

Supplementary Table 2. Comparison between the characteristics of two cases with two different *NR5A1* mutations at the same site.

Items	p.G22C	p.G22S
Mutated nucleotide	G>T	G>A
Mutated amino acids	G/C	G/S
<i>In silico</i> analyses	Subtle change in conformational structure	Moderately affects NR5A1 protein binding to DNA owing to glycine position
Affinity of DNA binding	Obviously decreased	Complete loss
Transcriptional activity	Obviously decreased	Obviously decreased
Nuclear localization	No change [#]	Intense subnuclear focal aggregates
Homozygous/heterozygous mutation	Heterozygous	Heterozygous
Source of variation	de novo	Maternal
Age at clinical evaluation	13 years	9 years
Race	Chinese	Indian
Patient history	Raised as female, the first visit triggered by right lower abdominal pain	Small penis, hypospadias, and undescended testis at the time of birth
Phenotype	Absent uterus, blind-side vagina, dysgenesis testicle in the inguinal area, undeveloped female external genitalia	Very small penis (~4 cm at age 5), penoscrotal hypospadias, and hypoplastic scrotum with reduced rugosity

[#]Owing to limitations of imaging equipment, detailed changes in nuclear localization were not observed.