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<b>Title</b>	A novel 33-Gene targeted resequencing panel provides accurate, clinical-grade diagnosis and improves patient management for rare inherited anaemias
<b>Author(s)</b>	Roy, Noémi B. A.; Wilson, Edward A.; Henderson, Shirley; Wray, Katherine; Babbs, Christian; Okoli, Steven; Atoyebi, Wale; Mixon, Avery; Cahill, Mary R.; Carey, Peter; Cullis, Jonathan; Curtin, Julie; Dreau, Helene; Ferguson, David J. P.; Gibson, Brenda; Hall, Georgina; Mason, Joanne; Morgan, Mary; Proven, Melanie; Qureshi, Amrana; Sanchez Garcia, Joaquin; Sirachainan, Nongnuch; Teo, Juliana; Tedgård, Ulf; Higgs, Doug; Roberts, David; Roberts, Irene; Schuh, Anna
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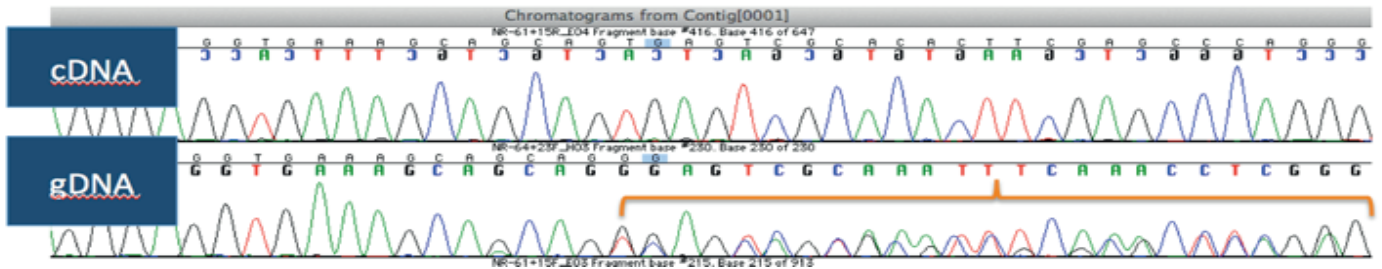


### Supplementary Fig 3. Sanger sequencing analysis of novel frameshift mutation in *CDAN1*.

(A) Sanger sequencing of complementary DNA (cDNA) and genomic DNA (gDNA) of cultured patient lymphocytes for patient 50 for exon 23 of *CDAN1*. Heterozygosity for a 7bp insertion in gDNA of *CDAN1* causing a frameshift (orange brackets) while cDNA, which shows only the expressed allele, is wild type in this region.

(B) Diagram illustrating nonsense mediated decay of the frameshifted allele and the resultant functional homozygosity.

A



B

