Table 1. Demographic, Hematologic and Clinical Characteristics of SLC25A38 CSA Patients. —, Unknown; ↓M:E, Decreased Myeloid:Erythroid Ratio; Abs Retic, Absolute Reticulocyte Count; ANC, Absolute Neutrophil Count; ASD, Autism Spectrum Disorder; C/B, CBC, Complete Blood Count; Complicated by; DD, Developmental Delay; DOL, Day of Life; DVT, Deep Venous Thrombosis; ECMO, Extracorporeal Membrane Oxygenation; F, Female; FTT, Failure to Thrive; GHD, Growth Hormone Deficiency; HGB, Hemoglobin; HTN, Hypertension; ID, Intellectual Disability; IVF, In Vitro Fertilization; LVH, Left Ventricular Hypertrophy; M, Male; MCV, Mean Corpuscular Volume; NP, Not Performed; PDA, Patent Ductus Arteriousus; PLT, Platelets; RDW, Red Cell Distribution Width; RS, Ring Sideroblasts; S/P, Status-Post; TX, Transfusion; WBC, White Blood Cell. *Reported by Kim MH et al. Clin Case Rep 2018; 6:1841-1844.

Patient	Relation to Proband	Sex	Year of Birth	Age at Presentation	Referral Country	Ancestry	Race/Ethnicity	Consanguinity	Age at CBC	TX Prior to CBC	HGB (g/dL)	MCV (fL)	RDW (%)	Abs Retic (M/μL)	WBC (K/μL)	ANC (cells/μL)	PLT (K/μL)	Bone marrow findings	RS (%)	Other Clinical Features
1.1	Self	M	2005	Infant	Lebanon	Lebanese	Arab	Yes	Infant	No	8.1	68	23.7	0.008	7.55	1480	484	↓M:E	17	None
2.1	Self	M	1996	Birth	US	Mexican	Hispanic	No	16	Yes	9.2	92	14.1	0.004	8.09	—	803	NP	—	DD,S/P splenectomy C/B thrombocytosis
2.2	Sibling	F	1997	Infant	US	Mexican	Hispanic	No	15	Yes	9.1	93	12.6	0.006	9.11	—	738	NP	—	None
3.1	Self	M	2000	Birth	Denmark	Danish	Caucasian	No	Infant	No	8.6	62	—	—	18.3	1010	562	Erythroid hypoplasia, Dyserythropoiesis	20	None
4.1	Self	F	1997	4 yrs.	US	Mexican/Filipino	Hispanic/Asian	No	14	No	8.4	67	27.7	0.028	3.6	1584	574	Initially normal, then normocellular with ↓M:E and RS	27	None
5.1	Self	M	—	—	UK	—	—	—	—	Yes	11.7	82	14.7	—	—	—	—	NP	—	—
6.1	Self	M	2004	Birth	Germany	—	Caucasian	No	Infant	Yes	6.2	70	—	0.010	5500	1320	405	Normocellular, ↓M:E	50	CM and IDDM improved after chelation therapy
7.1	Self	M	1999	Birth	Germany	—	Caucasian	No	1	No	5.1	61	—	—	8.1	1530	365	Normocellular, Erythroid hypoplasia	55	None
8.1	Self	M	1992	Birth	Australia	Lebanese	Caucasian	Yes	Infant	No	6.2	57	—	—	13.4	—	145	Dyserythropoiesis	>90	Phlebotomy post-HSCT
8.2	Sibling	M	1993	Birth	Australia	Lebanese	Caucasian	Yes	Infant	No	6.2	61	—	—	11.3	—	598	↓M:E	>90	Macrocephaly, Phlebotomy post-HSCT
9.1	Self	M	1986	14 yrs.	Kuwait	Arab	Arab	No	29	Yes	7.0	77	37.3	—	5.47	2850	375	Hypercellular, ↓M:E, Dyserythropoiesis	58	ID
10.1	Self	F	2005	Infant	Australia	Nepalese	South Asian	No	Infant	No	3.3	67	20.3	—	9.7	2100	677	↓M:E	12	No
11.1	Self	F	2017	Infant	Canada	Acadian	Caucasian	No	Infant	No	2.7	54	34.5	0.014	6.79	1.54	295	↓M:E	37	No
12.1	Self	F	1979	Infant	US	Northern European/ Native American	Caucasian/Native American	Yes	36	Yes	7.8	86	—	—	17.1	4340	820	Hypercellular, ↓M:E, Mild fibrosis	29	Cushingoid without history of excessive steroid use , DVT and thrombophlebitis, post-splenectomy
13.1	Self	F	2014	Prenatal	US	Northern European	Caucasian	No	Infant	Yes	3.3	75	16.4	—	2.6	600	480	Hypercellular, ↓M:E, Dyserythropoiesis	32	Unilateral corneal clouding, FTT, Congenital hypothyroidism, Congenital pulmonary hypertension requiring ECMO
14.1	Self	F	2011	Infant	Jordan	Jordanian	Arab	Yes	6	Yes	8.6	91	12.9	0.008	5.4	—	502	↓M:E, Hypercellular, erythroid hypoplasia	30	Box-shaped skull with hyperostotic calvarium
15.1	Self	F	2014	Birth	Egypt	Egyptian	Arab	Yes	3	Yes	6.0	86	35.1	—	13.5	—	717	Normocellular, Erythroid hypoplasia	Increased	Congenital pneumonia
16.1	Self	M	2008	Infant	Egypt	Egyptian	Arab	Yes	Infant	Yes	2.5	64	23.5	—	9.25	—	143	Hypercellular, ↓M:E, Dyserythropoiesis	37	None
17.1	Self	M	2015	Birth	Jordan	Jordanian	Arab	Yes	2	Yes	7.6	75	14.8	—	15.6	—	159	Normocellular	>50	Macrocephaly, Meningomyelocele, Club foot
17.2	Sibling	F	2017	Birth	Jordan	Jordanian	Arab	Yes	1	Yes	9.3	96	14.7	0.021	17.13	—	227	Normocellular	>90	None
18.1	Self	M	2006	Birth	US	Northern European	Caucasian	No	Infant	Yes	8.2	75	30.9	0.010	8.2	2378	280	Normocellular, Erythroid hypoplasia	None	Microphallus, Bicuspid aortic valve, ASD with aggressive behaviors, Speech delay, IVF conception
19.1	Self	M	2004	Infant	US	African American	Black	—	4	No	4.8	59	28	—	4	—	388	Hypercellular, ↓M:E, Dyserythropoiesis, Moderate fibrosis	Rare	—
20.1	Self	F	2009	Birth	US	Mexican	Hispanic	No	Infant	No	3.3	57	31.1	—	Normal	—	Normal	Hypercellular, ↓M:E	5-10	Phlebotomy post-HSCT
20.2	Sibling	F	2019	Birth	US	Mexican	Hispanic	No	Infant	No	5.9	70	37.6	0.078	5.8	1400	404	Hypercellular	Rare	PDA, LVH at birth. Coronary fistula.
21.1	Self	M	2010	Infant	US	European	Caucasian	—	1	No	1.3	60	—	—	1.8	—	142	Hypercellular, ↓M:E	Numerous	ASD, S/P splenectomy C/B thrombocytosis,
22.1	Self	F	2011	Birth	US	Guatemalan	Hispanic	No	Infant	No	2.7	—	—	—	52	2.23	100	Hypercellular	<10	DD, Syndromic facies, Behavioral outbursts/ASD, HSM, Pulmonary HTN, CM, Respiratory failure @ birth requiring ECMO
22.2	Sibling	F	2012	Birth	US	Guatemalan	Hispanic	No	DOL 0	Yes	9.1	74	32.2	—	20.2	8.7	326	Normocellular, Erythroid hypoplasia	Rare	Aortic root dilation
22.3	Sibling	M	2015	Birth	US	Guatemalan	Hispanic	No	—	—	—	—	—	—	—	—	—	NP	—	Hypospadias; PDA, Aortic root dilation
22.4	Sibling	M	2018	Prenatal	US	Guatemalan	Hispanic	No	DOL 0	Yes	13.8	77	24.7	0.220	32.5	14.95	585	NP	—	Hypospadias, In utero TX x 5
23.1	Self	F	2015	Birth	Australia	Pakistani	South Asian	Yes	2	Yes	8.5	77	—	—	9.2	—	189	Hypercellular, ↓M:E	>15	Asymptomatic b-ureidopropionase deficiency,UPB1, (homozygous c.873+1G>A),
24.1	Self	F	2019	Infant	US	European/Indian	Caucasian/South Asian	No	1	No	5.1	61	37.3	0.033	8.3	—	343	↓M:E	70	Frontal Bossing, "Box-Shaped" Head

Patient	SLC25A38 Mutations (NM_17875.3)						Homozygous
	Allele 1			Allele 2			
	cDNA	Protein	Type	cDNA	Protein	Type	
1.1	c.276+1G>A	p.?	SPL	c.276+1G>A	p.?	SPL	Yes
2.1	c.832C>T	p.Arg278X	X	c.832C>T	p.Arg278X	X	Yes
2.2	c.832C>T	p.Arg278X	X	c.832C>T	p.Arg278X	X	Yes
3.1	c.324_325del	p.Tyr109Leufs*43	FS	c.324_325del	p.Tyr109Lfs*43	FS	Yes
4.1	c.70-2A>C	p.?	SPL	c.70-2A>C	p.?	SPL	Yes
5.1	c.669_682del	p.Cys223Trpfs*67	FS	c.913T>C	p.X305Argext*28	EXT	No
6.1	c.277-2A>C	p.?	SPL	c.277-2A>C	p.?	SPL	Yes
7.1	c.207_214del	p.Met70Cysfs*80	FS	c.362del	p.Pro121Glnfs*26	FS	No
8.1	c.388G>A	p.Gly130Arg	MS	c.389G>A	p.Gly130Glu	MS	Yes
8.2	c.388G>A	p.Gly130Arg	MS	c.389G>A	p.Gly130Glu	MS	Yes
9.1	c.792+5G>C	p.?	SPL	c.792+5G>C	p.?	SPL	Yes
10.1	c.480dup	p.Ile161TyArgfs*12	FS	c.480dup	p.Ile161Tyrfs*12	FS	No
11.1	c.349C>T	p.Arg117X	X	c.349C>T	p.Arg117X	X	Yes
12.1	c.324_325del	p.Tyr109Leufs*43	FS	c.324_325del	p.Tyr109Leufs*43	FS	Yes
13.1	c.324_325del	p.Tyr109Leufs*43	FS	c.324_325del	p.Tyr109Lfs*43	FS	Yes
14.1	c.400C>T	p.Arg134Cys	MS	c.400C>T	p.Arg134Cys	MS	Yes
15.1	c.175C>T	p.Gln59X	X	c.175C>T	p.Gln59X	X	Yes
16.1	c.809dup	p.Phe271Leufs*24	FS	c.809dup	p.Phe271Leufs*24	FS	Yes
17.1	c.672delinsTT	p.Ile225Tyrfs*70	FS	c.672delinsTT	p.Ile225Tyrfs*70	FS	Yes
17.2	c.672delinsTT	p.Ile225Tyrfs*70	FS	c.672delinsTT	p.Ile225Tyrfs*70	FS	Yes
18.1	c.324_325del	p.Tyr109Leufs*43	FS	c.400C>T	p.Arg134Cys	MS	No
19.1	c.305G>A	p.Gly102Glu	MS	c.349C>T	p.Arg117X	X	No
20.1	c.832C>T	p.Arg278X	X	c.832C>T	p.Arg278X	X	Yes
20.2	c.832C>T	p.Arg278X	X	c.832C>T	p.Arg278X	X	Yes
21.1	c.349C>T	p.Arg117X	X	c.[161G>A;349C>T]	p.[Arg54His;Arg117X]	MS & X	No
22.1	c.457-1G>T	p.?	SPL	c.457-1G>T	p.?	SPL	Yes
22.2	c.457-1G>T	p.?	SPL	c.457-1G>T	p.?	SPL	Yes
22.3	c.457-1G>T	p.?	SPL	c.457-1G>T	p.?	SPL	Yes
22.4	c.457-1G>T	p.?	SPL	c.457-1G>T	p.?	SPL	Yes
23.1	c.475del	p.Glu159Argfs*7	FS	c.475del	p.Glu159Argfs*7	FS	Yes
24.1	c.401G>A	p.Arg134His	MS	c.560G>A	p.Arg187Gln	MS	No

Table 2: Genetic Characteristics of SLC25A38 CSA Patients in this Report. EXT, Extension/Stop-Lost; FS, Frameshift; MS, Missense; SPL, Splicing; X, Nonsense/Stop-Gained alleles

Table 3: Iron Status, Chelation Therapy and Outcomes of SLC25A38 CSA Patients. BX; Biopsy; CM, Cardiomyopathy; DFO, Deferoxamine; DFP, Deferiprone; DFX, Deferisirox; HE, Hepatic Encephalopathy; HSCT, Hematopoietic Stem Cell Transplantation; IDDM, Insulin-Dependent Diabetes Mellitus; MRI, Magnetic Resonance Imaging; NA, Not Applicable; ND, Not Determined; PHP, Panhypopituitarism; SS, Short Stature TX, Transfusion.

Patient	TX	Age at First TX	TX interval (wks)	TfSat (%)	Ferritin (mg/L)	Iron overload	Anemia/Iron Complications	Chelator(s)	HSCT?	Status	Cause of Death	Age at Last Follow-Up or Death
1.1	Yes	Infant	3	—	557	ND	None	DFX	No	Alive	NA	15
2.1	Yes	Neonate	3-5	—	—	Yes, Liver, Pancreas, Pituitary & Heart (MRI)	CM, PHP, IDDM, SS	DFX	No	Dead	CM	18
2.2	Yes	Neonate	3-5	—	1092	Yes	None	DFO, DFX, DFP	No	Alive	NA	23
3.1	Yes	Neonate	3-4	—	—	Yes	None	DFO or DFX	Yes	Alive	NA	21
4.1	Yes	8 yrs.	4	44	63	Yes, Liver, Pancreas and Heart (MRI)	SS	DFX & DFO	No	Alive	NA	21
5.1	Yes	—	3-5	—	—	—	—	—	—	—	—	15
6.1	Yes	Neonate	3-5	95	882	Yes, Liver and Heart	CM, IDDM	DFO	No	Alive	NA	14
7.1	Yes	Neonate	3-5	92	405	Yes, Liver (MRI)	None	DFO	No	Alive	NA	3
8.1	Yes	Neonate	4	—	—	Yes, Liver (BX)	None	DFO	Yes	Alive	NA	26
8.2	Yes	4 yrs.	4	—	—	Yes, Liver (BX)	None	DFO	Yes	Alive	NA	23
9.1	Yes	—	Rarely	—	2893	ND	SS	DFX	Yes	Alive	NA	32
10.1	Yes	Infant	4	—	1037	Yes, Liver (MRI)	None	DFX & DFP	No	Alive		15
11.1	Yes	Infant	3-4	—	269	Yes, Liver (MRI), No, Heart (MRI)	None	DFX	No	Alive		3
12.1	Yes	Infant	2	—	1373	Yes, Liver (MRI), No, Heart (MRI)	HE	DFX	No	Alive	NA	39
13.1	Yes	In utero	2	—	—	ND	None	DFX & DFO→ DFP & DFO	No	Alive	NA	6
14.1	Yes	Infant	2-3	116	832	Yes, Liver (BX)	None	DFX	Yes	Alive	NA	9
15.1	Yes	Neonate	6	—	362	ND	None	DFX	No	Alive	NA	6
16.1	Yes	Infant	4	—	596	ND	None	DFX	Yes	Alive	NA	12
17.1	Yes	Neonate	2-3	—	—	ND	None	No	No	Dead	Sepsis	3
17.2	Yes	Neonate	3	70	770	ND	None	No	Yes	Alive	NA	3
18.1	Yes	Neonate	2-3	—	~1000	Yes, Liver (BX and MRI)	SS, GHD	DFO→ DFX	Yes	Alive	NA	14
19.1	Yes	4 yrs.	4	100	1050	ND	None	DFX + IV DFO	No	Alive	NA	14
20.1*	Yes	Neonate	4-8	60	1817	Yes, Liver (MRI), No, Heart (MRI)	None	DFX	Yes	Alive	NA	11
20.2	Yes	Neonate	4-5	84	938	ND	None	DFX	No	Alive		1
21.1	Yes	Infant	4	62	945	Yes, Liver and Heart (MRI)	None	DFX & DFO→DFX & DFP	No	Alive	NA	10
22.1	Yes	Infant	2	95	1303	Yes, Liver (MRI),	None	DFX & DFO	No	Alive	NA	9
22.2	Yes	Infant	3	101	1431	Yes, Liver (MRI),	None	DFX & DFO	No	Alive	NA	8
22.3	Yes	Infant	3	95	1586	Yes, Liver (MRI),	None	DFX & DFO	No	Alive	NA	5
22.4	Yes	In utero	3	86	859	ND	None	No	No	Alive	NA	2
23.1	Yes	Neonate	4-5	_	500	ND	None	DFX	No	Alive	NA	5
24.1	Yes	Infant	4	91	632	ND	None	None	No	Alive	NA	2

Table 5. Hematopoietic Stem Cell Transplantation Outcomes of SLC25A38 CSA Patients. Ag, HLA-Antigen; AIHA, Autoimmune Hemolytic Anemia; BM, Bone Marrow; Bu, Busulfan; BX, Biopsy; CAM; Campath; CsA; Cyclosporine A; Cy, Cytoxan; Flu,

Fludarabine; aGVHD, Acute Graft vs. Host Disease; cGVHD, Chronic Graft vs. Host Disease; HSCT, Hematopoietic Stem Cell Transplantation; MRD, Matched Related Donor; MMSD, Mismatched Sibling Donor; MTX, Methotrexate; MUD, Matched Unrelated Donor; NA, Not Applicable; ND, Not Determined; PB, peripheral blood; PPX, Prophylaxis; PT, Post-Transplant; RIC, Reduced Intensity Conditioning; SD, Sibling Donor; TLI, Total Lymphoid Irradiation; Treo, Treosulfan; TT, Thiotepa; TX; Transfusion.

Patient	Year of HSCT	Age at HSCT (yrs.)	Pre-HSCT Ferritin	Donor type	Stem cell source	Conditioning Intensity	Conditioning	GVHD PPX	Most Recent Chimerism (%)	aGVHD	cGVHD	**Other HSCT Complications**	Follow up (yrs.)
3.1	2012	12	1500	MUD	BM	MAC	Bu/Cy	CsA/MTX	100%	None	None	None	8
8.1	2009	7	1825	MSD	BM	MAC	Bu/Cy/ATG	CsA/MTX	ND	None	Lung	None	11
8.2	2003	9	1700	MUD	BM	MAC	Bu/Cy/TT/ATG	CsA/MTX	100	None	Lung, Skin, Liver	None	17
9.1	2017	31	11154	MUD	BM	RIC	Flu/Bu	TAC/MTX	97	None	None	None	3
14.1	2018	8	ND	MMSD (1 Ag)	PB	RIC	Flu/Bu/ATG/TLI	PT-Cy/ CsA/ MMF	40 @12 mos. 4 @18 mos.	None	None	Graft failure	2
16.1	2018	11	750	MSD	PB	MAC	Bu/Cy/ATG	CsA	100	None	None	None	2
17.2	2020	3	ND	MRD	BM	MAC	ATG/Bu/Cy	CsA/MTX	Pending	None	NA	None	1 mo.
18.1	2012	6	342	MUD	BM	RIC	Flu/Treo/ATG	TA/MTX	100	Skin	Gut,Oral	TX-dependent post-HSCT AIHA, splenectomy	8
20.1	2013	4	783	MSD	BM	RIC	Flu/Bu/CAM	TAC/MTX	100	None	None	None	7

Table 6: The Currently Described SLC25A38 Mutation Spectrum. EXT, Extension/Stop-Lost; FS, Frameshift; MS, Missense; SPL, Splicing; X, Nonsense/Stop-Gained. †In at least two cases rs121918330 is in cis with rs144319567: c.161G>A; p.Arg64His. ‡Last base pair of exon predicted to affect splicing. ^Predicted to create a new splice acceptor site. ¶One patient is homozygous due to uniparental disomy.

cDNA NM_017875.3	Sequence Feature	Protein	Exon	Type	Number of Families	Number of Mutations @ Codon	SNP ID	gnomAD MAF	References
c.070-2A>C	—	p.?	2	SPL	1	1	—	—	Current
c.166C>A	CpG	p.Gln56Lys	2	MS	3	1	—	—	Fouquet et al., 2019; Kannengiesser et al., 2011
c.175C>T	—	p.Gln59X	2	X	2	1	—	—	Current; W. B. An et al., 2019
c.207_214del	Direct Repeat	p.Met70Cysfs*80	3	FS	1	1	—	—	Current
c.227_236del	—	p.Lys76Thrfs*17	3	FS	1	1	—	—	Shefer Averbuch et al., 2018
c.260G>A	—	p.Trp87X	3	X	1	1	—	—	W. An et al., 2015; W. B. An et al., 2019
c.276+1G>A	—	p.?	3	SPL	1	1	—	—	Current
c.276+1G>T	—	p.?	3	SPL	1	1	—	—	Ulirsch et al., 2019
c.277-1G>A	—	p.?	4	SPL	1	1	—	—	Guernsey et al., 2009
c.277-2A>C	—	p.?	4	SPL	1	1	—	—	Current
c.281T>A	—	p.Ile94Asn	4	MS	1	1	—	—	Liu et al., 2013
c.305G>A	—	p.Gly102Glu	4	MS	2	1	—	—	Current; Guernsey et al., 2009
c.324_325del	CT Repeat	p.Tyr109Leufs*43	4	FS	12	2	rs755447127	1.66E-04	Current; Fouquet et al., 2019; Guernsey et al., 2009; Kannengiesser et al., 2011; Le Rouzic et al., 2017
c.324_330del	CT Repeat	p.Tyr109X	4	X	1	2	—	—	Mehri et al., 2018
c.336_346del	—	p.Lys112Asnfs*37	4	FS	1	1	rs1301033567	—	Guernsey et al., 2009
c.349C>T†	CpG	p.Arg117X	4	X	15	1	rs121918330	1.59E-05	Current; Fouquet et al., 2019; Guernsey et al., 2009; Kakourou et al., 2016; Kannengiesser et al., 2011; Le Rouzic et al., 2017; Ravindra et al., 2020
c.362del	—	p.Pro121Glnfs*26	4	FS	1	1	—	—	Current
c.388G>A	—	p.Gly130Arg	4	MS	1	2	—	—	Current
c.389G>A	—	p.Gly130Glu	4	MS	1	2	rs762562272	3.98E-06	Guernsey et al., 2009
c.400C>T	CpG	p.Arg134Cys	4	MS	6	2	rs1293528130	3.98E-06	Current ; W. An et al., 2015; W. B. An et al., 2019; Fouquet et al., 2019; Kannengiesser et al., 2011; Le Rouzic et al., 2017
c.401G>A	CpG	p.Arg134His	4	MS	4	2	—	—	Current; Fouquet et al., 2019; Guernsey et al., 2009; Le Rouzic et al., 2017
c.409dup	—	p.Ala137Glyfs*16	4	FS	6	1	—	—	Ravindra et al., 2020
c.429_431deilinsAG	—	p.Ile144Alafs*3	4	FS	1	1	—	—	W. An et al., 2015; W. B. An et al., 2019
c.440T>A	—	p.Ile147Asn	4	MS	2	1	—	—	Fouquet et al., 2019; Kannengiesser et al., 2011
c.457-1G>T	—	p.?	4	SPL	1	1	rs1448237170	4.00E-06	Current
c.469G>C	—	p.Gly157Arg	4	MS	1	1	—	—	Liu et al., 2013
c.475del	CpG	p.Glu159Argfs*7	4	FS	1	1	—	—	Current
c.480dup	—	p.Ile161Tyrfs*12	5	FS	2	1	—	—	Current; Wong et al., 2015
c.560G>A	CpG	p.Arg187Gln	5	MS	6	2	rs121918331	7.96E-06	Current; W. An et al., 2015; W. B. An et al., 2019 Kannengiesser et al., 2011; Ravindra et al., 2020; Uminski et al., 2020
c.560G>C^	CpG	p.Arg187Prop.?	5	MS-SPL	1	2	rs121918331	7.96E-06	Guernsey et al., 2009
c.562G>C	—	p.Asp188His	5	MS	2	1	—	—	Ravindra et al., 2020
c.569C>G	—	p.Pro190Arg	5	MS	1	1	—	—	Kannengiesser et al., 2011
c.587T>C	—	p.Leu196Pro	5	MS	2	1	—	—	Fouquet et al., 2019; Le Rouzic et al., 2017
c.625G>C‡	CpG	p.Asp209Hisp.?	5	MS-SPL	4	1	rs1372117091	3.98E-06	Guernsey et al., 2009; Kannengiesser et al., 2011; Ravindra et al., 2020
c.626-2A>T	—	p.?	6	SPL	1	1	—	—	Ravindra et al., 2020
c.669_682del	—	p.Cys223Trpfs*67	6	FS	1	1	rs781372292	1.77E-05	Current
c.672delinsTT	—	p.Ile225Tyrfs*70	6	FS	2	1	—	—	Current; Shefer Averbuch et al., 2018
c.683G>T	—	p.Gly228Val	6	MS	4	1	rs755205622	3.98E-06	Kannengiesser et al., 2011; Mehri et al., 2018
c.689T>C	—	p.Leu230Pro	6	MS	1	1	—	—	Liu et al., 2013
c.790A>T	—	p.Lys264X	6	X	5	1	—	—	Fouquet et al., 2019; Guernsey et al., 2009; Le Rouzic et al., 2017; Ulirsch et al., 2019
c.792+5G>C	—	p.?	6	SPL	1	1	—	—	Current
c.809dup	—	p.Phe271Leufs*24	6	FS	1	1	—	—	Current
c.832C>G	CpG	p.Arg278Gly	6	MS	2	2	rs147431446	—	Kannengiesser et al., 2011; Le Rouzic et al., 2017
c.832C>T	CpG	p.Arg278X	6	X	5¶	2	rs147431446	3.18E-05	Current; Andolfo et al., 2020; Fouquet et al., 2019; Guernsey et al., 2009
c.858del	—	p.Ala287Glnfs*10	6	FS	1	1	—	—	Mehri et al., 2018
c.879T>G	—	p.Tyr293X	6	X	3	1	—	—	Fouquet et al., 2019; Guernsey et al., 2009; Le Rouzic et al., 2017
c.913T>C	—	p.*305Argext*28	6	EXT	2	1	rs1218815001	3.18E-05	Current; Guernsey et al., 2009