

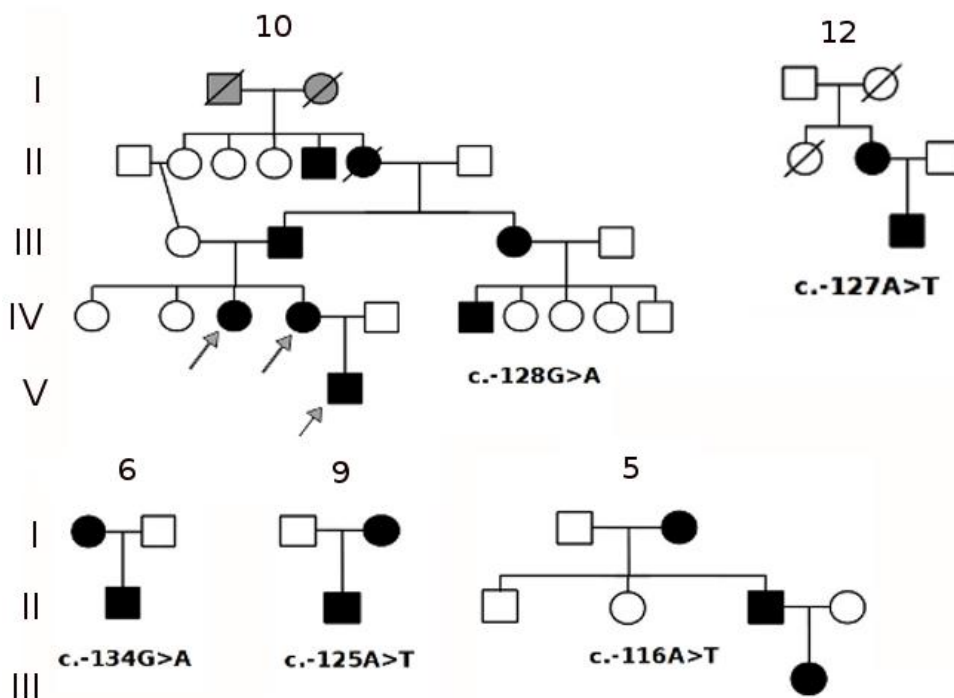
## Supplemental Data

### Mutations in the 5' UTR of *ANKRD26*, the Ankirin

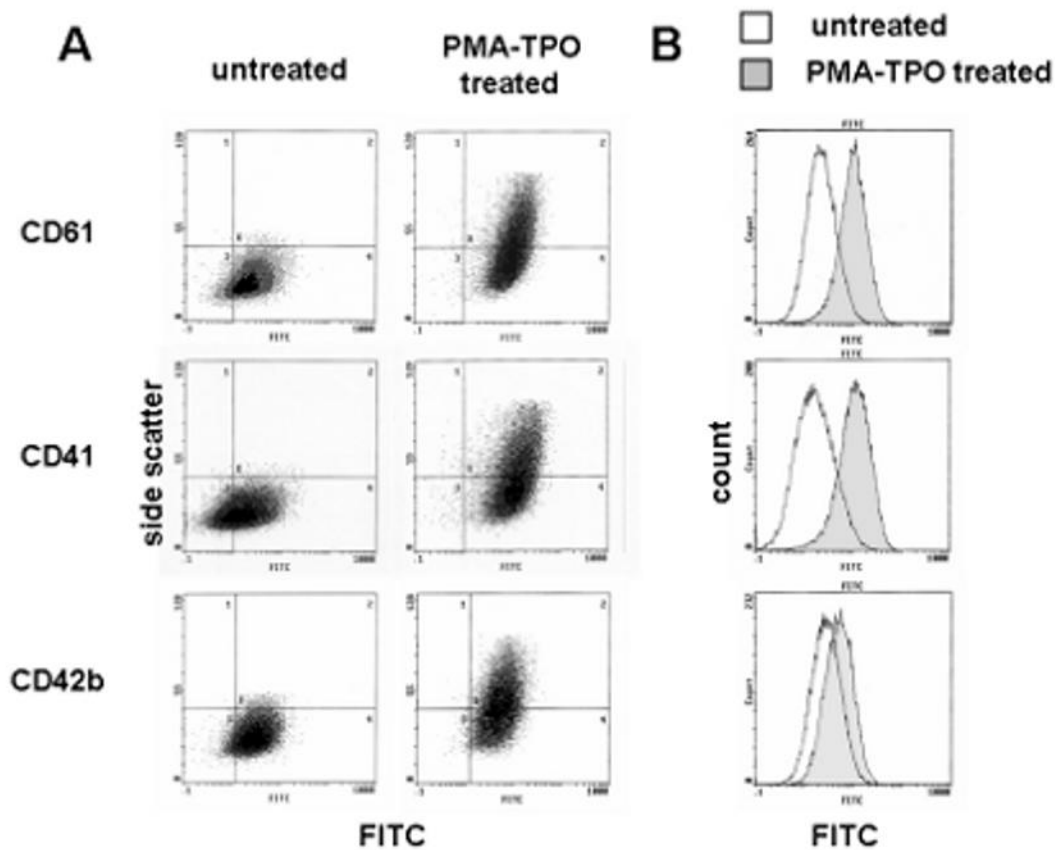
### Repeat Domain 26 Gene, Cause an Autosomal-Dominant

### Form of Inherited Thrombocytopenia, THC2

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**Figure S1. Five additional THC2 families with mutations in the 5'UTR of *ANKRD26*.** Number of the family and the corresponding mutation are reported above and under each pedigree, respectively. All affected individuals (black symbols) were carriers of the mutations. In family 10 only individuals indicated by arrows were analysed. Healthy relatives (white symbols) were all non carriers. Slashed symbols stand for deceased individuals.



**Figure S2. Flow cytometry assessment of megakaryocytic maturation of Dami cells.** Dami cells at  $1 \times 10^6$ /mL were cultured in modified RPMI 1640 medium (Lonza Ltd, Basel, Switzerland) for 48 hours with or without the addition of 100 nM PMA (Sigma-Aldrich, Milan, Italy) and 10 ng/mL TPO (PeproTech EC Ltd, London, UK). Maturation toward the megakaryocytic lineage of PMA/TPO-treated cells was then routinely assessed by flow cytometry. Dami cells treated with PMA and TPO showed an increase in surface expression of the specific markers associated with megakaryocytic differentiation and maturation CD41, CD61, and CD42b with respect to untreated cells (A and B, FITC). Megakaryocytic maturation was also typically associated with an increase in cell size, as shown by the increased light side scatter of treated cells compared to untreated (A).

Marker	Forward Primer	Reverse Primer	Position on Chr10
D10S586	5'-GAACAGCAGGATGTCTCG-3'	5'-TGAGACTTCATCACATGCGC-3'	24736107...24736260
D10S572	5'-TGGGTAGGAGGCCTTAG-3'	5'-TTGTAGAATGCCAGGTAGGG-3'	25677181...25677532
D10S1775	5'-CCTAGTCCCAGAGTCTTG-3'	5'-CACAGGTATTGCTGAGTGAG-3'	26433775...26433947
D10S111	5'-AATGTGGTAGCCCATCTC-3'	5'-ACATAGGTAGGGTCACACAG-3'	26810739...26810995
D10S593	5'-CAGGACCAACAACCAGAATA-3'	5'-CACATACCTGTTTGCCATTC-3'	27163370...27163563
CArepeat 1	5'-GTAAAGTACTACGCCAC-3'	5'-AATCATCTACTGTTGTAGCC-3'	27985712...27985881
CArepeat 2	5'-CAGAGAGTCAACAAGTAG-3'	5'-GATGATTGTTTTGTCTGCCA-3'	28391309...28391592
D10S174	5'-TTCTGTGAGCATCTGTACAG-3'	5'-TATGAGCCATCCCTAGCCAG-3'	28610794...28610943

**Table S1.** THC2 locus microsatellite markers, forward and reverse primers and physical position relative to the UCSC hg19 assembly.