Overview of Down Syndrome, Cerebral Palsy, and Epilepsy

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Objectives

- To understand the diagnosis and unique needs of Individuals with Down Syndrome, Epilepsy, and Cerebral Palsy.

- To learn how to support Individuals with Down Syndrome, Epilepsy, and Cerebral Palsy.

- To understand medical risks related to each diagnosis.
Unique Support Needs

DOWN SYNDROME
Down Syndrome Defined

- Down syndrome is a genetic variation of the 21st chromosome, which usually causes delay in physical, intellectual and language development.

- The exact cause of the chromosomal rearrangement and the primary prevention of Down syndrome are currently unknown.

- Down syndrome is one of the leading clinical causes of cognitive delay in the world – it is not related to race, nationality, religion or socio-economic status.
Incident

- Of all children born in this country annually, approximately 6,000 will have Down syndrome.

- There are about 350,000 to 400,000 people living in the United States with Down syndrome.

- According to a 2010 study reported by the CDC, the incidence of Down syndrome in the United States is estimated to be 1 in every 691 live births.
Risk Factors

- The likelihood of giving birth to a child with Down syndrome increases with maternal age; however, 80% of babies with Down syndrome are born to women under 35 years old, as women in that age group have the most babies.
Down syndrome is usually caused by an error in cell division called nondisjunction. It is not known why this occurs. However, it is known that the error occurs at conception and is not related to anything the mother did during pregnancy.
Diagnosis – Prenatal Screening

- “The Triple Screen” - These tests are usually done between 15 and 20 weeks of gestation.

- Sonograms (ultrasounds) - are usually performed in conjunction with other screenings.
Pre-natal Diagnostic Tests

- Amniocentesis is performed between 12 and 20 weeks gestation.
- Chorionic Villus Sampling (CVS) is conducted between 8 and 12 weeks.
- Percutaneous Umbilical Blood Sampling (PUBS) is performed after 20 weeks.
Genetic Screening

- Trisomy 21 (94%)
  - Trisomy 21 (nondisjunction) is caused by a faulty cell division that results in the baby having three #21 chromosomes instead of two. Prior to or at conception, a pair of #21 chromosomes in either the egg or the sperm fails to separate properly. The extra chromosome is replicated in every cell of the body. Ninety five percent of all people with Down syndrome have Trisomy 21.
Genetics – Con’t

- Mosaicism (2.4%)
  - Mosaicism occurs when nondisjunction of chromosome #21 takes place in one of the initial cell divisions after fertilization. When this happens, there is a mixture of two types of cells, some containing 46 chromosomes and some with 47. The cells with 47 chromosomes contain an extra 21st chromosome. Because of the “mosaic” pattern of the cells, the term mosaicism is used. This type of Down syndrome occurs in only one to two percent of all cases of Down syndrome.
Translocation (3.3%)

- Translocation accounts for only 3% to 4% of all cases. In translocation a part of chromosome #21 breaks off during cell division and attaches to another chromosome. The presence of an extra piece of the 21st chromosome causes the characteristics of Down syndrome. Unlike Trisomy 21, which is the result of random error in the early cell division, translocation may indicate that one of the parents is carrying chromosomal material that is arranged in an unusual manner.
Syndrome Presentation

- Low muscle tone
- Flat facial features, with a small nose
- Upward slant to the eyes
Presentation

- Speckling of iris – Brushfield’s spots
- Small skin folds on the inner corner of the eyes
- Small, abnormally shaped ears
Presentation

- Single deep crease across the center of the palm

- Hyperflexibility

- Enlarged tongue tends to stick out
Co-Morbid Conditions

- As many as 50% have heart anomalies
- 10-12% have gastrointestinal issues
- 60% Hearing problems
- 70% Vision problems
- 20% Celiac disease
- 45% Sleep apnea
- 50% Thyroid problems
- Increased risk of leukemia
- More susceptible-respiratory illness, pneumonia, aspiration, bronchiolitis syndromes, croup, and chronic lung disease
Treatment and Support

- Developmental Therapy
- Speech Therapy
- Physical Therapy
- Hippotherapy
- Music Therapy
Aging

- Increased hormonal problems than the general population
- Cataracts
- Conductive Hearing Loss
- Osteoarthritis
- 25% of adults over the age of 35 show signs of Alzheimer’s disease
Quality of Life

- Life expectancy for people with Down syndrome has increased dramatically in recent decades, from 25 in 1983 to 60 today
- Attend school, work, participate in decisions that affect them, and contribute to society in many ways.
Challenges

- Employment
- Social Isolation
- Health maintenance
Summary

Every child, every person, adds unique value to our world.
Welcome to Down Syndrome Indiana where we are dedicated to enhancing the lives of individuals with Down Syndrome through education, support and advocacy. We are glad you are here, and we hope that you become an active part of our family!
Resource – National Down Syndrome Congress
http://ndsccenter.org/
Unique Support Needs

EPILEPSY
"Epilepsy is the most common serious brain disorder worldwide. It has no age, racial, social class, national, or geographic boundaries."

Source: World Health Organization (WHO)
Definition

- Disorder of the brain’s electrical system
- Loss or change of consciousness
- Jerking and spasms
- Difficulty speaking
- 2 or more seizures
Epilepsy vs Seizure

- A seizure describes a brain dysfunction that occurs when the normal electrical impulses in the brain become disrupted.
  - approximately 70 percent of seizures occur without an identifiable cause
- Patients with epilepsy have seizures, but patients with a seizure do not necessarily have epilepsy
- Epilepsy is when an individual has two or more seizures
Symptoms

- Wide range of symptoms
- Staring
- Falling
- Fumbling with clothes
Absence Seizures

- Staring spells

- More common in children
Generalized Clonic Tonic Seizures

- Formally known as Grand Mal Seizures
- Stiffening of arms and legs
- Jerking movement
- May last up to 3 minutes
Partial Seizure

- Affects one side of brain
- Jerking
- Hallucinations
- Unaware
Causes

- Genetic factors
- Severe head injury
- Brain infection
- Stroke Oxygen deprivation

In 2/3 of people with epilepsy, a cause is never found
Diagnosis

- Medical exam and history
- EEG - Electroencephalogram
- Brain Scan
Health Risk

- Safety

- Suicide Risk

- SUDEP – Sudden Unexpected Death in Epilepsy
Home Safety
Home / About Epilepsy / Home Safety

Houses
The following are tips on how to make houses safer for people with epilepsy:

- Carpet the floors in your house or apartment with heavy pile and thick under padding.
- Pad sharp corners of tables and other furniture, look for rounded corners when you shop.
- Put guards around the fireplace or, preferably, close fireplace screens while a fire is burning.
- Don’t smoke or light fires when you’re by yourself.
- Don’t carry hot fireplace ashes or lighted candles through the house.
- Avoid space heaters that can tip over.
- Use curling irons or clothing irons with automatic shut off switches to prevent burns.
- Select chairs with arms to prevent falling.
- Make sure motor-driven equipment, such as a lawn mower, has a "dead man's" handle that will stop the machine if your hand releases normal pressure.

Bathrooms
- Hang bathroom doors so they open outwards instead of inwards (so that if someone falls against the door, it can still be opened).
- Put extra padding under carpeting in the bathroom.
- Hang an "Occupied" sign on the outside handle of the bathroom door, instead of locking it.
- Routinely check that the bathroom drain works properly before taking a bath or shower.
- If you fall frequently during seizures, consider using a shower or tub seat with a safety strap.
- Keep water levels in the tub low.
- Consider using a hand-held shower nozzle while seated in tub or shower.
- Set water temperature low so that you won’t be scalded if you lose consciousness while hot water is running.
- Avoid using electrical appliances, such as a hair dryer or electric razor, in the bathroom or near water.
Related Conditions

- ADHD
- Depression
- Mood Disorder
  - Most common - Major Chronic Depression
Treatment and Support

- Factors Influencing the Decision to Treat
  - Abnormal EEG
  - Previous Seizure
  - Other neurological impairment
  - Elderly
Treatment and Support

- Medications
  - Antiepileptic or anticonvulsant
  - Must maintain a therapeutic level
Treatment

○ Ketogenic Diet

○ High in fat, low in carbohydrates

○ Very strict diet
Treatment

- Vagus Nerve Stimulation – VNS
- “Pacemaker for the Brain”
- Surgical implant
Treatment

- Surgery used for people with partial seizures
- Treat underlying condition
First Aid for Seizures

- Time the seizure
- Clear the area of anything hard or sharp
- Loosen anything at the neck
- Turn the person onto his or her side
- Put something soft beneath the head
- Do NOT place anything in the mouth
- Call 911
Treatment for Status Seizures

- Status Epilepticus
- Hospital treatment
- Medication
- Oxygen
Seizure “Predicting” Dogs

- May be able to sense a seizure
- May keep individual safe
Epilepsy Research

Goals:

1. Increase the number of people who can fully control their seizures.
2. Reduce effects of treatment
Living with Epilepsy

- Challenges
  - Relationships
  - School
  - Employment
  - Driver license
  - Leisure activities
Epilepsy is a medical condition that produces seizures affecting a variety of mental and physical functions. It's also called a seizure disorder. When a person has two or more unprovoked seizures, they are considered to have epilepsy.

A seizure happens when a brief, strong surge of electrical activity affects part or all of the brain. One in 10 adults will have a seizure sometime during their life.

Watch an introductory video series on epilepsy and seizures on epilepsy.com
Unique Support Needs

CEREBRAL PALSY
Definition

- CP is a non-progressive disorder of motor control and function that is caused by damage to a child’s brain early in the course of development.
Cerebral Palsy

- Collection of motor disorders
- Not just one disorder
- Occurs due to brain injury early in life.
- NOT inherited
Cerebral Palsy

- Impaired motor control
- Abnormal muscle tone, posture, movements, balance and coordination
- Decreased function
- Variable severity
- Is permanent
3 major findings

- Decreased control of body movement or posture
- Static brain injury
- Occurrence of the brain injury either before birth, around the time of birth, or in the 1st few years of life
Cerebral Palsy

- Umbrella term
- Encompasses many different motor disorders.
- Many different causes.
- Wide variety of associated deficits.
Non-progressive Brain Injury

- But...Appearance of deficits may change as the child grows.
How Common in CP?

- Most common motor disability of childhood
- 1.5 to 4 cases per 1000 live births worldwide
- 3.3 per 1000 cases among 8 year olds
- CP, on average, occurs 1.2 times more frequently among boys than girls
- 8000 new cases per year in U.S.
What causes CP?

- 85 to 90% of cases are present at birth, or due to congenital causes.
Congenital Causes

- Neurologic damage due to fetal infection, poor oxygenation, prematurity, stroke
- Circulation deficits due to heart defects, blood clotting disorders
- Malformation of the brain
Perinatal Causes (around the time of birth)

- Premature infant with complications
- Stroke
- Meningitis/sepsis
- Severe, untreated jaundice
- Hypoxic-ischemic encephalopathy with acidosis
Postnatal Causes

- Infections/meningitis (viral or bacterial)
- Shaken baby syndrome
- Traumatic brain injury
- Stroke
Risk Factors

- Prematurity
- Multiple pregnancy
- Placental abruption
- Intrauterine infection
- Birth trauma causing anoxia (no oxygen)
- Maternal epilepsy
- Maternal hyper/hypo thyroid
- Chorioamnionitis
- Severe toxemia of pregnancy
Periventricular Leukomalacia

- Peri = around
- Ventricular = brain ventricle
- Leuko = white
- Malacia = scarring
Classification of CP

Classification is based on muscle tone and body movement patterns:

- **Spastic** – most common
- **Dyskinetic** – dystonic, athetoid
- **Ataxic** – least common
- **Hypotonic** – initially most worrisome
- **Mixed** – spastic / dystonic; spastic / athetoid; spasticity with hypotonia
Spastic CP

- Velocity dependent hypertonia
- Increased muscle tone in response to quick passive muscle stretches.
Dyskinetic

- Involuntary sustained or intermittent muscle contractions with twisting and repetitive, abnormal postures, or both.
Ataxic

- Jerky, unsteady, poorly coordinated movements
Hypotonic

- Floppy; low muscle tone, difficulty holding up trunk and head against gravity
Classification by Affected Body Parts

- Diplegic/Diparetic
  - Affects both legs much more than arms
Classification by Affected Body Parts

- **Quadriplegic/Quadriparetic**
  - Affects entire body/arms and legs
  - Spasticity and muscle control may be asymmetrical in arms and legs.
Signs and Symptoms - Infant

- Floppy at birth
- Exaggerated infant reflexes
- Difficulty sucking
- Motor skill delay
- Early hand preference
- Asymmetric arm or leg movement
- Other delayed milestones
Signs and Symptoms - Toddler

- Motor skill delay
- Abnormal mobility patterns – persistent army crawling, bunny hopping, bottom scooting
- Significant differences between motor and cognitive milestone
- Toe-walking; abnormal walking patterns
Diagnosis

- There is no one specific test
- Comprehensive history is reviewed: pregnancy, delivery, developmental milestones
- Family history; review risk factors
- Physical exam
- Neurological exam
- Eye exam
- Speech / language, occupational and physical therapy evaluations as needed
- Brain imaging
- Spinal cord imaging
Laboratory Work

- Not always routine
- CPK level in weak, low toned children.
- Urine studies for amino and organic acids, carnitine
- Blood levels for ammonia, lactate, pyruvate, acyl-carnitine and long chain fatty acids
- Chromosomal analysis and genetic studies
Differential Diagnosis
What else could it be?

- Muscular dystrophy
- Brain tumor
- Metabolic disorder
- Familial spastic paraparesis
- Neurodegenerative disease
## Associated Disorders

- Visual
- Sensory
- Hearing
- Learning
- Behavioral
- Psychological
- Oral motor
- Nutrition
- Respiratory
- Bowel/ bladder
- Epilepsy
Associated Disorder – Musculoskeletal

- **Bone health**: risk for decreased bone mineral density
- **Bone growth**: risk for joint deformities, length discrepancies of limbs
- **Foot**: bunions
- **Foot/ankle**: deformity
- **Knee**: flexion contractures, knock knee
- **Hip**: possible dislocation, flexion contractures
- **Spine**: kyphosis, lordosis, scoliosis
Windswept Posture

- Hip and knee flexion contractures with rotation of the thigh bones at the hip sockets; pelvic bone un-leveling
Associated Disorder – Musculoskeletal (cont)

- **Shoulders** - tight at sides of chest
- **Elbows** - elbow contractures and pronation deformities
- **Wrist** - flexion contractures with deviation towards the ulna or radial bone
- **Hands** - thumb in palm, finger deformities
Walking Patterns

- Scissored
- Crouched
Walking Patterns

- Toe Walking
- Stiff Knee
Walking Patterns

- In toeing / out toeing
Treatment - Multidisciplinary

- Pediatrician
- Neurologist
- Physiatrist
- Orthopedist
- Developmental pediatrician
- Gastroenterologist
- Pulmonologist
- Physical therapist
- Occupational therapist
- Speech and language pathologist
- Psychologist/neuropsychologist
- Orthotist
- Nutritonist
Common Habilitation Goal Areas

- Mobility skills/gross motor
- Fine motor skills
- Communication
- Socialization
- Oral motor skills
- ADL’s/ self care skills
- Behavioral regulation
- Spasticity management
- Muscle tone management
- Medical care
- Dental care
Common Goals

- Maintain or improve joint range-of-motion
- Manage spasticity and muscle tone decrease / increase as needed
- Muscle strengthening
- Improved physical endurance
- Maximize fine motor skills
- Improve balance and coordination
- Control drooling and improve swallowing
- Develop age-appropriate social and recreational skills
Traditional Therapies

- Occupational
- Physical
- Speech and oral-motor / swallowing
- Developmental
- Behavioral
- Recreational
- Music
Specific Techniques

- Stretching
- Strengthening
- Electrical stimulation
- Constraint-induced movement therapy
- Gait (walking) training - partial body weight supported on treadmill; pool therapy
- Robotics
- Hippotherapy
Robotics

- Robotic therapy offers an opportunity to treat secondary complications
  - High repetition
  - Consistency of movement
  - Increased intensity

- Robotics may actually create new neural pathways
Hippotherapy

- Physical therapy in which a patient sits or lies on the back of a horse for the therapeutic effect of the horse's movement
Bracing and Orthotics

- Upper extremities
- Lower extremities
- Spine
- Neuro-prosthesis
Adaptive Equipment

- Frequently prescribed to increase independence in mobility
- Used to facilitate safety and efficiency of care-giving.
Spasticity Management

- Stretching and Range of Motion
- Oral medications
- Chemical intervention
- Intrathecal baclofen
- Selective dorsal rhizotomy
Common Medications

**Oral Medications:**
- Baclofen
- Diazepam
- Clonidine
- Tizanidine
- Dantrolene

**Injected Medications:**
- Botox (BoNT-A)
- Myobloc (BoNT-B)
- Alcohol nerve and motor point blocks
Orthopedic Surgery

- Muscle tendon lengthening and transfers
- Spinal fusion
- Bony reconstruction of hip joints and ankles
- Femoral and tibial bone surgeries
Selective Dorsal Rhizotomy

- Neurosurgical technique for cutting sensory nerve rootlets in the lumbar and sacral spinal cord levels
- Decrease muscle tone
- May unmask underlying muscle weakness
- Permanent and not adjustable post-operatively
Intrathecal Baclofen Pump

- Surgical implant of pump and catheter (tubing) that delivers baclofen to the spinal fluid
- Dosage is adjustable
- Pump can be removed if ever desired
Lifetime Prognosis: Challenges with Growth

- Periods of rapid growth may bring increased contractures and periods of decreased coordination of motor skills.
- Increased caloric requirements with growth may present the need for caloric supplementation, by mouth or G-tube.
- Longer and heavier limbs are more difficult to move and control—there may be the appearance of decreasing motor function.
- However—sudden increases in spasticity deserve medical evaluation for constipation, UTI, pain, etc.
Quality of Life - QOL

- Perception of QOL is very subjective
- QOL in children with CP has been studied mostly through parental surveys
- Recent studies of direct child reports indicate that greater than 50% of children with CP perceive their psycho-social QOL as similar to non-disabled peers
- Children seem to incorporate early onset physical disabilities into the “sense of self”
Cerebral Palsy and Aging

- Approximately 500,000 people in the U.S. live with CP
- Health of adults with CP in generally “good”
- Functional status of adults with CP is not static and there is usually only modest decrease in function as there is for the general population
- Exercise can improve function
Cerebral Palsy and Aging

- Mortality: related to severity of impairments; those with most severe deficits rarely survive to 60 years.

- Those with mild / moderate impairments have survival rates close to the general population.
Health and Aging with CP

- Musculoskeletal: osteoarthritis, osteoporosis; hip, knee, and foot pain

- Spine: degenerative conditions; progression of scoliosis

- Peripheral nerve compression: carpal tunnel and ulnar nerve compression
Health and Aging with CP

- Respiratory issues: scoliosis and aspiration
- Swallowing deficits
- Intestinal obstruction
- Urinary tract infections
- Sexual dysfunction
Sexual Function and CP

- Adult women often receive limited health screens (PAP smears, etc.)
- Women require appropriate screens for use of oral and other contraceptives
- Women generally able to conceive and carry term pregnancies
- Adult males have normal function and fertility
- Adults require appropriate health screens and sex education/pregnancy prevention
Cerebral palsy is defined as a collection of motor disorders due to brain injury early in life of a baby or young child; it is a static condition but impacts development across the life span.
Summary

- The causes of CP are broad, but most causes are present at birth
- There are common subtypes base on the type of movement impairments and areas of body affected
- Management techniques include medical, surgical, therapeutic, orthotic, and adaptive equipment
Summary

- Associated medical issues can be extensive and highly variable.
- Aging issues commonly include pain and fatigue; but, exercise has been demonstrated to help maintain flexibility, strength, and function.
Resources

- [www.cerebralpalsy.org](http://www.cerebralpalsy.org)

- United Cerebral Palsy
  - [www.ucp.org](http://www.ucp.org)
Questions