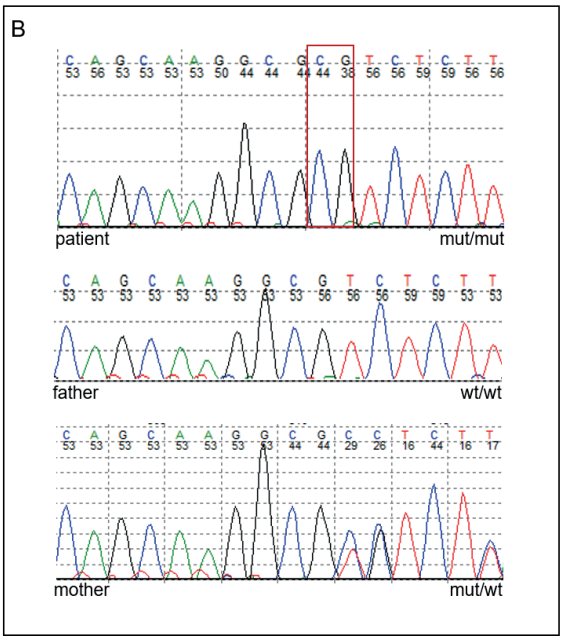
A

| Chrom | Position | Ref | Alt | Type | Consequence | ESP EUR | TG EUR | Gene Name | PolyPhen | SIFTcat | PHRED |
|---|---|---|---|---|---|---|---|---|---|---|---|
| 2 | 173356011 | G | GGC | INS | Frame shift | NA | NA | ITGA6 | NA | NA | 35 |



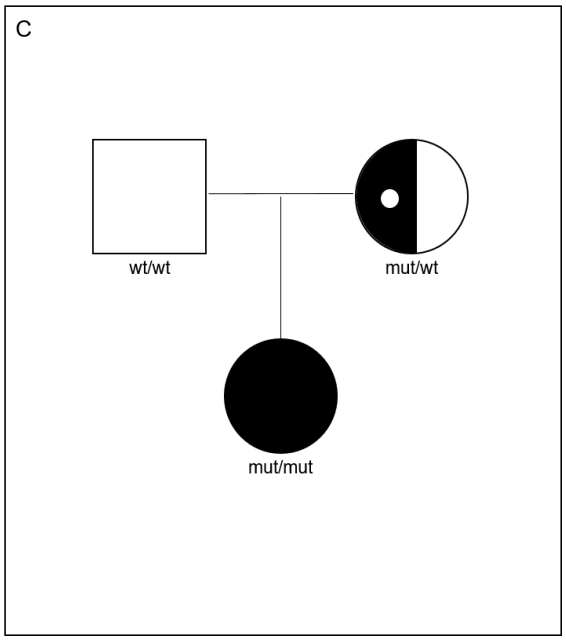




**Fig. S3. Genetic analysis.** (A) Results from whole exome sequencing identifying the ITGA6 mutation in exon 22. (B) Confirmation by Sanger sequencing and identification of a stop codon mutation c.2843_2844dupCGp.Ser949Argfs*15. The patient was homozygous for T to C mutation. The mother was heterozygous, and the father was not a carrier. (C) The ITAG6 pedigree, with *squares* representing male members, *circles* female members, shading members with autosomal recessive JEB, and the *dot* indicating the silent mutation carrier.