Medical Issues for Children with Down Syndrome

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Facts About Down Syndrome

• Chromosome condition that affects 1 in 700 to 1 in 1000 live births worldwide
• An extra copy of chromosome #21 is inherited
• Causes global developmental, speech, and mental delays
• Increased risk for many medical complications
Facts About Down Syndrome

• Occurs equally among all races, nationalities, and socio-economic backgrounds

• Genetic notation is 47, XY +21 or 47, XX +21

• Caused by “non-disjunction” (failure of chromosomes of egg or sperm to separate evenly before fertilization)
Facts About Down Syndrome

- Males and females equally affected
- Higher recurrence risk for females as they age, although 75% of babies with DS are born to women younger than 35 yrs
- No specific cause or prevention of DS is known
- Shortened life span (60’s), affected individuals typically need supervision to live independently
Common Physical Features

- Flat facial profile
- Upslanting palpebral fissures (outer corners of eyes)
- Epicanthal folds (inner corners of eyes)
- Flat nasal bridge
Common Physical Features

• Brushfield spots
Common Physical Features

- 5th finger clinodactyly (curving)
- Single transverse palmar creases
Common Physical Features

- Wide space between first and second toes
- Deep plantar creases (sole of foot)
Common Physical Features

• Brachycephaly (the back of the head is flattened)
Karyotype of trisomy 21
form of Down syndrome
(47, XX, +21)
Facts About Down Syndrome

- Recurrence risk is ~1% or age-related risk, whichever is higher (~1% at age 40)
- 95% babies born with DS have trisomy 21
- 2-3% have translocation DS (usually between chromosomes 14 and 21 or two chromosomes 21)
Facts About Down Syndrome

• If the child has translocation DS, parental chromosomes are suggested to see if one parent has a “balanced translocation”—if so, there is higher recurrence risk for DS in future pregnancies

• 2-3% have mosaic DS (some cells with 46 chromosomes, some cells with 47 chromosomes)
Diagnosis of Down Syndrome

- Suspected from selected physical features on exam
- Confirmed by chromosome analysis or FISH
- Genetic counseling recommended
Variability in Down Syndrome

- Risk for wide variety of medical concerns (cardiac disease, hypothyroidism, vision and hearing deficits, digestive issues, etc.)
- Variable levels of health
- Variable hypotonia, wide range of developmental delays
- Variable speech and cognitive abilities
Variability in Down Syndrome

• Variable personalities (calm/docile, stubborn/aggressive)
• May have autistic or obsessive-compulsive behaviors
• Growth tends toward short stature and weight gain: use DS growth charts to plot, available at www.growthcharts.com
Variability in Down Syndrome

- Future function impossible to predict (no "mild cases" of DS)
- Some children with DS are healthier than their typically developing siblings
- No two children with DS are exactly alike, even identical twins!
Concept of Preventive Management

- Down syndrome is common and has well-known medical complications which can be easily screened and effectively treated.
- Allows health care providers to supplement the initial diagnosis with a positive plan of action that can benefit patients and empower families (Wilson, 1999)
Complications in Down Syndrome

Medical complications will develop in 85% of children with Down Syndrome

Heart defects 50%
Hearing loss 75%
Obstructive Sleep Apnea 50-75%
Ear infections 50-70%
Eye disease 60%
Thyroid disease 15%
GI malformations 12%
C-spine instability 10%
Leukemia 1%
Hirschsprung disease 1%
Down Syndrome Preventive Medical Checklist

• Used by the PCP to help provide comprehensive care for children with DS
• Identifies various issues, screening, testing, referrals, etc. that are recommended at different ages
• Examples: Referral for echocardiogram at birth, yearly thyroid (T4/TSH) screening
Common health concerns in children with Down syndrome

Vision

- Congenital cataracts, strabismus (crossing), nystagmus (wiggling of pupils), lacrimal duct stenosis (tearing), refractive errors
- **Screening:** Annual pediatric ophthalmology consultation
Common health concerns in children with Down syndrome

Hearing/ENT

- Chronic ear infections, hearing loss, sinus infections, ear tubes are needed in about 1/3 of children with DS in our population
- Screening: ABR at birth and yearly until age 2-3, then audiology screening every 1-2 years, ENT consultation as needed
Common health concerns in children with Down syndrome

Respiratory

- Frequent respiratory infections, pneumonia, asthma/bronchitis, croup, aspiration

- **Screening:** Preventive medications, video swallow, referral to pulmonary or allergy specialist as needed
Common health concerns in children with Down syndrome

Cardiovascular

- ASD, VSD, AVSD, PDA, mitral valve prolapse are most common
- **Screening:** Echocardiogram/CXR and pediatric cardiology consultation at birth
Common health concerns in children with Down syndrome

Endocrine/metabolic

- Hypothyroidism, hyperthyroidism, obesity
- **Screening:** Annual free T4 and TSH, dietary counseling if overweight or underweight
Common health concerns in children with Down syndrome

Musculoskeletal

- Cervical spine instability, congenital hip dislocation, orthopedic concerns such as flat arches of feet
- Screening: Flexion/extension X-rays (Davis series) at age 3-5 years and as needed, counseling regarding sports activities, orthotics and/or orthopedic consultation if needed
Common health concerns in children with Down syndrome

Central/Peripheral Nervous System

• Obstructive sleep apnea, seizures, behavior problems due to depression, ADHD, autism, OCD, anxiety disorder, etc

• Screening: Sleep history/sleep study, behavioral and cognitive skills assessment, neurology or mental health consult as needed
Common health concerns in children with Down syndrome

Hematology/Oncology

• At birth, jaundice, high red cell counts, low platelet counts, and a newborn leukemia-like disorder are most often seen, usually resolve quickly

• **Screening:** Serial blood counts, bilirubin levels, phototherapy and transfusions as needed.
Common health concerns in children with Down syndrome

Gastrointestinal system

- Hirschsprung disease, imperforate anus, duodenal atresia, celiac disease, anal stenosis, chronic constipation
Common health concerns in children with Down syndrome

- **Screening:** Most gastrointestinal anomalies are present at birth. Antibody screening for celiac disease if chronic diarrhea or growth failure is present. Milk of Magnesia for routine constipation.
Common health concerns in children with Down syndrome

Dental

• Delayed dentition, poor spacing, random eruption of teeth and crowding are common

• **Screening:** initial pediatric dental evaluation at 12 mos age or when first teeth appear. Orthodontics referral in adolescence if needed.
Common health concerns in children with Down syndrome

**Immune system**
- More frequent infections, including ear, respiratory, gastrointestinal, skin, urinary tract infections
- Higher frequency of allergies
- **Screening:** routine immunizations plus RSV, Pneumococcal, seasonal flu, H1N1, rotovirus vaccines. Referral to allergist or other specialist as needed.
Common health concerns in children with Down syndrome

Adolescent issues

• Puberty comes at about the same time as other children; PMS and menstrual concerns for young women
• Weight gain, acne are common during this time
• Screening: Refer to adolescent counselor, clinic, dermatologist, and/or dietitian as needed. For young adult women: Pap smears if sexually active, yearly breast exam by clinician.
Role of the PCP in Preventive Management

• Address health issues of the newborn
• Present accurate information about Down syndrome to parents
• Refer parents for further genetic counseling
• Use a Down Syndrome Preventive Medical Checklist
• Use Down Syndrome Growth Charts
• Refer to specialists as appropriate throughout the life span
Importance of Multidisciplinary Approach to Care

• Down syndrome is a complex, multisystem disorder, best served by many disciplines and specialists.

• Coordination of care is key, typically by primary care practitioner.
Resources for Down syndrome:

- DS clinic at Children’s Medical Center Dallas is a weekly multidisciplinary specialty clinic that provides medical/nursing/psychosocial/therapy evaluations, genetic counseling, guidance, resources, and parent support. (call 214-456-2357)
DS Clinic Multidisciplinary Team Includes:

- Geneticist
- Genetic Counselor
- Social Worker
- Clinical Nurse Specialist
- Speech Therapist
- Physical Therapist
- Parent Representative from DS Guild of Dallas
- Dietitian by request
Referrals May be Made to the Following Specialists:

- Pediatric cardiology
- " ophthalmology (eye clinic)
- " audiology/ otolaryngology (ENT clinic)
- " dentistry
- " gastroenterology
- " endocrinology
- " dermatology
- " psychiatry
Resources for Down syndrome:

- Down syndrome preventive medical checklists/age specific health supervision guidelines, may be found at:
  www.ds-health.com
- DS specific growth charts at: www.growthcharts.com
Resources for Down syndrome:

The National Down Syndrome Congress
1370 Center Drive, Suite 102
Atlanta, Georgia 30338
Toll free: 1-800-232-NDSC (6372)
www.ndscccenter.org
Chris Burke: The potential of people with Down syndrome
“The ultimate goal of health care screening is for persons with Down syndrome to live enjoyable lives, free from undiagnosed and untreated chronic health concerns.”  (Van Allen, Fung, & Jurenka, Am J Med Genetics, 1999)
Thank you!

• Any questions?
• Please contact me at Joanna.Spahis@childrens.com or 214-456-2017.