



Down Syndrome Practice Tool Series 5 of 5

# **Medical Care for the Child with Down Syndrome from 13 to 21+ Years of Age**



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*When your child needs a hospital, everything matters.*

## Brief Overview of Down Syndrome

Down syndrome is the most common chromosomal condition in the United States. About 1 in every 700 babies born will be diagnosed with Down syndrome, totaling to approximately 6,000 babies each year.<sup>1</sup> Down syndrome is caused by the presence of the genetic material from a third copy of chromosome 21 (trisomy 21) or an unbalanced translocation between chromosome 21 and another chromosome. Approximately 95% of cases of down syndrome are sporadic, with no familial history.<sup>2</sup>

In addition to common physical features (e.g. hypotonia, small brachycephalic head, epicanthal folds, and flat nasal bridge), a number of life-time associated health conditions may affect individuals with Down syndrome. Some are age specific but can occur at a variety of ages.

More Common Conditions	%	Less Common Conditions	%
Hearing problems	75	Gastrointestinal atresias	11
Vision problems	60	Thyroid disease	4–18
Refractive errors	50	Seizures	1–13
Obstructive sleep apnea	50–75	Iron deficiency	10
Otitis media	50–70	Transient myeloproliferative disorder	10
Congenital heart disease	40–50	Celiac disease	5
Hypodontia/ delayed dental eruption	23	Anemia	3
Cataracts	15	Other (atlantoaxial instability, autism, Hirschsprung disease, leukemia)	1–2

Here, we summarize guidelines designed by the American Academy of Pediatrics to assist the pediatrician/primary care providers with medical care for an adolescent or young adult with Down syndrome between 13 and 21+ years of age.<sup>2</sup>

## Periodic Care and Follow Up Specific for Adolescents and Young Adults (13–21+ Years)

Several aspects of care require ongoing assessment throughout childhood and adulthood and should be reviewed at every physician visit, while others may be conducted at different time intervals depending on the patient's age and health care needs.

• Check hemoglobin	Annually
• Assess TSH to screen for hypothyroidism	Annually
• Instruct to contact physician for change in gait, change in use of arms or hands, change in bowel or bladder function, neck pain, head tilt, torticollis, or new-onset weakness	Biennially
• If patient has normal ear-specific hearing, assess behavioral audiogram	Annually
• Refer to pediatric ophthalmologist or ophthalmologist with experience with Down syndrome	Every three years
• Discuss physical and psychosocial changes though puberty, need for gynecologic care and the possibility of pregnancy in the pubescent female	Do once at this age
• Provide influenza vaccine	Annually

## Care and Follow Up Needs at All Visits or All Health Maintenance Visits (HMs)

• Assess the emotional status of parents/guardians and intrafamilial relationships	All HMs
• Review signs and symptoms of hypotonia and myopathy — refer for therapies	All HMs
• Discuss cervical spine positioning, especially for anesthesia or surgical or radiologic procedures	All HMs

## Care and Follow Up Needs at All Visits or All Health Maintenance Visits (HMsVs) (Continued)

• Advise risk of some contact sports and trampolines	All HMsVs
• Check for symptoms of celiac disease; if symptoms present, obtain tissue transglutaminase immunoglobulin (Ig) A & quantitative IgA	All HMsVs
• Assess for obstructive sleep apnea symptoms (see STOP-BANG approach)	All HMsVs
• Facilitate early intervention for physical, occupational, and speech therapy	All HMsVs
• Facilitate transitions when appropriate: <ul style="list-style-type: none"> <li>• Guardianship, financial planning, behavioral problems, school placement, vocational training, independence with hygiene (including routine gynecologic care for females) and self-care, group homes, and work settings</li> </ul>	HMsVs
• Discuss sexual development and behaviors, contraception, sexually transmitted diseases, and recurrence risk for offspring when appropriate	HMsVs
• If patient presents myelopathic signs or symptoms, obtain neutral position spine films	All visits
• If patient has congenital heart disease, monitor for signs & symptoms of congestive heart failure	All visits

## Assessing Obstructive Sleep Apnea in Adults with Down Syndrome

The STOP-BANG questionnaire should be used to screen for moderate to severe obstructive sleep apnea in all adults with Down Syndrome, but providers and care takers should be aware of other changes common in those with Down Syndrome, including behavioral changes, uncommon sleep positions, and restless sleep.

The STOP-BANG approach ([www.StopBang.ca/osa/Screening.php](http://www.StopBang.ca/osa/Screening.php)):

- Snoring
- Tired/fatigued regularly
- Observed pauses or choking and gasping
- Pressure — high blood pressure (not a reliable measure in Down Syndrome)
- Body Mass Index >35 kg/m<sup>2</sup>
- Age >50 years
- Neck size >16 inches
- Gender — male

## Situation Appropriate Screening

- Diabetes screening in those with elevate BMI (>20 kg/m<sup>2</sup>) or other health risks
- Cardiovascular/stroke screening to check lipid levels and treat based on risk evaluation
- Follow-up for congenital heart disease and transition from pediatric provider to adult provider as needed
- If prone to fractures or sedentary lifestyle, consider screening for osteoporosis every 2–3 years
- Changes in behavior, such as not enjoying things they used to, withdrawn, slowed movements/activities, and significant weight loss/gain
  - Consider treatable medical conditions, such as thyroid disease, celiac disease, anemia, obstructive sleep apnea, hearing/vision issues, and diabetes (less likely)
  - Consider depression/anxiety, especially in those aged 20–40 years but even in those with ≥40 years
  - Consider Down syndrome regression/Down syndrome disintegrative disorder which may not have an identifiable medical cause but can respond to various treatments- refer to the Down syndrome specialty clinic through Nationwide Children's or the Nisonger Center at The Ohio State University for further evaluation
  - Consider these changes may be a form of early dementia in those with ≥40 years

## Specialty Services at Nationwide Children's and When to Refer

### Down Syndrome Clinic

The Down Syndrome Clinic at Nationwide Children's Hospital is a family-centered clinic providing diagnostic and treatment services to children, adolescents, and young adults with Down syndrome and their families.

Families are linked with medical, educational, social and financial supports in their communities as indicated and resources identified to assist them in meeting the unique needs of the patient.

Our team of developmental pediatricians, psychologists, geneticists and advanced practice nurses maintain a close working relationship with the child's/patient's primary care physician throughout their care.

### Services We Offer

Services are covered by most insurance providers. To expedite insurance coverage we recommend a referral be sent to the clinic by the patient's primary care physician.

- Comprehensive evaluation — Monitoring for feeding issues, atlantoaxial instability and signs of celiac disease
- Assessments for Attention Deficit Hyperactivity Disorder or Autism Spectrum Disorders in individuals with Down syndrome
- Lab and radiology assessments, including recommended health supervision screening and focusing on individual concerns
- Behavior management counseling for families whose children are experiencing behavioral difficulties

### Reasons to Call

Parents/caregivers should notify the patient's pediatrician or the Down Syndrome Clinic for the following symptoms:

- Changes in gait
- Changes in use of arms or legs
- Changes in bowel or bladder habits
- New onset of snoring
- New onset of apnea when sleeping or new onset of sleepiness during the day
- New behavior or learning concerns

### Additional Resources

Parents and caregivers can find a full list of resources and support groups on the Nationwide Children's Downs Syndrome Clinic webpage. [NationwideChildrens.org/specialties/down-syndrome-clinic](https://www.nationwidechildrens.org/specialties/down-syndrome-clinic)

This tool is part of a 5-part series of practice tools. Please reference the other tools for information relevant to other age groups.

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**References:** 1) Centers for Disease Control and Prevention. Data and Statistics on Down Syndrome. Accessed 3/21/2021 at: <https://www.cdc.gov/ncbddd/birthdefects/downsyndrome/data.html>. 2) Bull MJ; Committee on Genetics. Health supervision for children with Down syndrome. *Pediatrics*. 2011 Aug;128(2):393-406. doi: 10.1542/peds.2011-1605. Epub 2011 Jul 25. Erratum in: *Pediatrics*. 2011 Dec;128(6):1212. PMID: 21788214 (<https://pubmed.ncbi.nlm.nih.gov/21788214/>).

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## Referrals and Consultations

Online: [NationwideChildrens.org/Request-An-Appointment](https://www.nationwidechildrens.org/Request-An-Appointment)

Phone: (614) 722-6200 or (877) 722-6220 | Fax: (614) 722-4000

Physician Direct Connect Line for 24-hour urgent physician consultations:

(614) 355-0221 or (877) 355-0221.

