



TuberOus SCLerosis Registry to Increase disease Awareness

# Quality of Life and Burden of Disease in Tuberous Sclerosis Complex: Findings from the TOSCA Research Project

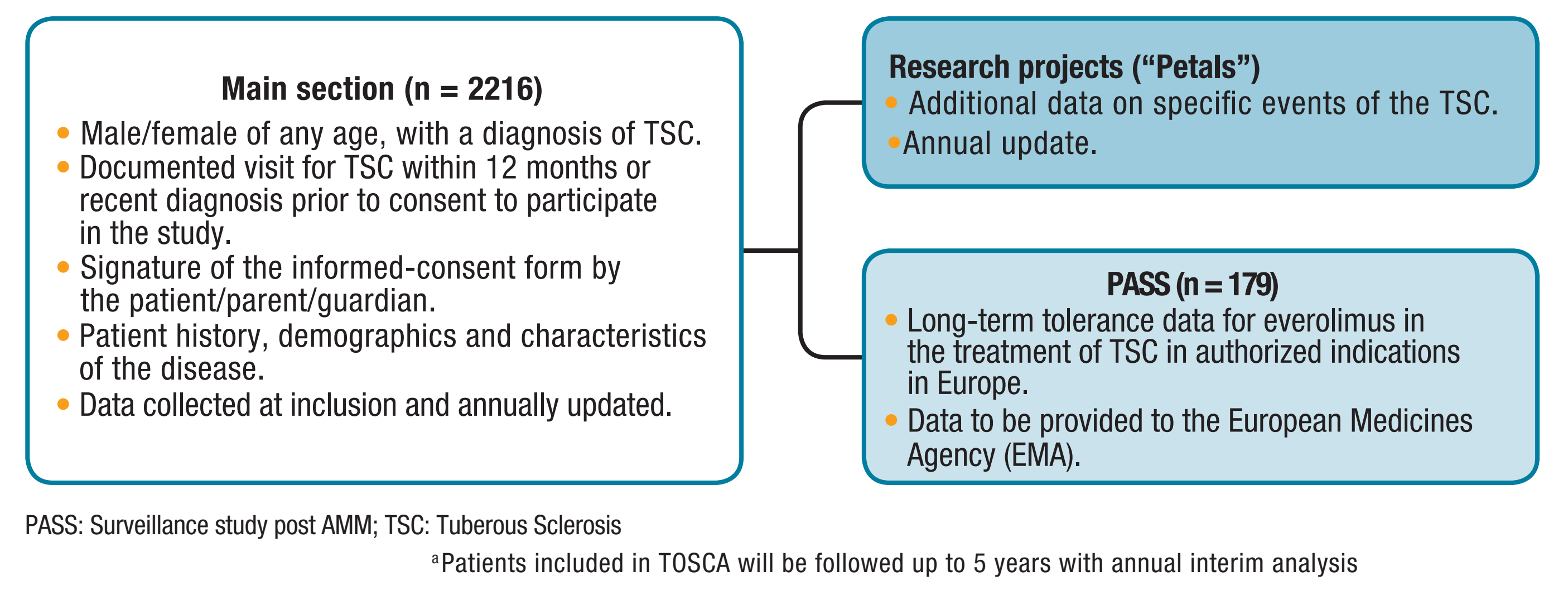
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## INTRODUCTION

- Tuberous sclerosis (TSC) is an autosomal dominant genetic disorder resulting from mutations in TSC1 and/or TSC2 genes in approximately 85% of patients<sup>1,2</sup>.
- TSC is characterized by the development of benign tumors or hamartomas in many organs such as the brain, kidneys, lungs, liver, eyes, and skin<sup>1, 2, 3</sup>.
- The major brain lesions involved in TSC are cortical tubers, subependymal nodules, giant cell subependymal astrocytomas, and abnormalities of radial white matter migration; associated with epilepsy and TAND (neuropsychiatric disorders associated with TSC)<sup>1</sup>.
- The aim of TOSCA - TuberOus SCLerosis registry to increase disease Awareness (Figure 1) is to supplement the current knowledge on the manifestations of TSC, their evolution as well as their response to treatments<sup>4</sup>.
- The additional research project *Quality of life and burden of disease in TSC* aims to assess the impact of TSC on these aspects of the lives of patients and caregivers.

Figure 1.TOSCA Registry Design<sup>a</sup>; the International Cohort Study on TSC



## RESULTS

### Population

Patients (N = 111)	
Self-assessment (n = 46)	Rating by a relative (n = 65)
→ Self-assessment of the child, n = 11	→ For a child, n = 55
→ Self-assessment of the adult, n = 35	→ For an adult, n = 10

### Patient Demographics and Clinical Features

	Children <sup>a</sup> (n = 66)	Adults <sup>a</sup> (n = 45)	Total (N = 111)
Gender, n (%)			
Male	26 (39.4)	13 (28.9)	39 (35.1)
Femal	40 (60.6)	32 (71.1)	72 (64.9)
Median age at the start of the research project, year	10,5 (3-18)	34 (18-65)	14 (3-65)
Duration of the TSCa, year	8.3 (1.6-17.7)	19.1 (1.8-41.0)	11.3 (1.6-41.0)
Patients with epilepsy, n	27	27	54
Duration of epilepsy at the beginning of the research project, year	10.8 (2.8-16.9)	29.6 (2.7-55.4)	14.0 (2.7-55.4)
Type of epilepsy <sup>b</sup> , n (%)			
Infantile spasms	19 (70.4)	7 (25.9)	26 (48.1)
Focal seizures	21 (77.8)	23 (85.2)	44 (81.5)
Infantile spasms and focal seizures	15 (55.6)	5 (18.5)	20 (37.0)
Others	11 (40.7)	11 (40.7)	22 (40.7)
Data on epilepsy, n (%)			
Controlled	18 (66.7)	22 (81.5)	40 (74.1)
Not controlled	9 (33.3)	5 (18.5)	14 (25.9)

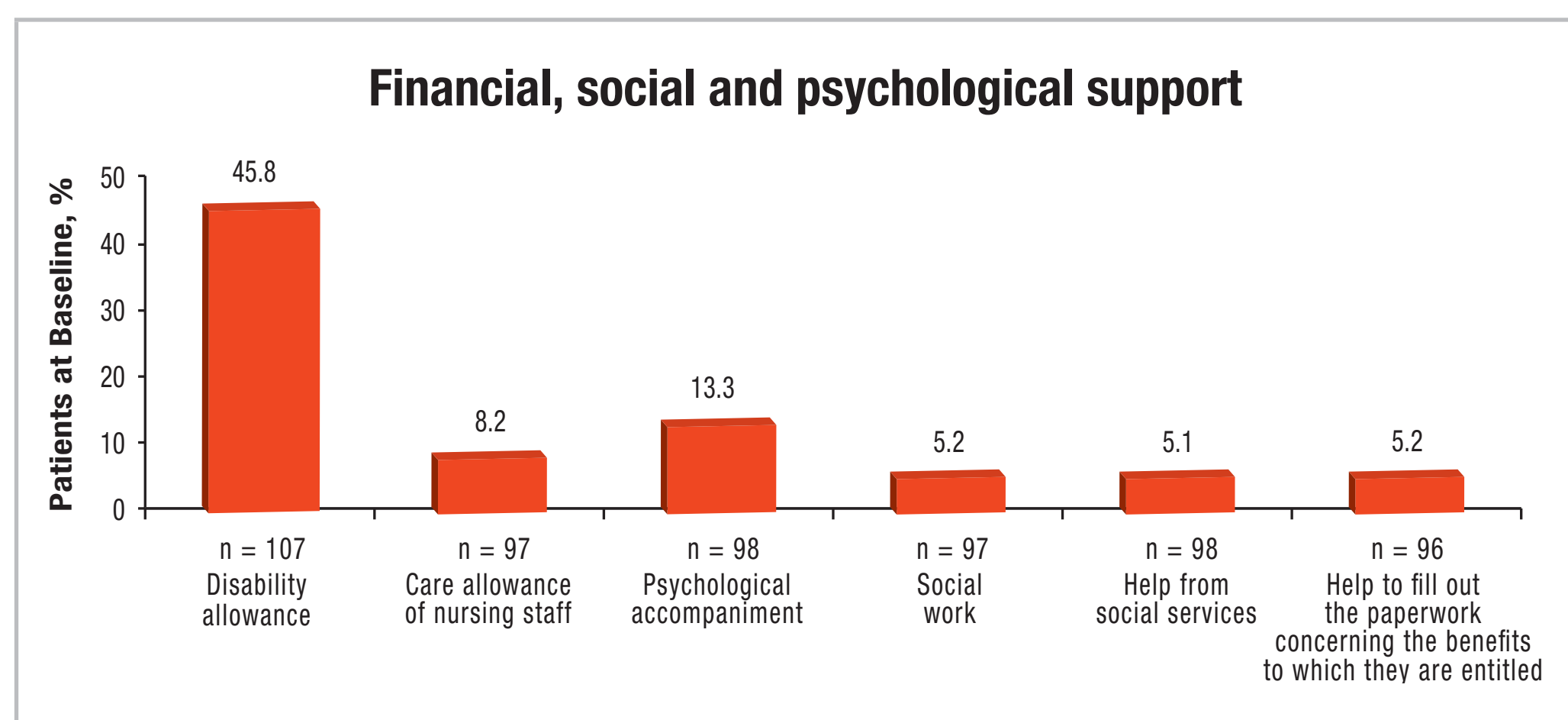
<sup>a</sup>The duration of the TSC was calculated from the date of diagnosis and the date of signature of consent for the research project

<sup>a</sup>A patient may have more than one type of seizure

<sup>a</sup>Data include self-assessments for children aged 11-17 and parent/caregiver assessments for children aged 10 years or unable to answer

<sup>a</sup>Data include adult self-assessment and parent/caregiver assessments for adults unable to respond

### TSC Burden: Assistance



#### Satisfaction of social services\*

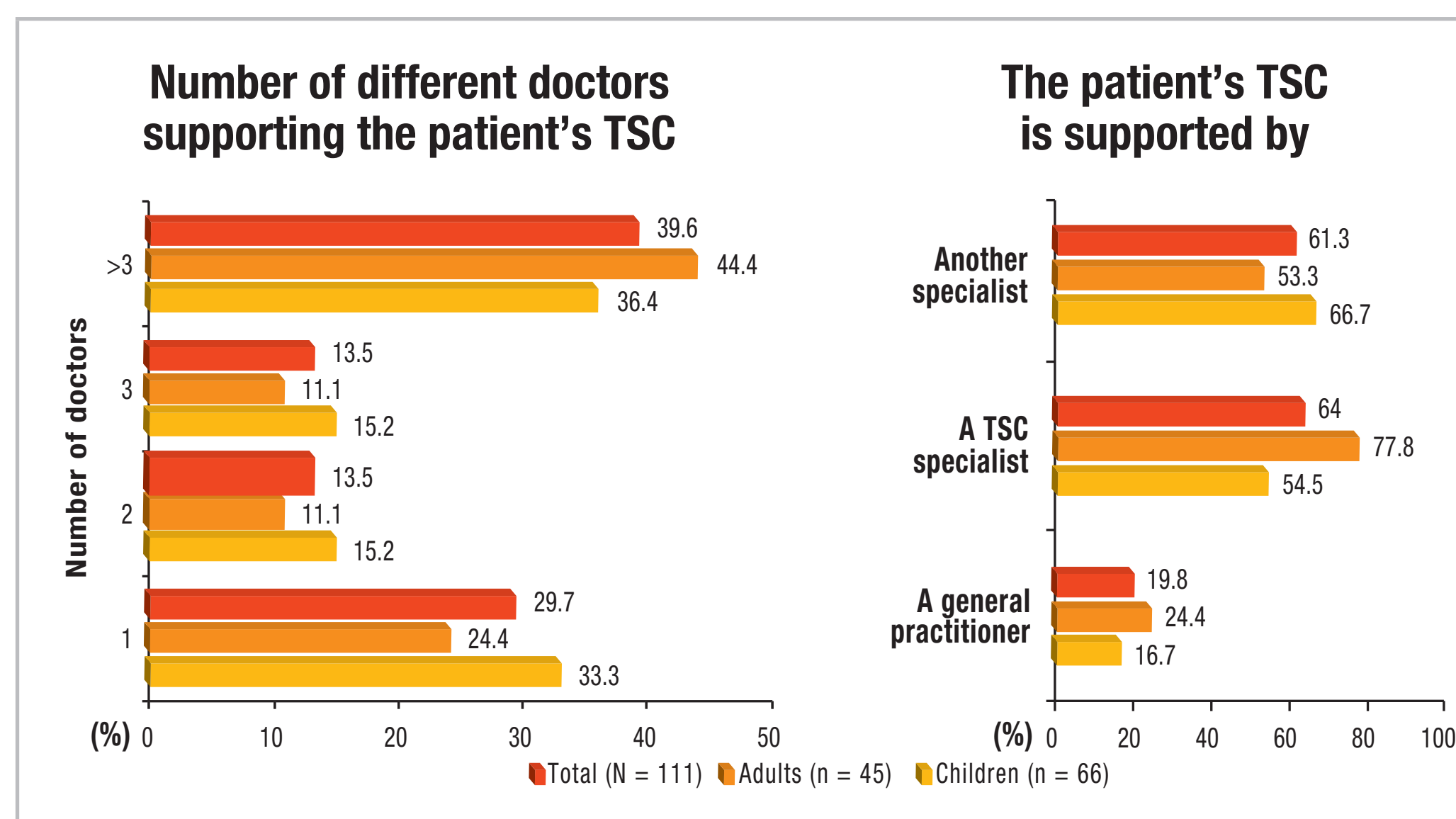
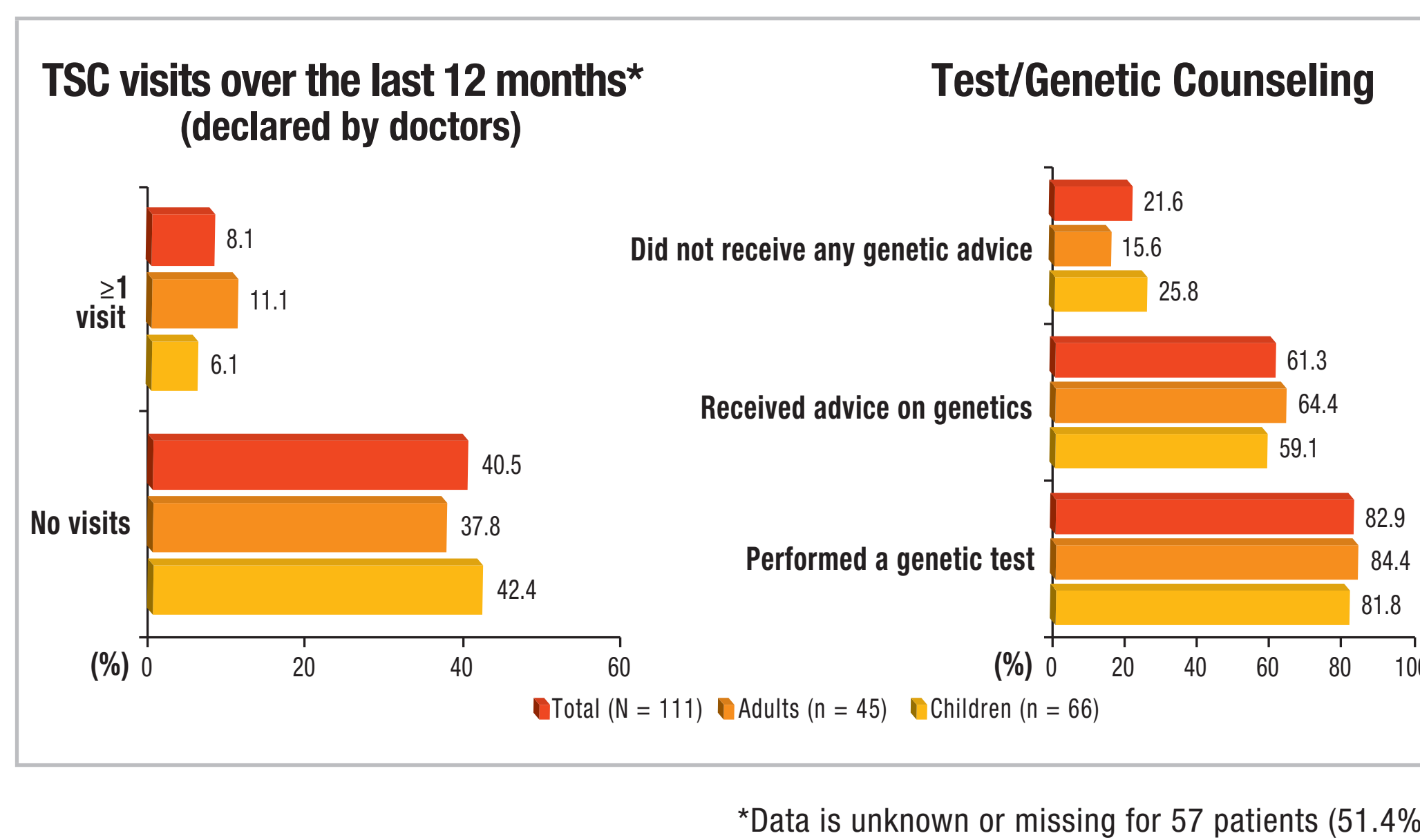
- 22.2% of adults (n = 10) and 6.1% of children (n = 4, close) are satisfied
- 40% of adults (n = 18) and 62% of children (n = 41) feel inadequately supported

#### Contact with the National TSC Patient Association

- About 40% of adults (n = 18) and 47% of children (n = 31) are in touch with the patients' association

\*13 adults (28.9%) and 16 children/relatives (24.2%) reported as "not applicable," and data are missing for 4 adults (8.9%) and 5 children/close relatives (7, 6%)

### TSC Burden: Health Needs



#### Transition from pediatrics to adults

31.1%

(14 out of 45 adults) have access to an adult care service in their area

46.7%

(21 out of 45 adults) experienced a simple transition from pediatrics to adults

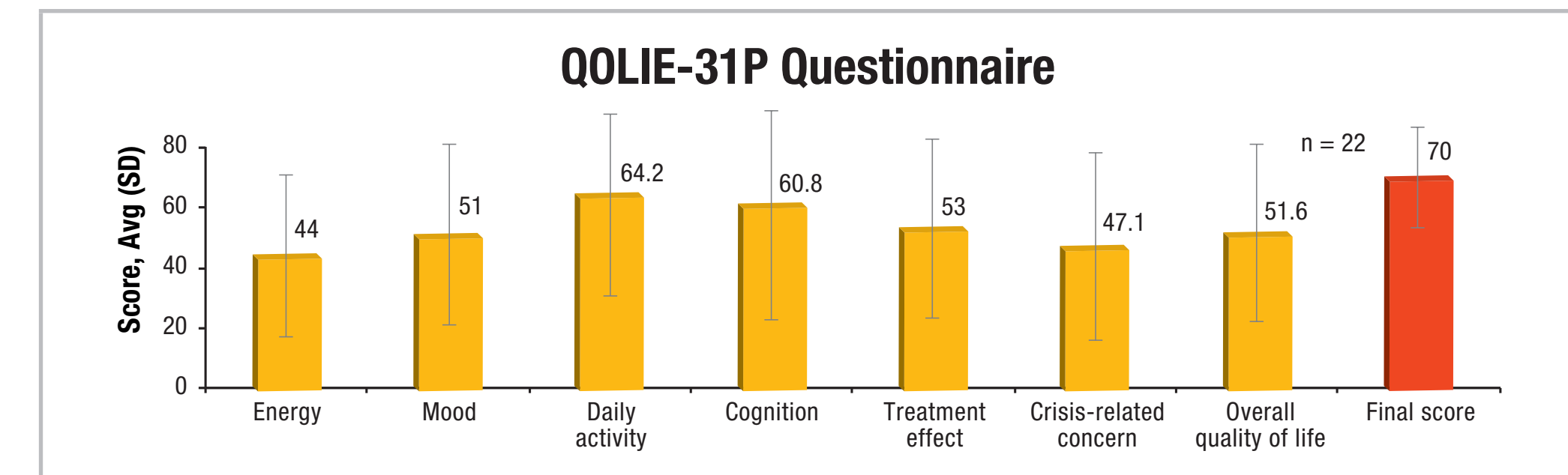
44.4%

(20 out of 45 adults) report having adequate support for their transition from pediatrics to adults

### Impact on career/education and income

	Children (n = 66)	Adults (n = 45)
<b>Impact on career or education of relatives (for children) or adults n (%)</b>		
Career Progression/Promotions	13 (19.7)	4 (8.9)
career choices	9 (13.6)	4 (8.9)
Loss of work	7 (10.6)	5 (11.1)
Part-time job rather than a full-time one	20 (30.3)	5 (11.1)
Education level reached	2 (3.0)	13 (28.9)
<b>Impact on relationships, n (%)</b>		
Familial relationship	21 (31.8)	9 (20.0)
Social relationships	28 (42.4)	22 (48.9)
Relationships with co-workers	10 (15.2)	5 (11.1)
<b>Average expenditure per year (in Euro)</b>		
Medicines	352.0 (762.20)	312.7 (538.03)
Round trip to the hospital for the TSC	269.4 (439.59)	202.5 (331.22)
Examinations/Analysis related to the TSC	205.7 (666.46)	138.9 (218.95)

### QOLIE-31P Questionnaire



### QOLCE Questionnaire

	Number of children with available data, n	QOLCE score, avg (SD)
Quality of life	50	48.5 (27.39)
Physical restriction	52	41.6 (24.77)
Energy/fatigue	50	54.0 (23.07)
Depression	51	68.8 (18.66)
Anxiety	50	58.1 (20.13)
Master/impotence	47	56.6 (18.83)
Self-esteem	47	63.1 (20.05)
Attention/concentration	49	35.6 (26.68)
Memory	41	53.4 (21.23)
Language	43	40.8 (26.61)
Other cognitive difficulties	47	28.5 (26.10)
Social interactions	40	52.9 (24.54)
Social activities	50	58.7 (37.00)
Shame	40	63.1 (37.55)
Behavior	52	48.5 (20.80)
General health	51	44.6 (26.61)
<b>Final score</b>		<b>50.2 (19)</b>

## WRAP UP

- These findings indicate that TSC has a significant impact on the health and social well-being of patients and their families.
- In general, patients are satisfied with the way their disease is managed. However:
  - There is always a need to improve communication between specialists and the organization of hospital visits
  - Very few patients received support from social services, a social worker or a psychologist
  - About 1/3 of patients were followed up by a TSC specialist
  - Genetic testing is done in about 80% of patients, but only 26% have benefited from genetic counseling
  - Up to 40% of families are not in touch with the national TSC patient association
- This can be significantly improved through educational, social and medical support.

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