Skeletal Dysplasias

Introduction

- Skeletal dysplasias are actually more than 380 conditions that involve abnormally developed bones and connective tissues. (The word dysplasia has Latin roots meaning *bad growth*.)

Causes

- Skeletal dysplasias can be due to many types of genetic abnormalities that are often difficult to determine as well as to treat. Skeletal dysplasias are caused by genetic mutations and can run in families. Often the disorders appear without any family history of skeletal dysplasia.

Effects

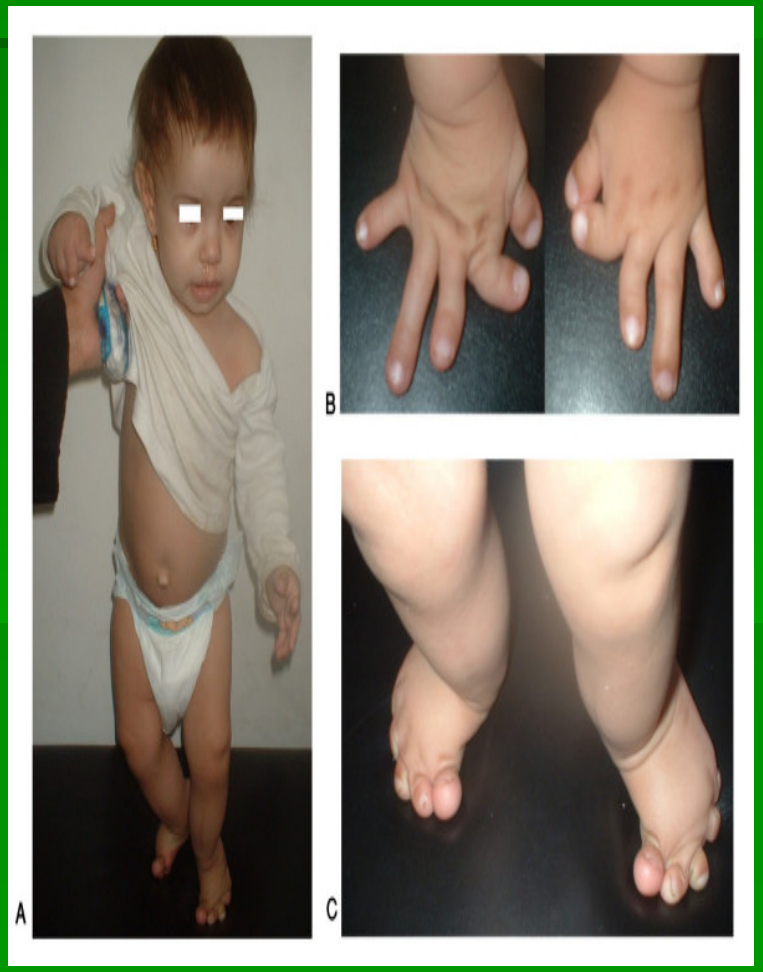
- Children with skeletal dysplasia frequently have pulmonary disease which can be life threatening. These pulmonary problems are due to multiple aetiologies including thoracic and craniofacial anomalies predisposing to restrictive lung disease, upper airway obstruction and central apnoea. Recognition of pulmonary disease and early intervention improves the survival and quality of life for these children.

Symptoms

- The specific symptoms will vary with the type of skeletal dysplasia and its cause. In general, skeletal dysplasia is associated with abnormalities in the size and shape of arms, legs, the trunk or the skull. As a result a person with skeletal dysplasia maybe unusually short with limbs that are out of proportion to the rest of the body.

Population

- No racial predilections are described.
- Males are primarily affected in X-linked recessive disorders. X-linked dominant disorders may be lethal in males.
- Otherwise, males and females are usually equally affected by skeletal dysplasias.
- Skeletal dysplasias are usually detected in the newborn period or during infancy.
- Some disorders may not manifest until later in childhood.



Treatment

- Medical care for individuals with skeletal dysplasia should be directed at preventing neurologic and orthopedic complications due to spinal cord compression, joint instability, and long bone deformity.
- Administer neonatal resuscitation and ventilatory support. Most infants with lethal skeletal dysplasias are stillborn or die within hours of birth. Given respiratory support, some infants with severe respiratory distress (eg, asphyxiating thoracic dysplasia) may survive.

Type of Mutation

- Genetic Mutation
- Achondroplasia group: These dysplasias are caused by mutations in the fibroblast growth factor 3 **gene** (FGFR3).
- Diastrophic dysplasia group: This group is caused by mutations in the diatrophic dysplasia sulfate transporter gene (DTDST).

Work Cited