Birth to 1 month

down syndrome care hub

Assessments & Health Visits for People Living with Down Syndrome

Guided Checklist

◯ Complete physical examination

Findings include: hypotonia, small brachycephalic head, epicanthal folds, flat nasal bridge, upward-slanting palpebral fissures, Brushfield spots, small mouth, small ears, excessive skin at nape of neck, single transverse palmar crease, short 5th finger with clinodactyly and wide spacing, and deep plantar groove between 1st and 2nd toes

◯ Genetic testing

Send blood sample for chromosome evaluation within the first 24-48 hours of life (use FISH)

◯ Counseling

- Diagnoses with parents; focus on strengths and positive experiences of other families living with DS
- Risks of DS recurrence; refer to genetic counseling as needed
- Potential clinical manifestations associated with DS and how to cope with long-term disabilities

◯ Feeding

- Refer for radiographic swallowing assessment for infants with marked hypotonia, slow feeding or choking, recurrent pneumonia or respiratory symptoms
- Unsuccessful breastfeeding; create regular feeding schedule and early supplementation until nursing pattern established

◯ Heart

Perform echocardiogram, refer to pediatric cardiologist to review results

◯ Hearing and vision

- Determine cataracts at birth, look for red eye reflex; prompt evaluation and treatment by ophthalmologist if detected
- Congenital hearing loss assessed by objective testing; brainstem auditory evoked response or otoacoustic emission at birth
- Follow-ups must be completed by 3 months

◯ Thyroid

- Risk for congenital hypothyroidism; obtain TSH concentration/screening
- Refer to pediatric endocrinologist to discuss management of condition if present

◯ Blood test

- Obtain CBC (complete blood cell count)
- Look for leukemoid reactions or transient myeloproliferative disorder, polycythemia common condition

◯ Stomach or bowel problems

- Perform clinical examination; assess for duodenal atresia or anorectal atresia/stenosis
- Constipation; evaluate for restricted diet, limited fluid intake, hypotonia, hypothyroidism, or gastrointestinal tract malformation (Hirschsprung disease)
- Assess for gastroesophageal reflux; refer for subspecialty intervention if severe

◯ Respiratory Issues and Infection

- Monitor or check for pneumonia/other infections, strider, wheezing or noisy breathing
- Give respiratory syncytial virus prophylaxis to those with severe and comorbid conditions
- Refer to pediatric pulmonologist, if severe, to assess for tracheal/airway abnormalities
- Discuss with parents the increased risk for respiratory tract infection in DS
- Developmental services
- If possible, advise parents and direct to Infant Development Program (IDP) services

◯ Resources

Provide information on DS specific support organizations or resources whenever possible