Foundation Australia
& New Zealand

22q11.2 Deletion Syndrome

- 22q11.2 Deletion Syndrome (22q Deletion) is a genetic disorder caused by a missing piece of the 22nd chromosome.
- 22q Deletion is said to occur in 1 in 2000 live births.
- 22q Deletion is the second most common cause of congenital heart defects and developmental delays.
- 22q Deletion is the most common genetic syndrome associated with cleft palates or palate abnormalities.
- 22q Deletion also causes other major medical problems such as immune, gastrointestinal, endocrine, skeletal, kidney, and ear, nose and throat issues.
- Most people with 22q Deletion will have some form of anxiety, stress or depression, however this can be managed.
- 22q Deletion can affect every system in the human body with nearly 200 mild to serious health and developmental issues.
- 22q Deletion is also known as **DiGeorge or VCFS**

22q.org.au

Upon initial diagnosis the standard assessments for all ages include but are not limited to:

- Cardiology - approx. 75% of patients with 22q11.2 DS will have a congenital heart defect
- Endocrinology
- Immunology - 22q Deletion can cause many other problems such as immunodeficiency leading to difficulty fighting infection and autoimmune disease
- Speech/Language/Developmental Assessments, including the palate - most children will have some form of speech impairment
- A Renal Ultrasound (to check the kidneys)
- X-rays of the neck (in children old enough to cooperate and where the bones are well ossified – so ~ 3 to 4 years of age)

Some individuals have many of the above issues and others have almost none. The condition is extremely variable.

For a full list of recommended assessments please refer to the **Practical Guidelines for Managing Patients with 22q11.2DS**. These guidelines have been produced by the International 22q Society of professionals and can be found on our website 22q.org.au