Physical Variances and Craniofacial Abnormalities
Medical Staff and Volunteer Training

Objectives – What's the target?

- Physical Variances
  - Review basic pathophysiology of the most common diagnoses in children
  - Discuss common complications and treatments
- Craniofacial Abnormalities
  - Review basic pathophysiology of the most common diagnoses in children
  - Discuss common complications and treatments
- Skeletal Dysplasia
  - Review basic pathophysiology of skeletal dysplasia in children
  - Discuss common complications and treatments
- Discuss considerations for campers with these types of diagnoses

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

Physical Variances

Physical Variances

Arthrogryposis (arthrogryposis multiplex congenital) is characterized by congenital, non-progressive joint contractures in at least 2 areas of the body.

Arthrogryposis occurs in about 1 in 3,000 live births in the U.S.

These joint contractures are due to decreased fetal movement during development. Decreased fetal movement can be caused by:

- Fetal abnormalities in neurogenic, muscular, or connective tissue development
- Physical limitations to intrauterine growth
- Maternal disorders (i.e. infection, trauma, drug use)

Bamshad, M., Van Heest, A.E., and Pleasure, D. (2009). Arthrogryposis: a review and update. Journal of Bone and Joint Surgery, 91(4), 40-46. doi: 10.2106/JBJS.I.00281

Physical Variances

People with arthrogryposis usually have normal lifespan, and their abilities depend on the locations of the contractures.

The most severe complications occur when the spine is curved (scoliosis) and can damage the spinal cord or cause respiratory problems.

Complications of arthrogryposis include:

- Symmetric deformities, with increasing severity distally (the hands and feet are usually the most severely deformed)
- Joint rigidity
- Joint displacement (especially in hips and knees)
- Muscular atrophy
- Decreased deep tendon reflexes, although sensation is usally present

Associated conditions and malformations include:

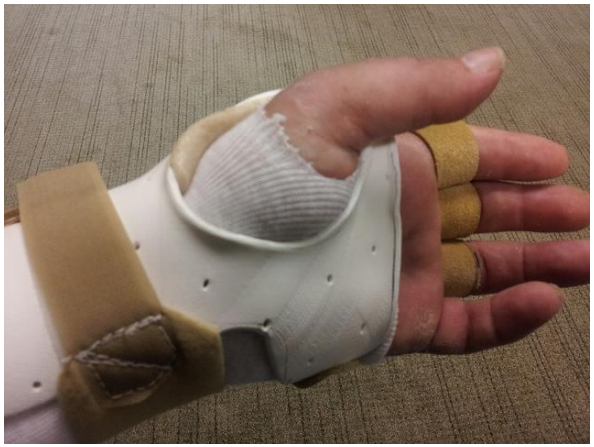
- Cardiac defects
- Structural abnormalities of the kidneys, bladder, and ureters
- Craniofacial malformations (may affect CNS development)
- Nervous system and sensory deficits
- Respiratory problems due to structural abnormalities of the airways and muscle development
- Muscle malformations or deficits in muscle function
- Connective tissue and skin changes

Bamshad, M., Van Heest, A.E., and Pleasure, D. (2009). Arthrogryposis: a review and update. Journal of Bone and Joint Surgery, 91(4), 40-46. doi: 10.2106/JBJS.I.00281

Physical Variances

There is no cure for arthrogryposis, and treatments focus on maximizing function of the lower body for ambulation and the upper body for independence.

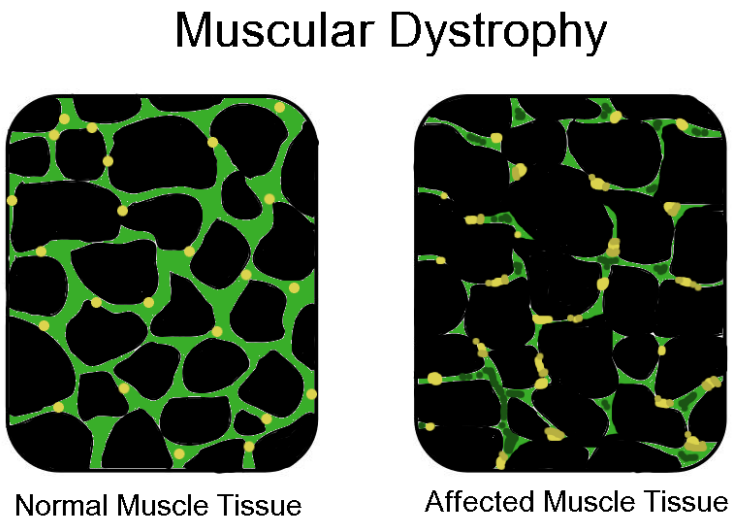
- Early gentle manipulation and stretching after birth can improve active and passive range of motion.
- Early physical therapy is important to improve strength and mobility.
- Physical therapy and splinting of joints to maintain mobility is preferred to casting or other prolonged immobility.
- Surgery, or multiple stages of surgeries, is often indicated to release the soft tissues around contracted joints.



Physical Variances

There are 9 types of muscular dystrophy, the most severe of which is Duchenne Muscular Dystropy (DMD).

DMD is a genetic disorder that results in an absence of the protein dystrophin, which helps keep muscle cells intact, and is characterized by progressive muscle weakness and degeneration.

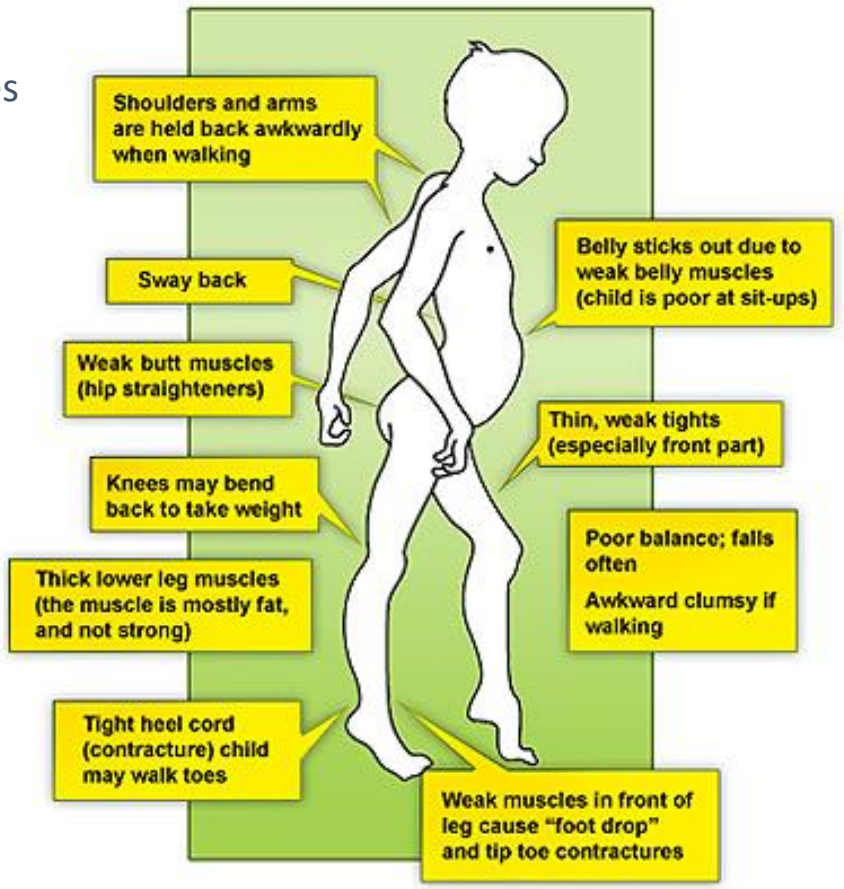


The genetic flaw that leads to DMD is an X-linked chromosome. The illness **primarily affects males.**

Females act as carriers of this gene, and can rarely be affected by the disease.

Physical Variances – Duchenne Muscular Dystrophy

- Symptoms (often clumsiness) appear between ages 3 and 5.
- The first muscles affected are around the core (hips, thighs, shoulders), and lead to the gait changes shown in this image.
- Skeletal muscles of the arms and legs are affected next.
- Children often begin transitioning to using a wheelchair during the school age years (7-12).
- By the teen years, the heart and respiratory muscles are affected.
- Life expectancy used to be the teen years, but survival into the early 30s is now more common.



Muscular Dystrophy Association. (2016). Duchenne muscular dystrophy. Retrieved from https://www.mda.org/disease/duchenne-muscular-dystrophy

Physical Variances

Major complications of Duchenne muscular dystrophy include:

- Heart function
  - In the teen years, cardiac muscle may weaken and lead to cardiomyopathy
  - Children with DMD should be followed closely by a cardiologist
  - There is some evidence that cardiac decline can be slowed by beginning treatment with ACE inhibitors and/or beta blockers at early signs of change.

- Respiratory function
  - Around age 10, the diaphragm and intercostal muscles begin to weaken, making inspiration and expiration less effective.
  - Decreased ability to cough can lead to respiratory infections and pneumonia
  - Headaches, poor concentration, poor sleep, and fatigue can indicate declining respiratory function

- Neurological function
  - DMD doesn't affect the nerves, so smooth muscle functions are maintained (including bowel and bladder control)
  - About 1/3 of children with DMD have a mild learning disability
  - Muscle deterioration is not painful, but muscle cramps or other pain may occur

Muscular Dystrophy Association. (2016). Duchenne muscular dystrophy. Retrieved from https://www.mda.org/disease/duchenne-muscular-dystrophy

Physical Variances

Treatment of DMD includes:

- Mobility and positioning aids
  - Use of splints, walkers, and wheelchairs is common to assist in maintaining mobility and independence as muscles deteriorate.
- Management of physical variances (i.e. scoliosis, contractures)
  - Surgery, AFOs, and other supports may be indicated
- Physical therapy
  - Some exercise is helpful for strengthening muscles, but too much may cause damage. PT can help maintain mobility and independence.
- Medications
  - Prednisone, or other steroids, may slow the deterioration process of DMD
- Respiratory support
  - Initially, non-invasive support (i.e. BiPAP, CPAP) while sleeping can improve respiratory status.
  - As the disease progresses, some people require mechanical ventilator support, often through a tracheostomy.

Muscular Dystrophy Association. (2016). Duchenne muscular dystrophy. Retrieved from https://www.mda.org/disease/duchenne-muscular-dystrophy

Cerebral Palsy

What is Cerebral Palsy?

Cerebral palsy (CP) describes a group of neurological disorders that affect muscle coordination and movement. CP is caused by a malformation or damage to the brain during development. CP is described as either congenital or acquired, depending on the timing of the malformation or brain damage.

Congenital CP (before or during birth)	Acquired CP (after 28 days of life)
85-90% of cases (<10% from lack of oxygen during birth)	Much smaller percentage, 10-15% of cases
Causes: Unknown specific cause	Causes: infection (encephalitis, meningitis), injury (abuse, MVA), obstruction of blood flow to brain (i.e. stroke, hemorrhage, or vascular problem)
Risk factors: low birth weight, maternal infection during pregnancy, multiple births, assisted reproductive technology pregnancy, jaundice, problems during the delivery	Risk factors: infancy, low birth weight

Cerebral Palsy

- CP is the most common motor disability in children
- About 1 in 323 children are diagnosed with CP
- CP is more common in boys than in girls.
- In 2008 CDC data, CP was more common in black children than white, with similar occurrence in Hispanic and white children
- Over half of the children with CP had a co-occurring illness
  - 41% had co-occurring epilepsy
  - 6.9% had co-occurring autism spectrum disorder
- A 2006 study of children with CP in Atlanta reported that over 40% also had an intellectual disability, and nearly 1 in 4 children with CP also had both epilepsy and an intellectual disability

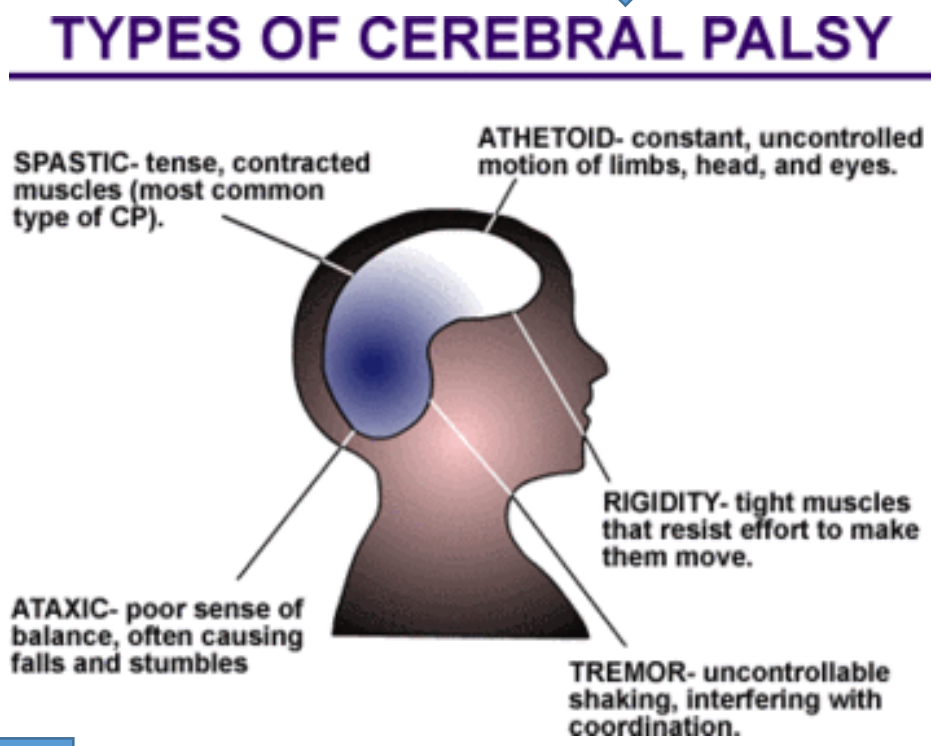
Centers for Disease Control. (2015). Data and statistics for cerebral palsy. Retrieved from http://www.cdc.gov/ncbddd/cp/data.html

Types of Cerebral Palsy

People with dyskinetic (includes athetoid) CP may have difficulties swallowing, sucking, talking, and walking. Their muscle tone may vary between too tight and too loose frequently.

Spastic CP affects about 80% of all people with CP.

Tight, contracted muscles may affect one side more than the other (hemiplegia), lower body more than upper (diplegia), or all four limbs (quadriplegia).



A person can have more than one type of CP. The most common mixed CP is spastic-dyskinetic.

Centers for Disease Control. (2015). Facts about cerebral palsy. Retrieved from http://www.cdc.gov/ncbddd/cp/facts.html

Cerebral Palsy Associated Complications

Intellectual disability and autism-spectrum disorder are conditions that are often associated in children with cerebral palsy.

There is an incredibly wide range of functioning levels and abilities among this group. NEVER assume a child's ability.

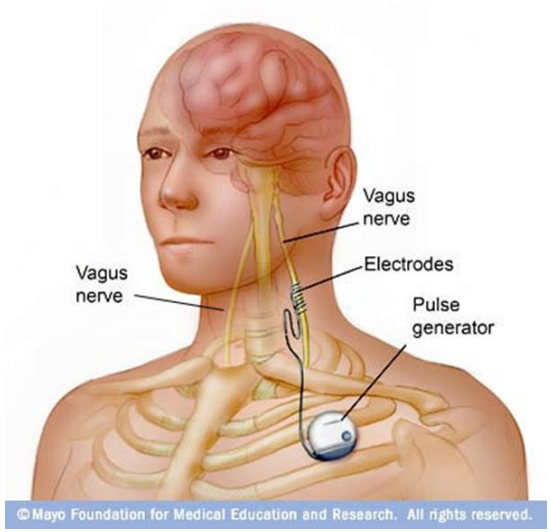
The Victory Junction Camper Care Form will list any co-morbidities.

Counselors are trained to communicate with these campers and manage behaviors, along with the full time program staff.

As a medical volunteer, you can help by supporting and understanding.

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

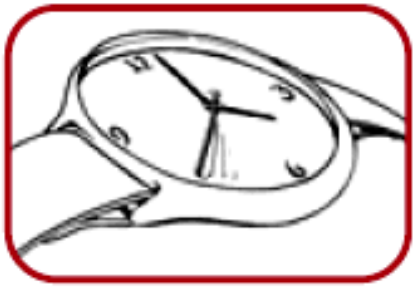
Crowd control

- **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.

Cerebral Palsy and Mobility Aids

Campers with CP may use a variety of mobility aids, including:

- Wheelchair (power or manual)
- Ankle-foot orthotics (AFOs)
- Crutches
- Walker
- Gait trainer
- Braces

Cerebral Palsy and Mobility at Victory Junction

Cabin counselors and volunteers are responsible for helping campers as needed with their ADL's, including use of AFOs and braces.

As a medical volunteer, you can help by:

- Ensuring the cabin counselors and volunteers are comfortable with proper use of these devices
- Inform the cabin counselors and volunteers about any additional instructions (i.e. a camper that must wear AFOs when ambulating, but not if using a wheelchair, or a camper that wears wrist braces on/off every 2 hours)
- Ensure the camper's skin integrity is checked with daily care, and that counselors notify the nurse if they notice any breakdown

Mobility at Victory Junction

Campers may use a wheelchair for mobility.

Wheelchair safety at camp:

- Every should wear closed-toe shoes.
- Brakes on!
- Don’t encourage racing.

A camper’s wheelchair is an extension of him/herself.

Wheelchair etiquette at camp:

- Allow independence, do not push a camper’s chair without asking.
- Avoid leaning on a camper’s wheelchair.
- Get on the camper’s level.

As a medical volunteer, you should help reinforce this etiquette with counselors and volunteers.

Cerebral Palsy at Victory Junction

What we want you to know at camp:

- Some campers will have trouble eating
- Some campers may have difficulty communicating
- Some campers may need help with toileting and changing
- Do not assume these campers have intellectual disabilities.

Cerebral Palsy at Victory Junction

How can you help the campers?

- Check their skin frequently for pressure sores or breakdown
  - Each camper will be checked every night for pressure sores
    - Check all extremities and trunk

- Report any new or unusual seizures to the medical team
  - Some campers may have several seizures a day at their baseline and that's ok!

- Listen to your campers and note any subtle changes!

- A camper said it best, "Don't dis my ability!"

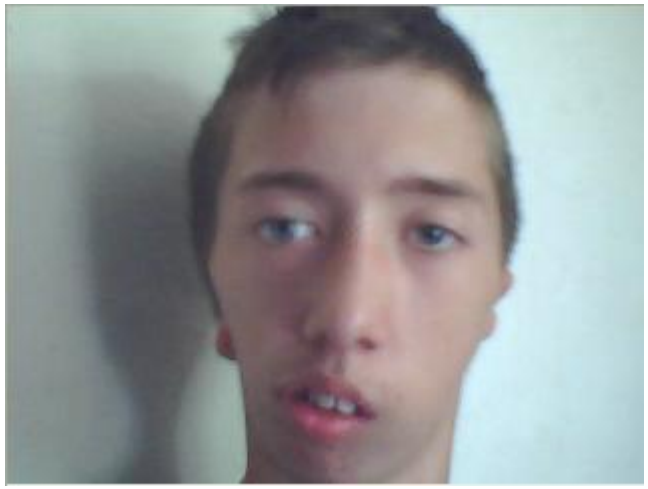
Other diagnoses at camp:

- Spinal muscular atrophy
- Spinal cord injury
- S/p amputations
- Severe scoliosis

Craniofacial Anomalies

Craniofacial Anomalies

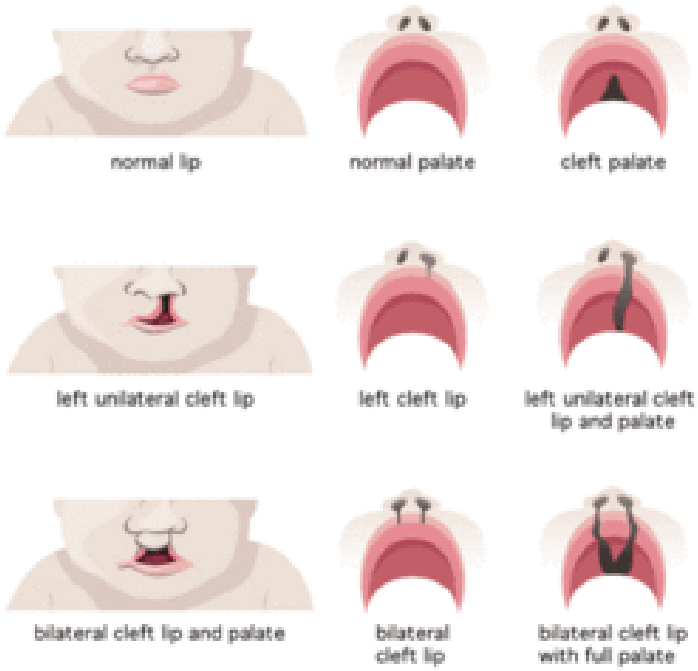
Variations in the development of the skull and facial bones are a diverse group of diagnoses called craniofacial anomalies. The two most common are cleft lip/palate and hemifacial microsomia.



Craniofacial Anomalies

There is no single known cause of craniofacial anomalies, rather they are thought to be due to the combined impact of genetics and environment during fetal development.

Women who do not take sufficient folic acid during pregnancy may have higher risk of having a baby with congenital anomalies, including cleft lip and/or palate.



The lips form during weeks 4-7 of pregnancy.

The palate forms during weeks 6-9 of pregnancy.

An infant may have one or both anomalies if the lip and/or palate doesn't join completely as it forms.

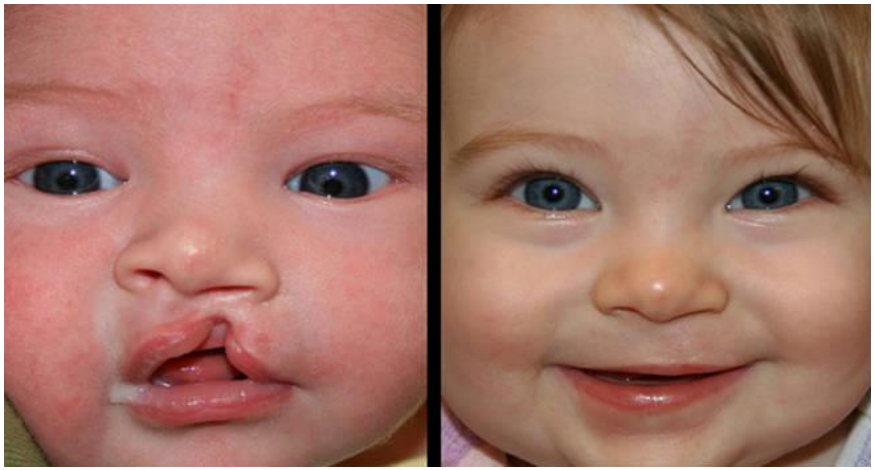
CDC estimates that, each year in the U.S., about

- 2,650 babies are born with a cleft palate
- 4,440 babies are born with a cleft lip with or without a cleft palate
- Cleft lip and palate are the most common craniofacial anomalies

Centers for Disease Control. (2015). Facts about cleft lip and cleft palate. Retrieved from http://www.cdc.gov/ncbddd/birthdefects/cleftlip.html

Craniofacial Anomalies

The primary complications of a cleft lip and/or palate is difficulty feeding, speaking clearly, and frequent ear infections.



Even after correction, some children may require support from speech therapy, specialized dental and orthodontic care, and an ENT specialist.

Surgical correction of a cleft lip typically occurs within the first few months of life.

Surgical correction of a cleft palate is recommended prior to 18 months of age.

Early surgery can help with feeding, speech development, hearing, breathing, and appearance.

Centers for Disease Control. (2015). Facts about cleft lip and cleft palate. Retrieved from http://www.cdc.gov/ncbddd/birthdefects/cleftlip.html

Craniofacial Anomalies

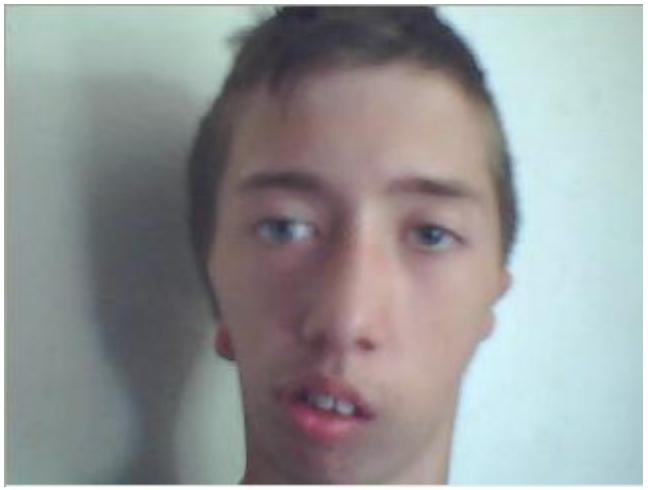
The second most common facial congenital defect is **hemifacial microsomia**.

This means that a portion of one side of the face, usually the ear and jaw, is small and underdeveloped. While their face looks different, most babies born with hemifacial microsomia do not have medical problems.

Ears
•Microtia (small ear)
•Aural atresia (no ear canal)
•Preauricular tags or facial tags (tags of skin in front of the ear)
•Other ear differences

Face
•Facial palsy (difficulty with muscle movement)
•Small cheekbone
•Epibulbar dermoid (pinkish-white growth on the eye)
•Macrostomia (wide mouth)
•Cleft lip and palate

Jaw and teeth
•Trismus (limited opening of the mouth)
•Shortness of lower jaw
•Crooked lower jaw
•Malocclusion (bad bite)



Seattle Children's Hospital. (2016). Craniofacial (hemifacial) microsomia. Retrieved from http://www.seattlechildrens.org/medical-conditions/chromosomal-genetic-conditions/hemifacial-microsomia/

Craniofacial Anomalies

Treatment of hemifacial microsomia includes:

- Surgical repair or restructuring of the facial bones and/or ear
- Speech therapy, special dental care, hearing evaluation
- Evaluation of the kidneys and heart
  - Children with CFA have a 10-15% chance of having kidney problems
- Feeding assistance
  - Often, children with CFA have trouble with breast and bottle feeding, and trouble coordinating breathing and swallowing. They may need feeding devices or nutritionist consultation.

Seattle Children's Hospital. (2016). Craniofacial (hemifacial) microsomia. Retrieved from http://www.seattlechildrens.org/medical-conditions/chromosomal-genetic-conditions/hemifacial-microsomia/

Skeletal Dysplasia

Skeletal Dysplasia

Skeletal dysplasia is a medical term for over 200 disorders characterized by short stature and abnormalities in cartilage and bone growth, resulting in disproportion of the long bones, head, and spine and abnormal skeletal size and shape.

Some people with skeletal dysplasia call this dwarfism.

The 4 most common skeletal dysplasias are thanatophoric dysplasia, achondroplasia, osteogenesis imperfecta, and achondrogenesis.

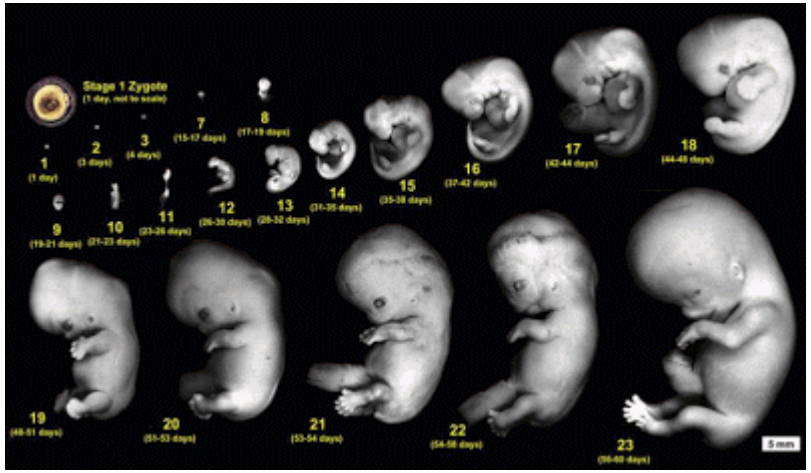
Thanatophoric dysplasia and achondrogenesis account for 62% of all lethal skeletal dysplasias.

Achondroplasia is the most common nonlethal skeletal dysplasia.

Chen, H. (2015). Skeletal dysplasia. Retrieved from http://emedicine.medscape.com/article/943343-overview#a6

Skeletal Dysplasia

Most skeletal dysplasias can be detected by ultrasound during fetal development.



Overall incidence of skeletal dysplasia is about 1 in 4-5,000 live births.

For infants with prenatally detected skeletal dysplasia, approximately 13% are stillborn, and 44% die during the perinatal period.

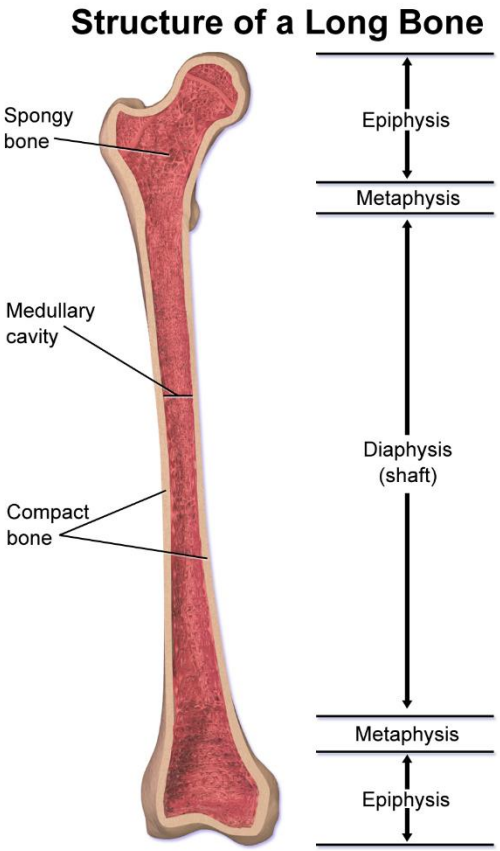
Some types of skeletal dysplasia are lethal, causing fetal death or stillbirth.

Measures, such as a scheduled Cesarean delivery, can decrease the risk of CNS complications from vaginal delivery because of the large fetal head and instability of C1-C2.

Chen, H. (2015). Skeletal dysplasia. Retrieved from http://emedicine.medscape.com/article/943343-overview#a6

Skeletal Dysplasia

Many of the gene mutations associated with skeletal dysplasias encode proteins that play critical roles in the growth plate.



FRFG3 gene mutation causes achondroplasia, hypochondroplasia, and thanatophoric dysplasia.

Clinical presenation of achondroplasia includes disproportionately short limbs with an enlarged head and average size trunk.

Chen, H. (2015). Skeletal dysplasia. Retrieved from http://emedicine.medscape.com/article/943343-overview#a6

Skeletal Dysplasia

The majority of treatment for skeletal dysplasia is supportive.

- Prevention of CNS and orthopedic complications due to spinal cord compression, joint instability, and long bone deformity.
- Monitoring height, weight, and head circumference. Prevention of obesity is important.
- Obstructive sleep apnea is common, and may require non-invasive respiratory support during sleep (i.e. BiPAP or CPAP).
- Growth hormone treatment is controversial and needs more research.
- Surgical intervention for severe scoliosis or bone lengthening procedures may be indicated or available to some people with skeletal dysplasias.

Camper Considerations

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

- Encouraging and allowing independence. Most of these campers are cognitively appropriate for their age.
- Using assistive devices. Many campers will use special devices to allow them to perform ADLs, such as eating and brushing teeth, independently.
- Minimize walking long distances. For many children with skeletal dysplasia and physical variances, this can result in quick fatigue and pain.
- Ensure set up of BiPAP/CPAP as needed at bedtime.

Thank you!

References

Bamshad, M., Van Heest, A.E., and Pleasure, D. (2009). Arthrogryposis: a review and update. *Journal of Bone and Joint Surgery*, *91*(4), 40-46. doi: 10.2106/JBJS.I.00281

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Seattle Children's Hospital. (2016). Craniofacial (hemifacial) microsomia. Retrieved from http://www.seattlechildrens.org/medical-conditions/chromosomal-genetic-conditions/hemifacial-microsomia/