

Figure 1 - Electropherogram of the genetic region surrounding variant c.1334C>T p.(Ala445Val) identified in a NTG patient.

Heterozygous transition of a C to a T at nucleotide 1334, changing the codon GCA to GTA and causing an Alanine to Valine amino acid substitution at position 445. The nomenclature used in Figure 1 for sequence variation is according to reference⁴⁵.