Dominant inheritance of a novel integrin β_3 mutation associated with a hereditary macrothrombocytopenia and platelet dysfunction in two Italian families

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Citation: Gresele P, Falcinelli E, Giannini S, D'Adamo P, D'Eustacchio A, Corazzi T, Mezzasoma AM, Di Bari F, Guglielmini G, Cecchetti L, Noris P, Balduini CL, and Savoia A. Dominant inheritance of a novel integrin β_3 mutation associated with a hereditary macrothrombocytopenia and platelet dysfunction in two Italian families. Haematologica 2009; doi:10.3324/haematol.2008.002246

Online Supplementary Table S1. Flow cytometry of platelet membrane glycoproteins (GP) from the proband, controls and one patient with typical congenital Glanzmann's thrombasthenia (GT).

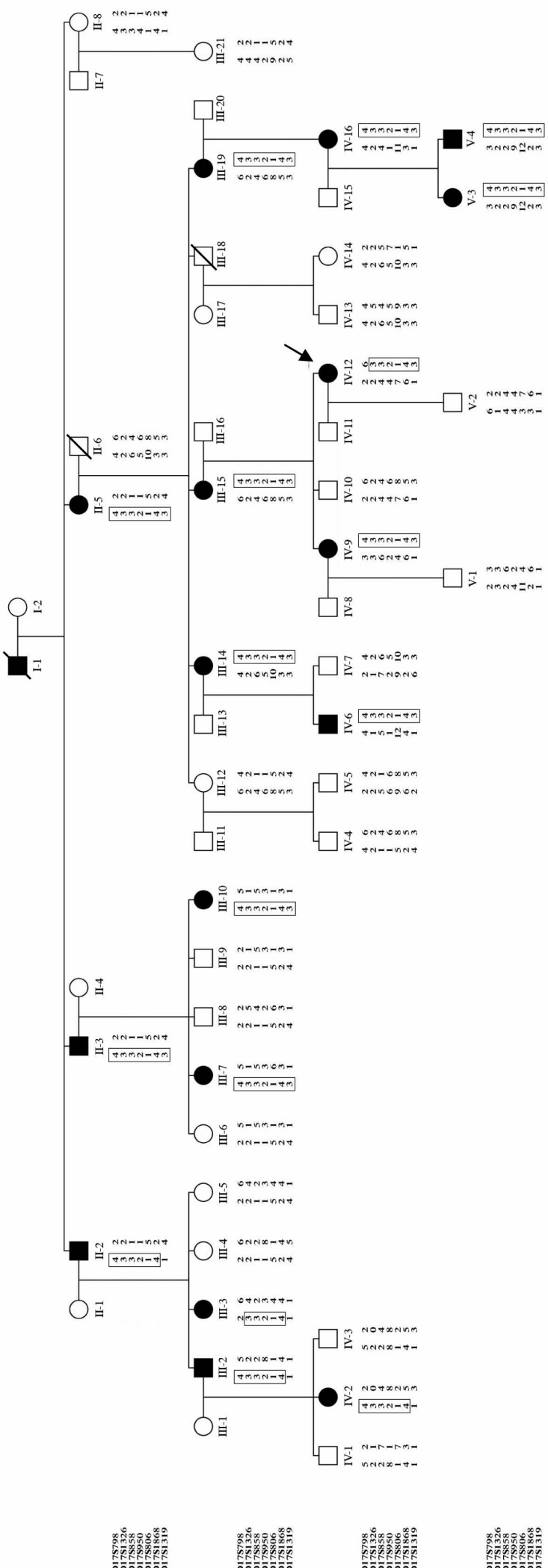
A. Mean values expressed as absolute values or as % of controls.

		Mean fluorescence intensity			% of controls	
Molecule	Antibody	Controls	Proband	GT	Proband	GT
GPIbα	SZ2	33.3±5.5	49.9	32.6	149	97
GPIX	SZ1	10.0±1.6	17.8	12.9	178	129
GPIV	FA6-152	19.1±6.1	18.9	13.2	98	69
GPIIa	5.6E	9.3±1.4	12	7.4	129	79
CD9	ALB6	50.2±11	48.9	51.8	97	103
CD51	AMF7	1.8±0.05	2	1.8	111	100
GPIIb	SZ22	4.5±1.06	3	0.6	66	13.6
	P2	60.2±6.6	30.5	1.1	50	3.3
GPIIIa	SZ21	28.3±10.3	13.9	1	49	8.12
	SAP	15.0±1.9	6.3	0.6	42	4
	AP-3	6.6±1.7	2.6	n.a.	39	n.a.
GPIIb/IIIa	A2A9/6	26.1±3.3	14.2	0.5	54	5.7
	AP-2	15.8±3.8	5.1	n.a.	32	n.a.

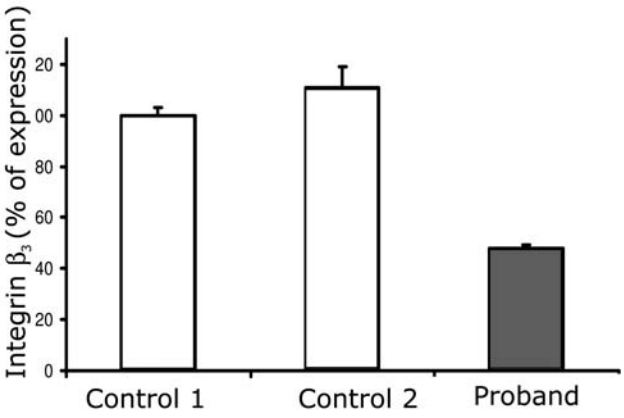
B. Mean values for GPIIb/IIIa normalized to those of GPIbα (Ratio) because of the larger volume of platelets.

		RATIO			% of controls	
Molecule	Antibody	Controls	Proband	GT	Proband	GT
GPIIb	SZ22	0.15±0.02	0.06	0.02	41	13
	P2	1.50±0.27	0.61	0.03	40	2
GPIIIa	SZ21	0.91±0.25	0.27	0.03	30	4
	SAP	0.31±0.20	0.12	0.02	40	6
	AP-3	0.13±0.09	0.05	n.a.	40	n.a.
GPIIb/IIIa	A2A9/6	0.73±0.04	0.28	0.01	38	1.4
	AP-2	0.41±0.04	0.10	n.a.	25	n.a.

n.a.: not available.



Online Supplementary Figure S1. Family pedigree with haplotype reconstruction for informative markers on chromosome 17 between markers D17S798 and D17S1319. The at-risk haplotype is boxed.



Online Supplementary Figure S2. Real-time PCR of purified platelet mRNA for β_3 normalized to B2MG of two controls and the proband, using primers laying upon the deletion and capable of amplifying the wild type allele only. Control 1 was used as a calibrator. RT-PCR confirming the 120pb deletion was carried out using the following primers: Ex 12 forward: CGTTACTGCCGTGACGAGATTGAG; Ex 14-15 reverse: CAGTGGGTTGTTGGCTGTGTCC. A 342 bp corresponding to the wild type band was found both in the controls and the proband (*data not shown*). In addition a 222 bp band, corresponding to the mutated allele, was evident in the proband only (*data not shown*).