Variant Confirmation Report

Sample ID:	22GK1188

Description:

Gene	TGFB2
Exon	6/8
Sequence Variant	ENST00000366929.4:c.979C>T ENSP00000355896.4:p.Arg327Trp
Variant Location (GRCh37)	1:218609452
Allele Balance	0.63
Allele Depth (REF,ALT)	76,129
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: CM126585 HGMD Classification: DM . Lindsay (2012) Nat Genet 44: 922 PubMed: 22772368

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