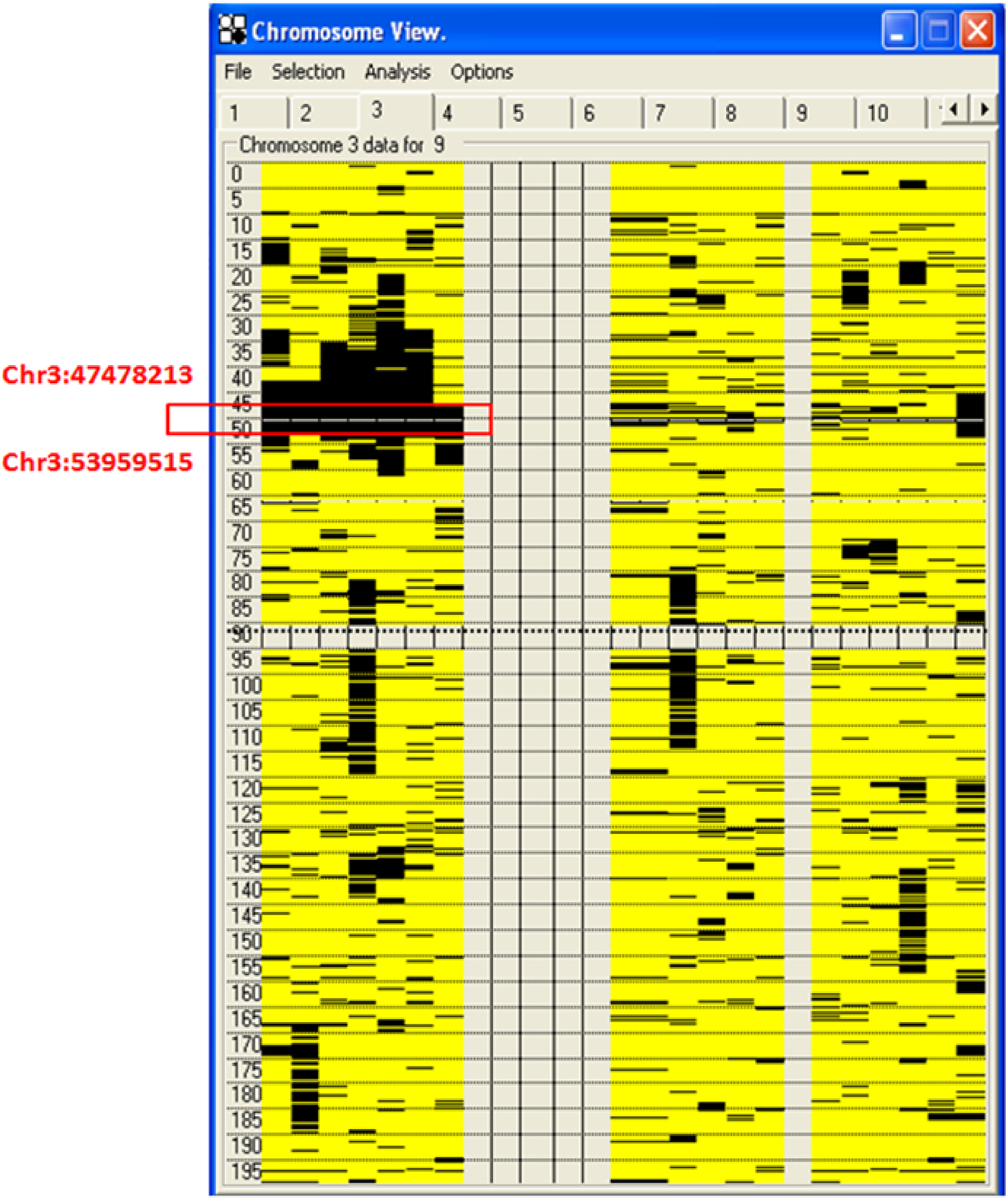
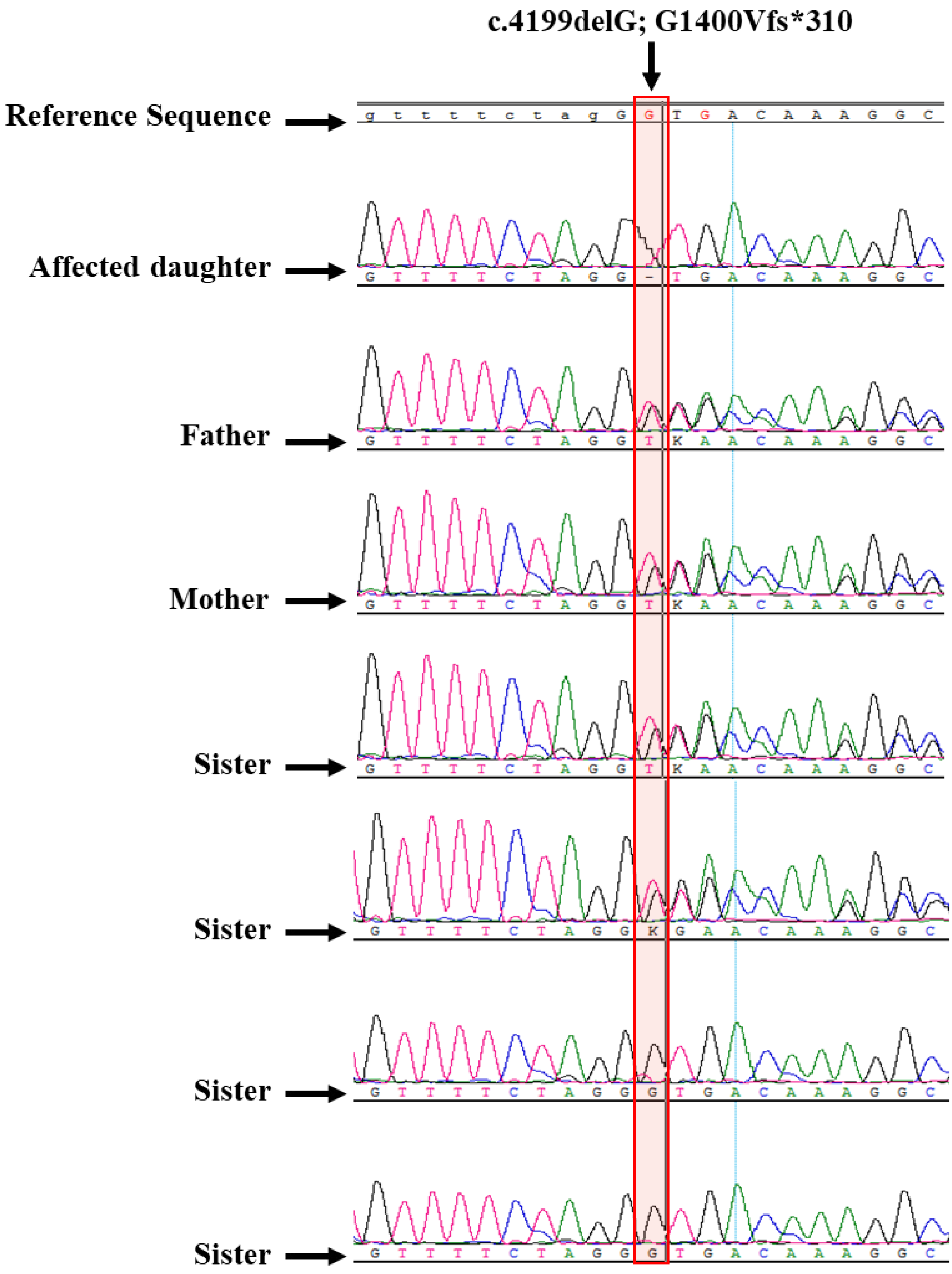
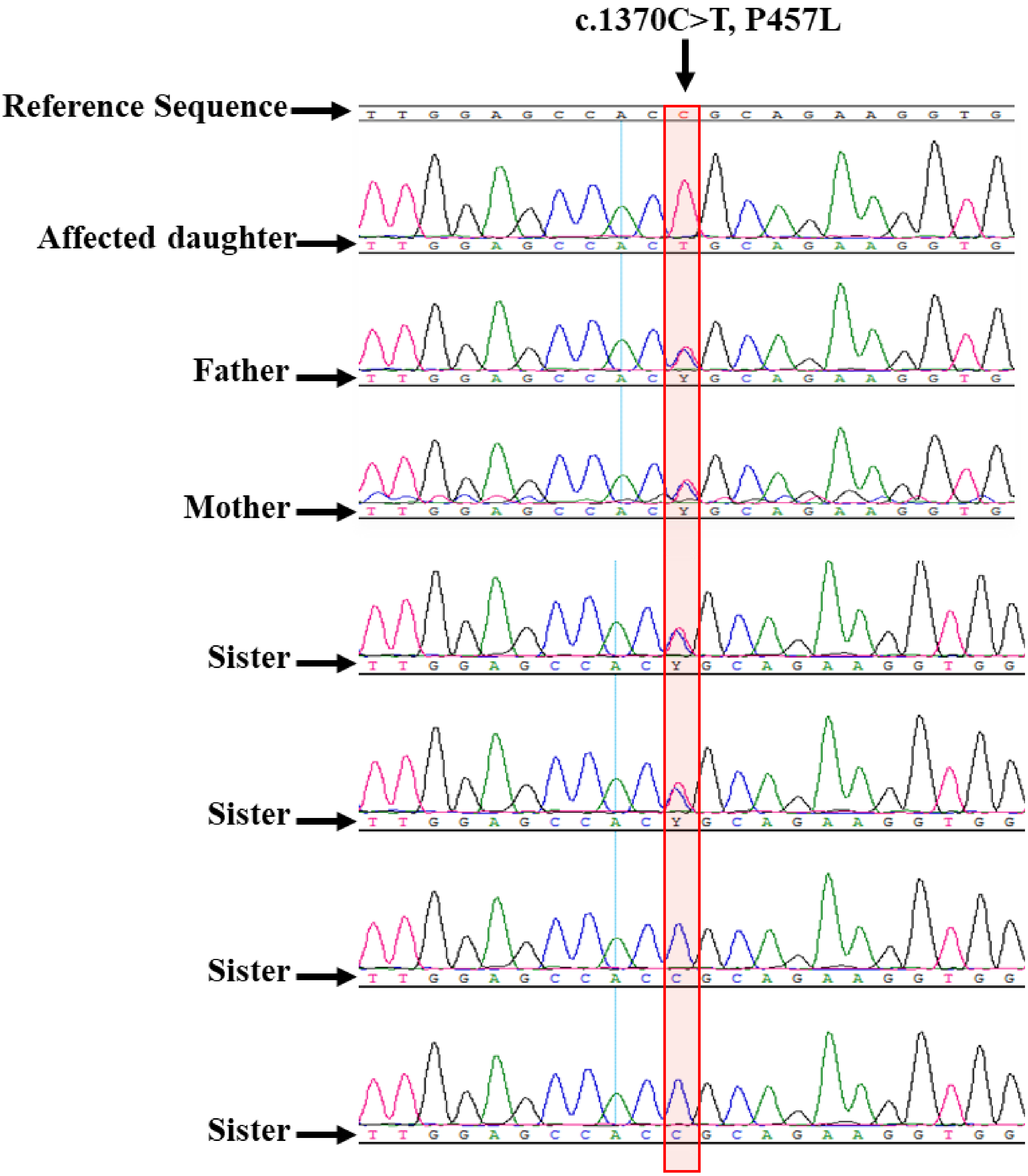
Supplementary Figure 1. Clinical features of some patients as described in Table 1. (a) Intact bullae in right thigh, (b) Bullae over dorsum of left hand, (c) Erosions in left side of neck, (d) Erosions and Erythema, (e) Erosions and Post inflammatory hypopigmentation, (f) Erosions over trunk, (g) Atrophy scar in left hand, (h) Scars and erosions, (i) Milia, (j) Crusted erosion in the neck, (k) Crusted erosions, (l) Anonychia, Loss of nail and nail dystrophy, (m) Loss of nail and nail dystrophy, (n) Loss of toe nails and erythema in right foot, (o) Nail loss and crusted erosions, (p) Fusion of fingers and milia, (q and r) Pseudosyndactyly (mitten-hand deformity) and Anonychia of both pictures



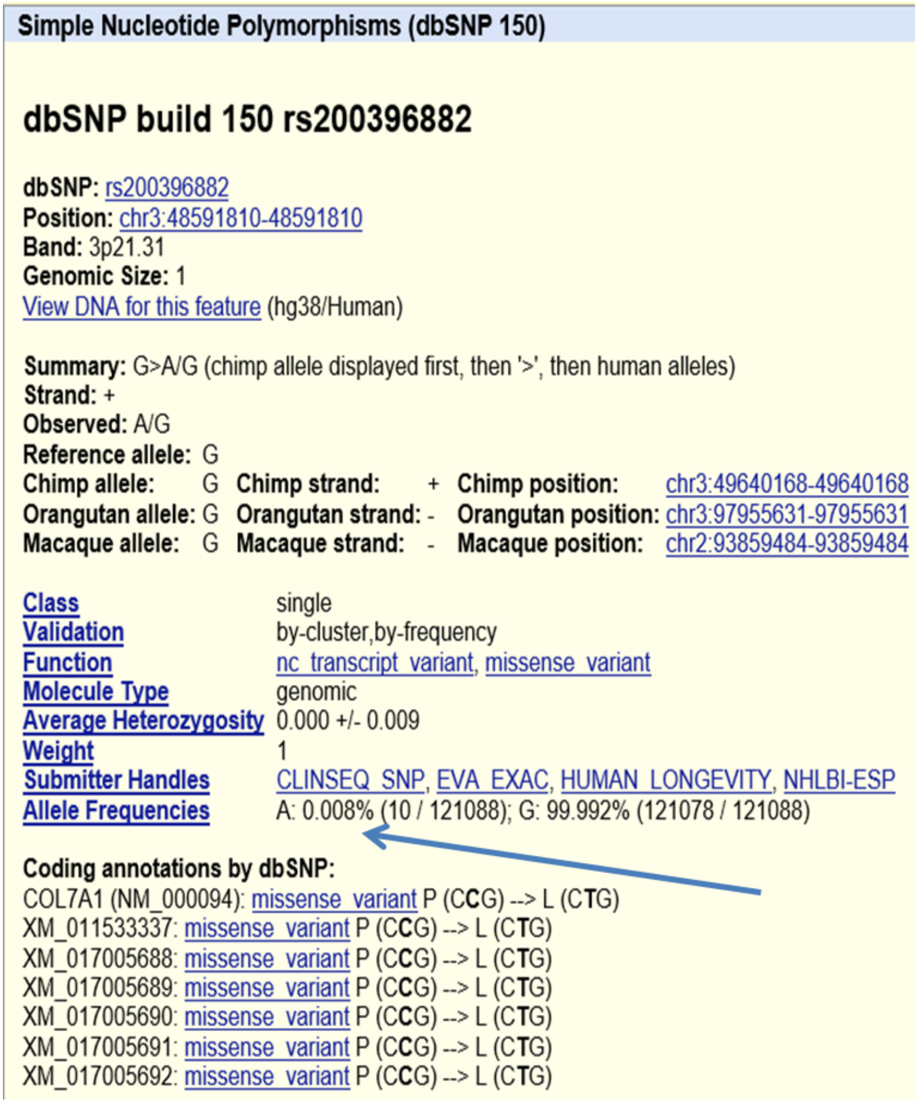
Supplementary Figure 2. shows the output of homozygosity mapping of the AutoSNPa software. The homozygous region (left side) shared by all 9 patients, but not with other normal family members, is boxed in red and the codon locations are indicated. The right side represents the homozygous regions of the parents and the normal siblings



Supplementary Figure 3. showing the chromatograms of a representative family sequencing, marked with a star in figure 1. The sex and relationship of all family members are shown. Two father, mother and two of the sisters are heterozygous while the other two are normal. The mutation is highlighted with a pink box



Supplementary Figure 4. showing the chromatograms of the second mutation. The missense mutation is shown for the same family in the supplementary figure 3. The patient is homozygous for the mutation. The father, mother and two of the sisters are heterozygous while the other two sisters are normal. The mutation is highlighted with a pink box



Simple Nucleotide Polymorphisms (dbSNP 150)

dbSNP build 150 rs200396882

dbSNP: rs200396882
Position: chr3:48591810-48591810
Band: 3p21.31
Genomic Size: 1
View DNA for this feature (hg38/Human)

Summary: G>A/G (chimp allele displayed first, then '>', then human alleles)
Strand: +
Observed: A/G
Reference allele: G

Chimp allele:	G	**Chimp strand:**	+	**Chimp position:**	chr3:49640168-49640168
Orangutan allele:	G	**Orangutan strand:**	-	**Orangutan position:**	chr3:97955631-97955631
Macaque allele:	G	**Macaque strand:**	-	**Macaque position:**	chr2:93859484-93859484

Class	single
Validation	by-cluster,by-frequency
Function	nc_transcript_variant, missense_variant
Molecule Type	genomic
Average Heterozygosity	0.000 +/- 0.009
Weight	1
Submitter Handles	CLINSEQ_SNP, EVA_EXAC, HUMAN_LONGEVITY, NHLBI-ESP
Allele Frequencies	A: 0.008% (10 / 121088); G: 99.992% (121078 / 121088)

Coding annotations by dbSNP:
COL7A1 (NM_000094): missense_variant P (CCG) --> L (CTG)
XM_011533337: missense_variant P (CCG) --> L (CTG)
XM_017005688: missense_variant P (CCG) --> L (CTG)
XM_017005689: missense_variant P (CCG) --> L (CTG)
XM_017005690: missense_variant P (CCG) --> L (CTG)
XM_017005691: missense_variant P (CCG) --> L (CTG)
XM_017005692: missense_variant P (CCG) --> L (CTG)

Supplementary Figure 5. presents the UCSC genomic browser output describing the second mutation characteristics. What is important here is the frequency of the change is 0.008% (pointed to it by an arrow)

Supplementary Table 1. List of primers used in this study

Exon	Forward primer	Reverse primer	Product size
Promoter	GGGACCTTGGTTTCCCTAAG	AGCCCGAGTCCTGGTCTT	624
1	CCCGAGATCAGGGACTTTTC	TGGAGTTGGCTGGGTTGT	411
2	ATCCCAAGTCCCAGTGAGG	GGTTTCCAGGGTCTCCTCC	422
3, 4	TGATACCCGTAACCCTCACC	ACTCAGTCACCCACATGCTC	539
5, 6	GAGTGACAGGTCAGCTGGG	AGCTGGGACGTTCACCTG	689
7, 8	GGCAGGAGGTAGGATGTCAG	TGGGCCTTGCAGACTCAG	551
9, 10	AGATGTGGGCTGAGGGG	GCCTGCAAGATAACAGGGTC	557
11	CCTTTCCTGGAACTTGGTG	CTCCAACAGGACCAGAGCT	448
11, 12	GTGAGACTGGTCAGTGCGG	TGGAGGTACACTCAGACCCC	575
13, 14	GTCCATCACTGCCATCGTC	TCAGAGGGAAATGCTGGC	563
15	AGGGACCAATGAGGGTATGG	GAGAAGCAAGGGTCTGCAAG	356
16, 17	AGACTCCCATCATCTTCCCC	CTGGGCAATCAGGAACACAC	586
18, 19	GACTTTCAGATCACCCCTGC	CTCCAATCCAGAGCACAAGC	555
20	GGTCTGAGAGGCTGGGC	CTACCTTGCGGAGTCTGCC	329
21	AACAGAGCCAGTGAAGTGGG	GAGTCACTCAGAGTCGGGG	353
22, 23	CAATAAGACAAACCGGACCC	GGGAACAAGTTACTGAAGCGG	565
24	TCCTGAGGCATCTGTCACAC	CCTATTGGGTGGTCAGGAGA	374

25, 26	CCCTGATGTGTTTCTCCAGG	GTCCAGTGGCTGCATGATAG	552
27, 28, 29	AGTAGGCTGATGGGGAAGGG	ATCAGTGAGACCTGTGCTGC	566
30, 31	GGGGAATAGAGAATGGGTTGA	TGTAAAAACCCCGAGACAGC	355
32, 33, 34	CCTTGTTGATGGGGTGAGG	TGCACAGGGTCTGGTGTG	579
35, 36	TGTCTAGGGGCTTCTTCCAG	ATCAAAGGGTCACAAGGGC	464
37	CCCCATTTGAACCCACATA	CTGACCTGCTGTTCTCTCC	335
37, 38, 39	ACCCCATGACCCTCATCAC	ACACGGGTGGGAAGACC	549
40, 41, 42	GGCTGACTTGATGATGTCCC	TGGAAAAGAAGAGGTCAGAGC	689
43	GGTGAGGCTGGAGAGAAGGT	TGAGGAGCAGGGGTAGCA	265
44	AGTCTTACCCCTGCCAACCT	GTGGCCCCTGAATGTAGAGA	315
45, 46	GACCAGGAGTGGGACTTTTG	CACTGTGGAATCACAGCCC	393
47, 48, 49, 50	TTCCATGTGACCTCCATTCC	GATGCAGGACGCTCGAAG	700
51, 52, 53	CAACCTCAAGGCTTCTGGG	CCCTCAGCCTTTCCTATCAC	652
56, 57, 58	AGTTGAGCCTGGGGCAAAC	CCTGAGGATAGGGGAGGAAG	656
59	GGGTGAGTGTGATTGGTCCT	CCACACCCTCTCACCTTTTC	271
60	GCCCACAGGTGAGTGATG	GGTCACCCTGTGGAAAATAGA	253
61	TGGGGGTTTCTCAGGACA	AATGAGGCTGGGGTCTAGG	273
62	GCCTTAGGCTCACAGGGA	ACCAACTCTCTCGGATGCTG	234
63, 64	TTTTGGGGTCCTCTTAGTCC	TTGCTACAGATCTTGGCTGTG	290
65, 66, 67	AACCATGGAGCTACAGGCAC	ATACGGCAGAACACAAAGGG	617
68, 69, 70	AGCCATGATGTTTCAATGGG	ATGGAGCCTCATGGCAAG	514
71, 72	GCTTTCCTGAGGCCGTG	GAAGAGAGAATGCTGGTGGC	466
73	GAAGTGGCTCAGTGGGTTGT	AAATGCAAATAGCGGGTGAG	409
74	GAGTGAGGGAAGAGGGGTTG	CCCTGCAGGAAACAAGAAAA	227
75	AGGAGAACGTGGAGAACAGG	CATATTTCTGGGGACCAAGC	274
76, 77, 78	CCCTATAACCCTCTGTGATCCTG	AGACCCCAGCCCTGCAC	584
79	CTCTGTCTAGGGGGATGGTG	AGACATGCACACACACAGCA	234
80	ATTGACCCCCAAGGAAAAAG	TCTGATGAGGGGGAGTTAGC	270
81, 82, 83	TGACCAGTACTGCCTCAGTTTC	GAGGAAGAGAAAGTTCAGGGC	596
84, 85, 86	CCTAGCTCACAGGTTAGGG	TGCCCCACTCCTCATATTTC	554
87, 88, 89	TGGGCCTGAAATATGAGGAG	GGTGGGTAAACTATGGGTCG	566
90	CCGCATATTTAAGCTCTGGC	CAAGAGGCCCCTTATGC	370
91A	GAAGGCTCAGTCCAGACACC	AACCCCAACACGTCCACTC	530
91B	AGATCCCAGGACACACCTGA	CTCAGACATGCGACCAAGAA	376
92, 93, 94	GAGCGTCCCTCTGTCTGTG	CCCCTCAAGACTGGGAAC	742
95, 96, 97	TTCCCAGTCTTGAGGGG	CATTGACTCAGAGGTTGGAAATC	627
98	CACAGGACCCTCACCTCACT	TGTACCCCAGGACTCTCTGC	512
99, 100	CACAGGACCCTCACCTCACT	CAGGCAGGAATCAGAGGAGT	454
101, 102	AAGGGTGACAAGGTGAGTGTG	CCCTCCTGCCCTCACAG	477
103, 104	GGGCTCGTTGTATTCTAAGCC	AGCTACCACACTGGTGGGAC	545
105, 106, 107	GTCCCCCACCTCCACAGTAG	CGATCAGGCCCTCCTTG	821
108	GGAGAGATGGTGAGTGTGGG	ATGACCCGACCATGAGCTTC	251
109, 110	GTTGGGGTCACAGTGGAGTT	ATCAGAGTCAGGCAGGTTGG	393
111	ATGGGAGTTCAGGGAGGTTC	CCAACCTGCCTGACTCTGAT	517
112	CAGAGGCCCCGAAGGACT	CTACCTCCCTCACCCAGCAC	543
113	AGAGAGGGGAGCAGGTGAGT	TACAGGGCAAGCAGTCAAGA	537
114, 115	TCTCTACCCCTCTGCCTGTG	GCCCTGCATTCATGGACAC	464
116, 117	GGAAGGTGAGGACAGCTGAA	GTCTCCTCCAGGACCCTGAC	777
118	GTAGGGTCCCCTTCCATGTT	CTGTACCCATGCAGAATAATACCA	644