female</td>
<td>12</td>
<td>28.3%</td>
</tr>
</tbody>
</table>

Turner females are excluded from the subsequent study.

Score of DSD-SS

M M F F

P < 0.001

95% of subjects in male showed a score of equal to or higher than 19, whereas 91 % of female exhibit a score of less than 19.

Score of the components of external genitalia

Scrotum/Labia

P < 0.001

M M F F

Uterus

P < 0.01

Gonadal position

P = 0.04

Score of internal genitalia and gonads

Uterus

P < 0.01

Gonadal position

P = 0.04

Testosterone production during mini-puberty or after hCG stimulation

Testosterone > 150 ng/dl*

M F

16/18 5/11

P = 0.01

88.9% 45.9%

* Evaluated after hCG test or during mini-puberty
**: Chai-square test

Summary
✓ DSD-SS well corresponds to the assigned gender in subjects with 45,X/46,XY mosaicism.
✓ This newly developed scoring system is useful to objectively and comprehensively evaluate the clinical features of DSD.

Nothing to Disclose

Masanobu KAWAI

Poster presented at:

Sex differentiation, gonads and gynecology or sex endocrinology

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