22q11.2 Deletion Syndrome

Health Watch Table

(Includes DiGeorge Syndrome (DGS), Velocardiofacial Syndrome (VCFS), Shprintzen Syndrome, Conotruncal Anomaly Face Syndrome (CTAF), Caylor Cardiofacial Syndrome, and Autosomal Dominant Opitz G/BBB Syndrome)

1. Head, eyes, ears, nose, throat

Considerations	Recommendations
▸ *Children and Adults*: 15% have strabismus in addition to other ocular issues (e.g., cataracts, retinal problems) ▸ Conductive and/or sensorineural hearing loss (often unilateral) occur in 45% and 10% respectively ▸ Most have chronic otitis media ▸ 69% have palatial abnormalities, particularly velopharyngeal insufficiency (VPI) that is often associated with hyper-nasal speech, some of whom have submucosal cleft palate, and a small minority have overt cleft palate, which can lead to nasal regurgitation	▸ Refer to an ophthalmologist for assessment at diagnosis and during preschool years ▸ Refer to an audiologist for evaluation in infancy (or when diagnosed) and every 6 months up to 8 years of age, then annually until adulthood ▸ Often need regular ear cleaning to remove cerumen ▸ Examine the palate in infancy and evaluate for feeding problems and/or nasal regurgitation and, if warranted by clinical findings, refer to a cleft palate team ▸ Refer to a speech and language pathologist for assessment by 1 year of age, sooner if warranted or when diagnosis is made ▸ Evaluate nasal speech quality

2. Dental

Considerations	Recommendations
▸ *Children and Adults*: Retrognathia (over-bite) is common and may cause dental malocclusion ▸ Significant dental issues are a recognized part of the syndrome (e.g., enamel hypoplasia and chronic caries)	▸ Refer to a dentist in early childhood ▸ Advocate and ensure for appropriate dental care

3. Cardiovascular

Considerations	Recommendations
▸ *Children and Adults*: Between 40% and 74% have congenital heart defects, most commonly of the conotruncal type (e.g., Tetralogy of Fallot, interrupted aortic arch, ventricular septal defect)	▸ At the time of diagnosis, complete a cardiovascular assessment, including EKG and echocardiogram ▸ Refer to a cardiologist as warranted by clinical findings

4. Respiratory

Considerations	Recommendations
▸ *Children*: Congenital malformations may lead to upper and/or lower airway obstructions ▸ Most airway concerns resolve spontaneously with time but some require surgical intervention (e.g., Robin sequence)	▸ Refer to an ENT surgeon for evaluation as warranted by clinical findings ▸ Consider a pre-op anesthesia consultation regarding narrow airways prior to the first surgery

4. Respiratory (continued)

Considerations	Recommendations
Adults: In order of prevalence, there is an increased frequency of recurrent pneumonia, atelectasis, asthma, and chronic obstructive pulmonary disease	Consider periodic pulmonary function studies and referral to a pulmonologist as warranted by clinical findings

5. Sleep

Considerations	Recommendations
Children: Congenital malformations may lead to airway obstructions and obstructive sleep apnea (OSA)	Undertake a sleep study in infancy and then as warranted by clinical findings after 3 years of age
Adults: Those with uncorrected congenital malformations remain at risk for OSA	Undertake a sleep study as warranted by clinical findings

6. Gastrointestinal

Considerations	Recommendations
Children and Adults: Feeding difficulties, related to pharyngeal and gastrointestinal tract hypotonia, commonly lead to failure to thrive Dysphagia and constipation, with or without gastrointestinal structural anomalies, are common Gastroesophageal reflux also is common 20% develop gallstones	Refer to a gastroenterologist and feeding specialist (e.g., speech-language pathologist) Treat constipation If difficulty swallowing pills, adapt medication regime (e.g., provide with liquid medication, crush pills) If there are unexplained gastrointestinal findings or changes in behavior or weight, investigate for constipation, GERD, peptic ulcer disease, and pica. Screen annually for manifestations of GERD and manage accordingly. If introducing medications that can aggravate GERD, monitor more frequently for related symptoms. Consider obtaining an abdominal ultrasound in adults to assess for gallstones

7. Genitourinary

Considerations	Recommendations
Children and Adults: Up to 33% may have renal tract anomalies 10% may develop renal failure in adulthood	Undertake a renal ultrasound at the time of diagnosis Maintain surveillance for urinary tract infections (UTIs) Determine creatinine levels at diagnosis and annually thereafter

8. Sexual function

Considerations	Recommendations
Children and Adults: People with the 22q11.2 deletion syndrome are fertile and have a 50% chance of transmitting the 22q11.2 deletion to children	Referral for genetic counseling may be appropriate

9. Musculoskeletal

Considerations	Recommendations
▸ *Children and Adults*: Many have skeletal abnormalities, most commonly vertebral or rib anomalies ▸ A minority have short stature during childhood that improves by adulthood	▸ Undertake cervical spine X-rays after age 4 years to assess for vertebral anomalies and instability on flexion/extension (five views: flexion, extension, AP, lateral, and open mouth) ▸ Arrange chest X-ray to evaluate for thoracic vertebral anomalies ▸ Provide clinical evaluation for scoliosis at diagnosis, during preschool, and periodically thereafter

10. Neurological

Considerations	Recommendations
▸ *Children and Adults*: Impairments due to reduced muscle tone and motor delay are common in children ▸ Seizures are frequently associated with hypocalcemia ▸ 40% of adults have recurrent (often hypocalcemic) seizures ▸ Cord compression may occur related to skeletal anomalies	▸ Undertake a neurodevelopmental assessment of infants with particular attention to reduced muscle tone and motor delay ▸ Refer to a physical therapist (PT) and/or occupational therapist (OT), as needed ▸ Ascertain history with attention to seizures ▸ Consider checking serum ionized calcium following any seizure activity ▸ Include EEG examination in evaluation if indicated ▸ Symptoms of cord compression are an indication for an emergent referral to a neurologist or neurosurgeon

11. Behavioral / mental health

Considerations	Recommendations
▸ *Children and Adults*: Conditions such as autism spectrum disorder (ASD), attention deficit disorder (ADD), attention deficit hyperactivity disorder (ADHD), and obsessive-compulsive disorder (OCD) are common ▸ Treatable anxiety disorders and depression are common ▸ Behavioral differences may begin at a young age ▸ 60% of adults have a psychiatric disorder ▸ Schizophrenia can become apparent in adolescence and 25% develop schizophrenia or other psychotic disorders in adulthood	▸ Ascertain comprehensive behavioral and mental health history ▸ Refer to a psychiatrist if evidence of ASD, ADD, ADHD, or OCD occurs ▸ Screening children for psychiatric issues before age 10 years may provide an opportunity for early intervention ▸ Assess for psychiatric illness with attention to changes in behavior, emotional state and thinking, including hallucinations or delusions and at-risk behaviors (e.g., sexual activity, alcohol/drug use) in teens and adults ▸ Refer to a psychiatrist as warranted by clinical findings

12. Endocrine

Considerations	Recommendations
▸ *Children and Adults*: 60% have episodic hypocalcemia (often missed when mild or transient) ▸ Hypocalcemia is due to hypoparathyroidism in children and adults ▸ Long-term calcium supplementation can lead to renal calculi	▸ Measure serum ionized calcium concentration in neonates, then annually to assess for hypoparathyroidism ▸ Assess calcium levels in infancy, every 3 to 6 months, every 5 years through childhood, and every 1 to 2 years thereafter ▸ Be vigilant regarding risk of hypocalcemia with acute illness and childbirth

12. Endocrine (continued)

Considerations	Recommendations
▸ Hypo- and hyperthyroidism have been reported in children and adults ~ 4% have growth hormone deficiency ~ 35% of adults are obese ~ 20% of adults have hypothyroidism ~ 5% of adults have hyperthyroidism	▸ All patients should have vitamin D supplementation; those with documented hypocalcemia and/or relative or absolute hypoparathyroidism may require prescribed hormonal forms supervised by endocrinologist ▸ Refer to an endocrinologist as warranted by clinical and laboratory findings and for initial management of hypocalcemia ▸ Consider densitometry to assess for osteopenia earlier than in general population ▸ Undertake T4 and TSH baseline screening ▸ Treat with standard thyroid replacement or antithyroid therapy where warranted ▸ Monitor growth and growth hormone levels annually and consider endocrinology assessment for poor growth

13. Hematological

Considerations	Recommendations
▸ *Children and Adults*: Autoimmune diseases (e.g., thrombocytopenia, juvenile rheumatoid arthritis, Grave's disease, vitiligo, neutropenia, hemolytic anemia) may be more common than in the general population ▸ 10% develop splenomegaly	▸ Monitor with CBC; thyroid function annually or if concerns arise ▸ Investigate arthritis problems for juvenile rheumatoid arthritis and refer to a rheumatologist as warranted

14. Infectious disease/immunization

Considerations	Recommendations
▸ *Children and Adults*: Congenital thymic aplasia is recognizable in infancy ▸ 77% have immune deficiency (although thymic aplasia is rare, thymic hypoplasia is common); improvement in T-cell production may occur over time ▸ 75% have chronic otitis media and frequent respiratory infections ▸ Irradiated blood products have been used when blood replacement has been necessary ▸ Recurrent upper and lower respiratory tract infections are common in adults	▸ In addition to obtaining a CBC with differential in newborns, consider undertaking flow cytometry. ▸ For infants, minimize exposure to infectious diseases and withhold live vaccines initially. Refer infants to an infectious disease specialist to assess regarding influenza vaccines, CMV-negative irradiated blood products and RSV prophylaxis ▸ Measure absolute lymphocyte count following initial diagnosis and refer to an immunologist if count is low. At age 9 to 12 months (prior to live vaccines), assess flow cytometry, immunoglobulins and T-cell function ▸ Evaluate immune status before offering any live vaccines ▸ Treat respiratory and other infections aggressively in children and adults

15. Dermatological	
Considerations	**Recommendations**
▸ *Children and Adults*: 35% have seborrhea or dermatitis ▸ 25% have severe acne	▸ Examine skin as part of routine care ▸ Treat as per general population, with referral to dermatologist as needed

16. Other	
Considerations	**Recommendations**
▸ Incidence: 1/4000, but more likely higher and many without typical features ▸ Huge variability in level of developmental disability and the number and severity of associated features ▸ IQ: The majority of affected people with 22q11 deletion fall in the high mild to borderline range; moderate to severe rates and average levels of IQ are less common ▸ A selection bias in reported studies may result in overestimating some prevalence rates	

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Expert Clinician Reviewer

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Modified with permission of Surrey Place Centre. This tool was reviewed and adapted for U.S. use by physicians on the Toolkit's Advisory Committee.

Resources

- 10 published 22q11.2 deletion syndrome health care guidelines reviewed and compared. (For full list of references see http://ddprimarycare.surreyplace.ca/wp-content/uploads/2018/03/HWT_22q11.2del.pdf. Accessed August 2025.
- 22q11.2 Deletion syndrome websites that may be helpful for families and caregivers
  www.c22c.org. Accessed August 2025. www.22q.org. Accessed August 2025.

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