THE AUSTRALIAN RETT SYNDROME STUDY

funded by
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Reporting Clinician | *APSU Dr. Code* | *Date of report* / /

PATIENT : DEMOGRAPHIC AND FAMILY INFORMATION

1. *Child's first name* (First 2 letters) 2. *Child's surname* (First 2 letters) 3. *Date of Birth* / /

4. *Sex* **M** **F** 5. *Post Code of Mother*

6. *Usual place of residence of child* Parental Home Group Home Hostel Hospital Other

7. *Are you aware of any other specialists being involved in the care of this child?* Yes No Don't know

Please specify

If this patient is primarily cared for by another physician who you believe will report the case then there is no need at this stage to complete the remainder of this questionnaire. Please keep the patient's name and other details in your records. If no other report is received for this child we will contact you for further information.

8. *Child's country of birth*

9. *Mother's country of birth* Australia Other *if other, please specify*

10. *If born in Australia is she an Aboriginal or Torres Strait Islander?* Yes No

11. *Father's country of birth* Australia Other *if other, please specify*

12. *If born in Australia is he an Aboriginal or Torres Strait Islander?* Yes No

ANTENATAL AND PERINATAL HISTORY

13. *Were there any problems during the pregnancy*? Yes No Don't know

14. *If yes, please specify*

15. *Were there any abnormalities during the perinatal period ?* Yes No Don't know

16. *If yes, please specify*

17. *Mode of delivery* Normal Vacuum Forceps Breech Elective caesarean Emergency caesarean

18. *Presentation* Vertex Breech Other Don't know

19. *Please give Apgar score at*: 1 minute 5 minutes

20. *Please indicate the degree of resuscitation required at the birth by recording the appropriate number in the box.*

1=none, 2=suction only, 3=oxygen therapy, 4=bag & mask, 5=endotracheal intubation, 6=ext. cardiac massage & ventilation, 8=other

21. *Please indicate (by a tick) any complications of labour and delivery (you may provide more than one option)*

1=precipitate delivery
2=fetal distress
3=prolapsed cord
4=cord tight around neck
5=cephalopelvic disproportion
6=PPH (=>500ml)
7=other

22. *What was the birth weight?* ☐☐☐☐ g Don't know ☐

23. *Please give gestational age if available* ☐☐ weeks Don't know ☐

24. *Please provide head circumference (HC) at birth if available* ☐☐ cm

25. *If HC at birth not known was head circumference considered to be in normal range for first month of life?* Yes ☐ No ☐ Don't know ☐

26. *Was there later slowing of head growth?* Yes ☐ No ☐ Don't know ☐

27. *If yes, at what age?* ☐☐ months. *If available please provide a copy of the head circumference growth chart*

DEVELOPMENTAL HISTORY

28. *Was development initially within normal limits?* Yes ☐ No ☐ Don't know ☐

29. *Is there now evidence of an apparently severe or profound intellectual handicap?* Yes ☐ No ☐ Don't know ☐

30. *At what age did normal development deteriorate?* ☐☐ months

31. *Was there any significant illness or injury prior to the time of developmental deterioration?* Yes ☐ No ☐ Don't know ☐

32. *Was there loss of previously acquired hand skills?* Yes ☐ No ☐ Don't know ☐

33. *If yes, at what age?* ☐☐ months

34. *Has a fixed pattern of hand movements developed?* (see APSU protocol) Yes ☐ No ☐ Don't know ☐

35. *Has there been a loss of communication skills?* Yes ☐ No ☐ Don't know ☐

36. *If yes, at what age?* ☐☐ months

37. *Is expressive language now impaired?* Yes, severe ☐ Yes, moderate to mild ☐ No ☐ Don't know ☐

38. *Is receptive language now impaired?* Yes, severe ☐ Yes, moderate to mild ☐ No ☐ Don't know ☐

39. *Was there a period of social withdrawal?* Yes ☐ No ☐ Don't know ☐

40. *If yes, at what age?* ☐☐ months

41. *Has this withdrawal persisted?* Yes ☐ No ☐ Don't know ☐

42. *Has the child ever walked?* Yes ☐ No ☐ Don't know ☐

43. *Is the child currently able to walk?* Yes ☐ No ☐ Don't know ☐

44. *If yes, is the gait stiff of clumsy?* Yes ☐ No ☐ Don't know ☐

45. *Is the posture stiff or clumsy?* Yes ☐ No ☐ Don't know ☐

INVESTIGATIONS, FURTHER CLINICAL HISTORY AND PRESENT CONDITION

46. *Has the child ever experienced: (please tick the appropriate boxes)*

Significant constipation ☐ Scoliosis ☐ Breathing abnormality ☐ Seizures ☐ Significant sleep disturbances ☐

47. *Is the child currently on or ever been on anticonvulsant medication?* Yes ☐ No ☐ Don't know ☐

Can you provide information on:

48. *Karyotype*	Normal ☐	Abnormal ☐	Not done ☐	Don't know ☐
49. *Urine metabolic screen*	Normal ☐	Abnormal ☐	Not done ☐	Don't know ☐
50. *Urine mucopolysaccharides*	Normal ☐	Abnormal ☐	Not done ☐	Don't know ☐
51. *WBC Lysosomal enzymes*	Normal ☐	Abnormal ☐	Not done ☐	Don't know ☐
52. *EEG*	Normal ☐	Abnormal ☐	Not done ☐	Don't know ☐
53. *Sleep study*	Normal ☐	Abnormal ☐	Not done ☐	Don't know ☐
54. *CT Scan*	Normal ☐	Abnormal ☐	Not done ☐	Don't know ☐
55. *MRI*	Normal ☐	Abnormal ☐	Not done ☐	Don't know ☐
56. *Ophthalmological Exam*	Normal ☐	Abnormal ☐	Not done ☐	Don't know ☐
57. *X-rays of Hands and Feet*	Normal ☐	Abnormal ☐	Not done ☐	Don't know ☐

58. *Please provide details of the results of any x-rays carried out.*

59. *Has DNA been collected on this child?* Yes ☐ No ☐ Don't know ☐

60. *Please provide details of any molecular analysis carried out.*

61. *Please state the initial diagnosis given if known.*

62. *Is the diagnosis now considered to be*: **Definite** Rett Syndrome ☐ **Possible** Rett Syndrome ☐

63. *Who made the diagnosis?* *and when?*

64. *Has the diagnosis been communicated to the parents?* Yes ☐ No ☐

65. *Have the parents been provided with information about the Rett Syndrome Association of Australia?* Yes ☐ No ☐

CLINICAL EXAMINATION AND MEASUREMENT

66. *Date of examination:* / /

67. *Weight (kg)* 68. *Height (cm)* 69. *Head circ. (cm)*

70. *Are there any dysmorphic features?* Yes ☐ No ☐ Don't know ☐

71. *If yes, please specify*

72. *Is there any evidence of spasticity?* Yes ☐ No ☐ Don't know ☐

73. *We welcome any additional comments regarding this family or Rett syndrome in general.*

THANK YOU VERY MUCH FOR YOUR TIME