Table SI. Summary of the clinical features of reported patients with pathogenic *CARMIL2* variants

		Ref. 2						Ref. 8				Ref. 9				Ref. 10							Ref. 11			Ref. 12	Ref. 13				
	Present patient	A1	A2	A3	B1	B2	C1	1 A:II-2	2 B:II-1	3 C:II-1	4 C:II-3	P1.1	P1.2	P2.1	P2.2	F1P1	F1P2	F1P3	F2P1	F2P2	F3P1	F3P2	1	2	3		P1	P2	P3	P4	P5
Age of first reported symptoms (years)	1.5	<1	1	< 2	<1	5	9	1		2		7	11	1	1	3	5	1	2	9	3	<1	4	<1	<1	10	<1	<1	5	6	4.5
Type of *CARMIL2* variant	ns	ms	ms	ms	ns	ns	ms	ms	ms	ms	ms	fs	fs	fs	fs	fs	fs	fs	fs	fs	ms	ms	sp	sp	sp	ms	sp	sp	fs	fs	fs
Skin and cutaneous annexes																															
Warts								+	+	+	+	+		+							+	+	+	+							
Molluscum contagiosum	++				+	+		+	+	–	–													+	+						
Viral skin infections								–	–	+	+												+	+	+						
Atopic dermatitis-like lesions	++				+	+	+	+	+	–	+	+			+	+	+	+		+	+	+	+		+		+				
Photodermatitis								–	–	+	++																				
Psoriasis-like lesions	+	+	+	+		+		+	+	+	+													+	+	+				+	
Dermatophytic lesions								+	+	–	+										+	+		+	+						+
CMC with perionyxis		+	+	+																											
Subcutaneous abscesses					+	+							+			+	+	+	+	+	+	+	+	+							
Alopecia	++	+																													
Lung and respiratory airways																															
Nasal congestion/polyps	++							+	++	–	(+)										+							+		+	+
Respiratory infections	+	+	+	+	+	+		(+)	–	(+)	+	+	+	+	+			+	+	+	+	+	+	+	+		+	+		+	+
Asthma	+				+	+	+	+	+	(+)	+										+		+					+			
Tuberculosis		+					+																								
COPD	+							+	+	–	[a]							+	+	+	+										
Digestive system																															
Gastritis					+			+																			+	+			
Stenosis of oesophagus	+	+														+	+					+				+			+		
Diarrhoea	+							+	–													+				+	+	+	+	+	+
IBD	+								–	+	+											+				+	+	+	+	+	+
Malabsorption	++																									+	+	+	+	+	+
Stomatitis/cheilitis	++							(+)	–	+	+															+	+	+	+	+	+
Pyorrhoea and gingivitis	++																			+											
Arthritis and arthralgia	+																													+	
Muscle cramps								+	+	–	(+)																				
EBV+ smooth muscle tumours										[b]		+	+	+	+																
Other		[c]							[d,e]		[e]																				

[a]Lung scarring; [b]malignant leiomyosarcoma; [c]viral meningoencephalitis; [d]haemophagocytic lymphohistiocytosis; [e]BK virus cystitis.
COPD: chronic obstructive pulmonary disease; IBD: inflammatory bowel disease; EBV: Epstein-Barr virus; ms: missense variant; ns: nonsense variant; fs: frameshift (truncating) variant; sp: splice site variant.