22q11.2 Deletion Syndrome Australian Paediatric Surveillance Unit	*APSU Office Use Only*	
	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	*Version 1_13.04.2015*	

REPORTING CLINCIANS DETAILS

1. 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 know

9. 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 ☐ DK

22. If known, please report the size of the deletion and the location of the deletion *(specify)*:_____ ☐ DK

23. Were any other genetic abnormalities identified? *(specify)*_____ ☐ DK

24. If familial, is the deletion maternal or paternal? ☐ Maternal ☐ Paternal ☐ DK

25. Do any siblings have the syndrome? ☐ Yes ☐ No ☐ DK ☐ No siblings

26. 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 "dadda") _____ (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.