Recommendations for periodic assessments and management of adults with 22q11.2DS

Source: Table 1 of Updated clinical practice recommendations for managing adults with 22q11.2 deletion syndrome (2023)

The boxes indicate items recommended by 22q experts. Please put a **CHECK** ☑ in the box when completed.

Assessments and Management	At Diagnosis or Initial Assessment	At Follow-up (Every 1-2 years)
Genetic		
• Parental genetic testing (FISH, MLPA, or microarray)[a]	☐	
• Genetic counseling (including recurrence risk, update on natural history, management)	☐	☐
• Family planning, reproductive and prenatal counseling	☐	☐
• Additional genetic testing[b]	If applicable	
General		
• Consultation with clinician(s) experienced with 22q11.2DS[c]	☐	☐
• Comprehensive history-taking (including family history), systems review, and medication review	☐	☐
• Assessment of need for/coordination with specialist(s) providing care	☐	☐
• Nutritional assessment; diet and exercise counseling	☐	☐
• Sleep evaluation (consider polysomnography), sleep hygiene recommendations	☐	☐
• Vaccination counseling, other standard preventive health care measures	☐	☐
• Assessment of functioning (including hygiene), care/supports (family/community/government), safety issues (e.g. financial, internet)	☐	☐
Physical examination and additional diagnostic tests		
• BMI, resting heart rate, blood pressure	☐	☐
• 22q11.2DS-relevant laboratory tests[d]	☐	☐
• Echocardiogram	☐^	
• Abdominal ultrasound	☐^	
• Routine care/hearing, vision, dental assessment[e]	☐	☐
Targeted clinical assessments[f]		
• CNS—psychiatric, neurologic, neurocognitive assessments (including for anxiety, psychosis, seizures, movement disorders, formal testing of cognitive and adaptive functioning/ADL)	☐	☐
• Congenital cardiac (ACHD) and cardiovascular risk assessment	☐	☐
• Endocrinology	☐	☐
• Genitourinary, obstetrics/gynecology assessment (including contraception, pregnancy risks, and safe sex counseling)	☐	☐
• Hematology, gastroenterology, orthopedic/rheumatology, respirology, immunology, otolaryngology, ophthalmology, dermatology	☐	☐

a – Strategy depending on test availability.
b – When rare recessive condition associated with a gene in the 22q11.2 deletion region is suspected or atypical phenotypic features are observed.
c – Having seen several adult patients with 22q11.2DS both in consultation and follow-up (if possible).
d – CBC and differential, thyroid-stimulating hormone, (pH-corrected ionized) calcium, magnesium, creatinine, lipid profile, glucose, and HbA1c; other examples are parathyroid hormone, electrolytes, and liver function tests (especially alanine aminotransferase); checking CBC and calcium preoperatively and postoperatively, as well as regularly during pregnancy, also recommended.
e – Follow-up intervals may be longer.
f – Consideration of referral to and collaboration with (medical) specialists in individual cases; especially in cases with complex diagnosis and/or complex management, taking into account the variability in natural history between patients and increased risk of many health issues.

☐^ indicates if not previously performed as an adult or in recent years, and with a low threshold for late-onset manifestations of 22q11.2DS, including aortic root dilation, gallstones, fatty liver, and nephrocalcinosis.

Abbreviations:
22q11.2DS, 22q11.2 deletion syndrome; ACHD, adult congenital heart disease; ADL, activities of daily living; BMI, body mass index. CBC, complete blood count; CNS, central nervous system; FISH, fluorescence in situ hybridization; MLPA, multiplex-ligation dependent probe amplification.

Source:
Updated clinical practice recommendations for managing adults with 22q11.2 deletion syndrome. Boot E. et al. Genet Med. 25(3): 100344, 2023.