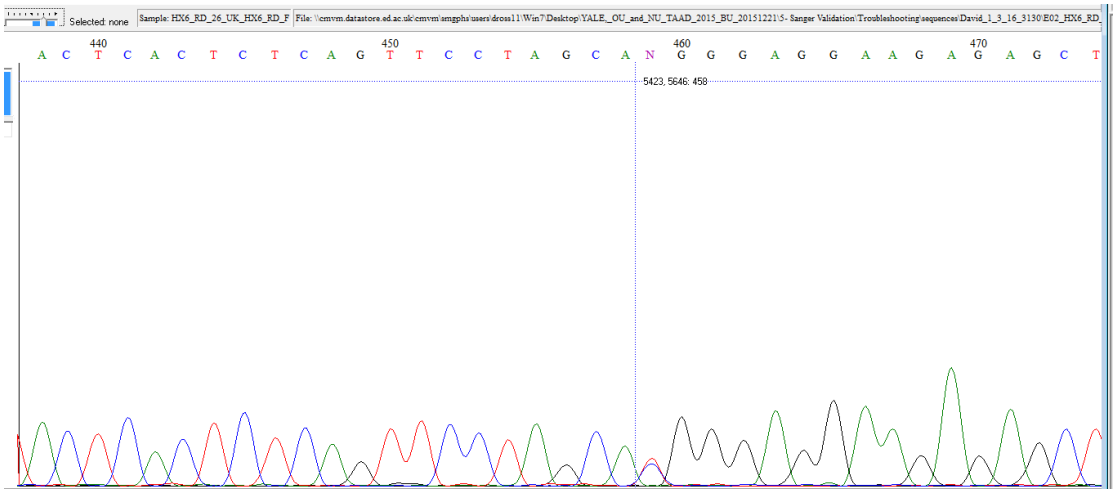
Variant Confirmation Report

Sample ID:	26JW1055

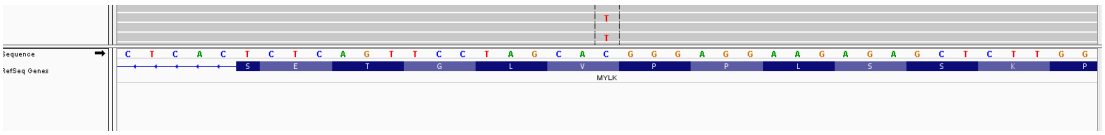
Description:

Gene	MYLK
Exon	20/34
Sequence Variant	ENST00000360304.3:c.3637G>A ENSP00000353452.3:p.Val1213Met
Variant Location (GRCh37)	3:123401086
Allele Balance	0.59
Allele Depth (REF,ALT)	103,146
Allele Frequency (ESP,ExAC,dbSNP)	0.00769 0.00003 -
Variant Found	Y
Comment	This mutation has been asserted as a likely disease-causing muatation in the HGMD database HGMD Accession: CM107678 HGMD Classification: DM? Wang (2010) Am J Hum Genet 87: 701 PubMed: 21055718 Date of Variant Class Change From DM to DM?: 2012-04-10 00:00:00

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