RESOURCE INFORMATION ON COMMON DIAGNOSES OF CHILDREN SERVED IN ECI PROGRAMS

Note: The information following is gathered from a number of resources (listed at the end of the resource guide) and is a general overview only. Information is sometimes contradictory from one source to another and this chart is not intended to be used as the authoritative source regarding any diagnoses. New information and research emerges daily regarding these diagnoses and the EIS is encouraged to use available websites and libraries to stay abreast of developments specific to the children and families served.

Down syndrome
Down syndrome is a chromosomal defect that causes mental retardation and physical abnormalities.

Overview:
Down syndrome is a genetic disorder in which the person affected carries an extra chromosome usually caused by an error in cell division called non-disjunction trisomy 21. However, two other types of chromosomal abnormalities, mosaicism and translocation are also implicated in Down syndrome. Regardless of the type a person may have, all people with Down syndrome have an extra, critical portion of the number 21 chromosome present in all, or some, of their cells. Nondisjunction trisomy 21 is a faulty cell division occurring prior to or at conception, resulting in three number 21 chromosomes instead of two. Mosaicism occurs when nondisjunction of the 21st chromosome takes place in one of the initial cell divisions after fertilization, resulting in a mixture of two types of cells, some containing 46 chromosomes and some containing 47. Translocation occurs in only three or four percent of people with Down syndrome and occurs when part of the 21st chromosome breaks off during cell division and attaches to another chromosome.

Down syndrome affects about 1 in 600 birth; incidence in the community is lower because many children die in infancy with related disorders including congenital heart deformities. Incidence of Down syndrome increases with maternal age. Children with Down syndrome have varying degrees of disability; most have related health problems.

Common characteristics:
In Down syndrome, both physical and cognitive development is delayed. Infants tend to be quiet, rarely cry, and have floppy muscle tone (hypotonia) and loose ligaments. Typically, a child with Down syndrome has a small head, a flat broad face, slanting eyelids, and a depressed nasal bridge. The tongue is often large and prominent. The ears are small and low set. The hands are short and broad with a single crease across the palm. The fingers are short; the fifth finger often has two instead of three sections and curves inward. Many children with Down syndrome also have heart defects. About 90 percent of children with Down syndrome have hearing deficits and many are prone to recurrent ear infections. Vision impairments are common because of changes in the cornea and lenses.

Developmental implications:
Children with Down syndrome have increased risk of heart disease and leukemia. With these disorders life expectancy is reduced; otherwise most children with Down syndrome live into adulthood. The degree of mental retardation ranges from mild to severe. Early intervention can maximize the potential of children with Down syndrome.

Facial Clefts
Facial clefts are congenital defects of the skull and face.

Overview:
Cleft lip and cleft palate are the most common defect of the skull and face. Cleft lip is an incomplete joining of the upper lip, usually below the nose. Cleft palate is an abnormal passageway through the roof of the mouth into the airway of the nose.

Early surgical repair facilitates typical development. The goal of surgical repair is to close the lip and palate clefts, leaving as little scarring as possible. Frequently, several separate operations at different stages of
development are necessary to meet the goal. Surgical repair of the lip and nose is usually scheduled two to 10 weeks after birth. Subsequent surgery corrects the shape of the nose and lip for medical and cosmetic reasons. Repair of the palate is usually performed between 2 and 24 months of age. The goal is to fuse the segments of the palate to allow normal development of the upper jaw and to promote normal speech and breathing.

Common characteristics:
Facial clefts are disfiguring and impede feeding, hearing, dental, and language development. Speech development is delayed; fluency is often restricted. Many children with facial clefts have frequent middle ear infections. Clefts that involve the gums affect the growth and positioning of the teeth.

Developmental implications: Feeding difficulties begin at birth because the opening in the baby’s mouth or lips prevents normal sucking. Dental devices, including prosthetic palates, permit infants to get adequate nutrition while developing jaw muscles used in sucking. Even with the devices, feeding times may be lengthy. Repeated ear infections can lead to hearing loss; careful monitoring is essential. Oral surgery and orthodontia are often necessary to reposition misaligned teeth to attain speech fluency. Incomplete surgical repair of the soft palate allows air to pass through the nose during speech; therapy can effectively counter this difficulty.

Hydrocephalus
Excessive cerebrospinal fluid in the brain.

Overview:
Cerebrospinal fluid (CSF) usually circulates through the brain ventricles and around the brain and spinal canal. When circulation or absorption of the fluid is blocked, the accumulation of fluid puts pressure on the brain, forcing it against the skull and damaging or destroying tissue. Risks for developing hydrocephalus include congenital defects, tumors, intrauterine infection, central nervous system infection (encephalitis and meningitis), injury during the birth process, and trauma before or after birth.

Common characteristics:
Symptoms of hydrocephalus vary depending on the cause of the obstruction of fluid, the age at which the malfunction occurs, and the extent of damage to brain tissue. Early symptoms include enlarged head, bulging fontanelles, separated sutures, vomiting, and muscle spasticity. Later symptoms include decreased cognitive ability, developmental lags across all domains, restricted movement, uncontrolled eye movements, headaches, poor coordination, and difficulty feeding. Surgical intervention, including removing the obstruction or implanting a shunt, is usually successful in promoting the normal circulation of CSF.

Developmental implications:
Untreated hydrocephalus has a 50-60 percent death rate; survivors have varying degrees of cognitive, physical, and neurological disabilities. Early treatment enables most children to have a normal life span; one third will have normal cognitive function but some neurological difficulties may persist.

Seizures Disorders and Infantile Spasms
Seizures are sudden, brief attacks of altered consciousness, motor activity, sensory disturbance, or inappropriate behavior caused by abnormal electrical discharges in the brain.

Overview:
Children are commonly affected by two types of seizures: infantile spasms and febrile seizures. Infantile spasms are marked by sudden muscle tension (bent arms, straight legs, thrust torso and head), and usually last only a few minutes. They generally occur in children younger than 3 but frequently evolve into seizure disorders later in life. Many children with infantile spasms have associated intellectual impairment or neurodevelopmental delays that usually continue into adulthood. Infantile spasms are difficult to control with antiepileptic drugs.
Febrile seizures generally occur in children from 5 months to 5 years of age and result from elevated body temperature related to trauma or illness. Children who have febrile seizures are only slightly more likely to develop seizures later in life.

Seizure disorders are a frequent and sometimes serious neonatal problem that can occur with any disorder that affects the central nervous system. Infections, hypoglycemia, inborn errors of metabolism, and trauma are common causes of seizures. Prognosis is directly related to the cause. The longer seizure activity continues the more likely the infant is to have neurological impairment including cerebral palsy and mental retardation. Seizures can be generalized with all of the brain cells involved or partial when the dysfunction is limited to one part of the brain; responses to seizures are determined by the part of the brain affected. If the electrical discharge is limited to a tiny section of the brain the child may notice only an odd taste, vision, or smell; if it involves a large area the child may have a convulsion with jerking and spasms throughout the body. A seizure usually lasts for 2 to 5 minutes. Epilepsy is a recurring seizure disorder. Other seizures include those caused by fever, an imbalance of body fluids or chemicals, or withdrawal from alcohol or other drugs.

When seizures are caused by identifiable medical conditions (tumor, infection, or chemical imbalance, for example) that condition is treated first. Further medication may not be required. If there is no underlying cause, medication can frequently prevent or diminish seizure activity.

**Common characteristics:**
Symptoms of seizure disorders include involuntary movement of arms and legs, periods of confusion or inattentiveness, episodes of staring, periods of unresponsiveness, unconsciousness with incontinence, and unconsciousness followed by excessive fatigue. Some seizures are accompanied by distorted perceptions including smells and sounds.

**Developmental implications:**
Careful monitoring of antiseizure drugs is critical so that seizures can be prevented. Frequent and recurring seizures are associated with minor brain damage and related learning difficulties.

**Cerebral Palsy**
Cerebral palsy is a condition characterized by poor muscle control, spasticity, paralysis, and other neurological impairments.

**Overview:**
Cerebral palsy is the result of brain injury that occurs during pregnancy, during birth, after birth, or before the age of 5. About 15 percent of the cases of cerebral palsy are caused by trauma or poor oxygen supply during labor and delivery. Premature infants are particularly vulnerable. Severe illness and trauma including meningitis, sepsis, and severe dehydration can cause brain injury in young children that may result in cerebral palsy.

There are four main categories of cerebral palsy:

- Spastic, in which muscles are stiff and weak;
- Choreaathetoid, in which muscles spontaneously move slowly and without normal control;
- Ataxic, in which coordination is poor and movement shaky; and
- Mixed, in which two types (usually spastic and choreoathetoid) are combined.

**Common characteristics:**
Symptoms of cerebral palsy usually appear in the first year of life and include poor head control, feeding difficulties, and delays in motor development. Later symptoms can range from clumsiness, poor balance, and difficulty moving and controlling limbs to severe spasticity. Cerebral palsy is difficult to diagnose before a baby is one year old, especially if the baby was born prematurely. Common indicators of this neurological disability include the following:

- sucking difficulty with breast or bottle
- difficulty feeding
• lack of muscle tone
• awkward positioning and postures (for example, one arm stretched out, the other above the head with neck turned toward outstretched arm)
• startles excessively
• impaired speech and language development
• purposeless gestures and motor activity
• delay in reaching cognitive and physical developmental milestones
• body is excessively stiff or floppy
• recurrent seizures
• crossed eyes
• deafness
• crawls awkwardly, using only hands or relying on one arm and leg on the same side of the body

Developmental implications:
Cerebral palsy cannot be cured but medical and therapeutic intervention can minimize complications and support independence. In all forms of cerebral palsy expressive language may be difficult to understand because of difficulty controlling facial muscles. Most children with cerebral palsy have other disabilities including some degree of cognitive delay; some have severe mental retardation. However, about 40 percent of children with cerebral palsy have normal or near-normal intelligence. About 25 percent of children with cerebral palsy—most often those with the spastic type—have recurrent seizures.

Spina Bifida
Spina Bifida means cleft spine, an incomplete closure in the spinal column.

Overview:
Spina bifida is a congenital neural tube anomaly. It can take two forms. Spina bifida occulta is characterized by an opening in one or more vertebrae of the spinal column and is generally asymptomatic unless the underlying spinal cord is affected. More serious is Spina bifida cystica or myelomeningocele, in which a portion of the spinal cord protrudes through the spinal column.

Spina bifida results from the failure of the spine to close properly during the first month of pregnancy. In severe cases, the spinal cord protrudes through the back and may be covered by skin or a thin membrane. Surgery to close a newborn’s back is generally performed within 48 hours after birth to minimize the risk of infection and to preserve existing function in the spinal cord.

Common characteristics:
Spina bifida occulta affects about 40 percent of the population but because there are no symptoms, few are aware of the defect. The effects of myelomeningocele, however, can include muscle weakness, loss of sensation, or paralysis below the area of the spine affected, and loss of bowel and bladder control. Hydrocephalus is present in most children with myelomeningocele. A surgically positioned shunt can relieve the build-up of cerebrospinal fluid and the resultant brain damage, seizures, or blindness. Many children with myelomeningocele have diminished mobility skills and rely on crutches, walkers, braces, or wheelchairs.

Developmental implications:
Most children with spina bifida have normal or above normal intelligence. Repeated surgeries and other extensive medical care may limit the child’s exposure to activities and interactions and lead to social and cognitive delays. Some children with spina bifida and hydrocephalus may experience learning difficulties and need special interventions to help educators understand how best to accommodate their learning needs. Some children require catheterization to mitigate the diminished sensations and muscle weakness that interfere with bowel and bladder control.

Failure to Thrive
Failure to thrive is a condition in which a child does not grow as expected.
Overview:
Failure to thrive is determined when the growth failure is significant and not transient. The term usually applies to young children, especially those under 2 years of age. The child’s condition may have an underlying physical disorder or illness that affects the ability to take in, absorb, process, or retain food. If a child has trouble eating due to prematurity or a facial cleft, for example, nutrition may be inadequate. Other causes include most acute or chronic diseases like cancer, juvenile arthritis, cystic fibrosis, chronic liver disease, and inborn errors of metabolism. Infections (parasites, urinary tract infections, and tuberculosis, for example) place great demands on the body and force it to use nutrients rapidly, sometimes bringing about short- or long-term failure to thrive. Alternatively, psychological, social, or economic factors may play a role. Chronically apathetic and unresponsive caregivers impede the social interactions and emotional ties that influence a child’s growth and development.

Common characteristics:
Because cognitive, social, emotional, and physical growth and development are interrelated, children with this diagnosis have delays in all domains of development. Characteristics include:

- apathy
- irritability
- stunted physical size
- unresponsiveness
- cognitive delays
- delays in reaching developmental milestones like sitting up, walking, and talking.
- Additional characteristics are related to the underlying causations.

Developmental implications:
Most diagnoses of failure to thrive are made in infants and toddlers in the first few years of life, a crucial period of physical and mental development. After birth, a child’s brain grows as much in the first year as it will grow during the rest of the child’s life. Poor nutrition during this period can have permanent negative effects on a child’s mental development. There may be an underlying medical condition or disease that, if treated, will alleviate the problem. Untreated and undiagnosed failure to thrive can result in significant developmental delay. Even with treatment, in about one third of these children, mental development, especially verbal skills, remains below normal. About half the children continue to have social and emotional problems including eating and nutritional disorders.

Microcephaly
Microcephaly is a rare, neurological disorder in which the circumference of the head is smaller than the average for the age and gender of the child. It correlates with having a brain that is also small.

Overview:
Microcephaly may have genetic causes or be the result of exposure to toxic substances during fetal development. The disorder may stem from a variety of conditions that cause abnormal brain growth (exposure to radiation, infection, maternal diabetes, substance abuse), and is often associated with chromosomal disorders.

Common characteristics:
Children with microcephaly are born with a normal or reduced head size that fails to grow in proportion to the rest of the face. Characteristics include small head, large face, receding forehead, loose often wrinkled scalp, high pitched cry, delayed motor and speech functions, varying severity of mental retardation, seizures, and broad range of motor impairment from clumsiness to spastic quadriplegia.

Developmental implications:
Generally, life expectancy for children with microcephaly is low; prognosis for typical cognitive development is poor. The degree of associated abnormalities may dictate interventions that prevent or minimize impairments and maximize supports for functioning at home and in the community.
Inborn Errors of Metabolism (IEM)
Inborn errors of metabolism result from a group of rare genetic disorders in which the body is unable to metabolize foods normally.

Overview:
The buildup of the product that cannot be metabolized can cause an array of symptoms. The most frequently diagnosed IEMs in infants and toddlers relate to disorders of protein metabolism (Sickle-cell anemia and Phenylketonuria or PKU), disorders of carbohydrate metabolism (fructose intolerance, galactosemia), lysosomal storage disorders (mucopolysaccharidoses or MPS), and mitochondrial disorders. IEMs can affect single or multiple organ systems and result in manifestations ranging from acute, life-threatening deterioration over hours, to episodic, infrequent symptoms, and asymptomatic conditions with insidious degeneration over decades.

Common characteristics:
Symptoms of IEMs become apparent at varying ages. Typically, symptoms appear with a significant imbalance in metabolic function. The onset and severity may be exacerbated by environmental factors, such as diet and illness.

Disorders of carbohydrate or protein metabolism and disorders of energy production tend to present in the neonatal period or early infancy and tend to be unrelenting and rapidly progressive. Less severe variants of these diseases usually present later in infancy or childhood and tend to be episodic. IEM disorders manifested by subtle neurological or psychiatric features often go undiagnosed until adulthood.

Neonates with inborn errors that result in defects in energy production and utilization often have dysmorphic features; skeletal malformations; cardiopulmonary compromise; abnormalities in organ structure; and severe and generalized hypotonia.

Infants and toddlers often have dysmorphic or coarse features, skeletal abnormalities, abnormalities of the hair or skin, ataxia, hypotonia or hypertonia, and visual and auditory disturbances. Additional characteristics include recurrent episodes of vomiting, poor feeding habits, seizures, lethargy, failure to thrive, dilated cardiomyopathy, jaundice, liver dysfunction, developmental delay, and coma.

Developmental implications: IEM forms vary in age of onset, clinical severity, and mode of inheritance. Rapid diagnosis, restricted diet, and consistent medication are requirements for most IEM survivors.

Pervasive Developmental Disorder
Pervasive Developmental Disorder (PDD) is a class of neurological disorders, usually evident by the age of 3, that affect a child’s ability to communicate, understand language, play, and relate to others.

Overview:
Five disorders are identified under the category PDD: autistic disorders, Rett’s disorder, childhood disintegrative disorder, Asperger’s disorder, and pervasive developmental disorder, not otherwise specified (PDDNOS). Due to the similarity of behaviors associated with autism and PDD, use of the term pervasive developmental disorder has caused some confusion among parents and professionals. However, the treatment and educational needs are similar for both diagnoses.

The cause of PDD is not clear or conclusive. Current research seems to point to neurological damage and biochemical imbalances in the brain as influencing factors. These disorders are not caused by psycho-logical factors.

Related disorders:
Autism Spectrum Disorders
**Autism Spectrum Disorders**

Autism Spectrum Disorders (ASD) are neurological impairments, usually apparent before the age of 3 that affect a child’s ability to communicate, understand language, play, relate to other people, and engage appropriately with the objects, events, and activities in the environment.

**Overview:**

Autism, a class of Pervasive Developmental Disorder, is a brain disorder (generally thought to be caused by abnormalities in the brain’s structure or function) that typically affects aspects of a child’s language, cognitive, social, and emotional development. ASDs are four times more common in boys than girls. Children with autism vary widely in abilities, intelligence, and behaviors. Some have significant language delays and do not speak; others have limited language that frequently includes repeated phrases and vocalizations. Some children with autism appear to be closed off and shut down; others engage in repetitive behaviors and rigid patterns of thinking. While diverse in symptoms and deficits, children with autism share documented social, communication, motor, and sensory impairments that affect behavior in predictable ways.

Autistic spectrum disorder is one of the disabilities specifically defined in the Individuals with Disabilities Education Act (IDEA), the federal legislation under which children and youth with disabilities receive special education and related services.

**Several related disorders commonly accompany autism, including:**

- Tuberous sclerosis, a genetic condition that causes abnormal tissue growth in the brain. Although rare, about one-fourth of the children affected with tuberous sclerosis also have autism.
- Seizures, aberrant electrical discharges in the brain, occur in about one-third of the children with autism. Most seizures can be controlled with medication.
- Fragile X syndrome, named for a defective piece of the X-chromosome, affects about 10 percent of people with autism. Children with Fragile X syndrome are more likely to have mental retardation as well as unusual physical features not typical of autism.

**Common Characteristics:**

Children with ASDs commonly have impairments in social, language, emotional, cognitive, and motor skills. Mental retardation affects about 75 to 80 percent of people with autism.

**The following characteristics are common in children with ASD but not all children will exhibit all behaviors.**

- engages in repetitive activities and stereotyped movements
- avoids eye contact
- prefers being alone; solitary play usually repetitive
- resists environmental change or change in daily routines
- has unusual responses to sensory experiences
- resists being touched, hugged, or cuddled
- oblivious to voices and attempts at communication
- impaired expressive and receptive communication
- repeats sounds, phrases, and sentences (echolalia)
- difficulty relating to people, objects, and events
- uneven cognitive performance or cognitive delays resulting from communication and behavior dysfunction
- reverses normal use of pronouns; uses you instead of I or me when referring to self

**Developmental implications:**

Early diagnoses and appropriate educational programs impact the developmental outlook for children with autism and related disorders. Although the symptoms persist throughout life, typically a child’s prognosis is determined by the strength of language skills by the age of 7.

**Rett’s Disorder**

Rett’s Disorder is diagnosed primarily in females in whom development proceeds in an apparently typical fashion until about 18 months of age. At that time there is a noted change in behavior including regression or
loss of abilities (especially gross motor skills); loss of speech, reasoning, and social engagement; impaired expressive and receptive language; and severe psychomotor delay. Hand use becomes less functional, gestures are repetitive and meaningless and frequently include hand-wringing or hand-washing.

**Childhood Disintegrative Disorder**

Childhood Disintegrative Disorder is a clearly apparent regression in multiple areas of functioning (motor skills, bowel control, social behaviors, and language fluency) following at least 2 years of apparently typical development. This condition is extremely rare.

**Asperger’s Disorder**

Asperger’s Disorder is characterized by a lack of social skills, poor coordination and concentration, and restricted, repetitive, and stereotyped patterns of behavior and activities but normal intelligence and adequate language skills. While many children with autism have mental retardation, those with Asperger’s Disorder usually have average to above-average cognitive abilities.

**Pervasive Developmental Disorder Not Otherwise Specified (PDDNOS)**

Pervasive Developmental Disorder Not Otherwise Specified (PDDNOS) is a diagnosis used when a child has severe and pervasive impairment in the development of social, communication, and motor skills but does not meet the criteria for other PDD categories.

**Common characteristics:**

Most children with PDD show the following characteristics. Note, however, that subtle differences and variations are commonly present; there is no set pattern of symptoms.

- Avoids eye contact
- Unresponsive to human voice
- Indifferent to affection
- Resistant to change
- Ritualistic or compulsive behaviors
- Limited facial responsiveness
- Passive acceptance or refusal of physical contact
- Fails to bond with primary caregivers
- Impaired cognitive skills
- Unusual responses to sensory experiences
- Avoids interactions, especially with other children
- Limited or abnormal speech skills

**Developmental implications:**

Approximately 60-80 percent of children with PDD also have mental retardation. Levels of PDD vary, however, with some children learning to function quite well in society with very limited support and others gaining only minimal skills.

**Resources used in developing this chart include:**

American Medical Association, [www.ama-assn.org](http://www.ama-assn.org)


Centers for Disease Control, [www.cdc.gov](http://www.cdc.gov)

Division of Early Childhood of the Council for Exceptional Children, [www.dec-sped.org](http://www.dec-sped.org)

Family Village, [www.familyvillage.wisc.edu](http://www.familyvillage.wisc.edu)

Merck Manual, [www.merck.com](http://www.merck.com)

National Center for Education in Maternal and Child Health, [www.ncemch.org](http://www.ncemch.org)

National Down Syndrome Society, [www.ndss.org](http://www.ndss.org)