CAMT-MPL: congenital amegakaryocytic thrombocytopenia caused by *MPL* mutations - heterogeneity of a monogenic disorder - a comprehensive analysis of 56 patients

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Table S1: Detailed patient information

CAMT-ID	sex	consan-guineous parents	Intron/ Exon	cDNA	Zygosity	predicted protein	THPO plasma levels [pg/ml]	TP detected at birth? (plt count [G/l])	platelet rise? (max plt count)	Bleeding pet/epis/ICH other	First signs of multi-lineage cytopenia* [mo] or HSCT (if earlier) (if negative, age of last information is provided in brackets[mo])	Bone marrow findings (age [mo])	Cytogenetics	CAMT group	Therapy (other than HSCT) plt/RBC/Ig/Steroids other	HSCT / outcome	non-hematological findings	mentioned in earlier publications (Ref)
CAMT001	f	y	E2	c.127C>T	hom	p.Arg43Ter	NT	y (17)	n (20)	y/n/n	0	12: normocellular, MK↓↓ 18: hypocellular, MK↓↓	NAD	I	n/n/n/y	0.6 y / +	no	1,2,3
CAMT006	f	n	E3	c.268C>T	hom	p.Arg90Ter	1606	y (36)	n (38)	y/n/n	8	6: normocellular, MK↓	NAD	I	y/n/n/n	0.8 y / +	no	1,3
CAMT007	m	y	E3	c.305G>C	hom	p.Arg102Pro	1310 - 3290	y (14)	y (90)	y/n/y	50	16: normocellular, MK↓↓ 82: hypocellular, MK↓↓	NAD	II	y/n/n/n oprelvekin	7.0 y / 7.1 y death	no	1,2,3
CAMT009	f	y	E3	c.378delT	hom	p.Phe126LeufsTer5	1659 - 4250	y (21)	n (31)	y/y/y	6	2: normocellular, MK↓↓	t2;11 (1/19)	I	y/y/y/y fresh frozen plasma oprelvekin	4.2 y / ND	statomotoric retardation, speech delay, nystagmus, strabismus	1,2,3
CAMT011	f	n	E5	c.823C>A	het	p.Pro275Thr	1295-3226	n (46 at age 6 w)	y (100)	y/y/n gum bleeding	- (250)	normocellular, MK↓↓	NAD	II	n/n/y/y	_	high palate	1,2,3
			E3	c.305G>C	het	p.Arg102Pro												
CAMT012	m	n	E3	c.305G>C	hom	p.Arg102Pro	940 - 1708	y (16)	y (163)	y/n/n	48	66: hypocellular, MK↓↓	NAD	II	y/n/y/n	4.1 y / late graft failure, 6.4 y 2nd HSCT / +	hypertelorism, epicanthus	1,2,3
CAMT013	f	y	E3	c.235_236delCT	hom	p.Leu79GlufsTer84	2674	y (30)	n (40)	y/n/y	0	42: hypocellular, MK↓↓	47XX (additional marker chromosome in 94%)	I	y/y/y/y	3.8 y / +	hydrops fetalis, global retardation, visual impairment, hypertelorism, strabismus, nystagmus	1,2,3
CAMT015	f	n	E3	c.305G>C	hom	p.Arg102Pro	NT	y (14)	n (22)	y/n/n	0	1: normocellular, MK↓↓	ND	I*	y/n/y/n	1.9 y / 2.5 y death	no	2
CAMT017	f	y	E2	c.127C>T	hom	p.Arg43Ter	1092	y (25)	n (25)	y/n/n	9	9: normocellular, MK↓↓	ND	I	y/y/y/y	1.9 y / + (GvHD II)	statomotoric retardation, strabismus, nystagmus	1,2,3
CAMT018	f	y	E2	c.127C>T	hom	p.Arg43Ter	679 - 4412	y (18)	n (42)	y/n/y	53	11 : normocellular, MK↓↓ 53: hypocellular, MK↓↓ 88: SAA, MK↓↓	ND	I	y/y/y/y	7.4 y / +	ventriculomegaly, strabismus	2,3
CAMT019	f	y	E2	c.127C>T	hom	p.Arg43Ter	NT	n (49 at age 3 mo)	n (46)	y/n/n	43	6: normocellular, MK↓↓	ND	I	y/n/n/n	3.6 y / +	no	3
CAMT030	m	n	I1	c.79+2T>A	hom	splice defect (p.Asp27fs)	NT	y (11)	n (20)	ND	0	0: hypocellular, MK↓↓	ND	I	n/n/y/n	2.7 y / no engraftment, 2nd HSCT / +	no	3
CAMT031	f	y	E3	c.378delT	hom	p.Phe126LeufsTer5	NT	y (20)	n (ND)	y/n/n	48	ND	ND	I	y/y/n/n	4.7 y / + (GvHD)	no	3
CAMT033	f	n	E3	c.378delT	het	p.Phe126LeufsTer5	NT	y (ND)	n (49)	ND	ND / HSCT: 35	0: normocellular, MK↓↓	ND	I	ND	2.9 y / +	no	3
			I2	c.213-1G>A	het	splice defect (p.Arg71fs)												
CAMT034	f	y	E3	c.378delT	hom	p.Phe126LeufsTer5	2872	y (20)	n (21)	y/n/n	ND / HSCT: 25	0: normocellular, MK↓↓	ND	I	n/n/y/n	2.1 y / +	no	3
CAMT036	m	y	E2	c.127C>T	hom	p.Arg43Ter	NT	y (6)	n (28)	y/n/n	ND / HSCT: 30	0: nomocellular, MK↓↓ 3: normocellular, MK↓↓	ND	I	y/n/y/n	2.5 y / + (delayed engraftment)	arachnoid cyst, strabismus	2,3
CAMT039	f	n	E3	c.378delT	het	p.Phe126LeufsTer5	583	y (4)	n (ND)	y/n/y rectorrhagia	18	1: normocellular, MK↓↓	NAD	I	y/n/y/n	1,8 y / 2.3 y death	no	3
			E3	c.367C>T	het	p.Arg123Ter												
CAMT043	f	y	E3	c.304C>T	hom	p.Arg102Cys	NT	y (<20)	ND	ND	HSCT: 87	72: hypocellular, MK↓↓	M7 (45%) at age 50 mths	II	y/y/n/n G-CSF	7.3 y / +	no	3,4
CAMT050	f	y	E4	c.460T>C	hom	p.Trp154Arg	NT	y (12)	y (110)	n/n/n	HSCT: 35	hypocellular, MK↓↓	ND	II	y/n/y/n	2.9 y / +	no	3
CAMT052	f	n	E8	c.1305G>C	hom	p.Trp435Cys	1212	y (29)	y (110)	y/y/n	24	0: normocellular, MK↓↓	NAD	II	y/n/y/n	3.5 y (planned) / ND	no	2,3
CAMT055	m	y	E5	c.770G>T	hom	p.Arg257Leu	1271	y (20)	y (100)	y/n/n	37	15: normocellular, MK↓ 36: hypocellular, MK↓↓	NAD	II	y/n/n/n	3.2 y / +	atopic dermatitis	3
CAMT058	m	n	E4	c.407C>T	het	p.Pro136Leu	401 - 2585	n (60 at age 25 mo)	y (154)	y/y/n	106	104:hypocellular, MK↓↓	NAD	II	y/n/n/n	8.9 y / + (after 2 x donor lymphocytes)	eczema	3
			E12	c.1781T>G	het	p.Leu594Trp												
CAMT059	m	y	E8	c.1230G>A	hom	p.Trp410Ter	766	y (17)	ND	y/n/n GI bleeding	<12	1: normocellular, MK↓↓	NAD	I	y/n/y/y	1.0 y / +	high palate, small uvula	
CAMT067	m	n	E3	c.305G>C	het	p.Arg102Pro	NT	y (10)	ND	y/n/n	85	75: normocellular, MK↓↓ 85: hypocellular, MK↓↓	09/2003: 46XY 11/2004: monosomy	II	y/n/n/n oprelvekin	7.8 y (planned) / ND	obstrucrive sleep apnea, short stature, speech delay	3
			E3	c.311T>C	het	p.Phe104Ser												
CAMT075	m	n	E3	c.305G>C	hom	p.Arg102Pro	NT	y (14)	y (175)	y/n/n	8	ND	NAD	II	ND/ND/y/n	3.0 y / +	mild eczema	3
CAMT082	m	n	E3	c.305G>C	het	p.Arg102Pro	840	y (16)	y (70)	y/y/n	HSCT: ND	11: hypocellular, MK↓↓	ND	II	y/n/n/n	ND	strabismus	
			I3	c.391+5G>C	het	Splice defect												
CAMT083	m	n	E8	c.1305G>C	hom	p.Trp435Cys	NT	(y) (fetal)	ND	y/n/y	ND	normocellular, MK↓↓	ND	_	-	-	cerebellar hypoplasia, Dandy-Walker malformation, hydrocephalus	
CAMT087	m	y	E2	c.127C>T	hom	p.Arg43Ter	1932	y (ND)	ND	ND	ND / HSCT: 8	ND	ND	I	y/n/n/n	0.7 y / ND	no	
CAMT092	f	n	I3	c.391+5G>C	het	Splice defect	1472	n (<10 at age 18 mo)	(late detection)	y/y/n	120	16: normocellular, MK↓ 126: hypocellular, MK↓↓	NAD	II	ND/ND/y/y	11.0 y / +	no	5
			E9	c.1378C>T	het	p.Gln460Ter												
CAMT098	m	n	I1	c.79+2T>A	het	Splice defect (p.Asp27fs)	1244	n (32 at age 15 mo)	(late detection)	y/y/n	"late"	23: normocellular, MK↓↓	NAD	II	y/n/n/n	2.5 y / +	no	
			E4	c.404C>G	het	p.Pro135Arg												
CAMT101	m	y	E6	c.883G>C	hom	p.Asp295Tyr	1563	n (37 at age 24 mo)	(late detection)	y/n/n	>60	66: hypocellular, MK↓↓	NAD	II	y/n/n/y	planned	no	5
CAMT102	f	y	E6	c.883G>C	hom	p.Asp295Tyr	1468	n (79 at age 4 y)	(late detection)	y/n/n	ND	ND	NAD	II	n/n/n/n	5.5 y (planned) / ND	no	5
CAMT108	f	n	E3	c.378delT	het	p.Phe126LeufsTer5	864	y (3 at age 24 d)	n (ND)	y/n/y (?) gum bleeding, easy bruising upper extremities	<3	1: hypocellular, MK↓↓	NAD	I	y/y/n/n G-CSF	1.5 y / +	arachnoid cyst, nystagmus	6
			I11	c.1653delG	het	p.Lys553ArgfsX75												
CAMT113	f	n	I2	c.212+1G>A	het	Splice defect	NT	y (35)	y (110)	n (?)/n/n	55	55: hypocellular, MK↓↓	NAD	II	n/n/n/n Cotrimoxazol	ND	no	
			I3	c.391+5G>C	het	Splice defect												
CAMT122	f	y	E12	c.1742C>T	hom	p.Pro581Leu	1716	n (ND)	(late detection)	y/n/n	41	ND	ND	II	n/n/n/n	planned (info from age 3.8 y)	ND	
CAMT123	f	y	E4	c.460T>C	hom	p.Trp154Arg	564	y (6)	ND	y/y/y	25	0: normocellular, MK↓↓ 32: hypocellular, MK↓↓	ND	?	y/y/n/n	planned, waiting for donor	psychomotor retardation	7
CAMT125	f	y	E4	c.407C>A	hom	p.Pro136His	NT	y (ND)	ND	ND	72 (severe AA)	ND	ND	II	ND	6.9 y / +	no	
CAMT130	f	n	I3	c.391+5G>C	het	Splice defect	953	y (ND)	y (ND)	y/y/n	45	45: normocellular, MK↓↓	NAD	II	y/y/n/n	4.3 y / + (GVHD, AFP)	no	
			E5	c.769C>T	het	p.Arg257Cys												
CAMT133	f	y	E8	c.1180C>T	hom	p.Pro394Ser	437	ND	ND	y/n/n	>72	92: hypocellular, MK↓↓	ND	II	y/y/n/n	8.0 y / +	no	
CAMT136	m	y	E9	c.1361G>C	hom	p.Arg454Pro	1715	ND	ND	ND	>21	20: normocellular, MK↓↓	ND	II	ND Eltrombopag	-	no	
CAMT137	f	n	I3	c.391+5G>C	het	Splice defect	NT	n (65 at age 4 w)	y (65) (late detection)	n/n/n	HSCT: 43	ND	ND	II	n/n/n/n	3.6 y / +	no	
			E5	c.769C>T	het	p.Arg257Cys												
CAMT138	f	y	E9	c.1390A>G	hom	p.Arg464Gly	1363	n (6 at age 3 y)	(late detection)	y/y/n	>36	44: hypocellular, MK↓↓ 107: SAA, MK↓↓	NAD	II	y/y/n/n	8.9 y / +	hypopigmentation, skin lesions	
CAMT140	m	y	E8	c.1180C>T	hom	p.Pro394Ser	2767	n (ND)	y (ND)	ND	- (96)	normocellular, MK↓↓	ND	II	n/n/n/n	ND	insulin dependent diabetes,	
CAMT144	f	n	E3	c.304C>T	het	p.Arg102Cys	2284	y (25)	ND	y/n/y	8	3: normocellular, MK↓↓	NAD	?	y/y/y/y	planned (info from age 0.9 y)	strabismus	
			E4	c.407C>T	het	p.Pro136Leu												
CAMT157	m	n	E4	c.506T>A	hom	p.Leu169His	2230	y (3)	y (65)	y/y/y	30	3x normocellular, MK↓↓ 33: hypocellular, MK↓↓	NAD	II	y/y/y/y G-CSF	5.1 y / 6.1 y death	arachnoid cyst, strabismus	
CAMT159	f	y	E1	c.23T>G	hom	p.Met8Arg	1591	n (8 at age 9 mo)	(late detection)	y/y/n	24	9: normocellular, MK↓↓	ND	II	y/y/y/y	planned (info from age 7 y)	ND	
CAMT160	m	y	E3	c.235_236delCT	hom	p.Leu79GlufsX84	>4000	y (7)	n (<10)	y/y/y	- (10)	10: normocellular, MK↓↓	ND	I	y/n/y/y	ND	psychomotor retardation, Dandy-Walker malformation	
CAMT163	f	y	E9	c.1431G>A	hom	p.Trp477Ter	1685	ND	ND	ND	<36	ND	ND	?	ND	3.8 y / +	no	
CAMT167	f	y	E5	c.805T>C	hom	p.Trp269Arg	1493	y (ND, 54 at age 7w)	y (54)	y/n/y	- (12)	8: hypocellular, MK↓↓	NAD	II	y/n/y/y	planned (info from age 1 y)	psychomotor retardation	
CAMT168	m	y	E11	c.1571T>G	hom	p.Leu524Arg	1154	n (TP at age 8 mo)	(late detection)	ND	<48	44: hypocellular, MK↓↓	ND	II	y/y/y/y	planned (info from age 3.7 y)	ND	
CAMT169	m	n	E3	c.305G>A	het	p.Arg102His	1756	ND	ND	n/y/n	22	51: hypocellular, MK↓↓	ND	?	y/y/n/n	5.4 y (planned)	no	
			E3	c.235_236delCT	het	p.Leu79GlufsX84												
CAMT178	f	n	E3	c.305G>C	het	p.Arg102Pro	NT	y (12)	n	y/n/n	- (8)	normocellular, MK↓↓	ND	?	y/n/n/n	planned (info from age 2.2 y) waiting for donor	strabismus	
			E4	c.506T>A	het	p.Leu169His												

CAMT-ID	sex	consan-guineous parents	Intron/ Exon	cDNA	Zygosity	predicted protein	THPO plasma levels [pg/ml]	TP detected at birth? (plt count [G/l])	platelet rise? (max plt count)	Bleeding pet/epis/ICH other	First signs of multi-lineage cytopenia* [mo] or HSCT (if earlier) (if negative, age of last information is provided in brackets[mo])	Bone marrow findings (age [mo])	Cytogenetics	CAMT group	Therapy (other than HSCT) plt/RBC/Ig/Steroids other	HSCT / outcome	non-hematological findings	mentioned in earlier publications (Ref)
CAMT179	f	n	E4	c.407C>G	het	p.Pro136Arg	NT	y (19)	y (119)	y/n/n	ND	ND	ND	II	y/n/n/n	ND	no	
			E6	c.944T>G	het	p.Phe315Cys												
CAMT180	m	y	E2	c.127C>T	hom	p.Arg43Ter	881	y (ND)	ND	ND	ND / HSCT	ND	ND	?	ND	3.3 y / ND	ND	
CAMT181	f	n	E3	c.305G>C	het	p.Arg102Pro	586	y (4)	ND	y/n/y	- (5)	normocellular, MK↓↓	ND	?	y/n/n/n	0.7 y / +	no	
			I9	c.1469-2A>T	het	Splice defect												
CAMT183	f	n	E4	c.506T>A	hom	p.Leu169His	NT	y (1)	n (23)	y/y/y	- (18)	normocellular, MK↓↓	NAD	II	y/y/y/n	planned (info from age 1.8 y)	colpocephaly, agenesis corpus callosum, optic nerve hypopplasia	

Table S1: Detailed patient information

Abbreviations: d: day(s), epis: epistaxis, f: female, het: heterozygous, hom: homozygous, HSCT: hematopoietic stem cell transplantation, ICH: intracerebral hemorrhage, Ig: immunoglobulins, m: male, MK: megakaryocytes, mo: month(s), n: no, NAD: no abnormality detected, ND: no data, NT: not tested, pet: petechiae, plt: platelet(s), RBC: red blood cells, w: week(s), y: year(s)

* First signs of multilineage cytopenia: first reports about neutropenia, anemia or reduced cellularity in bone marrow

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Table S2: MPL mutations

	genomic (GRCh38)	Exon/Intron	CDS	type	protein	MutationTaster[1]	SIFT[2]	PROVEAN[3]	gnomAD[4] Freq.	dbSNP[5]	ClinVar[6]	HGMD[7]	incidence	alleles
a) nonsense mutations														
	Chr1:g.43338146C>T	E2	c.127C>T	nonsense	p.Arg43Ter	disease causing / 1			9/246204	rs148434485	RCV000411190.1	CM010061	ho: n=7; het: n=0	14
	Chr1:g.43338597C>T	E3	c.268C>T	nonsense	p.Arg90Ter	disease causing / 1			1/246262	rs763144679		CM010062	ho: n=1; het: n=0	2
	Chr1:g.43338696C>T	E3	c.367C>T	nonsense	p.Arg123Ter	disease causing / 1			1/246238	-		CM061128	ho: n=0; het: n=1	1
	Chr1:g.43346856G>A	E8	c.1230G>A	nonsense	p.Trp410Ter	disease causing / 1				-			ho: n=1; het: n=0	2
	Chr1:g.43348912C>T	E9	c.1378C>T	nonsense	p.Gln460Ter	disease causing / 1				-			ho: n=0; het: n=1	1
	Chr1:g.43348965G>A	E9	c.1431G>A	nonsense	p.Trp477Ter	disease causing / 1				-			ho: n=1; het: n=0	2
b) frame shift mutations														
	Chr1:g.43338564_43338565del	E3	c.235_236delCT	frame shift	p.Leu79Glufs*84	disease causing / 1.000			12/277118	rs587778514	RCV000121536.1 RCV000503692.1	CD010110	ho: n=2; het: n=1	5
	Chr1:g.43338707del	E3	c.378delT	frame shift	p.Phe126LeufsX5	disease causing / 1.000			11/277158	rs587778515	RCV000121537.1 RCV000255711.1	CD010111	ho: n=3; het: n=3	9
	Chr1:g.43352304delG	E11 (I11)	c.1653delG (c.1653+1delG)	frame shift (splicing)	p.Lys553ArgfsX75 (splice defect)	disease causing / 1.000			8/276684	rs755257605		CD076831	ho: n=0; het: n=1	1
c) splice site mutations														
	Chr1:g.43337929T>A	I1	c.79+2T>A	splicing (donor)	Splice defect (p.Asp27fs)	disease causing / 1.000			96/267198	rs146249964	RCV000254762.1 RCV000586535.3 RCV000122423.1	CS061304	ho: n=1; het: n=1	3
	Chr1:g.43338232G>A	I2	c.212+1G>A	splicing (donor)	Splice defect	disease causing / 1.000			5/245248	rs142565191			ho: n=0; het: n=1	1
	Chr1:g.43338541G>A	I2	c.213-1G>A	splicing (acceptor)	Splice defect (p.Arg71fs)	disease causing / 1.000				rs867404262		CS061305	ho: n=0; het: n=1	1
	Chr1:g.43338725G>	I3	c.391+5G>C	splicing (donor)	Splice defect	disease causing / 1.000			28/277018	rs752453717	RCV000414099.1	CS055604	ho: n=0; het: n=5	5
	Chr1:g.43349261A>T	I9	c.1469-2A>T	splicing (acceptor)	Splice defect	disease causing / 1.000							ho: n=0; het: n=1	1
d) missense mutations														
	Chr1:g.43337871T>G	E1	c.23T>G	missense	p.Met8Arg	polymorphism / 0.963	Deleterious (0.02)	Neutral (-1.53)	0				ho: n=1; het: n=0	2
	Chr1:g.43338633C>T	E3	c.304C>T	missense	p.Arg102Cys	disease causing / 1.000	Deleterious (0.03)	Deleterious (-2.81)	3/246270	rs763568293		CM055993	ho: n=1; het: n=1	3
	Chr1:g.43338634G>A	E3	c.305G>A	missense	p.Arg102His	disease causing /0.983	Tolerated (0.3)	Neutral (-1.08)	2/246270	-			ho: n=0; het: n=1	1
	Chr1:g.43338634G>C	E3	c.305G>C	missense	p.Arg102Pro	disease causing / 0.995	Tolerated (0.07)	Neutral (-2.30)	104/277190	rs28928907	RCV000121539.1 RCV000015221.26	CM002058	ho: n=4; het: n=5	13
	Chr1:g.43338640T>C	E3	c.311T>C	missense	pPhe104Ser	disease causing / 0.975	Deleterious (0.00)	Deleterious (-4.89)		-		CM061130	ho: n=0; het: n=1	1
	Chr1:g.43339283C>G	E4	c.404C>G	missense	p.Pro135Arg	disease causing / 0.995	Deleterious (0.00)	Deleterious (-5.12)	2/245764	rs761623764			ho: n=0; het: n=1	1
	Chr1:g.43339286C>A	E4	c.407C>A	missense	p.Pro136His	disease causing / 0.991	Deleterious (0.00)	Deleterious (-5.46)	3/245820	rs764904424		CM002059	ho: n=1; het: n=0	2
	Chr1:g.43339286C>T	E4	c.407C>T	missense	p.Pro136Leu	disease causing / 1.000	Deleterious (0.00)	Deleterious (-5.60)	2/245820	rs764904424		CM061125	ho: n=0; het: n=2	2
	Chr1:g.43339286C>G	E4	c.407C>G	missense	p.Pro136Arg	disease causing / 0.995	Deleterious (0.00)	Deleterious (-5.52)	1/245820	rs764904424			ho: n=0; het: n=1	1
	Chr1:g.43339339T>C	E4	c.460T>C	missense	p.Trp154Arg	disease causing / 0.984	Deleterious (0.00)	Deleterious (-9.75)	2/246240	rs758428763		CM061127	ho: n=2; het: n=0	4
	Chr1:g.43339385T>A	E4	c.506T>A	missense	p.Leu169His	disease causing / 0.903	Deleterious (0.00)	Deleterious (-3.48)		-			ho: n=2; het: n=1	5
	Chr1:g.43340042C>T	E5	c.769C>T	missense	p.Arg257Cys	disease causing / 1.000	Deleterious (0.01)	Deleterious (-5.31)	15/276484	rs121913611	RCV000015219.22	CM002060	ho: n=0; het: n=2	2
	Chr1:g.43340043G>T	E5	c.770G>T	missense	p.Arg257Leu	disease causing / 1.000	Deleterious (0.03)	Deleterious (-4.53)		-		CM061126	ho: n=1; het: n=0	2
	Chr1:g.43340078T>C	E5	c.805T>C	missense	p.Trp269Arg	disease causing / 0.994	Deleterious (0.00)	Deleterious (-10.14)		-			ho: n=1; het: n=0	2
	Chr1:g.43340096C>A	E5	c.823C>A	missense	p.Pro275Thr	disease causing / 0.985	Tolerated (0.11)	Deleterious (-4.94)	1/245852	rs28928908	RCV000015224.26	CM010064	ho: n=0; het: n=1	1
	Chr1:g.43340416G>C	E6	c.883G>C	missense	p.Asp295Tyr	disease causing / 0.998	Deleterious (0.01)	Deleterious (-3.20)		-			ho: n=2; het: n=0	4
	Chr1:g.43340477T>G	E6	c.944T>G	missense	p.Phe315Cys	disease causing / 0.999	Deleterious (0.01)	Deleterious (-3.25)	1/246262	rs750244518			ho: n=0; het: n=1	1
	Chr1:g.43346806C>T	E8	c.1180C>T	missense	p.Pro394Ser	disease causing / 0.981	Deleterious (0.00)	Neutral (-2.43)		-			ho: n=2; het: n=0	4
	Chr1:g.43346931G>C	E8	c.1305G>C	missense	p.Trp435Cys	disease causing / 1.000	Deleterious (0.00)	Deleterious (-6.27)	1/244636	rs1006158872		CM061129	ho: n=2; het: n=0	4
	Chr1:g.43348895G>C	E9	c.1361G>C	missense	p.Arg454Pro	polymorphism / 1.000	Tolerated (0.33)	Neutral (-1.59)		-			ho: n=1; het: n=0	2
	Chr1:g.43348924A>G	E9	c.1390A>G	missense	p.Arg464Gly	polymorphism / 0.993	Deleterious (0.03)	Deleterious (-3.64)	1/155566	-			ho: n=1; het: n=0	2
	Chr1:g.43352221T>G	E11	c.1571T>G	missense	p.Leu524Arg	disease causing / 0.807	Deleterious (0.01)	Deleterious (-2.78)		-			ho: n=1; het: n=0	2
	Chr1:g.43352606C>T	E12	c.1742C>T	missense	p.Pro581Leu	disease causing / 0.981	Tolerated (0.59)	Neutral (-2.10)		-			ho: n=1; het: n=0	2
	Chr1:g.43352645T>G	E12	c.1781T>G	missense	p.Leu594Trp	polymorphism / 0.986	Deleterious (0.03)	Neutral (-2.06)	1/246264	-	RCV000584875.1	CM061131	ho: n=0; het: n=1	1

n=6 not yet described in CAMT, but in one of the public
n=11 not yet published and in none of the public

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Table S3: CAMT patients with only one allele affected from deleterious MPL mutations

patient-ID	sex	intron/ exon	cDNA	zygosity	predicted protein	THPO level
CAMT139	f	E9	c.1390A>G	hetero	R464G	880 pg/ml
CAMT065	m	E2	c.127C>Y	hetero	R43X	ND
CAMT086	m	I3	c.391+5G>C	hetero	splicing defect	ND
CAMT088	f	E3	c.378delT	hetero	F126LfsX5	1786 pg/ml
CAMT129	m	E11	c.1612G>C	hetero	V538L	937 pg/ml
CAMT073	f	E7	c.1069C>T	hetero	R357X	2101 pg/ml
		E4	c.585T>C	hetero	P195P	

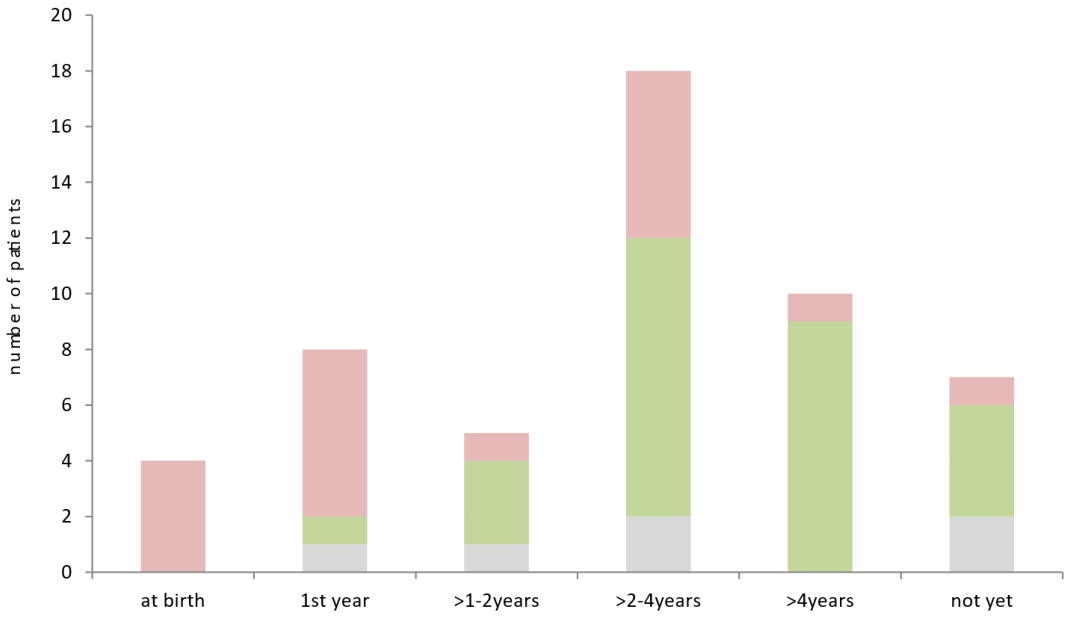

Figure S1: Development of multi-lineage cytopenia in CAMT-MPL patients
The figure summarizes the time of first reports about neutropenia, anemia, reduced cellularity in the bone marrow, or HSCT (if earlier) for patients in this study. red: CAMT I, green: CAMT II, grey: CAMT group unclear)