



Steve Roberds, PhD
Chief Scientific Officer
Tuberous Sclerosis Alliance
801 Roeder Road, Suite 750
Silver Spring, MD 20910
United States of America

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Dear Steve,

I hope this letter finds you well. I am writing to share the results of our TSC Family Survey conducted during 2014. This was not a formal research project but used by our organisation to better understand our constituents and will influence our work. I hope these results are informative to the individuals involved in the upcoming workshop to update the research plan for Tuberous Sclerosis.

As you know Tuberous Sclerosis Australia is the only Tuberous Sclerosis focussed organisation in Australia and represents the thousands of individuals with TSC in Australia and their families. Australian researchers have participated in several international clinical trials and our organisation has also funded some smaller projects.

The family survey was conducted by Tuberous Sclerosis Australia and promoted only through our existing contact database. The survey was answered by 88 individuals with TSC and their caregivers living around Australia in both urban and regional areas.

One part of the survey explored which aspects of Tuberous Sclerosis most significantly affected the respondents. Our analysis of this data revealed that **TSC associated neuropsychiatric disorders (TAND, includes behaviour, mental health and cognition and learning challenges) are a major concern for families. Epilepsy and control of seizures was also identified as an area of significant impact.**

I encourage the workshop attendees to ensure the research plan for Tuberous Sclerosis appropriately prioritises both TAND and epilepsy. I have included more information about our survey and more detailed results on the following pages. I am happy to be contacted for any further information and I wish you and all the attendees well with the workshop.

Yours Sincerely,

Clare Stuart
General Manager
clare@tsa.org.au
+61 410 685 181



Except from survey results - Part 2: How does TSC affect them?

Results

- The respondents were provided with a list of conditions commonly associated with TSC. They were asked to report if these affected themselves or the person they cared for. The choices were “yes”, “no” or “I don’t know”. Table 1 shows each condition with the most common at the top:

	Number of respondents indicating “Yes”	% of respondents
Skin problems	77	88%
Epilepsy	70	80%
Learning difficulties	66	75%
Kidney tumours or cysts	63	72%
Intellectual disability	56	64%
Challenging behaviour	51	58%
Other (e.g. eyes, liver)	42	48%
Autism	37	42%
SEGA brain tumour	29	33%
Mental Health issues	22	25%
Mobility problems	21	24%
ADHD	14	16%
Lung issues (e.g. LAM)	9	10%

Table 1: Reported frequency of conditions associated with TSC

- Figure 7 shows the frequency of each condition in the order they were asked:

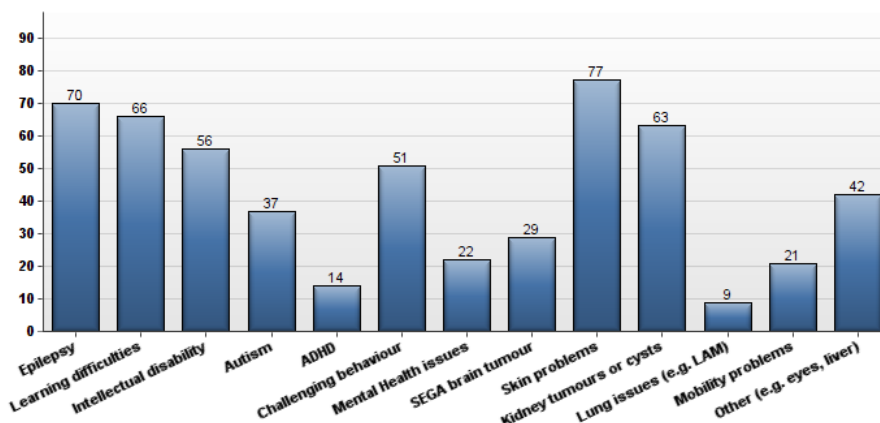


Figure 1: Reported frequency of conditions associated with TSC



- The next question asked respondents to report on the impact that these conditions had on their lives. The full results are shown in figure 8.

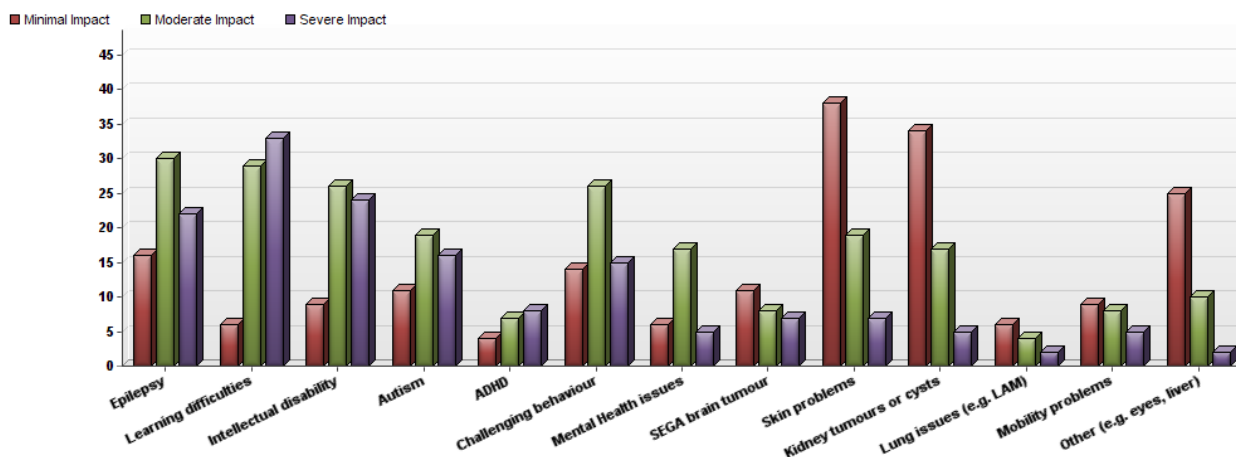


Figure 2: Impact of TSC associated conditions on respondents' lives

- The conditions that were reported most as having severe impact were:
 - Learning difficulties
 - Intellectual disability
 - Epilepsy
 - Autism
 - Challenging behaviour
- This section also asked respondents to describe how TSC affects them. 65 out of 88 respondents (74%) provided an open text response. This qualitative data has been analysed for themes Clare Stuart, who has both a personal and professional experience of TSC. The common themes in the responses are shown in table 2.



Theme	Frequency
TSC Associated Neuropsychiatric disorders (TAND) including behaviour, developmental delays, mental health and education.	26
Epilepsy and seizures	17
Socialising	12
Fear of what will happen in the future	10
Access to medical specialists knowledgeable in TSC	10
Kidneys	8
Financial burden of TSC	8
Time taken for medical appointments	8
Impact of TSC on family relationships	7
Lack of independence	7
SEGA brain tumour	5
Skin conditions associated with TSC	5
Other medical aspects of TSC	5
Identifies as being mildly affected by TSC	4
Genetics and family planning	4

Table 2: Themes identified in open text responses to how TSC impacts them

- Open text that were representative of some of these themes were:

My son doesn't have a diagnosis of autism, however his behaviour is definitely on the spectrum. His health is great at the moment and his epilepsy controlled with medication. He is driving and holding down two permanent part time jobs. My biggest concern is his social interactions, in the home, at work and with minimal friends. I worry about what the future holds and whether he will be ever able to independent. I worry that he will be lonely.

Parents of a teenager with TSC

Our son is 'mildly' affected by TSC. The biggest impact is psychological, emotional and behavioural. The physical (skin) symptoms and the regular doctors tests make our son aware he is "different"; which doesn't help with the anxiety that comes with his condition.

To all others he seems normal except for funny looking skin and this makes it hard to explain to others and to him, his immaturity, his anxiety, his inattentiveness, learning difficulties, bad behaviour and angry outbursts. Managing his behaviour has had the biggest impact on our lives and in particular the lives of his siblings who find it very hard to cope. Getting assistance with this aspect of his condition has been very very difficult.

Elderly parents of an adult with TSC