Lymphangioleiomyomatosis screening upon transition in tuberous sclerosis complex

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Introduction & Objectives

- TSC is a rare multisystem autosomal-dominant genetic disorder.
- Diagnosis: primarily in infancy based on neurological and dermal features.
- TSC is associated with LAM with onset in younger adults, **negatively affecting mortality and morbidity.**
- TSC guidelines recommend screening of women with TSC for LAM at 18 years.
- **However, many patients are not screened mostly due to neurological impairment.**

Objective: To examine the presence of LAM in the transition from childhood to adult life.

Methods

- Prospective cohort study including TSC patients 18-26 y/o evaluated for LAM.
- TSC diagnosis: latest criteria (2021)
- LAM diagnosis: characteristic HRCT findings
- From March to October 2025, 9 out of 115 TSC patients of the Greek pediatric cohort were evaluated.

Results

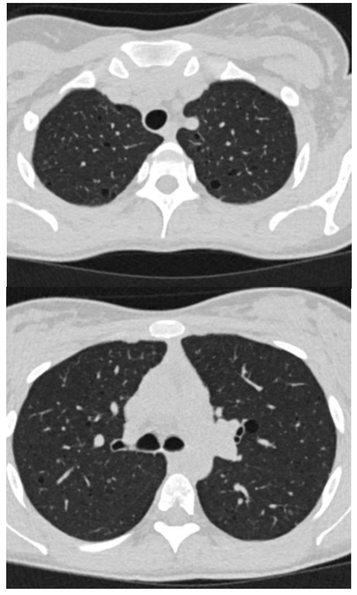
Table 1.Baseline Characteristics of TSC patients (n=9)

Non parametric variables	Parameter	Median (range)
56% female	Age at inclusion (yr)	18 (17-22)
100% non-smokers	Age at diagnosis	2.3 (1-5.4)
Hx of epilepsy 7 (78%)	BMI (kg/m^2)	24.6 (23.3-27.6)
Learning difficulties 5 (56%)	$SatO_2$ (%)	98 (97.5-98)
TAND 5 (56%)		
SENs 9 (100%)		
SEGAs 3 (33.3%)		
Angiomyolipoma 3 (33.3%)		
mTOR treatment 3 (33.3%)		

Table 2. Diagnostic criteria for TSC

Major	Minor
Hypomelanotic macules ≥3	Confetti lesions
Angiofibromas ≥3	Dental pits >3
Ungual fibromas ≥2	Intraoral fibromas ≥2
Shagreen patch	Retinal achromic patch
Retinal hamartomas	Renal cysts
Cortical dysplasias	Non-renal hamartomas
SENs	Sclerotic bone lesions
SEGA	
Cardiac rhabdomyoma	
LAM	
AMLs ≥2	

Definite TSC:
2 major OR
1 major + ≥2 minor OR
genetic diagnosis
*AMLs + LAM→ not definite TSC



HRCT documented LAM in 1 26 yo female patient

Figure 1. HRCT with LAM findings

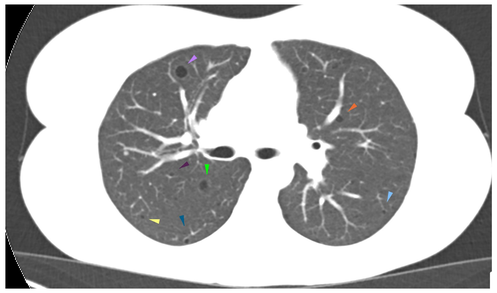


Figure 2. Angiofibromas

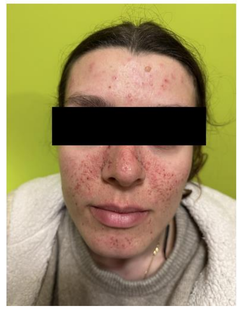


Figure 3. Angiomyolipomas

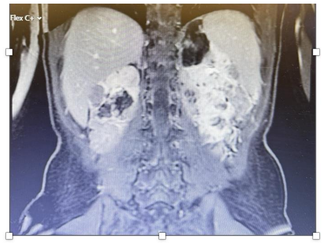


Figure 4. Hypomelanotic macules

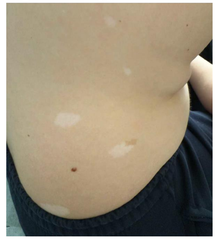


Figure 5. Shagreen patches

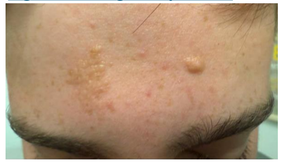


Figure 6. Subependymal nodules

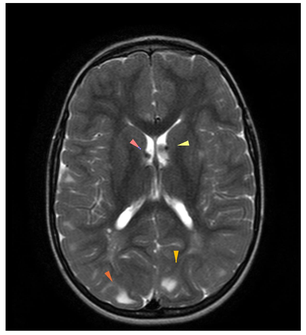
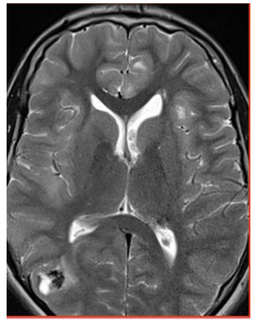
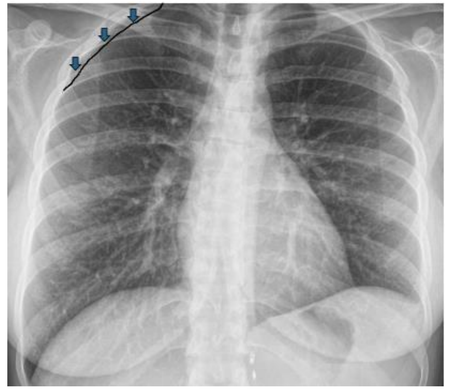


Figure 7. SEGA



All data are collected in an anonymized manner after Medical Ethics Committee approval (Β'ΠΝΕΥΜ, ΕΒΔ 583/29-08-2025).

Patient presented dyspnea and was complicated with pneumothorax shortly after the diagnosis (FEV1% 80%pred, DLCO% 73% pred).



Conclusions

- In TSC, screening for LAM upon transition necessitates close collaboration between experts (pediatric and adult neurologists, pulmonary physicians).
- Screening and specialized care could prevent avoidable lung damage, progression of disease, complications and further disability.

References
1) Rebaine Y et al., Eur Respir Rev 2021;30:200348.
2) Johnson J et al., Orphanet J Rare Dis. 2024;19:137.