Variant Validation Report Key

Gene	FBN1
Exon	55/66
Sequence Variant	ENST00000316623.5:c.6700G>A ENSP00000325527.5:p.Val2234Met
Variant Location (GRCh37)	chr15:48725102
Allele Balance	0.45
Allele Depth (REF,ALT)	576,471
Allele Frequency (ESP,ExAC,dbSNP)	0.06160% 0.07162% 0.00020%
Variant Confirmed by Sanger	Y
Comment	This mutation has been asserted as a likely disease-causing mutation in the HGMD database HGMD Accession: CM067394 HGMD Classification: DM? Tjeldhorn (2006) Genet Test 10: 258 PubMed: 17253931 Date of Variant Class Change From DM to DM?: 01/06/2015 Groth (2015) Genet Med : PubMed: 25812041

The Exon numbering has been extracted from the ensembl transcript ID webpage

Ensembl transcript and protein identifiers with HGVS nomenclature for the description of the sequence variant

Alternative (ALT) allele depth / Reference (REF) allele depth = allele depth ratio

Tab delimited figures representing the allele frequencies of the sequence variant in the Exome Sequencing Project, Exome Aggregation Consortium and dbSNP databases respectively. A dash means no frequency information was provided in the respective database.

The variant found in the analysis of the MiSeq-Fluidigm assay, was confirmed by sanger sequencing

The likely biological/clinical outcome

HGMD Only – HGMD accession number, classification and primary literature report associated with the sequence variant.

DM; Denotes a mutation reported to be disease-causing

DM?; Denotes a mutation reported as likely disease-causing

HGMD Only – this only appears along with variants classified as DM?. It provides a description of the date in which the variant was changed from disease-causing to likely disease-causing along with the publication (if any) which is associated with this change in classification.

If the variant was found in ClinVar, the comment section is represented as follows:

Comment	This mutation has been asserted as likely pathogenic in the ClinVar database dbSNP ID: rs397515808 ClinVar Classification: Likely Pathogenic Clin Genet. 2013 Nov;84(5):453-63. doi: 10.1111/cge.12257.

The likely biological/clinical outcome

ClinVar Only – dbSNP ID, ClinVar classification and primary literary report associated with the sequence variant.