

## 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,dbS	- - -
Variant Found	Y
Comment	<p>This mutation has been asserted as a disease-causing mutation in the HGMD database</p> <p>HGMD Accession: CM126585 HGMD Classification: DM . Lindsay (2012) Nat Genet 44: 922 PubMed: 22772368</p>

Validation:

Fluidigm Image:

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