

Immunoglobulin heavy and light chain gene features are correlated with primary cold agglutinin disease onset and activity

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doi:10.3324/haematol.2016.153411

Suppl. Table 1: Patient cohort

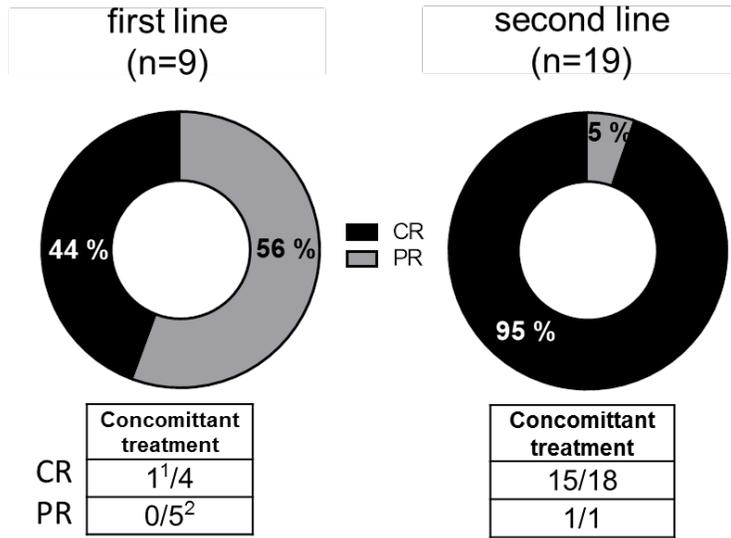
Pt #	Gender	Genetic	Mutation	Protein level	FAS Domain	Age of onset [yrs]	Clinical presentation	Status at last follow up
1	F	FAS Htz	Ex 9 c.688-689dup	p.Ser230fs Arg Ter12	IC	0,0	LP/AI (bone infiltration)	Remission
2	M	FAS Htz	Ex 9 c.709 G>C	p.Ala237Pro	IC	1,0	LP/AI (pancytopenia)	Remission
3	F	FAS Htz	Ex 7 c.586 C>T	p.Gln196Ter	IC	5,0	LP	Remission
4	M	FAS Htz	Ex 8 c.671 T>G	p.Leu224Ter	IC	0,3	LP- AI(AIHA)	Remission
5	M	FAS Htz	Ex 8 c.652-2A>T	P.Glu218fsMet Ter3	IC	0,2	LP/AI(AIHA)	Remission
6	M	FAS Htz	Ex 9 c.794A>G	p.Asp265Gly	IC	12,0	LP/AI(AIHA)	Remission
7	M	FAS Htz	Ex 6 c.506-3C>T	p.Gly169_Trp189del	TM	11,0	AI (ITP)	Remission
8	F	FAS Htz	Ex 9 c.748C>T	p.Arg250Ter	IC	9,0	LP	Remission
9	F	sFAS	Ex 9 c.812_814del	p.Ala271del	IC	0,6	LP	Remission
10	F	sFAS	Ex9 c.768C>G	p.Arg250Gly	IC	0,8	LP/AI(AIHA)	Remission
11	M	sFAS	Ex 9 c.815 A>G	p.Glu272Gly	IC	3,4	LP/AI (ITP)	Remission
12	F	FAS Htz	Ex 6 c.506-2 A>G	p.Gly169_Trp189del	TM	14,0	AI (AIHA)	Remission
13	F	FAS htz	Ex 7 c.613_617del	p.Glu205fsProTer4	IC	3,0	LP/AI(AIHA)	PR
14	F	FAS hmz	Ex 4 c.327T>G	p.Cys143Arg	EC	0,2	LP/AI(AIHA)	Remission
15	M	FAS htz	Ex 4 c.410_411del	p.Ser137fsTyrTer3	EM	4,0	LP/AI(AIHA)	Remission
16	F	FAS Htz	Ex 3 c.275_276delCA	p.Thr92ArgfsTer13	EC	1,6	LP/AI(AIHA/ITP/NTP)	Remission
17	F	sFAS	Ex 9 c.712G>T	p.Gly238Ter	IC	0,5	LP	Remission
18	M	FAS Htz	Ex 9 c.779A>G	p.Asp260Gly	IC	0,5	LP	Remission
19	M	FAS Htz	Ex 6 c.536T>G	p.Leu179Arg	TM	4,0	LP/AI(AIHA)	Remission
20	M	sFAS	IVS7 c.652-2A>T	Splice site	IC	3,7	LP/AI(AIHA)	PR
21	M	sFAS	IVS7 c.652-1G>A	Splice site	IC	2,3	LP/AI(AIHA)	Remission
22	M	sFAS	Ex 8 c.657_658delAG	p.Val220GlyfsTer6	IC	1,0	LP/AI(ITP)	off test
23	F	sFAS	Ex 9 c.778G>T	p.Asp260Tyr	IC	0,8	LP/AI(Anemia)*	Remission
24	M	FAS Htz	Ex 4 c.405T>A	p.Cys135Ter	EC	2,0	LP/AI(AIHA)	Remission
25	F	FAS Htz	Ex 8 c.676_+1G/C	p.Pro217fs	IC	0,7	LP	PR
26	M	FAS Htz	Ex 3 c.471_474delGACA	p.Thr76fs	EC	1,8	LP/AI(AIHA)	Remission
27	F	FAS Htz	Ex 6 c.568G>T	p.Val190Leu	TM	2,5	LP	Remission
28	M	FAS Htz	Ex 9 c.792T>G	p.Asn264Lys	IC	26,0	LP	off test, PR with MMF

Suppl. Table 1: Table of patient characteristics, mutations, and clinical manifestations.

M=male, F=female; IC=intracellular, TM=transmembrane, EC=extracellular; LP=lymphoproliferation, AI=autoimmunity, AIHA=autoimmune hemolytic anemia, ITP=immune thrombocytopenia, NTP=autoimmune neutropenia, PR=partial remission.

*P23 suffered from anemia requiring transfusion, but Coombs-test was negative.

Suppl. Fig. 1: Clinical rapamycin response at 6 months



Suppl. Fig. 1: Clinical response to rapamycin 6 months after treatment initiation.

Pie-charts show percentages of complete and partial remission of patients receiving rapamycin as a first line treatment (left) or second line treatment (right).¹The patient received steroids concomitantly to rapamycin initiation. ²Two of these five patients had documented malcompliance.