

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 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.

45,X/46,XY mosaicism

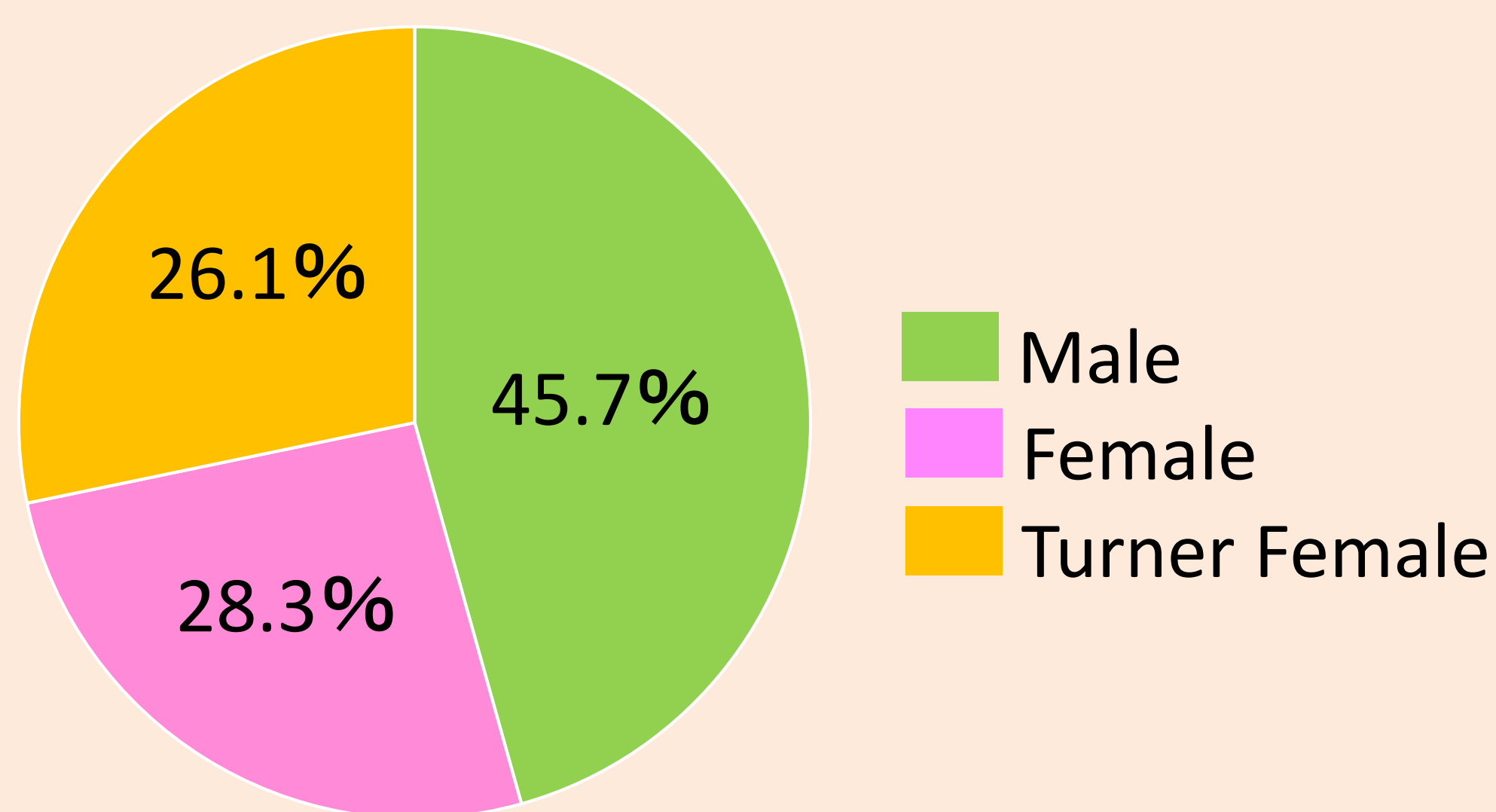
- ✓ A rare sex chromosomal DSD
- ✓ A broad spectrum of clinical features ranging from normal male to almost female phenotype
- ✓ Left-right difference in clinical features is often observed.
- ✓ Gender is often difficult to be assigned

DSD scoring system (DSD-SS)

I	Scrotum/Labia
I -①	Growth (Score of 0, 2, 4, 6)
I -②	Fusion (Score of 0, 3, 6)
II	Glans penis size (Scores of 0 to 6)
III	Urethral orifice position (0 to 6)
IV	Urogenital sinus (Scores of 0 or 6)
V	Uterus (Scores of 0 or 6)
VI	gonadal position (Scores of 0-6)

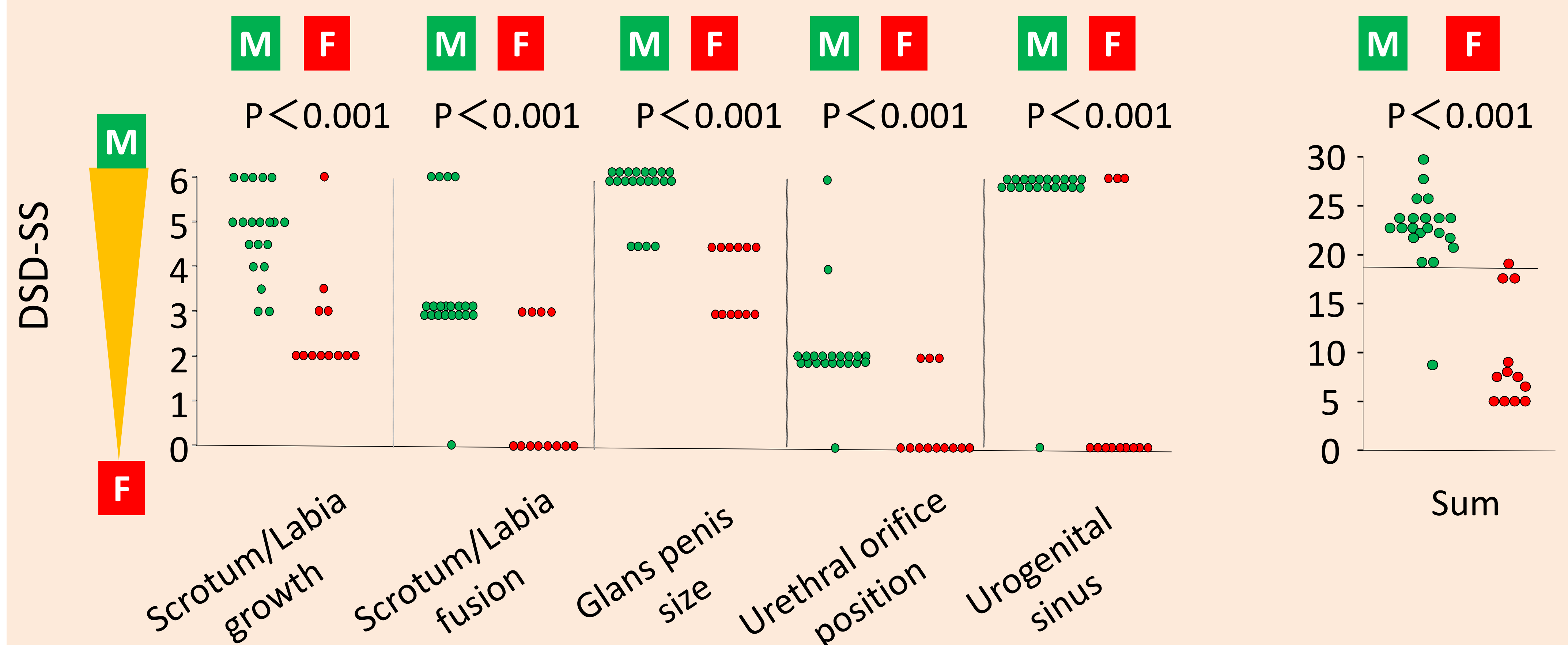
- ✓ Average value is calculated when there is difference between left and right.
- ✓ The normal male pattern receives the highest score of 42.

Summary of 46 subjects

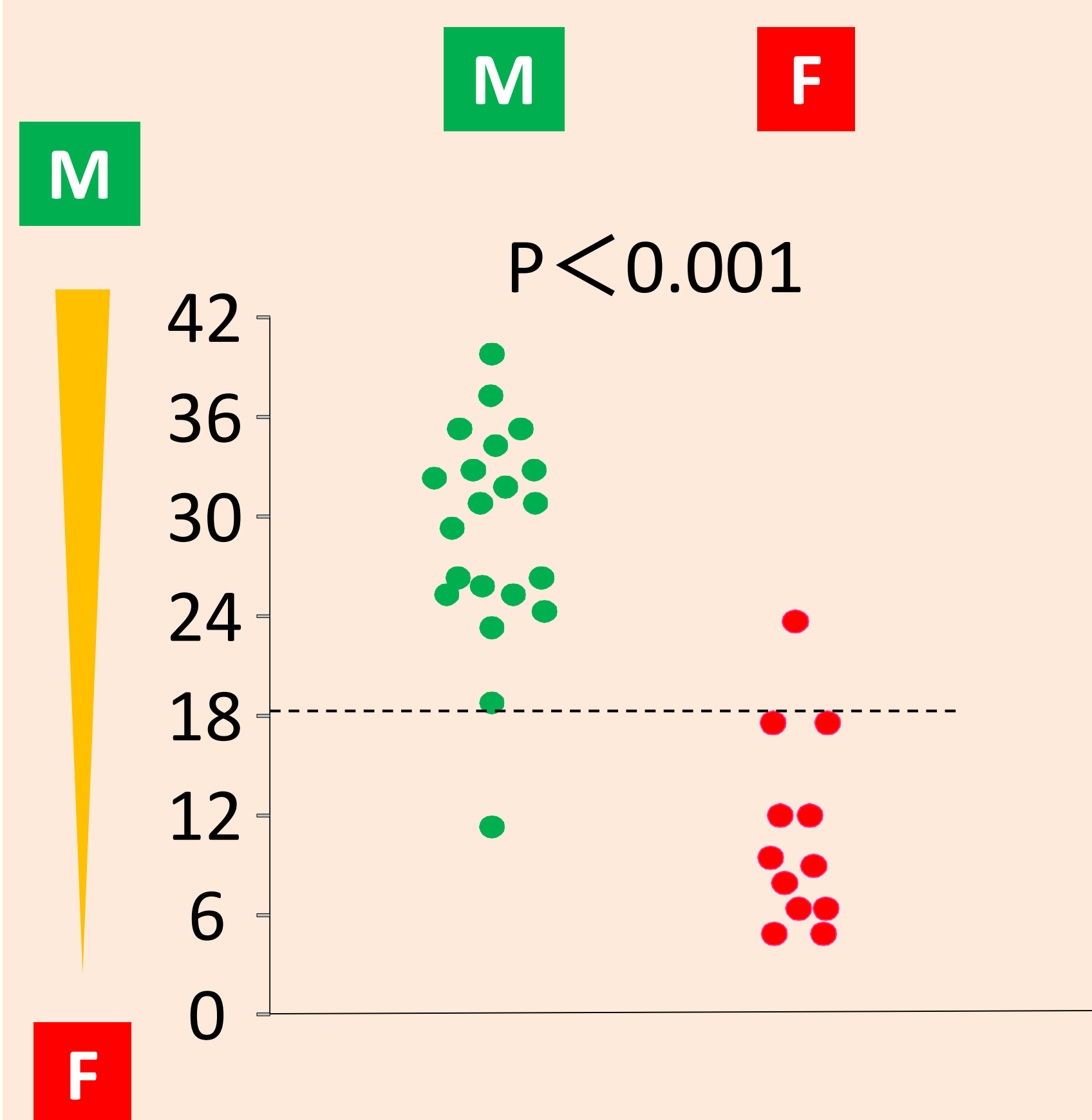


✓ Turner females are excluded from the subsequent study.

Score of the components of external genitalia

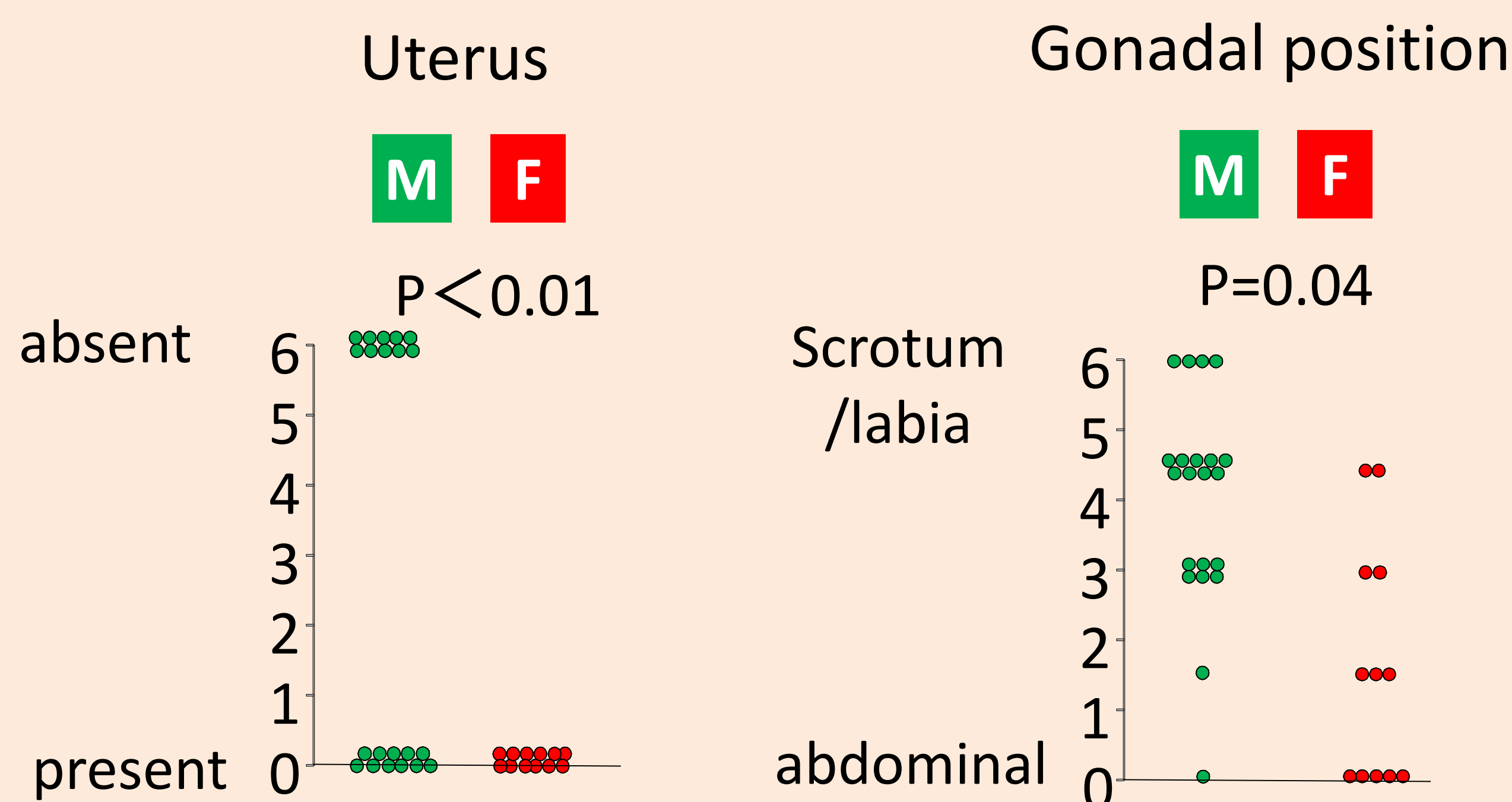


Score of DSD-SS



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 internal genitalia and gonads



DSD-SS	Male		P value	Female	
	Uterus(+)	Uterus(-)		Uterus(+)	P value vs M with U
External genitalia	23 (22-25)	22 (19-23)	0.08	8 (5-15.375)	0.0005
Gonadal position	5 (4.125-6)	3 (3-4.5)	0.14	2 (0-3)	0.04

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.

COI

Nothing to Disclose
Masanobu KAWAI

Testosterone production during mini-puberty or after hCG stimulation

	M	F	P value**
Testosterone > 150 ng/dl*	16/18 88.9%	5/11 45.5%	0.01

*: Evaluated after hCG test or during mini-puberty

** : Chai-square test



OWCH

