

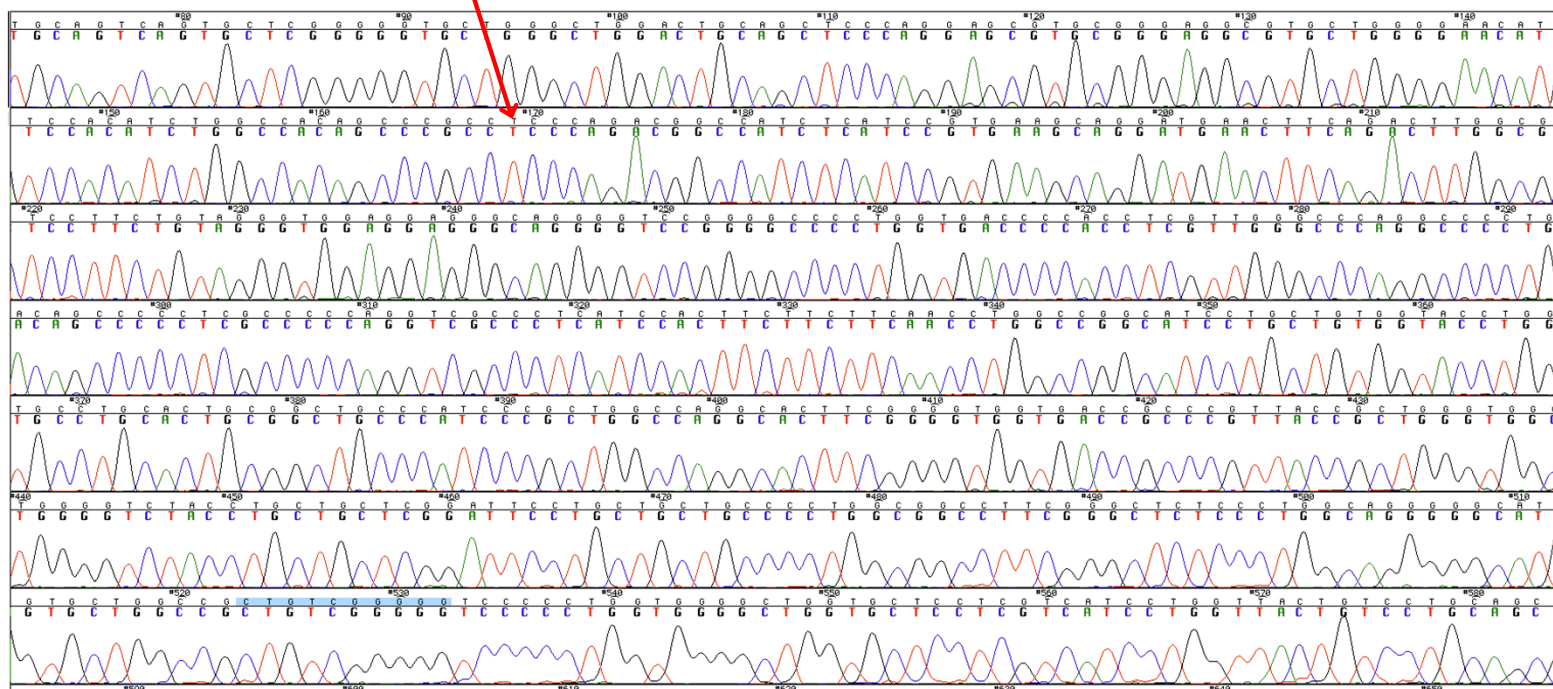
**Supplemental Data for “Digenic Heterozygous Mutations in SLC34A3 and SLC34A1
Cause Dominant Hypophosphatemic Rickets with Hypercalciuria”**

Supplemental Figure 1. Sanger sequencing of *SLC34A3* and rs28434439 alternative alleles.

Sanger sequencing of subcloned DNA fragments showing mutant (A) and wild type (B) alleles, demonstrating that the *SLC34A3* mutant allele and the rs28434439 alternative allele are not linked together (Panels A through H). PCR sequences from subject III-6 shows heterozygosity (T/C) for rs28434439 (Panel I) indicating presence of paternal wild type allele (A), and by extension, confirming the maternal origin of mutant *SLC34A3* allele. These observations confirm the imputation results that the shared haplotype between father and mother carries the *SLC34A3* mutation.

A. (II-4)

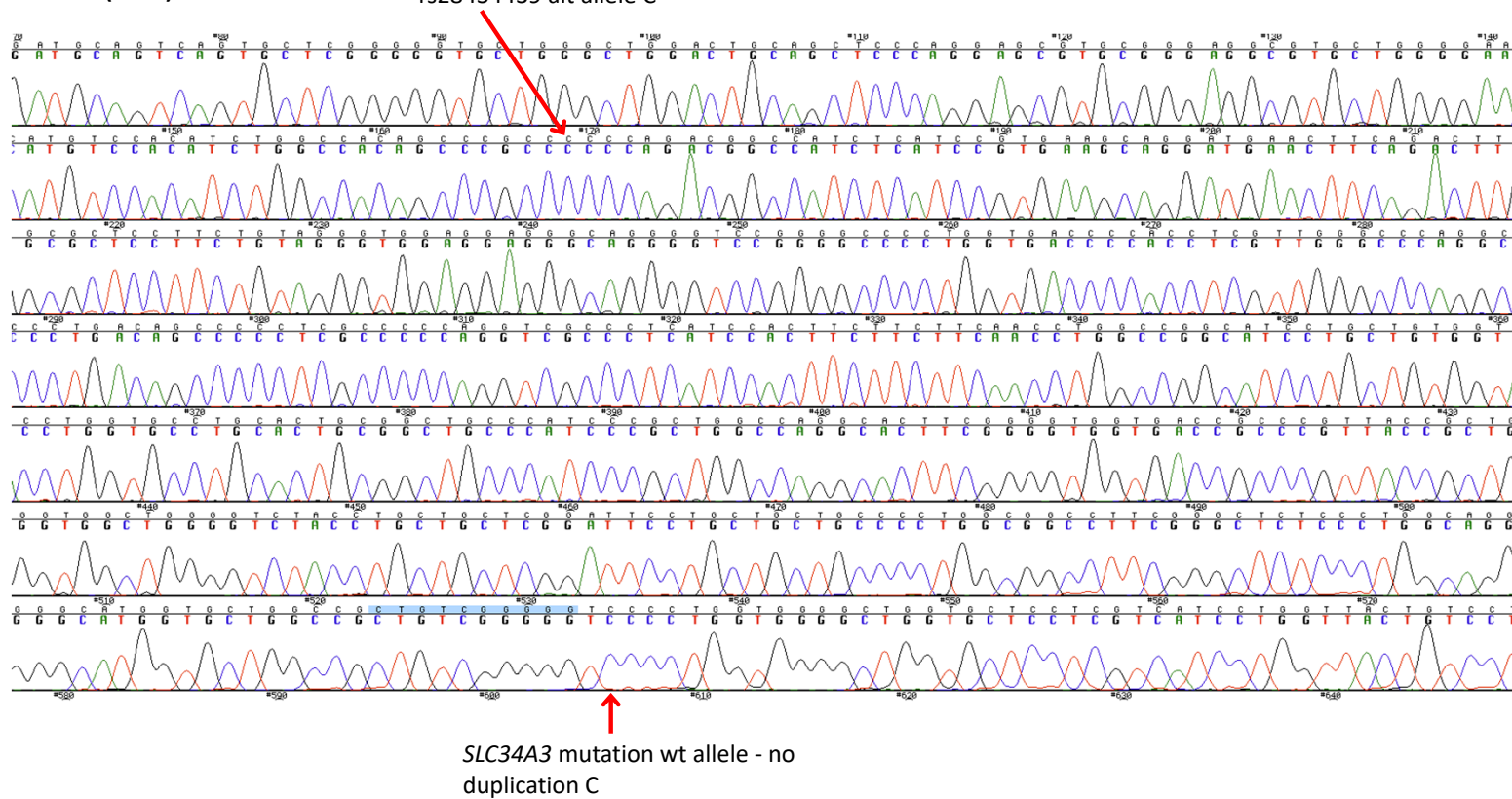
rs28434439 wt allele T



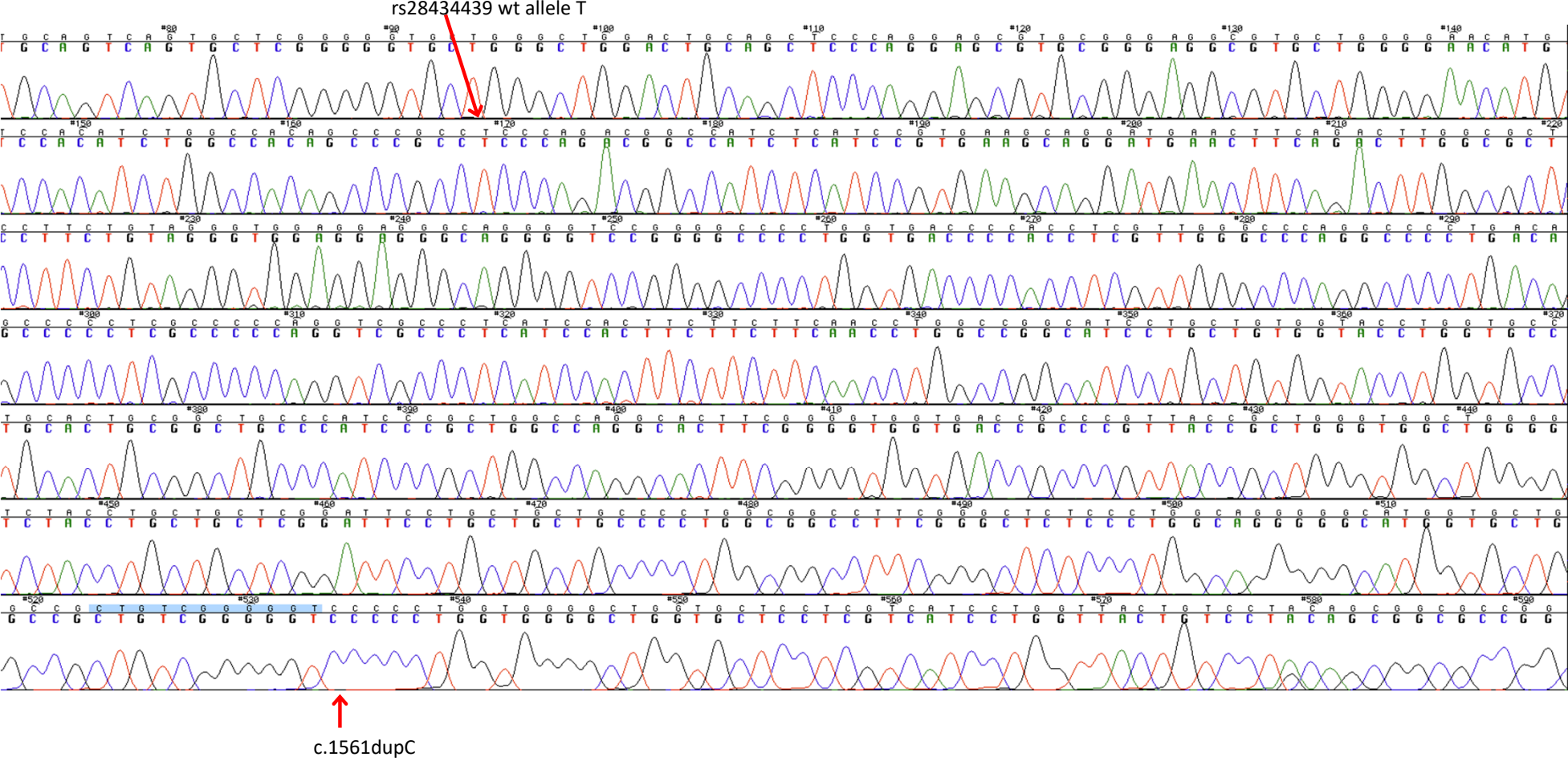
c.1561dupC

B. (11-4)

rs28434439 alt allele C

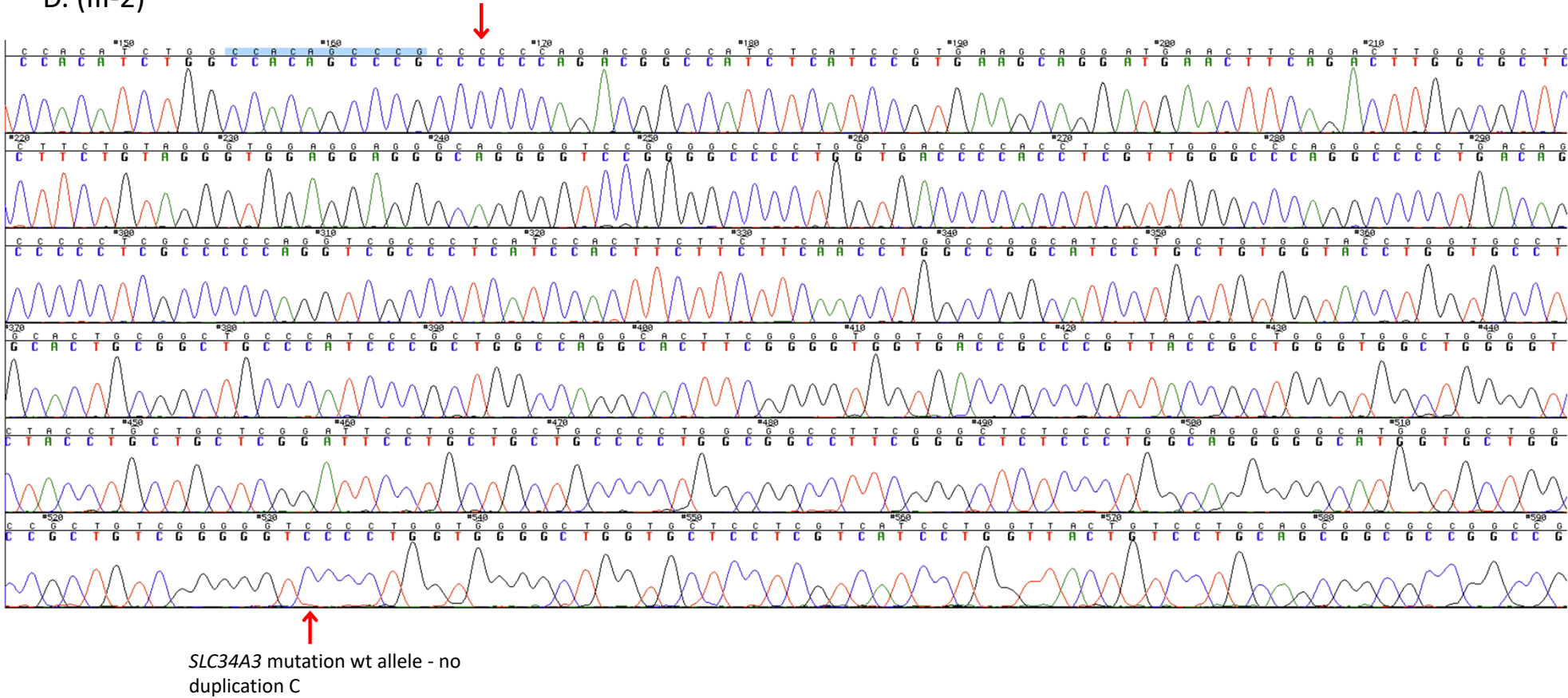


C. (III-2)

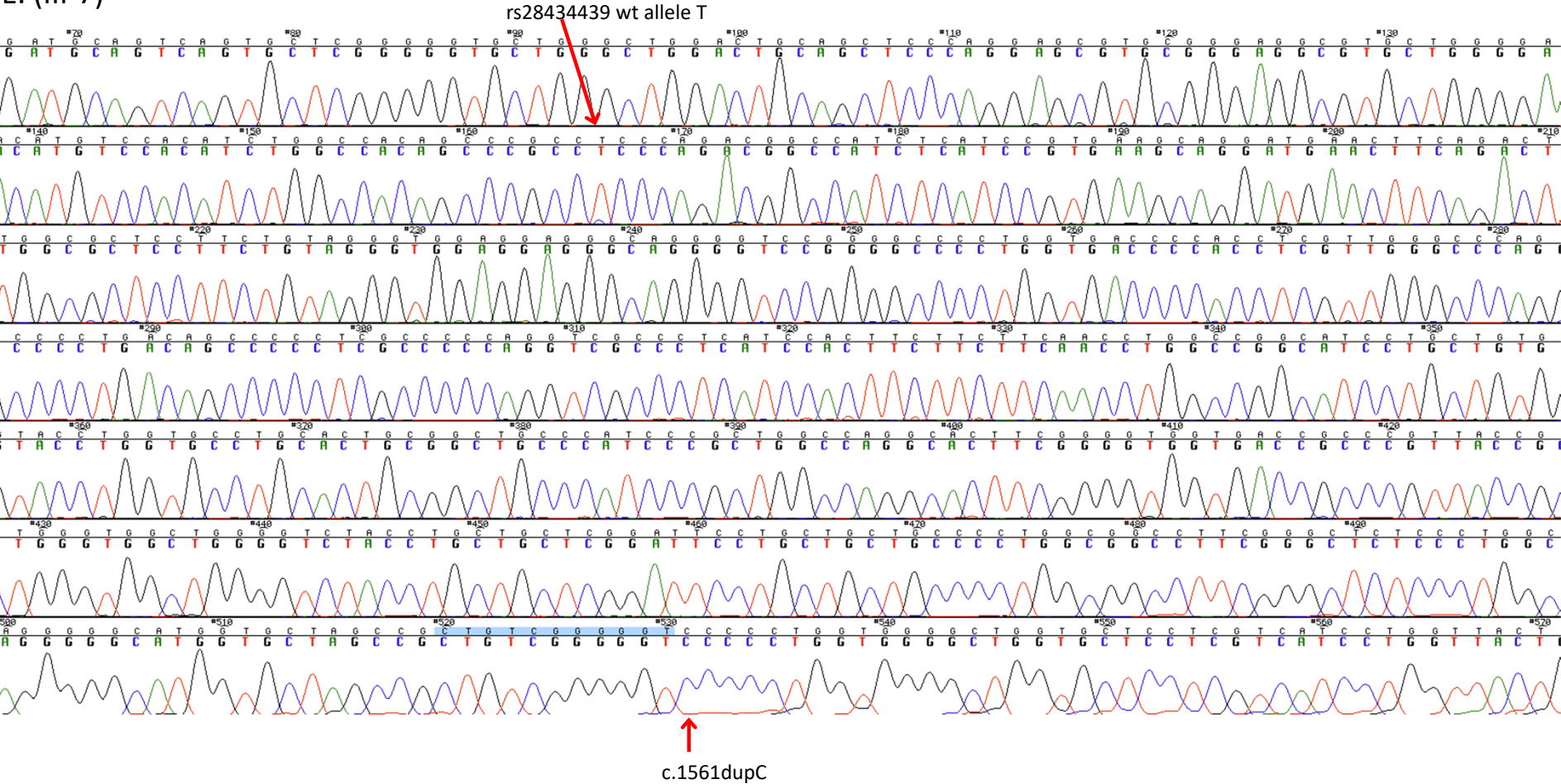


D. (III-2)

rs28434439 alt allele C

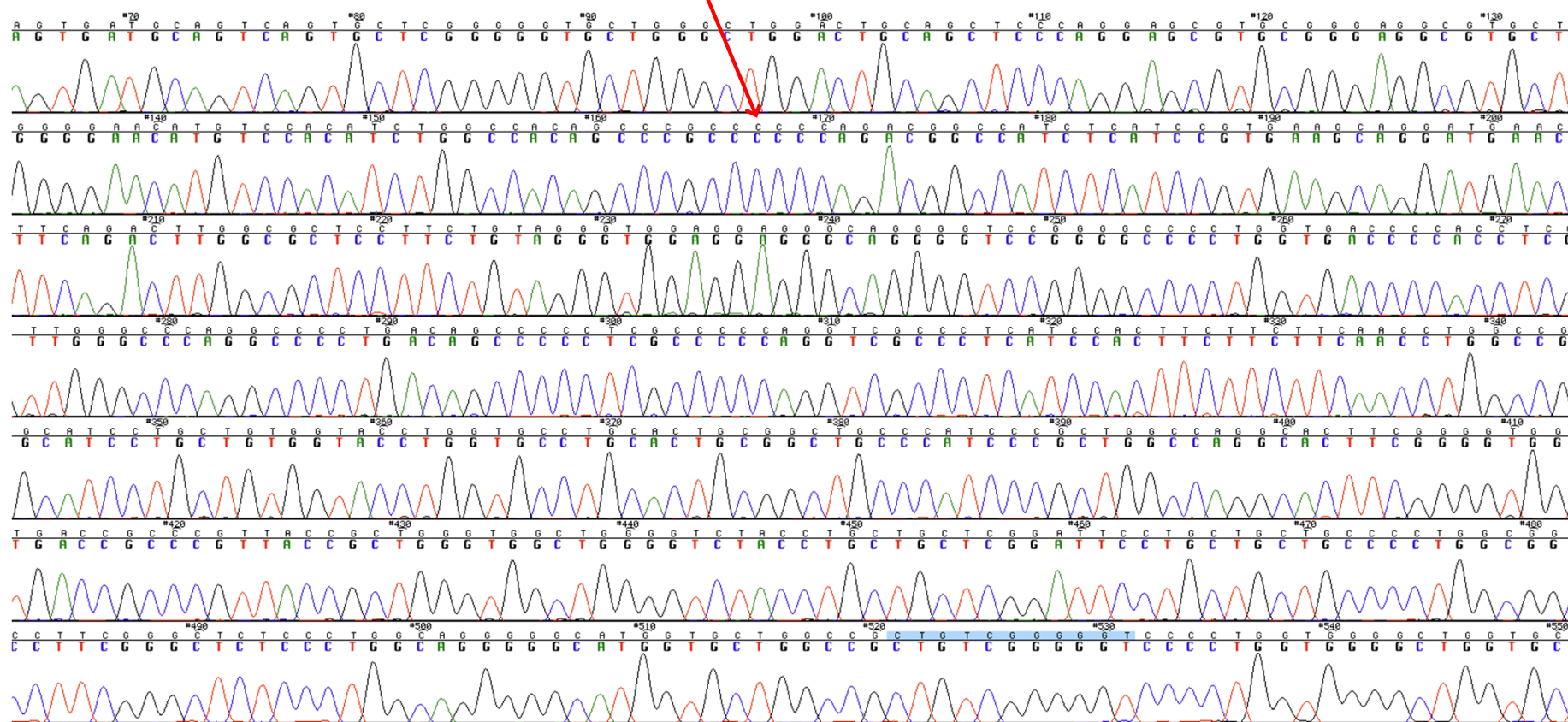


E. (III-7)



F. (III-7)

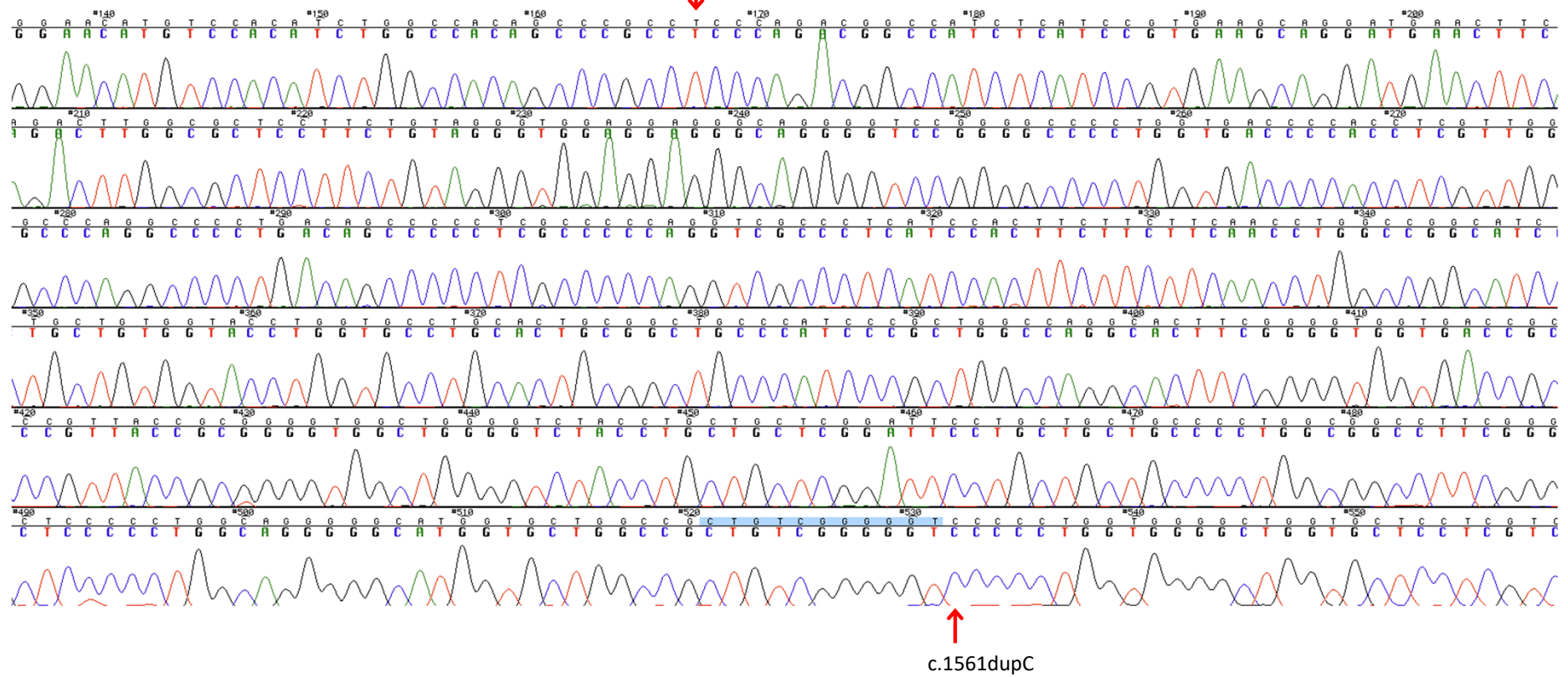
rs28434439 alt allele C



SLC34A3 mutation wt allele - no
duplication C

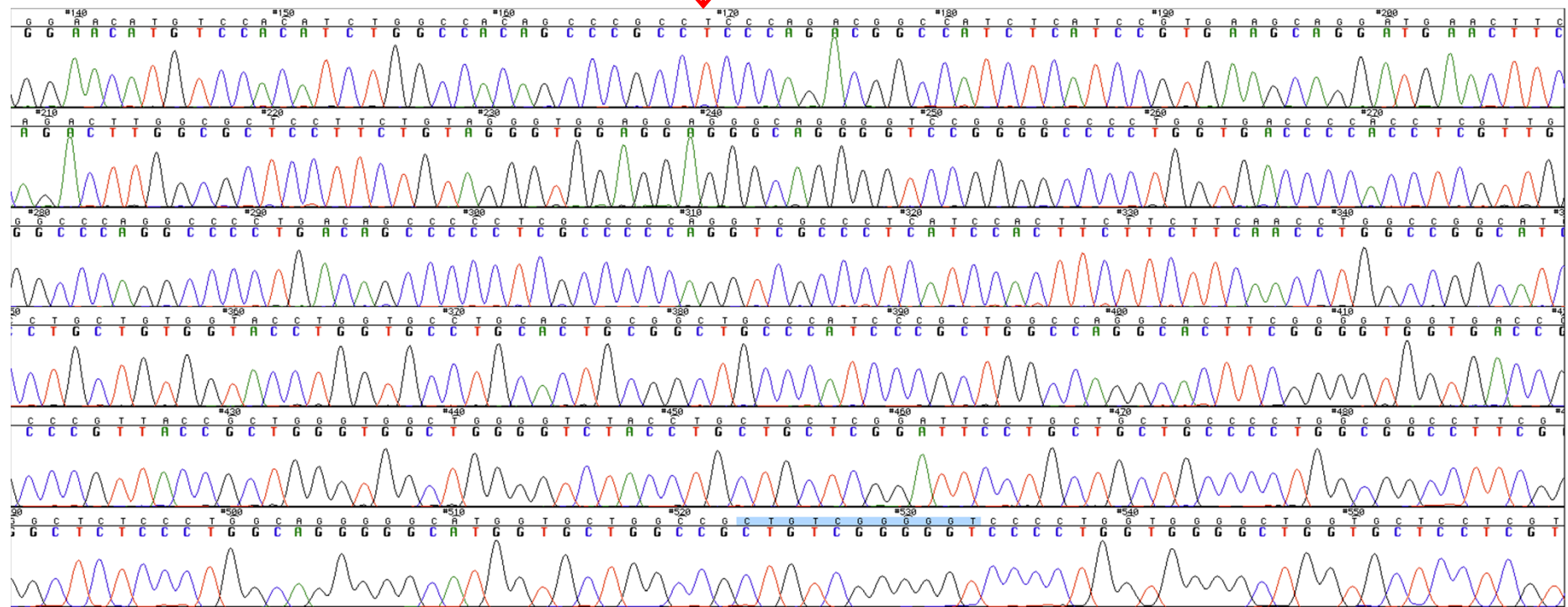
G. (II-5)

rs28434439 wt allele T



H. (II-5)

rs28434439 wt allele T*

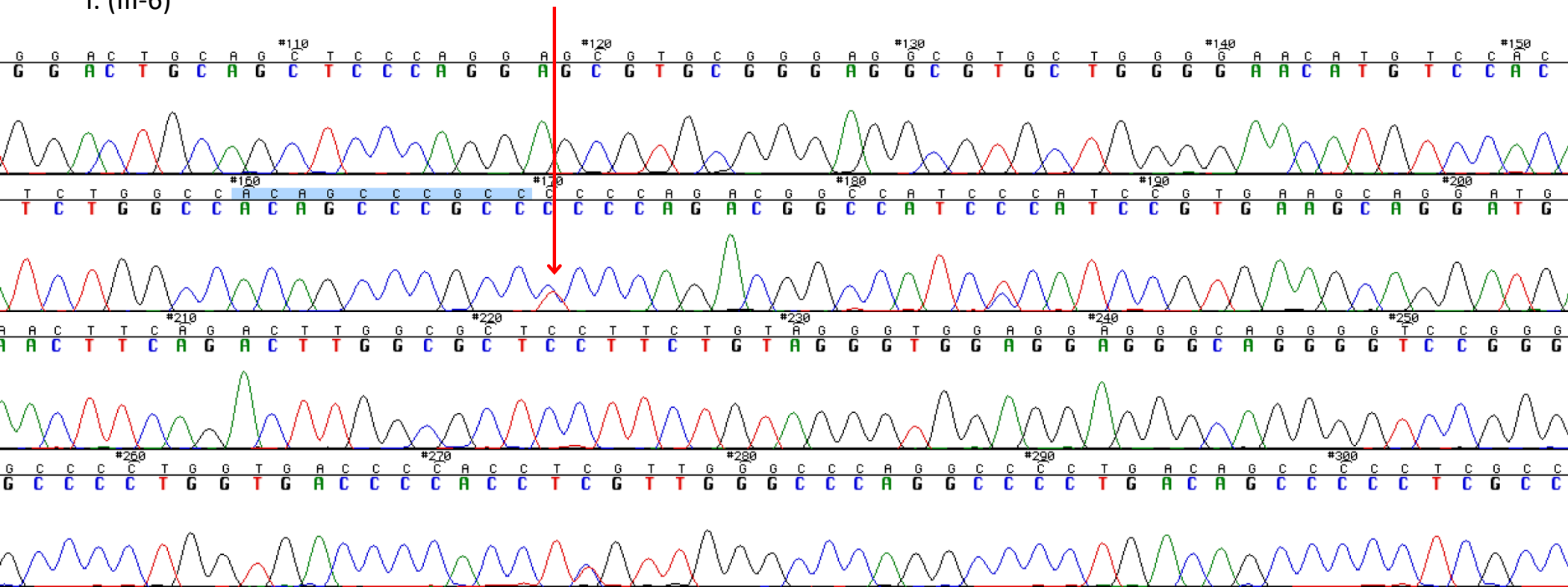


* Mother has TT alleles for rs28434439 as shown in Table 2

SLC34A3 mutation wt allele - no
duplication C

I. (III-6)

rs28434439 heterozygous T/C*



* This provides evidence that this subject carries father's WT allele