

Down Syndrome

Health Care Guidelines

Neonatal (Birth - 1 Month):

- Review parental concerns. Chromosomal karyotype; genetic counseling, if necessary.
- If vomiting or absence of stools, check for gastrointestinal tract blockage (duodenal web or atresia or Hirschsprung disease).
- Evaluation by pediatric cardiologist including echocardiogram. Subacute endocarditis prophylaxis - (SBE) in susceptible children with cardiac disease.
- Exam for plethora, thrombocytopenia.
- Review feeding history to insure adequate caloric intake.
- Thyroid function test - check on results of state-mandated screening at birth.
- Auditory Brainstem Response (ABR) or Otoacoustic Emissions (OAE) test to assess congenital sensorineural hearing (at birth or three months).
- Pediatric Ophthalmological evaluation (by 6 months) for screening purposes.
- Discuss Early Intervention (infant stimulation) and refer for enrollment in local program.
- Referral to local Down syndrome parent group or family support and resources as indicated.

Infant (1-12 months):

- General neurological, neuromotor and musculoskeletal examination.
- TSH and T4- Thyroid Function Test (6 & 12 months).
- Evaluation by pediatric cardiologist including echocardiogram (if not done at birth). Consider progressive pulmonary hypertension if VSD or atrioventricular septal defect and little or no symptoms of heart failure.
- Subacute bacterial endocarditis prophylaxis-SBE (as indicated).
- Feeding consult, especially if constipated. Consider Hirschsprung disease.
- Well-Child Care: immunizations.

- Auditory brainstem response (ABR) or otoacoustic emission (OAE) test to assess congenital sensorineural hearing (by 3 months if not already done or if suspicious).
- Ear, Nose and Throat exam (as needed), especially if suspicious of otitis media.
- Well balanced, high fiber diet.
- Vision Exam (by 6 months and annually; earlier if nystagmus, strabismus or indications of poor vision).
- Discuss Early intervention and refer for enrollment in local program if not yet done.
- Application for Supplemental Security Income (SSI), depending on family income.
- Consider estate planning and custody arrangements; continue family support.

Childhood (2-12 years):

- Continue usual well-child care with immunizations. (Pneumococcal Vaccine at age 2).
- Audiologic evaluations yearly.
- Ophthalmologic evaluations yearly
- Thyroid (TSH & T4) yearly
- Nutrition guidance yearly
- Begin Dental Exams at 2 years and continue every 6 months thereafter.
- Celiac Screening at age 2. (IgA antiendomysium antibodies and total IgA).
- Parent Support
- Developmental and Educational Services Continue.
- Neck X-rays & Neurological Exam (Cervical Spine x-rays: flexion, neutral and extension, between 3-5 years of age. Repeat as needed for Special Olympics participation).
- Neurological Exam yearly.

Adolescent (13 to 18 years):

- Usual Preventative Care.
- Audiological Evaluation yearly.
- Ophthalmologic Evaluation yearly.
- Thyroid (TSH & T4) yearly.
- Nutrition guidance annually.
- Dental Exam yearly.
- Parent Support
- Developmental and Educational Services.
- Neck X-rays and neurological exam. X-rays @ 3-5 years. Repeat for sport participation.
- Neurological Exams Yearly.

Age 16 - adulthood:

- Assess Contraceptive need if sexually active.

Adulthood (more than 18 years)

- TSH and T4 Thyroid Function Test (annually)
- Auditory Testing (every 2 years)
- Cervical Spine X-rays (as needed for sports)
- Ophthalmologic examination, looking especially for keratoconus and cataracts. (Every 2 years)
- Clinical Evaluation of the heart to rule out mitral/aortic valve problems.
Echocardiogram-ECHO (as indicated)
- Reinforce need for subacute bacterial endocarditis prophylaxis (SBE) in susceptible adults with cardiac disease.

- General physical and neurological exam. Check for atlanto-axial dislocation.
- Routine adult health care.
- Clinical evaluation for sleep apnea.
- Low calorie, high fiber diet. Regular exercise. monitor for obesity.
- Clinical Evaluation of functional abilities. (consider accelerated aging); monitor loss of independent living skills.
- Neurological referral for early symptoms of dementia: decline in function, memory loss, ataxia, seizures, and incontinence of urine and/or stool.
- Monitor for behavioral or emotional changes and/or mental health problems.
- Psych referral if needed.
- Continue speech and language therapy (as indicated).

For Females:

- Baseline Mammography (at 40 years; follow up every other year until 50 years, then annually).
- Pap smear and pelvic exam (every 1-3 years. after first intercourse. If not sexually active and manual exam is not possible, screen pelvic ultrasound (every 2-3yrs.)

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