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Supplemental Data

Using VAAST to Identify an X-linked Disorder Resulting in Lethality in Male Infants Due to N-Terminal Acetyltransferase Deficiency

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Table S1. All Genotyped Samples (in the Shared Haplotype Region) from Family 1

Sample Name	Marker	Allele 1	Allele 2
III-4 (recent male)	**DXS1227 (Xq27.2)**		**90**
II-2 (carrier mother)		**82**	**90**
III-2 (unaffected brother)		**82**	
I-2 (carrier grandmother)		**82**	**90**
II-2 (carrier aunt)		**82**	**82**
II-5 (carrier aunt)		**82**	**82**
II-6 (FFPE tissue)			**90**
III-7 (FFPE tissue)		**82**	
III-4 (recent male)	**DXS8043 (Xq27.3)**		**170**
II-2 (carrier mother)		**156**	**170**
III-2 (unaffected brother)		**156**	
I-2 (carrier grandmother)		**156**	**170**
II-2 (carrier aunt)		**156**	**170**
II-5 (carrier aunt)		**156**	**170**
II-6 (FFPE tissue)			**170**
III-7 (FFPE tissue)			**170**
III-4 (recent male)	**DXS8091 (Xq28)**		**83**
II-2 (carrier mother)		**70**	**83**
III-2 (unaffected brother)		**70**	
I-2 (carrier grandmother)		**70**	**83**
II-2 (carrier aunt)		**70**	**83**
II-5 (carrier aunt)		**70**	**83**
II-6 (FFPE tissue)			**83**
III-7 (FFPE tissue)			**83**
III-4 (recent male)	**DXS1073 (Xq28)**	**310**	
II-2 (carrier mother)		**310**	**310**
III-2 (unaffected brother)		**310**	
I-2 (carrier grandmother)		**310**	**321**
II-2 (carrier aunt)		**310**	**310**
II-5 (carrier aunt)		**310**	**310**
II-6 (FFPE tissue)		**310**	
III-7 (FFPE tissue)		**310**	

Table S2. Gene Coverage in Family 1 in Shared Haplotype 14 MB Region

gene	name	exonlength	Coverage count	Gene coverage
NM_005638	*VAMP7*	663	76	0.11
NM_001145149	*VAMP7*	540	66	0.12
NM_001144064	*CXorf51*	327	57	0.17
NM_005840	*SPRY3*	8584	4180	0.49
NM_002186	*IL9R*	1566	2149	1.37
NM_001136273	*ZFP92*	1250	6090	4.87
NM_001666	*ARHGAP4*	2841	198326	69.81
NM_001164741	*ARHGAP4*	2961	210991	71.26
NM_001025243	*IRAK1*	1902	146954	77.26
NM_001025242	*IRAK1*	2049	161731	78.93
NM_001569	*IRAK1*	2139	169309	79.15
NM_080701	*TREX2*	711	56311	79.20
NM_001163257	*PLXNB3*	5799	473352	81.63
NM_006014	*LAGE3*	432	35802	82.88
NM_005393	*PLXNB3*	5730	475950	83.06
NM_001039582	*PNCK*	1281	118389	92.42
NM_001007524	*F8A3*	1116	104709	93.83
NM_001007523	*F8A2*	1116	104709	93.83
NM_014235	*UBL4A*	474	48986	103.35
NM_020994	*CTAG2*	633	66416	104.92
NM_005334	*HCFC1*	6108	644690	105.55
NM_004699	*FAM50A*	1020	108286	106.16
NM_006280	*SSR4*	522	55629	106.57
NM_001135740	*PNCK*	1083	118024	108.98
NM_001145255	*IKBKG*	963	106751	110.85
NM_002910	*RENBP*	1284	143731	111.94
NM_001395	*DUSP9*	1155	133833	115.87
NM_001099856	*IKBKG*	1464	170283	116.31
NM_001099857	*IKBKG*	1260	148104	117.54
NM_003639	*IKBKG*	1260	148104	117.54
NM_001009613	*SPANXN4*	300	35746	119.15
NM_014370	*SRPK3*	1704	204129	119.79
NM_032512	*PDZD4*	2310	278822	120.70
NM_017518	*HAUS7*	1107	137547	124.25
NM_001146151	*AVPR2*	930	117577	126.43
NM_001142391	*SLC10A3*	1347	171581	127.38
NM_021806	*FAM3A*	693	89835	129.63
NM_181312	*TAZ*	837	109944	131.35
NM_005629	*SLC6A8*	1908	250823	131.46
NM_001142805	*SLC6A8*	1878	248368	132.25

NM_000033	*ABCD1*	2238	296381	132.43
NM_000116	*TAZ*	879	116737	132.81
NM_004135	*IDH3G*	1182	157553	133.29
NM_001142392	*SLC10A3*	1576	212032	134.54
NM_019848	*SLC10A3*	1576	212032	134.54
NM_017514	*PLXNA3*	5616	756963	134.79
NM_174869	*IDH3G*	1143	154064	134.79
NM_006730	*DNASE1L1*	909	125626	138.20
NM_001009934	*DNASE1L1*	909	125626	138.20
NM_001009932	*DNASE1L1*	909	125626	138.20
NM_001009933	*DNASE1L1*	909	125626	138.20
NM_001142806	*SLC6A8*	1563	224607	143.70
NM_172377	*CTAG2*	543	79306	146.05
NM_181313	*TAZ*	747	109944	147.18
NM_000054	*AVPR2*	1116	164406	147.32
NM_001130997	*FAM58A*	685	101245	147.80
NM_181311	*TAZ*	789	116737	147.96
NM_001110792	*MECP2*	1497	224298	149.83
NM_001110556	*FLNA*	7944	1193079	150.19
NM_001456	*FLNA*	7920	1191123	150.39
NM_000117	*EMD*	765	115182	150.56
NM_007150	*ZNF185*	2070	313025	151.22
NM_134445	*CD99L2*	573	88663	154.73
NM_000402	*G6PD*	1638	254551	155.40
NM_004992	*MECP2*	1461	229822	157.30
NM_032882	*PNMA6A*	1957	309141	157.97
NM_134446	*CD99L2*	642	103384	161.03
NM_001042351	*G6PD*	1548	252605	163.18
NM_003491	*ARD1A/NAA10*	708	116410	164.42
NM_005362	*MAGEA3*	1010	168223	166.56
NM_152274	*FAM58A*	745	124274	166.81
NM_031462	*CD99L2*	789	138330	175.32
NM_001711	*BGN*	1107	195912	176.98
NM_001183	*ATP6AP1*	1413	258088	182.65
NM_003492	*TMEM187*	1133	208791	184.28
NM_024332	*BRCC3*	951	176266	185.35
NM_001017980	*VMA21*	306	56795	185.60
NM_003372	*VBP1*	594	110381	185.83
NM_000425	*L1CAM*	3774	704030	186.55
NM_001143963	*L1CAM*	3747	700493	186.95
NM_024003	*L1CAM*	3762	703539	187.01
NM_020061	*OPN1LW*	1095	207583	189.57
NM_013364	*PNMA3*	3415	667881	195.57
NM_001018055	*BRCC3*	876	176239	201.19

NM_001001344	*ATP2B3*	3663	742325	202.65
NM_024082	*PRRG3*	696	142405	204.60
NM_004709	*CXorf1*	2175	450642	207.19
NM_001080485	*ZNF275*	1290	272184	211.00
NM_001289	*CLIC2*	744	157176	211.26
NM_021949	*ATP2B3*	3522	749865	212.91
NM_001166461	*MPP1*	1341	293285	218.71
NM_001102576	*CSAG1*	237	52823	222.88
NM_153478	*CSAG1*	237	52823	222.88
NM_002436	*MPP1*	1401	312633	223.15
NM_033085	*FATE1*	552	123719	224.13
NM_001166460	*MPP1*	1350	302988	224.44
NM_001586	*TEX28*	1233	278056	225.51
NM_001139457	*BCAP31*	942	215987	229.29
NM_002024	*FMR1*	1899	440871	232.16
NM_005491	*MAMLD1*	2325	543360	233.70
NM_001081573	*GAB3*	1764	412959	234.10
NM_080612	*GAB3*	1761	412558	234.27
NM_001493	*GDI1*	1344	315670	234.87
NM_005342	*HMGB3*	603	142303	235.99
NM_001166462	*MPP1*	1311	310033	236.49
NM_080720	*H2AFB3*	509	120669	237.07
NM_001018025	*MTCP1*	324	80149	247.37
NM_177456	*MAGEC3*	1180	292522	247.90
NM_003828	*MTMR1*	1998	501537	251.02
NM_138702	*MAGEC3*	1932	497438	257.47
NM_005745	*BCAP31*	741	191643	258.63
NM_001139441	*BCAP31*	741	191643	258.63
NM_001363	*DKC1*	1545	405920	262.73
NM_001142463	*DKC1*	1530	403674	263.84
NM_171998	*RAB39B*	642	171222	266.70
NM_016249	*MAGEC2*	1188	317683	267.41
NM_152578	*FMR1NB*	768	208573	271.58
NM_173493	*PASD1*	2322	641988	276.48
NM_005366	*MAGEA11*	1290	360301	279.30
NM_001011544	*MAGEA11*	1203	343389	285.44
NM_019863	*F8*	651	186117	285.89
NM_001145933	*TKTL1*	1773	506976	285.94
NM_012253	*TKTL1*	1791	512677	286.25
NM_021048	*MAGEA10*	1175	336728	286.58
NM_001011543	*MAGEA10*	1175	336728	286.58
NM_021049	*MAGEA5*	440	126819	288.23
NM_005462	*MAGEC1*	3429	989927	288.69
NM_000132	*F8*	7056	2046969	290.10

NM_004961	*GABRE*	1521	443792	291.78
NM_001145934	*TKTL1*	1623	475981	293.27
NM_002025	*AFF2*	3936	1155849	293.66
NM_004344	*CETN2*	519	152631	294.09
NM_002362	*MAGEA4*	1506	444020	294.83
NM_001011548	*MAGEA4*	1506	444020	294.83
NM_001011549	*MAGEA4*	1506	444020	294.83
NM_001011550	*MAGEA4*	1506	444020	294.83
NM_006013	*RPL10*	645	191534	296.95
NM_000252	*MTM1*	1812	544031	300.24
NM_018196	*TMLHE*	1266	383237	302.71
NM_006123	*IDS*	1032	313421	303.70
NM_001012989	*UBE2NL*	1155	354027	306.52
NM_001166401	*MAGEA8*	1563	480864	307.65
NM_001166400	*MAGEA8*	1563	480864	307.65
NM_005364	*MAGEA8*	1563	480864	307.65
NM_000202	*IDS*	1653	519671	314.38
NM_001144006	*SLITRK2*	3415	1084285	317.51
NM_001144009	*SLITRK2*	3415	1084285	317.51
NM_001144004	*SLITRK2*	3415	1084285	317.51
NM_001144010	*SLITRK2*	3415	1084285	317.51
NM_001144003	*SLITRK2*	3415	1084285	317.51
NM_001144007	*SLITRK2*	3415	1084285	317.51
NM_001144008	*SLITRK2*	3415	1084285	317.51
NM_032539	*SLITRK2*	3415	1084285	317.51
NM_001144005	*SLITRK2*	3415	1084285	317.51
NM_001009614	*SPANXN1*	219	69555	317.60
NM_175743	*MAGEA2*	1500	483833	322.56
NM_052926	*PNMA5*	1470	475993	323.80
NM_001103150	*PNMA5*	1470	475993	323.80
NM_001103151	*PNMA5*	1470	475993	323.80
NM_001129765	*NSDHL*	1122	366022	326.22
NM_015922	*NSDHL*	1122	366022	326.22
NM_001166550	*IDS*	1383	461914	333.99
NM_001327	*CTAG1B*	543	183463	337.87
NM_139250	*CTAG1A*	543	183463	337.87
NM_023934	*FUNDC2*	570	194246	340.78
NM_001009609	*SPANXN3*	426	145991	342.70
NM_173078	*SLITRK4*	2564	893820	348.60
NM_001018024	*MTCP1NB*	207	75629	365.36
NM_018558	*GABRQ*	1899	697085	367.08
NM_005140	*CNGA2*	1995	732889	367.36
NM_032508	*TMEM185A*	1053	411578	390.86
NM_004988	*MAGEA1*	995	391414	393.38

NM_000808	*GABRA3*	1479	581868	393.42
NM_004224	*GPR50*	1854	840571	453.38
NM_178124	*CXorf40A*	477	222061	465.54
NM_001013845	*CXorf40B*	477	226563	474.97
NM_001166387	*MAGEA12*	1020	485475	475.96
NM_001166386	*MAGEA12*	1020	485475	475.96
NM_005367	*MAGEA12*	1020	485475	475.96
NM_001048181	*OPN1MW2*	1095	523946	478.49
NM_000513	*OPN1MW*	1095	523946	478.49
NM_004909	*CSAG2*	333	160163	480.97
NM_001129828	*CSAG3*	333	160163	480.97
NM_001009615	*SPANXN2*	543	265691	489.30
NM_001080848	*CSAG2*	384	211170	549.92
NM_001129826	*CSAG3*	384	211170	549.92
NM_175868	*MAGEA6*	1504	882627	586.85
NM_005363	*MAGEA6*	1504	882627	586.85
NM_001080790	*MAGEA9B*	1013	610417	602.58
NM_005365	*MAGEA9*	1527	1000365	655.12
NM_016153	*HSFX1*	1272	974432	766.06
NM_001164415	*HSFX2*	1272	974432	766.06
NM_001017991	*H2AFB2*	348	268548	771.69
NM_012151	*F8A1*	1131	1008804	891.96
NM_153488	*MAGEA2B*	1010	1117400	1106.34
NM_175742	*MAGEA2*	1010	1118700	1107.62
NM_005361	*MAGEA2*	1010	1118700	1107.62
NM_001017990	*H2AFB1*	356	412992	1160.09

Figure 1. Sanger Sequencing Validation of *NAA10* in Family 1. III-4: Recent affected boy (hemizygous: c.109T>C; p.S37P)

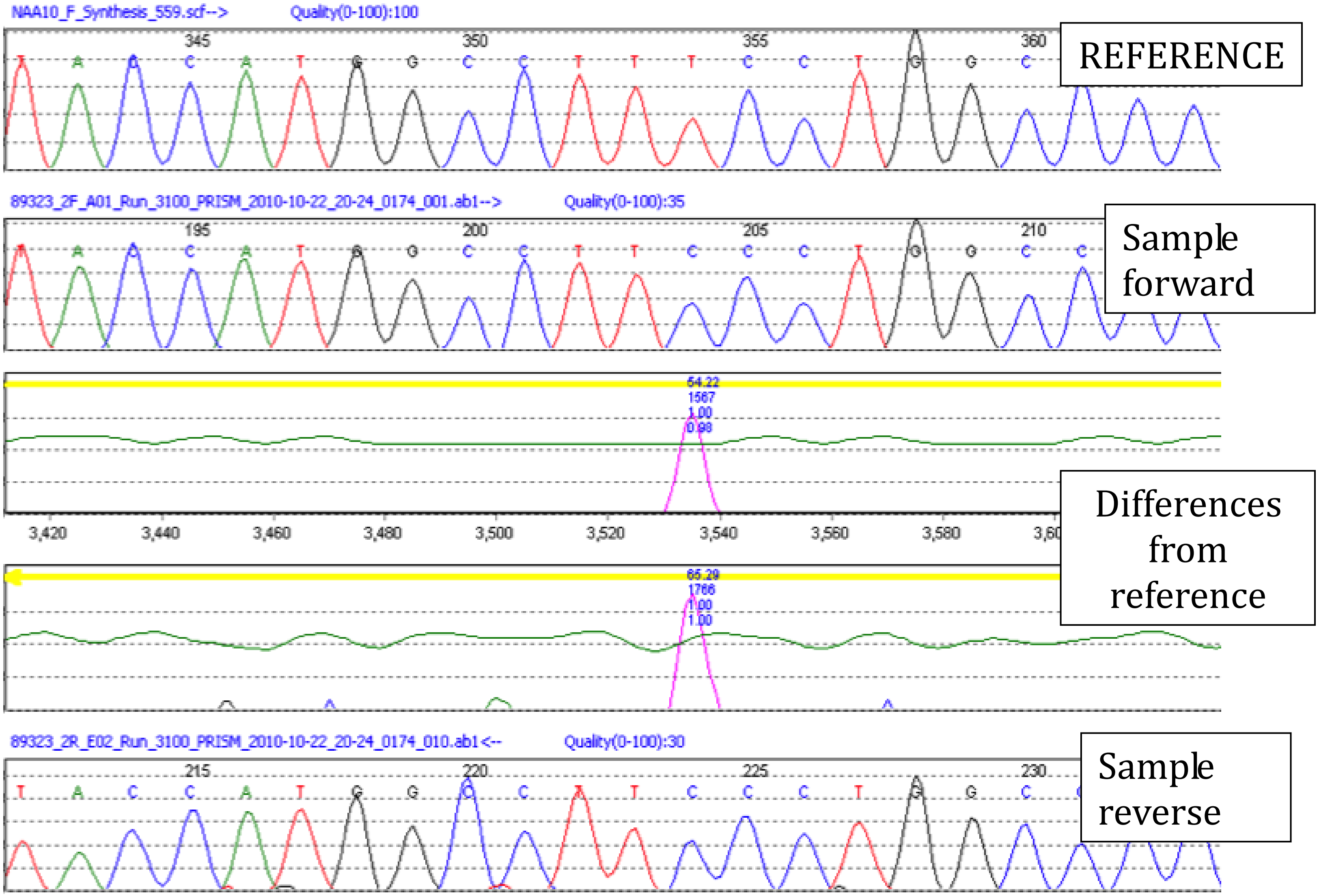

Figure S2. Sanger Sequencing Validation of *NAA10*. III-2: Unaffected brother (wild-type)

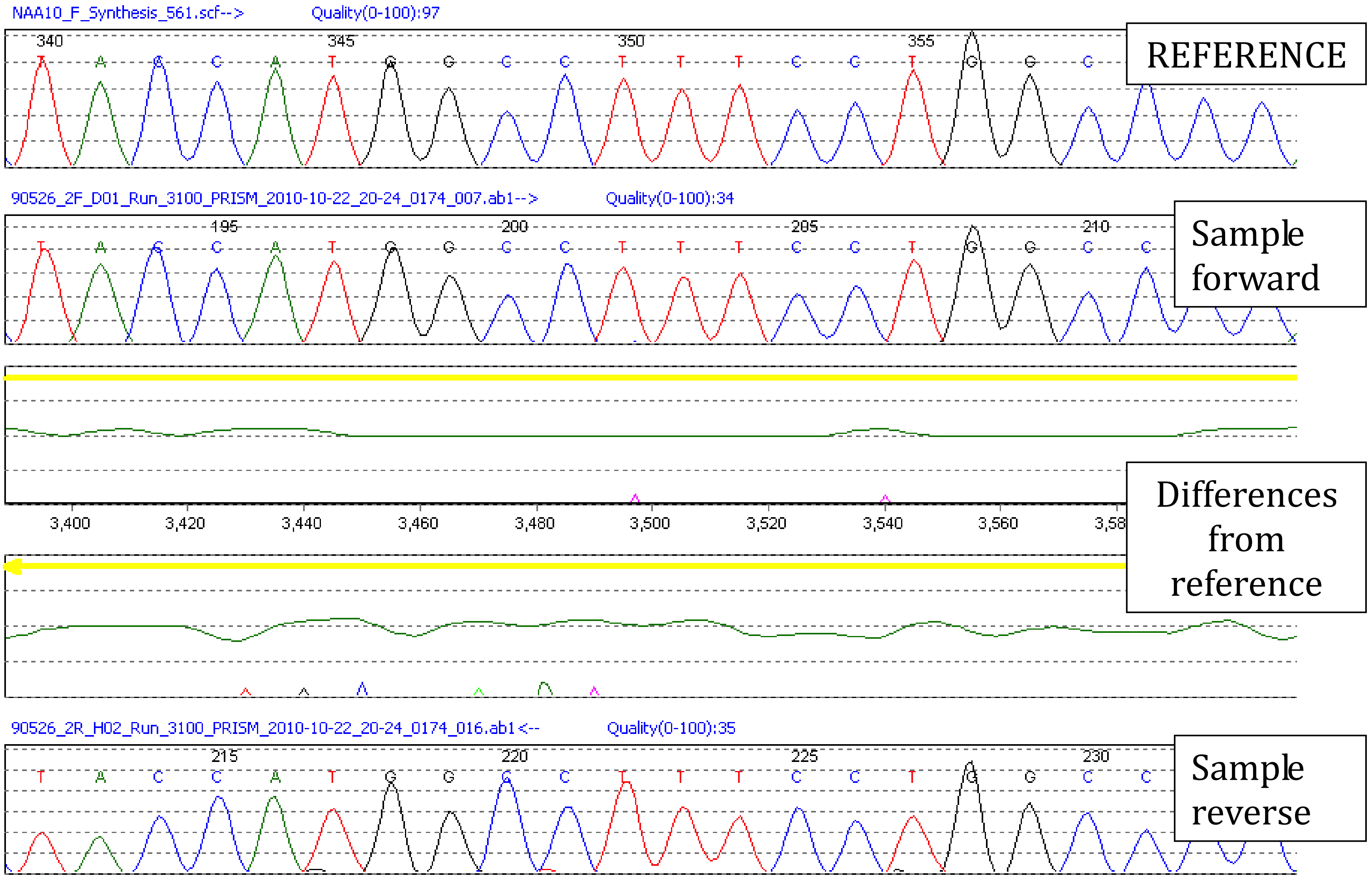

Figure S3. Sanger Sequencing Validation of *NAA10*. II-3: Aunt, Carrier (heterozygous: c.109T>C; p.S37P)

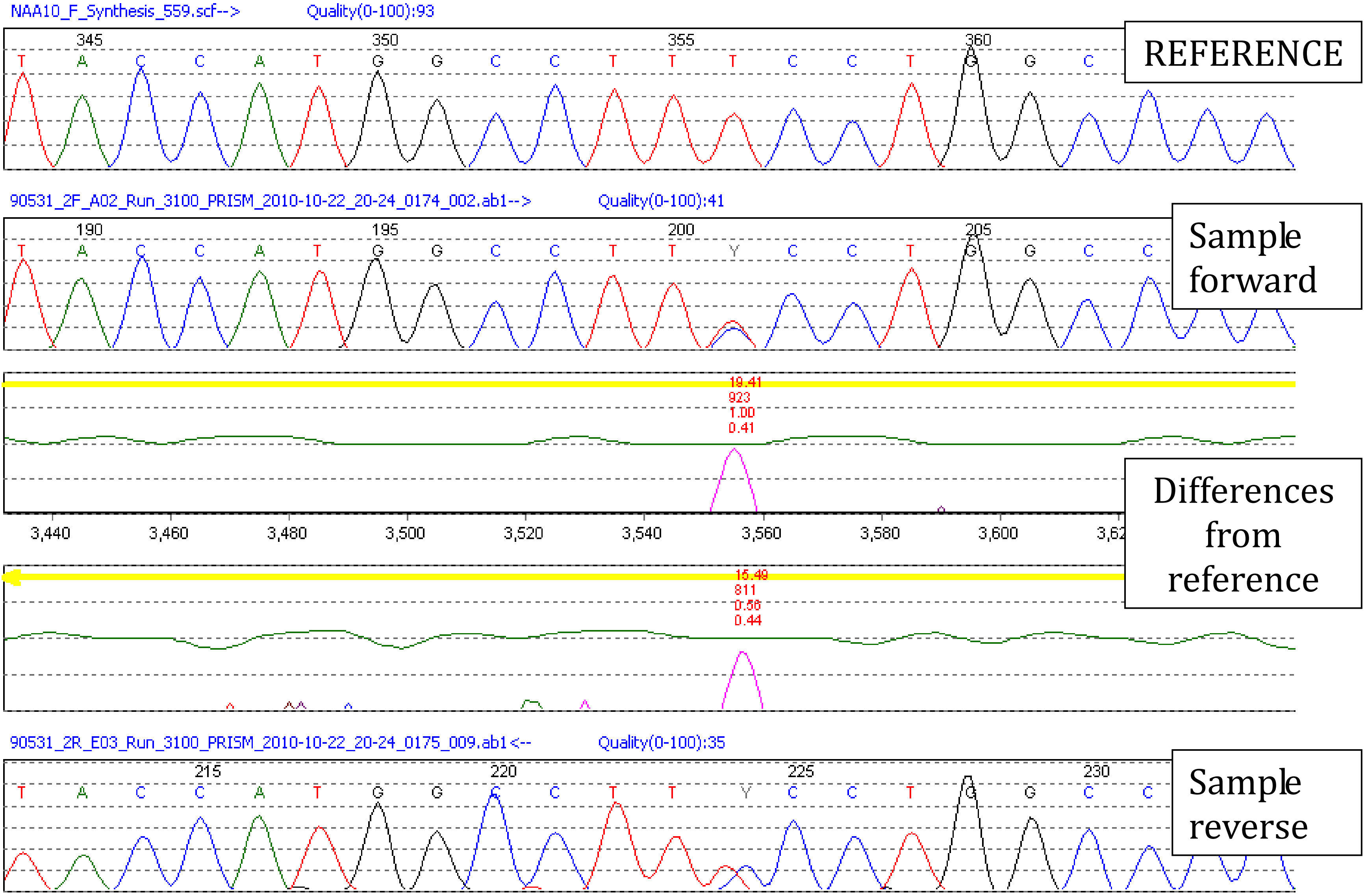

Figure S4: Sanger Sequencing Validation of *NAA10*. PCA-50-08-3A: FFPE sample from affected boy (hemizygous: c.109T>C; p.S37P)

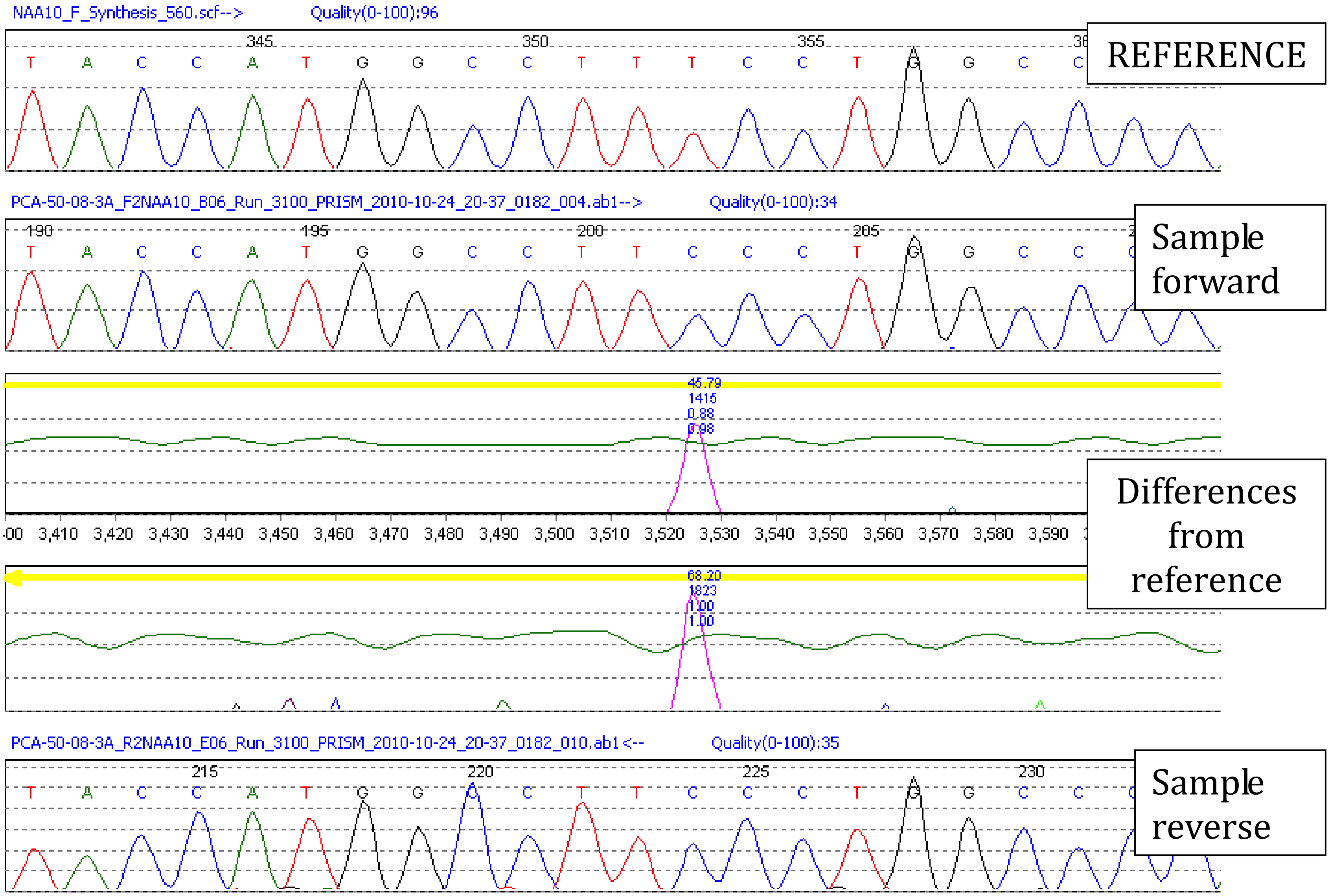

Figure S5. Sanger Sequencing Validation of *NAA10*. PA-25-88-2: FFPE sample from affected boy (hemizygous: c.109T>C; p.S37P)

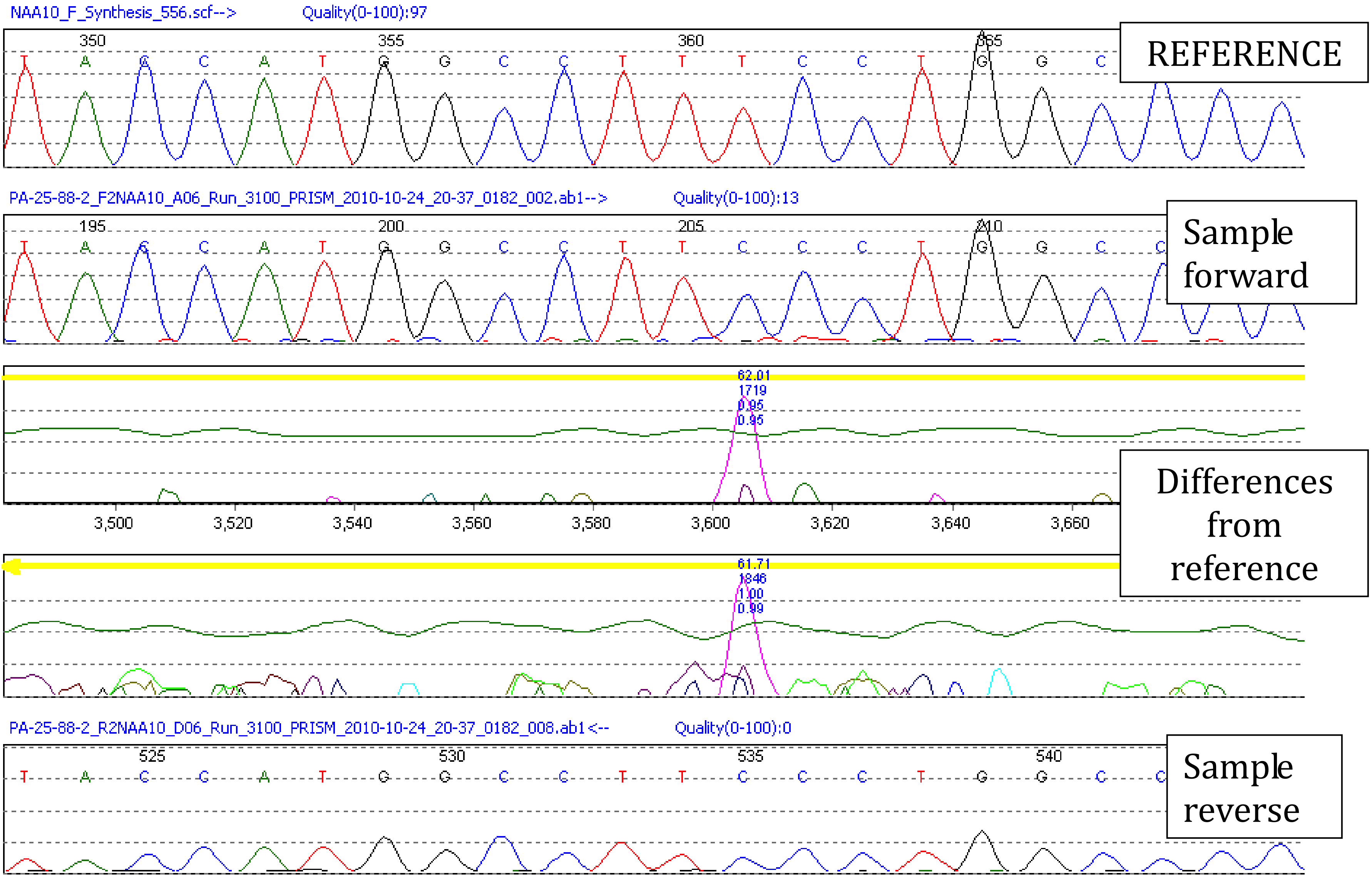

Figure S6. Picture of III-6 in Family 1