

cyp17a1^{ihb144/+} (AB) (CZRC catalog ID: CZ 292)

Nature of the mutation

Between 226 bp to 234 bp of the wild-type *cyp17a1* coding sequence, TGTATT, is deleted. The mutated *cyp17a1* codes for a truncated protein containing 234 aa, of which 519 aa are identical to wildtype *cyp17a1*.

Sense Strand Sequence

cyp17a1

GCAGTGCTGTTTCAGAAGAGCTGGAGGACACTCAGTTGAATATAGCTGACAATGGCTG
AAGCACTCATCCTGCCCTGGCTGCTCTGTTTAAGCCTGTTCTCCGCAGTGACTCTGGC
AGCTCTGTATCTCAAACAGAAGATGAATGGATTTGTGCCAGCAGGAAATAGATCTCC
TCCAAGTCTCCCTTCACTGCCCATCATCGGGAGTCTGATGAGCCTGGTGAGCGACAGT
CCTCCGCACATCTTCTTTTCAGGACCTGCAGAAGAAATACGGAGATCTGTATTCCCTCA
TGATGGGCTCCACAAACTGCTCATTGTGAACAACCACCATCATGCGAAGGAGATCC
TGATCAAAAAAGGAAAAATATTTGCAGGGAGGCCACGGACTgtaagtacataatcacattcagttt
aattagccatttttagctgtttaattttgtaatttaaatatggcccatctgcatatggttaaactaaattacatattcattcattcatttcttttggc
ttagtccctttattaatcagaggtcgtcagagcagaatgaactgcc

Uppercase: Exon/coding sequence

Lowercase: intron/noncoding sequence

atcg : Forward/Reverse primer

Genotyping assay

Primers:

CZ292_forward: 5' GCAGTGCTGTTTCAGAAGAGCT 3'

CZ292_reverse: 5' GGCAGTTCATTCTGCTCTGA 3'

PCR program:

95°C 5min

95°C 30 sec

60°C 30 sec

72°C 30 sec

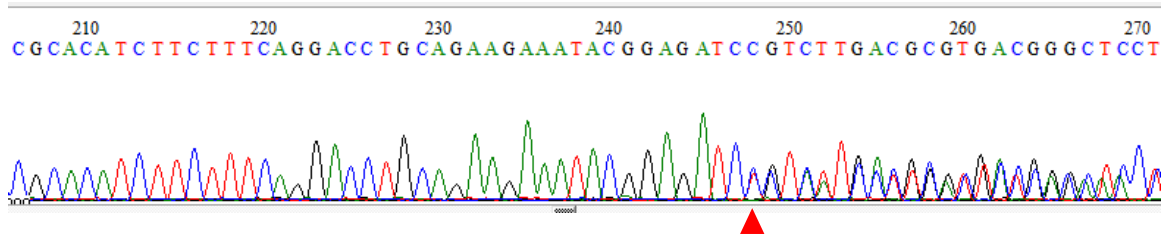
72°C 8min

4°C hold

} 30 Cycles

Product size: 565 bp

The sequencing results of the CZ292(+/-):



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WT      GCAGTGCTGTTCAGAAGAGCTGGAGGACACTCAGTTGAATATAGCTGACAATGGCTGAAG
CZ292   GCAGTGCTGTTCAGAAGAGCTGGAGGACACTCAGTTGAATATAGCTGACAATGGCTGAAG
*****

WT      CACTCATCCTGCCCTGGCTGCTCTGTTAAGCCTGTTCTCCGCAGTACTCTGGCAGCTC
CZ292   CACTCATCCTGCCCTGGCTGCTCTGTTAAGCCTGTTCTCCGCAGTACTCTGGCAGCTC
*****

WT      TGTATCTCAAACAGAAGATGAATGGATTTGTGCCAGCAGGAAATAGATCTCCTCCAAGTC
CZ292   TGTATCTCAAACAGAAGATGAATGGATTTGTGCCAGCAGGAAATAGATCTCCTCCAAGTC
*****

WT      TCCCTTCACTGCCCATCATCGGGAGTCTGATGAGCCTGGTGAGCGACAGTCCTCCGCACA
CZ292   TCCCTTCACTGCCCATCATCGGGAGTCTGATGAGCCTGGTGAGCGACAGTCCTCCGCACA
*****

WT      TCTTCTTTCAGGACCTGCAGAAGAAATACGGAGATCTGTATTCCTCATGATGGGCTCCC
CZ292   TCTTCTTTCAGGACCTGCAGAAGAAATACGGAGATCTGTATTCCTCATGATGGGCTCCC
*****

WT      ACAAAGTCTCATTGTGAACAACCACCATCATGCGAAGGAGATCCTGATCAAAAAAGGAA
CZ292   ACAAAGTCTCATTGTGAACAACCACCATCATGCGAAGGAGATCCTGATCAAAAAAGGAA
*****

WT      AAATATTTGCAGGGAGGCCACGGACTgtaagtacatacaatcacattcagtttaattagc
CZ292   AAATATTTGCAGGGAGGCCACGGACTgtaagtacatacaatcacattcagtttaattagc
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Reference:

Gang Zhai, Tingting Shu, Yuguo Xia, Xia Jin, Jiangyan He, Zhan Yin. Androgen signaling regulates the transcription of anti-Müllerian hormone via synergy with SRY-related protein SOX9A. *Science Bulletin* Volume 62, Issue 3, 15 February 2017, Pages 197–203.

<http://doi.org/10.1016/j.scib.2017.01.007>