Variant Confirmation Report

Sample ID:	EO_21_18

Description:

Gene	FBN1
Exon	38/66
Sequence Variant	ENST00000316623.5:c.4640C>T ENSP00000325527.5:p.Thr1547Ile
Variant Location (GRCh37)	15:48760242
Allele Balance	0.73
Allele Depth (REF,ALT)	25,66
Allele Frequency (ESP,ExAC,dbSNP)	- 0.00331 0.00040
Variant Found	Y
Comment	This mutation has been asserted as a likely disease-causing muatation in the HGMD database HGMD Accession: CM145768 HGMD Classification: DM? Lerner-Ellis (2014) Mol Genet Metab 112: 171 PubMed: 24793577 Date of Variant Class Change From DM to DM?: 2014-06-09 00:00:00 Lerner-Ellis (2014) Mol Genet Metab 112: 171 PubMed: 24793577

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