WESTERN CAPE ASSOCIATION FOR PERSONS WITH DISABILITIES
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Focus on... Achondroplasia *(Dwarfism)*

What is Dwarfism?
There are many conditions and diseases that can cause short stature. Some of these conditions involve a primary bone disorder -- the bones do not grow and develop normally. These conditions are called *skeletal dysplasias* or *chondrodystrophies*. People with disproportionate short stature often refer to themselves as dwarfs or short-statured persons. More than 100 specific skeletal dysplasias have been identified. Of these, achondroplasia is the most common. It occurs in all races and with equal frequency in males and females, and affects about one in every 25,000 children. Achondroplasia is recognisable at birth and after the 24th week of gestation using ultrasound. It is estimated that there are about 10,000 individuals with achondroplasia in the United States.

Characteristics
- Moderately enlarged head
- Average height and size trunk
- Short limbs
- Underdevelopment of the mid-third of the face so that the nasal bridge is relatively low or flat
- Short fingers with excessive separation of the fingertips, especially the third and fourth fingers

Challenges of disability
Children with achondroplasia may **reach motor milestones of development slowly**. For instance, good head control may not occur until the infant is three or four months old, because it takes longer to develop the muscular strength necessary to control the large head. Though there are exceptions, many children do not walk until relatively late, often between 24 and 36 months. Ultimately, overall development is usually normal.
Weight control is a frequent and lifelong problem for many people with this disorder. Both children and adults must be careful of their nutrition because they are prone to add excess weight.
Children with achondroplasia have a tendency toward **chronic ear, nose and throat infections** in the first five or six years of life, probably due to abnormal drainage of the tube from the middle ear to the throat. The basic cause is faulty development of the bone structure. If these infections are not recognised and treated, or are resistant to treatment, the child may sustain significant hearing loss. Each infection should be treated promptly and the child's hearing tested regularly. Many children with achondroplasia require ear "tubes" for treatment of recurrent ear infections.
Orthodontic problems caused by overcrowding of teeth (especially those of the upper jaw) may occur. Malocclusion (poor bite) often results and makes good oral hygiene difficult. In addition to ordinary dental care, orthodontic treatment may be necessary.
Young children with achondroplasia are at increased risk for specific **neurologic and respiratory problems** in some cases. These result from smaller openings in the skull, particularly at the foramen magnum (or "large window") at the base of the skull. In some cases, surgery is required to enlarge this opening and relieve pressure on the base of the brain and spinal cord.
Common **orthopaedic problems** in childhood include a reverse curvature (kyphosis) of the lower spine in infancy, and bowed legs, which may develop any time after children begin to walk. The spinal kyphosis usually resolves without treatment, but may require bracing or surgery if it persists. Bowed legs may be treated by bracing or orthopaedic surgery.
Older children and adults frequently experience fatigue, numbness, or pain in the lower back and thighs. Often these complaints are simple **muscular problems** that do not require special care. If they are persistent or severe, the person should be evaluated by a physician, perhaps a neurologist. Nerve or spinal-cord problems are common because of the narrow spinal canal, particularly in the lower back.