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Investigation of mutations in the SRY, SOX9, and DAX1 genes in sex reversal patients from the Sichuan region of China

L. Chen, X.P. Ding, X. Wei and L.X. Li

Bio-resource Research and Utilization Joint Key Laboratory of Sichuan-Chongqing, Institute of Medical Genetics, College of Life Science, Sichuan University, Chengdu, China

Correspondence author: X. P. Ding
E-mail: Brainding@scu.edu.cn

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ABSTRACT. We investigated the molecular genetic mechanism of sex reversal by exploring the relationship between mutations in the sex-determining genes *SRY*, *SOX9*, and *DAX1* with genetic sex reversal disease. Mutations in the three key genes were detected by polymerase chain reaction (PCR) and sequencing after karyotype analysis. The mutations detected were then aligned with a random sample of 100 normal sequences and the NCBI sequence database in order to confirm any new mutations. Furthermore, the copy number of *SOX9* was measured by fluorescence quantitative PCR. Seven of the 10 male sex reversal patients (46, XX) contained an excess copy of the *SRY* gene, while one of the eight female sex reversal patients (46, XY) was lacking the *SRY* gene. Additionally, a new mutation (T-A, Asp24Lys) was detected in one female sex reversal patient (46, XY). No other mutation was detected in the analysis of *SOX9* and *DAX1*, with the exception of an insertion mutation (c.35377791insG) found in the testicular-specific enhancer (TESCO) sequences in an *SRY*-positive

female sex reversal patient (46, XY). Eight of the 18 sex reversal cases (44.4%) showed obvious connections with *SRY* gene translocations, mutations, or deletions, which was significantly higher than that reported previously (33.3%), indicating a need to further expand the range of sample collection. Overall, these results indicated that the main mechanism of sex reversal are not associated with mutations in the coding regions of *SOX9* and *DAX1* or copy number variations of *SOX9*, which is consistent with results of previous studies.

Key words: Sex reversal; *SRY*; *SOX9*; *DAX1*; Gene mutation; Real-time fluorescence quantitative PCR

