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Expression of Mutant Ectodysplasin-A Gene in Hypohidrotic Ectodermal Dysplasia

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ABSTRACT To investigate the protein expression changes of c.878T>G mutant and wild-type Ectodysplasin-A (EDA) gene on prokaryotic vectors from hypohidrotic ectodermal dysplasia (HED) families. The c.878T>G mutant and wild-type EDA genes were amplified, screened, and double-enzyme digested to obtain the target fragments. The recombinant plasmids were constructed and introduced into *Escherichia coli*. Lactosidase (IPTG) induces the expression of the target gene. The EDA protein expression content was determined by Western blot and enzyme-linked immunosorbent method. On the prokaryotic expression vector, the c.878T>G mutant and wild-type EDA protein expression increased with the increase of induction time, and the mutant protein expression was lower than the wild-type protein expression at the same point ($P<0.05$). In conclusion, the c.878T>G gene mutation in this family caused one amino acid of the EDA protein to become another amino acid, and the c.878T>G mutation site can lead to a significant decrease in the expression of EDA protein on the prokaryotic expression vector, is highly pathogenic and may be the cause of the HED family.