

A *de novo* splice donor mutation in the thrombopoietin gene causes hereditary thrombocythemia in a Polish family

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Supplementary Table S1. Sequencing primers for the *THPO* gene.

Primer	Forward	Primer	Reverse
370	5'-GGCTAGGGAGGGGCAGAG-3'	368	5'-GAGGCAGGCTAGGCAGTGA-3'
1313	5'-AAGGTCCATCAGCCGATCTC-3'	1314	5'-TCTCTCCTGTTCTCCTCCC-3'
1315	5'-GCTGGACGGGAAAATTCCAG-3'	1316	5'-TTTGGACGGGTTGAGTGGAC-3'
1365	5'-AGCCATCCAAGCCACTGGAC-3'	1366	5'-CTGGACCAGGGTCCACTCTC-3'
385	5'-CAAATCTTCCCGTGGTGTG-3'	545	5'-CTCATGTCCCCTTTCCTTAG-3'
1367	5'-TTGGTGAATACAGCACAGC-3'	1368	5'-GATGGGCTAACAGTTCTGGG-3'
1371	5'-CTCAACATATTGCGCCCGTT-3'	1372	5'-ATCGCTTGTACCTGGGAGGC-3'
1373	5'-ATGGTGCGATCTCGGCTCAC-3'	1374	5'-GGGTGAACAGATGCTGGGGA-3'
497	5'-TTCAGGGCTTTGGCAGTTCC-3'	408	5'-GTCTGCTGTGAAGGACATGG-3'
371	5'-TGCAAGGCTAACGTGTGC-3'	425	5'-AGCAGGCAGCAGGACAGGTG-3'
365	5'-TGACCAGCTGTGCGGAGAC-3'	386	5'-CCAGCAGAAGGGTCACTGC-3'
433	5'-ATTCACTGTGCTGGCTACTC-3'	186	5'-CCACGAGTTCATTCAAGAG-3'
227	5'-GGTGCGTTTCTGATGCTTG-3'	364	5'-TGAGACAATGCTGATGTCGG-3'
465	5'-TGAACAGGATACACGAACTC-3'	452	5'-GAGTCCCCTGAGAATGATA-3'

Supplementary Table S2. Microsatellite markers in the *THPO* locus.

Markers	Sequence	Size
D3S3578-F	FAM-GATTGGGTTACAGAGTAGCCACAC	200-242bp
D3S3578-R	GTTTCTTCTGAAATGTAGACTGCCCTTTG	
TPO1-F	HEX-AAAATAGCTGGACAGGAAAAC	150-160bp
TPO1-R	GCAGCATTAAAGTGATTAGACTG	
TPO2-F	GTTTCTTATGAATGGGCTGGATCTGG	139bp
TPO2-R	FAM-CTAACTGGTTGGATTGCTGG	
TPO4-F	FAM-CAGCCTTTTACAGAGTAGCAA	190bp
TPO4-R	GTTTCTTGTAGACTGCCCTTTGTGTCAG	
TPO5-F	FAM-GACGTGGCCTCCCCTTTCAA	91BP
TPO5-R	GTTTCTTCCCCTGTGTTCCCCAGTCAAC	
TPO6-F	FAM-AGAGATCACACCACTGAACT	244BP
TPO6-R	GTTTCTTGACAAGGAACTTAGTCACGT	

Supplementary Table S3. Primers used to detect single nucleotide polymorphisms in the *THPO* locus.

	ID	Alleles	Frequency	Heterozygosity	Sequence	Size
1	rs3792299	A	0.2	0.32	F: CTGTTAGCCCATCTCTTGGC	171 bp
		G	0.8		R: TGCACAAAGCTGTAAGTGGG	
2	rs956732	A	0.437	0.492	F: TCTGGGCAGAGTAGGGTGGG	112 bp
		G	0.563		R: CCTGCCAGGCAGTCTCTTCC	
3	rs2280740	A	0.127	0.222	F: TACTCCCAGGAAGACACCAT	172 bp
		G	0.873		R: TAAGCAGGCTAGTCTCCAT	
4	rs16858759	C	0.029	0.057	F: AACGGGAATAGGTCAAAGGC	152 bp
		T	0.971		R: GGCCACCCTAACCCAATCTA	
5	rs10513797	A	0.155	0.215	F: TCTGTACGAGCATTTCACACA	265 bp
		G	0.845		R: GACAAGAGCAAACTCCGTC	
6	rs9870912	C	Unknown	N.D.	F: GGGTGGGGAGCCTTAGGAGT	126 bp
		T	Unknown		R: AGCAGAGGCTGATGACCAGC	
7	rs2700322	A	Unknown	N.D.	F: GAAGCAGAGGCTGATGACCA	166 bp
		T	Unknown		R: AGAGAGAAGCGCACTGCCTC	
8	rs1126665	A	Unknown	N.D.	F: ACAACTGGGACCCACTTGCC	96 bp
		G	Unknown		R: TGGGTTCCAAGGAGGCTCTG	
9	rs6142	A	Unknown	N.D.	F: CCTCCTTGGAACCCAGGTAA	169 bp
		C	Unknown		R: AGTACCCAGGCATTGTGGCT	
10	rs6141	A	0.632	0.487	F: TGAACAGGATACACGAACTC	762 bp
		G	0.368		R: GAGTCCCCTGAGAATGATA	

Supplementary Table S4. Summary of clinical data of 12 affected members of the Dutch family with thrombocytosis.

UPN	Sex	Date of Birth	Date of Dx	Date of last follow up or DOD	Platelets 150-450 x10 ⁹ /L (lowest and highest observed values)	WBC 3.5-10.0 x10 ⁹ /L (at last follow up)	RBC 4.2-6.3 x10 ¹² /L (at last follow up)	Hemo-globin 120-140 F 140-180 M g/L (at last follow up)	spleno-megaly (at last follow up)	hepato-megaly (at last follow up)	thrombocytosis-associated symptoms	important co-morbidity
I-2	F	1908	2-1974	5-1986	650-875	na	na	na	na	na	none	none
II-2	F	1932	7-1991	4-2003	772-1270	5.3	3.4	114	-	-	died 04-2003 (myelofibrosis)	none
II-3	M	1934	4-1986	1/2002 died 3-2006 (stroke)	701-1200	7.0	4.9	158	+	-	Erythromelalgia, gangrene of a toe 1968, transient paresis/left carotid stenosis, secondary fibrosis, stroke in 2006	hypertension
II-7	F	1940	7-1991	1/2002	880-1120	10.6	4.0	130	-	-	Paresthesia, acrocynosis, cold tip feeling	none
II-8	M	1948	7-1991	Lost to follow up	578	7.3	5.4	152	-	-	Transient paresis and sensibility loss of arms	none
III-4	F	1967	12-1989	1/2002	907-1280	8.6	4.7	124	-	-	Ecchymosis menorrhagia	none
III-5	M	1968	12-1989	8-2003	625-1240	9.5	5.4	154	-	-	Cold tip feeling, paresthesia	none
III-6	F	1970	7-1991	1/2002	791-1010	8.8	4.8	129	-	-	Same above	none
III-7	F	1978	7-1991	1/2002	657-976	7.6	5.4	154	-	-	Cold tip feeling	none
III-14	F	1979	7-1991		763						none	none
IV-3	M	1990	2-1991	1/2002	1100-1700	8.0	4.4	114	+	-	none	Leukemoid reaction at birth
IV-7	F	1997	1/2002	1/2002	890	7.9	4.1	109	-	-	none	none

UPN, unique patient number; Dx, diagnosis; DOD, date of death; WBC, white blood cells; RBC, red blood cells; Reference values are given for platelets, WBC, RBC, and hemoglobin; F, female, M, male; lowest and highest platelet values are given where available; na, data not available