MC1R Gene Sequencing Results
Analysis Report for JM
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Myredhairgene.com
1015 Edwards Road
Burlingame, CA 94010
650-348-2958
info@myredhairgene.com

Conclusion

Status: Red Hair Carrier

The sample from JM indicates that she has a DNA change in one copy of his MC1R gene, which cause amino acid change from a Arginine to a Cystine at position 151 (R151C). This change is associated with red hair color. JM is a carrier of red hair trait.

Human MC1R Gene Sequences

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atg gct gtg cag gga tcc cag aga aga ctt ctg ggc tcc ctc aac tcc acc ccc aca gcc 60
 M   A   V   Q   G   S   Q   R   R   L   L   G   S   L   N   S   T   P   T   A  20

atc ccc cag ctg ggg ctg gct gcc aac cag aca gga gcc cgg tgc ctg gag gtg tcc atc 120
 I   P   Q   L   G   L   A   A   N   Q   T   G   A   R   C   L   E   V   S   I  40

tct gac ggg ctc ttc ctc agc ctg ggg ctg gtg agc ttg gtg gag aac gcg ctg gtg gtg 180
 S   D   G   L   F   L   S   L   G   L   V   S   L   V   E   N   A   L   V   V  60

gcc acc atc gcc aag aac cgg aac ctg cac tca ccc atg tac tgc ttc atc tgc tgc ctg 240
 A   T   I   A   K   N   R   N   L   H   S   P   M   Y   C   F   I   C   C   L  80

gcc ttg tcg gac ctg ctg gtg agc ggg agc aac gtg ctg gag acg gcc gtc atc ctc ctg 300
 A   L   S   D   L   L   V   S   G   S   N   V   L   E   T   A   V   I   L   L 100

ctg gag gcc ggt gca ctg gtg gcc cgg gct gcg gtg ctg cag cag ctg gac aat gtc att 360
 L   E   A   G   A   L   V   A   R   A   A   V   L   Q   Q   L   D   N   V   I 120

gac gtg atc acc tgc agc tcc atg ctg tcc agc ctc tgc ttc ctg ggc gcc atc gcc gtg 420
 D   V   I   T   C   S   S   M   L   S   S   L   C   F   L   G   A   I   A   V 140

gac cgc tac atc tcc atc ttc tac gca ctg cgc tac cac agc atc gtg acc ctg ccg cgg 480
 D   R   Y   I   S   I   F   Y   A   L   R   Y   H   S   I   V   T   L   P   R 160

gcg cgg cga gcc gtt gcg gcc atc tgg gtg gcc agt gtc gtc ttc agc acg ctc ttc atc 540
 A   R   R   A   V   A   A   I   W   V   A   S   V   V   F   S   T   L   F   I 180

gcc tac tac gac cac gtg gcc gtc ctg ctg tgc ctc gtg gtc ttc ttc ctg gct atg ctg 600
 A   Y   Y   D   H   V   A   V   L   L   C   L   V   V   F   F   L   A   M   L 200

gtg ctc atg gcc gtg ctg tac gtc cac atg ctg gcc cgg gcc tgc cag cac gcc cag ggc 660
 V   L   M   A   V   L   Y   V   H   M   L   A   R   A   C   Q   H   A   Q   G 220

atc gcc cgg ctc cac aag agg cag cgc ccg gtc cac cag ggc ttt ggc ctt aaa ggc gct 720
 I   A   R   L   H   K   R   Q   R   P   V   H   Q   G   F   G   L   K   G   A 240

gtc acc ctc acc atc ctg ctg ggc att ttc ttc ctc tgc tgg ggc ccc ttc ttc ctg cat 780
 V   T   L   T   I   L   L   G   I   F   F   L   C   W   G   P   F   F   L   H 260

ctc aca ctc atc gtc ctc tgc ccc gag cac ccc acg tgc ggc tgc atc ttc aag aac ttc 840
 L   T   L   I   V   L   C   P   E   H   P   T   C   G   C   I   F   K   N   F 280

aac ctc ttt ctc gcc ctc atc atc tgc aat gcc atc atc gac ccc ctc atc tac gcc ttc 900
 N   L   F   L   A   L   I   I   C   N   A   I   I   D   P   L   I   Y   A   F 300

cac agc cag gag ctc cgc agg acg ctc aag gag gtg ctg aca tgc tcc tgg tga         954
 H   S   Q   E   L   R   R   T   L   K   E   V   L   T   C   S   W   *         317
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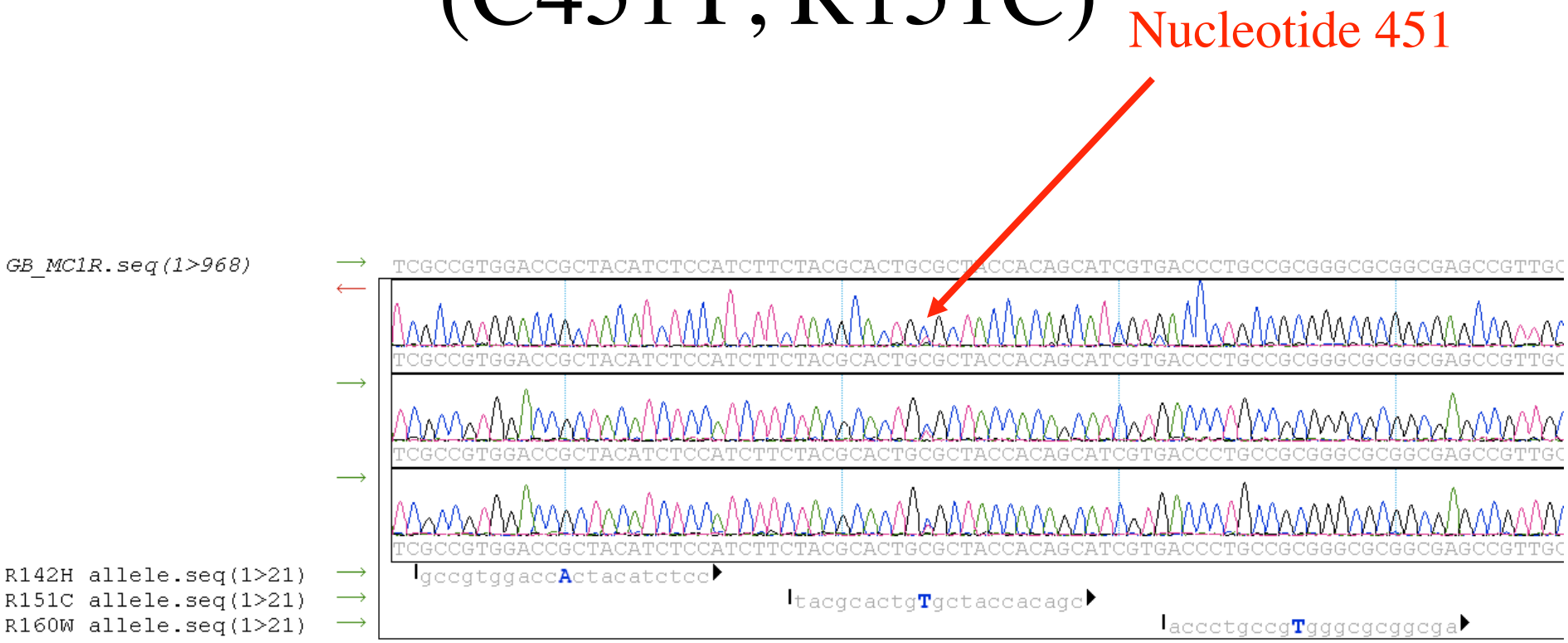
Red color letters indicate positions where nucleotide (lower case) and amino acid (upper case) sequence variations (SNP - single nucleotide polymorphism) are known to cause red hair phenotype in individuals.

Known Red Hair Color Variants

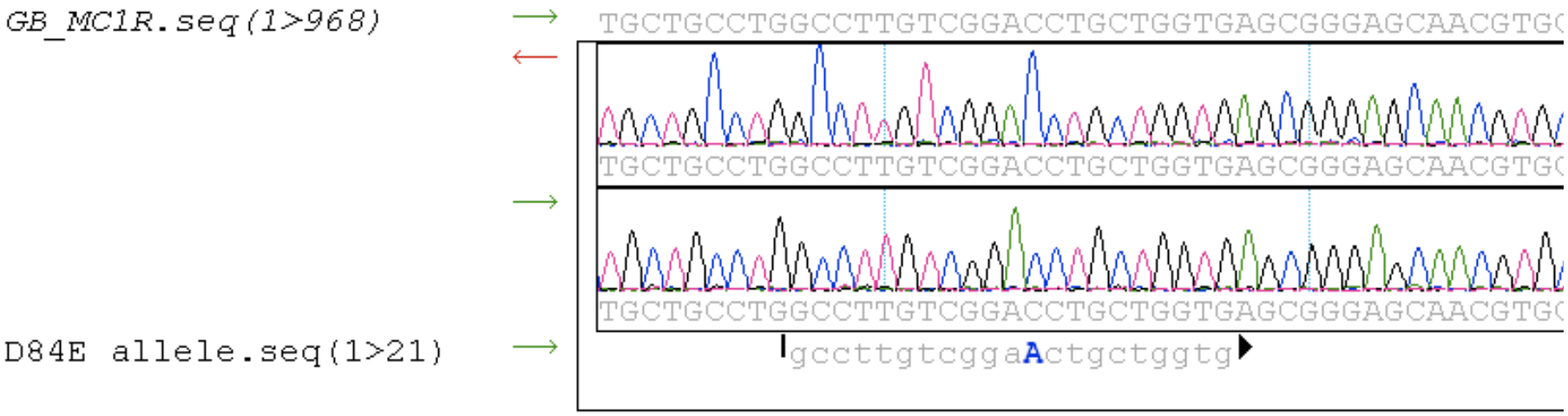
RHC variatns	Not Red	Red
Asp84Glu	GAC	GAA
Arg142His	CGC	CAC
Arg151Cys	CGC	TGC
Arg160Trp	CGG	TGG
Asp294His	GAC	CAC

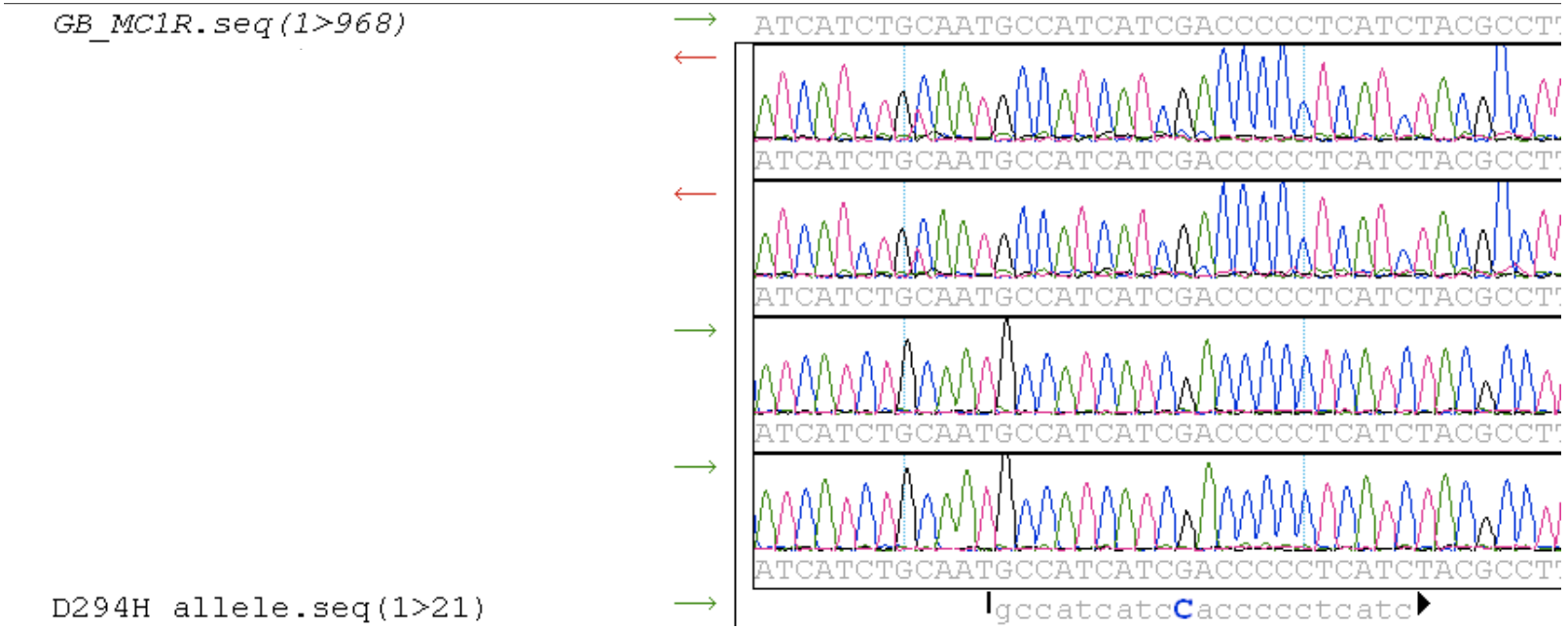
Results: JM's MC1R Variants (C451T, R151C)



Both copies of the MC1R gene are sequenced at once. A single peak means that both copies of the gene share the letter at that position. So at position 449 there is a T and at 450 there is a G. If there are two peaks at one position then this means that two MC1R genes are different at that position. So in JM's case, one MC1R gene has a T and the other has a C at position 451. The C at this position causes a change at amino acid position 151 which changes from Arginine (R) to Cysteine (C) and can cause red hair.

JM's MC1R sequences at other common red hair causing positions are regular





How Red Hair Works

Hair color is determined by the amount of two pigments called eumelanin and phomelanin that are in your hair. The amount of eumelanin in your hair gives you a range from blonde to black—a little eumelanin and you are blonde, an intermediate amount, brown, and a lot, black. Red comes into the equation with phomelanin. The more phomelanin in your hair, the redder it is.

Your hair color is a mixture of how much eumelanin and phomelanin is in your hair. For example, strawberry blonde is a little of each, auburn is some eumelanin and phomelanin and a redhead is very little eumelanin and lots of phomelanin.

Your cells decide how much of these melanins to put in your hair with genes. The key gene involved in red hair is called MC1R. (If you are interested in some current theories on inheritance of hair color besides red, please see http://www.thetech.org/genetics/ask.php?id=39.)

The MC1R gene comes in two versions--red and not-red. The not-red version is able to change phomelanin into eumelanin. So people with this version do not have red hair. The red version of MC1R cannot change phomelanin into eumelanin and so the red pigment builds up causing red hair.

Remember, we have two copies of each of our genes--one from mom and one from dad. To get red hair, both copies of your MC1R gene need to be the red version. In other words, you need to get the red version from your mom and your dad.

People with one version of each type of MC1R don't have red hair but they can pass the gene version on to their kids. These people are called carriers. Carriers are often freckled and sometimes have red highlights in their hair. Also, male carriers can have a red moustache or beard.