Genetic characterization of sixteen 46,XX males with disorders of sex development

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Objective
To investigate the phenotype-genotype association by genetic analysis of sixteen 46,XX males with disorders of sex development (DSD).

Methods
Clinical data of sixteen 46,XX males were collected. The karyotypes of sixteen patients were determined by the application of conventional chromosome G banding of peripheral blood. Fluorescence in situ hybridization (FISH) and multiple polymerase chain reaction (PCR) were utilized to detect and locate the SRY gene region. Multiplex ligation-dependent probe amplification (MLPA) was applied to analyze the gender related genes copy number.

Results
The karyotype analysis of sixteen patients showed that all were 46,XX males. Among these 46,XX males, 15 had SRY gene and no mutation was found. One patient had no SRY gene and gender related gene copy number abnormalities were not found by MLPA.

Conclusion
The primary cause of 46,XX DSD is the hidden SRY gene translocated to the X chromosome. There is an alter way beyond the traditional gender determination in the patient of male phenotype with SRY negative.