Variant Confirmation Report

Sample ID:	22SA765

Description:

Gene	COL3A1
Exon	-
Sequence Variant	ENST00000317840.5:c.2022+2T>C -
Variant Location (GRCh37)	2:189863446
Allele Balance	0.41
Allele Depth (REF,ALT)	148,102
Allele Frequency (ESP,ExAC,dbSNP)	- - -
Variant Found	Y
Comment	This mutation has been asserted as a disease-causing muatation in the HGMD database HGMD Accession: CS1412566 HGMD Classification: DM Pepin (2014) Genet Med 16: 881 PubMed: 24922459

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