

Adult Rubinstein-Taybi Syndrome Patient Input for Management Guidelines

INTRODUCTION

We are the European Reference Network ERN-ITHACA (see [About ERN ITHACA - ERN ITHACA \(ern-ithaca.eu\)](#) and we are collecting information about adult individuals (>18 years old) with Rubinstein-Taybi syndrome (RTS). We aim to develop guidelines for the management of their specific needs. We would be grateful if you would fill in the following questionnaire!

- None of these questions are required but your complete responses will be of great help
- As it is most likely that a caregiver may answer the questions, we worded these accordingly
- There should be only one respondent from each family
- Your submission is anonymous, but if you want to include your email in the “Final Comments” Question number 63, at the end of this questionnaire, we will be able to contact you in case we have any questions
- Please review your answers before submitting them, to ensure they are as complete and correct as possible

A. GENERAL QUESTIONS

1. Are you a caregiver or a patient? (*one option possible*)
 - a. Caregiver
 - b. Patient
2. If you replied 'Caregiver' in Question 1, could you specify (for example parent, doctor etc)
_____ (*free text answer*)
3. How old is the person with RTS?
_____ (*free text answer*)
4. Where do you live (please specify country only)?
_____ (*free text answer*)
5. Where does the person with RTS live? (*multiple options possible*)
 - a. At home, with their main caregiver full-time (for example family)
 - b. In supported living services: a living arrangement with support from carers for everyday tasks
 - c. On their own/Independently
 - d. Other
6. If you replied 'Other' in Question 5, could you specify?
_____ (*free text answer*)
7. At what age was the person with RTS diagnosed? (*one option possible*)
 - a. Prenatally (before birth)
 - b. Newborn/Infancy (less than 1 year of age)
 - c. 1-5 years old
 - d. 6-10 years old
 - e. Older than 10 years old
 - f. I do not know
8. Has the RTS diagnosis been confirmed with genetic testing? (*one option possible*)
 - a. Yes, CREBBP – related
 - b. Yes, EP300 – related
 - c. No
 - d. I do not know
 - e. Test in progress/awaiting results
 - f. Other
9. If you replied 'Yes, CREBBP-related' or 'Yes, EP300' or 'Other' in Question 8, could you specify (for example, if known to you, you can write here the specific gene change)
_____ (*free text answer*)
10. What is the gender of the person with RTS? (*one option possible*)
 - a. Female

- b. Male
 - c. Other
 - d. Prefer not to answer
11. Does the person with RTS have any of the following medical problems that have impacted their quality of life in adulthood? (*multiple options possible*)
- a. Yes, behavioral/psychiatric problems
 - b. Yes, gastrointestinal problems
 - c. Yes, skin problems
 - d. Yes, sleep problems
 - e. Yes, other problems (there will be room to explain later in the questionnaire)
 - f. No, none of the above

B. GASTROINTESTINAL PROBLEMS

12. If the person with RTS currently has or has had gastrointestinal problems in adulthood, can you find them in the list below? (*multiple options possible*)

- a. Reflux
- b. Constipation
- c. Abdominal migraine
- d. Blood in stool
- e. Chronic diarrhea
- f. Diverticulitis / Diverticulosis
- g. Eosinophilic esophagitis
- h. Esophageal varices
- i. Gallstones
- j. Hirschsprung disease
- k. Inflammatory Bowel Disease (i.e. Crohn's, Ulcerative Colitis, etc)
- l. Irritable bowel disease
- m. Liver fibrosis
- n. Liver problems
- o. Malrotation
- p. Pancreatitis
- q. Recurrent/Cyclic vomiting
- r. Small Bowel Obstruction
- s. Unsure
- t. Other

13. If you replied 'Constipation' in Question 12, how frequently are stools coming typically in a week?

_____ (*free text answer*)

14. If you replied 'Constipation' in Question 12, what is the person with RTS using to manage their constipation? (*multiple options possible*)

- a. A specific diet
- b. Medications
- c. Other

15. If you chose one or more options in Question 14, could you specify (for example what kind of diet etc)

_____ (*free text answer*)

16. If you replied 'Constipation' in Question 12, was a specific management/treatment more helpful than others and if so, which one?

_____ (*free text answer*)

17. If you replied 'Other' in Question 12, could you specify?

_____ (*free text answer*)

C. SKIN PROBLEMS

18. If the person with RTS currently has or has had skin problems in adulthood, can you find them in the list below? (*multiple options possible*)

- a. Heavy or hypertrophic scars that can occur after an injury to the skin, also known as keloids
- b. Abnormal scarring of other type
- c. Poor wound healing
- d. Increased skin infections
- e. Ingrowing or infections of their fingernails or toenails that require treatment with medication
- f. Very dry skin
- g. Eczema
- h. A yellowish or greenish pigmentation of the skin and, sometimes, of the white of the eyes, also known as jaundice
- i. Swelling of hands and feet
- j. A purplish or reddish, slightly elevated area of skin made up of blood vessels, also known as hemangioma
- k. A pilomatricoma, sometimes called a pilomatrixoma; a rare, noncancerous tumor that grows in hair follicles. It looks and feels like a hard lump on the skin. It is most common on the head and neck, but can appear anywhere on the body.
- l. Other
- m. Unsure
- n. No

19. If you replied 'Heavy or hypertrophic scars that can occur after an injury to the skin, also known as keloids' in Question 18, is there itching of the keloids?

- a. Yes
- b. No

20. If you replied 'Yes' in Question 19, does the itching have influence on daily life?

- a. Yes
- b. No

21. If you replied 'Yes' in Question 19, how has the itching been managed or treated?

(*multiple options possible*)

- a. Surgery
- b. Laser
- c. Steroids
- d. Other medication
- e. Other management

22. If you chose one or more options in Question 21, could you specify (for example which medication etc)

_____ (*free text answer*)

23. If you chose one or more options in Question 21, was a specific management more helpful than others and if so, which one?

_____ (*free text answer*)

24. If you replied 'Other' in Question 18, could you specify?
_____ (*free text answer*)

D. SLEEP PROBLEMS

25. If the person with RTS currently has or has had sleep problems in adulthood, can you find them in the list below? (*multiple options possible*)
- a. Difficulty falling asleep
 - b. Difficulty staying asleep/awaking frequently during the night
 - c. Increased need for sleep
 - d. Falling asleep suddenly during the day/Narcolepsy
 - e. Breathing repeatedly stops and starts during sleep. Or the person snores loudly and feels tired even after a full night's sleep. May also be known as Sleep Apnea
 - f. Unpleasant or uncomfortable sensations in the legs and an irresistible urge to move them. Symptoms commonly occur in the late afternoon or evening hours, and are often most severe at night when a person is resting, such as sitting or lying in bed. Can also be known as Restless Legs Syndrome or Periodic Limb Movement Disorder
 - g. Other
 - h. Unsure
 - i. No
26. If you replied 'Increased need for sleeping' in Question 25, which one of the options below best describes the problem? (*multiple options possible*)
- a. Increased nighttime sleeping
 - b. Increased daytime sleeping
 - c. Increased need to sleep immediately after lunch
 - d. Mood change if not allowed to sleep
 - e. Other
27. If you replied 'Other' in Question 26, could you specify?
_____ (*free text answer*)
28. If you replied 'Other' in Question 25, could you specify ?
_____ (*free text answer*)

E. BEHAVIORAL OR PSYCHIATRIC PROBLEMS

29. If the person with RTS currently has or has had behavioral/psychiatric problems in adulthood, can you find them in the list below? (*multiple options possible*)

- a. Aggression toward others
- b. Anxiety
- c. Autism Spectrum Disorder
- d. Depression
- e. Difficulty maintaining attention, hyperactivity and impulsive behaviour. May also be known as Attention Deficit Hyperactivity Disorder (ADHD)
- f. High levels of frustration
- g. Limited interests and repetition (routine/words/order of doing things)
- h. Loss of interest in things that used to interest them
- i. Loss of skills that they used to be able to perform
- j. Mood abnormalities or disorder/sudden mood changes
- k. Recurring, unwanted thoughts, ideas or sensations (obsessions) that make them feel driven to do something repetitively (compulsions), also known as Obsessive Compulsive Disorder (OCD)
- l. Self-harm/Aggression toward themselves
- m. Sudden onset of reduced activity
- n. Other
- o. Unsure
- p. No

30. If you replied 'Other' in Question 29, could you specify?

_____ (*free text answer*)

31. When did the behavioral/psychiatric problems start? (*multiple options possible*)

- a. During childhood
- b. During puberty
- c. In adulthood

32. If you chose more than one option in Question 29 and the problems started at different times each, could you specify?

_____ (*free text answer*)

33. How were the behavioral/psychiatric problems managed? (*multiple options possible*)

- a. Change of environment
- b. Psychotherapy
- c. Medications

34. If you chose more than one option in Question 29 and each problem was managed differently, could you specify?

_____ (*free text answer*)

35. Was a specific management more helpful than others and if so, which one and for which behavioral problem?

_____ (*free text answer*)

36. Which of the behavioral problems are the most important for the person with RTS?

_____ (*free text answer*)

37. Which of the behavioral problems are influencing most the life of the person with RTS?

_____ (*free text answer*)

F. OTHER PROBLEMS

38. Are there other problems that the adult person with RTS has? (*multiple options possible*)

- a. Binge eating
- b. Cancer
- c. Decreased mobility/any difficulties with moving around
- d. Diabetes
- e. Hearing difficulties or loss
- f. High pain threshold
- g. Hypersensitivity to noise and crowded places
- h. Hypertension or other problems with the blood vessels and the heart
- i. Menopause
- j. Osteoporosis
- k. Vision difficulties or loss
- l. Other

39. If you replied 'Other' in Question 38, could you specify?

_____ (*free text answer*)

E. SURGERY

40. Has the patient with RTS had any surgery during adulthood? (*one option possible*)

- a. Yes
- b. No

41. If you replied 'Yes' in Question 40, what was the original problem?

_____ (*free text answer*)

42. If you replied 'Yes' in Question 40, how many times has the adult person with RTS had surgery?

_____ (*free text answer*)

43. If you replied 'Yes' in Question 40, what type of surgery did the person have?

_____ (*free text answer*)

44. If you replied 'Yes' in Question 40, did the surgery resolve the issue?

_____ (*free text answer*)

45. Do you have any other comments about surgery in an adult person with RTS?

_____ (*free text answer*)

E. EVERYDAY LIFE

46. Does the adult RTS person you are/are aware of work? (*one option possible*)
- Yes
 - No
47. If you replied 'Yes' in Question 46, could you specify what work the person does?
_____ (*free text answer*)
48. Does the adult RTS person you are/are aware of maintain social activities? (*one option possible*)
- Yes
 - No
49. If you replied 'Yes' in Question 48, could you specify?
_____ (*free text answer*)
50. Does the adult RTS person you are/are aware of drive a car? (*one option possible*)
- Yes
 - No
51. Is the adult RTS person you are/are aware of able to take transportation independently? (*one option possible*)
- Yes
 - No
52. Is the adult RTS person you are/are aware of able to do grocery shopping independently? (*one option possible*)
- Yes
 - No
53. A question caregivers and parents often ask is whether individuals with RTS have been sexually active. We would be grateful if you would consider answering whether the adult RTS individual you are/know has been sexually active (*one option possible*)
- Yes
 - No
 - I do not know
 - Prefer not to answer
54. If you replied 'Yes' in Question 53, could you specify? (*multiple options possible*)
- Stimulation of one's own genitalia (masturbation)
 - With a partner
 - Other
55. If you replied 'Other' in Question 54, could you specify?
_____ (*free text answer*)
56. Are there other problems that the adult person with RTS has that you would like us to know about?
_____ (*free text answer*)

F. MEDICAL FOLLOW-UP

57. Who is following up the medical issues of the adult person you are/are aware of with RTS?

- a. The general practitioner
- b. A specialist in the hospital
- c. A reference center where I/he/she are seen by many specialists
- d. Other
- e. No one. I/he/she has not seen any doctor in adult life

58. If you replied 'A specialist in the hospital' in Question 57, could you specify which specialty?

_____ (free text answer)

59. If you replied 'A reference center' in Question 57, could you specify which center?

_____ (free text answer)

60. If you replied 'Other' in Question 57, could you specify?

_____ (free text answer)

61. How often does the adult person you are/are aware of with RTS have health checks?

- a. Once a year
- b. Twice a year
- c. Once every 3 years
- d. Once every 5 years
- e. Only if there are specific concerns
- f. Other

62. If you replied 'Other' in Question 61, could you specify?

_____ (free text answer)

63. Any other, final comments?

_____ (free text answer)