Care for Children with Down Syndrome
Changing Prejudice and Building Acceptance of Children with Disabilities
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Why Discuss Down Syndrome

- Most common pattern of malformation in humans (1:660 newborns)
  - Large number of institutionalized children
- Avoiding institutionalization and deinstitutionalization of children with Down syndrome is an example of the system changes needed to promote family care for disabled children.
History of Down Syndrome

• 1866 - John Langdon Haydon Down
  • Superintendent of the Earlswood Asylum for idiots in Surrey, England

• 1959 - Extra chromosome present
  • Patricia Jacobs (England) Jerome Lejune (France)

• 1960’s - Diagnostic ability increased institutionalization of children with Down syndrome in the US
Evolution of the Contemporary Approach to Children with Down Syndrome

- 1958- National Defense Education Act (NDEA)
  - Training leadership personnel in teaching children with mental retardation.
- 1964- Stedman and Eichorn reported adverse effects of institutionalization on children with Down syndrome
Evolution of the Contemporary Approach to Children with Down Syndrome

1972- Pennsylvania Association for Retarded Children v. Commonwealth of Pennsylvania. Contested state law that specially allowed public schools to deny services to children “who have not attained a mental age of five years” at the time they would ordinarily enroll in first grade.

Every child has a right to a free and appropriate public education program, regardless of mental ability.
Evolution of the Contemporary Approach to Children with Down Syndrome

- 1973- Parents and professional caregivers formed the Down Syndrome Congress
- 1970’s- Decreasing institutionalization rates.
- 1981- *Down Syndrome Preventive Medical Checklist*
- 1982- Indiana “Baby Doe”
  - against Federal law to withhold food and medical treatment from handicapped babies
Summary

• Recognize the potential
• Correct diagnosis
• Opportunities must be available
  • Partnerships to improve child and family outcomes
    • Parents
    • Professionals
      • Medical
      • Educational
    • Government
• Change in public opinion
Postnatal Diagnosis

Physical Features in the Newborn

• Flat facial profile - 90%
• Poor Moro reflex - 84%
• Hypotonia - 80%
• Hyperflexibility of joints - 80%
• Upward-slanting palpebral fissures - 80%
• Excessive skin on back of neck - 80%
• Small, abnormally shaped ears - 60%
• Short 5th finger - 60%
• Single palmar crease - 45%
Chromosomal Confirmation

- Sporadic nonfamilial trisomy 21 (95%)
- Unbalanced translocation (3-5%)
  - 75% de novo
  - 25% familial
- Mosaicism (1-2%)
Medical Care for Children with Down Syndrome

Medical management, home environment, early intervention, education, and vocational training can significantly affect the level of functioning of children and adolescents with Down syndrome and facilitate their transition to adulthood.

American Academy of Pediatrics Committee on Genetics 2011

Improves Outcome
Improves Quality of Life for Child and Family
Medical Problems Common in Down Syndrome

- Hearing problems 75%
- Vision problems 60%
  - Cataracts 15%
  - Refractive errors 60%
- Obstructive sleep apnea 50-75%
- Otitis media 50-70%
- Congenital heart disease 40-50%
- Dental problems 23%
- Intestinal atresias 12%
- Thyroid disease 4-18%
- Seizures 1-13%
- Hematologic problems
  - Anemia 3%
  - Iron deficiency 10%
  - Transient myeloproliferative disorder 10%
  - Leukemia 1%
- Celiac disease 5%
- Atlantoaxial instability 1-2%
- Autism 1%
- Hirschsprung disease < 1%
Ongoing Assessment and Counseling Throughout Childhood

- Personal support available to the family
- Participation in a family-centered medical home
- Age-specific Down syndrome-related medical and developmental conditions
- Financial and medical support programs for which the child and family may be eligible
- Injury and abuse prevention with special consideration of developmental skills
- Nutrition and activity to maintain appropriate weight
Health Supervision
Newborn to 1 Month

- Heart defects 50%
- Feeding problems
- Cataracts
- Congenital hearing loss
- Gastrointestinal Problems
  - Atresia or stenosis
  - Constipation
  - Gastroesophageal reflux
- Respiratory Problems
  - Hypotonia
  - Stridor
- Hematologic Problems
  - Transient myeloproliferative disorder 10%
  - Polycythemia 18-64%
- Hypothyroidism 1%
Anticipatory Guidance
Newborn to 1 Month

- Increased susceptibility to respiratory infections
- Cervical spine positioning
- Efficacy of early intervention
- Information about parent support groups
- Strengths of the child and positive family experiences
- Review individual resources for support
- Counseling on how to inform siblings and other family members
- Genetic counseling
- Advice on alternative treatments
Health Supervision
1 Month to 1 year

- Monitor infants with heart defects for evidence of failure
- Eye examination
- Repeat hearing screen at 6 months
- Assessment of middle ear disease
- Check for iron deficiency
- Monitor weight and weight-for-height
- Appropriate immunizations
- Repeat thyroid tests at 6 and 12 months
- Monitor for signs of neurologic dysfunction (seizures)
- **Monitor for obstructive sleep apnea**
  - Heavy breathing
  - Snoring
  - Restless sleep
  - Frequent waking
  - Apneic pauses
  - Daytime sleepiness
  - Behavior problems
Anticipatory Guidance
1 Month to 1 Year

• Cervical spine positioning
• Efficacy of early intervention
• Information about parent support groups
• Assess emotional status of parents and intrafamilial relationships.
• Genetic counseling
• Advice on alternative treatments
Health Supervision
1-5 Years

- History and physical examination
- Annual hearing testing
- Attention to serous otitis media 50-70%
- Annual eye exam
  - 50% refractive error

- Look for myelopathic signs
  - Change in gait or use of arms or hands
  - Change in bowel or bladder function
  - Neck pain, stiff neck, head tilt or torticollis
  - Weakness
Atlantoaxial Instability

- Increased risk in children with Down syndrome
- Routine x-rays not recommended
- Certain contact sports should be avoided
- X-rays in symptomatic children
Health Supervision
1-5 Years

- Annual thyroid testing
- Evaluate for sleep apnea
- Cardiac follow-up

- Continue to monitor for neurologic dysfunction
- Annual hemoglobin
- Appropriate immunizations
Health Supervision
1-5 Years

• Review symptoms of celiac disease
  • Diarrhea
  • Constipation
  • Slow growth
  • Unexplained failure to thrive
  • Anemia
  • Abdominal pain or bloating
  • Refractory developmental or behavioral problems

• For symptomatic children
  • Tissue transglutaminase immunoglobulin A level and quantitative IgA
Normal Duodenum

Celiac Disease
Anticipatory Guidance
1-5 Years

- Review and coordinate early intervention
- Review child’s behavioral and social progress
- Discuss delayed tooth eruption and hypodontia
- Encourage optimal diet and exercise
- Discuss complementary and alternative treatments
- **Address issues of sex education**
  - Proper names for body parts
  - Respect personal space
  - Good touch - bad touch
  - Discuss increased risk of sexual exploitation
Health Supervision
5-13 Years

- History and physical
- Growth assessment
- Annual hearing test
- Eye exam every 2 years
- Annual thyroid test
- Annual hemoglobin
- Assess skin dryness
- Cardiology follow-up
- Assess symptoms of gluten sensitivity
- Monitor for neurologic dysfunction
  - Cervical spine
  - Sports participation
- Assess symptoms of sleep apnea
Anticipatory Guidance 5-13 Years

• Review child’s development and appropriateness of school placement and developmental intervention
• Monitor for behavior problems that can interfere with social function
  • Attention problems
  • Noncompliant behavior
  • Obsessive compulsive behaviors
  • Wandering off
• Encourage independence with hygiene and self-care
• Encourage parents to teach, model and respect privacy at home and in the community.
Anticipatory Guidance
5-13 Years

Discuss issues related to sexuality

- Provide guidance on healthy, normal and typical sexual development and behaviors
- Emphasize the need for understandable information
- Encourage opportunities for advancing comprehension of sexuality
- Provide routine gynecologic counseling and care
  - Discuss premenstrual behavioral problems
  - Discuss management of menses
Health Supervision
13-21 Years

- Annual hemoglobin
- Annual thyroid testing
- Annual hearing test
- Eye exam every 3 years
  - Keratoconus
- Hair and skin care
- Cardiology
  - Known abnormalities
  - Evolving problems
    - Mitral and aortic valvular disease
- Monitor for signs of neurologic dysfunction
  - Review importance of cervical spine positioning
    - Athletic participation
- Review symptoms of gluten sensitivity
- Review symptoms of obstructive sleep apnea
Anticipatory Guidance
13-21 Years

Discuss issues related to sexuality
• Provide guidance on healthy, normal and typical sexual development and behaviors
• Emphasize the need for understandable information
• Encourage opportunities for advancing comprehension of sexuality
• Discuss the need for contraception and prevention of sexually transmitted disease and the degree of supervision required
• Provide routine gynecologic care
  • Discuss premenstrual behavioral problems
  • Discuss management of menses
Parent Involvement

Chișițnău, Moldova
Kazan, Russia
Diagnosis

Prenatal Diagnosis

• First Trimester (detection rate 82-87%)
  • Maternal age, nuchal translucency ultrasonography, maternal b-hcg, pregnancy associated plasma protein A (PAPP-A)
• Second Trimester (detection rate 80%)
  • Quad screen
    • Maternal age, maternal serum hCG, unconjugated estriol, alpha-fetoprotein (AFP).
• Integrated Screening
  Detection rate 95%
• False Positive rate about 5%
Prenatal Screening for Down Syndrome

Gidiri, M., et al., BJOG 2007;114:458-461
Reece’s Rainbow
International Down Syndrome Orphan Ministry
Adoption Grants, Advocacy, Birth Family Support

![Graph showing data from 2006 to 2010.](chart)

![Image of a father and baby.](family)
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