

# Recommendations for periodic assessments and management of **children and adolescents** with 22q11.2DS

Source: Table 1 of [Updated clinical practice recommendations for managing children 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	Annual/Biennial	0-1y	1-5y	6-12y	13-18y
<b>Genetic</b>						
• Genetic testing (proband: MLPA or microarray; FISH if only available method) (parents: MLPA or FISH) <sup>a</sup>	<input type="checkbox"/>					
• Genetic counseling (etiology, natural history, recurrence risk, prenatal/preconception screening/diagnostics)	<input type="checkbox"/>	<input type="checkbox"/>				<input type="checkbox"/>
• Remaining allele/exome sequencing (when appropriate) <sup>b</sup>	<input type="checkbox"/>					
<b>General</b>						
• Consultation with clinician(s) experienced with 22q11.2DS <sup>c</sup>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Comprehensive history-taking (including family history)	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Physical examination	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Nutritional assessment, feeding, swallowing, GERD, constipation, and growth	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Neurologic and developmental assessment (neurologic exam, milestones, sacral dimple, neuroimaging as needed)	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Assessment of history of infections, allergy, asthma, autoimmunity, and malignancy	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Assessment of access to specialized health care and community, developmental, and government resources	<input type="checkbox"/>		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<b>Other clinical assessments</b>						
• Cardiac evaluation (using echocardiogram and EKG; determine arch sidedness)	<input type="checkbox"/>					
• Long term follow-up for all with CHD; transition to GUCH if CHD		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Periodic screening for arrhythmias/EKG abnormalities and dilated aortic root <sup>d</sup>				<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Periodic EKG screening in at-risk patients (antiepileptic/neuropsychiatric treatment, hypocalcemia, thyroid disease)		<input type="checkbox"/>				
• Referral to cleft-palate team to assess for overt cleft, SMCP, and VPD (nasendoscopy/videofluoroscopy as needed) <sup>e</sup>	<input type="checkbox"/>		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Evaluation of speech and language by speech-language pathologist <sup>f</sup>	<input type="checkbox"/>		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Evaluation by otolaryngologist for recurrent otitis media and possible laryngo-tracheo-esophageal anomalies	<input type="checkbox"/>		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Evaluation of hearing using audiogram +/- tympanometry	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Ophthalmic evaluation/vision (refractive errors, strabismus, exotropia, sclerocornea, coloboma, ptosis)	<input type="checkbox"/>		<input type="checkbox"/>	<input type="checkbox"/>		
• Dental evaluation (measure saliva secretion rate from 6 y) <sup>g</sup>				<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Endocrinologic assessment (PTH, calcium, magnesium, creatinine, TSH, and free T4; GH studies as needed)	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Consider clinical (multidisciplinary) feeding and/or swallowing evaluation including assessment of airway <sup>h</sup>			<input type="checkbox"/>	<input type="checkbox"/>		
• Renal and bladder ultrasound	<input type="checkbox"/>					
• Immunologic assessment: T- and B cell phenotyping <sup>i</sup>	<input type="checkbox"/>		<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>
• Immunologic assessment: IgG, IgA, IgM, IgE levels (not before 6 months)			<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>
• Immunologic assessment: vaccine responses <sup>j</sup>			<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>
• Complete blood count and differential	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Routine scoliosis screening with scoliometer and with x-ray when clinically indicated					<input type="checkbox"/>	<input type="checkbox"/>
• Radiography of the cervical spine at age ~4 y to exclude instability <sup>k</sup>				<input type="checkbox"/>		
• Sleep evaluation (consider polysomnography pre and post VPD repair), sleep hygiene recommendations				<input type="checkbox"/>	<input type="checkbox"/>	
<b>Cognitive development, academic functioning, and child psychiatry</b>						
• Assessment of cognitive/learning capacities including language domains with standardized measures	<input type="checkbox"/>			<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Assessment of adaptive functioning (e.g. daily living skills)	<input type="checkbox"/>			<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
• Psychiatric assessment (ASD, ADHD/ADD, anxiety, and psychotic disorders)	<input type="checkbox"/>			<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Table 1 provides recommendations for periodic assessment and management of children and adolescents with 22q11.2 deletion syndrome at diagnosis, annually/biannually, and by age.

**Abbreviations:**

- ADD, attention deficit disorder
- ADHD, attention deficit hyperactivity disorder
- ASD, autism spectrum disorders
- CHD, congenital heart disease
- EKG, electrocardiogram
- FISH, fluorescence in situ hybridization
- GERD, gastroesophageal reflux disease
- GH, growth hormone
- GUCH, grown-up congenital heart disease
- MLPA, multiplex-ligation dependent probe amplification
- PTH, parathyroid hormone
- SLP, speech language pathologist
- SMCP, submucosal cleft palate
- TSH, thyroid stimulating hormone
- VPD, velopharyngeal dysfunction

- a – Proband and parents; strategy depending on test availability.
- b – When rare recessive condition associated with 22q11.2 region is suspected or atypical phenotypic features observed.
- c – Having seen many pediatric patients with 22q11.2DS both in consultation and in follow-up.
- d – Applies to children with and children without known CHD.
- e – Consider velopharyngeal port imaging (e.g., nasopharyngoscopy or speech videofluoroscopy) with cleft team (SLP and surgeon) when adequate speech output and articulation skills are present to allow for valid diagnostic imaging.
- f – Should include assessment of speech (e.g., articulation, resonance, voice), receptive and expressive language, and social/pragmatics skills.
- g – Dental assessment not relevant before age 2 years.
- h – Consider videofluoroscopic swallow study or fiberoptic endoscopic evaluation of swallowing if any signs or symptoms of aspiration.
- i – T cell phenotyping: CD3, CD4, CD8 cell counts (+ CD4/CD45RA), B cell count (CD19) and switched memory B cells (CD19 or CD20+, CD27+IgM-).
- j – Include antibodies against tetanus, diphtheria, and pneumococci.
- k – Especially important before VPD surgery to exclude instability; can be performed from age 4 years when sufficient bony ossification has occurred.
- l – Increased risk for obstructive sleep apnea after VPD surgery.

**Source:**  
[Updated clinical practice recommendations for managing children with 22q11.2 deletion syndrome](#)  
 Óskarsdóttir et al.  
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