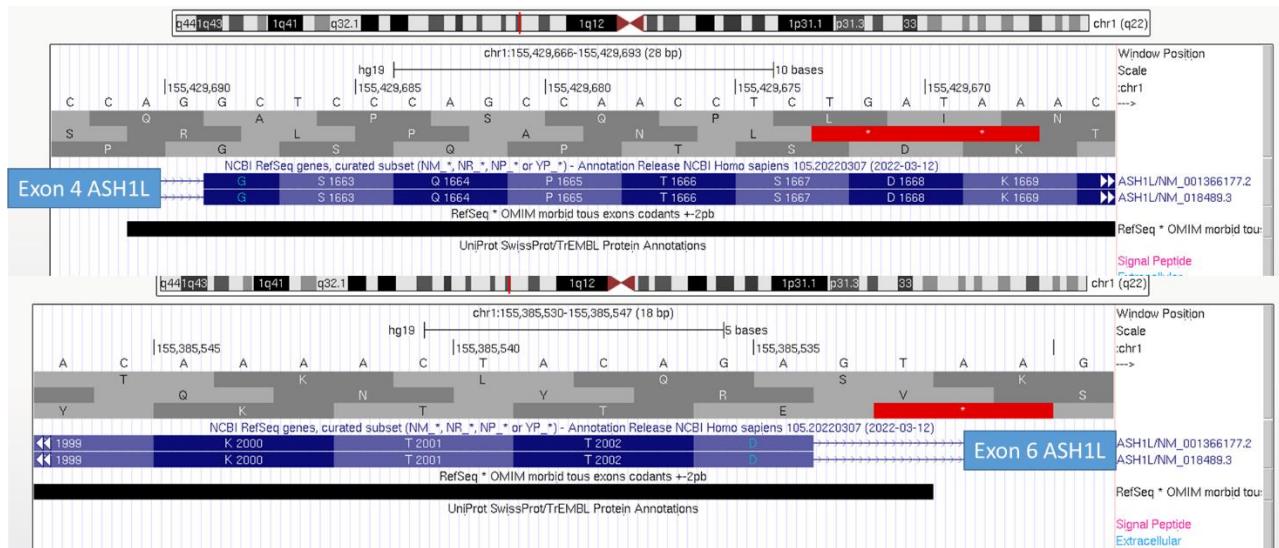


Supplementary material



Supplementary Figure 1.

Reverse UCSC genome browser view of exons 4 and 6 of ASH1L, with the nucleotide track in full to visualize the three genomic frames. The end of exon 6 is directly followed by exon 4: the last amino acid of exon 6 contains only 2 nucleotides (GA), which normally codes for Asp, but with the frameshift mutation (C>G), it results in p.Asp>Glu. With the track of the three possible frames on UCSC, the sequence continues on the second frame line, starting with a Leu, and has a stop codon at the 7th position.

Caractéristiques du variant	
Niveau Génomique	
Version du génome :	GRCh37
Chromosome :	Chr1 (q22)
gADN:	g.155365344G>C
Type:	Substitution
Niveau du Transcript	
cADN :	NM_018489.3(ASH1L):c.6009C>G
Position:	Exon 7
Exon naming: Systematic numbering (1..n)	
Niveau des protéines	
Coding Effect:	Missense
pNomen:	p.(Asp2003Glu)
Compare AA:	
Effet possible sur le site d'epissage le plus proche	
Voir les prédictions dans la fenêtre d'épissage	
Outils Externes	
VariantValidator	Mutalyzer

Supplementary Figure 2.

Simulation using Alamut software by creating a variant on the first codon of exon 7, c.6009C>G, to identify the first changed amino acid.