Angelman Syndrome Development Project

Questionnaire Booklet

Angelman Syndrome Clinic
Dept of Developmental Assessment
St. George Hospital
Sydney, Australia

Postal Address:
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Letter to Parents/Caregiver

Dear Parents/Caregivers,

This questionnaire aims to obtain a better understanding of the behaviour and natural history of persons with Angelman Syndrome. It will add to the information collected by the Angelman Syndrome Clinic, St George Hospital.

This questionnaire is confidential. No information identifying a person will be released in any publication arising from this study.

Participation in the questionnaire is voluntary. You will receive the same level of services regardless of whether or not you participate.

Whilst the questionnaire takes about 30 minutes to complete, you may wish to complete it in stages.

Quite apart from the fact that this questionnaire may substantially help with the understanding of the development of your own child, it may also assist parents and carers with similar problems in the near future. The results of this survey will be published in the Angelman Syndrome Association's Newsletter.

We would like to thank the parents and the Angelman Syndrome Association for their input. We acknowledge the support of the St George Hospital for making its resources available.

Thank you in anticipation for completing this questionnaire. Should you have any questions, please contact:

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CONSENT FORM

As the person filling out the Questionnaire for the child/adult with Angelman syndrome, please complete the following:

Name	DOB

Address
Phone

Your relationship to child/adult with Angelman syndrome. Please ☑ as appropriate.

- ❑ Mother
- ❑ Father
- ❑ Guardian
- ❑ Other *(please specify)*

I have read and understand the Information Sheet and the Letter on the preceding page and am agreeable to participate in this questionnaire. I understand that participation is voluntary and I can withdraw from the study at any time.

Signed		Date / /
Name		

This questionnaire asks general and medical questions about the person with Angelman syndrome. Other parts of this questionnaire booklet are being developed.

The following questions apply both to a child with Angelman syndrome or an adult with Angelman syndrome. They could be living with a family or in the care of others. To keep it short, we have referred to the person with Angelman syndrome as "your child". But please understand that this also refers to adults with Angelman syndrome and to those not living with their family.

The questions require either a short written answer or ticking a box (ie ☑).

Please note that you may need to tick more than one box in some questions.

1. Name of child/adult with Angelman syndrome:

First Name	Family Name

2. Date of birth (and age) of child.

/ / day month year	years

3. Sex of child. Please ☑ the appropriate box.

☐ Male
☐ Female

4. Does the child live with you?

☐ Yes
☐ No. *Please specify*

Address

5. What are your main concerns about the child?

6. What things does your child do best? *Please specify***.**

7. What things does your child particularly enjoy? *Please specify.*

QUESTIONS 8 TO 14 ASK ABOUT SERVICES ASSISTING WITH YOUR CHILD'S CARE

8. Which of the following medical practitioners have been involved in your child's care? Please ☑ as appropriate.

☐ **General Practitioner:** *If so, please specify.*

Name	Address

☐ **Paediatrician:** *If so, please specify.*

Name	Address

☐ **Neurologist**: *If so, please specify*.

Name	Address

☐ **Geneticist**: *If so, please specify*.

Name	Address

☐ **Other**: *If so, please specify*.

Name	Address

9. Does your child receive any of the following?

☐ Physiotherapy
☐ Occupational therapy
☐ Speech therapy
☐ Behaviour therapy

10. Which of the following educational/training facilities does your child attend?

☐ Early intervention services
☐ Special needs pre-school
☐ Special class
☐ Special school
☐ Adult day program
☐ Other. *Please specify.*

☐ None of the above

11. Which of the following support services do you receive at home?

☐ Caseworker (eg social worker, community nurse)
☐ Home care (ie help with domestic duties)
☐ Home nursing care
☐ Home respite care (ie paid carer visiting your home)
☐ Other. *Please specify.*

☐ None of the above

12. Does he/she receive respite care away from home?

☐ No
☐ Yes. *If so, please specify the usual frequency during the last year.*
  ☐ less than 1 to 2 days/month
  ☐ 1 to 2 days/month
  ☐ greater than 1 to 2 days/month

13. Does he/she live away from his family on a permanent basis?

☐ No
☐ Yes. *Please specify*
  ☐ Group home
  ☐ Institution (eg hospital)
  ☐ Other. *Please specify.*

14. Does your child receive any of the following?

☐ Child Disability Allowance
☐ Invalid Pension
☐ Other benefit/allowance. *Please specify.*

☐ None of the above

QUESTIONS 15 TO 18 ASK ABOUT THE FAMILY BACKGROUND

15. Mother's

Name

Date of Birth | Place of Birth

Ethnic Background | Occupation

Has the mother had any miscarriages?

☐ No

☐ Yes. If so, please indicate the number of miscarriages and stage in pregnancy.

Has the mother had any stillbirths?

☐ No

☐ Yes. If so, please indicate the number of stillbirths.

Is there anyone with intellectual or neurological problems on the mother's side of the family? *Please specify.*

16. Father's

Name

Date of Birth | Place of Birth

Ethnic Background | Occupation

Is there anyone with intellectual or neurological problems on the father's side of the family? *Please specify.*

17. In some cultures, it is customary for relatives to marry (especially cousins). Are the mother and father related in any way?

☐ No

☐ Yes. *Please specify.*

18. Does your child with Angelman syndrome have any brothers or sisters? *Please specify.*

Name	Sex	DOB	Birth Weight

Have any of the brothers or sisters had problems with development (eg slow with walking, talking or learning)? *Please specify.*

Name	Problems

QUESTIONS 19 TO 33 ASK ABOUT THE PREGNANCY, LABOUR, BIRTH AND NEWBORN PERIOD OF YOUR CHILD WITH ANGELMAN SYNDROME

19. Please indicate if any of these problems occurred during the pregnancy.

- ☐ Threatened miscarriage
- ☐ Sugar diabetes
- ☐ High blood pressure
- ☐ Any infection
- ☐ Any other illness

If you ticked any of the above, please give details of when during the pregnancy and treatment required, if any.

20. Did the mother take any drugs/medications (either prescribed or not prescribed) during the pregnancy?

- ☐ No
- ☐ Yes. *Please specify.*

21. Did the mother drink any alcohol during the pregnancy?

- ☐ No
- ☐ Yes. *Please specify.*

22. Did the mother smoke during the pregnancy?

- ☐ No
- ☐ Yes. *Please specify.*

23. Did the mother have an ultrasound during the pregnancy?

- ☐ No
- ☐ Not sure
- ☐ Yes. As far as you know, the test was
  - ☐ Normal
  - ☐ Abnormal. *Please specify.*
  - ☐ Not sure

24. Did the mother have an amniocentesis during the pregnancy?

- ☐ No
- ☐ Not sure
- ☐ Yes. As far as you know the test was
  - ☐ Normal
  - ☐ Abnormal. *Please specify.*
  - ☐ Not sure

25. Did the mother have a chorionic villus biopsy during the pregnancy?

- ☐ No
- ☐ Not sure
- ☐ Yes. As far as you know the test was
  - ☐ Normal
  - ☐ Abnormal. *Please specify.*
  - ☐ Not sure

26. Please indicate how labour began. If the baby was born by planned Caesarean section, please go to question 28.

- ☐ Spontaneous onset of labour
- ☐ Induction of labour (eg labour brought on by drip or breaking the waters)

27. How long was the labour?

- ☐ Under 12 hours
- ☐ 12 to 24 hours
- ☐ Over 24 hours

28. At what stage in the pregnancy was the baby born?

- ☐ Around the due date (full-term)
- ☐ Late (after 42 weeks)
- ☐ Early (before 37 weeks). *Please specify how many weeks early.*

	weeks

29. The baby was born by:

- ☐ Normal delivery
- ☐ Breech delivery
- ☐ Forceps delivery
- ☐ Caesarean section
- ☐ Other. *Please specify.*

30. Please specify the following (if you can recall).

Birth Weight	
Birth Length	
Head circumference	

31. In the newborn period, did your baby require any of the following?

- ☐ Oxygen
- ☐ Assistance with breathing (ventilation)
- ☐ A drip (intravenous fluids)
- ☐ Tube-feeding (naso-gastric tube)
- ☐ Light therapy (phototherapy) for yellow skin (jaundice)
- ☐ Other. *Please specify.*

32. Was your baby born in hospital?

- ☐ No
- ☐ Yes. *If so, please specify hospital.*

33. How long did your baby remain in hospital (if born in hospital)?

Weeks	Days

QUESTIONS 34 TO 38 ASK ABOUT YOUR CHILD AS AN INFANT (ie first year of life)

34. As an infant (ie first year of life) did your child have any of the following feeding problems?

- ☐ Difficulty sucking
- ☐ Difficulty swallowing
- ☐ Strong forward movement of tongue (tongue-thrusting)
- ☐ Persistent vomiting (reflux)
- ☐ Failure to thrive (consistently underweight)

35. Was your baby breast fed?

- ☐ No
- ☐ Yes. *Please specify for how long*

Months

36. As an infant (ie the first year of life) did your child have any of the following problems?

- ☐ Turn in the eye (squint)
- ☐ Fits (convulsions)
- ☐ Jittery movements
- ☐ Other medical problems. *Please specify.*

37. How would you describe your infant (first year) when being held?

- ☐ Floppy
- ☐ Stiff
- ☐ Normal
- ☐ Combination of above. *Please specify.*
- ☐ Not sure

38. Which, if any, of the following would apply to your infant (first year)?

- ☐ Early or persistent smiling
- ☐ Frequent or persistent crying
- ☐ Happy
- ☐ Irritable
- ☐ Quiet
- ☐ Sleeping problems. *Please specify.*
- ☐ None of the above

QUESTIONS 39 TO 51 ASK ABOUT YOUR CHILD'S DEVELOPMENT

39. When did you first become concerned about your child's development?

Years	Months

40. What were you concerned about?

41. Which of the following developmental milestones has your child reached? As far as you can recall, when did he/she reach the milestone?

☐ Sitting unsupported	years	months
☐ Crawling	years	months
☐ Bottom shuffling	years	months
☐ Standing alone	years	months
☐ Walking alone	years	months

42. Which of the following applies to your child's speech?

- ☐ Babbles
- ☐ Less than 3 single (clear) words
- ☐ 3 to 6 single (clear) words
- ☐ More than 6 single (clear) words
- ☐ Puts 2 (clear) words together
- ☐ Puts 3 or more (clear) words together

43. Has your child had an intelligence (IQ) test?

- ☐ No
- ☐ Not sure
- ☐ Yes. *If so, please specify.*

Name of person / agency who performed test	Age of child at assessment

44. As far as you are aware, what is his/her level of intellectual handicap:

- ☐ Severe
- ☐ Moderate
- ☐ Mild
- ☐ Other. *Please specify.*

45. Which of the following applies to your child's mobility

- ☐ Unable to walk; uses stroller or wheelchair
- ☐ Walks with assistance
- ☐ Walks alone but unsteadily
- ☐ Walks well

46. Which of the following applies to your child's feeding?

- ☐ Needs to be fed
- ☐ Finger feeds
- ☐ Feeds self with spoon
- ☐ Uses fork to stab food
- ☐ Uses knife for spreading
- ☐ Eats with knife and fork

47. Which of the following applies to your child?

- ☐ Right handed
- ☐ Left handed
- ☐ Ambidextrous (use both hands equally well)
- ☐ Not sure

48. Which of the following applies to your child's dressing?

- ☐ Needs to be undressed and dressed
- ☐ Can take some clothes off by self
- ☐ Can undress self completely
- ☐ Can put some clothes on by self
- ☐ Can dress self completely

49. Which of the following applies to your child's bathing?

- ☐ Needs to be bathed
- ☐ Needs assistance with bathing
- ☐ Can bathe independently

50. Which of the following (if any) applies to your child's toileting?

- ☐ Incontinent of urine ('wee') by day
- ☐ Incontinent of urine ('wee') by night
- ☐ Incontinent of faeces ('poos') by day
- ☐ Incontinent of faeces ('poos') by night
- ☐ Wears nappies/incontinence aids during the day
- ☐ Wears nappies/incontinence aids during the night

51. Which of the following applies to your child's independence when toileting?

- ☐ Fully dependent on others
- ☐ Toilet-timed for urine ('wee')
- ☐ Toilet-timed for faeces ('poos')
- ☐ Goes by self to toilet to pass urine ('wees'), but needs supervision
- ☐ Goes by self to toilet to pass stools ('poos'), but needs supervision
- ☐ Fully independent

QUESTIONS 52 TO 54 ASK ABOUT YOUR CHILD'S BEHAVIOUR

52. Does your child have any of the following behaviours?

- ☐ Overactive, restless
- ☐ Poor attention span, unable to attend to one activity for any length of time
- ☐ Easily distracted from tasks
- ☐ Impulsive, acts before thinking

53. Does your child have any of the following behaviours?

- ☐ Chews or mouths objects
- ☐ Eats non-food items
- ☐ Fussy eater or has food fads
- ☐ Gorges food

54. Does your child have any of the following behaviours?

- ☐ Bursts of laughter
- ☐ Hand flapping
- ☐ Fascination for water
- ☐ Sleeps too little; disturbed sleep

PLEASE COMPLETE QUESTIONS 55 TO 65 IF YOUR CHILD HAS HAD FITS (EPILEPTIC SEIZURES). IF YOUR CHILD HAS NOT HAD FITS, PLEASE GO TO QUESTION 66.

55. What was your child's age when he/she first had a fit?

	Years

56. During the last year, has your child had a fit?

- ☐ Yes
- ☐ No. If so, at what age was the last fit?

	Years

Please go to Question 61 if you answered 'No'

57. If your child had fits during the last year, how often did they usually occur?

- ☐ Once per day or more frequently
- ☐ Once per week
- ☐ Once per month
- ☐ Once every few months
- ☐ Once per year

58. During the last year, how frequently have your child's fits been occurring?

- ☐ More often
- ☐ Less often
- ☐ About the same

59. There are different types of fits, such as generalised tonic-clonic (grand mal) and absences (petit mal). While some children have only one type of fit, others have more than one type. With this in mind, how many types of fits have your child had during the last year?

- ☐ Once type of fit
- ☐ Two types of fits
- ☐ Three or more types of fits
- ☐ Not sure

60. With regards to the previous question, please briefly describe the type of fit(s).

Main Type	
Second Type	
Other(s)	

61. Have doctors given a name to the type of fit(s) described above? *If so, please specify.*

Main type	
Second type	
Other(s)	

62. Have any anti-convulsant medications ever been tried to prevent fits occurring?

- ☐ No
- ☐ Yes. If so, which of the following have been tried?
  - ☐ Tegretol (carbamazepine)
  - ☐ Frisium (clobazam)
  - ☐ Rivotril (clonazepam)
  - ☐ Mogadon (nitrazepam)
  - ☐ Valium (diazepam)
  - ☐ Phenobarbitone (phenobarb)
  - ☐ Dilantin (phenytoin)
  - ☐ Epilim (valproate)
  - ☐ Sabril (vigabatrin)
  - ☐ Lamictal (lamotrigine)
  - ☐ Other *(please specify)*

63. Has your child ever been given rectal Valium (diazepam)?

- ☐ No
- ☐ Yes
- ☐ Unsure

64. Which of the above medication (if any) is your child taking at present. *Please specify.*

65. In your opinion, what anticonvulsant medication (if any) worked best?

QUESTIONS 66 TO 77 ASK ABOUT YOUR CHILD'S GENERAL HEALTH

66. Does your child have any visual problems?

- ☐ No
- ☐ Yes. If so, please tick as appropriate:
  - ☐ Cross-eyed (turn or squint)
  - ☐ Jerky eye movements (nystagmus)
  - ☐ Short sighted (myopia)
  - ☐ Far sighted (hypermetropia)
  - ☐ Other. *Please specify.*

67. Does your child have any hearing problems?

- ☐ No
- ☐ Yes. If so, please tick as appropriate
  - ☐ Glue ears (conductive deafness)
  - ☐ Nerve deafness (sensorineural deafness)
  - ☐ Hyperacute (very sensitive) hearing
  - ☐ Other. *Please specify.*

68. Does your child have any dental problems?

- ☐ No
- ☐ Yes. *Please specify*.

69. Does your child tend to dribble saliva from his/her mouth?

- ☐ No
- ☐ Yes. If so, please specify whether this is:
  - ☐ Most of the time
  - ☐ Sometimes
  - ☐ Rarely

70. Please indicate if your child has:

- ☐ Blond hair
- ☐ Blue eyes
- ☐ Lighter skin colour than parents
- ☐ Lighter skin colour than brothers/sisters
- ☐ Facial features different to other family members. *Please specify.*

71. Please indicate if your child has:

- ☐ Jerky movements
- ☐ Flat feet
- ☐ Sway back (lumbar lordosis)
- ☐ Curved back (scoliosis)

72. If your child is MALE and has reached puberty (development of pubic hair), *please specify* age of onset.

Years

73. If your child is FEMALE and has commenced her periods, *please specify* age of onset.

Years

74. Please indicate if your child has had any operations.

- ☐ No
- ☐ Yes. *Please specify.*

75. Does your child have any other health problems, not already indicated in this questionnaire.

- ☐ No
- ☐ Yes. *Please specify.*

76. Please specify your child's current regular medications.

77. Please specify if your child uses any of the following equipment aids.

- ☐ Foot orthotic (splint)
- ☐ Ankle foot orthotic (splint)
- ☐ Walking frame
- ☐ Wheelchair
- ☐ Other. *Please specify.*

QUESTIONS 78 TO 82 ASKS ABOUT TESTS THAT YOUR CHILD MAY HAVE HAD

78. Has your child had an EEG (brain wave test)?

- ☐ No
- ☐ Not sure
- ☐ Yes. If so, as far as you are aware, what was the result?
  - ☐ Normal
  - ☐ Abnormal. *Please specify.*
  - ☐ Not sure

As far as you can recall, when and where was the test done?

79. Has your child had a brain (cerebral) CT scan?

- ☐ No
- ☐ Not sure
- ☐ Yes. If so, as far as you are aware, what was the result?
  - ☐ Normal
  - ☐ Abnormal. *Please specify.*
  - ☐ Not sure

As far as you can recall, when and where was the test done?

80. Has your child had a brain (cerebral) MRI scan?

- ☐ No
- ☐ Not sure
- ☐ Yes. If so, as far as you are aware, what was the result?
  - ☐ Normal
  - ☐ Abnormal. *Please specify.*
  - ☐ Not sure

As far as you can recall, when and where was the test done?

81. Has your child had a chromosome test?

- ☐ No
- ☐ Not sure
- ☐ Yes. If so, as far as you are aware, what was the result?
  - ☐ Normal
  - ☐ Abnormal. *Please specify.*
  - ☐ Not sure

As far as you can recall, when and where was the test done?

82. Has your child had a FISH or DNA test?

- ☐ No
- ☐ Not sure
- ☐ Yes. If so, as far as you are aware, what was the result?
  - ☐ Normal
  - ☐ Abnormal. *Please specify*
  - ☐ Not sure

As far as you can recall, when and where was this test done?

THE FOLLOWING QUESTIONS ASK FOR YOUR COMMENTS ON THE QUESTIONNAIRE

83. Did you have any difficulties in filling out this questionnaire? If so, please comment.

84. Are there any issues concerning your child which have not been adequately covered in the questionnaire? If so, please comment.

85. Are there any other comments you would like to make?

Thank you for completing this questionnaire.

Please return the completed questionnaire to:

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