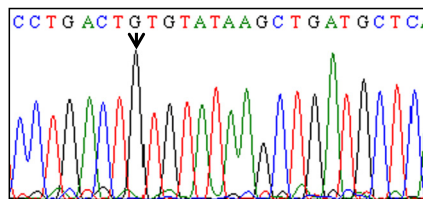
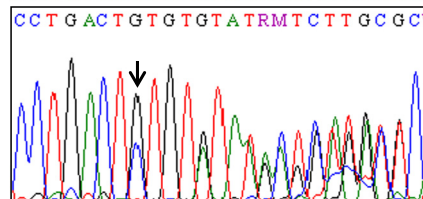
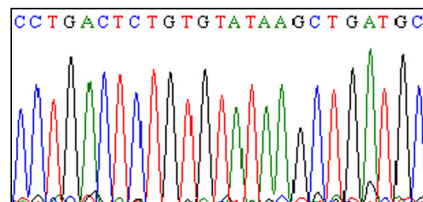


A**c.2468_2469delCT (p.S823Cfs*2)**

Patient



Carrier



Normal

B**Ser 823**

HumanMutant	RDQGRQTYLPQPAICPDCV
HumanWt	RDQGRQTYLPQPAICPDSVYKLM LSCWRRDTKNRPSFQEI H L L L L Q Q G D E
Chimpanzee	RDQGRQTYLPQPAICPDSVYKLM LSCWRRDTKNRPSFQEI H L L L L Q Q G D E
Rhesus	RDQGRQTYLPQPAICPDSVYKLM LSCWRRDTKNRPSFQEI H L L L L Q Q G D E
Dog	RDQGRQIYLPQPVICPDSVYKLM LSCWRRDTK H RPSFQEI H L L L L Q Q G D E
Cow	RDQGRQTYLPQPAICPDSVYKLM LSCWRRDTK H RPSFQEI H L L L L Q Q V D D
Mouse	RDQGRQIYLPQPALCPDSVYKLM LSCWRR ETK H RPSFQEI H L L L L Q Q G A E
Rat	RDQGRQIYLPQPAVC PDSVYKLM LSCWRR ETK H RPSFQEI H L L L L Q Q G A E