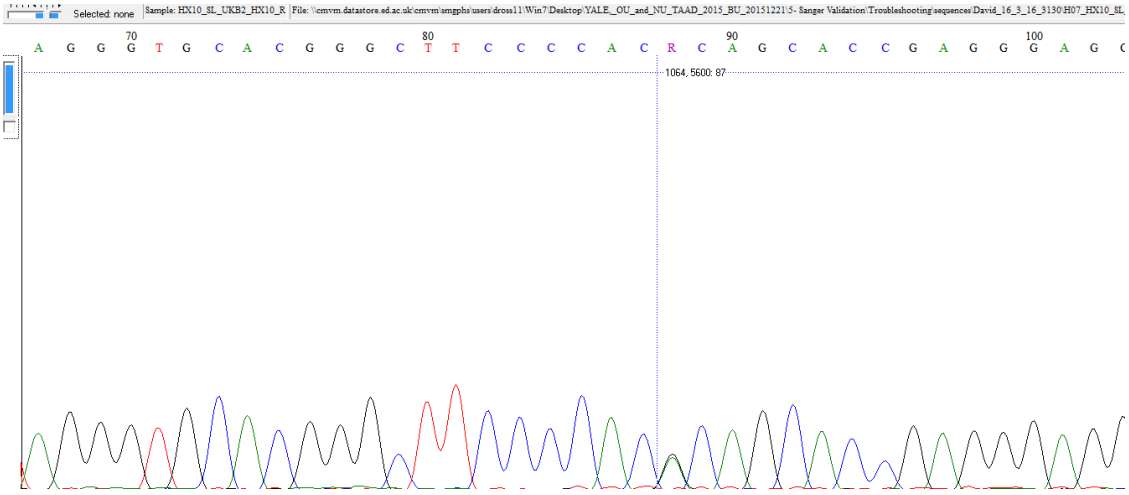
Variant Confirmation Report

Sample ID:	21SL0085

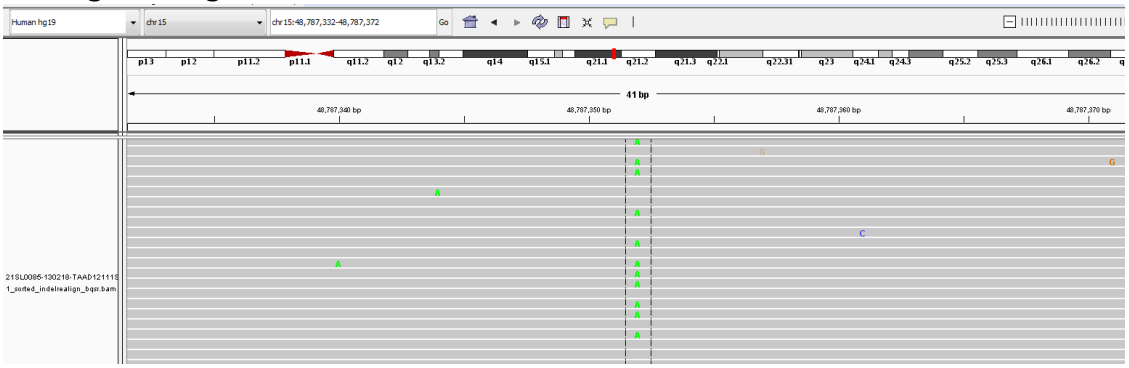
Description:

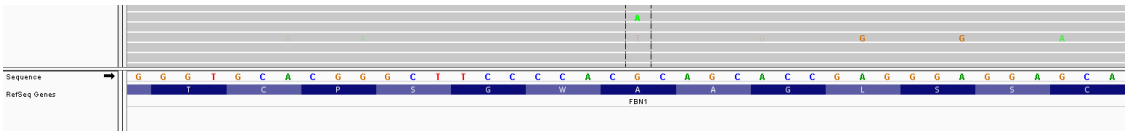
Gene	FBN1
Exon	22/66
Sequence Variant	ENST00000316623.5:c.2645C>T ENSP00000325527.5:p.Ala882Val
Variant Location (GRCh37)	15:48787352
Allele Balance	0.45
Allele Depth (REF,ALT)	138,111
Allele Frequency (ESP,ExAC,dbSNP)	- - -
Variant Found	Y
Comment	This mutation has been asserted as a disease-causing muatation in the HGMD database HGMD Accession: CM042039 HGMD Classification: DM Loeys (2004) Hum Mutat 24: 140 PubMed: 15241795

Validation:



Fluidigm Image:





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