Current Medical Science 41(3),2021
DOI https://doi.org/10.1007/s11596-021-2375-5

Table S1 The clinical data of petients with EBV-LPDs (including the genetic variants list)

																					Relative genetic mutations													
Case	Gender	Age (years)	Diagnosis	Highest EBV titers copies/mL	WBC ($\times10^9$/L) (3.50–9.50)	Neutrophil ($\times10^9$/L) (1.80–6.30)	lymphocyte ($\times10^9$/L) (1.10–3.20)	Hb (g/L)	PLT ($\times10^9$/L)	ALT (U/L)	AST (U/L)	LDH (U/L)	TG (mmol/L)	Fibrinoge (ng/L)	High triglycerides or low fibrinogen (0/1)	Highest ferritin (μg/L)	Fever (0/1)	Hepatosplenomegaly (0/1)	Superficial lymph node enlargement (0/1)	Rash (0/1)	Hemophagocytic symptom of bone marrow	Total mutation	*LYST*	*UNC13D*	*ITK*	*PRF1*	*STXBP2*	*AP3B1*	*GZMB*	*RAB27A*	*XIAP*	*STX11*	*GNLY*	*CD27*	*SRGN*
1	Male	8	CAEBV-T/NK	7660000	4.25	2.07	1.86	104	520	83	98	434	2.83	1.95	1	205.1	1	1	0	0	0	AP3B1 1234T>C, Y412H						AP3B1 1234T>C, Y412H							
2	Male	51	CAEBV-T/NK	74600000	8.2	2.96	4.61	116	272	1280	116	232	1.56	3.42	1	470.30	1	0	0	0	0														
3	Male	18.6	CAEBV-T/NK	305000	12.05	0.82	8.8	145	134	299	225	848	1.7	2.2	1	267.60	1	1	1	0	0														
4	Male	29.3	CAEBV-T/NK	1090000	4.43	3.15	0.79	105	185	47	27	299	2.01	3.04	1	15.00	1	1	1	1	0	LYST 8368A>C, K2790Q	LYST 8368A>C, K2790Q												
5	Female	28.3	CAEBV-T/NK	10200000	3.72	2.02	1.18	132	180	16	22	166	1.68	2.53	1	127.50	0	0	1	0	0	LYST 8368A>C, K2790Q	LYST 8368A>C, K2790Q												
6	Female	4.5	CAEBV-T/NK	53200	6.85	2.46	3.73	126	401	56	62	277	1.63	3.42	1	470.30	1	0	1	0	0														
7	Male	4.4	CAEBV-T/NK	20200000	4.73	1.55	2.81	83	141	97	116	636	2.32	1.96	1	200.30	1	1	0	0	0														
8	Male	17.3	CAEBV-T/NK	737000	26.06	1.35	22.45	140	133	257	147	882	3.87	1.96	1	670.00	0	1	1	0	0	XIAP 962C>G, A321G									XIAP 962C>G, A321G				
9	Male	12	CAEBV-T/NK	1750000	5.28	2.59	2.01	116	158	56	62	378	2.42	2.12	1	167.3	1	1	1	0	0	CD27 779C>T, P260L												CD27 779C>T, P260L	
10	Male	11.2	CAEBV-T/NK	11700000	3.64	2.17	1.41	133	36	331	709	1262	1.03	1.3	1	4171.2	1	1	0	1	0	LYST 10526G>A, R3509Q	LYST 10526G>A, R3509Q												
11	Female	20.6	CAEBV-T/NK	26100000	4.16	3.16	0.82	115	121	67	75	572	1.65	3.26	1	249.90	1	1	1	0	0														
12	Male	18	CAEBV-T/NK	3830000	9.37	7.11	1.88	159	105	49	42	344	0.95	2.92	1	367.30	1	1	1	0	0	XIAP.894_898de, K299fs									XIAP.894_898de, K299fs				
13	Female	12.2	CAEBV-T/NK	9800000	8.79	2.18	5.10	108	302	130	120	537	1.42	2.51	1	482.8	1	1	0	0	0	RAB27A 560G>A, R187Q								RAB27A 560G>A, R187Q					
14	Male	46.2	CAEBV-T/NK	6870000	2.54	1.64	0.54	119	141	167	134	301	3.24	1.65	1	1207.3	1	1	1	0	1														
15	Male	23.0	CAEBV-T/NK	652000	9.14	7.42	1.01	120	122	84	39	378	2.42	2.24	1	68.7	0	1	1	0	0	LYST 8368A>C, K2790Q; PRF1 889C>T, R297W; LYST 3456T>G, I1152M	LYST 8368A>C, K2790Q; LYST 3456T>G, I1152M			PRF1 889C>T, R297W									
16	Female	10.1	CAEBV-T/NK	320000	2.79	2.15	0.53	113	121	16	52	662	0.91	2.87	1	462	1	1	0	0	0	PRF1 674G>A, R225Q				PRF1 674G>A, R225Q									
17	Male	29.3	CAEBV-T/NK	490000	5.59	2.56	2.02	141	123	320	127	232	2.13	2.33	1	411.7	1	0	0	1	0														
18	Female	13.1	CAEBV-T/NK	12300000	3.97	1.96	1.66	97	245	152	162	308	1.45	2.52	1	71.7	0	1	0	1	0														
19	Male	18	CAEBV-T/NK	187000	1.54	0.84	0.29	86	170	57	85	1150	2.18	1.15	1	1698.10	1	1	1	1	0	LYST 8368A>C, K2790Q	LYST 8368A>C, K2790Q												
20	Female	49	CAEBV-T/NK	17800000	4.78	2.57	1.75	124	131	16	27	196	1.37	2.61	1	111.60	1	0	1	0	0	LYST 3103A>G, K1035E	LYST 3103A>G, K1035E												
21	Female	29.0	CAEBV-T/NK	7450	5.89	3.03	2.34	143	258	22	22	204	2.01	1.93	1	94.9	0	0	0	0	0														
22	Male	42.0	CAEBV-T/NK	18400	4.67	2.83	1.24	143	218	27	26	178	1.56	3.87	1	77.20	0	0	1	0	0														
23	Male	16	CAEBV-T/NK	1e7	3.79	1.85	1.50	95	73	109	79	507	1.34	1.80	1	1380	1	0	0	0	0														
24	Female	35	CAEBV-T/NK	1430000	3.79	1.52	1.89	90	160	17	41	320	3.13	2.82	1	97.1	1	1	0	1	0														
25	Male	26	CAEBV-T/NK	2760000	2.79	1.52	0.77	112	157	14	18	443	1.72	3.37	1	313.80	1	1	1	0	0	AP3B1 2081G>A, S694N						AP3B1 2081G>A, S694N							
26	Female	28.7	CAEBV-T/NK	3250000	1.52	0.51	0.88	106	83	136	220	617	2.74	2.43	1	796.70	1	1	1	0	0														
27	Male	20	CAEBV-T/NK	2730000	6.90	3.66	2.76	137	214	33	24	245	2.77	3.83	1	89.2	1	0	0	0	0														
28	Male	34.0	CAEBV-T/NK	139000	5.97	3.58	1.49	141	254	20	17	181	2.21	2.82	1	377.80	0	0	1	0	0														
29	Female	3	CAEBV-T/NK	10400000	8.25	3.90	2.85	97	372	13	16	311	1.26	3.40	1	57.3	1	1	0	1	0	ITK 1741C>T, R581W			ITK 1741C>T, R581W										
30	Male	12.0	CAEBV-T/NK	1090000	5.71	3.63	1.41	101	112	10	18	274	2.14	2.53	1	66.30	1	0	1	0	0														
31	Female	60	CAEBV-T/NK	10800000	4.05	2.71	0.96	125	190	35	42	340	1.26	2.73	1	311.1	1	0	0	1	0														
32	Female	21	CAEBV-T/NK	865000	3.64	2.42	0.82	117	281	17	23	240	1.47	3.32	1	122.80	1	0	1	1	0	RAB27A 11G>T, G4V								RAB27A 11G>T, G4V					
33	Female	30	CAEBV-T/NK	1e7	2.77	1.40	1.08	98	218	64	76	242	2.34	2.11	1	149.60	1	1	1	0	0														
34	Female	34.0	CAEBV-T/NK	439000	2.44	1.15	0.97	112	111	73	104	315		1.38	1	92.4	0	1	1	0	0														
35	Male	60.0	CAEBV-T/NK	167000	5.12	3.29	1.28	135	126	11	18	471	3.32	4.03	1	169.50	0	1	1	0	0	LYST 8368A>C, K2790Q	LYST 8368A>C, K2790Q												
36	Male	37.0	CAEBV-T/NK	10000000	7.51	4.36	2.18	129	106	45	24	379	3.26	5.48	1	396.7	1	1	1	1	0	ITK 1283A>G, Q428R			ITK 1283A>G, Q428R										
37	Male	16	CAEBV-T/NK	2760000	5.44	2.93	1.43	159	190	20	28	235	1.7	3.85	1	282.20	1	0	1	1	0														
38	Male	24	CAEBV-T/NK	12000000	4.35	3.28	0.72	115	56	40	156	679	1.79	3.16	1	603.9	1	1	1	0	0	ITK 1741C>T, R581W			ITK 1741C>T, R581W										
39	Female	26	CAEBV-T/NK	9450000	9.61	7.07	1.74	115	156	42	17	283	4.80	1.86	1	441.00	1	1	1	0	0	GZMB 630C>A, N210K							GZMB 630C>A, N210K						
40	Male	49	CAEBV-T/NK	357000	2.23	0.91	1.05	134	267	47	57	414	3.11	1.98	1	362.20	1	1	1	0	1	LYST 2150A>G, N717S; UNC13D 1607G>T, R536L	LYST 2150A>G, N717S	UNC13D 1607G>T, R536L											
41	Female	17.9	CAEBV-T/NK	7200000	3.29	2	0.97	110	211	421	193	289	2.09	3.06	1	210.80	1	0	1	0	0	UNC13D 2588G>A, G863D		UNC13D 2588G>A, G863D											
42	Male	56.5	CAEBV-T/NK	116000	5.43	3.63	0.91	94	432	51	47	186	1.96	7.7	1	1804.10	1	0	1	0	0	UNC13D 2800G>A, A934T		UNC13D 2800G>A, A934T											
43	Male	54	CAEBV-T/NK	4040	4.52	2.22	1.85	140	150	14	17	514	2.42	2.36	1	1201	1	0	0	0	0	UNC13D 2756C>T, S919F		UNC13D 2756C>T, S919F											
44	Female	24	CAEBV-T/NK	1520000	5.83	5.05	0.51	118	202	425	287	266	0.91	3.74	1	112.00	1	1	1	0	0	LYST 8368A>C, K2790Q; UNC13D 2240G>A, S747N	LYST 8368A>C, K2790Q	UNC13D 2240G>A, S747N											
45	Male	44.0	CAEBV-T/NK	151000	6.57	4.18	1.67	144	227	12	17	160	2.43	2.84	1	206	0	0	1	0	0														
46	Female	36	CAEBV-T/NK	45000	10.81	5.37	4.11	129	387	22	19	183	1.76	2.68	1	155.70	1	0	1	0	0	UNC13D 3067C>T, R1023C		UNC13D 3067C>T, R1023C											
47	Male	66.1	CAEBV-T/NK	2680000	5.74	3.52	1.55	142	284	19	18	173	0.88	3.77	1	236.6	0	0	0	0	0	PRF1 674G>A, R225Q				PRF1 674G>A, R225Q									
48	Female	30.4	CAEBV-T/NK	4440000	1.43	0.8	0.5	88	192	56	58	290	2.21	2.63	1	506.30	1	1	0	1	0	UNC13D 118-308C>T		UNC13D 118-308C>T											
49	Male	16.6	CAEBV-T/NK	45400000	2.74	1.38	0.86	112	84	22	32	282	0.95	2.55	1	145.70	0	1	1	0	0														
50	Male	20.9	CAEBV-T/NK	13000000	7.78	2.78	4.4	149	139	54	36	189	1.13	2.2	1	283.70	1	1	1	1	0	UNC13D 1189G>A, A397T; UNC13D 2588G>A, G863D		UNC13D 1189G>A, A397T											
51	Male	0.8	EBV-HLH	10100	1.18	0.31	0.62	117	205	2246.5	6653.9	796	2.34	0.56	1	2565.00	1	1	0	0	1	AP3B1 2081G>A,S694N; GZMB 187G>C, A63P						AP3B1 2081G>A, S694N	GZMB 187G>C, A63P						
52	Male	1.4	EBV-HLH	49190000	3.36	1.15	1.58	80	47	470	745	6122	3.88	0.6	1	82690.00	1	1	0	0	1														
53	Female	14.1	EBV-HLH	13000000	1.2	0.16	0.69	70	29	265	745	1818	4.51	0.81	1	8000.00	1	1	1	0	0	AP3B1 2012C>T, S671F						AP3B1 2012C>T, S671F							
54	Female	47.8	EBV-HLH	7660000	1.37	0.84	0.36	65	21	15	60	997	1.93	0.91	1	6008.80	1	1	1	0	1	LYST 8368A>C, K2790Q	LYST 8368A>C, K2790Q												
55	Female	4.7	EBV-HLH	137000	0.87	0.51	0.23	91	21	249	525	1867	2.01	1.54	1	9000.00	1	1	0	0	1														
56	Female	25.2	EBV-HLH	357000	1.51	0.9	0.53	81	83	14	37	690	3.72	2.94	1	8000.00	1	1	0	0	1														
57	Male	3.5	EBV-HLH	116000	1.44	0.47	0.83	83	109	26	63	998	2.15	2.22	1	1116.70	1	1	1	0	0														
58	Male	62.4	EBV-HLH	3830000	2.09	1.71	0.26	86	53	79	109	1867	1.96	2.05	1	8000.00	1	1	1	0	1	RAB27A 560G>A, R187Q								RAB27A 560G>A, R187Q					
59	Female	45.9	EBV-HLH	11700000	2.57	1.53	0.49	66	22	21	12	78	3.58	4.14	1	3851.8	1	1	0	0	1	LYST 1975T>C, C659R	LYST 1975T>C, C659R												
60	Male	19	EBV-HLH	9450000	3.66	1.92	0.91	91	35	25	27	366	2.35	1.78	1	1065	1	1	0	0	1														
61	Male	32.8	EBV-HLH	1520000	0.09	0.04	0.02	88	28	67	57	1073	3.29	2.14	1	5830.30	1	0	0	0	1														
62	Male	2.1	EBV-HLH	9700000	9.69	1.07	7.26	95	102	109	96	864	1.35	1.94	1	2827.3	1	1	0	1	1	RAB27A 523G>A, E175K; STX11 91G>A, E31K								RAB27A 523G>A, E175K		STX11 91G>A, E31K			
63	Male	45.2	EBV-HLH	4040	7.67	4.1	2.41	112	61	57	86	1867	2.21	0.86	1	3606.50	1	1	1	0	1	ITK 176G>A, R59Q			ITK 176G>A, R59Q										
64	Male	28.5	EBV-HLH	10400000	2.82	0.78	1.98	106	14	63	153	1209	2.88	0.51	1	4573.60	1	1	1	0	1														
65	Female	21	EBV-HLH	2680000	1.12	0.85	0.19	100	56	233	245	938	2.84	1.65	1	1805	1	1	0	0	0	LYST 4952G>C, C1651S	LYST 4952G>C, C1651S												
66	Male	32	EBV-HLH	10000000	1.33	0.93	0.23	76	59	57	98	245	3.84	1.58	1	2477.3	1	1	0	0	0	GZMB 358C>T, R120W							GZMB 358C>T, R120W						
67	Male	2.3	EBV-HLH	127000	1.73	0.19	1.45	85	52	94	163	595	1.27	1.54	1	1810.8	1	0	0	0	0														
68	Male	4.7	EBV-HLH	6800000	2.69	1.10	0.88	93	230	22	51	386	1.74	0.93	1	24412	1	0	0	0	1	LYST 7661A>G, Q2554R	LYST 7661A>G, Q2554R												
69	Male	55	EBV-HLH	17800000	7.36	6.63	0.33	103	39	198	244	1143	1.29	1.94	1	8869.2	1	0	0	0	1														
70	Female	28.1	EBV-HLH	11300000	4.61	3.01	0.77	71	212	52	225	822	5.94	0.67	1	17560	1	1	0	0	0	LYST 5549A>T, H1850L; UNC13D 1545T>G, N515K; PRF1 10C>T, R4C	LYST 5549A>T, H1850L	UNC13D 1545T>G, N515K		PRF1 10C>T, R4C									
71	Male	42	EBV-HLH	1750000	1.37	0.78	0.44	127	59	31	30	365	2.00	2.83	1	1264.5	1	1	0	0	1														
72	Female	23	EBV-HLH	43000	2.82	1.21	0.79	88	133	13	15	207	3.24	1.89	1	795.2	1	1	0	0	0														
73	Male	17.9	EBV-HLH	10000000	1.02	0.26	0.56	59	47	66	71	570	5.21	0.36	1	15233.00	1	1	0	0	1	LYST 7661A>G, Q2554R; PRF1 65del, P22fs; PRF1 147C>A, D49E	LYST 7661A>G, Q2554R			PRF1 65del, P22fs; PRF1 147C>A, D49E									
74	Male	17.6	EBV-HLH	4440000	1.21	0.94	0.18	108	6	158	561	4780	3.35	2.27	1	66981.00	1	1	0	1	0														
75	Female	29	EBV-HLH	59300	1.9	1.56	0.11	94	152	73	27	237	2.17	1.21	1	1090.7	1	1	1	1	0	LYST 830A>T, H277L; STXBP2 592A>C, T198P	LYST 830A>T, H277L				STXBP2 592A>C, T198P								
76	Female	12.6	EBV-HLH	13000000	6.01	3.95	1.7	110	101	57	24	217	3.56	0.85	1	185.80	1	0	0	0	1	ITK 1741C>T, R581W			ITK 1741C>T, R581W										
77	Male	37	EBV-HLH	320000	1.53	1.11	0.40	98	156	46	19	158	2.21	2.07	1	3405.4	1	0	0	0	1														
78	Female	73	EBV-HLH	429000	2.65	2.24	0.29	69	31	10	24	335	1.78	3.02	1	2046	1	1	0	0	0														
79	Male	24	EBV-HLH	7000000	2.89	0.92	1.66	102	133	125	117	979	2.19	1.67	1	3312	1	0	0	0	1														
80	Male	24	EBV-HLH	10000000	2.58	0.79	1.52	93	158	357	1270	1867	11.42	1.57	1	>50000	1	0	0	0	1														
81	Female	24	EBV-HLH	2750000	0.9	0.62	0.13	73	38	21	81	1512	2.19	2.91	1	7664	1	1	0	0	0	LYST 8368A>C, K2790Q	LYST 8368A>C, K2790Q												
82	Male	55.0	EBV-HLH	106000	2.31	1.53	0.57	100	79	57	103	716	3.72	1.82	1	>50000	1	1	0	0	1														
83	Female	38.3	EBV-HLH	652000	3.07	0.41	2.46	102	50	91	88	652	2.69	1.41	1	14488.00	1	1	0	0	1	LYST 8368A>C, K2790Q; UNC13D 2588G>A, G863D	LYST 8368A>C, K2790Q	UNC13D 2588G>A, G863D											
84	Female	7	EBV-HLH	87900	4.37	0.52	3.63	109	51	101	121	901	2.41	2.69	1	662.2	1	1	0	0	0	STXBP2 953C>T, T318M					STXBP2 953C>T, T318M								
85	Female	18.5	EBV-HLH	865000	1.27	0.62	0.58	129	37	200	215	772	3.55	3.87	1	6108.00	1	0	0	0	1	LYST 452A>G, H151R; UNC13D 2756C>T, S919F	LYST 452A>G, H151R	UNC13D 2756C>T, S919F											
86	Male	16	EBV-HLH	1690000	2.33	1.16	0.79	101	433	92	35	208	2.32	2.46	1	951.2	1	0	1	0	1														
87	Male	34	EBV-HLH	10200000	1.21	0.41	0.66	43	36	97	122	626	4.42	0.71	1	4722.20	1	1	0	0	1	UNC13D 2588G>A, G863D		UNC13D 2588G>A, G863D											
88	Male	30.6	EBV-HLH	2350000	4.55	4.02	0.44	121	186	274	165	651	1.73	3.03	1	16008.00	1	0	0	0	0	UNC13D 2677C>T, R893W; RAB27A 11G>T, G4V		UNC13D 2677C>T, R893W						RAB27A 11G>T, G4V					
89	Male	21.5	EBV-HLH	9800000	3.22	2.09	0.92	93	29	134	295	1608	4.32	1.25	1	8000.00	1	1	0	0	0	UNC13D 2897G>A, R966Q		UNC13D 2897G>A, R966Q											
90	Male	68.4	EBV-HLH	45000	0.39	0.03	0.27	73	13	36	60	594	1.27	1.4	1	32810.00	1		0	0	0	UNC13D 2972C>T, P991L		UNC13D 2972C>T, P991L											
91	Male	1.4	EBV-HLH	5620000	0.85	0.01	0.81	95	2	20	17	130	2.26	3.69	1	998.70	1	0	0	1	1	UNC13D 3134C>T, T1045M		UNC13D 3134C>T, T1045M											
92	Male	60	EBV-HLH	12000000	0.7	0.49	0.2	56	71	279	288	1379	3.56	1.29	1	23002	1	1	0	0	1	UNC13D 3229_3235delCGGGCCA, R1077fs		UNC13D 3229_3235delCGGGCCA, R1077fs											
93	Male	2	EBV-HLH	2760000	2.91	1.47	1.01	72	49	15	22	331	4.21	1.65	1	13378	1	0	0	0	1	UNC13D 2219C>T, T740M		UNC13D 2219C>T, T740M											
94	Female	44	EBV-HLH	737000	5.64	4.18	1.32	96	183	67	34	176	1.27	2.39	0	71.2	1	1	0	0	1	UNC13D 3067C>T, R1023C; UNC13D 2764G>A, A922T		UNC13D 3067C>T, R1023C; UNC13D 2764G>A, A922T											
95	Female	4.7	EBV-HLH	500000000	9.52	5.88	2.35	160	13	164	475	613	1.23	2.03	0	7316.00	1	1	0	0	1	LYST 8368A>C, K2790Q; UNC13D 1232G>A, R411Q	LYST 8368A>C, K2790Q	UNC13D 1232G>A, R411Q											
96	Male	29	EBV-HLH	187000	3.06	1.63	1.19	92	29	71	35	168	1.67	1.24	1	1367.2	1	1	0	0	1	UNC13D 2588G>A, G863D; SRGN 233C>T, T78M; UNC13D 1978_1979insATTACCG, V660fs		UNC13D 2588G>A, G863D; UNC13D 1978_1979insATTACCG, V660fs											SRGN 233C>T, T78M
97	Male	83	EBV-HLH	7200000	1.22	1,05	0.12	39	6	<5	17	333	1.52	1.17	1	1819.60	1	1	0	0	1	UNC13D 1201_1202delinsAA, S401N		UNC13D 1201_1202delinsAA, S401N											
98	Female	25.3	EBV-HLH	59200	1.66	0.95	0.57	84	32	154	253	1225	4.67	0.35	1	3481.60	1	1	1	0	0	PRF1 916G>A, G306S; PRF1 65del, P22fs				PRF1 916G>A, G306S; PRF1 65del, P22fs									
99	Male	52	EBV-HLH	175000	9.59	6.44	2.06	76	58	11	11	433	1.885	5.09	1	1870.6	1	0	1	0	1	STXBP2 358C>T, R120C					STXBP2 358C>T, R120C								
100	Female	52	EBV-HLH	167000	4.76	3.81	0.89	115	89	44	14	431	3.41	2.09	1	1579.3	1	1	0	0	1														
101	Male	59	EBV-HLH	6210000	2.44	1.46	0.52	116	35	21	33	330	2.16	2.10	1	1726.00	1	1	0	0	1														
102	Male	19.3	ENKTL	12300000	12.47	8.51	3.45	145	139	16	16	279	1.9	2	1	95.00	1	1	1	0	0	LYST 4578T>A, N1526K	LYST 4578T>A, N1526K												
103	Female	30.7	ENKTL	8020000	1.7	0.96	0.37	103	142	17	26	449	3.27	3.04	1	486.00	1	0	1	1	0														
104	Female	19.6	ENKTL	361000	2.96	1.38	1.23	121	137	35	73	718	2.07	2.63	1	88.6	1	1	0	1	0														
105	Male	49	ENKTL	557000	8.81	5.42	2.21	133	297	11	16	192	2.21	2.54	1	322.70	1	1	0	0	0	LYST 8368A>C, K2790Q	LYST 8368A>C, K2790Q												
106	Female	31.0	ENKTL	6890	6.73	4.12	1.95	121	209	<5	21	188	1.98	2.79	1	167.2	0	0	0	0	0														
107	Male	28.4	ENKTL	1390	3.62	2.45	0.82	124	236	14	16	165	1.32	3.08	1	223.2	0	0	0	0	0														
108	Female	53.3	ENKTL	197000	4.64	2.65	1.34	115	162	15	18	189	1.71	3.09	1	150.1	0	0	1	0	0														
109	Male	28.0	ENKTL	8800000	3.78	2.12	1.43	122	197	49	109	248		1.46	1	533	0	0	0	0	0														
110	Male	72.0	ENKTL	57800	4.81	2.66	1.62	120	192	<5	10	192	1.21	4.23	1	167.5	0	0	0	0	0	GZMB 737G>A, R246H; GNLY 280C>T, R94W							GZMB 737G>A, R246H				GNLY 280C>T, R94W		
111	Female	27	ENKTL	4120	3.63	2.86	0.34	106	141	64	41	305	3.14	3.07	1	145.2	1	0	0	1	0														
112	Male	45.0	ENKTL	698	8.94	6.03	1.84	144	389	92	43	274	1.13	3.63	1	247.8	0	0	0	0	0														
113	Male	39.9	ENKTL	15100	12.39	9.24	1.24	146	170	76	21	172	0.99	3.3	1	89.4	0	0	0	0	0														
114	Male	62.0	ENKTL	796	5.93	4.69	0.84	112	102	11	6	219	2.12	5.31	1	94	0	0	0	0	0	LYST 8368A>C, K2790Q	LYST 8368A>C, K2790Q												
115	Male	26.0	ENKTL	355	8.25	5.73	1.52	132	217	14	21	268	3.32	2.33	1	312.7	0	0	0	0	0														
116	Male	27	ENKTL	32200	10.36	9.31	0.66	104	323	24	46	538	2.01	1.26	1	5756.60	1	1	1	1	0	PRF1 500A>G, Y167C				PRF1 500A>G, Y167C									
117	Female	32	ENKTL	44500000	5.32	4.36	0.46	104	166	26	36	342		1.65	1	170.4	1	1	1	0	0														
118	Male	26.0	ENKTL	7900000	4.7	3.9	0.56	81	20	98	95	667		0.5	1	5471.2	0	1	0	0	1														
119	Male	35.0	ENKTL	567	3.19	1.23	1.15	128	201	11	19	181	0.88	2.32	1	332.8	0	0	0	0	0														
120	Male	19	ENKTL	6060	3.02	1.41	1.26	128	284	25	17	269	2.14	4.09	1	358.50	1	1	0	0	0														
121	Male	19	ENKTL	232000	5.07	2.98	1.22	128	184	25	33	248	1.66	3	1	75.60	1	1	1	0	0	UNC13D 3229_3235del, R1077fs; AP3B1 1022G>A, R341H		UNC13D 3229_3235del, R1077fs				AP3B1 1022G>A, R341H							
122	Female	73.0	ENKTL	530000	7.73	3.48	2.02	107	268	17	36	219	4.21	2.21	1	125.70	0	0	1	1	0	AP3B1 914A>G, K305R						AP3B1 914A>G, K305R							
123	Female	52	ENKTL	18100	11.94	9.97	1.18	94	107	17	19	352	1.04	4.49	1	2427.80	1	1	0	0	0	UNC13D 1280G>A, R427Q		UNC13D 1280G>A, R427Q											
124	Female	13.7	ENKTL	10000000	3.38	1.66	1.47	78	82	145	180	1195	1.5	0.5	1	983.00	1	1	1	1	0														
125	Female	45.1	ENKTL	19500000	2.82	1.57	0.96	80	102	91	33	207	5.12	0.84	1	300.00	1		1	0	0	PRF1 674G>A, R225Q				PRF1 674G>A, R225Q									
126	Male	10.9	ENKTL	1480000	8.66	7.96	0.4	100	175	18	70	1240	1.21	2.59	1	1107.10	1	1	1	0	0														
127	Male	18.5	ENKTL	910000	5.49	2.19	2.7	143	261	200	71	162	1.06	3.07	1	321	1	1	1	0	0														
128	Male	30.1	ENKTL	60700	11	10.14	0.52	92	109	16	30	290	0.99	1.28	1	482.30	1	0	0	0	0	STXBP2 1663A>G, R555G					STXBP2 1663A>G, R555G								
129	Male	32	ENKTL	530000	1.77	1.45	0.18	111	86	15	25	370	1.7	3.54	1	651.10	1	1	0	0	0														
130	Female	28	ANKL	45500	5.1	4.27	0.61	135	102	45	72	887	2.6	0.87	1	17226	1	0	0	0	0	LYST 5275G>A, V1759M	LYST 5275G>A, V1759M												
131	Male	25.9	ANKL	17900000	0.9	0.7	0.14	99	8	41	40	422	2.59	2.48	1	882.20	1	1	1	0	0														
132	Female	15.4	ANKL	141000	1.43	0.64	0.74	99	68	218	144	1125	2.08	1.59	1	4059.30	1	1	0	0	1														
133	Male	22.7	ANKL	10400000	3.37	2.94	0.34	113	85	48	63	402	2.78	2.8	1	422.50	1	1	0	0	1														
134	Male	24.3	ANKL	20200000	5.28	4.4	0.53	101	36	84	134	1514	2.6	0.55	1	20204.00	1	0	0	0	0														
135	Male	26.4	ANKL	17200000	11.23	2.91	8.02	121	118	475	707	2054	4.66	1.03	1	17686.00	1	1	0	0	0														
136	Female	48	ANKL	34100	0.68	0.31	0.24	78	15	44	49	701	3.15	1.11	1	1897.60	1	1	0	0	1														
137	Male	68.6	ANKL	3280000	2	0.64	1.19	107	40	23	75	594	1.48	0.64	1	3515.8	1	1	0	0	1														
138	Male	27	ANKL	941000	1.34	1.01	0.28	120	104	1326	2589	1867	2.11	3.23	1	979.4	1	1	0	0	0														
139	Female	58.5	ANKL	7950000	1.8	1.42	0.21	81	26	31	21	339	1.81	1.49	1	864.6	1	0	0	0	1														
140	Male	20	ANKL	350000	2.41	1.67	0.41	122	98	35	43	1433	2.83	1.45	1	8987.2	1	0	0	0	0														
141	Male	22.7	ANKL	5340000	2	1.6	0.36	98	30	181	705	1867	3.35	1.27	1	8000.00	1	1	0	0	0														
142	Male	51	ANKL	4020000	4.49	3.44	0.8	102	34	110	122	1773	5.9	1.9	1	43244.00	1	1	0	0	0														
143	Female	24	ANKL	7610000	1.53	0.98	0.42	99	35	19	50	694	4.64	1.46	1	480.50	1	1	1	0	1	STXBP2 953C>T, T318M					STXBP2 953C>T, T318M								
144	Male	31	ANKL	3600000	2.45	1.97	1.67	56	102	83	99	583	1.34	4.03	1	489.2	1	1	0	0	1														
145	Male	30	ANKL	560000	0.67	0.31	0.34	84	32	102	311	517	2.34	1.80	1	14500	1	1	0	0	1														
146	Male	22.9	ANKL	2100000	1.17	0.74	0.3	80	37	34	36	776	0.95	1.3	1	1256.6	1	0	0	0	1	UNC13D 3229_3235delCGGGCCA, R1077fsSTXBP2 1663A>G, R555G		UNC13D 3229_3235delCGGGCCA, R1077fs			STXBP2 1663A>G, R555G								
147	Male	25	ANKL	15400000	2.89	1.88	0.82	124	59	44	47	1344	2.42	2.11	1	1882	1	1	0	0	1														
148	Male	26	ANKL	37000	1.49	1.02	1.43	117	35	15	31	255	1.63	3.26	1	1256.5	1	1	1	0	0	LYST 1109A>G, Q370R	LYST 1109A>G, Q370R												
149	Male	22	ANKL	7500000	1.24	0.81	0.93	79	29	68	87	1624	2.36	3	1	7856.2	1	1	0	0	1														
150	Male	19	ANKL	488000	4.24	3.45	1.75	114	89	23	25	1126	2.51	3.32	1	14700	1	1	0	0	1														
151	Male	45	ANKL	83000	1.03	0.67	1.24	110	15	41	40	179	3.32	3.42	1	1792.4	1	0	0	0	0	LYST 10833T>A, H3611Q	LYST 10833T>A, H3611Q												
152	Female	18	ANKL	13500000	0.88	0.67	1.67	102	19	37	38	684	1.04	2.79	1	3120	1	1	0	0	1														
153	Male	28	ANKL	4300000	0.72	0.5	0.96	58	36	193	342	1057	2.52	2.61	1	5255	1	1	0	0	1														
154	Male	16	ANKL	7900000	2.13	1.56	2.85	121	41	25	23	788	1.9	2.84	1	10200	1	1	1	0	0	PRF1 272C>T, A91V				PRF1 272C>T, A91V									
155	Male	25	ANKL	946000	5.79	3.71	4.4	108	154	86	135	1945	2.53	2.73	1	1522.2	1	1	0	0	1														
156	Male	63	ANKL	11300000	1.57	1.10	0.53	125	73	46	78	806	2.54	2.2	1	3577.8	1	1	0	0	1														
157	Male	17	ANKL	149000	0.45	0.1	0.82	90	46	41	54	415	0.95	2.82	1	2200	1	1	0	0	0	STXBP2 1459G>A, V487M					STXBP2 1459G>A, V487M								
158	Male	36	ANKL	9800000	5.21	4.20	0.34	113	75	142	169	622	0.99	2.82	1	1346	1	1	0	0	1														
159	Male	57	ANKL	25600000	1.12	0.71	1.15	87	11	456	870	854	2.22	2.87	1	8120	1	1	0	0	1														
160	Male	24	ANKL	177000	0.5	0.21	0.22	58	6	23	22	120	3.54	1.41	1	3102.4	1	0	0	0	1	LYST 1541G>A, R514Q	LYST 1541G>A, R514Q												
161	Male	21	ANKL	6700	3.47	2.76	0.18	112	71	106	154	2244	2.09	2.32	1	1734.6	1	1	0	0	1														
162	Female	71	ANKL	481000	2.53	1.52	0.57	117	71	43	74	998	6.85	3.78	1	3640.6	1	0	0	0	1	LYST 10526G>A, R3509Q	LYST 10526G>A, R3509Q												
163	Male	23.6	ANKL	6520000	2.61	1.4	0.6	80	30	66	186	1867	3.95	1.01	1	12066.00	1	1	0	0	0	LYST 8368A>C, K2790Q; PRF1 10C>T, R4C	LYST 8368A>C, K2790Q			PRF1 10C>T, R4C									
164	Male	16	ANKL	6700000	1.36	0.91	0.51	49	154	17	19	722	2.11	3.07	1	577	1	0	1	0	0	ITK 1741C>T, R581W			ITK 1741C>T, R581W										
165	Female	21	ANKL	12000000	1.18	0.68	0.31	81	56	37	39	228	2.83	0.56	1	14668.0	1	0	1	0	0	UNC13D 2179C>T, R727W		UNC13D 2179C>T, R727W											
166	Male	55	ANKL	721000	1.22	0.99	4.61	94	39	33	31	1756	1.06	3.54	1	35122	1	1	0	0	1														
167	Male	42	ANKL	7900	0.56	0.22	1.55	94	33	65	112	506	1.65	3.23	1	1225.5	1	1	0	0	0														
168	Male	18	ANKL	10000000	0.9	0.65	1.67	63	31	952	1340	2651	2.11	3.42	1	553.4	1	0	0	0	0														
169	Female	17	ANKL	6700000	6.32	4.25	0.34	103	122	92	122	1924	1.65	2.59	1	3346.2	1	0	0	0	0														
170	Female	19	ANKL	452000	1.35	1.06	0.82	96	30	35	65	931	1.63	2.25	1	2578.8	1	0	0	0	1														
171	Male	21	ANKL	155000	0.34	0.1	0.21	54	30	40	34	372	2.24	2.96	1	2679.3	1	1	0	0	0														
172	Male	34	ANKL	10000000	1.26	0.67	0.28	115	73	48	44	313	2.74	1.56	1	1236.60	1	1	0	0	0														
173	Female	28	ANKL	7270000	2.74	1.77	0.68	92	78	122	139	459	2.05	1.54	1	749.3	1	1	1	0	1														
174	Female	20.9	ANKL	21100000	5.82	2.71	2.06	102	55	744	1409	819	4.01	1.05	1	3050.70	1	1	0	0	1														
175	Male	26.4	ANKL	5790000	1.28	1.02	0.14	122	74	85	202	480	2.74	0.78	1	2532.40	0	1	0	0	1														
176	Female	23	ANKL	2500000	1.43	0.87	1.84	105	23	105	121	388	1.06	3.07	1	3467.3	1	1	1	0	0	LYST 8368A>C, K2790Q	LYST 8368A>C, K2790Q												
177	Male	52.9	ANKL	95400000	1.32	0.97	0.19	110	32	109	362	2951	2.18	1.51	1	50000.00	1	1	1	0	1														

Table S2 Identification of 94 genetic variants with allele frequency less than 0.01 according to the gnomAD

Chr	Start	End	Ref	Alt	Gene_symbol	Referenced coding sequence	Effect_refGene	Mutation_type	AA_change_refGene	gnomAD_exome_ALL	ExAC_ALL	ClinVar	COSMIC
chr5	77471628	77471628	T	C	*AP3B1*	NM_003664.4	Nonsynonymous	SNV	*AP3B1*: NM_001271769:exon10:c.928A>G:p.T310A AP3B1:NM_003664:exon10:c.1075A>G:p.T359A	2.85E–05	3.297e–05	–	–
chr5	77458772	77458772	A	G	*AP3B1*	NM_003664.4	Nonsynonymous	SNV	*AP3B1*: NM_001271769:exon13:c.1087T>C:p.Y363H AP3B1:NM_003664:exon13:c.1234T>C:p.Y412H	4.07E–06	8.245e–06	–	–
chr5	77409744	77409744	C	T	*AP3B1*	NM_003664.4	Nonsynonymous	SNV	*AP3B1*: NM_001271769:exon19:c.1934G>A:p.S645N AP3B1:NM_003664:exon19:c.2081G>A:p.S694N	1.222e–05	–	–	–
chr5	77409666	77409666	G	A	*AP3B1*	NM_003664.4	Nonsynonymous	SNV	*AP3B1*: NM_001271769:exon19:c.2012C>T:p.S671F AP3B1:NM_003664:exon19:c.2159C>T:p.S720F	–	–	–	–
chr5	77298814	77298814	G	A	*AP3B1*	NM_003664.4	Nonsynonymous	SNV	*AP3B1*: NM_001271769:exon27:c.3050C>T:p.S1017F AP3B1:NM_003664:exon27:c.3197C>T:p.S1066F	5.687e–05	6.59e–05	–	–
chr5	77477359	77477359	T	C	*AP3B1*	NM_003664.4	Nonsynonymous	SNV	*AP3B1*: NM_001271769:exon8:c.767A>G:p.K256R AP3B1:NM_003664:exon8:c.914A>G:p.K305R	–	–	–	–
chr5	77473181	77473181	C	T	*AP3B1*	NM_003664.4	Nonsynonymous	SNV	*AP3B1*: NM_001271769:exon9:c.875G>A:p.R292H AP3B1:NM_003664:exon9:c.1022G>A:p.R341H	0.0002	0.0003	–	–
chr12	6560554	6560554	C	T	*CD27*	NM_001242.4	Nonsynonymous	SNV	*CD27*: NM_001242:exon6:c.779C>T:p.P260L	8.195e–06	1.674e–05	–	–
chr2	85924653	85924653	C	T	*GNLY*	NM_006433.3	Nonsynonymous	SNV	*GNLY*: NM_006433:exon4:c.280C>T:p.R94W GNLY:NM_001302758:exon5:c.361C>T:p.R121W GNLY:NM_012483:exon5:c.235C>T:p.R79W	1.219e–05	8.242e–06	–	ID=COSM1208428; OCCURENCE=1 (large_intestine)
chr2	85924746	85924746	G	A	*GNLY*	NM_006433.3	Nonsynonymous	SNV	*GNLY*: NM_006433:exon4:c.373G>A:p.G125R GNLY:NM_001302758:exon5:c.454G>A:p.G152R GNLY:NM_012483:exon5:c.328G>A:p.G110R	5.28e–05	4.946e–05	–	ID=COSM1208429; OCCURENCE=1 (large_intestine)
chr14	25101646	25101646	C	G	*GZMB*	NM_004131.5	Nonsynonymous	SNV	*GZMB*: NM_001346011:exon3:c.187G>C:p.G63R GZMB:NM_004131:exon3:c.223G>C:p.G75R	–	–	–	–
chr14	25101306	25101306	G	A	*GZMB*	NM_004131.5	Nonsynonymous	SNV	*GZMB*: NM_001346011:exon4:c.322C>T:p.R108W GZMB:NM_004131:exon4:c.358C>T:p.R120W	3.667e–05	3.302e–05	–	ID=COSM1369436, COSM1369435; OCCURENCE=1 (large_intestine)
chr14	25100355	25100355	G	T	*GZMB*	NM_004131.5	Nonsynonymous	SNV	*GZMB*: NM_001346011:exon5:c.630C>A:p.N210K GZMB:NM_004131:exon5:c.666C>A:p.N222K	4.062e–06	8.238e–06	–	–
chr14	25100284	25100284	C	T	*GZMB*	NM_004131.5	Nonsynonymous	SNV	*GZMB*: NM_001346011:exon5:c.701G>A:p.R234H GZMB:NM_004131:exon5:c.737G>A:p.R246H	2.032e–05	1.648e–05	–	–
chr5	156667172	156667172	C	G	*ITK*	NM_005546.3	Nonsynonymous	SNV	*ITK*: NM_005546:exon10:c.952C>G:p.L318V	8.129e–06	8.239e–06	–	–
chr5	156671322	156671322	A	G	*ITK*	NM_005546.3	Nonsynonymous	SNV	*ITK*: NM_005546:exon13:c.1283A>G:p.Q428R	–	–	–	–
chr5	156675967	156675967	C	T	*ITK*	NM_005546.3	Nonsynonymous	SNV	*ITK*: NM_005546:exon16:c.1741C>T:p.R581W	0.0009	0.0008	Accession: 352476 phneotype: Lymphoproliferative syndrome Clinical significance: Uncertain_significance	–
chr5	156635937	156635937	G	A	*ITK*	NM_005546.3	Nonsynonymous	SNV	*ITK*: NM_005546:exon2:c.176G>A:p.R59Q	1.219e–05	8.28e–06	–	ID=COSM175471; OCCURENCE=2 (large_intestine)
chr5	156641310	156641310	A	G	*ITK*	NM_005546.3	Nonsynonymous	SNV	*ITK*: NM_005546:exon4:c.434A>G:p.Q145R	2.439e–05	4.944e–05	–	–
chr1	235952116	235952116	T	C	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon13:c.4573A>G:p.I1525V LYST:NM_001301365:exon13:c.4573A>G:p.I1525V	8.146e–06	8.276e–06	–	–
chr1	235952111	235952111	A	T	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon13:c.4578T>A:p.N1526K LYST:NM_001301365:exon13:c.4578T>A:p.N1526K	0.0002	0.0002	–	–
chr1	235945298	235945298	C	G	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon15:c.4952G>C:p.C1651S LYST:NM_001301365:exon15:c.4952G>C:p.C1651S	2.035e–05	2.477e–05	–	–
chr1	235940548	235940548	C	T	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon17:c.5275G>A:p.V1759M LYST:NM_001301365:exon17:c.5275G>A:p.V1759M	–	–	–	–
chr1	235940508	235940508	T	C	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon17:c.5315A>G:p.Q1772R LYST:NM_001301365:exon17:c.5315A>G:p.Q1772R	2.44e–05	3.297e–05	–	–
chr1	235938298	235938298	T	A	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon18:c.5549A>T:p.H1850L LYST:NM_001301365:exon18:c.5549A>T:p.H1850L	1.632e–05	8.341e–06	–	–
chr1	235929436	235929436	G	C	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon21:c.6064C>G:p.P2022A LYST:NM_001301365:exon21:c.6064C>G:p.P2022A	–	–	–	–
chr1	235922378	235922378	C	T	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon23:c.6775G>A:p.V2259I LYST:NM_001301365:exon23:c.6775G>A:p.V2259I	6.113e–05	7.433e–05	–	–
chr1	235914629	235914629	T	C	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon28:c.7661A>G:p.Q2554R LYST:NM_001301365:exon28:c.7661A>G:p.Q2554R	2.032e–05	8.243e–06	–	–
chr1	235909698	235909698	G	A	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon29:c.7910C>T:p.T2637M LYST:NM_001301365:exon29:c.7910C>T:p.T2637M	5.688e–05	4.947e–05	–	–
chr1	235909637	235909637	T	A	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon29:c.7971A>T:p.Q2657H LYST:NM_001301365:exon29:c.7971A>T:p.Q2657H	–	–	–	–
chr1	235904729	235904729	G	C	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon31:c.8351C>G:p.S2784C LYST:NM_001301365:exon31:c.8351C>G:p.S2784C	8.147e–06	8.286e–06	–	–
chr1	235897950	235897950	T	G	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon32:c.8368A>C:p.K2790Q LYST:NM_001301365:exon32:c.8368A>C:p.K2790Q	0.0008	0.0008	Accession: 296373 phneotype: Chédiak–Higashi syndrome Clinical significance: Uncertain_significance	–
chr1	235860421	235860421	C	T	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon46:c.10526G>A:p.R3509Q LYST:NM_001301365:exon46:c.10526G>A:p.R3509Q	0.0002	0.0003	Accession: 296354 phneotype: Chédiak–Higashi syndrome Clinical significance: Uncertain_significance	–
chr1	235840887	235840887	A	T	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon49:c.10833T>A:p.H3611Q LYST:NM_001301365:exon49:c.10833T>A:p.H3611Q	–	–	–	–
chr1	235973009	235973009	T	C	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon5:c.1109A>G:p.Q370R LYST:NM_001301365:exon5:c.1109A>G:p.Q370R	–	–	–	–
chr1	235972878	235972878	A	G	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon5:c.1240T>C:p.C414R LYST:NM_001301365:exon5:c.1240T>C:p.C414R	3.257e–05	4.129e–05	–	–
chr1	235972577	235972577	C	T	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon5:c.1541G>A:p.R514Q LYST:NM_001301365:exon5:c.1541G>A:p.R514Q	6.511e–05	2.474e–05	–	–
chr1	235972460	235972460	C	A	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon5:c.1658G>T:p.C553F LYST:NM_001301365:exon5:c.1658G>T:p.C553F	–	–	–	–
chr1	235972143	235972143	A	G	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon5:c.1975T>C:p.C659R LYST:NM_001301365:exon5:c.1975T>C:p.C659R	3.671e–05	4.15e–05	–	–
chr1	235971968	235971968	T	C	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon5:c.2150A>G:p.N717S LYST:NM_001301365:exon5:c.2150A>G:p.N717S	0.0005	0.0004	–	–
chr1	235973666	235973666	T	C	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon5:c.452A>G:p.H151R LYST:NM_001301365:exon5:c.452A>G:p.H151R	0.0001	6.594e–05	–	ID=COSM1581212; OCCURENCE=1 (haematopoietic_and_lymphoid_tissue)
chr1	235973288	235973288	T	A	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon5:c.830A>T:p.H277L LYST:NM_001301365:exon5:c.830A>T:p.H277L	8.143e–06	–	–	–
chr1	235969333	235969333	T	C	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon6:c.3103A>G:p.K1035E LYST:NM_001301365:exon6:c.3103A>G:p.K1035E	–	–	–	–
chr1	235969329	235969329	T	G	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon6:c.3107A>C:p.E1036A LYST:NM_001301365:exon6:c.3107A>C:p.E1036A	5.691e–05	5.774e–05	–	–
chr1	235967903	235967903	A	C	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon7:c.3456T>G:p.I1152M LYST:NM_001301365:exon7:c.3456T>G:p.I1152M	–	–	–	–
chr1	235967880	235967880	A	G	*LYST*	NM_000081.3	Nonsynonymous	SNV	*LYST*: NM_000081:exon7:c.3479T>C:p.L1160S LYST:NM_001301365:exon7:c.3479T>C:p.L1160S	–	–	–	–
chr10	72360649	72360649	G	A	*PRF1*	NM_005041.4	Nonsynonymous	SNV	*PRF1*: NM_001083116:exon2:c.10C>T:p.R4C PRF1:NM_005041:exon2:c.10C>T:p.R4C	0.0009	0.0014	–	–
chr10	72360512	72360512	G	T	*PRF1*	NM_005041.4	Nonsynonymous	SNV	*PRF1*: NM_001083116:exon2:c.147C>A:p.D49E PRF1:NM_005041:exon2:c.147C>A:p.D49E	4.115e–06	8.551e–06	–	–
chr10	72360387	72360387	G	A	*PRF1*	NM_005041.4	Nonsynonymous	SNV	*PRF1*: NM_001083116:exon2:c.272C>T:p.A91V PRF1:NM_005041:exon2:c.272C>T:p.A91V	0.0294	0.0311	Accession: 13718 phneotype: Hemophagocytic lymphohistiocytosis, familial, 2, susceptibility to|Familial hemophagocytic lymphohistiocytosis|not provided Clinical significance: Conflicting_interpretations_of_pathogenicity	–
chr10	72360159	72360159	T	C	*PRF1*	NM_005041.4	Nonsynonymous	SNV	*PRF1*: NM_001083116:exon2:c.500A>G:p.Y167C PRF1:NM_005041:exon2:c.500A>G:p.Y167C	–	–	–	–
chr10	72360594	72360594	G	-	*PRF1*	NM_005041.4	Nonsynonymous	Frameshift deletion	*PRF1*: NM_001083116:exon2:c.65delC:p.P22fs PRF1:NM_005041:exon2:c.65delC:p.P22fs	1.271e–05	1.08e–05	–	–
chr10	72358407	72358407	C	T	*PRF1*	NM_005041.4	Nonsynonymous	SNV	*PRF1*: NM_001083116:exon3:c.1070G>A:p.R357Q PRF1:NM_005041:exon3:c.1070G>A:p.R357Q	0.0004	0.0004	–	–
chr10	72358324	72358324	G	A	*PRF1*	NM_005041.4	Nonsynonymous	SNV	*PRF1*: NM_001083116:exon3:c.1153C>T:p.R385W PRF1:NM_005041:exon3:c.1153C>T:p.R385W	0.0002	0.0002	–	ID=COSM42670; OCCURENCE=2 (large_intestine), 1 (breast), 1 (endometrium), 1 (central_nervous_system)
chr10	72358803	72358803	C	T	*PRF1*	NM_005041.4	Nonsynonymous	SNV	*PRF1*: NM_001083116:exon3:c.674G>A:p.R225Q PRF1:NM_005041:exon3:c.674G>A:p.R225Q	0.0002	0.0001	–	–
chr10	72358588	72358588	G	A	*PRF1*	NM_005041.4	Nonsynonymous	SNV	*PRF1*: NM_001083116:exon3:c.889C>T:p.R297W PRF1:NM_005041:exon3:c.889C>T:p.R297W	8.123e–06	8.237e–06	–	–
chr10	72358561	72358561	C	T	*PRF1*	NM_005041.4	Nonsynonymous	SNV	*PRF1*: NM_001083116:exon3:c.916G>A:p.G306S PRF1:NM_005041:exon3:c.916G>A:p.G306S	2.437e–05	2.472e–05	–	–
chr15	55527122	55527122	C	A	*RAB27A*	NM_183236.2	Nonsynonymous	SNV	*RAB27A*: NM_004580:exon2:c.11G>T:p.G4V RAB27A:NM_183234:exon3:c.11G>T:p.G4V RAB27A:NM_183235:exon3:c.11G>T:p.G4V RAB27A:NM_183236:exon3:c.11G>T:p.G4V	3.278e–05	7.4e–05	–	–
chr15	55520896	55520896	G	A	*RAB27A*	NM_183236.2	Nonsynonymous	SNV	*RAB27A*: NM_004580:exon4:c.254C>T:p.T85M RAB27A:NM_183234:exon5:c.254C>T:p.T85M RAB27A:NM_183235:exon5:c.254C>T:p.T85M RAB27A:NM_183236:exon5:c.254C>T:p.T85M	2.034e–05	2.483e–05	–	ID=COSM963022; OCCURENCE=1 (endometrium)
chr15	55516177	55516177	G	A	*RAB27A*	NM_183236.2	Nonsynonymous	SNV	*RAB27A*: NM_004580:exon5:c.377C>T:p.P126L RAB27A:NM_183234:exon6:c.377C>T:p.P126L RAB27A:NM_183235:exon6:c.377C>T:p.P126L RAB27A:NM_183236:exon6:c.377C>T:p.P126L	–	–	–	–
chr15	55497848	55497848	C	T	*RAB27A*	NM_183236.2	Nonsynonymous	SNV	*RAB27A*: NM_004580:exon6:c.523G>A:p.E175K RAB27A:NM_183234:exon7:c.523G>A:p.E175K RAB27A:NM_183235:exon7:c.523G>A:p.E175K RAB27A:NM_183236:exon7:c.523G>A:p.E175K	–	–	–	–
chr15	55497811	55497811	C	T	*RAB27A*	NM_183236.2	Nonsynonymous	SNV	*RAB27A*: NM_004580:exon6:c.560G>A:p.R187Q RAB27A:NM_183234:exon7:c.560G>A:p.R187Q RAB27A:NM_183235:exon7:c.560G>A:p.R187Q RAB27A:NM_183236:exon7:c.560G>A:p.R187Q	0.0005	0.0004	Accession: 235346 phneotype: not provided Clinical significance: Uncertain_significance	–
chr10	70863632	70863632	C	T	*SRGN*	NM_002727.3	Nonsynonymous	SNV	*SRGN*: NM_001321054:exon2:c.65C>T:p.T22M SRGN:NM_002727:exon3:c.233C>T:p.T78M SRGN:NM_001321053:exon4:c.233C>T:p.T78M	3.661e–05	2.487e–05	–	–
chr6	144507855	144507855	G	A	*STX11*	NM_003764.3	Nonsynonymous	SNV	*STX11*: NM_003764:exon2:c.91G>A:p.E31K	4.063e–05	4.967e–05	–	–
chr19	7707702	7707702	C	T	*STXBP2*	NM_006949.3	Nonsynonymous	SNV	*STXBP2*: NM_001127396:exon11:c.944C>T:p.T315M STXBP2:NM_001272034:exon11:c.986C>T:p.T329M STXBP2:NM_006949:exon11:c.953C>T:p.T318M	0.0004	0.0004	–	–
chr19	7712054	7712054	G	A	*STXBP2*	NM_006949.3	Nonsynonymous	SNV	*STXBP2*: NM_001127396:exon17:c.1450G>A:p.V484M STXBP2:NM_001272034:exon17:c.1492G>A:p.V498M STXBP2:NM_006949:exon17:c.1459G>A:p.V487M	0.0004	0.0005	–	–
chr19	7712331	7712331	A	G	*STXBP2*	NM_006949.3	Nonsynonymous	SNV	*STXBP2*: NM_001127396:exon18:c.1621A>G:p.M541V STXBP2:NM_001272034:exon18:c.1663A>G:p.M555V STXBP2:NM_006949:exon18:c.1630A>G:p.M544V	4.116e–06	–	–	–
chr19	7705818	7705818	C	T	*STXBP2*	NM_006949.3	Nonsynonymous	SNV	*STXBP2*: NM_001127396:exon6:c.349C>T:p.R117C STXBP2:NM_001272034:exon6:c.391C>T:p.R131C STXBP2:NM_006949:exon6:c.358C>T:p.R120C	9.746e–05	0.0001	Accession: 286335 phneotype: not specified Clinical significance: Uncertain_significance	–
chr19	7706933	7706933	A	C	*STXBP2*	NM_006949.3	Nonsynonymous	SNV	*STXBP2*: NM_001127396:exon8:c.583A>C:p.T195P STXBP2:NM_001272034:exon8:c.625A>C:p.T209P STXBP2:NM_006949:exon8:c.592A>C:p.T198P	4.071e–06	8.334e–06	–	–
chr17	73832762	73832762	C	T	*UNC13D*	NM_199242.2	Nonsynonymous	SNV	*UNC13D*: NM_199242:exon14:c.1189G>A:p.A397T	5.7e–05	8.27e–05	–	–
chr17	73832750	73832751	CT	AA	*UNC13D*	NM_199242.2	Nonsynonymous	Nonframeshift substitution	*UNC13D*: NM_199242:exon14:c.1201_1202delinsAA:p.Ser401Asn	–	–	–	–
chr17	73832719	73832719	C	T	*UNC13D*	NM_199242.2	Nonsynonymous	SNV	*UNC13D*: NM_199242:exon14:c.1232G>A:p.R411Q	0.0006	0.0006	–	–
chr17	73832671	73832671	C	T	*UNC13D*	NM_199242.2	Nonsynonymous	SNV	*UNC13D*: NM_199242:exon14:c.1280G>A:p.R427Q	5.694e–05	8.298e–05	–	ID=COSM3421908; OCCURENCE=1 (large_intestine)
chr17	73839908	73839908	C	T	*UNC13D*	NM_199242.2	Nonsynonymous	SNV	*UNC13D*: NM_199242:exon14:exon14:c.c.118-308C>T:p.?	–	–	–	–
chr17	73831987	73831987	A	C	*UNC13D*	NM_199242.2	Nonsynonymous	SNV	*UNC13D*: NM_199242:exon18:c.1545T>G:p.N515K	–	–	–	–
chr17	73831848	73831848	C	A	*UNC13D*	NM_199242.2	Nonsynonymous	SNV	*UNC13D*: NM_199242:exon19:c.1607G>T:p.R536L	1.219e–05	8.263e–06	–	–
chr17	73831015	73831015	C	A	*UNC13D*	NM_199242.2	Nonsynonymous	SNV	*UNC13D*: NM_199242:exon21:c.1978G>T:p.V660F	–	–	–	–
chr17	73831014	73831015	GC	ATTACCG	*UNC13D*	NM_199242.2	Nonsynonymous	Frameshift insertion	*UNC13D*: NM_199242:exon23:c.1978_1979insATTACCG:p.Val660fs	–	–	–	–
chr17	73830525	73830525	G	A	*UNC13D*	NM_199242.2	Nonsynonymous	SNV	*UNC13D*: NM_199242:exon23:c.2179C>T:p.R727W	2.997e–05	–	–	–
chr17	73830485	73830485	G	A	*UNC13D*	NM_199242.2	Nonsynonymous	SNV	*UNC13D*: NM_199242:exon23:c.2219C>T:p.T740M	0.0001	9.323e–05	–	ID=COSM984488; OCCURENCE=1 (endometrium)
chr17	73830464	73830464	C	T	*UNC13D*	NM_199242.2	Nonsynonymous	SNV	*UNC13D*: NM_199242:exon23:c.2240G>A:p.S747N	3.714e–05	4.453e–05	–	–
chr17	73827216	73827216	C	T	*UNC13D*	NM_199242.2	Nonsynonymous	SNV	*UNC13D*: NM_199242:exon27:c.2588G>A:p.G863D	0.0003	0.0004	–	–
chr17	73826691	73826691	G	A	*UNC13D*	NM_199242.2	Nonsynonymous	SNV	*UNC13D*: NM_199242:exon28:c.2677C>T:p.R893W	1.254e–05	–	–	–
chr17	73826517	73826517	G	A	*UNC13D*	NM_199242.2	Nonsynonymous	SNV	*UNC13D*: NM_199242:exon29:c.2756C>T:p.S919F	4.057e–05	2.257e–05	–	–
chr17	73826509	73826509	C	T	*UNC13D*	NM_199242.2	Nonsynonymous	SNV	*UNC13D*: NM_199242:exon29:c.2764G>A:p.A922T	8.654e–05	0.0001	–	–
chr17	73826473	73826473	C	T	*UNC13D*	NM_199242.2	Nonsynonymous	SNV	*UNC13D*: NM_199242:exon29:c.2800G>A:p.A934T	9.831e–06	1.999e–05	–	–
chr17	73839274	73839274	G	A	*UNC13D*	NM_199242.2	Nonsynonymous	SNV	*UNC13D*: NM_199242:exon3:c.227C>T:p.T76M	0.0004	0.0005	–	–
chr17	73826166	73826166	C	T	*UNC13D*	NM_199242.2	Nonsynonymous	SNV	*UNC13D*: NM_199242:exon30:c.2897G>A:p.R966Q	0.0001	0.0001	–	–
chr17	73825047	73825047	G	A	*UNC13D*	NM_199242.2	Nonsynonymous	SNV	*UNC13D*: NM_199242:exon31:c.2972C>T:p.P991L	2.621e–05	1.851e–05	–	–
chr17	73824952	73824952	G	A	*UNC13D*	NM_199242.2	Nonsynonymous	SNV	*UNC13D*: NM_199242:exon31:c.3067C>T:p.R1023C	0.0001	0.0003	–	–
chr17	73824885	73824885	G	A	*UNC13D*	NM_199242.2	Nonsynonymous	SNV	*UNC13D*: NM_199242:exon31:c.3134C>T:p.T1045M	8.629e–05	0	–	–
chrX	123034137	123034141	ATTAA	-	*XIAP*	NM_001204401.1	Nonsynonymous	Frameshift deletion	*XIAP*: NM_001167:exon3:c.894_898del:p.Lys299fs	–	–	–	–
chrX	123022553	123022553	C	G	*XIAP*	NM_001204401.1	Nonsynonymous	SNV	*XIAP*: NM_001167:exon3:c.962C>G:p.A321G XIAP:NM_001204401:exon3:c.962C>G:p.A321G	0.0003	0.0003	Accession: 367794 phneotype: Lymphoproliferative syndrome Clinical significance: Likely_benign	–
chrX	123034437	123034437	G	T	*XIAP*	NM_001204401.1	Nonsynonymous	SNV	*XIAP*: NM_001167:exon6:c.1194G>T:p.Q398H XIAP:NM_001204401:exon6:c.1194G>T:p.Q398H	5.035e–05	5.727e–05	–	–
chrX	123034504	123034504	A	G	*XIAP*	NM_001204401.1	Nonsynonymous	SNV	*XIAP*: NM_001167:exon6:c.1261A>G:p.S421G XIAP:NM_001204401:exon6:c.1261A>G:p.S421G	–	–	–	–