

Medical Issues in adults with CHARGE syndrome

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This questionnaire is intended for:

- Adults (18 years or older) with CHARGE syndrome who are able to complete the questionnaire themselves.

OR

- A parent or legally authorized substitute decision-maker completing the questionnaire on behalf of an adult (18 years or older) with CHARGE syndrome who is unable to complete the questionnaire independently.

Thank you for participating. The purpose of this questionnaire is to better understand the medical experiences and healthcare needs of adults with CHARGE syndrome. The questionnaire is anonymous. Please answer each question as accurately as possible. You may skip any question you do not wish to answer.

Warm regards,

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Claudia Junghans – 1. President CHARGE Syndrom e.V., Germany

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Demographic Information

1. Gender of the person with CHARGE

- ☐ Female
- ☐ Male
- ☐ Other
- ☐ Prefer not to say

2. Age group

Please check one

- ☐ 18-29
- ☐ 30-39
- ☐ older than 40

3. Country of residence

4. Who is completing this questionnaire?

- ☐ Person with CHARGE Syndrome
- ☐ Parent
- ☐ legal substitute decision maker

CHARGE Syndrome Eligibility Questions

You can check more than 1 box depending on your situation.

5. Has a doctor or healthcare provider ever told you that you (or the person you care for) have CHARGE syndrome?

- ☐ Yes
- ☐ No
- ☐ Not sure

6. Was the diagnosis confirmed with genetic testing (for example, a change/mutation in the CHD7 gene)?

- ☐ Yes
- ☐ No
- ☐ Not sure

7. Which of the following CHARGE syndrome features have been diagnosed?

- ☐ Coloboma (eye abnormality)
- ☐ Heart defect
- ☐ Choanal atresia/narrow nasal passages
- ☐ Short stature
- ☐ Genital differences (a small penis, undescended testicles, or differences of the labia)
- ☐ Ear abnormalities or hearing loss
- ☐ Balance problems
- ☐ Feeding/swallowing difficulties
- ☐ Cranial nerve problems

Medical Questions

Please check all boxes that apply to your medical or health issues.

You can click more than one box depending on your situation.

For this questionnaire, "adolescence" refers to the period from 18 years of age onwards. Please answer questions based on your health from age 18 to the present.

8. Vision and Eye Health:

Do you have vision problems?

- ☐ Yes
- ☐ No
- ☐ Not sure

9. Since adolescence, how has your vision changed?

- ☐ Improved
- ☐ Stayed about the same
- ☐ Gradually worsened / new vision problems developed

10. Describe all vision issues and the age at which they worsened or you developed new symptoms.

11. When was your last eye examination?

- ☐ Within the past year
- ☐ 1–2 years ago
- ☐ More than 2 years ago
- ☐ Never / unsure

12. Hearing status:

Which best describes your hearing?

- ☐ No hearing loss
- ☐ Mild hearing loss
- ☐ Moderate hearing loss
- ☐ Severe hearing loss
- ☐ Deaf in one ear
- ☐ Deaf in both ears
- ☐ Other

13. Since adolescence, how has your hearing changed?

- ☐ Improved
- ☐ Stayed about the same
- ☐ Gradually worsened / new hearing problems developed

14. What hearing supports do you currently use?

- ☐ Hearing aids
- ☐ Cochlear implant
- ☐ Bone conduction device
- ☐ None
- ☐ Other

15. Describe all hearing issues and the age at which they worsened or you developed new symptoms.

16. When was your last hearing assessment.

- ☐ Within the past year
- ☐ 1–2 years ago
- ☐ More than 2 years ago
- ☐ Never / unsure

17. What is your primary form of communication?

- ☐ Spoken speech
- ☐ Sign language (visual or tactile)
- ☐ AAC or symbol-based communication (e.g., device, app, picture system)
- ☐ Gestures/body language
- ☐ Mixed methods (no single main form)
- ☐ Other

18. Since adolescence (approximately age 18), have you noticed any changes in the way you communicate?

- ☐ Yes
- ☐ No
- ☐ Not sure

19. If yes, please describe the changes in your communication and indicate approximately when you first noticed them.

20. Do you have facial palsy?

- ☐ Yes
- ☐ No
- ☐ Not sure

21. Since adolescence, how has your facial palsy or facial weakness changed?

- ☐ Improved
- ☐ Stayed about the same
- ☐ Gradually worsened / new symptoms developed

22. Describe facial palsy issues and the age at which they worsened or you developed new symptoms, and note if surgical treatment was received.

23. Heart:

Do you have heart conditions?

- ☐ Yes
- ☐ No
- ☐ Not sure

24. Since adolescence, how have your heart-related conditions changed?

- ☐ Improved
- ☐ Stayed about the same
- ☐ Gradually worsened / new symptoms developed

25. Describe heart issues and the age at which they worsened or you developed new symptoms.

26. Feeding (including swallowing and reflux):

Do you have feeding or swallowing difficulties, including reflux?

- ☐ Yes
- ☐ No
- ☐ Not sure

27. Since adolescence, your feeding or swallowing issues have:

- ☐ Improved
- ☐ Stayed about the same
- ☐ Gradually worsened / new symptoms developed

28. Describe feeding or swallowing issues and any devices or treatment used and the age at which they worsened or you developed new symptoms.

29. Stomach, gut and constipation:

Do you have stomach, gut, or bowel problems (for example constipation, abdominal pain, dumping syndrome, cyclical vomiting, or bowel motility issues)?

- ☐ Yes
- ☐ No
- ☐ Not sure

30. If yes, how have these stomach, gut, or bowel problems changed since adolescence?

- ☐ Improved
- ☐ Stayed about the same
- ☐ Gradually worsened / new symptoms developed

31. Describe the stomach, gut or bowel issues and the age at which they worsened or you developed new symptoms.

32. Secretions:

Do you have problems with excessive or difficult-to-manage secretions (for example, drooling, thick saliva, frequent mucus, difficulty clearing secretions, or frequent suctioning)?

- ☐ Yes
- ☐ No
- ☐ Not sure

33. How have these secretion-related problems changed since adolescence?

- ☐ Improved
- ☐ Stayed about the same
- ☐ Gradually worsened / new symptoms developed

34. Describe secretion-related issues and the age at which they worsened or you developed new symptoms.

35. Breathing, Airway, and Nasal Health (Including Choanal Atresia):

Do you have any breathing, airway, or nasal problems (frequent chest infections or pneumonia, choanal atresia, airway narrowing, or a tracheostomy)?

- ☐ Yes
- ☐ No
- ☐ Not sure

36. If yes, how have these breathing or airway problems changed since adolescence?

- ☐ Improved
- ☐ Stayed about the same
- ☐ Gradually worsened / new symptoms developed

37. Describe breathing or airway issues and the age at which they worsened or you developed new symptoms.

38. Sleep apnea:

Do you have sleep-related breathing problems (such as sleep apnea, loud snoring, or breathing pauses during sleep)?

- ☐ Yes
- ☐ No
- ☐ Not sure

39. How have these sleep-related breathing problems changed since adolescence?

- ☐ Improved
- ☐ Stayed about the same
- ☐ Gradually worsened / new symptoms developed

40. Describe the sleep-related breathing issues and the age at which they worsened or you developed new symptoms.

41. Upper Tract Infections:

Do you have frequent or recurrent upper tract infections (for example, sinus infections, ear infections or postnasal drip)?

- ☐ Yes
- ☐ No
- ☐ Not sure

42. How have these infections changed since adolescence?

- ☐ Improved
- ☐ Stayed about the same
- ☐ Gradually worsened / new symptoms developed

43. Describe upper tract infections-related issues and the age at which they worsened or you developed new symptoms

44. Kidneys (i.e. urinary tract issues/infections):

Do you have any kidney or urinary tract problems (for example kidney abnormalities, urinary tract infections, reflux up the ureters, or difficulty emptying your bladder)?

- ☐ Yes
- ☐ No
- ☐ Not sure

45. How have the kidney or urinary tract problems changed since adolescence?

- ☐ Improved
- ☐ Stayed about the same
- ☐ Gradually worsened / new symptoms developed

46. Describe kidney or urinary issues and the age at which they worsened or you developed new symptoms.

47. Spine problems:

Do you have spine problems (such as scoliosis, kyphosis, abnormal posture, or spinal curvature)?

- ☐ Yes
- ☐ No
- ☐ Not sure

48. How have the spine problems changed since adolescence?

- ☐ Improved
- ☐ Stayed about the same
- ☐ Gradually worsened / new symptoms developed

49. Describe spine issues, the type of spine management you received and the age at which they worsened or you developed new symptoms.

50. Balance and Mobility:

Do you have balance or mobility problems (for example poor balance, frequent falls, delayed motor skills, dizziness, or difficulty walking)?

- ☐ Yes
- ☐ No
- ☐ Not sure

51. How have the balance or mobility problems changed since adolescence?

- ☐ Improved
- ☐ Stayed about the same
- ☐ Gradually worsened / new symptoms developed

52. Describe the balance or mobility issues, any mobility aids used and the age at which they worsened or you developed new symptoms.

53. Osteoporosis:

Do you have Osteoporosis?

- ☐ Yes
- ☐ No
- ☐ Not Sure

54. If yes, have you had a bone density (DEXA) scan?

- ☐ Yes
- ☐ No
- ☐ Not Sure

55. If you have osteoporosis, how old were you at the time of diagnosis?

56. If you have osteoporosis, what treatment have you received (e.g calcium supplements, Vitamin D (include mg/IU), bisphosphonates (specify which ones).

57. Growth hormones?

Were growth hormones given (daily injections given for short stature)?

- ☐ Yes
- ☐ No

58. If yes, at what age did you start receiving growth hormones?

59. Are you still using growth hormones?

- ☐ Yes
- ☐ No
- ☐ Not sure

60. If no, at what age did you stop using growth hormones?

61. Sex Hormones:

Did you receive hormone treatment (for example testosterone or estrogen therapy)?

- ☐ Yes
- ☐ No
- ☐ Not sure

62. If known at what age did treatment start?

63. Are you currently receiving any sex hormone treatment as an adult?

- ☐ Yes
- ☐ No
- ☐ Not sure

64. If no, at what age did the treatment stop?

65. Do you have any additional comments about your endocrine health (including an absent or reduced sense of smell)?

66. Seizures:

Do you ever experience seizures or be diagnosed with a seizure disorder?

- ☐ Yes
- ☐ No
- ☐ Unsure

67. If yes, how have your seizures changed since adolescence?

- ☐ Improved
- ☐ Stayed about the same
- ☐ Gradually worsened / new symptoms developed

68. Describe the seizures and the age at which they worsened or you developed new symptoms.

69. Postural Orthostatic Tachycardia Syndrome (POTS):

Were you diagnosed with Postural Orthostatic Tachycardia Syndrome (POTS)

- ☐ Yes
- ☐ No
- ☐ Not sure

70. If yes, how have POTS or autonomic symptoms changed since adolescence?

- ☐ Improved
- ☐ Stayed about the same
- ☐ Gradually worsened / new symptoms developed

71. Describe the POTS symptoms, any treatments received and the age at which they worsened, or you developed new symptoms.

72. Pain:

Do you experience recurrent pain (for example migraines, muscle or joint pain, abdominal pain, or other chronic pain)?

- ☐ Yes
- ☐ No
- ☐ Not sure

73. If yes, how has your pain changed since adolescence?

- ☐ Improved
- ☐ Stayed about the same
- ☐ Gradually worsened / new symptoms developed

74. Describe the pain issues and the age at which they worsened or you developed new symptoms.

75. Dental problems:

Do you have any dental or oral health problems (for example missing or malformed teeth, delayed tooth eruption, enamel defects, frequent cavities, gum problems, or jaw issues)?

- ☐ Yes
- ☐ No
- ☐ Not sure

76. How have your dental or oral health problems changed since adolescence?

- ☐ Improved
- ☐ Stayed about the same
- ☐ Gradually worsened / new symptoms developed

77. Describe dental issues and the age at which they worsened or you developed new symptoms.

78. Fatigue:

Do you experience ongoing fatigue or low energy levels?

- ☐ No fatigue
- ☐ Mild fatigue – noticeable but does not interfere with daily activities
- ☐ Moderate fatigue – interferes with some daily activities
- ☐ Severe fatigue – significantly limits daily activities
- ☐ Not sure

79. If fatigue has worsened or newly appeared since adolescence, please describe the changes and the age at which they began.

80. Please list and describe any further medical issues that have not been mentioned above (i.e. Hypoglycemia, diabetes and obesity):

81. Do you feel your current medical follow-up addresses your health needs as an adult?

- ☐ Yes
- ☐ No
- ☐ Not sure

82. If you answered *No* or *Unsure*, list and describe what is not being addressed.

Thank you for your cooperation

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