Variant Confirmation Report

Sample ID:	BW_42_25

Description:

Gene	COL5A2
Exon	53/54
Sequence Variant	ENST00000374866.3:c.4295A>T ENSP00000364000.3:p.Asp1432Val
Variant Location (GRCh37)	chr2:189899700
Allele Balance	0.51
Allele Depth (REF,ALT)	403,412
Allele Frequency (ESP,ExAC,dbSNP)	0.03844 0.05840 -
Variant Found	Y
Comment	This mutation has been asserted as a disease-causing muatation in the HGMD database HGMD Accession: CM021075 HGMD Classification: DM Grond-Ginsbach (2002) Neurology 58: 1103 PubMed: 11940702

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