LIMB DEFORMITIES

Embryology

The limb buds are identifiable by the 4th week of uterine life. In the next 4 weeks they grow and differentiate rapidly in a proximodistal sequence. The skeletal elements are found in these limb buds. Most limb defects occur in the embryonic stage of development.

Aetiology

This can be due to genetic disorders and non-genetic disorders. The later may be due to various causes like exposure to teratogenic agents or abnormal inter uterine conditions.

Genetic disorders

1. Chromosomal abnormalities

Additions, deletions and changes in chromosomal structure usually have serious effects. Affected foetuses are either still-born or become infants with severe physical and mental abnormalities. Examples with orthopaedic relevance are Down syndrome and Turner's syndrome.

2. Single gene disorders.

These are due to addition, deletion or subtraction of nucleotides in DNA. The abnormality may then be passed on to the future generation according to simple Mendelian principles.

3. Polygenic and multifactorial disorders

These are disorders that only occur when a genetic predisposition combines with an appropriate environmental "trigger". E.g. Gout is common in families with hyperuricemia, but it requires an exposure to uricogenic diet, and the acute onset is usually triggered trauma (e.g. bumping the toes).

3. Non-genetic disorders

Many developmental abnormalities are secondary to exposure to teratogenic agents like drugs e.g. thalidomide, ionizing radiations and viruses e.g. rubella. Certain groups of disorders may also be due to the limbs of the developing foetus being subjected to abnormal pressure for prolonged time. Many foot deformities e.g. talipes equinovarus deformities are thought to be due to this.

Patterns of inheritance

- Autosomal dominant disorders
- Autosomal recessive disorders
- X-linked disorders

Classification of limb deformities

The American Society for the Surgery of the Hand and the International Federation of Societies for the Surgery of the Hand classification is currently the most internationally accepted classification:

1. *Failure of formation of parts (Arrest of development)*
   a) Transverse deficiency: All the part of the limb distal to the defect is absent. It is also sometimes and erroneously referred to as congenital amputations, but true congenital amputations are secondary to constricting bands.
   b) Longitudinal deficiencies: A complete segment of the limb (e.g. the forearm) fails to develop so that the distal part of the limb (e.g. the hand) is now directly attached to the arm Thalidomide babies most usually had deformities of this type.
   One of the bones in the forearm or the leg may also fail to develop leading to another type of longitudinal disorder.
2. *Failure of differentiation (separation) of part.*
   The basic unit has developed but the final form is not completed. Examples are syndactyly and arthrogryposis multiplex congenital.
3. *Duplication*
   These usually occurred as a result of insult to the limb bud and ectodermal cap in the early stage of their development. Examples are polydactyl.
4. *Overgrowth (Gigantism)*
   The whole or part of the limb may be affected by overgrowth; usually they are due to skeletal overgrowth with normal-appearing soft parts.
5. *Undergrowth (hypoplasia)*
   This denotes defective or incomplete development of parts. This may involve the entire extremity or its divisions. It may also involve a particular type of tissue e.g. nerves, skin and nails etc.
6. *Congenital constriction band syndrome*
   A focal necrosis along the course of a limb may happen in the early post-embryonic stage if development. This may lead to annular constrictions when they heal as circular scars. When they are severe, they may result in intrauterine gangrene and foetal amputation may occur (True congenital amputation).
7. *Generalised skeletal abnormalities*
   The limb deformities are only a part of the general abnormalities the patient has developed e.g. Marfan's syndrome, achondroplasia etc.

Clinical features

1. *Pre natal*

The following conditions are associated with congenital deformities and may be indications for pre-natal screening:

- High maternal age
- High paternal age (achondroplasia)
- Previous history of chromosomal disorder
- Polyhydramnios
- Oligohydramnios

2. *Post-natal*
   a) Onset (Congenital or noticed post-natally?)
   b) Pregnancy history
      - Maternal illness during pregnancy?
      - Antenatal exposure to ionizing radiation e.g. x-rays
      - Antenatal intake of teratogenic drugs
   c) Birth History
      - Presentation
      - Type of delivery e.g. C/S, SVD or induced.

3. Family history
- Are the order siblings similarly affected?
- Are other relatives also affected
- Are the older generations also affected?
- Are all the sexes equally affected?

Investigations

In most cases the diagnosis is quite obvious, but certain investigations may be done to determine the underlying genetic defect. Others e.g. x-rays are done to establish the underlying skeletal abnormality. Maternal alpha feto-protein and Foetal imaging (USS) may be done in the pre natal period.

Management

These depends on the 1) diagnosis, 2) severity of the deformity, 3) pattern of inheritance, 4) mental capacity of the patient, and 5) his social aspiration

1. Counselling

Explain as much as possible about the deformity to the parents. The likely outcome of treatment, what will be required of the family and risk of other siblings developing the same abnormality should be discussed with the family.

2. Correction of deformity

1. Passive stretching and serial application of corrective casts
2. Surgeries
   - Release of contractures
   - Tendon transfers
   - Tenotomy
   - Corrective osteotomies
   - Limb lengthening
   - Amputation
3. Rehabilitation
   - Orthosis (callipers)
   - Prosthesis
   - Special schools
   - Walking aids e.g. Crutches and wheel chairs.
   - Vocational training.