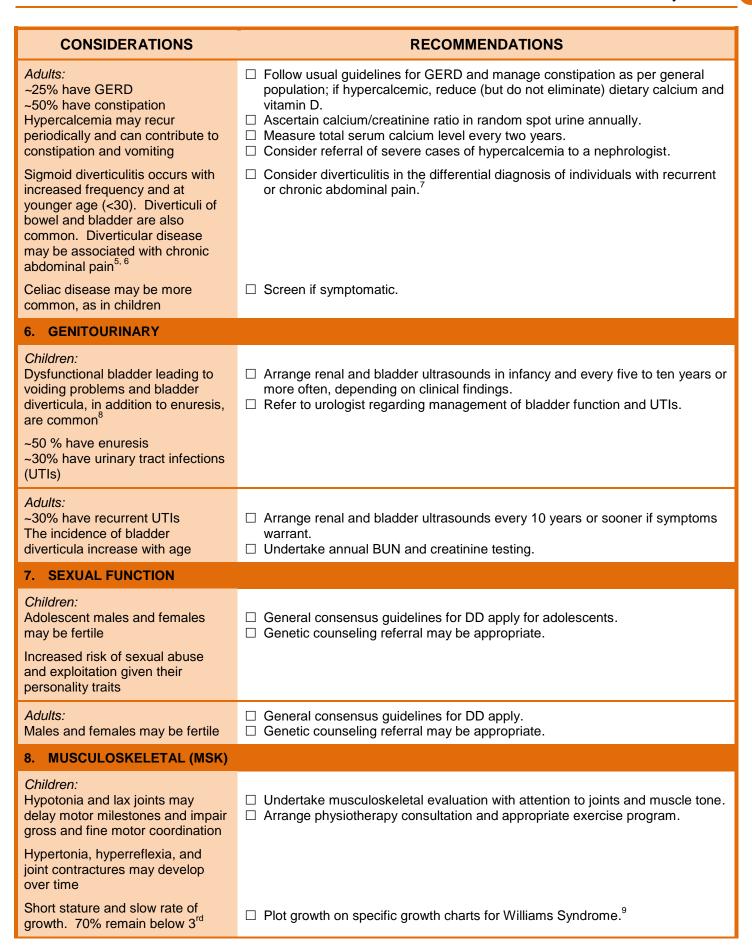


Health Watch Table — Williams Syndrome Forster-Gibson and Berg 2013

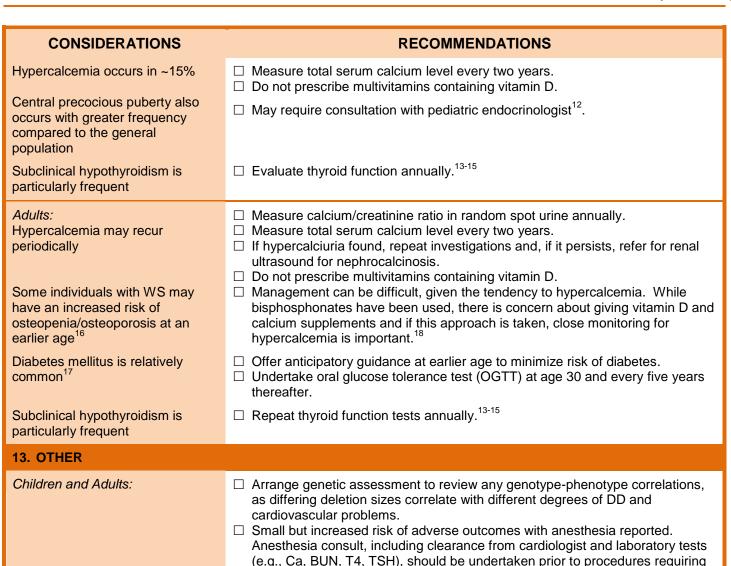
CONSIDERATIONS	RECOMMENDATIONS			
1. HEENT (Head, Eyes, Ears, Nose, Throat)				
Children: ~60% have chronic otitis media	 □ Arrange hearing assessment at 6-12 months and annual screen thereafter. □ Check for wax accumulation if indicated. □ Refer for tympanoplasty, if indicated. 			
Nearly all have characteristic auditory profile including hyperacusis [84-100%] and paradoxic affinity to music. Early onset phonophobia (fear of noises) also common	 Encourage caregivers to reduce exposure to loud noises (e.g., electric household appliances and machines, fireworks, thunder) and recommend earplugs in noisy environments. Use headphones with music in uncomfortable situations, e.g., school-bus ride or watching a noisy event. 			
Noise tolerance improves with age				
Strabismus and refractive errors such as hyperopia are common	 Arrange ophthalmologic exam by 1 year and vision screening annually thereafter. 			
Amblyopia and reduced depth perception can cause difficulty in negotiating uneven surfaces and stairs	☐ See also #8, MUSCULOSKELETAL regarding mobility.			
Blocked tear ducts are common				
Hoarse, deep voice				
Adults: Vulnerable auditory system with risk of hearing loss	 Refer for audiology examination at age 30 and every five years thereafter to assess sensorineural hearing loss. Check for wax accumulation, if indicated. Follow general consensus guidelines for DD for vision assessment. 			
2. DENTAL				
Children: Missing or small teeth, malocclusion and other dental anomalies are common. Visual- motor integration difficulties interfere with dental hygiene	 □ Arrange early dental evaluation; recommend dental cleaning every four months by adolescence and advise caregiver to provide daily supervision or assistance with brushing and flossing. □ Make orthodontic referral by 8 years, as needed. 			
Adults:	 □ Refer for dental cleaning every four months and recommend use of sealants. □ Supervise brushing and flossing. 			
3. CARDIOVASCULAR				
Children: Stenosis of medium to large-sized arteries is common; typically supravalvular aortic stenosis (SVAS) in ~45% and/or peripheral pulmonary stenosis (PPS) in ~37% ¹ Mitral valve prolapse (MVP), usually mild, may be present in	 □ Refer for cardiology evaluation in infancy and annually to age 5, then every 3-5 years (or more frequently if clinically warranted). □ Close and ongoing monitoring by a cardiologist should be determined by severity of findings. 			
~15%				

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CONSIDERATIONS	RECOMMENDATIONS				
Spontaneous improvement without surgical or catheter-based interventions of mild to moderate lesions may occur in childhood					
Hypertension (HTN) is common in adolescence but may present earlier	 Measure BP annually in both arms and legs (to detect stenoses), preferably in a relaxing environment and using manual cuffs, to minimize anxiety-related response. If HTN is present, assess for arterial stenosis, renal disease, and hypercalcemia. 				
Adults: SVAS is common. Pulmonary artery stenosis may have resolved	 Arrange cardiac evaluation for mitral valve prolapse, aortic insufficiency, and arterial stenosis. 				
HTN is common in adults and may be severe	 Measure BP as in children. If HTN is present, assess for arterial stenosis, renal disease, and hypercalcemia. 				
4. RESPIRATORY					
Children and Adults: Sleep disorders, such as sleep anxiety, night waking, restless sleep and daytime sleepiness appear to be fairly common and improve with increased age Habitual snoring, obstructive sleep apnea (OSA), have been reported	 Ascertain comprehensive sleep history and refer for sleep study as appropriate. Consider the possibility that sleep disturbance may have a negative impact on daytime behavior before diagnosis of behaviourally defined disorder is entertained.² The use of Melatonin to improve sleep patterns has received anecdotal support.³ 				
5. GASTROINTESTINAL					
Children: Early failure to thrive, colic, Gastroesophageal Reflux Disease (GERD), vomiting and, hypercalcemia, are common	 □ Undertake clinical evaluation and history with attention to GERD and feeding difficulties. □ Recommend small, frequent, high caloric density meals for infants. □ Consider feeding and oral motor therapy for child with significant feeding difficulty. □ Measure total serum calcium level every two years. □ Ascertain calcium/creatinine ratio in random spot urine annually. 				
Constipation is variously due to hypercalcemia, to low muscle tone and to low-fibre diet; when chronic, diverticular disease often occurs and occasional bowel perforation has also occurred	 Manage constipation as per general population (e.g., institute regular, routine toilet regimen) and if hypercalcemic, reduce (but do not eliminate) dietary calcium and vitamin D (see #12 ENDOCRINE). Advise parent that chronic constipation can lead to diverticular problems. Consider referral of severe cases to nephrologist (infants may require therapy with pamidronate). 				
Sensory issues, notably texture aversion may affect feeding	 Consider extending period of feeding with pureed food to circumvent sensory aversion to some food textures. Consultation with a feeding specialist (speech therapist) can help with transition to solid food. 				
Increased frequency of celiac disease has been reported ⁴	☐ Screen once after 3 years of age or if patient is symptomatic.				
Hernias (inguinal and umbilical) due to connective tissue abnormality, are relatively common and may occur at any age	☐ Ascertain if present at regular/routine medical visits.				



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CONSIDERATIONS	RECOMMENDATIONS			
percentile for mid-parental height Adolescents may have awkward gait, scoliosis, kyphosis and lordosis				
Adults: Hypertonia, hyper-reflexia, and joint contractures may develop over time. Adults may have awkward gait, scoliosis, kyphosis and lordosis	 ☐ Undertake musculoskeletal evaluation with attention to joints and muscle tone. ☐ Arrange physiotherapy consult and appropriate exercise program. 			
9. NEUROLOGICAL				
Children: Hypotonia, mild cerebellar and extrapyramidal signs are common ¹⁰	 □ Refer to neurologist, if indicated. □ Consider Occupational Therapy (OT) referral for sensory integration. □ Consider evaluation for Chiari I malformation if individual complains of headache, dysphagia, dizziness or weakness. 			
Adults: There have been some reports of cerebrovascular accidents, possibly due to intracranial stenosis and/or hypertension 11	 Investigation for and management of symptoms of stroke should follow guidelines for the general population. 			
10. DERMATOLOGICAL				
Children and Adults:	☐ Recommend diligent use of sunscreen to reduce vitamin D absorption and hypercalcemia.			
11. BEHAVIOURAL/MENTAL HEALTH				
Children: Full-scale IQ can be misleading with most in mild to moderate range of impairment Typical cognitive profile includes: strengths in verbal short-term memory and language; unusual degree of interest and enjoyment of music; weakness in fine motor skills (e.g., buttoning, handwriting, drawing) contrast with strength in visual recognition (e.g., reading achievement) Common behavioural concerns include overfriendliness, excessive empathy, attention deficit, anxiety, and specific phobias	 □ Arrange developmental and neuropsychological evaluations to assist in developing early intervention and special education programs, and vocational training programs. □ Make psychiatric referral, if appropriate. □ Consider occupational therapy (including sensory integration), physiotherapy and speech language therapy, as well as behavioural and pharmacological therapy for anxiety and other disorders. 			
Adults: May have increasing anxiety and social withdrawal	☐ Follow general consensus guidelines for DD, keeping specific behavioural concerns in mind.			
12. ENDOCRINE				



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RESOURCES

Williams Syndrome websites that may be useful for families and caregivers:

Canadian Association for Williams Syndrome www.caws-can.org
The Williams Syndrome Association (WSA) (USA) www.williams-syndrome.org
Williams Syndrome Foundation (UK) – Clinical guidelines
http://wsf.bigorangesoftware.co.uk/vebo/index.php?idPage=102

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Thanks to the following clinicians for their review and helpful suggestions.

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