



Neurological and Genetic Disorders Medical Staff and Volunteer Training



Neurological and Genetic Disorders

Objectives – What's the target?

- Neurological Disorders
 - Review basic pathophysiology of the most common neurological disorders in children
 - Discuss common complications
 - Discuss treatment of neurological disorders in children
 - Discuss considerations for campers
- Genetic Disorders
 - Review basic pathophysiology of the most common genetic disorders in children
 - Discuss common complications
 - Discuss treatment of selected genetic disorders
 - Discuss considerations for campers



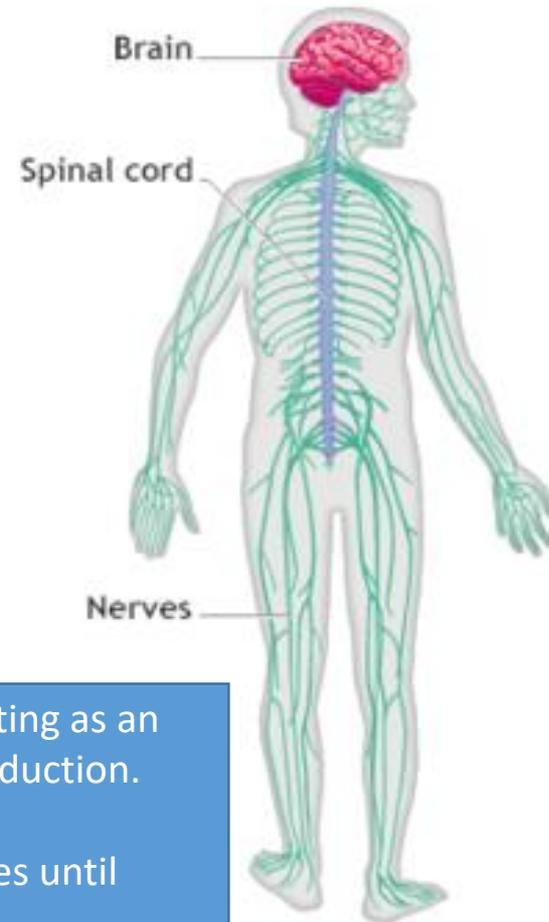
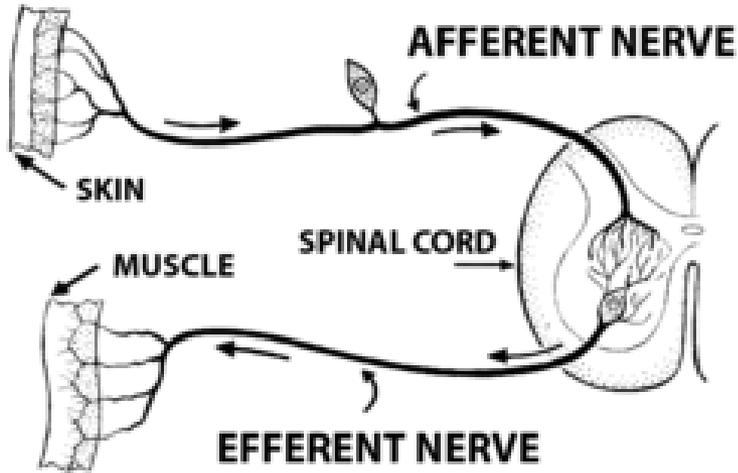
The target is camper safety! Better understanding of the diseases our campers have means better care!

Neurological Disorders



Central Nervous System

The central nervous system includes the brain, spinal cord, and nerves.



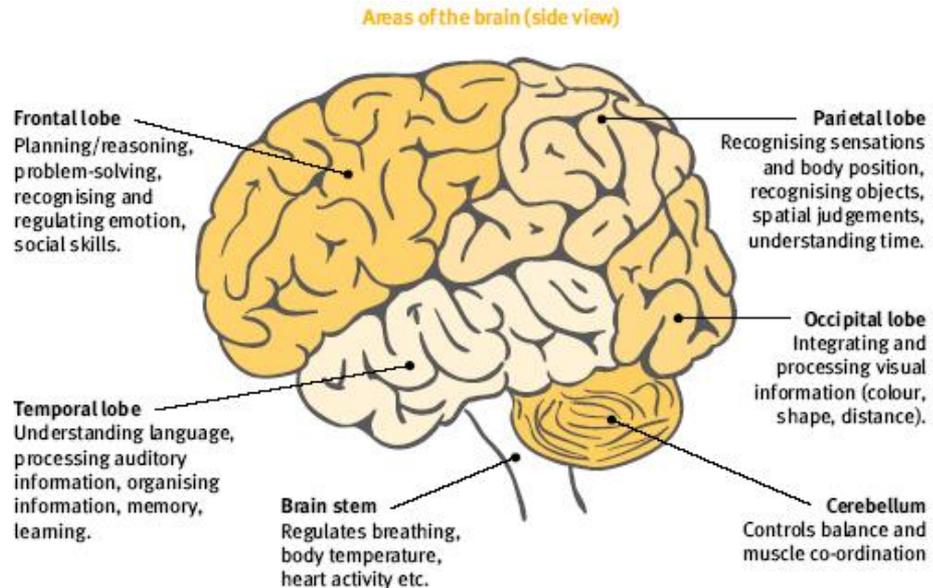
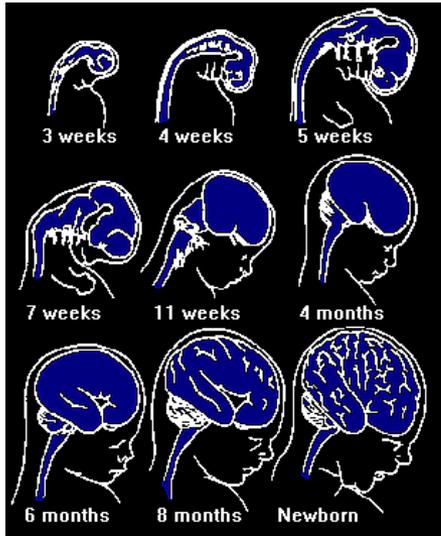
Nerves communicate sensory information to the CNS and motor information from the CNS to muscles.

Myelin is a fatty substance that surrounds some neurons, acting as an electrically insulating layer, and increasing the speed of conduction.

Myelination begins during fetal development and continues until adolescence.



Central Nervous System Development

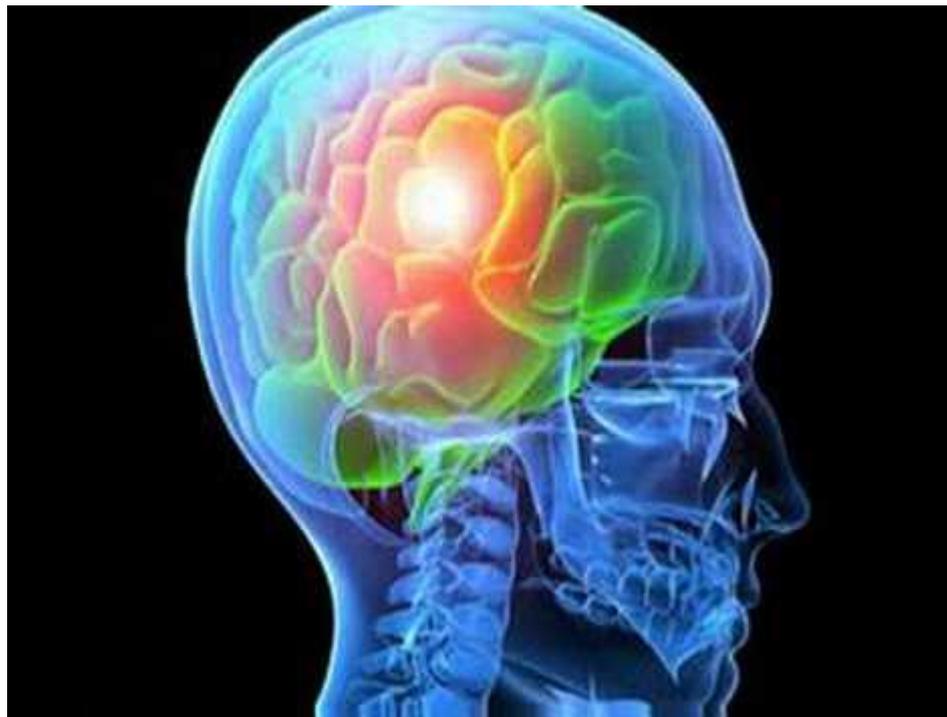


The cerebellum is affected in those with cerebral palsy.

Brain development is a process that occurs in stages over time, with 3 key factors.

- **Genetics** – Genes create templates for proteins essential to the development process.
- **Environment** – provides essential input that influences the developing system
- **Time** – Each step of the developmental process depends on the availability of the appropriate elements at the appropriate time. A flaw in the developmental process will affect the following stages.

Neurological Disorders



Epilepsy is one of the most common neurological disorders, and is also called a “seizure disorder.”

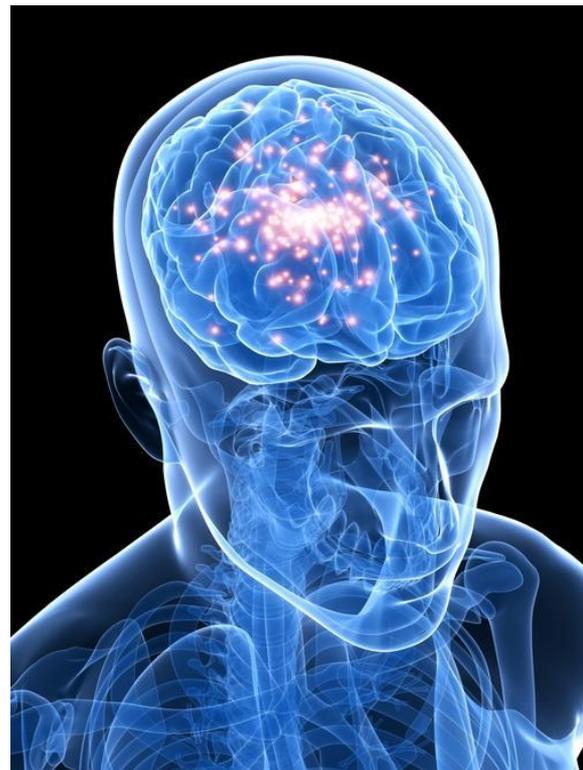
There are many types of seizures, and various types will look different and affect the person in different ways.

About 1% of children in the U.S. have a diagnosis of epilepsy, which was approximately 750,000 children ages 0-17 in 2013.



Neurological Disorders

Epilepsy is diagnosed by a specialist provider, based on the medical history, exam, and testing of the electrical activity in the brain.



Known causes of epilepsy include:

- Traumatic brain injury
- Stroke
- Brain tumor
- Severe brain infection
- Associated neurological or genetic disorder that affects the brain development and function

About 2/3 of epilepsy diagnoses do not have a specific known cause.

About 40% of epilepsy diagnoses are thought to have a genetic cause.

Neurological Disorders

Types of seizures

Focal Seizures (Partial Seizures)

- Affect one area of the brain
- Simple focal seizures – affect a small area of the brain, seizures may cause a strange sensation (i.e. smell) or twitching
- Complex focal seizures – this type of seizure can make a person confused, making it difficult to answer questions for a few minutes

Generalized Seizures

- Affect both sides of the brain
- Absent (petit mal) seizures – may look like a person is staring off for a few seconds, or rapid blinking
- Tonic-Clonic (grand mal) seizures – the person may fall, have shaking or jerking in parts of their body, or lose consciousness

A seizure can also begin in a specific area (focal) and become generalized.

Not every seizure is due to epilepsy. Seizures can happen for many reasons.



Neurological Disorders

People with epilepsy may have several seizures every day, or may have a seizure very rarely.

Words to describe seizures:

Tonic - Muscles become stiff

Atonic – Muscles become relaxed

Clonic- Parts of the body shaking or jerking

Myoclonic – Short jerking in parts of the body

3 phases of a seizure:

- **Aura** – symptoms that appear seconds before a seizure, vary based on the location of seizure activity but may include a smell/sound/vision change, sensation (i.e. dizziness, nausea, numbness), or feeling (i.e. fear, anxiety)
- **Ictal** – the visible seizure activity period
- **Post-ictal** – Immediately following a seizure, lasting about 5-30 minutes. A person may be somnolent, disoriented, have a headache, feel numbness or partial paralysis, or lose bowel/bladder control. This is different for each individual.

Neurological Disorders

There is no cure for epilepsy, but medications can help control and limit seizure activity.

Medication	Type	Common side effects
Levetiracetam (Keppra)	Anticonvulsant – treats partial onset, tonic-clonic, or myoclonic seizures	Fatigue, weakness, achiness, decreased appetite
Topiramate (Topamax)	Anticonvulsant – also used to treat migraines	Dizziness, drowsiness, mood change, flushing, taste change, nausea
Clonazepam (Klonopin)	Benzodiazepine – treats anxiety and certain types of seizures	Drowsiness, dizziness, difficulty with memory, difficulty with balance
Carbamazepine (Tegretol)	Anticonvulsant – decreases nerve impulses, also can treat nerve pain	Drowsiness, dizziness, difficulty with balance, nausea
Lamotrigine (Lamictal)	Anticonvulsant – treats seizure disorders	Dizziness, headache, insomnia, vision change, poor coordination, fatigue
Valproic acid (Depakene)	Anticonvulsant – treats various types of seizures, as well as bipolar disorder and migraines	GI distress, flu-like symptoms, insomnia, drowsiness, dizziness, weakness, tremors
Phenytoin (Dilantin)	Anticonvulsant – slows nerve impulses	GI distress, tremors, weakness, rash
Oxcarbazepine (Trileptal)	Anticonvulsant – decreases nerve impulses	Drowsiness, dizziness, tremors, vision change, GI distress, fever
Diazepam (Valium)	Benzodiazepine – can be used in adjuvant therapy to control seizures	Drowsiness, weakness, decreased coordination
Phenobarbital	Barbituate – slows nervous system activity and impulses	Drowsiness, dizziness, feeling restless



Neurological Disorders



Seizures lasting > 5 minutes are an emergency.

A person with a seizure disorder may have a medication to be given to stop the seizure in this event.

The most common are:

Diastat (diazepam) or AcuDial – given rectally

Midazolam (Versed) – given intranasally



Campers that have a PRN seizure medication may keep it nearby (with the counselor) if needed.



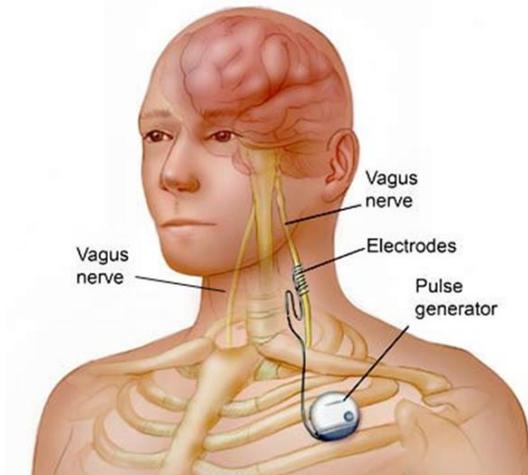
Putting diazepam up the rectum works faster than injecting it into a muscle.



Camp Life

- Campers may carry emergency anti-seizure medication with them
- Some campers will have multiple magnets for a vagal nerve stimulator (this is used to prevent or help stop a seizure once it has started).
- The magnet will need to be with the designated counselor for that camper at all times!!!
- Talk to the parents about the characteristics of their child's seizure
- Some campers may have special, very strict, diets used to control their seizures

Report any suspected seizure activity to your Unit Nurse or the Body Shop!



Seizure First Aid

STAY CALM!!!!

Don't forget to call for help! (x2000)

Crowd control

- **In the pool, keep camper's head above water**
- **On a boat, lay the camper down and bring the boat to shore**



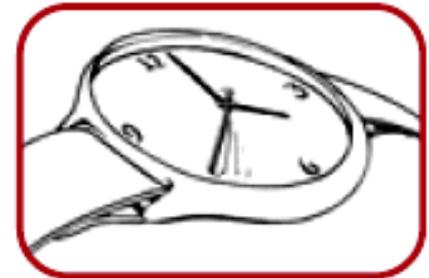
1. Cushion head, remove glasses.



2. Loosen tight clothing.



3. Turn on side and keep airway clear.



4. Note the time a seizure starts and the length of time it lasts.



5. Don't put anything in mouth.



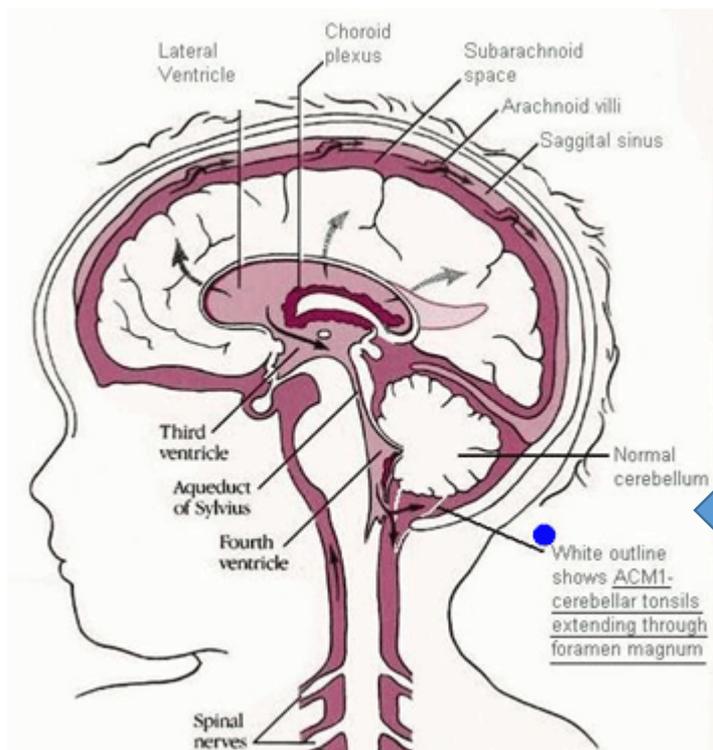
6. Don't hold down.

7. As seizure ends...offer help.

Neurological Disorders

Chiari malformations are structural abnormalities in the cerebellum.

The cerebellum is the portion of the brain responsible for coordination and balance.



Normally, the cerebellum and parts of the brain stem sit in an indented space of the skull above the foramen magnum (a funnel shaped opening to the spinal canal).

A **Chiari malformation** causes a portion of the cerebellum to sit below the foramen magnum.



Neurological Disorders

A Chiari malformation can be classified as primary or secondary:

Primary Chiari malformations are much more common, and caused by defects in the fetal development of the brain and spinal cord.

This could be due to **genetic defects** or lack of nutrients.

Secondary Chiari malformations can occur if too much CSF is drained from the spinal column, such as in a traumatic injury or infection.

Arnold Chiari malformations.

Chiari type I: elongation of cerebellar tonsils extending in vertebral canal

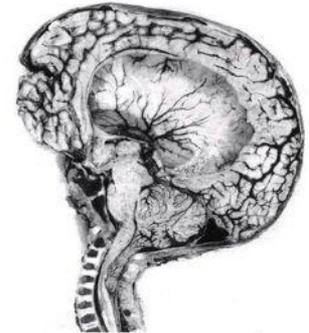
Chiari II: elongation of inferior vermis and brain stem with their displacement in cervical spinal canal with myelomeningocele and hydrocephalus; posterior fossa is shallow, torcular is low, sometimes craniolacuna, polymicrogyria

Chiari III: extremely rare, bony defect at occipito cervical level with herniation of cerebellum into encephalocele

type 1



type 2



Dr. G. Bhanu Prakash



Neurological Disorders

Chiari malformations are classified into types.

Chiari malformations are associated with spina bifida (myelomeningocele), tethered cord, and hydrocephalus, but also occur independently of these neurological diagnoses.

Type I

- Most common, least severe type
- Usually diagnosed in adolescent/adult years
- Portion of the cerebellum extends into the foramen magnum, does NOT involve the brain stem
- Only type that can be acquired

Type II

- “Classic”
- Portions of the brain stem and cerebellum extend into the foramen magnum
- Usually accompanied by myelomeningocele spina bifida

Type III

- Most severe type
- Cerebellum and brain stem protrude into the spinal canal
- Occasionally, a part of the fourth ventricle will also protrude
- This type causes severe neurological defects.



Neurological Disorders

Symptoms of Chiari malformation include:

- Neck pain
- Balance problems, poor coordination
 - Muscle weakness
 - Numbness in arms and legs
 - Dizziness
 - Vision change or blurred vision
- Headache (aggravated by straining, coughing)
- Hearing loss or ringing/buzzing in the ears
 - Difficulty swallowing

People with Type I malformation may not have symptoms, or have mild symptoms noted later in life. Infants with Chiari malformation may have difficulties feeding, growing, and with irritability and a stiff neck or back.



Neurological Disorders

Treatment for Chiari malformations aim to manage symptoms.

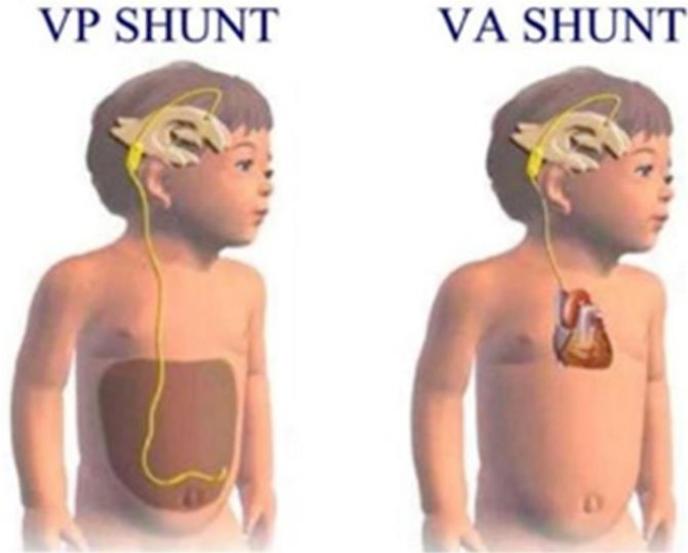
- Medication
 - May help with headaches and pain associated with Chiari malformation
- Surgery
 - Correct myelomeningocele, hydrocephalus, or other underlying associated conditions
 - Posterior fossa decompression – usually only performed on adults, relieves pressure on the spinal column
 - Spinal laminectomy – remove the lamina (curved outer bone of spinal column) to create space and relieve pressure on the spinal cord and nerves



Hydrocephalus

Hydrocephalus is a birth defect that impacts the brain, and some children with CP, who also have hydrocephalus, have had a shunt placed.

A shunt allows this extra fluid to drain from the ventricles into the peritoneal space (VP) or atrium (VA), reducing pressure on the brain.



In a camper with a shunt, we are concerned about:

- Change in level of consciousness (extreme agitation, behavior change, extreme/unusual fatigue)
- Headaches
- Vomiting

Information about a camper's shunt is found on the Camper Care Form in CampSite.

Neurological Disorders

Help the counselors keep these kids safe and healthy at camp by:

- Avoid triggers for seizures. (i.e. flashing lights)
- Encouraging campers to get enough rest, as lack of sleep can aggravate symptoms of neurological disorders.
- Staying hydrated! Always important.
- Understanding what a seizure looks like for each camper – each one is an individual.
- If needed, keep seizure emergency medications with the camper (using the carabiner system with the counselors).
- Straining or coughing may aggravate neck or headache pain in campers with Chiari malformations. Avoid activities that may strain campers.



Genetic Disorders



Genetic Disorders

Genetic disorders can be caused by several different types of defects that occur in the process of cellular multiplication. There may be too few or too many chromosomes, or the information may be in the wrong location on the chromosome.

Monosomy = too few chromosomes

- Turner syndrome (X)

Trisomy = too many chromosomes

- Klinefelter syndrome (XXY)
- Down syndrome (Trisomy 21)

Deletion = a portion of the chromosome is missing

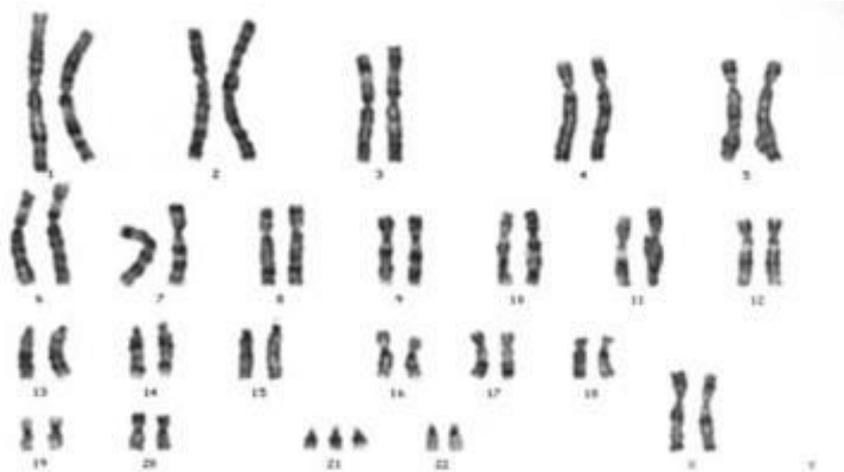
- Cri-du-chat syndrome (chromosome 5 deletion)
- Williams syndrome (chromosome 7 deletion)

Translocation = portions of chromosome rearranged

- If the translocation is balanced, the person may be healthy



Genetic Disorders



Genetic disorders are diseases caused by defects in DNA. They may be due to a single gene, chromosome abnormalities, or multiple factors within the DNA.

This photo shows the **most common genetic disorder, Down syndrome**, in which there are 3 copies of the 21st chromosome instead of 2. Down syndrome is also called trisomy 21.



Genetic Disorders – Down syndrome

Down syndrome affects **1 in every 691** babies born in the U.S, or **6,000 babies each year**. Approximately 400,000 Americans have Down syndrome.



Physical characteristics of individuals with Down syndrome often include:

- Decreased muscle tone
 - Small stature
 - Broad, flat nose
 - Small ears
 - Upward slanting eyes
- Single deep crease in center of palms

Genetic Disorders – Down syndrome

Many times, genetic disorders make a person more likely to have other medical problems.

For example, children with Down syndrome commonly also have:

- Hearing loss (up to 75% of people with DS are affected)
- Obstructive sleep apnea (50-75%)
- Ear infections (50-70%)
- Eye or vision problems (up to 60%)
- Congenital heart defects (50%)

Children with Down syndrome are at higher risk than children without DS for:

- Intestinal blockage at birth requiring surgery
- Hip dislocation
- Thyroid disease
- Anemia
- Leukemia in infancy or early childhood (and have higher or lower risk with different types of leukemia because of their DS)
- Hirschsprung disease

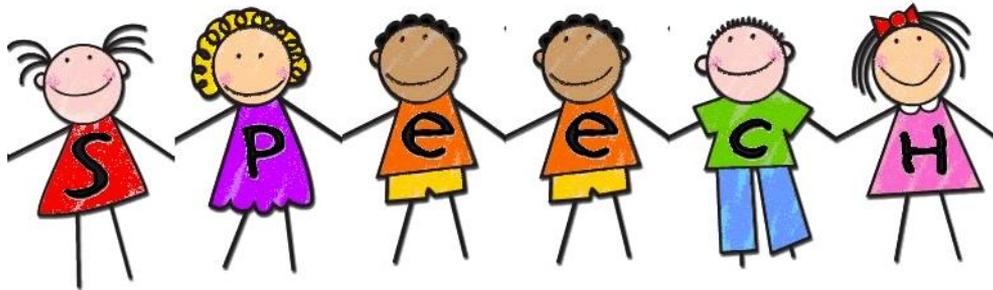


Genetic Disorders

Genetic disorders are chronic conditions. While associated medical conditions may be treated or repaired, the genetic disorder is lifelong.

Treatment of Down syndrome, and many genetic disorders, aims to optimize the child's physical and mental functioning.

Early and intensive intervention with Physical, Occupational, and Speech Therapy is recommended for children with Down syndrome.



For some children, learning sign language improves their ability to communicate.



Genetic Disorders – Mitochondrial Disease

Over 90% of mitochondrial diseases in children are caused by mutations in nuclear DNA, which contains genes involved in all functions of mitochondrial functioning.

Mitochondrial disease presents differently in each child, but can affect all systems.

- Neurologic
 - Most commonly affected (45% of children present with these signs)
 - Migraines, stroke-like episodes, spasticity, ptosis, ataxia, peripheral neuropathy, muscle weakness (proximal>distal, upper>lower extremities)
 - 20% show intellectual dysfunction
- GI
 - Anorexia, frequent emesis, abdominal pain, diarrhea, constipation (can be severe enough to cause pseudo-obstruction)
- Musculoskeletal
 - Significant muscle weakness
 - Typically normal or only slightly elevated CK levels
- Skin
 - Eczema, hypertrichosis, vitiligo
- Endocrine
 - Growth failure (20%)
 - Hypoparathyroidism, hypothyroidism, diabetes (insipidus or mellitus), adrenocorticotrophic hormone deficiency
- Sensory
 - Sensorineural deafness reports range from 7-26% of children
- Cardiac
 - Arrhythmias, cardiac murmurs, cardiomyopathy, sudden death
 - Anemia, thrombocytopenia

The mitochondria are the powerhouses for the body – mitochondrial disease is like a car running with minimal power.



Genetic Disorders

Treatment for genetic disorders differs for each disorder, but may include:

- Bone marrow transplantation
 - Certain genetic disorders can be cured by this process, such as inborn metabolism disorders and anemias
 - A person needs to have a donor match to be eligible
 - The BMT process can be life threatening and result in long-term side effects
- Medications
 - Treat the symptoms and complications of the genetic disorder, such as seizures, attention difficulties, malnutrition or malabsorption, pain
- Physical, Occupational, and Speech therapy
 - Focus on maximizing functioning and communication
- Assistance in school
 - Some children with genetic disorders benefit from assistance in school for medical, behavioral, and intellectual support



Genetic Disorders

Help the counselors keep these kids safe and healthy at camp by:

- Remembering each child is an individual! Genetic disorders affect each individual differently, even if they have the same diagnosis!
- Learning about the child's methods of communication. Some may use a communication device, sign language, or other non-verbal ways to communicate their needs.
- Ensuring the campers get enough rest! Take breaks as needed and keep a close eye on campers.
- Staying hydrated! This is particularly important for campers with mitochondrial disorders.
- Staying cool! This is also very important for campers with mitochondrial disorders.



Thank
you!



References

Koenig, M.K. (2008). Presentation and diagnosis of mitochondrial disease in children. *Pediatric Neurology*, 38(5), 305-313. doi: [10.1016/j.pediatrneurol.2007.12.001](https://doi.org/10.1016/j.pediatrneurol.2007.12.001)

National Down Syndrome Society. (n.d.) Down syndrome. Retrieved from <http://www.ndss.org/Down-Syndrome/>

National Institute of Neurological Disorders and Stroke. (2016). Chiari malformation fact sheet. Retrieved from http://www.ninds.nih.gov/disorders/chiari/detail_chiari.htm

University of Chicago Medicine. (2016). Epilepsy. Retrieved from <http://www.uchospitals.edu/specialties/neurosciences/epilepsy/index.html>

