

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 born 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

- A. 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.
- B. Thyroid function. A one in four chance of hypothyroidism or hyperthyroidism.
- C. Calcium blood level. A greater than 50% life time risk of hypocalcaemia due to hypoparathyroidism, particularly at times of stress eg pregnancy, surgery. Endocrinologist referral for assessment and management advice.
- D. Renal function where ultrasound (must be done in ALL patients at least once) has shown single kidney or other renal abnormality.
- E. 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.
- F. 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).
- G. 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, enamel underdeveloped due to hypocalcaemia predispose to decay. Bacterial endocarditis risk for those with cardiac conditions. Dental supervision mandated.

Turn OVER THE PAGE for RED FLAG INDICATORS AND REFERRALS IN 22Q11DS

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

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

STD's: 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.

Seizures 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 UNNECESSARY DELAY IN MANAGEMENT, A COMMONLY RECOGNISED PROBLEM IN THESE COMPLEX PATIENTS

Free downloadable article:

[Practical guidelines for managing adults with 22q11.2 deletion syndrome.](#)

Fung WL, et al. Genet Med. 2015 ;17:599-609.

(Drs Habel and Kumararatne on behalf of UK 22q11DS Guideline Group, 2016)