Short-Stature Conditions

What are short-stature conditions?

- More than 100 specific conditions have been identified that can cause short stature. Some of these conditions involve a genetic bone disorder that causes the bones to not grow and develop normally.
- Growth-hormone deficiency (GHD) is a rare disorder characterized by the inadequate secretion of growth hormone (GH) from the pituitary gland in the brain.
- Achondroplasia is the most common short-stature condition. Children affected with achondroplasia have very short arms and short legs, while their trunks are normal size. Their heads are often large.
- Growth-hormone deficiency and achondroplasia are not related.
- Many people with these conditions are most comfortable with the term "little people" to describe their conditions.

How common are they?

- Achondroplasia occurs in people of all races and with equal frequency in males and females, and it affects about 1 in every 26,000 children.
- An estimated 10,000 individuals in the United States have achondroplasia.
- Growth-hormone deficiency has been identified in 1 in every 3,800 babies.

What are some common characteristics of children who have achondroplasia or of achondroplasia as children present with it?

- Features or effects of achondroplasia include
  - Short arms and short legs
  - A large head with a prominent forehead
  - A small midface with a flattened nasal bridge
  - Spinal curvature and back and neck problems
  - Short fingers and toes and extra space between middle and ring fingers
  - Crowded or crooked teeth
  - Bowleg (genu varum) or knock knee (genu valgum) deformity
  - Frequent ear infections
  - Vision problems
  - Hearing loss
  - Respiratory and breathing problems
  - Extra fluid within the brain (hydrocephalus)
  - Normal intelligence
- Overall, development is usually normal, yet children with achondroplasia may reach motor milestones of development slowly.
  - For instance, good head control may not occur until the infant is 7 or 8 months of age, because it takes longer to develop the muscular strength necessary to control a larger head.
  - Although there are exceptions, many of these children do not walk until relatively late, often between 2 years and 3 years (ie, 24 months and 36 months) of age.
- Weight control is a frequent and lifelong problem for many people with this disorder. Children and adults must be careful of their nutrition because they tend to gain weight easily.

What are some common characteristics of children who have growth-hormone deficiency or of growth-hormone deficiency as children present with it?

- Most children are normal size at birth.
- Over the first few years after birth, the child does not grow at the same rate as other children.
- These children are small but with proportional limbs and facial features.
- Once GHD is diagnosed, the child will receive injected GH daily.

Who might be on the treatment team?

- All children need a primary health care professional to coordinate routine preventive care services and specialist care.
- Children with achondroplasia will often see pediatric specialists in dentistry, orthopedics (bone), and otolaryngology (ears, nose, and throat) for their complications.
- Children with GHD often see a pediatric endocrinologist (hormones).
- Physical and occupational therapy may be needed to help these children achieve normal motor milestones.
- Sometimes, surgery is done to help with some of the related physical problems.
- Children who are younger than 3 years (ie, 36 months) may receive therapies through *early intervention* services.
- For children 3 years and older, *special education and related services* are available through public schools to provide the accommodations necessary for school achievement and adaptation.

Short-Stature Conditions (*continued*)

What adaptations may be needed?

Physical Environment and Other Considerations

Care Plans may include

- Adaptive equipment to support the head and spine of these children when they are younger.
- As these children grow, physical adaptations (eg, lowered doorknobs, lowered blackboards, foot supports for desks and toilets) can be used for many activities of daily living to promote independence.
- Many children will appear younger than their same-age peers because of their short statures. Be sure to take their ages and normal intelligences into account as you interact with them.
- Many children are at risk for teasing because of their physical appearances. Work to foster self-confidence with the child as well as understanding among the child's classmates.
- For achondroplasia
  - When conducting physical activity in class, be aware that jumping can cause unnecessary stress on joints, especially of the spine. Low-impact activity is encouraged.
  - Gymnastics and contact sports should be avoided because of potential risk of spine injury. Swimming and biking are encouraged. Adaptive foot pedals on bicycles to accommodate short limbs are helpful.
  - Be aware of possible hearing loss in a child who does not respond to you.

What should be considered an emergency?

Notify parents/guardians immediately for

- Unexplained numbness or tingling in the arms and legs
- Change in gait when walking
- Change in bowel or bladder control
- Severe headache with vomiting

What are some resources?

- Child Growth Foundation: www.childgrowthfoundation.org
- Human Growth Foundation: http://hgfound.org, 1-800-451-6434
- Little People of America: www.lpaonline.org
- MAGIC Foundation: www.magicfoundation.org, 1-800-362-4423

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