Variant Confirmation Report

Sample ID:	1290

Description:

Gene	COL5A1
Exon	6/66
Sequence Variant	ENST00000371817.3:c.849delC ENSP00000360882.3:p.Glu284LysfsTer3
Variant Location (GRCh37)	9:137620574
Allele Balance	0.59
Allele Depth (REF,ALT)	101,147
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