

Supplementary File S1. In silico predictions

GATA1 gene (Xq11.23):

NM_002049.4

Patient 1.

GATA1 c.220+2T>C; p.?

intron 2

Splicing site mutation

Position 48791331

gnomAD: 0

Class 5 pathogenic according to ACMG-AMP (Richards et al., Genet Med 2015 ; Amendola et al., Am J Hum Genet 2016).

No publications about this variation; no metascoring; no clinVar description

CADD= 26.4

PHRED=26.4

Rawscore: 3.91

Varsome data:

Individual predictions: bayesDel addAF=damaging; bayesDel noAF= damaging

FATHMM-MKL= damaging

Mutation taster disease causing

Patient 2.

GATA1 c.2T>C; p.(Met1Thr)

exon 2

start loss allelic variation

position 48791111

rs587776451

gnomAD=0

Class 5 pathogenic according to ACMG-AMP (Richards et al., Genet Med 2015 ; Amendola et al., Am J Hum Genet 2016).

4 publications: Bluteau et al, Blood; Ludwig LS et al., Nature med 2014; Parrella et al., S J Pediatr Blood and Cancer; Kanezaki et al., Blood

Clinvar: pathogenic RCV001382885, RCV000144252, RCV000754842

CADD=21.9

PHRED=21.9

Rawscore=2.35

Metascore: 4/4:

METALR=0.8322 damaging;

METASVM=0.5822 Damaging;

METARNN=0.8873 Damaging;

Revel=0.56 pathogenic

Individual predictions = 11/12 damaging

Bayesdel addAF score 0.7434 damaging;

Bayesdel noAF score 0.8299 damaging;

mutation taster disease causing;

sift/sift4G score 0 damaging

Electropherogram of **GATA1 c.220+2T>C**; p.?

