Division of Genetic and Genomic Medicine

Down Syndrome Practice Tool Series 1 of 5

Medical Care for the Child with Down Syndrome from Birth to 1 Month

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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.[1] 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.[2]

In addition to common physical features (e.g. hypotonia, small brachycephalic head, epicanthal folds, and flat nasal bridge), a number of associated health conditions may affect babies with Down syndrome. Here, we summarize guidelines designed by the American Academy of Pediatrics to assist the pediatrician with medical care for a child with Down syndrome from birth to 1 month of age.[2]

Prenatal screening/diagnosis of Down Syndrome

The American College of Obstetricians and Gynecologists recommends that all pregnant women be offered the option of screening testing for Down syndrome. When discussing screening/screening results, remind patients/families that prenatal screening is not equal to diagnosis.

Screening ≠ Diagnosis ; Negative Screen ≠ No Diagnosis

Prenatal screening results suggesting a diagnosis of Down syndrome need to be confirmed with invasive testing or postnatal testing.

- If a karyotype from chorionic villous sampling/amniocentesis (diagnostic) → No additional testing needed
- If any non-invasive/cell-free DNA test (NIPS) and family declined invasive screening → postnatal cytogenetic analysis (preferably a STAT karyotype) to confirm the diagnosis and rule out a chromosome translocation

Before jumping into providing medical guidelines, if prenatal diagnosis is known, remember to first congratulate the parents on the birth of their child prior to providing information on Down syndrome.

In the postnatal setting, clinical suspicion is grounds to start a discussion about diagnosis but keep in mind that the diagnosis still has to be confirmed.

When giving diagnosis/sharing a clinical suspicion, do so in a private setting. Delivery of the "unexpected news" (never referred to as "bad news") should come from should be a provider who is knowledgeable about the condition (physician, nurse, genetic counselor, social worker) and, if possible, someone who the parents/family knows and is comfortable with (e.g. someone previously involved in the mother and child's care).

- Be prepared, honest, and positive (Be aware of the possibilities for people with Down syndrome)
- Do not whisper, say "I'm sorry", or give inaccurate information
- Provide resources such as from the Down Syndrome Association of Central Ohio (dsaco.net) or other local organization AND the required Ohio Department of Health Down Syndrome Fact Sheet (https://odh.ohio.gov/wps/portal/gov/odh/know-our-programs/down-syndrome/down-syndrome)

Periodic Care and Follow Up Specific for Babies From Birth to 1 Month

Several aspects of care require ongoing assessment throughout childhood 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.

• Confirm diagnosis with chromosomal analysis	Do if not done previously
• Referral to Genetics for evaluation and support	Do once at this age
• Provide parent-to-parent contact, support groups, current books and pamphlets (see Additional Resources)	Do if not done previously
• Conduct echocardiogram (regardless of prenatal echocardiogram)	Do once at this age
• Check red reflex to screen for cataracts	Do once at this age
• Conduct newborn hearing screen and complete follow-up	Do once at this age
• Collect history, conduct physical exam for duodenal or anorectal atresia	Do once at this age
• Evaluate complete blood count (CBC) to rule out transient myeloproliferative disorder, polycythemia	Do once at this age
• Assess TSH (if born in state that uses T4 in newborn screening; see www.newsteps.org/data-resources/reports/screening-methodologies-and-targets-report and under "Screening Target" select "T4")	Do once at this age
• If family requests, review physical exam for evidence of trisomy 21 with parents	Do once at this age
• If needed, conduct radiographic swallowing assessment if marked hypotonia, slow feeding, choking with feeds, recurrent or persistent respiratory symptoms, failure to thrive	Do once at this age
• Discuss risk of respiratory infection due to aspiration/ congenital heart disease	Do once at this age

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

Assess the emotional status of parents and intrafamilial relationships	All HMVs
Review signs and symptoms of hypotonia and myopathy- refer for therapies, including Help-Me-Grow if appropriate	All HMVs
Discuss cervical spine positioning, especially for anesthesia or surgical or radiologic procedures	All HMVs
Discuss complementary & alternative therapies	All HMVs
If infant is experiencing constipation, evaluate for limited diet or fluids, hypotonia, hypothyroidism, gastrointestinal malformation, & Hirschsprung disease	All visits
If infant presents myelopathic signs or symptoms, obtain neutral position spine films	All visits
If infant has congenital heart disease, monitor for signs & symptoms of congestive heart failure	All visits

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/).

Specialty Services at Nationwide Children's and When to Refer

Down Syndrome Clinic

For initial assessments, please refer to the Genetics Clinic to discuss test results and general outcomes. We can also address reoccurrence risk and provide psychological support if needed.

The Genetics and Down Syndrome Clinic through the Developmental and Behavioral Health Department at Nationwide Children's Hospital is a family-centered clinic providing diagnostic and treatment services to children and adolescents 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 their child.

Our team of developmental pediatricians, psychologists, geneticists and advanced practice nurses maintain a close working relationship with the child'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 difficulty in infants, 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 use of arms or legs
- Changes in bowel or bladder habits
- New onset of snoring
- New onset of apnea when sleeping

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**

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

Referrals and Consultations

Online: 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.

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