

Variant Confirmation Report

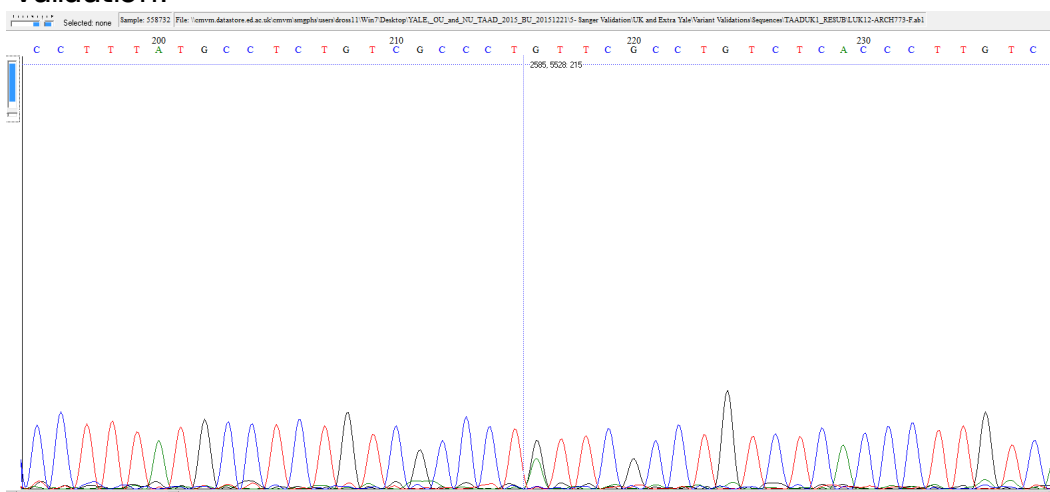
Sample ID:

ARCH773

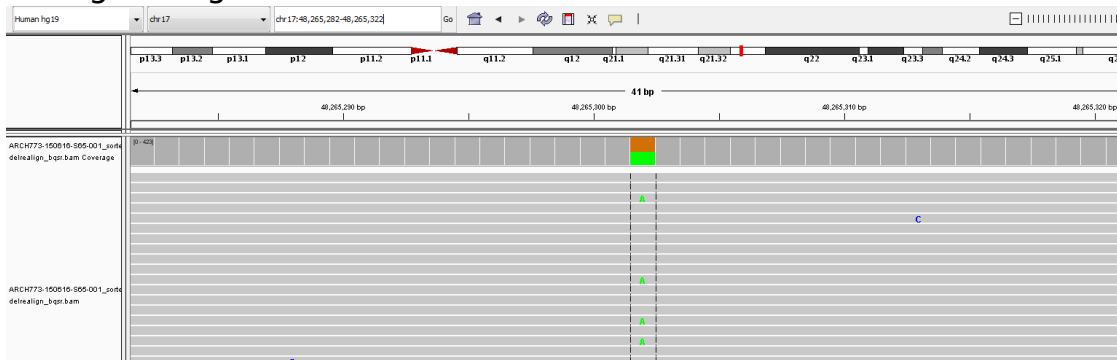
Description:

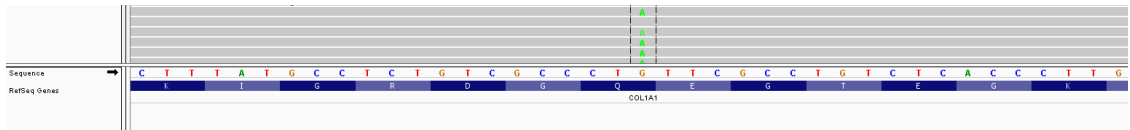
Gene	COL1A1
Exon	45/51
Sequence Variant	ENST00000225964.5:c.3304C>T ENSP00000225964.5:p.Gln1102Ter
Variant Location (GRCh37)	17:48265302
Allele Balance	0.42
Allele Depth (REF,ALT)	144,106
Allele Frequency (ESP,ExAC,dbSNP)	- - -
Variant Found	Y
Comment	This mutation introduces a prematurestop codon and is likely to be pathogenic

Validation:



Fluidigm Image:





The following information is for research purpose only. Any decisions made on the information sh
by an appropriate responsible clinician who may require further confirmation within a clinical labo



