

Appendix 1

In this file are contained clinical data of the single patients of our cohort, in order to help to understand the peculiarities of the disease onset and the progressive evolution of the clinical phenotype during a long-term follow-up. Please note that the difference between clinical profile at presentation and at diagnosis derives from the long time between the disease onset and the diagnosis of pSLE.

Patient 1

Age at onset: 15 years

Clinical presentation: Gastroenteritis, malar rash, photosensitivity

Time to diagnosis: 17 months

Duration of follow-up: 4 years

Clinical feature	Diagnosis	1 Year	3 Years
Acute cutaneous SLE	X	X	X
Malar Rash			
Photosensitivity	X	X	X
Articular Involvement	X	X	X
Renal Involvement			
Neurolupus			
Haematological Involvement	X	X	X
Haemolytic Anemia	X		
Leukopenia	X	X	X
Thrombocytopenia			

Patient 2

Age at onset: 15 years

Clinical presentation: Photosensitivity, malar rash

Time to diagnosis: 11 months

Duration of follow-up: 30 years

Clinical feature	Diagnosis	1 Year	3 Years	5 Years	10 Years
Acute cutaneous SLE	X				
Malar Rash	X				
Photosensitivity					
Articular Involvement	X	X			X
Renal Involvement		X	X	X	X
Neurolupus					
Haematological Involvement	X	X		X	X
Haemolytic Anemia					
Leukopenia	X	X		X	X
Thrombocytopenia					
Other					Pleuritis

Patient 3

Age at onset: 16 years

Clinical presentation: Arthritis

Time to diagnosis: 2 months

Duration of follow-up: 4 years

Clinical feature	Diagnosis	1 Year	3 Years
Acute cutaneous SLE	X	X	
Malar Rash	X	X	
Photosensitivity	X	X	
Articular Involvement	X	X	X
Renal Involvement		X	
Neurolupus			
Haematological Involvement			
Haemolytic Anemia			
Leukopenia			
Thrombocytopenia			

Patient 4

Age at onset: 17

Clinical presentation: Pleuritis, Pericarditis

Time to diagnosis: 1 month

Duration of follow-up: 7 years

Clinical feature	Diagnosis	1 Year	3 Years	5 Years
Acute cutaneous SLE				
Malar Rash				
Photosensitivity				
Articular Involvement				
Renal Involvement				
	X	X	X	X
Neurolupus				
Haematological Involvement				
Haemolytic Anemia				
Leukopenia				
	X	X	X	
Thrombocytopenia				
Other				
	Pleuro-pericarditis		Pleuro-pericarditis	Pleuritis

Patient 5

Age at onset: 15 years

Clinical presentation: Thrombocytopenia

Time to diagnosis: 62 months

Duration of follow-up: 4 years

Clinical feature	Diagnosis	1 Year	3 Years
Acute cutaneous SLE	X	X	X
Malar Rash	X		X
Photosensitivity	X	X	X
Articular Involvement			
Renal Involvement			
Neurolupus			
Haematological Involvement	X	X	X
Haemolytic Anemia			
Leukopenia			
Thrombocytopenia	X	X	X

Patient 6

Age at onset: 14

Clinical presentation: Nephritis, malar rash, photosensitivity

Time to diagnosis: 0 months

Duration of follow-up: 24 years

Clinical feature	Diagnosis	1 Year	3 Years	5 Years	10 Years
Acute cutaneous SLE	X	X	X	X	
Malar Rash	X	X	X	X	
Photosensitivity	X	X		X	
Articular Involvement					
Renal Involvement	X	X	X	X	X
Neurolupus					X
Haematological Involvement					
Haemolytic Anemia					
Leukopenia					
Thrombocytopenia					
Other			Vasculitis		Vasculitis

Patient 7

Age at onset: 15 years

Clinical presentation: Arthritis, proteinuria, malar rash, cytopenia

Time to diagnosis: 0 months

Duration of follow-up: 24 years

Clinical feature	Diagnosis	1 Year	3 Years	5 Years	10 Years
Acute cutaneous SLE	X				
Malar Rash	X				
Photosensitivity					
Articular Involvement	X				
Renal Involvement	X	X	X	X	X
Neurolupus					X
Haematological Involvement	X	X	X	X	X
Haemolytic Anemia					
Leukopenia	X				
Thrombocytopenia	X	X	X	X	X

Patient 8

Age at onset: 14 years

Clinical presentation: Malar rash, arthritis

Time to diagnosis: 0 months

Duration of follow-up: 17 years

Clinical feature	Diagnosis	1 Year	3 Years	5 Years	10 Years
Acute cutaneous SLE	X				
Malar Rash	X				
Photosensitivity					
Articular Involvement	X				
Renal Involvement				X	X
Neurolupus					
Haematological Involvement					
Haemolytic Anemia					
Leukopenia					
Thrombocytopenia					
Other			Pleuro-pericarditis		

Patient 9

Age at onset: 13 years

Clinical presentation: Malar rash

Time to diagnosis: 12 months

Duration of follow-up: 10 years

Clinical feature	Diagnosis	1 Year	3 Years	5 Years	10 Years
Acute cutaneous SLE	X	X	X	X	X
Malar Rash	X	X	X	X	X
Photosensitivity	X	X		X	X
Articular Involvement		X		X	X
Renal Involvement			X	X	X
Neurolupus					
Haematological Involvement				X	X
Haemolytic Anemia					
Leukopenia				X	X
Thrombocytopenia					

Patient 10

Age at onset: 16 years

Clinical presentation: Arthritis, Raynaud's phenomenon

Time to diagnosis: 26 months

Duration of follow-up: 19 years

Clinical feature	Diagnosis	1 Year	3 Years	5 Years	10 Years
Acute cutaneous SLE					
Malar Rash					
Photosensitivity					
Articular Involvement	X	X	X	X	
Renal Involvement					X
Neurolupus					X
Haematological Involvement					
Haemolytic Anemia					
Leukopenia					
Thrombocytopenia					

Patient 11

Age at onset: 12 years

Clinical presentation: malar rash

Time to diagnosis: 6 months

Duration of follow-up: not available (too recently diagnosed)

Clinical feature	Diagnosis
Acute cutaneous SLE	X
Malar Rash	X
Photosensitivity	X
Articular Involvement	X
Renal Involvement	
Neurolupus	
Haematological Involvement	
Haemolytic Anemia	
Leukopenia	
Thrombocytopenia	

Patient 12

Age at onset: 17 years

Clinical presentation: Arthritis, photosensitivity

Time to diagnosis: 0 months

Duration of follow-up: 5 years

Clinical feature	Diagnosis	1 Year	3 Years	5 Years
Acute cutaneous SLE	X	X	X	X
Malar Rash				
Photosensitivity	X	X	X	X
Articular Involvement	X		X	X
Renal Involvement				
Neurolupus				
Haematological Involvement	X	X		
Haemolytic Anemia				
Leukopenia	X	X		
Thrombocytopenia				

Patient 13

Age at onset: 15 years

Clinical presentation: Renal involvement

Time to diagnosis: 2 months

Duration of follow-up: 11 years

Clinical feature	Diagnosis	1 Year	3 Years	5 Years	10 Years
Acute cutaneous SLE					
Malar Rash					
Photosensitivity					
Articular Involvement					
Renal Involvement	X	X	X	X	X
Neurolupus					
Haematological Involvement				X	X
Haemolytic Anemia					
Leukopenia					
Thrombocytopenia				X	X

Patient 14

Age at onset: 14 years

Clinical presentation: Arthritis, Raynaud's phenomenon

Time to diagnosis: 12 months

Duration of follow-up: 15 years

Clinical feature	Diagnosis	1 Year	3 Years	5 Years	10 Years
Acute cutaneous SLE	X				
Malar Rash					
Photosensitivity	X				
Articular Involvement	X				
Renal Involvement					
Neurolupus					
Haematological Involvement	X	X	X	X	X
Haemolytic Anemia					
Leukopenia	X	X	X	X	X
Thrombocytopenia					
Other					Vasculitis

Patient 15

Age at onset: 17 years

Clinical presentation: Arthritis, cytopenia

Time to diagnosis: 0 months

Duration of follow-up: 16 years

Clinical feature	Diagnosis	1 Year	3 Years	5 Years	10 Years
Acute cutaneous SLE					
Malar Rash					
Photosensitivity					
Articular Involvement	X				X
Renal Involvement		X	X	X	X
Neurolupus					
Haematological Involvement	X	X	X	X	X
Haemolytic Anemia	X	X			
Leukopenia	X		X	X	
Thrombocytopenia					X

Patient 16

Age at onset: 15 years

Clinical presentation: Thrombocytopenia

Time to diagnosis: 67 months

Duration of follow-up: 10 years

Clinical feature	Diagnosis	1 Year	3 Years	5 Years	10 Years
Acute cutaneous SLE					
Malar Rash					
Photosensitivity					
Articular Involvement			X		
Renal Involvement	X				X
Neurolupus					
Haematological Involvement	X	X	X	X	X
Haemolytic Anemia					
Leukopenia	X		X		
Thrombocytopenia	X	X	X	X	X
Other				Vasculitis	Vasculitis

Patient 17

Age at onset: 15 years
Clinical presentation: Arthritis, renal involvement
Time to diagnosis: 34 months
Duration of follow-up: 13 years

Clinical feature	Diagnosis	1 Year	3 Years	5 Years	10 Years
Acute cutaneous SLE			X		X
Malar Rash			X		X
Photosensitivity			X		X
Articular Involvement	X		X		
Renal Involvement	X	X	X	X	X
Neurolupus					
Haematological Involvement		X	X	X	X
Haemolytic Anemia		X	X	X	X
Leukopenia					
Thrombocytopenia					

Patient 18

Age at onset: 13 years

Clinical presentation: Thrombocytopenia

Time to diagnosis: 52 months

Duration of follow-up: 15 years

Clinical feature	Diagnosis	1 Year	3 Years	5 Years	10 Years
Acute cutaneous SLE	X				
Malar Rash	X				
Photosensitivity					
Articular Involvement	X				
Renal Involvement	X	X	X	X	X
Neurolupus					
Haematological Involvement	X				
Haemolytic Anemia					
Leukopenia					
Thrombocytopenia	X				

Patient 19

Age at onset: 16 years

Clinical presentation: Arthritis

Time to diagnosis: 10 months

Duration of follow-up: 10 years

Clinical feature	Diagnosis	1 Year	3 Years	5 Years	10 Years
Acute cutaneous SLE	X	X			
Malar Rash	X	X			
Photosensitivity					
Articular Involvement	X				
Renal Involvement		X	X	X	X
Neurolupus					
Haematological Involvement	X				
Haemolytic Anemia	X				
Leukopenia	X				
Thrombocytopenia	X				

Patient 20

Age at onset: 15 years

Clinical presentation: Malar rash

Time to diagnosis: 0 months

Duration of follow-up: 26 years

Clinical feature	Diagnosis	1 Year	3 Years	5 Years	10 Years
Acute cutaneous SLE	X	X	X	X	X
Malar Rash	X	X	X	X	X
Photosensitivity	X	X	X	X	
Articular Involvement					
Renal Involvement					
Neurolupus					
Haematological Involvement					
Haemolytic Anemia					
Leukopenia					
Thrombocytopenia					

Patient 21

Age at onset: 15 years

Clinical presentation: Malar rash, xerostomia, xerophtalmia

Time to diagnosis: 86 months

Duration of follow-up: 10 years

Clinical feature	Diagnosis	1 Year	3 Years	5 Years	10 Years
Acute cutaneous SLE	X			X	X
Malar Rash	X			X	X
Photosensitivity	X			X	
Articular Involvement					
Renal Involvement					
Neurolupus					
Haematological Involvement	X				X
Haemolytic Anemia					
Leukopenia					
Thrombocytopenia	X				X

Patient 22

Age at onset: 14 years
Clinical presentation: Arthritis, cytopenia
Time to diagnosis: 0 months
Duration of follow-up: 18 years

Clinical feature	Diagnosis	1 Year	3 Years	5 Years	10 Years
Acute cutaneous SLE					
Malar Rash					
Photosensitivity					
Articular Involvement	X				
Renal Involvement		X		X	X
Neurolupus		X	X	X	
Haematological Involvement	X				X
Haemolytic Anemia	X				
Leukopenia	X				
Thrombocytopenia					X

Patient 23

Age at onset: 11 years

Clinical presentation: Haemolytic anemia

Time to diagnosis: 35 months

Duration of follow-up: 3 years

Clinical feature	Diagnosis	1 Year	3 Years
Acute cutaneous SLE			
Malar Rash			
Photosensitivity			
Articular Involvement			
Renal Involvement	X	X	X
Neurolupus			
Haematological Involvement	X		
Haemolytic Anemia	X		
Leukopenia			
Thrombocytopenia			

Patient 24

Age at onset: 15 years

Clinical presentation: Deep venous thrombosis, arthritis

Time to diagnosis: 0 months

Duration of follow-up: 28 years

Clinical feature	Diagnosis	1 Year	3 Years	5 Years	10 Years
Acute cutaneous SLE		X		X	
Malar Rash				X	
Photosensitivity		X		X	
Articular Involvement	X	X			
Renal Involvement					
Neurolupus		X		X	X
Haematological Involvement	X	X			
Haemolytic Anemia	X				
Leukopenia					
Thrombocytopenia	X	X			

Patient 25

Age at onset: 11,5 years

Clinical presentation: Malar rash, arthritis, renal involvement

Time to diagnosis: 1 month

Duration of follow-up: 17 years

Clinical feature	Diagnosis	1 Year	3 Years	5 Years	10 Years
Acute cutaneous SLE	X				
Malar Rash	X				
Photosensitivity					
Articular Involvement	X				X
Renal Involvement	X				
Neurolupus					
Haematological Involvement					X
Haemolytic Anemia					
Leukopenia					X
Thrombocytopenia					
Other	Pericarditis				