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Clinical and genetic characterization of complete androgen insensitivity syndrome in a Chinese family

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ABSTRACT. We studied a family with two cousins who were diagnosed with complete androgen insensitivity syndrome, an X-linked disorder caused by mutations in the androgen receptor gene. A pedigree analysis and a molecular study using PCR and DNA sequencing clarified each female family member's androgen receptor status and revealed a mutation consisting of the deletion of exon 2 and surrounding introns of the androgen receptor gene. Based on the relative nucleotide positions, we concluded that the deletion mutation in exon 2 and its surrounding introns was approximately 6000 to 7000 bp. This mutation, never previously fully characterized using DNA sequencing, was responsible for complete androgen insensitivity syndrome in this family. Pedigree analysis with a molecular study of the androgen receptor gene in affected families facilitates genetic counseling provided to family members.

Key words: CAIS; Phenotype; Deletion mutation; AR gene

