**ADULT 22q11DS GUIDELINES FOR HEALTH PROFESSIONALS**

Foreword:

When diagnosed in childhood, 22q11 is sometimes managed by a multidisciplinary approach. Once the transition into adult hood is made however, or if the patient is diagnosed in adulthood then these safety nets are not available to them and this is where GP and other health professional services become vital for their life long care to be the link to all areas of support that are needed to help them live a long and productive life.

To help assist your patient with their care plan we would be grateful if you could spare a few minutes to familiarise yourself with the care recommendations contained within this document which has been written by Dr Alex Habel, a leading expert on the condition and founder of the 22q11 clinic at Great Ormond Street.

by Mark Tripp, Trustee of Max Appeal

**Recommended medical assessments for adults with 22q11deletion syndrome (also known as di George Syndrome, Velocardiofacial Syndrome, Shprintzen Syndrome)**

It is the most common microdeletion syndrome in humans. Growing numbers of affected children surviving to adulthood is now the norm. Congenital heart disease, speech difficulties often linked to cleft palate or disordered palate function, reduced immunity, major psychological and behavioural difficulties, hypocalcaemia, presenting in childhood are commonly followed by treatable later-onset conditions. The concerns of them, their partners and family deserve careful assessment if they attend a doctor's surgery, social work department, or community based therapist. Most adults have borderline intelligence 70-84; 30% have mild intellectual disability (IQ 55-69). Particularly weak arithmetic ability renders many financially vulnerable. The above should be borne in mind when discussing and planning their management and appropriate referral to specialist colleagues.

**Regular (annual or 2 yearly) General Practitioner checks in Primary Care**

- Immune problems are relatively common: Full blood count for anaemia, platelet count and/or white blood cell count may be low due to frequently occurring immune disorders and warrant referral for further assessment.
- Thyroid function. A one in four chance of hypothyroidism or hyperthyroidism.
- Calcium blood level. A greater than 50% life time risk of hypocalcaemia due to hypoparathyroidism, particularly at times of stress e.g. pregnancy, surgery and adolescence. Endocrinologist referral for assessment and management advice.
- Renal function where ultrasound (must be done in ALL patients at least once) has shown single kidney or other renal abnormality.
- BMI check for overweight/obesity which may be more frequent and troublesome. Muscle weakness, reduced energy levels, mental health disorders and their treatment may contribute to overweight. Appropriate dietetic and activity advice and management should be offered.
- Existing disorders from childhood reviewed for symptoms. Ensure appropriate follow-up is in place for congenital cardiac disorders, palate function (which may deteriorate with age).


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- History and appropriate physical exam because of an increased tendency for:

  (i) Rheumatoid arthritis.
  (ii) Diabetes type 2.
  (iii) Vulnerable behaviours: on-line/social media contacts, introduced to substance abuse, gambling, sexual exploitation.
  (iv) Early onset Parkinsonism (under 50 years old).
  (v) Schizophrenia has a 25% incidence. Mental decline, dementia, seizures, and behaviour disorders especially panic attacks, depression, attention deficit. All much more frequent than in the general population.
  (vi) Dental hygiene: reduced immunity and enamel underdevelopment due to hypocalcaemia predispose to decay. Bacterial endocarditis risk for those with cardiac conditions. Dental supervision mandated.

**RED FLAG INDICATORS AND REFERRALS IN 22q11DS**

**Psychological. A complexity of symptoms and presentations for referral to Mental Health Services**

- Disordered thinking (Delusions, hallucinations, preoccupations, suspiciousness).
- Emotions disordered (increased anxiety, angry, sadness, inappropriate laughing, rapidly changing mood, hurt feelings).
- Behaviour (social withdrawal, impulsive, agitation, self injury, neglecting themselves).
- Accompanying Physical changes: more/less/changed sleep, energy levels, appetite; onset or worsening of tics, tremors, somatic complaints e.g. chest, GI symptoms

**STDs:** assess vulnerability including to internet and social media sites (to evaluate whether referral for appropriate safe sex counselling and/or possible social work involvement is indicated).

**Women with certain congenital cardiac lesions:** specific pregnancy and contraception education because pregnancies have increased risks of maternal and fetal/neonatal complications - see below.

**Pregnancy in mother with 22q11DS or an affected father:** Pregnancy complications from previous cardiac surgery, hypocalcaemia and premature delivery are more frequent. Refer immediately to antenatal diagnosis for a possibly affected fetus (50% chance of inheritance of 22q11DS). Include referral for genetic counselling at a level appropriate to the individual's understanding, to explore management options.

**Seizure**s of any type occur in 6%. Consider hypocalcaemia as well as epilepsy, brain malformations. Endocrine or Neurological referral, with urgency.

**Scoliosis** in teens can progress rapidly and more require surgery than in the general population. Adults with significant kyphosis need expert monitoring regularly for complications and progression.

**End note: Premature mortality in adults with 22q11DS, from a variety of causes, reminds clinicians to pay close attention to their medical and psychological health issues. EARLY REFERRAL TO AN APPROPRIATE CLINICAL SERVICE IS ADVISED TO AVOID UNNECCESSARY DELAY IN MANAGEMENT, A COMMONLY RECOGNISED PROBLEM IN THESE COMPLEX PATIENTS**

Authors: Drs Habel and Kumararatne on behalf of UK 22q11DS Guideline Group, 2016


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**Free downloadable documents:**
The National Consensus on the Diagnosis and Management of 22q11DS: https://www.maxappeal.org.uk/consensus-document

Practical guidelines for managing adults with 22q11.2 deletion syndrome: https://www.maxappeal.org.uk/adult-management-guidelines

**About Max Appeal:**
Max Appeal is the UKs charity supporting families affected by 22q11 syndromes.

Vision: Max Appeal's vision is of a Society where children and adults affected by 22q11.2 DS are valued and able to fulfil their potential.

Mission: To enable people with 22q deletion to lead an independent and economically prosperous life as possible with Max Appeal being the voice to achieve appropriate medical care, social support, employment and empowerment for individuals and their families.



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