

**Australian Paediatric Surveillance Unit  
STUDY PROTOCOL****The Australian 22q11.2 Deletion Syndrome Study****BACKGROUND**

22q11.2 deletion syndrome (22q11DS) is the most common microdeletion disorder with an estimated prevalence of ~1 in 4000 live births<sup>1-3</sup>. An identified deletion at 22q11.2 has been identified in the majority of individuals diagnosed with DiGeorge syndrome, velocardiofacial syndrome and conotruncal anomaly face syndrome, as well as in some people with autosomal dominant OpitzG/BBB syndrome and Cayler cardiofacial syndrome<sup>4</sup>. The syndrome has also been known as Sphrintzen syndrome and CATCH-22. The 22q11.2 deletion syndrome is a multi-system syndrome associated with congenital heart defects, palatal anomalies, immune deficiencies and characteristic facial features. The syndrome is arguably one of the most common causes of developmental delay and congenital cardiac abnormalities after Down syndrome<sup>5</sup>. People with 22q11DS often have intellectual disabilities in the mild range and there is a high prevalence of psychiatric disorders such as ADHD, autism-like symptoms and, in adulthood, psychosis.

However, the syndrome is clinically under-recognised and it has been suggested that the prevalence may be significantly higher than what the current literature proposes<sup>6</sup>. The diagnosis of 22q11.2 deletion syndrome is frequently delayed and sometimes a diagnosis is not achieved until adolescence or adulthood. This may be due to lack of familiarity with the syndrome from clinicians and further complicated by the large variability in phenotypic expression even within families, as explored by, for example, the Australian geneticist Anthony Lipson in the 1980's. In some people, the symptoms are very mild and they may not be diagnosed with the 22q11.2 deletion until they experience severe mental health problems later in life or they have a child who is diagnosed with the syndrome. However, it is unknown how many children with 22q11.2 deletion syndrome there are in Australia and, further, it is not clear what the health care needs are for this group of children. Delays in diagnosis due to a lack of familiarity can cause not only medical complications but also has an impact on the psychological wellbeing both of the child and family.

This study is the first in Australia to investigate the prevalence and incidence of 22q11.2 deletion syndrome and the associated health care needs of children with the syndrome. The current study will also establish a cohort of diagnosed cases and follow them over time to provide data on health care needs for people with 22q11DS.

***Clinicians notifying cases will also be invited to participate in a survey at the end of the case report form to identify their understanding of the syndrome and the perceived health care needs of children with the syndrome. This survey is optional.***

**STUDY OBJECTIVES**

1. To increase clinicians' awareness of 22q11.2 deletion syndrome by provision of information on clinical features and diagnostic criteria.
2. To estimate the birth prevalence of 22q11.2 deletion syndrome in Australian children seen by paediatricians.
3. To estimate the incidence (new cases identified) of children aged  $\leq 16$  years with 22q11.2 deletion syndrome in Australia over the study period.
4. To determine the age of diagnosis and the clinical features.
5. To determine the health services utilised by patients due to conditions associated with 22q11.2 deletion syndrome.

## CASE DEFINITION

Please report only **newly diagnosed** cases of children < 16 years of age with a genetically confirmed 22q11.2 deletion syndrome, whom you have seen within the last month and that you have not previously reported to the APSU.

## FOLLOW- UP NOTIFICATIONS

A questionnaire requesting further details will be forwarded to practitioners who report a case with 22q11.2 deletion syndrome. You can complete the questionnaire online or you can complete a paper copy. A copy of the questionnaire is enclosed for your information. Please, return the questionnaire even if you cannot fill out all the questions.

Clinicians will also be invited to contribute further to research on health care needs in this population. If you wish to learn more about this research, you will be asked to provide a valid email address and the researchers will provide further information. This is completely voluntary and you can choose if you would like to participate subsequent to perusing the information. A similar procedure has been used in other studies facilitated by APSU without problems.

Any doctor wanting information about the research should contact Dr Linda Campbell (see contact details below).

## INVESTIGATOR CONTACT DETAILS (\*Principal Investigator and contact person)

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