Skeletal Dysplasia

Professor Muhammad Aqeel Khan

Dr Tahir Ahmad

Learning Outcomes

- Definition
- Aetiology
- Clinical Manifestation
- Diagnosis
- Treatment
- Differential Diagnosis

Definition

- Skeletal dysplasia is **a category of rare genetic disorders that**
- **cause abnormal development of a baby's bones, joints, and cartilage**

- skeletal dysplasia affects different parts of the body in different children, the areas most often affected include the
- Legs
- Arms
- Ribcage
- Skull and
- Spine

Aetiology of Skeletal Dysplasia

- Skeletal dysplasias are caused by **genetic mutations** and can run in families.
- Often the disorders appear without any family history of skeletal dysplasia.

Clinical Manifestations

- Signs include:

  Abnormal growth in the

- Spine and
- Skull, and in
- The long bones of the arms and legs,

which can result in the individual being short in stature.

Diagnosis

Prenatal

- Skeletal dysplasia is often diagnosed during pregnancy by **prenatal ultrasound**.
- In general, the earlier skeletal dysplasia becomes detectable on an ultrasound, the more severe it tends to be.
- If a baby has a family history of skeletal dysplasia, genetic testing can detect the condition.

- postdelivery radiographs and autopsy, including histomorphic analysis of cartilage and bone

Treatment

- Though **there is** **no cure for skeletal dysplasia** There are a wide range of different treatment options depending on type
- As child matures, growth hormone therapy may be appropriate
- Orthopedic surgery may also be necessary

Prognosis

- **Certain skeletal dysplasia conditions can be fatal**
- These deaths occur when the bony chest cavity fails to grow normally, preventing normal lung development and resulting in lungs that are too small to support the baby's breathing after birth—a condition known as lethal pulmonary hypoplasia

- About half of fetuses with skeletal dysplasia are stillborn or die within the first six weeks of life.
- But not all children with dysplasias have severe medical problems. **Many of these children can live relatively normal lives**.

Short Stature Differential Diagnosis

- These disorders are the skeletal dysplasias and 372 have been documented.
- These disorders are classified using radiographic, clinical, and molecular data.

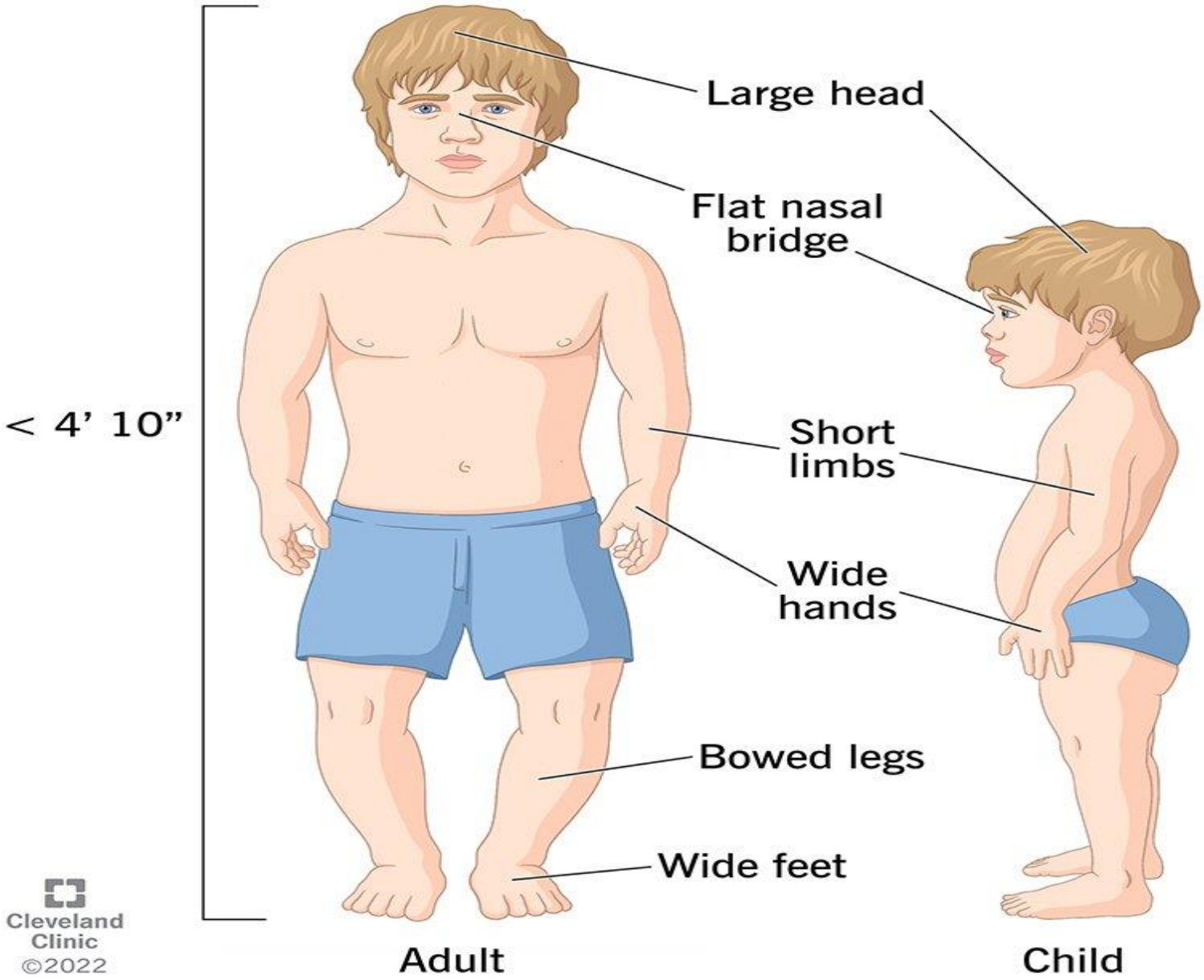
The most common dysplasias are

- **Osteogenesis imperfecta,**
- **Osteopetrosis and**
- **Achondroplasia,**


c
d
e

Dwarfism

Skeletal dysplasia



Classical features of Osteogenesis Imperfecta

- B: Blue Sclera
- D: Dental Imperfections (Dentinogenesis imperfecta with weak and discolored teeth)
- S: Sensironeural Hearing Loss
- M: Multiple fractures

Osteopetrosis(Marble Bone Diseases)

- Osteopetrosis is characterized by dense bones throughout the body.
- Symptoms include fractures, low blood cell production, and loss of cranial nerve function causing blindness, deafness, and/or facial nerve paralysis.
- Affected individuals may experience frequent infections of teeth and the bone in the jaw.

- Achondroplasia is a genetic condition affecting a protein in the body called the fibroblast growth factor receptor.
- In achondroplasia, this protein begins to function abnormally, slowing down the growth of bone in the cartilage of the growth plate

Normal and Achondroplasic sibs

Achondroplasia: Features

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