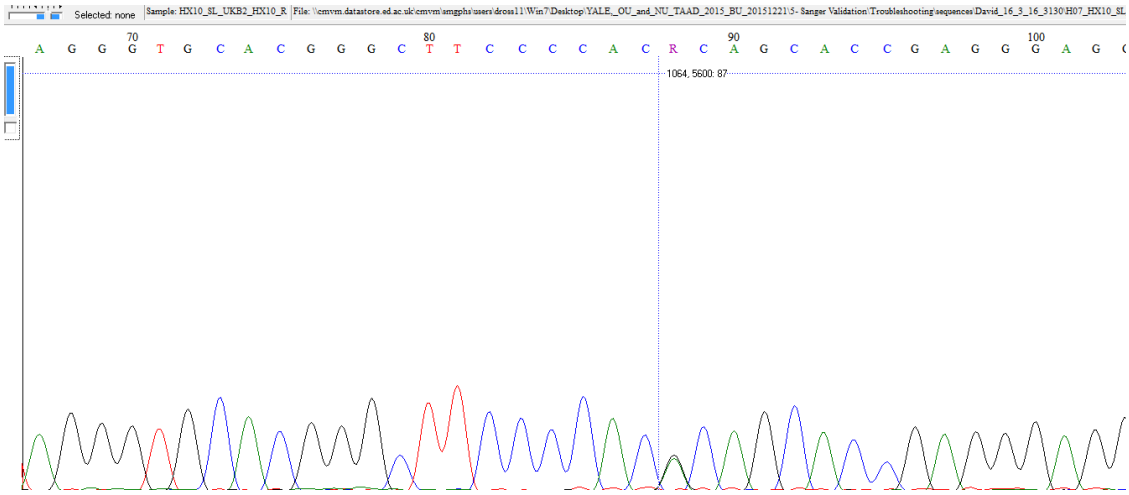
Variant Confirmation Report

Sample ID:	21SL0085

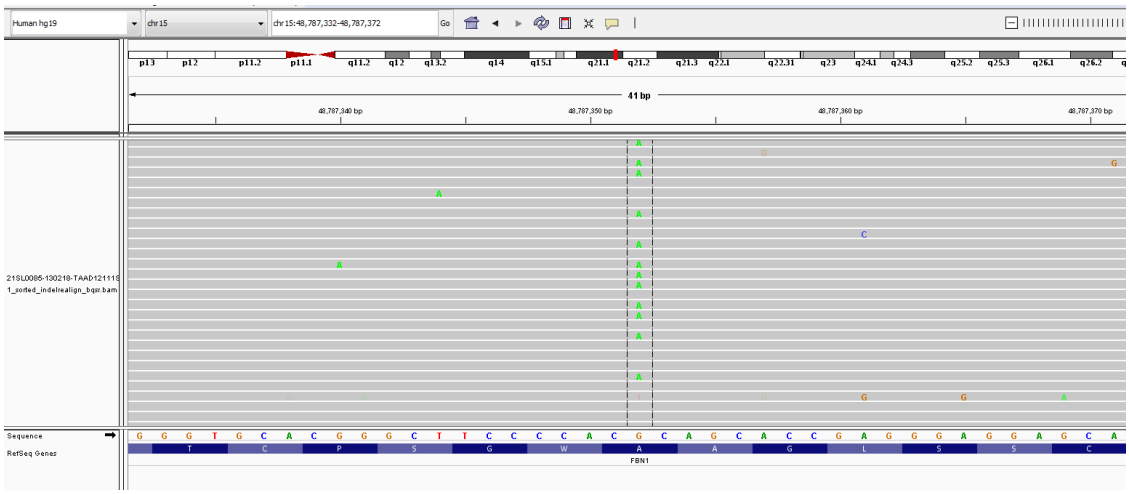
Description:

Gene	FBN1
Exon	22/66
Sequence Variant	ENST00000316623.5:c.2645C>T ENSP00000325527.5:p.Ala882Val
Variant Location (GRCh37)	15:48787352
Allele Balance	0.45
Allele Depth (REF,ALT)	138,111
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: CM042039 HGMD Classification: DM Loeys (2004) Hum Mutat 24: 140 PubMed: 15241795

Validation:



Fluidigm Image:



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