Variant Confirmation Report

Sample ID: JD_26_51

Description:

Gene	FBN1
Exon	58/66
Sequence Variant	ENST00000316623.5:c.7039_7040delAT ENSP00000325527.5:p.Met2347ValfsTer19
Variant Location (GRCh37)	15:48719927
Allele Balance	0.37
Allele Depth (REF,ALT)	106,61
Allele Frequency (ESP,ExAC,dbS	- - -
Variant Found	Y
Comment	This mutation has been asserted as a disease-causing muatation in the HGMD database HGMD Accession: CD020234 HGMD Classification: DM Korkko (2002) J Med Genet 39: 34 PubMed: 11826022

Validation:

Fluidigm Image:

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