

# CYP21A2 gene mutations analysis in 21 Chinese patients with salt-wasting form of congenital adrenal hyperplasia

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**Background:** Studies about the genetics of congenital adrenal hyperplasia caused by 21-hydroxylase deficiency (CAH) in Chinese children are less.

**Objective and hypotheses:** Study the genotypes of Chinese probands with salt-wasting form of congenital adrenal hyperplasia caused by 21-hydroxylase deficiency (CAH) and performed pedigree-based linkage analysis.

**Method:** We have performed genetic-testing (Method: qPCR and Sanger sequencing) in 21 probands with salt-wasting form of congenital adrenal hyperplasia caused by 21-hydroxylase deficiency (CAH), among which 19 patients were associated with pedigree-based linkage analysis.

**Results:** Through pedigree-based conditional linkage analysis, there are two families occurred de novo mutation. The frequency of the pathogenic allele respectively, were c. 293-13C>G 38.78%, EX1-10DEL 20.41%, c.1069C>T 10.2%, c. 955C>T 10.2%, c. 518T>A 8.16%, EX3DEL 6.12%, c. 1279C>T 2.04%, c. 1451\_1452delGGinsC 2.04%, c.740delA 2.04% in probands. The detected mutation c. 293-13C> G, EX1-10DEL, c. 1069C> T, c. 955C> T, c. 518T> A, EX3DEL in this study were the hot spots Maria's research paper. Because we only involve salt-wasting type, the frequency of mutation are different. In particular, the mutation c.740delA was found for the first time that can result in premature termination of protein synthesis, leading to loss of protein function. In addition, we also carried out statistical analysis in mutation frequency among pedigree parents that higher frequency mutation were still c. 293-13C> G and EX1-10DEL.

**Conclusion:** We found the new mutation point c.740delA, Others are hot spots. We found that pathogenic allele were homogenous due to partial or whole exon deletion which could lead to misjudgment in disease loci. To avoid diagnostic errors, it is recommended that large-scale deletion need to be tested first using qPCR and then detect point mutations.

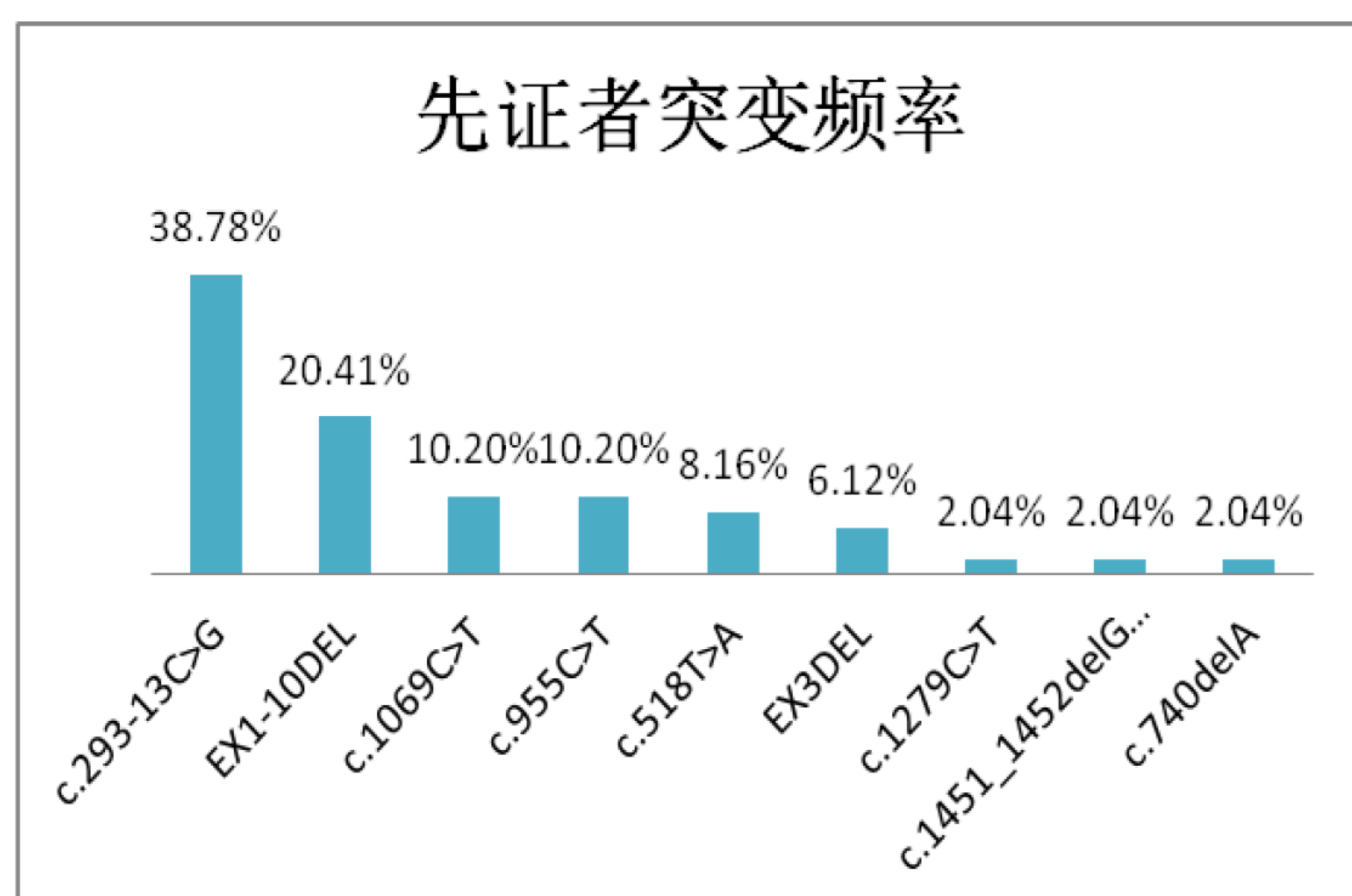


Fig 1 Statistical analysis in mutation frequency among probands

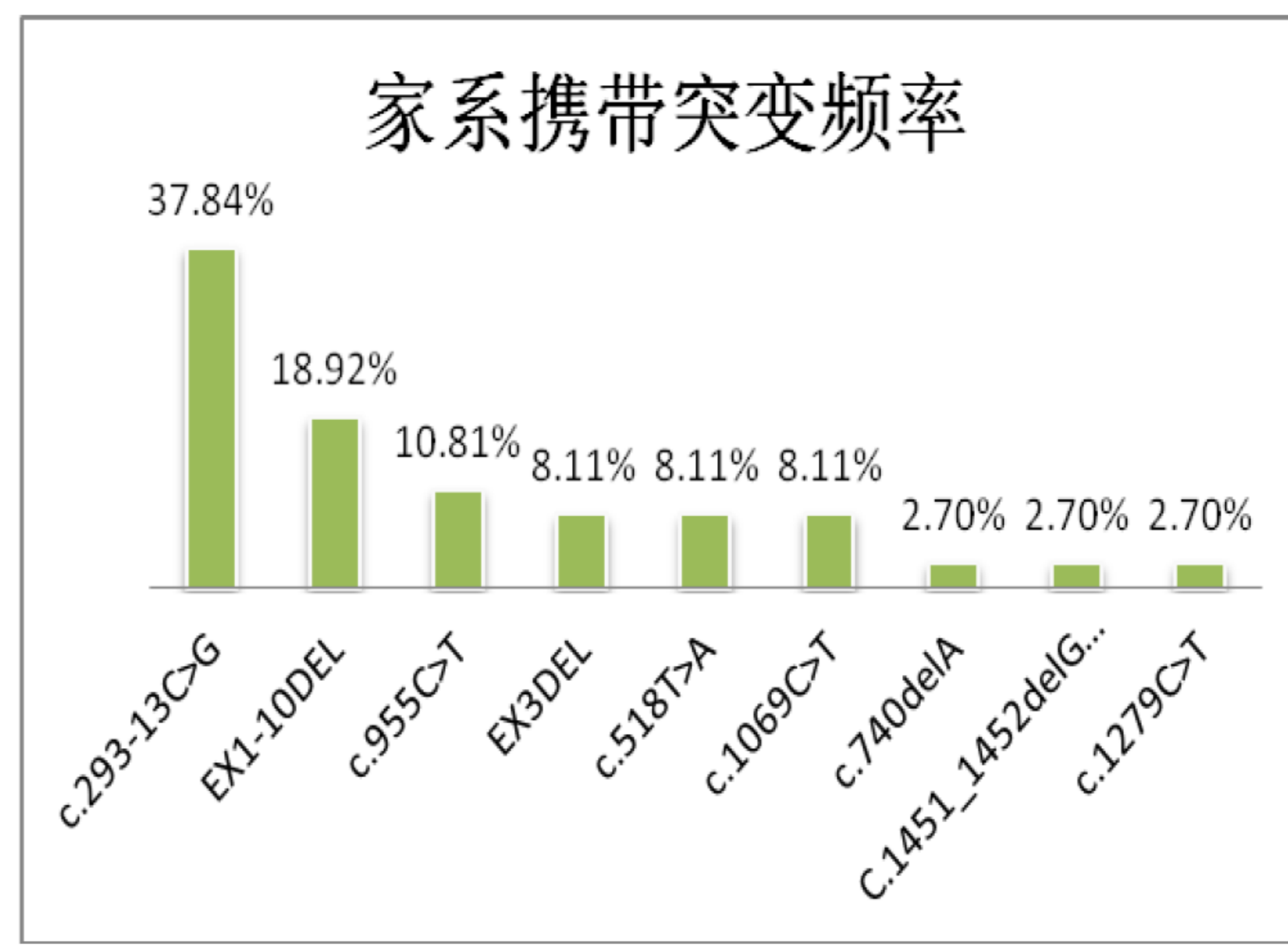


Fig 2 Statistical analysis in mutation frequency among pedigree parents

