

22q11.2 Deletion Syndrome

APSU Office Use Only

Australian Paediatric Surveillance Unit

Study ID #:

Please contact the APSU (02) 9845 3005; apsu@chw.edu.au If you have any questions about this form

Month/Year Report:

Instructions: Please answer each question by ticking the appropriate box or writing your response in the space provided. DK=Don't Know; NA = Not Applicable

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REPORTING CLINICIANS DETAILS1. APSU Dr Code/Name: / _____ 2. Date questionnaire completed: / / **PATIENT DETAILS** 3. First 2 letters of first name: 4. First 2 letters of surname: 5. Date of Birth: / / 6. Sex: M F 7. Postcode of family: 8. Child's ethnicity: Caucasian Asian African
 Middle Eastern Aboriginal or Torres Strait Islander Other (please specify) _____ Don't know9. Child's country of Birth Australia Other (please specify) _____**If this patient is primarily cared for by another physician who you believe will report the case, please complete the questionnaire details above this line and return to the APSU. 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 information requested in the remainder of the questionnaire.***The primary clinician caring for this child / young person is: Name:**Hospital:***MEDICAL HISTORY AND PREGNANCY HISTORY**

10. How old was the child when you first saw him/her in your practice? _____ years _____ months

11. How old was the child when concerns about their development were first raised? _____ years _____ months

11a. Were these concerns first raised by Parent/Caregiver Health professional Other (please specify) _____12. Did the patient originally receive an alternative diagnosis? Yes No (please specify) _____13. Who first suspected the 22q11.2 deletion syndrome? General Practitioner Paediatrician Physiotherapist
 Cardiologist Other (please specify): _____

14. Which features alerted you to suspect a diagnosis of 22q11.2 deletion syndrome? Please tick all items that apply.

- Developmental delay Learning Difficulties Behavioural problems Psychiatric issues
 Cardiac anomalies Palatal defects Feeding problems Hypernasal speech
 Immunodeficiency Hypocalcaemia Ear infections Seizures
 Thyroid abnormalities Dysmorphic features Other health problems (please specify): _____
 Other problems (please specify): _____

15. Were there any complications during the pregnancy? Yes No DK

15a. If yes, please specify complications: _____

16. Were there any complications during birth? Yes No DK

16a. If yes, please specify complications: _____

17. Was the child premature? Yes No DK

17a. If yes, what was the gestational age? _____ weeks

17b. If yes, what was the child's birth weight? _____ grams

18. Has the child ever been hospitalised? Yes No DK

18a. Please estimate the number of admissions you believe were related to 22q11.2 deletion syndrome. _____

DIAGNOSIS

19. At what age was the diagnosis made? _____ (months) _____ (years)

20. By what method was the diagnosis confirmed? Fish MLPA micro array other (please specify) _____21. Is the deletion de novo? Yes No DK22. If known, please report the size of the deletion and the location of the deletion (specify): _____ DK23. Were any other genetic abnormalities identified? (specify) _____ DK24. If familial, is the deletion maternal or paternal? Maternal Paternal DK25. Do any siblings have the syndrome? Yes No DK No siblings26. Do any siblings have developmental disabilities? Yes No DK No siblings

26a. If yes, please specify sibling disabilities: _____

27. Is there a family history of mental health problems? Yes No DK If yes, specify _____

28. Is there a family history of intellectual disability, learning problems? Yes No DK If yes, specify _____

DEVELOPMENTAL AND BEHAVIOURAL FEATURES

29. By what age were developmental milestones achieved? a.) Sitting _____ (mths) not yet achieved DK

b.) Walking _____ (mths) not yet achieved DK

c.) Talking (simple words i.e. "mama" or "dada") _____ (mths) not yet achieved DK

30. If your patient is **younger than 5 years**, is there a global developmental delay? Yes No DK

31. If your patient is **5 years or older**, please indicate level of intellectual disability (based on WISC-IV assessment, if known).

None (>85) Borderline (68-84) Mild (52-67) Moderate (36-51) Severe (20-35) Profound (<20) DK

32. What type of special educational needs does the patient have (specify): _____

33. Does the patient have any of the following (please tick all that apply):

Social Problems Repetitive behaviours Anxiety Compulsive behaviours Hallucinations

Severe temper tantrums Oppositional behaviours Aggression Attention problems Delusions

Withdrawn behaviours Other behavioural features (specify): _____

34. If appropriate, please provide additional information: _____

35. Has the child been diagnosed with any of the following in conjunction with the 22q11.2 deletion? (please tick all that apply)

Autism Spectrum Disorder Attention Deficit/Hyperactivity disorder Oppositional Defiant Disorder Anxiety

Depressive Disorder Sleep Disorder Learning Disability Schizophrenia or other psychotic disorder

Other psychiatric diagnoses (specify): _____

35a. If appropriate, please provide additional information: _____

36. Is the child currently accessing disability services? Yes No DK

MEDICAL HISTORY (please refer to table of clinical guidelines attached)

37. Has the patient suffered from any of the below conditions (please tick all that apply)?

Condition	Yes/No	If yes, please specify	Was the child referred (Y/N)	If yes, to what type of health professional (e.g., cardiologist, speech pathologist, etc)
Dysmorphic features (including face, hands, genitals)				
Cardiovascular (conotruncal/others)				
Palatal and related problems (e.g. hypernasal speech/VPI)				
Immune related (e.g. recurrent infections)				
Endocrine (e.g., hypocalcaemia)				
Gastroenterological (e.g. dysphagia, constipation)				
Genitourinary (e.g. urinary tract anomaly)				
Ophthalmological (e.g. strabismus)				
Skeletal (e.g. scoliosis)				
Haematology/oncology (e.g. thrombocytopenia)				
Neurological (e.g. epilepsy)				
Growth problems (e.g., failure to thrive)				
Dental (e.g. enamel hypoplasia)				
Other (please specify)				

Thank you very much for participating in this important study.

If you have time, we would very much appreciate it if you would complete these additional questions regarding your general experience of patients with 22q11.2 deletion syndrome.

YOUR GENERAL EXPERIENCE OF PATIENTS WITH 22q11.2 DELETION SYNDROME

1. How many patients with 22q11.2 deletion syndrome have you ever provided care for?
 None Less than 5 patients 5 to 10 patients More than 10 patients
2. How would you describe your understanding of 22q11.2 deletion syndrome?
 Poor Average Good Excellent
3. Do you see yourself as the primary healthcare provider for the above patient? Yes No
- 4a. If yes, do you think you should be the primary healthcare provider? Yes No
- 4b. If no, who do you think should be the primary care provider? *(specify)* _____
5. Have you experienced any barriers in providing care for children with 22q11.2 deletion syndrome? *(specify)* _____
6. What services do you think that your patients with 22q11.2 deletion syndrome will need? *(specify)* _____

7. Do you believe that these services are easily accessible to patients? Yes No DK Comment:*(specify)* _____

8. How useful did you find the clinical guidelines table?
 Not at all useful A bit useful Useful Very useful Extremely useful
9. Would you like additional information about 22q11.2 deletion syndrome? Yes No
- 9a. If yes, please supply a postal address: _____
10. Have you directed your patient and their family to the VCFS & 22q11 support group (www.vcfsfa.org.au)? Yes No
11. What is your medical specialty?
 Clinical Geneticist General Paediatrician Developmental Paediatrician Other *(specify)* _____

We are currently undertaking another research study of health care needs of families affected by 22q11.2 deletion syndrome. If you would like more information about this study OR if you would more information about published medical guidelines on the care of children with 22q11.2 deletion syndrome OR if you would like information about parent support opportunities for families. Please, provide a contact email address and we will get back to you shortly.

Full Name: _____

Email address: _____

Thank you for your help with this research project. Please return this questionnaire to the APSU in the reply-paid envelope or fax to 02 9845 3082 even if you don't complete all items. Australian Paediatric Surveillance Unit, Kid's Research Institute, Locked Bag 4001, Westmead NSW 2145.
The APSU is affiliated with the Royal Australasian College of Physicians (Paediatrics and Child Health Division) and Sydney Medical School, The University of Sydney. The APSU is funded by the Australian Government Department of Health and Ageing. This study has been approved by a Human Research Ethics Committee properly constituted under NHMRC guidelines.