

Table S1 Baseline characteristics and diagnostic timing (n=6)

Characteristic	Value
Patients, n	6
Age at index encounter, years, median (range)	30 (26–34)
Female sex, n (%)	4 (66.7)
Index encounter service, n (%)	
Dermatology	4 (66.7)
Neurology	1 (16.7)
Respiratory medicine	1 (16.7)
Adult diagnosis (age ≥ 18 years), n (%)	5 (83.3)
Age at diagnosis in adult-diagnosed cases, years, median (range)	29.1 (20.3–35.0)
Diagnostic delay in adult-diagnosed cases, years, median (range)	23.4 (6.1–31.9)
Childhood diagnosis, n	1 ^a

^aDiagnosis year discrepant across documents (1993 vs 1997); diagnostic timing for this case is approximate.

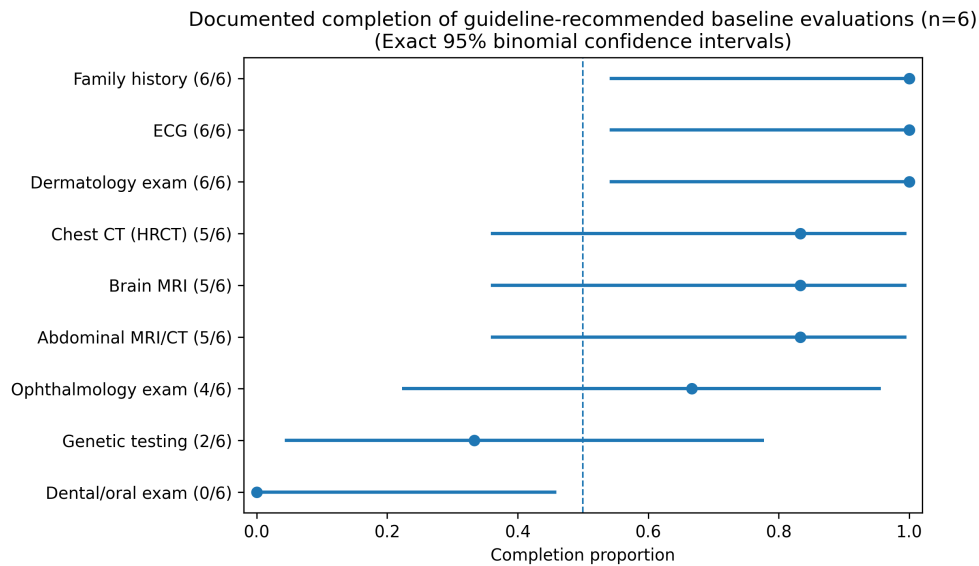


Fig. S1 Documented completion proportions for core baseline evaluation items (n=6). Dots indicate completion proportions and horizontal lines indicate exact 95% binomial confidence intervals (Clopper–Pearson).

Table S2 Clinical context at adult evaluation (n=6)

Domain / variable	Summary
Cutaneous manifestations	
Facial angiofibromas, n/N (%)	6/6 (100.0)
Hypomelanotic macules (Wood's lamp positive), n/N (%)	4/6 (66.7)
Ungual fibromas, n/N (%)	4/6 (66.7)
Shagreen patch, n/N (%)	4/6 (66.7)
Neurologic manifestations	
Seizure/convulsive episode history documented, n/N (%)	4/6 (66.7)
Brain MRI documented, n/N (%)	5/6 (83.3)
Renal involvement and kidney function	
Renal AML documented, n/N (%)	5/6 (83.3)
Maximal AML diameter, mm, median (range) ^b	50 (9.2–120)
Maximal AML diameter ≥ 40 mm, n/N (%) ^b	4/5 (80.0)
Renal cysts, n/N (%)	3/6 (50.0)
Systolic blood pressure, mmHg, median (range)	134.5 (118–148)
Diastolic blood pressure, mmHg, median (range)	81 (71–95)
eGFR, mL/min/1.73 m ² , median (range)	105.3 (31.0–123.8)
Proteinuria not negative (trace/positive), n/N (%)	3/6 (50.0)
Renal AML hemorrhage, n/N (%)	1/6 (16.7)
Renal intervention: embolization, n/N (%)	1/6 (16.7)
Renal intervention: partial nephrectomy history, n/N (%)	1/6 (16.7)
Pulmonary involvement	
Chest HRCT performed, n/N (%)	5/6 (83.3)
Typical LAM on HRCT, n/N (%)	2/6 (33.3)
Pulmonary function tests performed, n/N (%)	2/6 (33.3)
FEV ₁ (L), median (range) ^c	2.34 (2.09–2.58)
DLCO % predicted available, n/N (%) ^c	1/2 (50.0)
DLCO % predicted (single available value)	59
Cardiac and ophthalmologic	
ECG abnormal, n/N (%)	3/6 (50.0)
Echocardiography performed, n/N (%)	5/6 (83.3)
Ophthalmology exam performed, n/N (%)	4/6 (66.7)
Retinal hamartoma among those examined, n/N (%)	0/4 (0.0)
Genetics and family history	
Family history collected, n/N (%)	6/6 (100.0)
Family history positive, n/N (%)	2/6 (33.3)
Genetic testing performed, n/N (%)	2/6 (33.3)
Oral/dental	
Dental/oral evaluation performed, n/N (%)	0/6 (0.0)

^bCalculated among patients with documented AML diameter measurements (n=5).^cCalculated among patients with available pulmonary function tests (n=2).