Clinical Practice Guidelines for Management of Children With Down Syndrome: Part I

Daniel L. Ivan, MD, & Polly Cromwell, MSN, RN, CPNP

KEY WORDS
Down syndrome, practice guideline, trisomy 21

Down syndrome (DS), also known as trisomy 21, is a common genetic disorder that is the result of having an extra copy of chromosome 21. The Centers for Disease Control and Prevention (CDC) estimates that each year about 6000 babies are born with DS (an average of 1 in 691 infants born in the United States), without any predilection of race or socioeconomic class (CDC, 2011). The likelihood of having a baby with DS increases with advanced maternal age. If the odds are 1:1441 when the mother is 20 years old, they increase to 1:338 when the mother reaches 35 years of age and can be as high as 1:84 for a woman 40 years old (Morris, Mutton, & Alberman, 2002).

Approximately 95% of children with DS have this condition because of the presence of one extra chromosome 21. Another 3% to 4% of patients present with an unbalanced translocation; these patients have a karyotype with 46 chromosomes, but one of the chromosomes (usually chromosome 14) carries extra chromosomal material from chromosome 21. Twenty-five percent of children with an unbalanced translocation (0.75% to 1% of those diagnosed with DS) receive the affected chromosome from one of the parents, and thus additional familial studies and genetic counseling are necessary (Bull & the Committee on Genetics, 2011). The remainder of the patients with DS have mosaicism, in which case individuals have both normal cells and cells with an extra chromosome 21.

Apart from characteristic physical features (see the Physical Examination section), most children with DS have a variable degree of cognitive impairment, ranging from mild impairment (intelligence quotient [IQ] of 50 to 70) to severe impairment (IQ of 20 to 35). Children with DS carry a significant degree of morbidity because of associated conditions such as hearing loss (75%), otitis media (50% to 70%), eye disorders (60%), obstructive sleep apnea (50% to 79%), congenital heart defects (50%), gastrointestinal (GI) problems (12%), hip dislocation (6%), thyroid disease (4% to 18%), hematologic disorders (4% to 10%), and neurologic dysfunction (1% to 13%; Bull & the Committee on Genetics, 2011).

The complexity of this condition makes it challenging for primary care physicians, nurse practitioners, and physician assistants alike to render adequate care and follow-up. In 2011 the American Academy of

Section Editors
Robert J. Yetman, MD
Corresponding Editor
University of Texas School of Medicine
Houston, Texas

Polly F. Cromwell, MSN, RN, CPNP
Yale-New Haven Children's Hospital
Bridgeport Hospital Campus
Bridgeport, Connecticut

Daniel L. Ivan, Assistant Professor, Department of Pediatrics, Division of Community & General Pediatrics, University of Texas–Houston Medical School, Houston, TX.

Polly Cromwell, Pediatric Nurse Practitioner, Yale-New Haven Children's Hospital, Bridgeport Campus, and Pediatric Primary Care Center, Bridgeport, CT.

Conflicts of interest: None to report.

Correspondence: Daniel L. Ivan, MD, Department of Pediatrics, Division of Community & General Pediatrics, University of Texas–Houston Medical School, 6431 Fannin St, JJL-495, Houston, TX 77030; e-mail: Daniel.L.Ivan@uth.tmc.edu.

J Pediatr Health Care. (2014) *28*, 105-110.

0891-5245/$36.00

Published online July 26, 2013.

http://dx.doi.org/10.1016/j.pedhc.2013.05.002

Pediatrics (AAP) Committee on Genetics published updated guidelines for the management of children with DS, replacing those published in 2001 (AAP, Committee on Genetics, 2001). The recommendations in the new guidelines span from the prenatal period through adulthood (Bull & the Committee on Genetics, 2011). The purpose of this Practice Guideline, divided into two parts, is to summarize the new guidelines in a more direct manner for the primary care practitioner. Part One will address the health maintenance of patients with DS until their first birthday, whereas Part Two will focus on their care as these children transition to adulthood.

THE PRENATAL VISIT

In 2007, the American College of Obstetricians and Gynecologists (ACOG) recommended that all women be offered aneuploidy screening before 20 weeks of gestation regardless of maternal age. The detection rate of DS through combined prenatal screenings performed during the first and second trimester is 95% (ACOG, 2007). Screening during the first trimester takes into account the maternal age, along with measurement of fetal nuchal translucency through ultrasonography and maternal serum human chorionic gonadotropin and pregnancy-associated plasma protein A. During the second semester, screening again assesses the maternal age risk and also includes measurements of maternal serum human chorionic gonadotropin, unconjugated estriol, alpha fetoprotein, and inhibin levels.

Once the diagnosis is entertained, pediatric health care providers are often involved to counsel the families and should address the following topics:

- Prenatal laboratory and imaging studies that led to the diagnosis
- Available genetic counseling
- Decision to continue or terminate pregnancy (in cases of early prenatal diagnosis)
- Clinical manifestations and prognosis
- Additional fetal studies (e.g., echocardiogram and ultrasound of the GI tract)
- The need for follow-up with various subspecialists (e.g., a cardiologist, gastroenterologist, and ophthalmologist)
- Available treatments and intervention
- Delivery plan and neonatal care
- Local and national support groups

BIRTH TO 1-MONTH VISITS

History

- Review prenatal information (first- and second-trimester screening results and karyotype results).
- Review family history (previous pregnancies that ended in miscarriages, children born with DS, and developmental delay).

Physical Examination

Physical examination in the first 24 hours of life is a very important tool for diagnosing DS (Bull & the Committee on Genetics, 2011). Although the clinical presentation of children with DS is variable, the following physical features are quite suggestive:

- Epicanthal folds (i.e., skin fold of the upper eyelid, covering the medial canthus of the eye)
- Flat nasal bridge
- Narrow, upward-slanting palpebral fissures
- Brushfield spots (i.e., white or yellow spots seen on the anterior surface of the iris)
- Small, brachycephalic (flat) head
- Small mouth and small dysplastic ears
- Excessive skin on the nape of the neck
- Single (or bridged) palmar crease
- Short fifth finger with clinodactyly (i.e., a bent or curved finger due to mid phalanx hypoplasia)
- Generalized hypotonia (one of the most striking characteristics; Weijerman & de Winter, 2010)

Other common physical findings or symptoms in newborns with DS are:

- Head, eyes, ears, nose, throat
  - — Abnormally large anterior fontanelle
  - — Fine, soft, and sparse hair
  - — Midfacial hypoplasia
  - — Abnormal/asymmetric red reflex
  - — Large tongue compared to the size of the mouth
- Respiratory
  - — Stridor
  - — Wheezing
  - — Noisy breathing
- Cardiac
  - — Dyspnea
  - — Cyanosis
  - — Murmur(s)
- GI
  - — Easy fatigability with feedings
  - — Poor suck
  - — Emesis
  - — Abdominal distention
  - — Diastasis recti (separation of the rectus abdominis muscles into right and left halves)
  - — Imperforate anus
- Genitourinary
  - — Micropenis (in boys) or lower labial index (labia majora shorter and wider than normal in girls)
  - — Cryptorchidism
  - — Hypospadias
- Extremities
  - — Wide gap between the first and second toe
- Skin
  - — Pale, blotchy skin
  - — Persistent, worsening jaundice