Background: Tuberous sclerosis complex (TSC) is a multisystem genetic disorder with a global incidence of 1 per 6,000-20,000 live births (bit. 5 million people). 1-3

The purpose of this study was to assess the burden of illness and health-related quality of life in patients with TSC and their caregivers. A total of 143 patients (55 adults and 88 children) from 7 European countries were enrolled in this research project as part of the TOSCA study (Table 1). The main goals were to confirm that TSC and its manifestations have a severe impact on the children's QoL and emotional state of the caregiver. 4

Methods: TSC is a large-scale, non-interventional study designed with a "core" section to record background patient information, 6 research projects that focus on specific disease mechanisms, and a European post authorization safety study (PASS). Patients were included if they met the TSC diagnostic criteria, were aged between 11-18 years, and were living with epilepsy. All enrolled patients and their caregivers were asked to complete validated paper questionnaires in the local languages to collect information on QoL and EQ-5D. Table 1 summarizes the demographic characteristics of the study population. Patients with reporting problems, %

Table 1. Demographic Characteristics

<table>
<thead>
<tr>
<th>Age group (years)</th>
<th>Adults (n=55)</th>
<th>Children (n=88)</th>
<th>Overall (n=143)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sex, n (%)</td>
<td>Male</td>
<td>Female</td>
<td>Male</td>
</tr>
<tr>
<td></td>
<td>36</td>
<td>19</td>
<td>27</td>
</tr>
<tr>
<td></td>
<td>64%</td>
<td>46%</td>
<td>61%</td>
</tr>
<tr>
<td>Age at onset (years)</td>
<td>21±13.8</td>
<td>11±5.5</td>
<td>11±5.5</td>
</tr>
<tr>
<td>Duration of TSC (years)</td>
<td>11±10.2</td>
<td>28±17.4</td>
<td>27±11.8</td>
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</tr>
</tbody>
</table>

Results:

- Of the 143 enrolled patients, 55 adults and 88 children from European countries were enrolled in this research project as part of the TOSCA study. The main goals were to confirm that TSC and its manifestations have a severe impact on the children's QoL and emotional state of the caregiver.

- Among the 55 adults, 42 (76.4%) were able to complete the questionnaire independently, whereas caregivers completed the questionnaire for 13 patients.

- Among the 88 children, 78 (88%) were able to complete the questionnaires independently, whereas caregivers completed the questionnaire for 10 patients who required assistance.

- Fifty-nine patients (41.3%) reported that they were followed for TSC management by more than 3 physicians, whereas TSC was managed by a specialized team in 45 patients (31.6%).

- Of the 28 adults with epilepsy, 15 (53.6%) reported an impact on their camcorder/education due to TSC.

- Impact on family: The impact of the disease on the patients and their families was assessed by 29 families (52.7%). Social, familial, and professional relationships of the caregiver were impacted by the child's TSC condition in 29 families (52.7%).

- Twenty-six patients (33.3%) reported that their social care was impacted by TSC.

- Of the 143 enrolled patients, 42 (29.4%) were hospitalized in the last 12 months since the enrolment in this research project. Main reasons for hospitalization were seizures, complications of antiepileptic drugs, infections, and psychosocial problems (58%).

- Of the 55 enrolled adults, 18 patients (32.7%) reported availability of adult care in their area, and smooth transition from pediatric to adult care was reported by 16 patients (29.1%).

- Sixty-eight patients (47.6%) were in contact with a TSC association and 69 (48.3%) were not.

- Among the 88 children, 78 (88%) were able to complete the questionnaires independently, whereas caregivers completed the questionnaire for 10 patients who required assistance.

- Of the 55 adults, 38 were able to complete the questionnaire independently, whereas caregivers completed the questionnaire for 7 patients who required assistance.

- Among the 88 children, 11 (7.7%) were reported as having extreme anxiety/depression. On the thermometer scale of 0 to 100, (100 being the best state of health imaginable), 7.7% (4.2%) reported extreme anxiety/depression.

- Of the 55 adults, 38 were able to complete the questionnaire independently, whereas caregivers completed the questionnaire for 7 patients who required assistance.

Conclusions:

- TOSCA is the first multinational clinical study to evaluate EQ5D and QoL in patients with TSC. Overall, the results suggest that although TSC is considered a rare disease, it has a great impact on the patients' and their caregiver's health-related quality of life and overall well-being.

References:


Acknowledgments:

We would like to thank the patients and patient organizations who participated in this trial and their families and the staff at each site that assisted in the conduct of this research. This study was also supported by the National Epilepsy Center, Shizuoka Institute of Epilepsy and Neurological Disorders, NHO, 886 Urushiyama Aoi-ku Shizuoka, Japan; St. Sophia Children's Hospital, Athens, Greece; Kingswood C, et al. Eur J Paediatr Neurol 2016;20:296-308.

Figure 1. Satisfaction with Treatment Aspects (N=143)

- Figure 2. Summary of the EQ-5D Questionnaire

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