



Tuberous sclerosis complex: Concerns and needs of patients and parents from the transitional period to adulthood

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ABSTRACT

Introduction: Transitioning into adulthood and from pediatric services to adult healthcare are both challenging processes for young adults with rare chronic disorders such as tuberous sclerosis complex (TSC) and their parents. Adult healthcare systems are often less family-oriented and lack multidisciplinary care and experience with TSC, which can result in increased health risks and morbidity. Patient-driven data on care needs are necessary to optimize support for this vulnerable patient group.

Aim: The aim of this study was to explore the concerns and care needs of young adult patients with TSC in medical, psychological, and socioeconomic domains.

Method: A qualitative study was performed using semistructured interviews with 16 patients (median age: 21 years; range: 17 to 30) and 12 parents. Concerns and care needs were organized using the International Classification of Functioning, Disability, and Health (ICF).

Results: Main concerns involved mental and physical health, participation, self-management skills, family planning, and side effects of medications. Patients expressed the need for multidisciplinary care that is well-informed, easily accessible, and focused on the patient as a whole, including his/her family. Parents reported high stress levels.

Conclusion: The current study provides patient-driven information, allowing recommendations to facilitate the (transition of) care for young adults with TSC. In addition to seizures, tumor growth, and TSC-associated neuropsychiatric disorders (TAND), more attention is needed for concerns and care needs specific to the transitional period, participation, and environmental factors. Adult healthcare providers should offer expert multidisciplinary care for adult patients with TSC, including attention for parental stress.

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1. Introduction

Tuberous sclerosis complex (TSC) is a rare genetic disorder with a birth incidence of 1:6000 [1], caused by mutations in either the *TSC1* or

Abbreviations: ADHD, attention-deficit/hyperactivity disorder; ASD, autism spectrum disorder; ICF, International Classification of Functioning, Disability, and Health; ID, intellectual disability; LAM, lymphangioleiomyomatosis; MEC, Medical and Ethical Review Committee; mTOR, mammalian target of rapamycin; SEGA, subependymal giant cell astrocytoma; STSN, Stichting Tubereuze Sclerose Nederland; TAND, TSC-associated neuropsychiatric disorders; TSC, tuberous sclerosis complex; WHO, World Health Organization.

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the *TSC2* gene [2–4]. Through forming a *TSC1–TSC2* protein complex, the proteins regulate cell proliferation and differentiation and function as tumor-suppressor genes through the mammalian target of rapamycin (mTOR) complex 1 pathway [2,5]. Loss of function of these proteins causes growth of generally benign tumors in multiple organ systems, such as cutaneous features (96%), renal angiomyolipomas (80%), subependymal giant cell astrocytomas (SEGA; 20%), pulmonary lymphangioleiomyomatosis (LAM; 26–34%), and cardiac rhabdomyomas (18–58%) [6–13]. In addition, structural abnormalities and disturbances in neurobiological processes are associated with various neurological and psychiatric symptoms, such as epilepsy (70–90%), autism spectrum disorder (ASD; 20–50%), attention-deficit/hyperactivity disorder (ADHD; 30–50%), intellectual disability (ID; 50%), and depression and anxiety disorders (30–60%) [14–19].

The multisystem manifestations of TSC vary across age, and thus, require multidisciplinary care throughout the lifespan of patients [16], especially during the transition from childhood and adolescence into adulthood, when medical issues and concerns are changing. For instance, the most prevalent psychiatric diagnoses in childhood and early adolescence are developmental disorders such as ASD and ADHD while mood and anxiety disorders are more likely to become evident in late adolescence and adulthood [20]. Comorbid epilepsy further complicates the transitional period of patients with TSC, as these patients require more supervision which subsequently interferes with the development of autonomy [21]. For example, adolescents with epilepsy show difficulties with self-management skills such as medication compliance, resulting in their parents (88%) becoming frequently responsible for their medication intake [22]. Lack of experience with TSC in the adult healthcare system—which is often less family-centered and less multidisciplinary—adds to the complexity of the transitional period [23]. Even though there are guidelines drafted with regard to TSC, specific diagnostic and treatment guidelines for neuropsychiatric problems during the transitional period and adulthood do not exist [18]. However, the recently developed TSC-associated neuropsychiatric disorders (TAND) checklist emphasizes the importance of the neuropsychiatric disorders frequently encountered in patients with TSC [16].

Often, children with chronic genetic/congenital diseases are lost to follow-up during the transfer from pediatric to adult care (~88% of patients with neurofibromatosis type 1) [24,25]. Previous research regarding the transitional period found that young people who are severely affected by their health issues tend to communicate less about their care needs during transition [26]. These care gaps can have adverse health effects, especially in case of comorbidities such as ASD and epilepsy [21,27]. To facilitate this transfer in healthcare, patients and their families and physicians need to all be prepared for this transfer [28, 29]. As the course of TSC is variable and unpredictable, patients run the risk of being negatively affected by care gaps during transition.

Little is known about the concerns and needs of young adults with TSC in medical, psychological, and socioeconomic domains. Even though the TAND checklist encompasses a large part of this spectrum, transitional, socioeconomic, and contextual issues are not explicitly addressed [16]. Therefore, the aim of the current study was to use patient- and parent-driven data to assess which difficulties patients and parents experience and what concerns they have in medical, psychological, and socioeconomic domains during the transitional period. Using this information, we hope to find recommendations to provide optimal care for this vulnerable patient group in the transitional period and young adulthood.

2. Methods

A qualitative study was carried out to examine the concerns and care needs of young adults with TSC in medical, psychological, and socioeconomic domains, with a special emphasis on the transfer from pediatric to adult care and parental stress. Data were collected using focus groups and semistructured interviews with young adult patients and parents of young adults with TSC.

2.1. Participants

Using purposive sampling, patients were invited either by the Dutch Foundation for patients with TSC (Stichting Tubereuze Sclerosis Nederland; STSN) or by their healthcare providers via email. Patients were included if they were an adolescent transitioning into adult care or within the young adult age range (18–30 years old), with a definitive TSC diagnosis (confirmed either by genetic testing or based on the diagnostic criteria [4]). Parents were required to have a child with TSC of 18 years, or older, or younger and having transitioned into adult care. Two focus groups were created: one for patients (17–30 years) and one for parents. Patients that were not able to participate in the focus groups were individually interviewed.

2.2. Data collection

The semistructured interviews with focus groups lasted 90 min. During these interviews, patients and parents were explicitly asked to discuss their experiences regarding healthcare transfer and their concerns and care needs in various domains. The interview questions and probes were based on the literature on TSC, the TAND checklist, the literature on the transitional age and process, and clinical experience (see Table 1). Trained moderators (JP, PB, and AE) redirected the conversation when necessary to ensure that all care domains were discussed. None of the moderators were clinically involved with the patients. Other participants were individually interviewed by phone or in person, depending on the preference of the patients or parents. The same interview guide as in the focus groups was used for this group of participants. Both focus groups and individual interviews in person were audio- and video-recorded, but interviews by phone were only audio-recorded.

2.3. Data analysis

The recordings of the focus groups and interviews were transcribed verbatim. Transcripts were imported into the ATLAS.ti 6.2 analysis software [30] to assist with data organization and analyses. Each meaningful element in the transcript was coupled to a code, and these codes were organized into categories. These categories were subdivided into themes, which are a reflection of the topics discussed in the focus groups and interviews [31]. The transcripts and codes were continually reevaluated by researchers (PB, AE, and a research student) to ensure that these accurately represented the categories and themes. To enhance validity, the coding process and the emerging themes were regularly discussed with the senior author (AE) and were also presented and discussed with an expert panel (AR, GH, JP, LWH, and MV) until consensus was reached. Interviews were held until data saturation was reached. In this article, several quotes from the patients or parents are used, which were translated from Dutch to English, potentially resulting in slight deviations in meaning.

To provide a holistic interpretation of the patients' and parents' concerns and needs, the themes were classified based on the components of the International Classification of Functioning, Disability, and Health (ICF) model. The ICF classification is a framework for classifying health

Table 1
Abbreviated interview guide.

| Key questions: | Probes |
|--|---|
| (1) What are your worries about... | |
| (2) What are your care needs for... | |
| Transition from pediatric to adult care? | Transition into adult healthcare, change in physician, transition into adult life, transition into work, independent living |
| Medical issues? | Symptoms, TSC-related care, medication, care consultations |
| Psychological and behavioral issues? | Sleeping problems, ADHD, ASD, depression, anxiety |
| Social life? | Friendships, romantic relationships, family, loneliness, sexuality |
| Work and daily activities? | Work, school, independence, finances, daily living |
| Paramedical issues? | Language problems, nutrition, motor skills |

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