



Kelainan kongenital

Ruby RA
FKUMM

Definition (WHO)

- Congenital anomalies are also known as birth defects, congenital disorders or congenital malformations. Congenital anomalies can be defined as structural or functional anomalies, including metabolic disorders, which are present at the time of birth.

WHO

- Congenital anomalies (also referred as birth defects) affect approximately 1 in 33 infants and result in approximately 3.2 million birth defect-related disabilities every year
- The most common serious congenital disorders are heart defects, neural tube defects and Down syndrome
- About 110 000 cases of babies born with congenital rubella syndrome can be prevented through timely vaccination of the mothers during childhood and the reproductive years.
- An adequate intake of folic acid, iodine, vaccination, and adequate antenatal care are key.

PENYEBAB



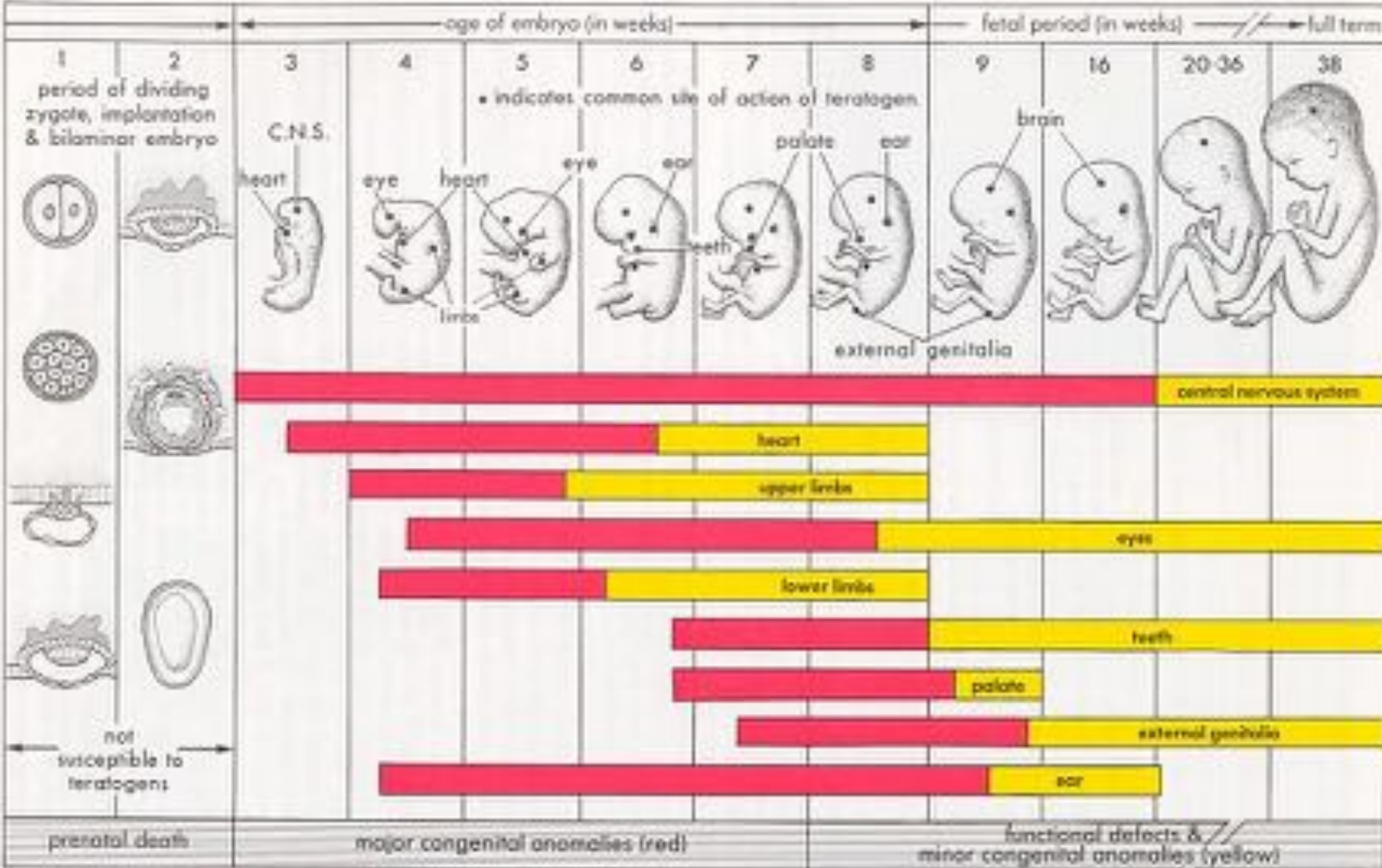
Genetik

- a. Sex atau autosomal khromosom
- b. dominan atau resesif

2. Lingkungan selama proses kehamilan

- a. pesticides, medicinal and recreational drugs, alcohol, tobacco, certain chemicals, high doses of vitamin A during the early pregnancy, and high doses of radiation
- b. infeksi
- c. trauma mekanis
- d. anoxia/hypoxia
- e. Nutrien → sosek

3. Kombinasi genetik dan lingkungan



Schematic illustration of the critical periods in human development. During the first two weeks development, of the embryo is usually not susceptible to teratogens. During these pre-embryonic stages, a teratogen either damages all or most of the cells, resulting in its death, or damages only a few cells, allowing the conceptus to recover and the embryo to develop without birth defects. *Red denotes highly sensitive periods* when major defects may be produced (e.g. amelia, absence of limbs). Yellow indicates stages that are less sensitive to teratogens when minor defects may be induced (e.g. hypoplastic thumbs)

Prevention

- Improving the diet of women throughout their reproductive years.
- Avoiding exposure to hazardous environmental substances (e.g. heavy metals, pesticides, some medicinal drugs) during pregnancy.
- Improving vaccination coverage, especially with rubella virus, for children and women.
- Increasing and strengthening education to health staff and others interested in promoting birth defects prevention.

Detection

- Preconception screening is used to identify persons at risk for specific disorders or at risk for passing one on to their children.
- Antenatal screening includes screening for advanced maternal age, Rhesus blood group incompatibility, and carrier screening. Ultrasound can be used to detect Down syndrome during the first trimester and serious fetal anomalies during the second trimester
- Newborn screening includes clinical examination and screening for haematological, metabolic, and hormonal disorders.

Syndactily

- kegagalan separasi (differensiasi) jari pada minggu ke 6-8 intra uterine
- 1 dari 2250 kelahiran
- 50% bilateral
- 57% antara jari III-IV
- pria 2x wanita
- 10-40% ada riwayat keluarga
- multifaktorial



syndactyly

Klinis :

- jari menjadi satu
- Simple (skin & soft tissue) & complex (tulang)
- complete & incomplete

Therapy :

- Teknik : - simple : Z plasty
 - complex : separasi tulang dan
Z plasty



Macrodactily

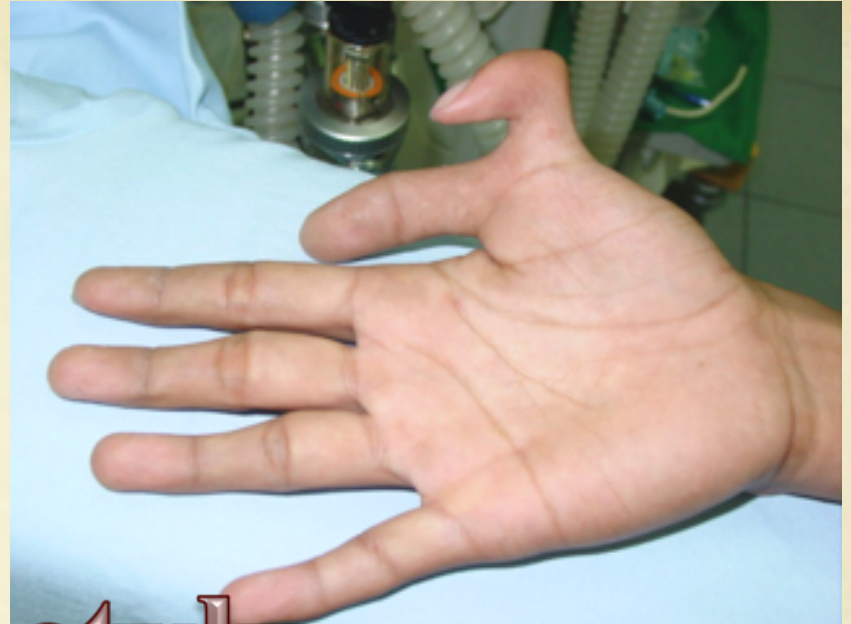




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polydactyly







neurofibroma





neurofibroma



Congenital Talipes Equino
Varus
(Club Foot)

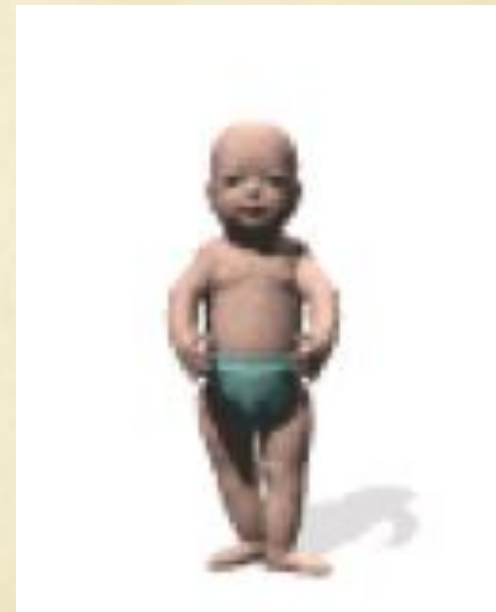
Intrauterine Position



The hips are always flexed and externally rotated, while the knees are usually flexed and the feet turned inward

PRINSIP TERAPI

Bertujuan mencapai kesempurnaan fungsi organ dengan mengkoreksi kelainan anatomis sedini mungkin



Makna Kata

Talipes = Talus = ankle

Pes = kaki

Equinus = jari lebih rendah

Varus = lateral kaki sebagai alas



OTEV

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CTEV



Gambaran Klinis

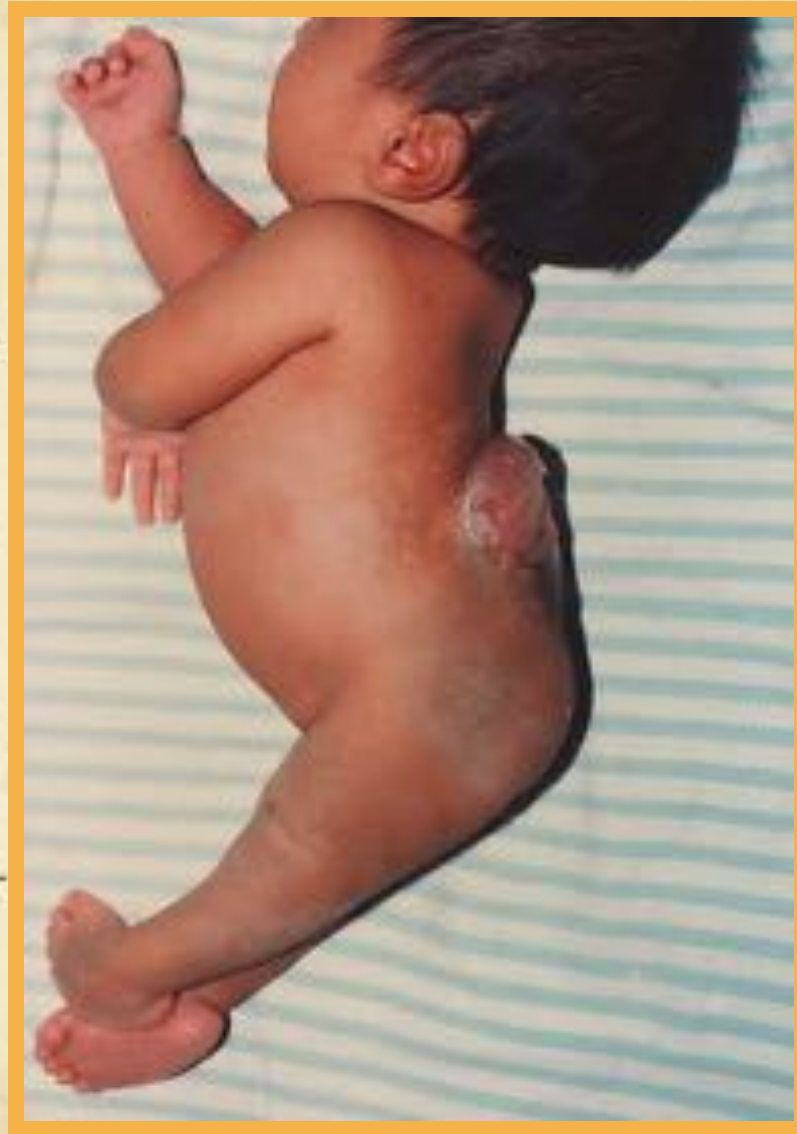
- Cavus - relative pronation of forefoot c.f. hindfoot
- Adduction – forefoot
- Varus – heel
- Equinus – heel
- Supination – midfoot



Test dorso flexi (24 jam pertama)
ibu jari tidak pada crista tibia

Differential Diagnosis

- Spina bifida
- Agenesis tibia
- Constriction band



- *Terapi :*
- a.Konservatif :
 - serial plasting selama 8 minggu
 - pada tipe fleksibel (subyektif)
 - umur \leq 5 bulan
 - sirkuler gips atas lutut (above knee)
- b.Operatif :
 - konservatif gagal
 - umur $>$ 5 bulan
 - tipe rigid
 - dilakukan post medial release (PMR) + Achilles tendon lengthening (ATL) kmd sirkular gips 6 minggu



NORMAL



MET. VARUS



CLUBFOOT

AP view



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Komplikasi



Rocker Bottom Foot

Komplikasi



Skin Blister



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Doni



Upper Extremity

Terminal - longitudinal



Paraxial hemimelia
(Radial)



Paraxial hemimelia
(ulnar)



Partial
adactyly
(ray 1)



Paraxial aphalangia
(ray 3)

Lower Extremity

Terminal - longitudinal



Paraxial hemimelia (fibular)



Partial adactyly

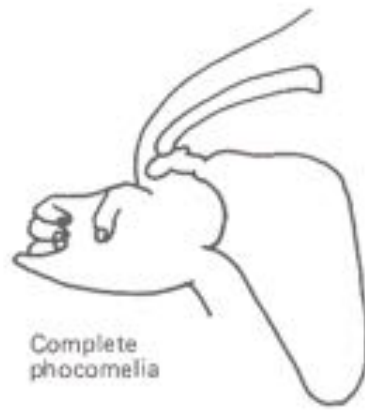


Paraxial Hemimelia (tibial)



Partial aphalangia

Intercalary - transverse



Complete phocomelia



Proximal phocomelia



Distal phocomelia

Intercalary - longitudinal

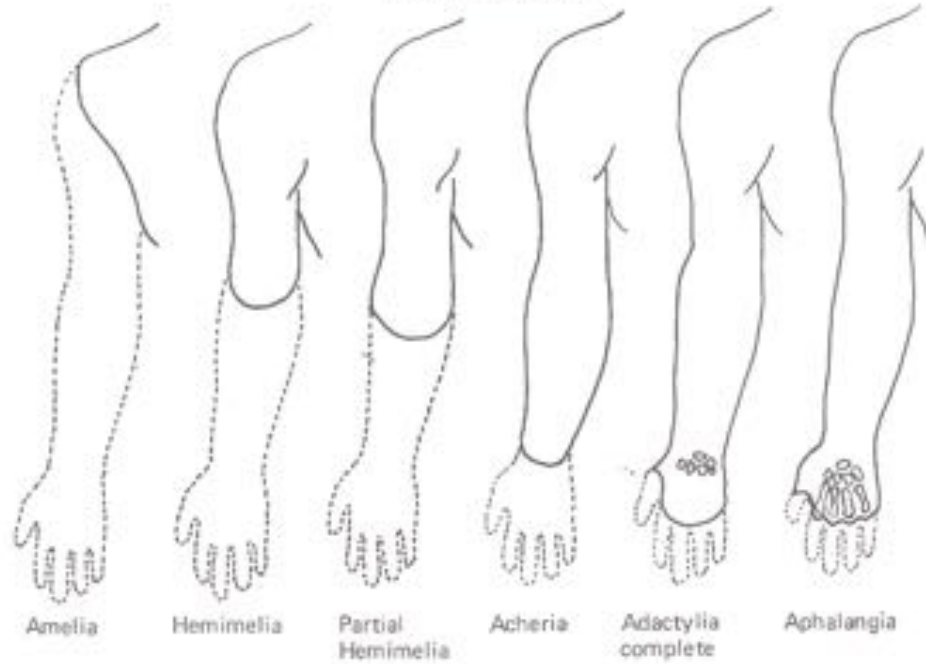


Paraxial hemimelia (radial)

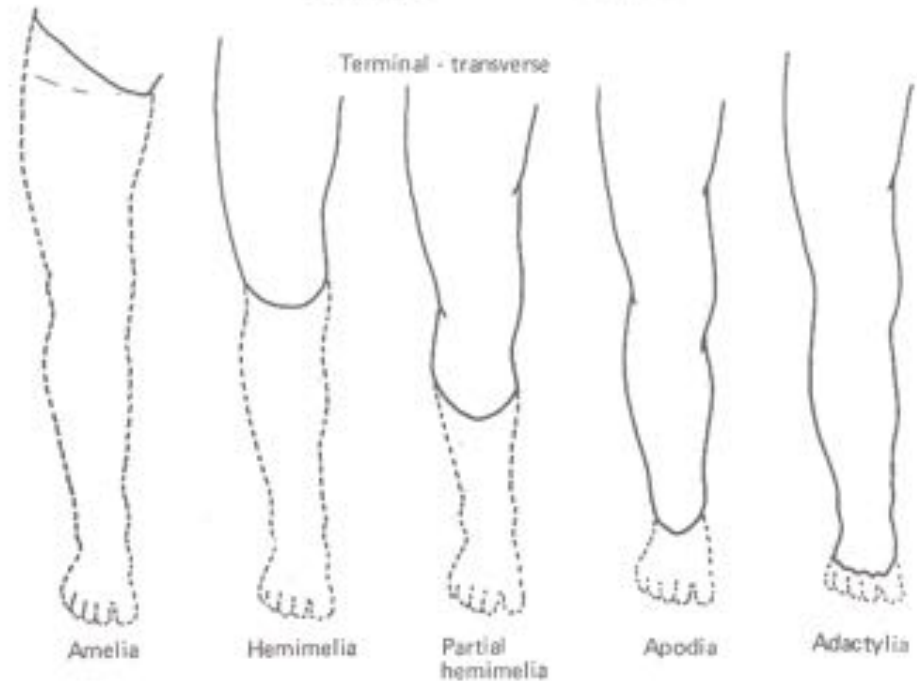


Paraxial hemimelia (ulnar)

Terminal transverse

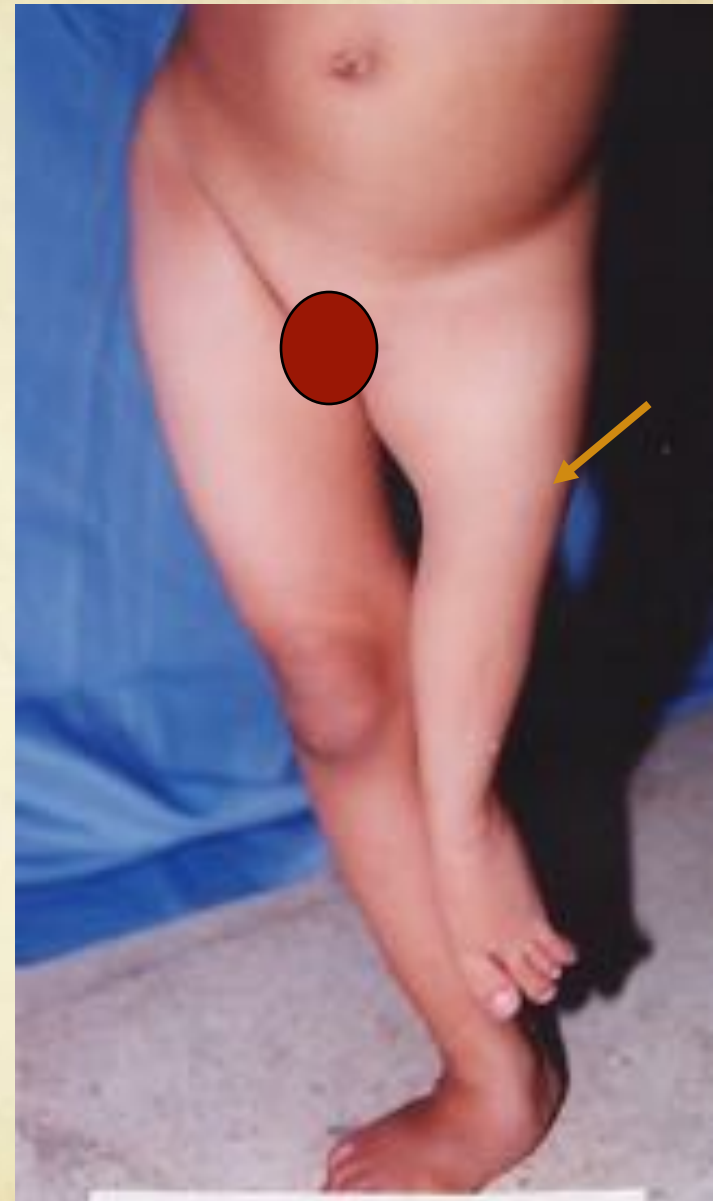


Terminal - transverse



→ PFFD

(Proximal Focal Femoral Deficiency)







PFFD (WANITA 4,5 TAHUN)



Osteogenesis Imperfecta

Ciri-ciri

- Hereditas autosomal
- Dominan atau resesif
- Tulang osteoporotik dan mudah patah
- Tulang bengkok (bowing)
- Ligamen hyperlaxity
- Blue sclera
- Dentinogenesis





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ANOMALI NEPHRIC SYSTEM

A. JUMLAH :

1. RENAL AGENESIS :

- Unilateral atau bilateral
- Didapati secara kebetulan
- Tanpa keluhan (bila unilateral)

2. SUPERNUMERARY KIDNEY : jarang

BILATERAL RENAL AGENESIS

- Lahir mati : (kebanyakan)
- Masa hamil : Oligo Hydramnion
- Tanda khas : " Potter Face "
- Yaitu
 - "Elfin Ears" → telinga lebar letak rendah
 - Hidung → flattening
 - Mata → lebar
- Laki-laki → lebih sering

ANOMALI NEPHRIC SYSTEM

B. RENAL EKTUPIK

Macam :

- Pelvic Ectopic Kidney
- Crossed Ectopic Kidney
- Thoracic Kidney
- Abdominal Kidney