Navigating the Patient Journey of Tuberous Sclerosis Complex (TSC) in Germany

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1. BACKGROUND

Tuberous Sclerosis Complex (TSC) is a rare multisystem genetic disorder requiring lifelong care [1]. This qualitative interview study as part of a master's thesis explored how patients, caregivers and healthcare professionals experience the TSC patient journey in Germany, focusing on access, coordination, continuity and treatment experiences, including facial angiofibromas (FA) [2].

2. METHODS

Study Design

Qualitative, exploratory study using semi-structured online interviews (April – June 2026)

Theoretical Framework

Integrated approach:
- Andersen's Behavioral Model
- Chronic Care Model
- Concept of Continuity of Care

Participants

(*n* = 20)
- 3 adult patients
- 11 caregivers
- 6 healthcare professionals specialized in TSC

Data Analysis

Qualitative content analysis (deductive-inductive approach) using ATLAS.ti; reported following SRQR guidelines

3. RESULTS

Thematic model of the TSC patient journey

Cross-cutting theme: Living with facial angiofibromas (FA)

Paediatric care | **Transition to adult care** | **Adult care and long-term management**

1 Recognition & diagnosis
- early signs
- delayed diagnosis
- referral pathways

2 Access to specialised TSC care
- TSC centres
- specialist expertise
- regional availability

3 Care coordination & navigation
- family-led navigation
- appointments & reports
- bureaucracy

4 Transition to adult care
Potential disruption of continuity
- transfer of responsibility
- loss of familiar providers
- risk of care gaps
- fragmented adult services

5 Continuity in adult care
- long-term follow-up
- informational continuity
- management continuity

6 Treatment, monitoring & daily life
- treatment effectiveness
- treatment burden
- adherence
- psychosocial burden

Ongoing coordination and family-led navigation across the care pathway

Living with FA

visibility · psychosocial relevance · treatment access · adherence & reimbursement

Although FA can cause significant psychosocial distress and reduced quality of life due to their visibility in some patients, consistent topical treatment is frequently hindered by local follow-up prescription barriers and is sometimes deprioritized due to systemic TSC symptoms and everyday usability.

Quote of a healthcare professional regarding topical sirolimus/rapamycin:
"It is approved. It can be prescribed, and the health insurance has to cover the costs. […] There are special budgets for this. You have to submit an application once […]. Then the health insurance knows, and then it is excluded from the budget calculation."

Recurring patterns identified across the interviews

Recognizing and accessing TSC care
Gaining access to specialized TSC care is often a non-standardized process that depends heavily on the early recognition of complex symptoms, proactive referrals, and geographical proximity to expert centers.

Coordinating fragmented care
Due to highly fragmented healthcare structures, caregivers act as informal care coordinators who constantly manage appointments, prescriptions, and the flow of medical information between specialized centers and local providers.

Maintaining continuity over time
Continuity of care is particularly vulnerable during the transition to adulthood, where patients often lose the integrated structure of multidisciplinary paediatric care and must instead navigate isolated, organ-specific adult services.

Managing treatment and daily burden
While medical therapies provide essential clinical stability, they create a significant daily burden due to extensive monitoring requirements and the critical need to adapt medical routines to patients' cognitive and behavioral needs.

4. CONCLUSION

The primary challenge in German TSC care is not just accessing specialized expertise, but making this expertise coordinated and continuous. There is a significant implementation gap between guideline-based lifelong surveillance and its actual organization in routine care. Improving the TSC patient journey requires establishing central care coordinators, implementing structured transition pathways, and developing shared-care models to reduce the administrative burden on families and make specialized expertise usable in everyday life.

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References: [1] Northrup et al. Updated International Tuberous Sclerosis Complex Diagnostic Criteria and Surveillance and Management Recommendations. *Pediatr Neurol* 2021, 123, 50-66. [2] Submitted for journal publication.

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