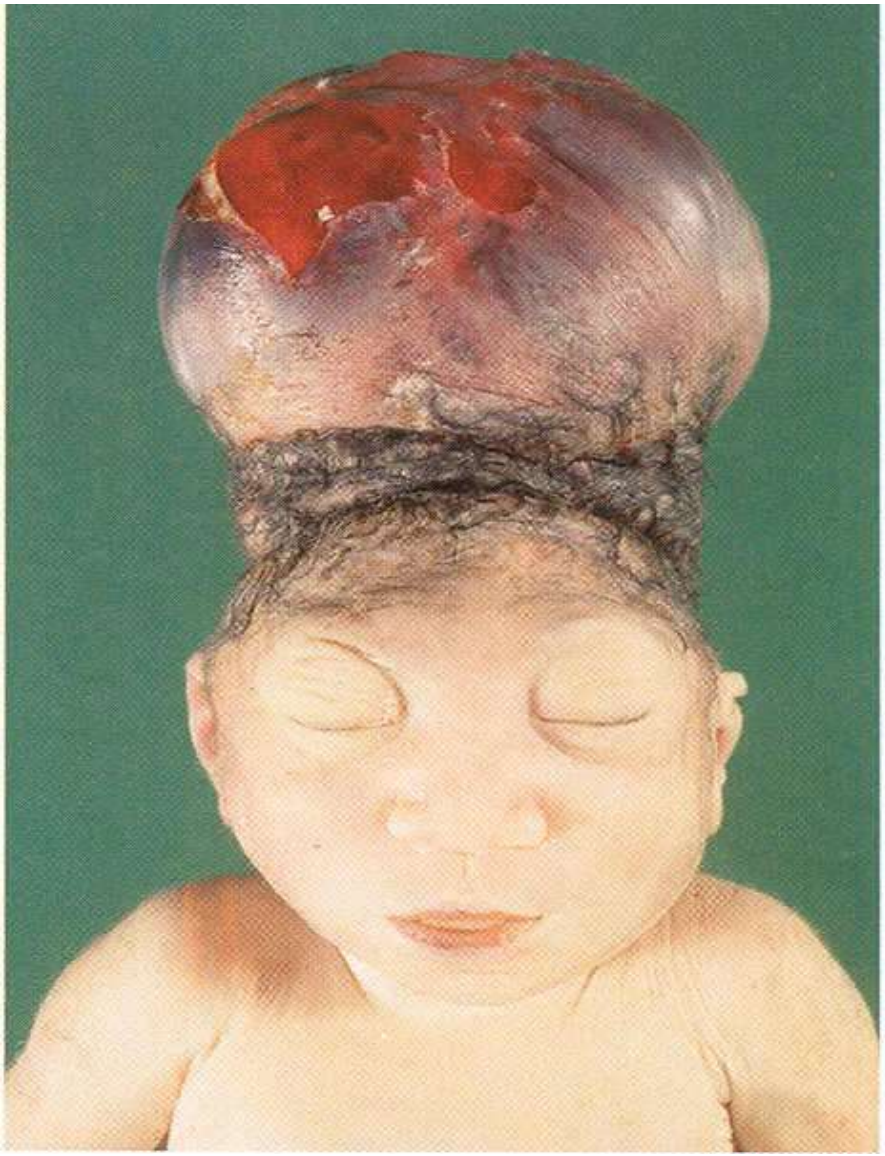
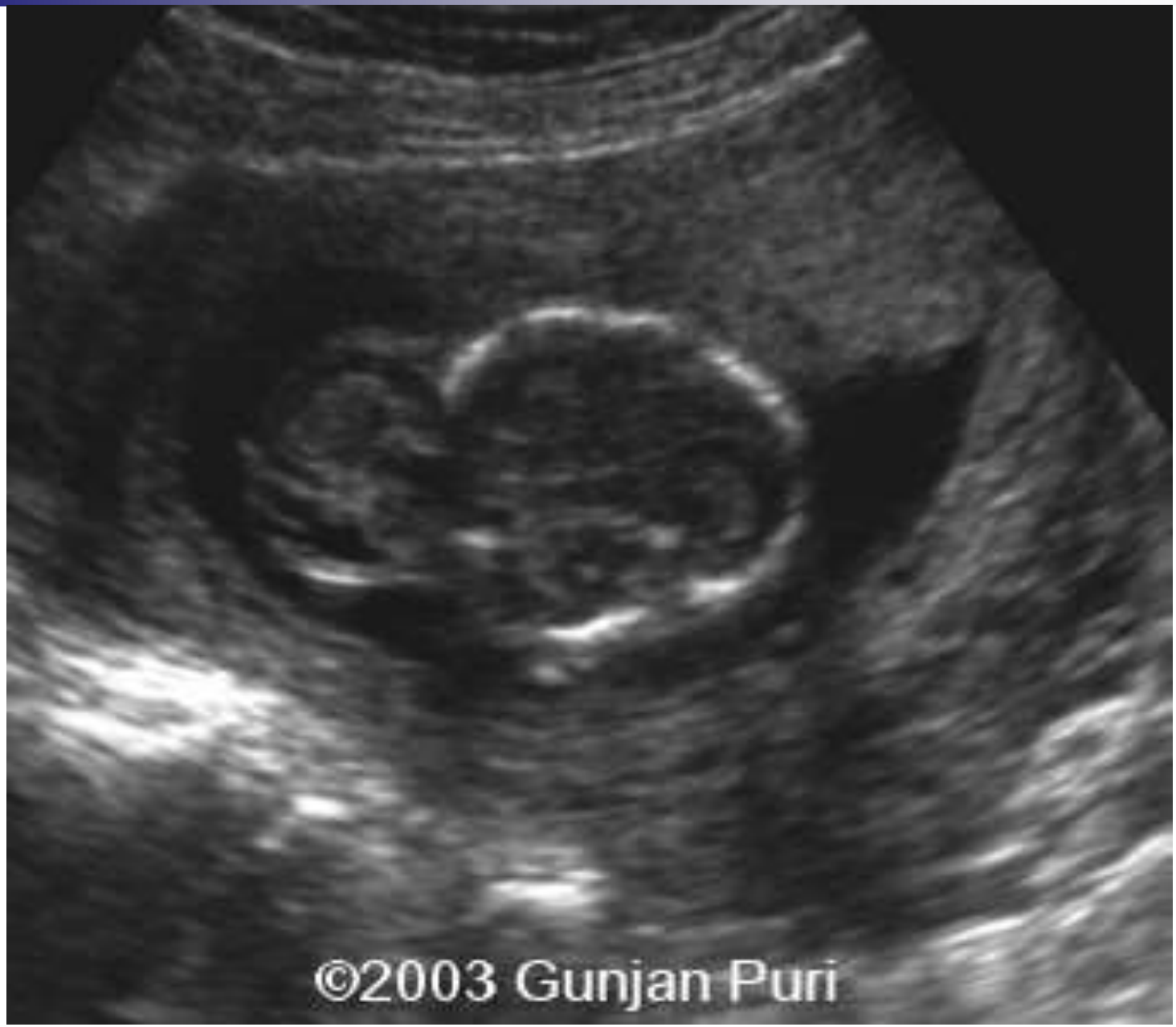
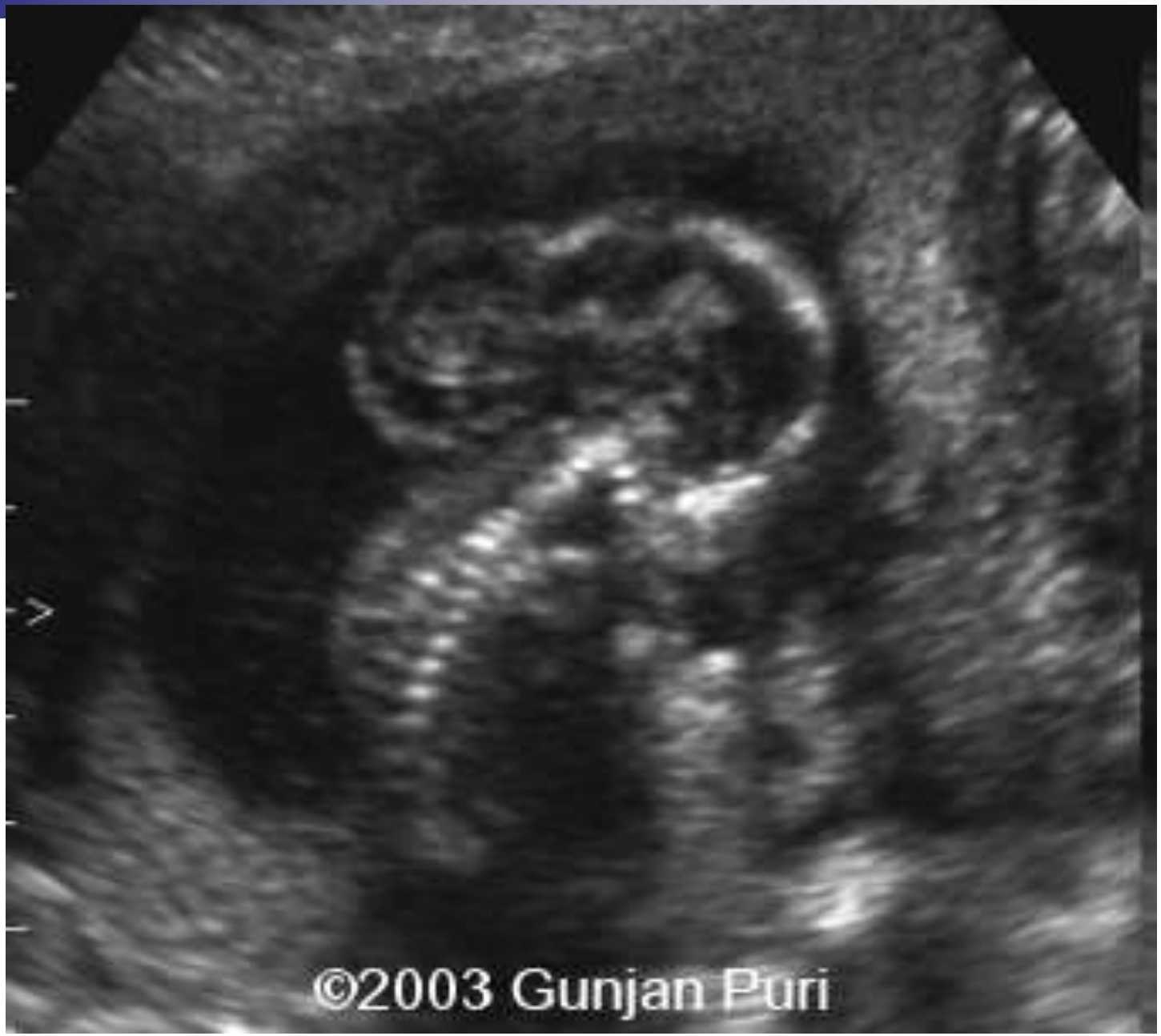
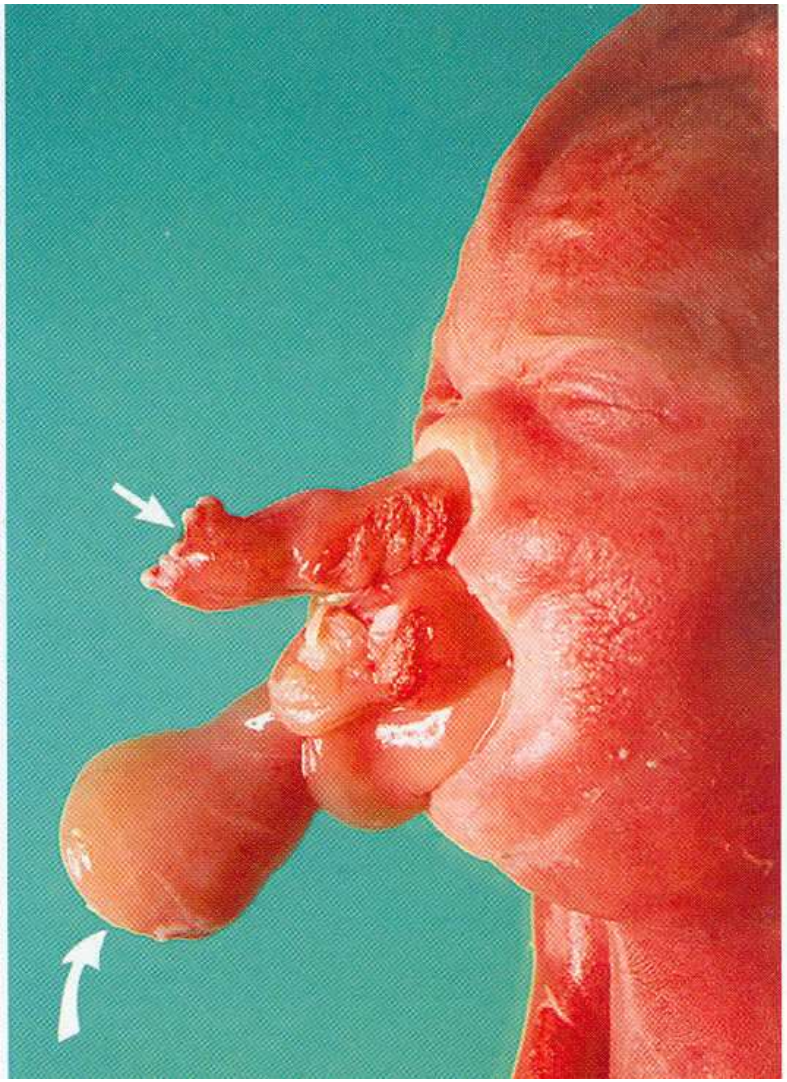


Large encephalocele

- 1-3 in 10,000 births
- Recurrence is 25% with Meckel-Gruber Syndrome
- Can be easily diagnosed in late 1st trimester

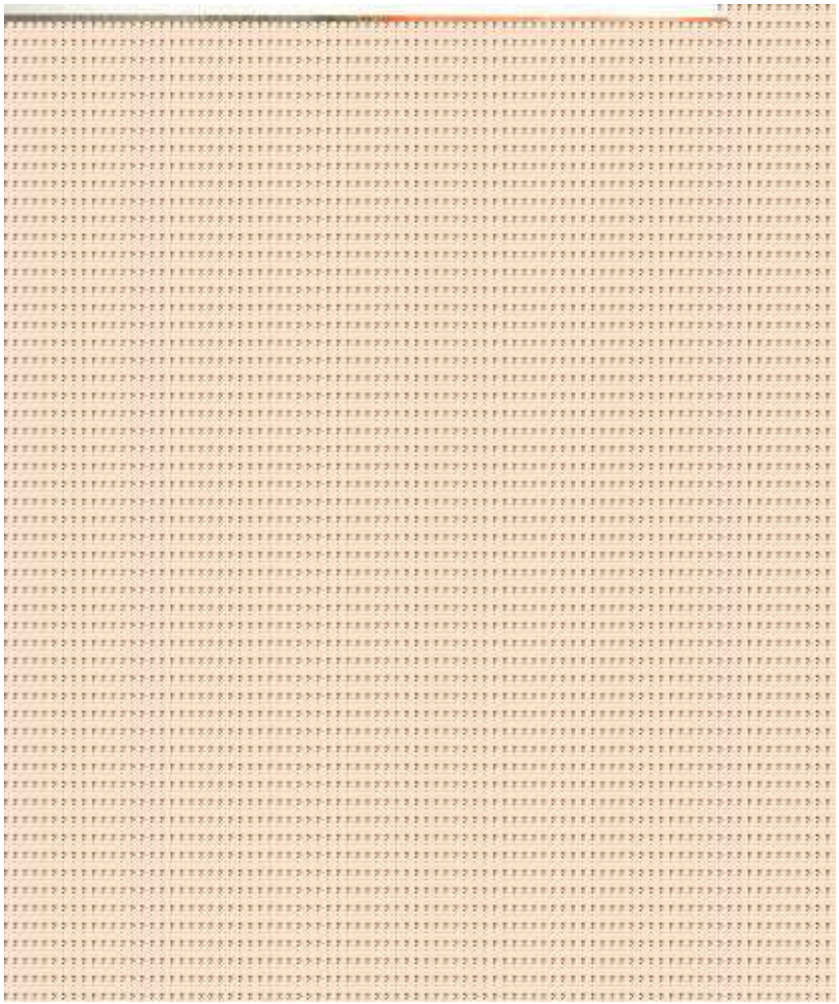

©2003 Gunjan Puri


©2003 Gunjan Puri



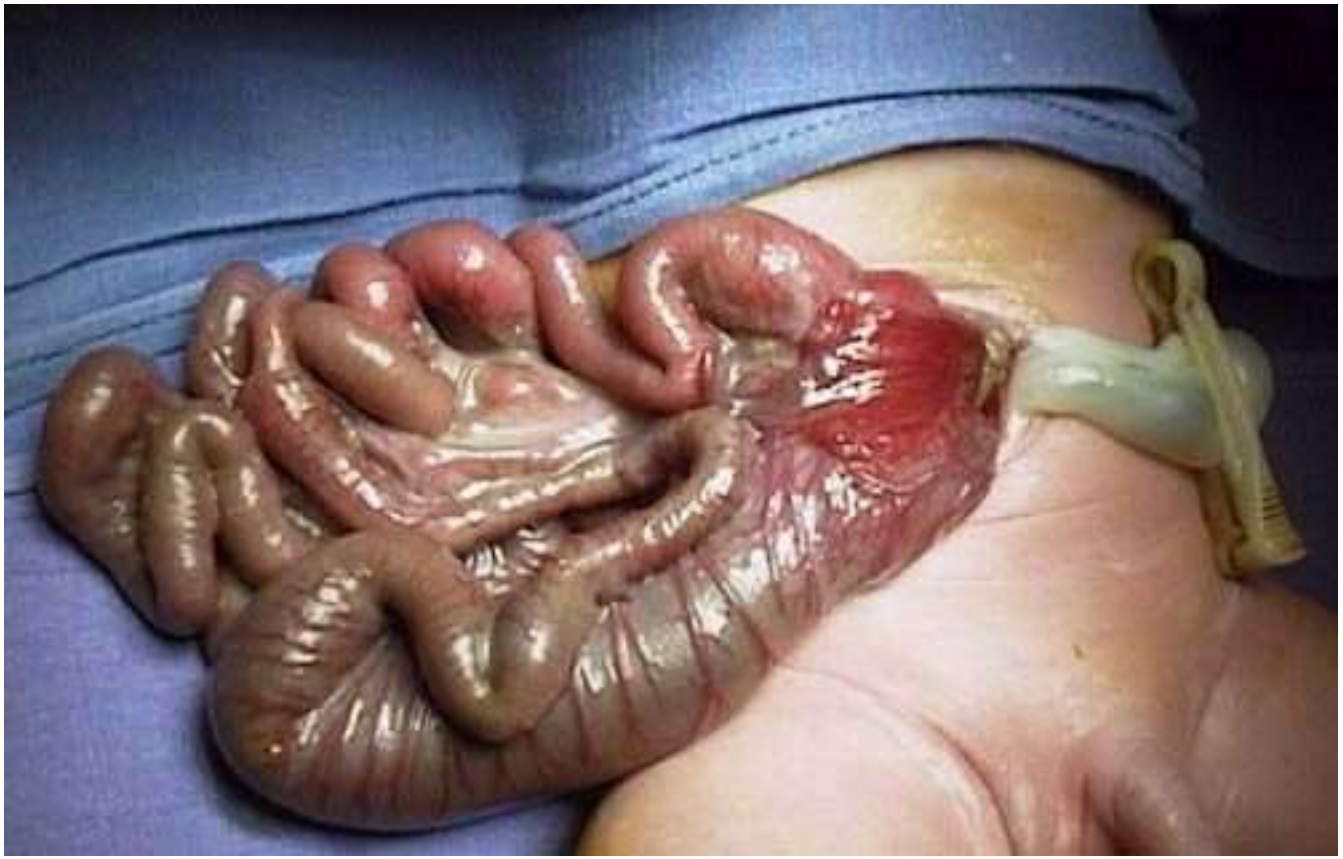
Oral teratoma (epignathus)

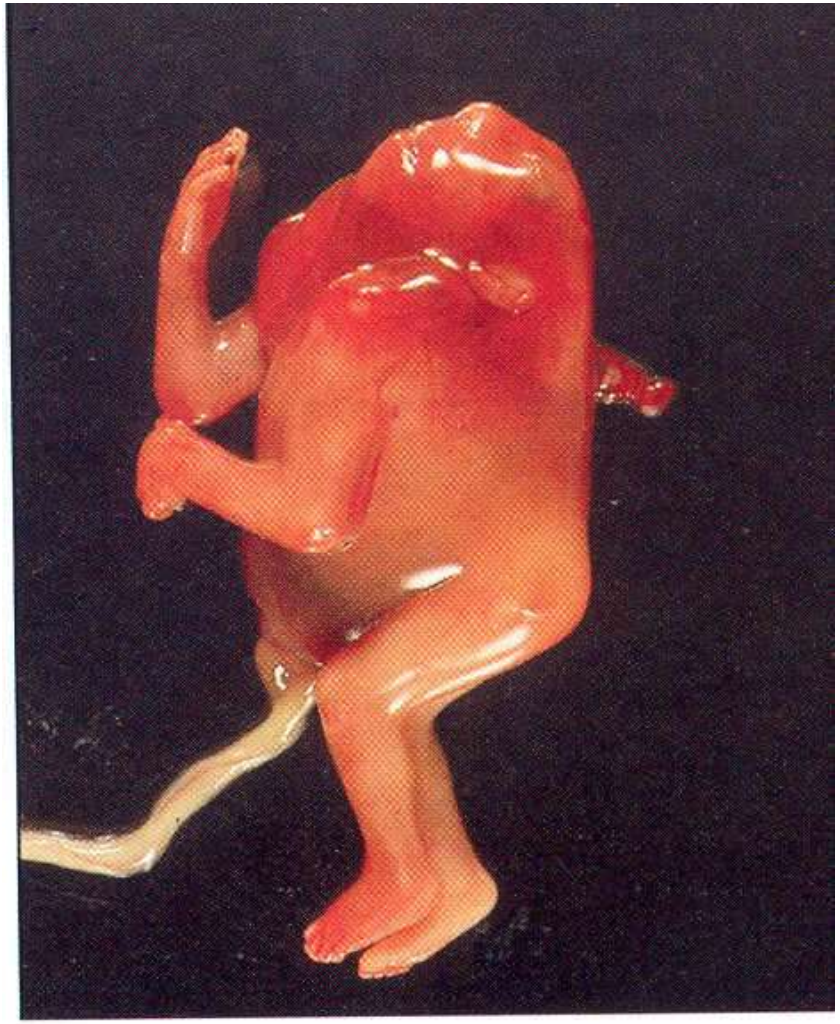
- Rare
- 2 % of all childhood teratomas
- C/S and tracheostomy prior to surgical excision



Gastroschisis

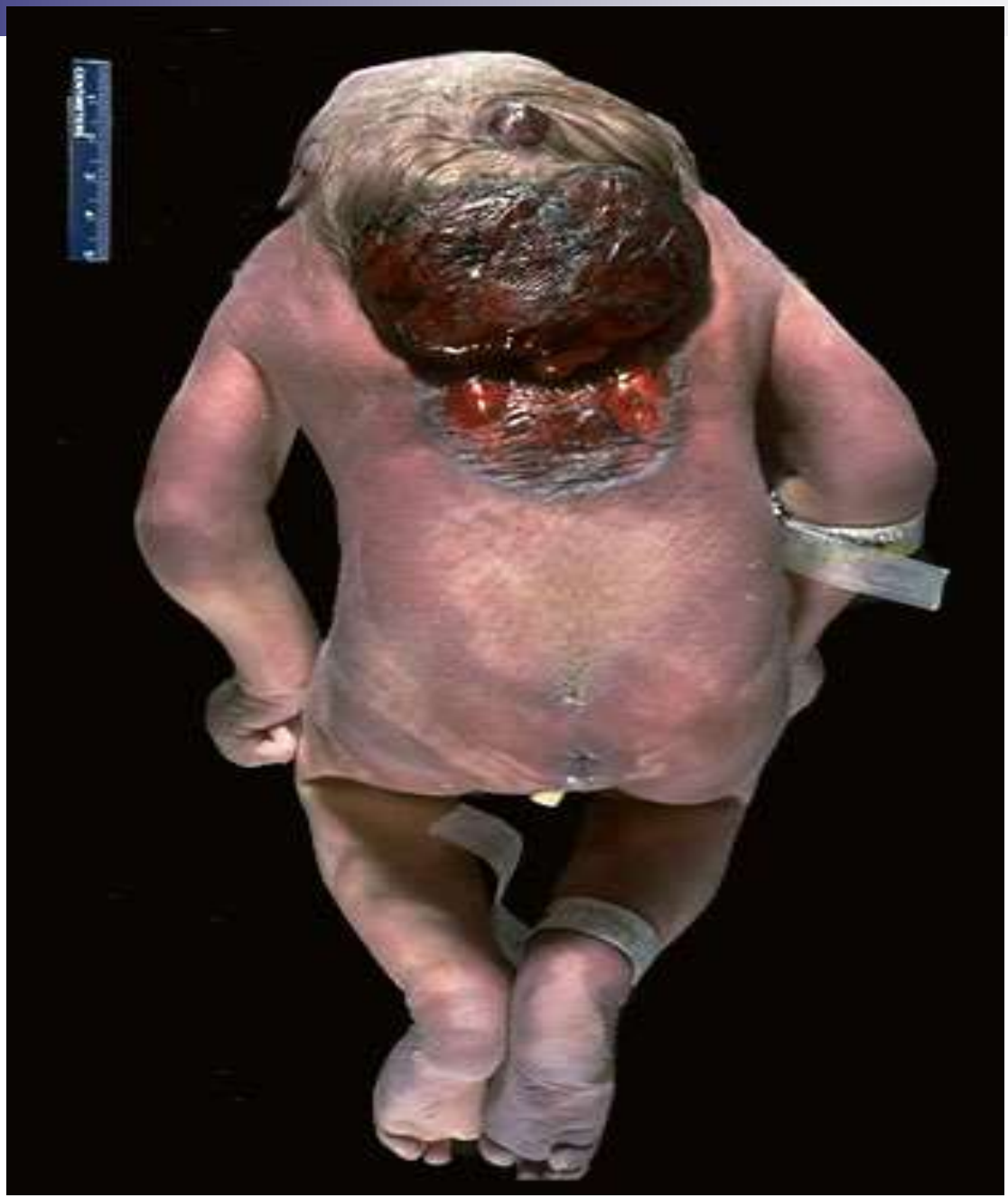
- 1 in 3,000 births
- Isolated sporadic
- May benefit from C/S
- Can be diagnosed early 2nd trimester

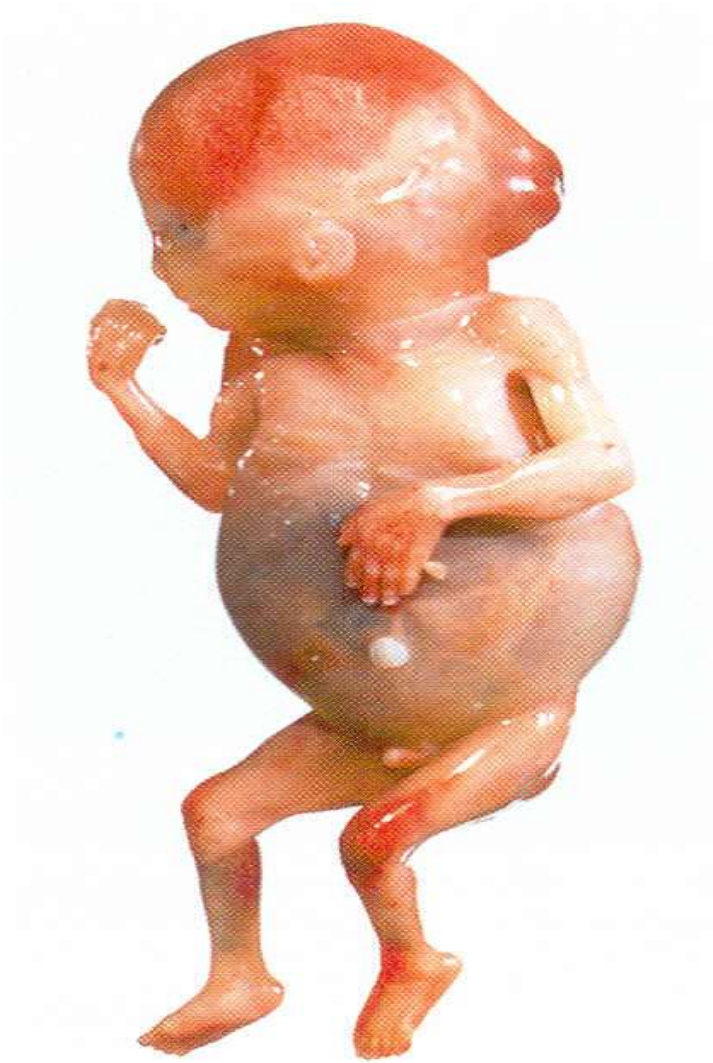




Iniencephaly

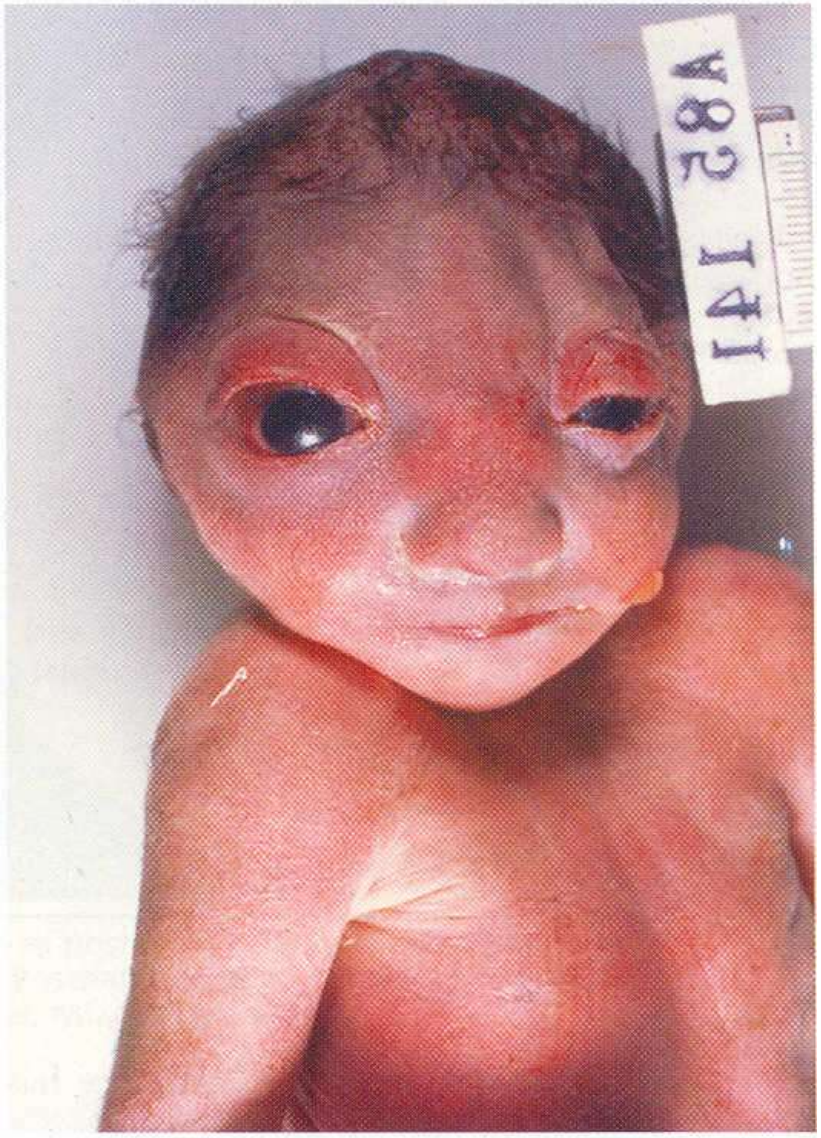
- 1-10 in 10,000
- Rare NTD
- Absent occiput (extremedorsal flexion)
- Rachischisis
- Lethal in most cases





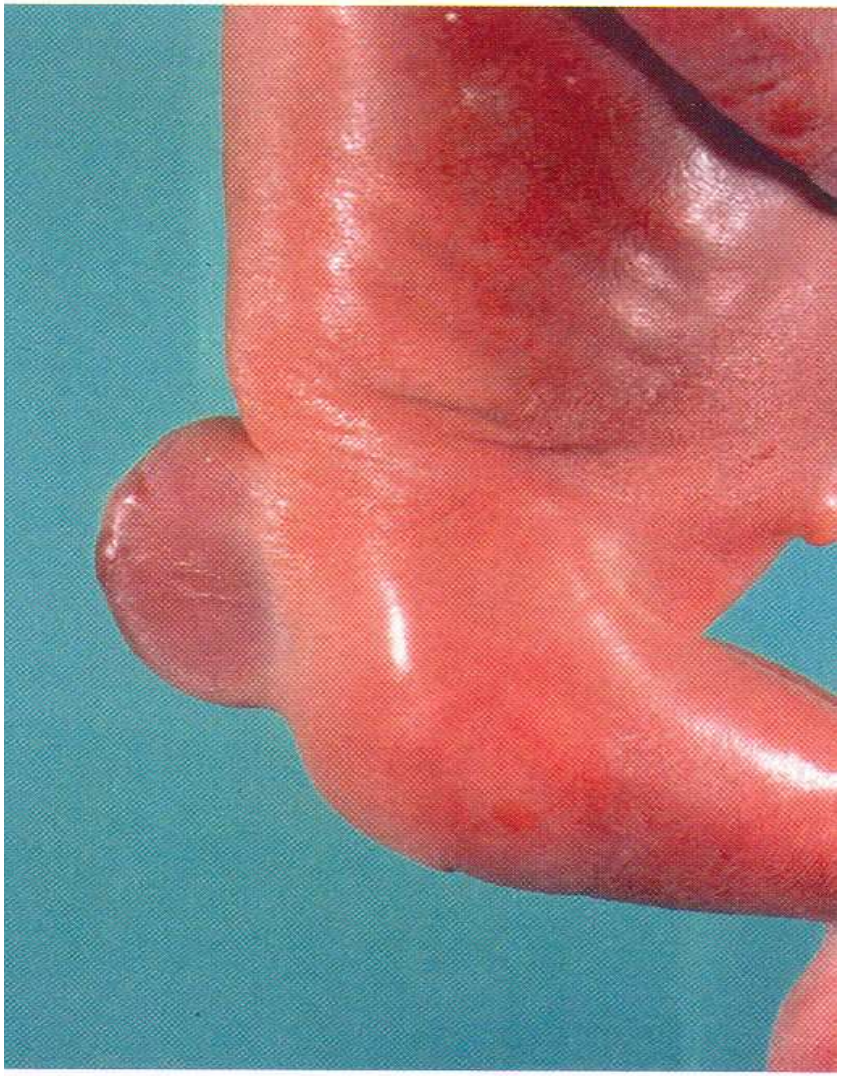
Meckel-Gruber Syndrome

- 1 in 9,000
- Autosomal recessive
- Occipital encephalocele-polydactyly-polycystic kidneys
- Diagnosed as early as 13 weeks
- Fatal disease



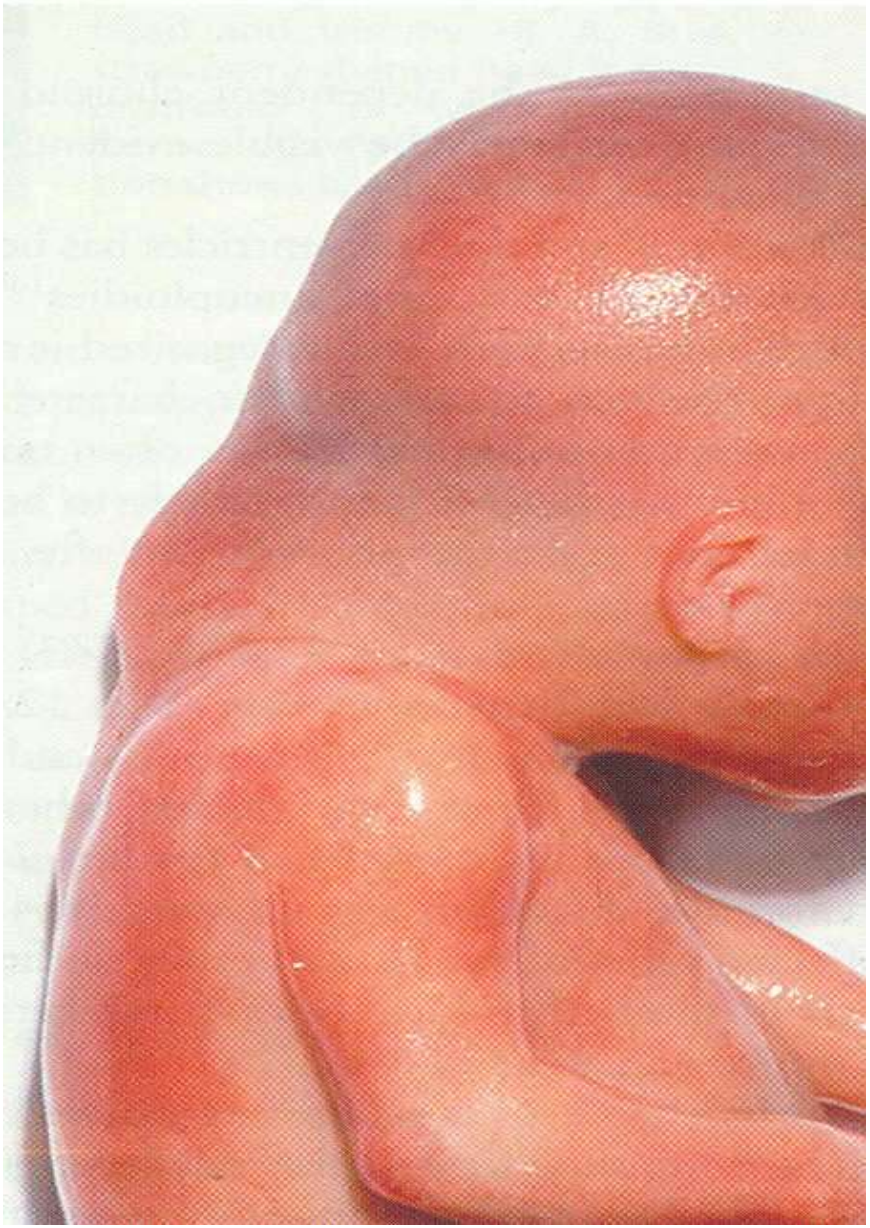
Microcephaly

- 1-2 in 1,000 births
- Multiple causes (infections, teratogenic, genetic)
- Wide range of severity
- Lethal if associated with trisomy 13, 18



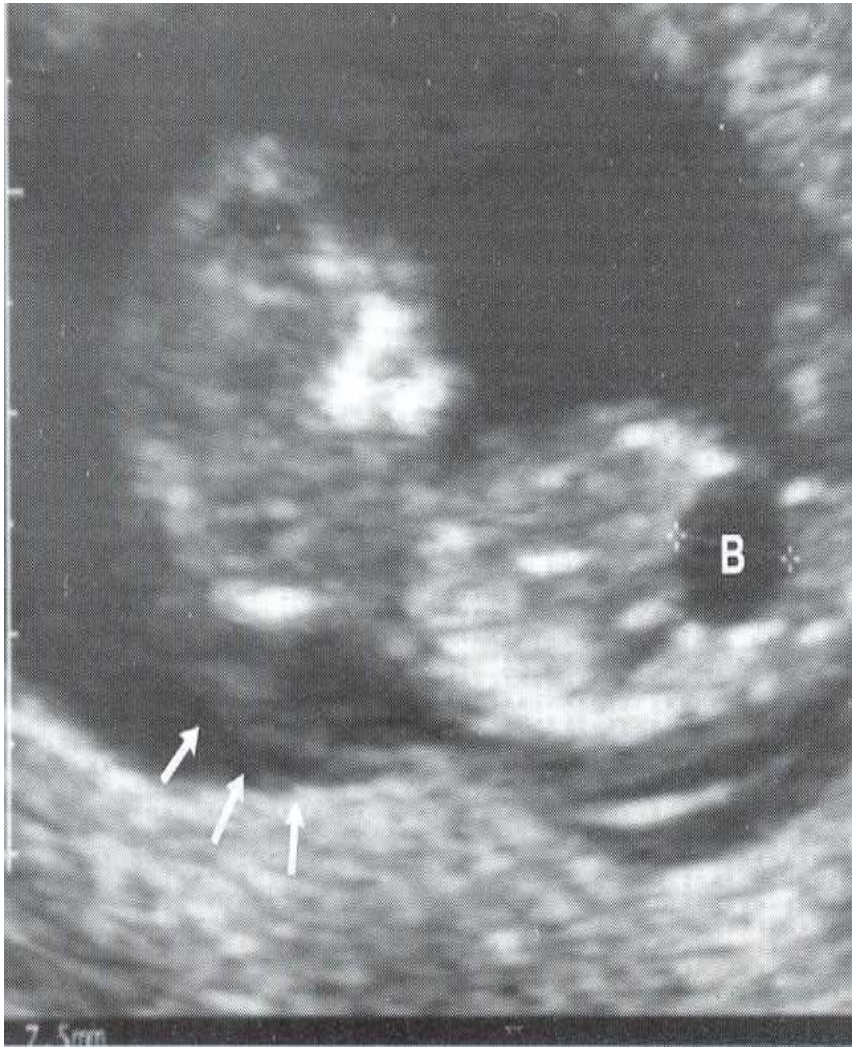
Spina bifida

- 1 in 1,000 bifida
- AFP + U/S diagnosis (85-100% of cases)
- Recurrence is reduced by 71% with folic acid



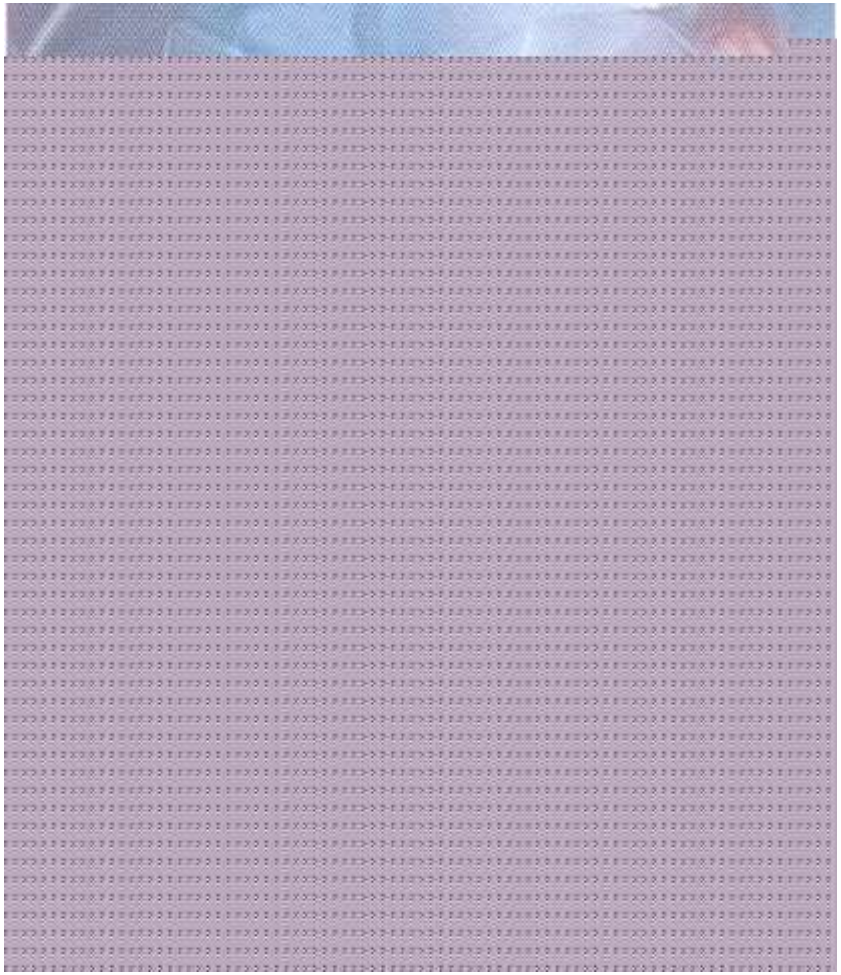
Nuchal thickening

- Marker for aneuploidy
- Mid trimester scan
- >5mm
- Sensitivity 40%



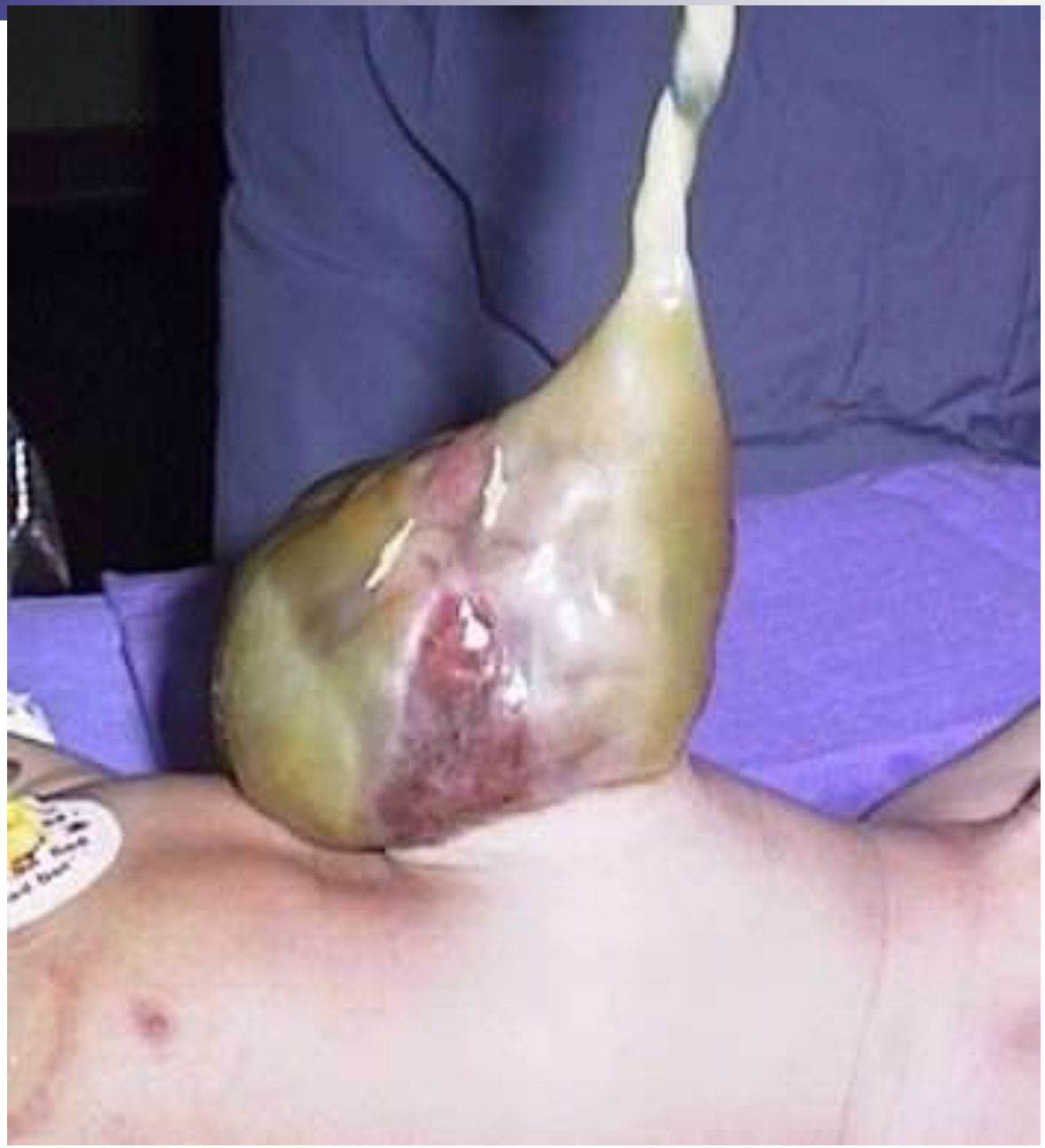
Trisomy 13

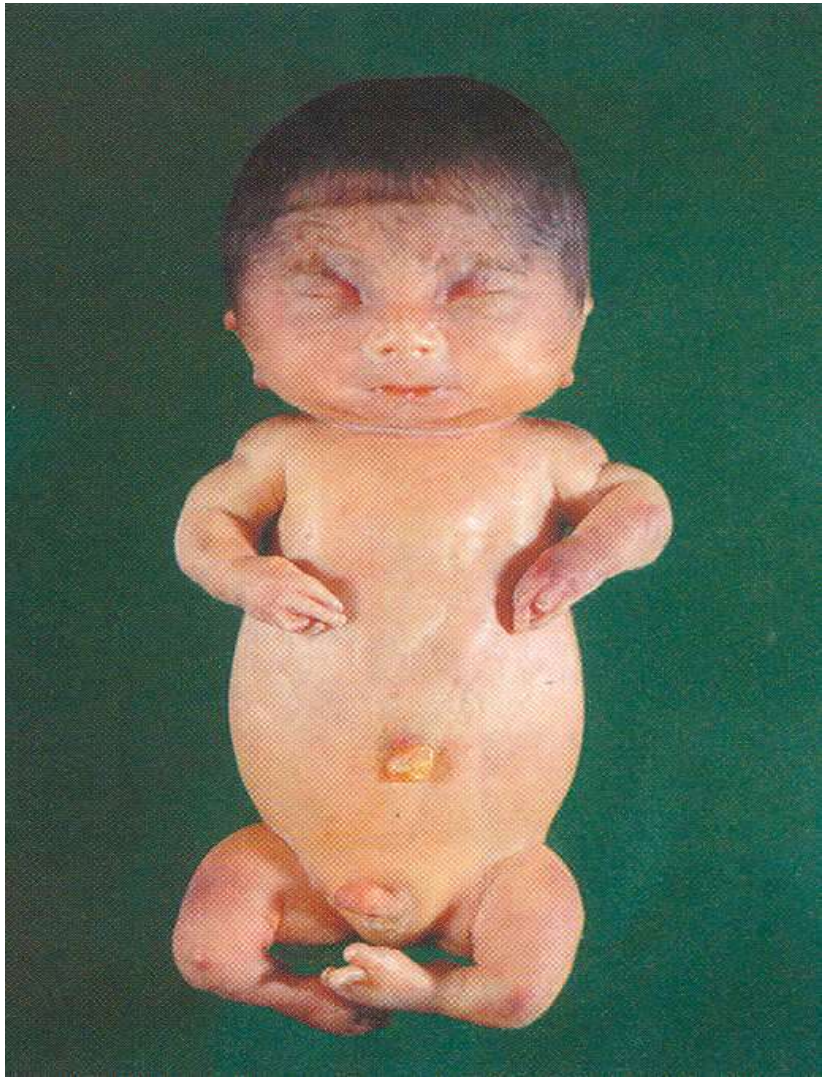
- 11 Weeks scan
- Megacystis
- Increased nuchal translucency



Omphalocele

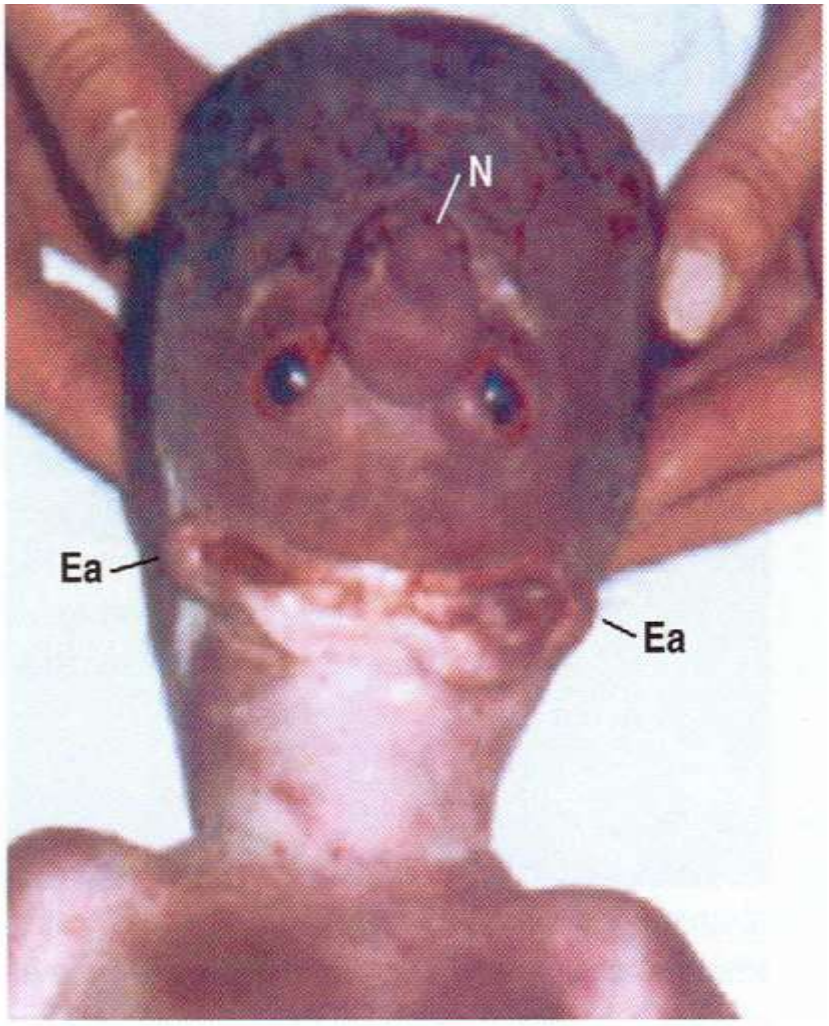
- 1 in 4,000
- (30-50%) Aneupolidy rate (T18, T13, 45x0)
- Prognosis depends on associated anomalies





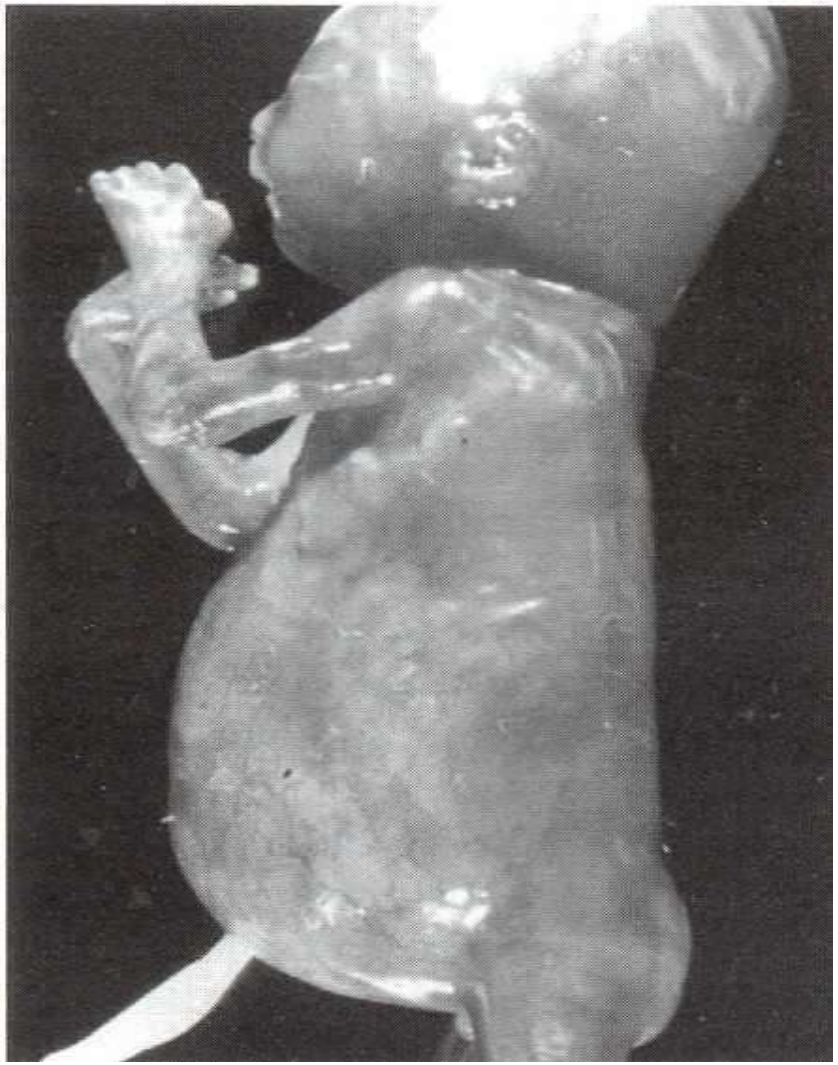
Osteogenesis imperfecta

- 0.5 in 10,000
- Severe forms cause multiple fractures
- Type 2 is lethal



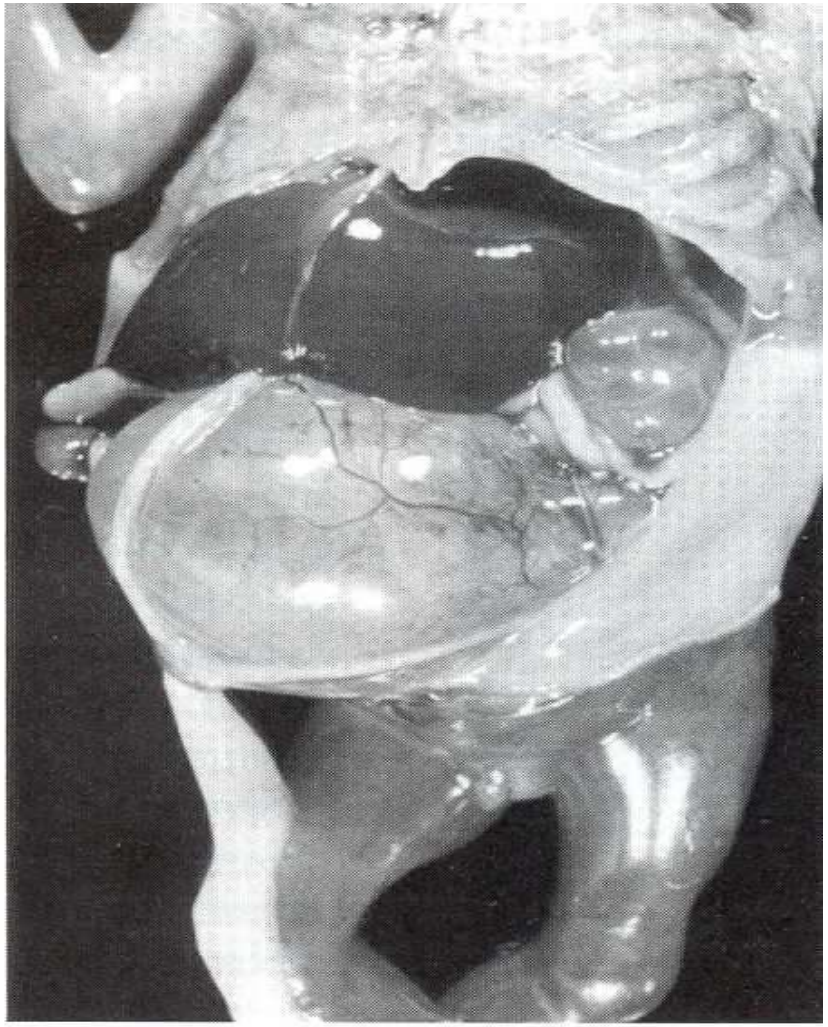
Otocephaly

- Very rare
- Failure of assent of ears
- Agnathia, proposis, holoprosencephaly
- Lethal condition in general



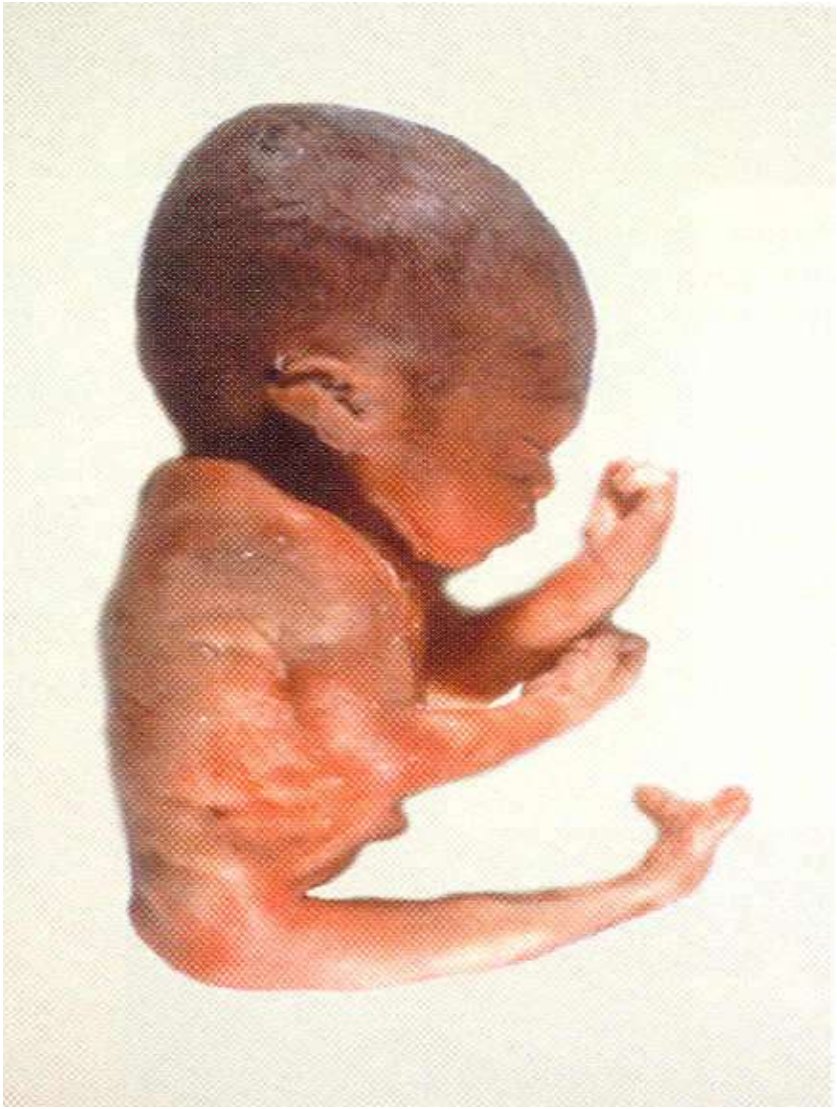
Posterior urethral valve

- 1 in 5,000 boys
- Severe cases are lethal



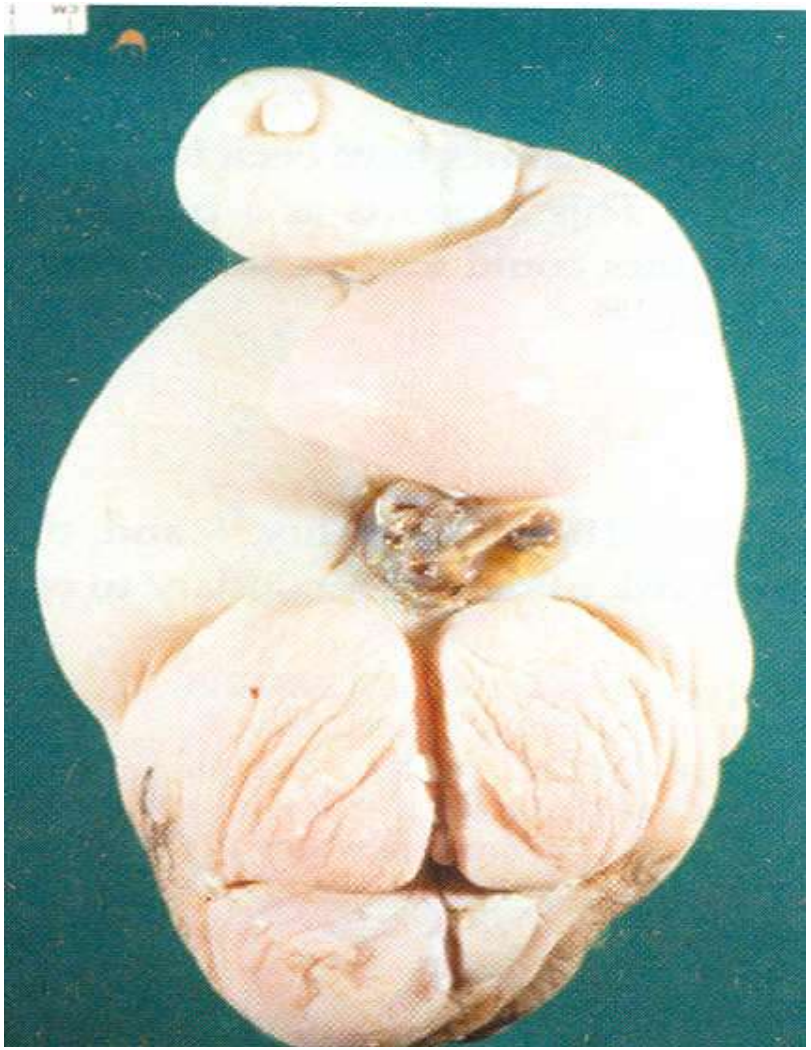
Posterior urethral valve

- Distended bladder



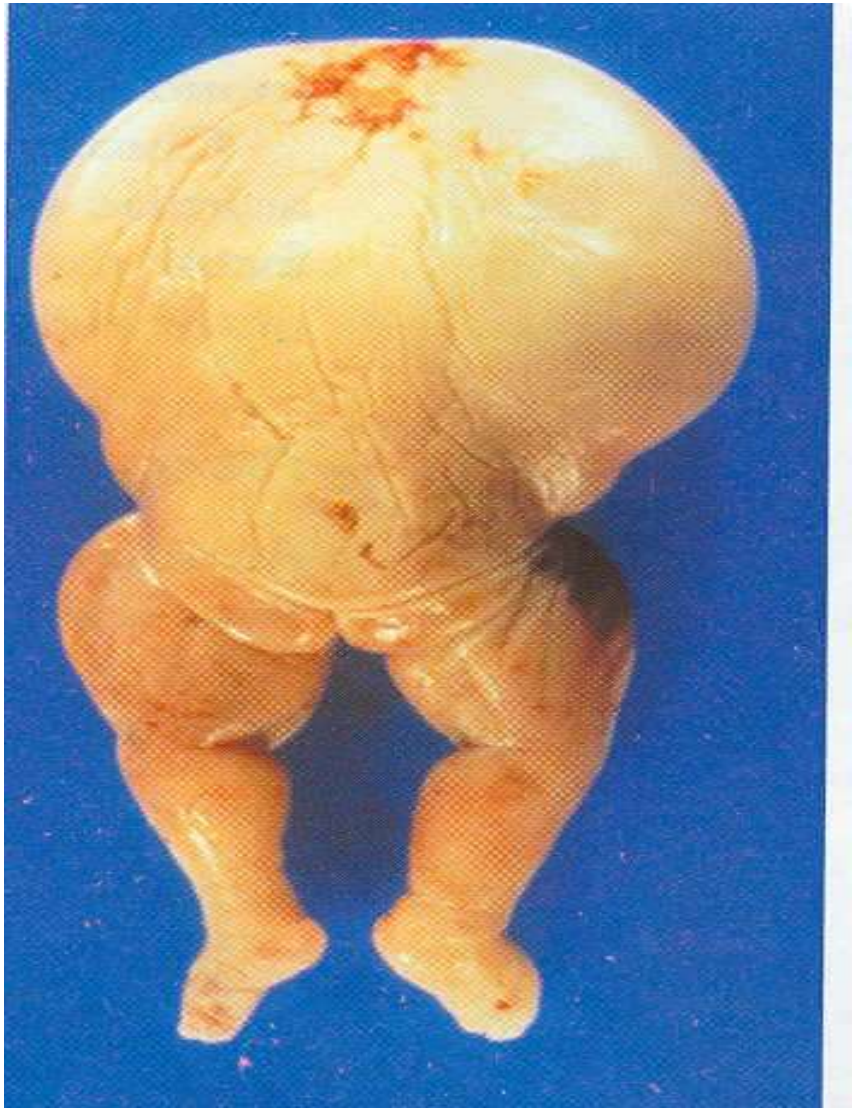
Sirenomelia

- Fusion of lower limbs
- Renal agenesis
- A hydramnios
- Lethal condition

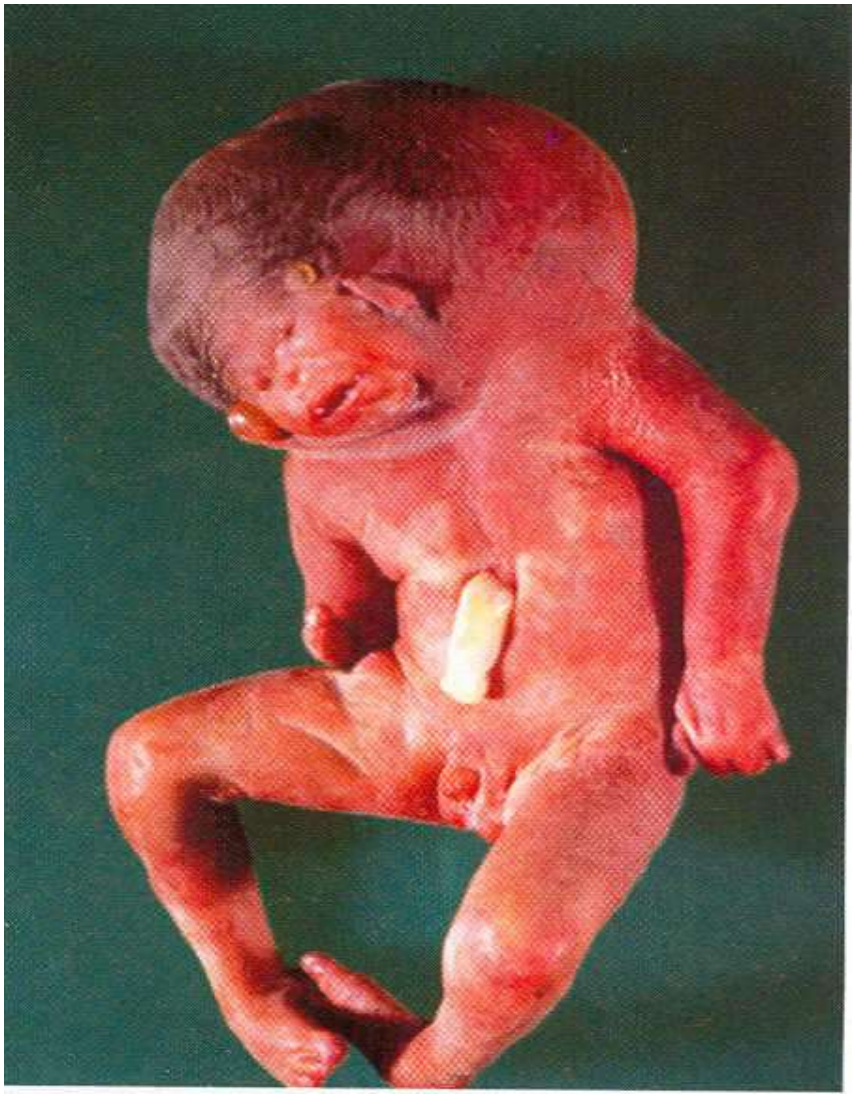


A cardiac twins

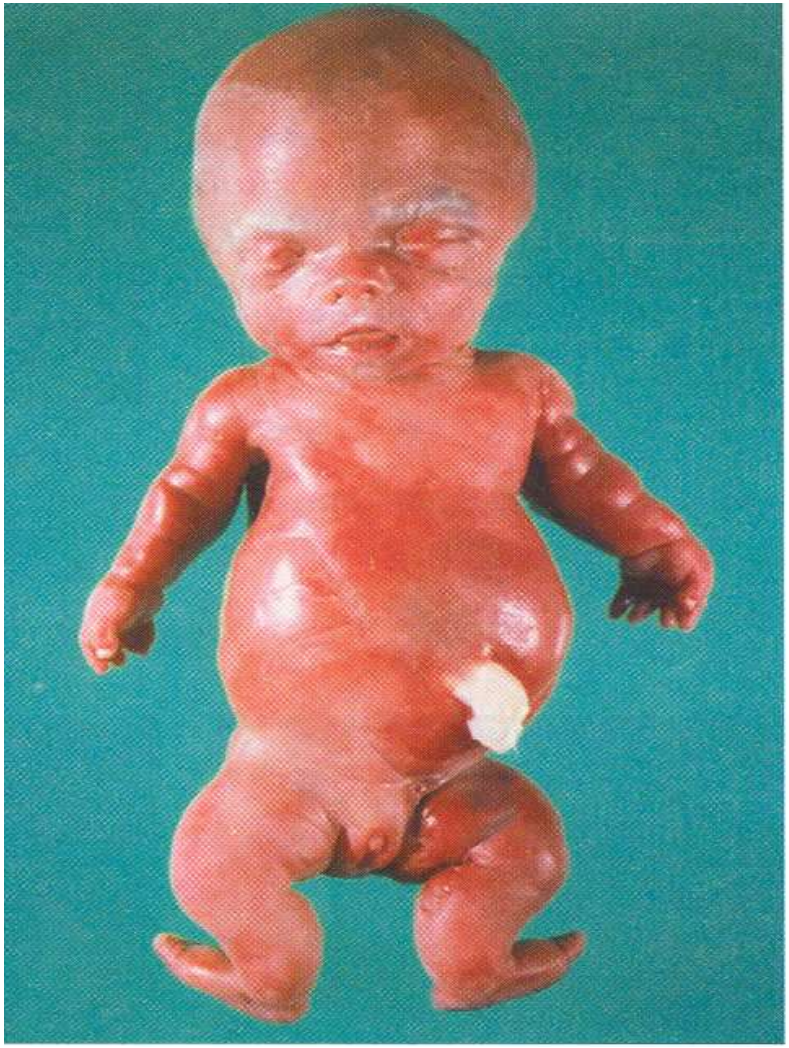
- 1 in 35,000 pregnancies
- Parasitic twin
- Mainly lower body development
- Severe deformity or non-development of upper body



A cardiac twins

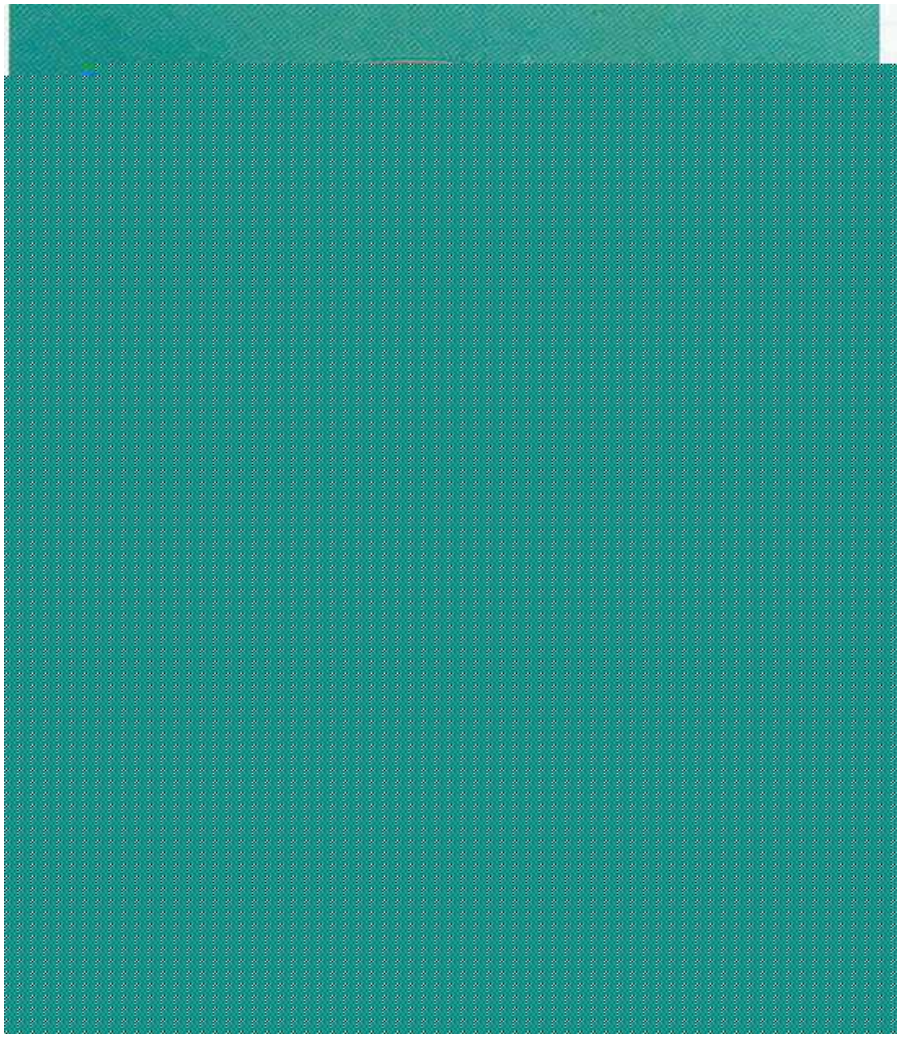


A cardiac twins



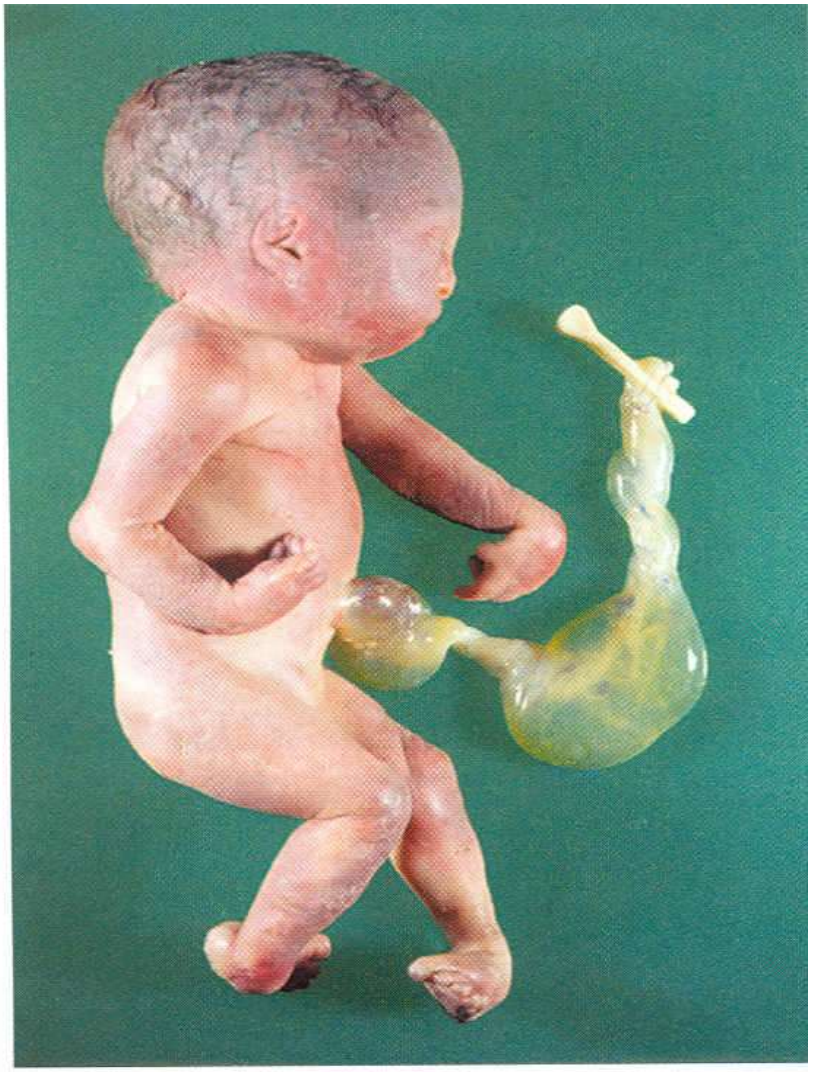
Thanatophoric dysplasia

- 0.7 in 10,000
- Severe short limbs
- Narrow thorax – polydramnios
- Lethal condition



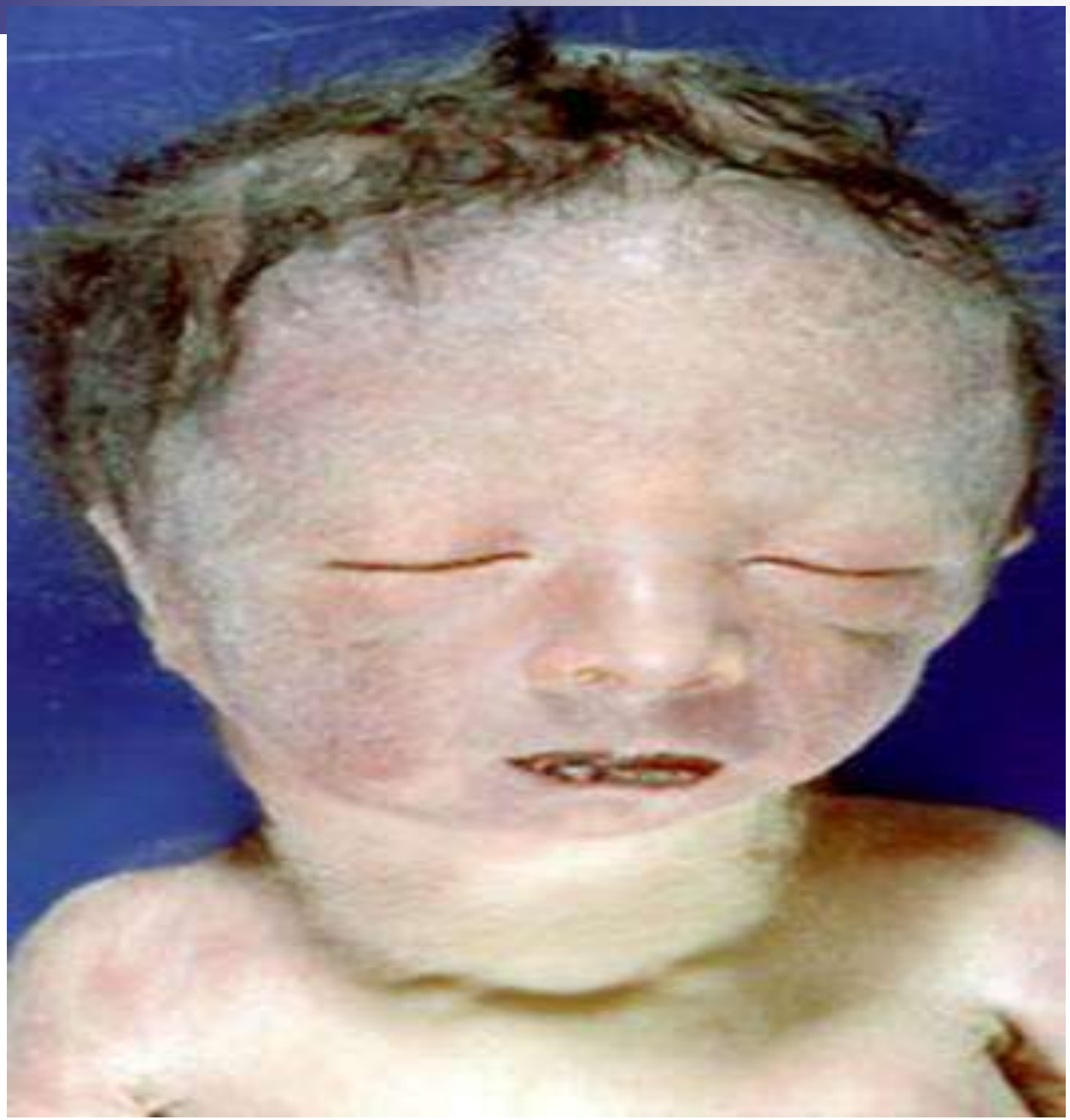
Trisomy 13

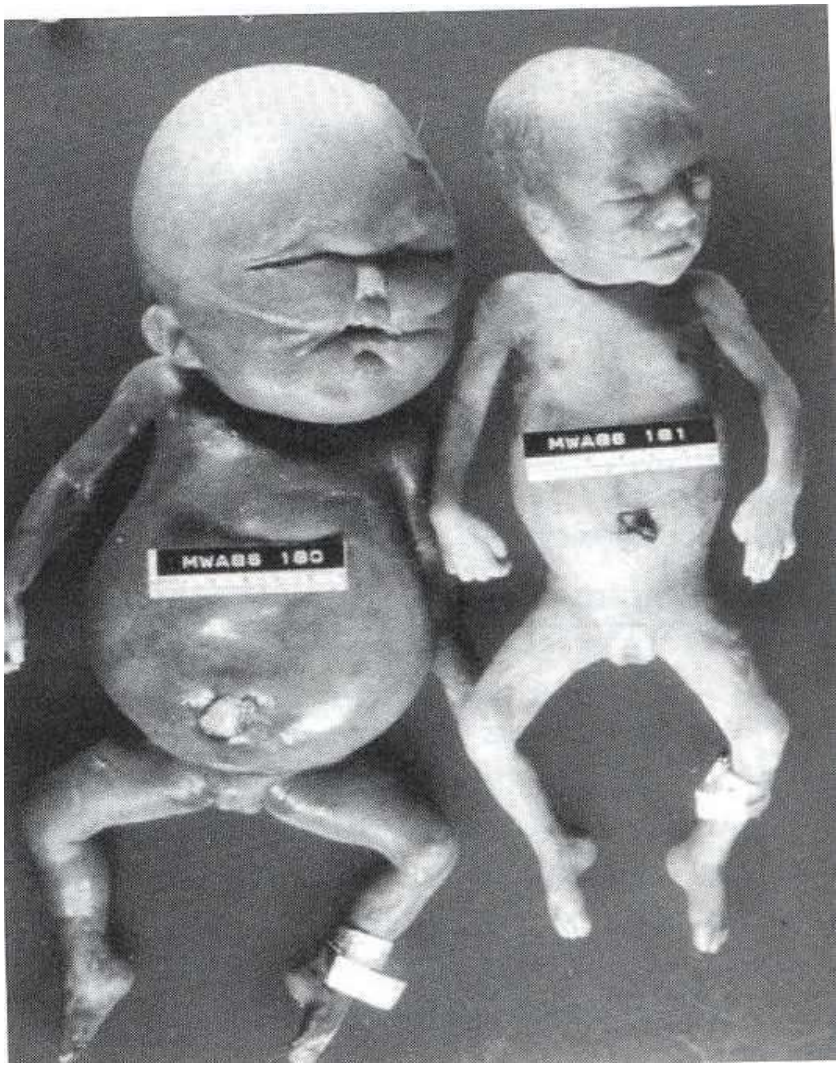
- 1 in 6,000
- Microcephaly
- Proposcis
- Cyclopia
- Holoprosencephaly
- Lethal condition



Trisomy 18

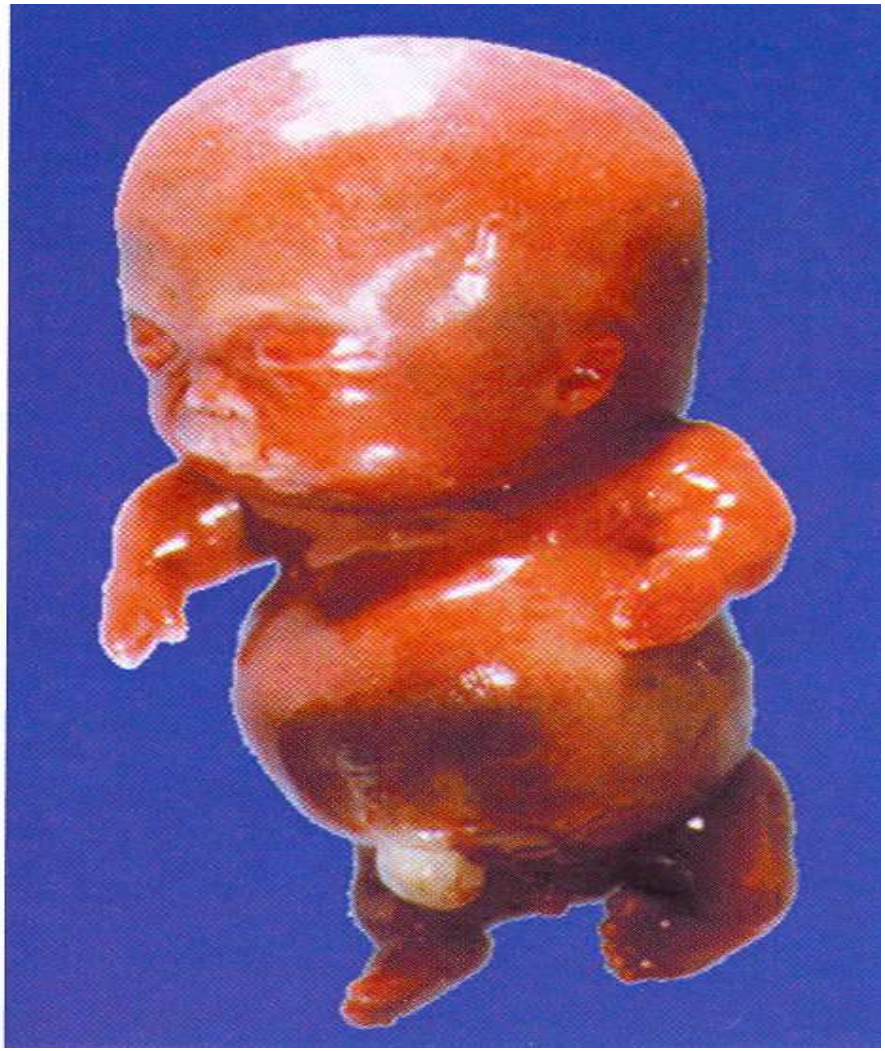
- 1 in 3,000
- Clinched hands
- Club feet
- Microcephaly
- Micrognathia
- Omphalocele
- Lethal condition





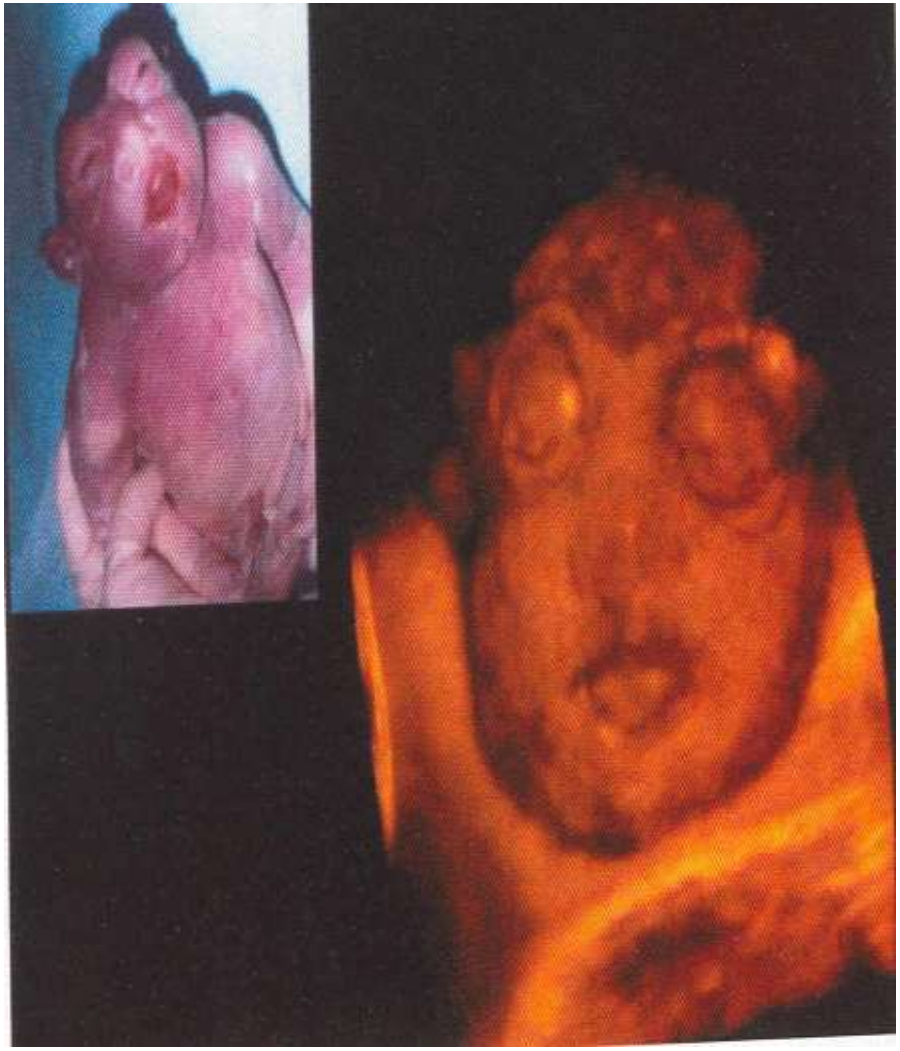
Twin-To-Twin Transfusion Syndrome

- Monochorionic twins



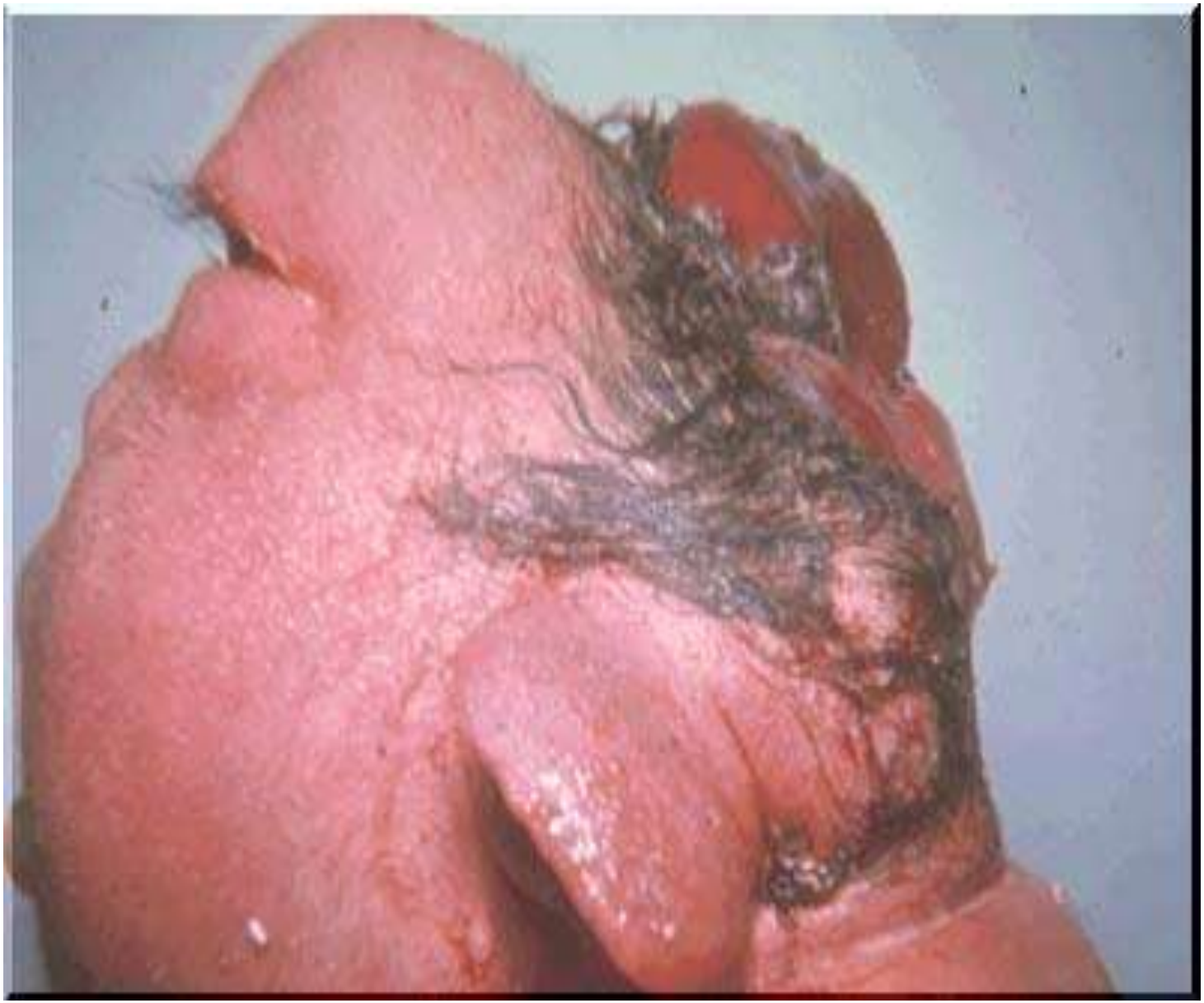
Achondrogenesis

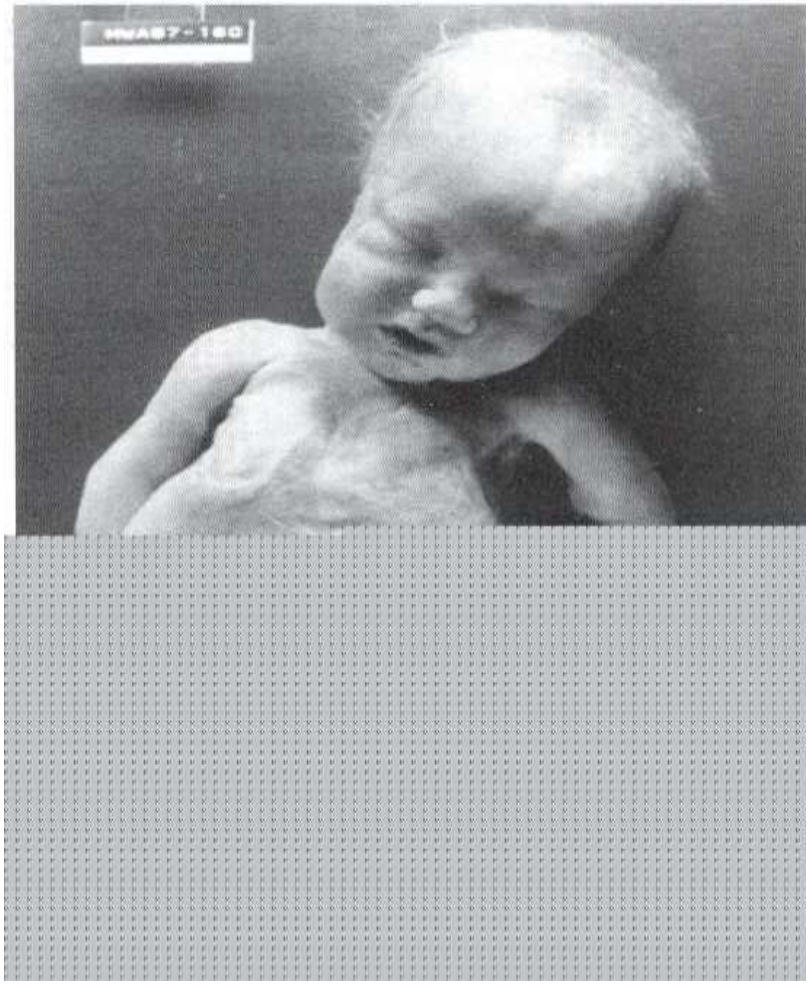
- Rare condition
- Autosomal recessive
- Very short limbs
- Lethal condition



Anencephaly

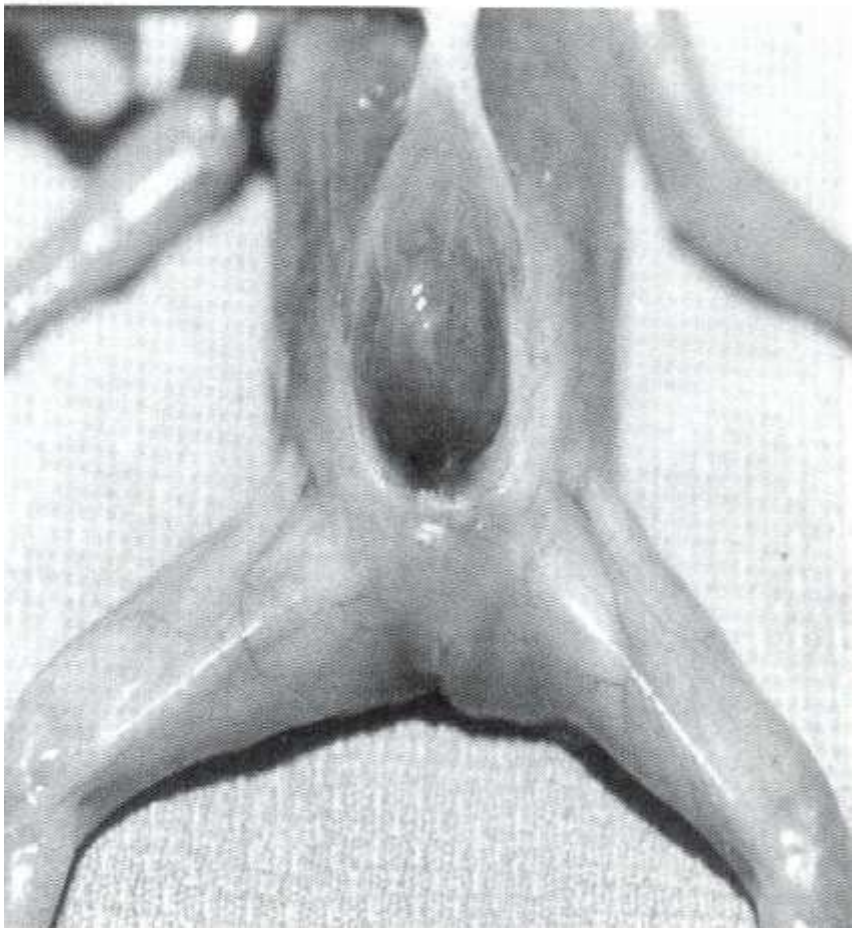
- 1 in 1,000
- Female: Male 4 to 1
- Multifactorial NTD
- Lethal condition
- Recurrence after one child is x10
- Folic acid prevent 71% of recurrences





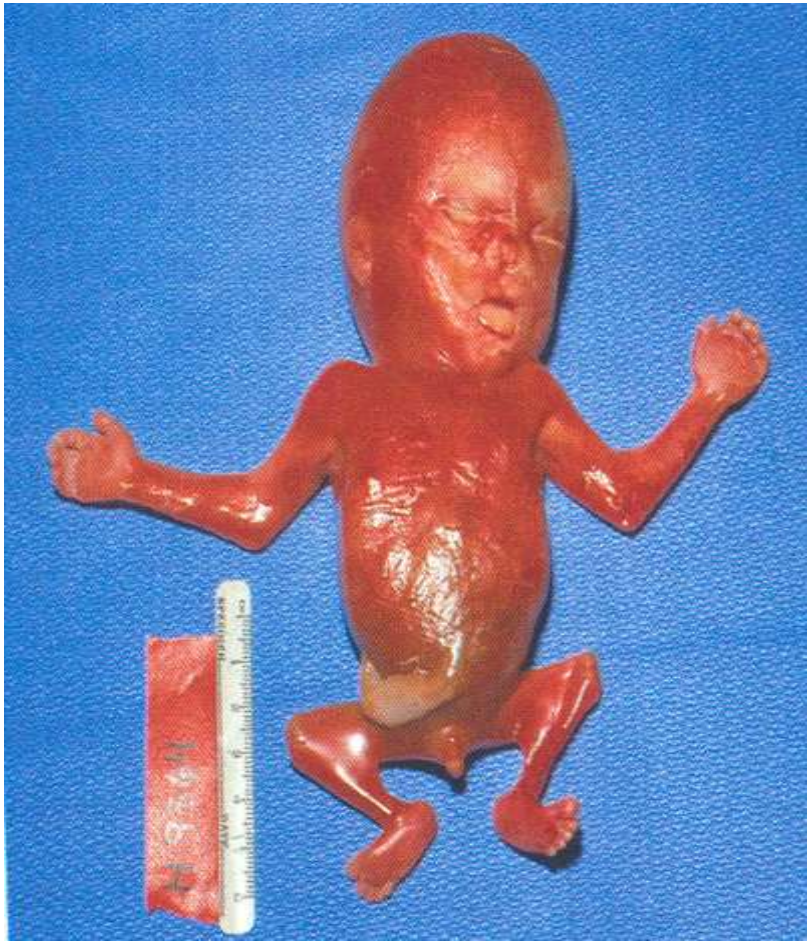
Arthrogryposis

- Wide range of disorders
- Fixed contractures of limbs
- Majority of cases lethal



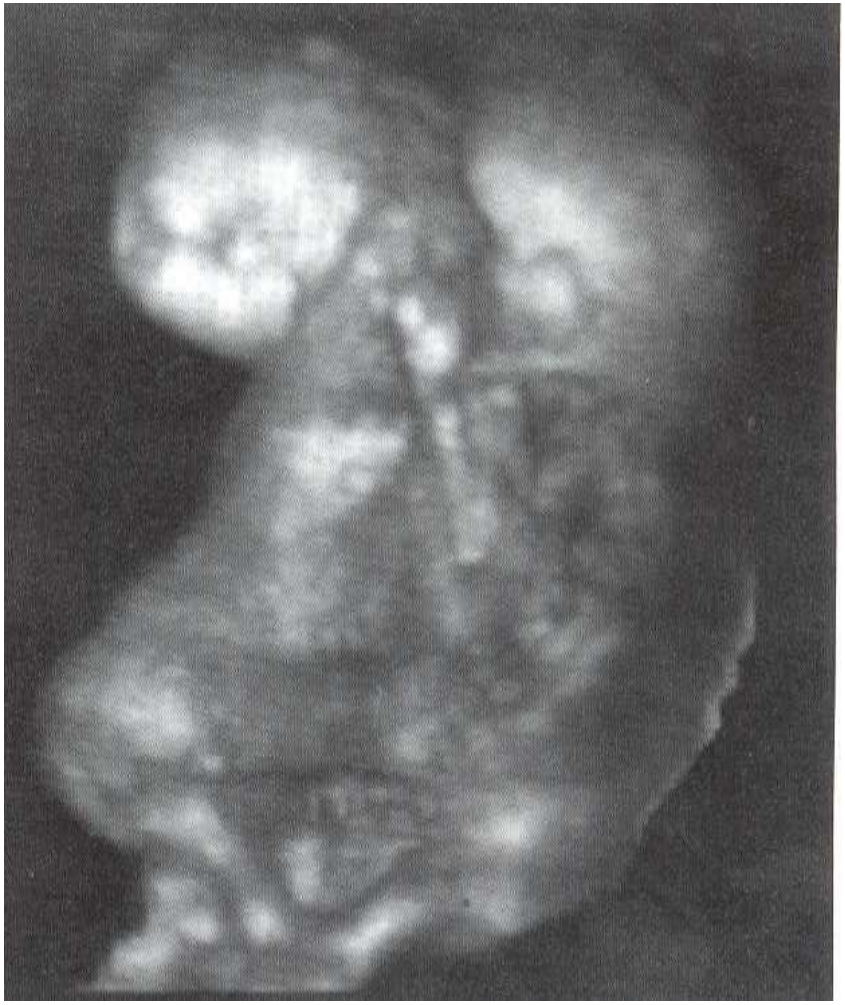
Bladder extrophy

- 1 in 30,000
- Can be corrected post natally
- May have urine and fecal incontinence
- 4% may develop bladder Ca



Sacral agenesis

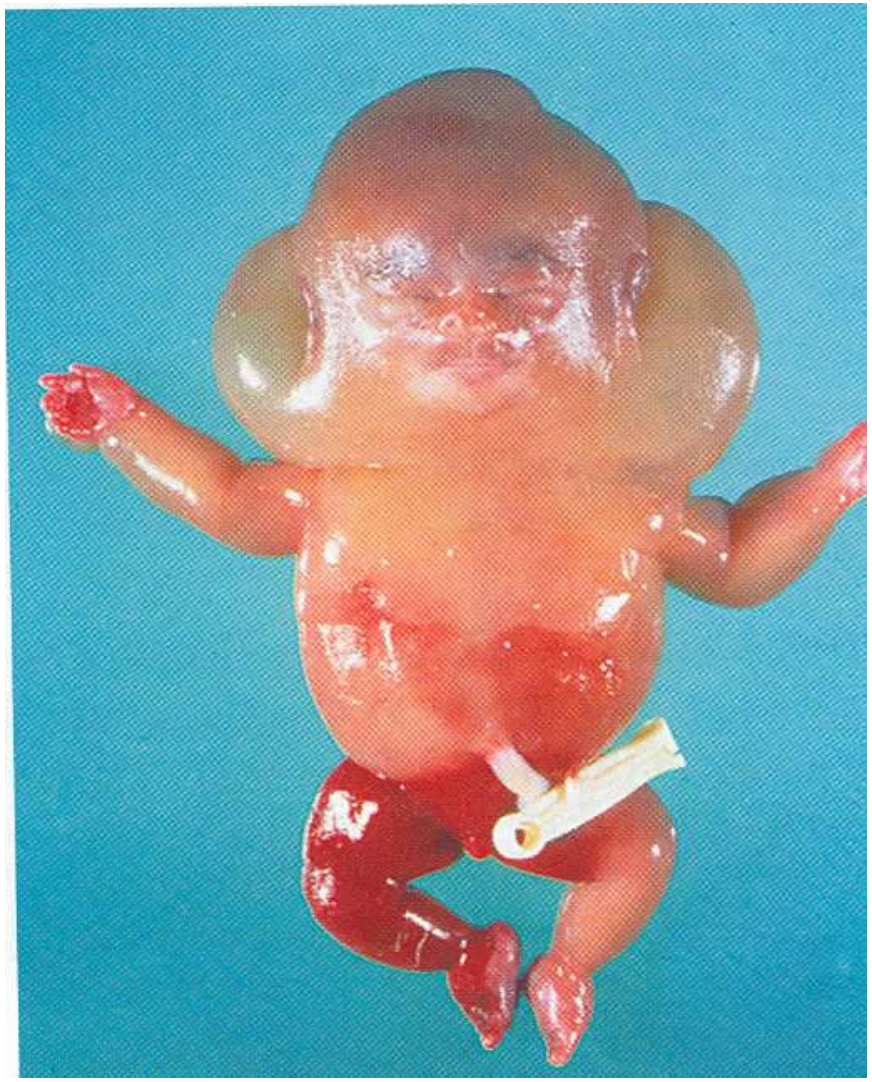
- 16% of cases seen with D.M.
- Diagnosis is made by Short Crown-ramp length
- Renal agenesis can be seen
- Neurologic impairment (bowel+bladder dysfunction)



Conjoined twins

- 1 in 50,000
- Female : Male (3:1)
- Monoamniotic monochorionic
- 1st trimester diagnosis

Conjoined twins

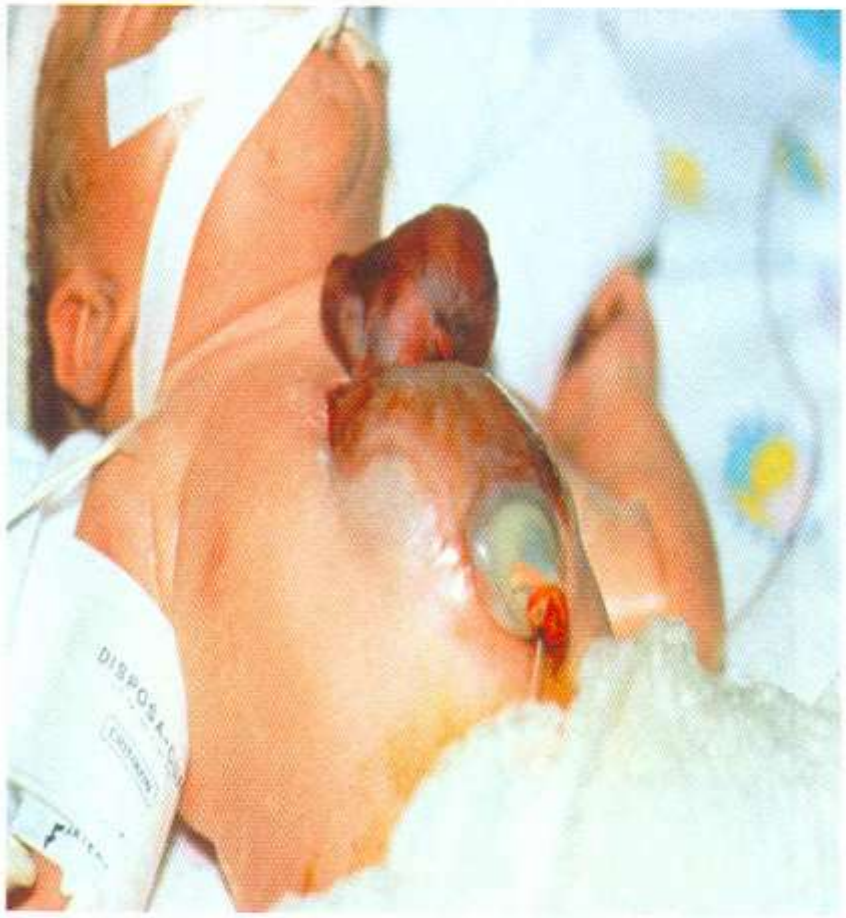


Cystic hygroma

1 in 200 abortuses

1 in 1,000 births

- Lymphatic accumulation
- 40-50% chance of 45x0 in 2nd trimester diagnosis
- Trisomies more common in 1st trimesters diagnosis



Ectopia cordis

Severe abnormality

Most cases die in few days of life