

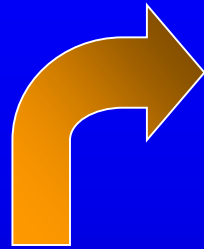
# Anomalias Congênitas: Dismorfologia

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[esramos@fmrp.usp.br](mailto:esramos@fmrp.usp.br)

**Congênito**

**≠**

**Genético**



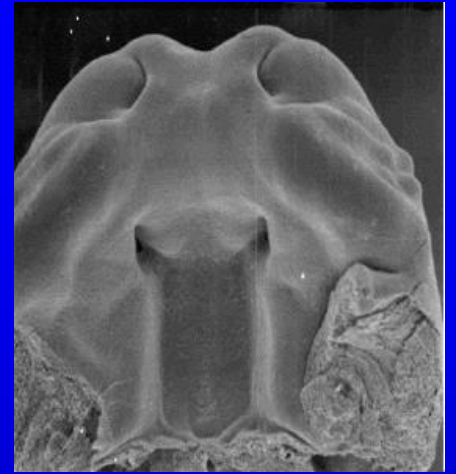
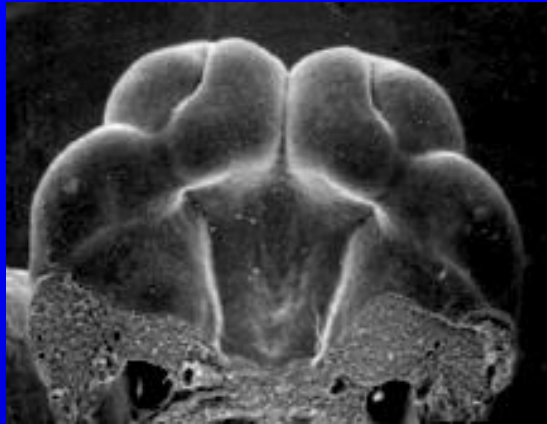
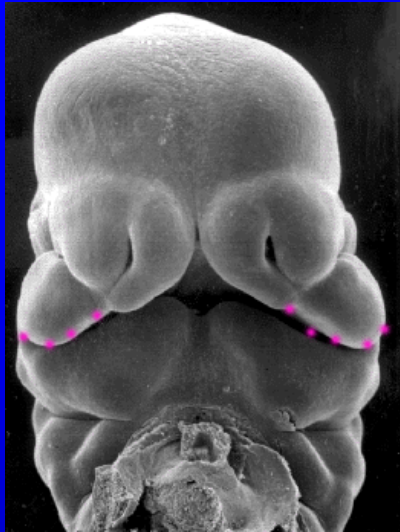
**Defeitos  
congênitos**

# 1. Morfogênese incompleta (agenesia, hipoplasia)



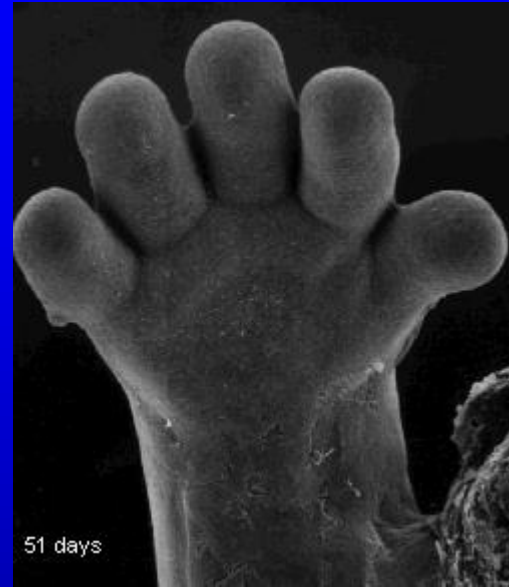
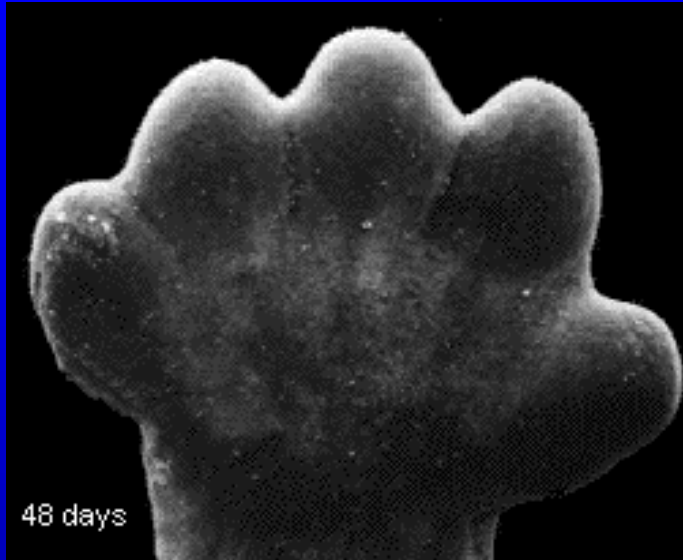
- 1. Morfogênese incompleta (agenesia, hipoplasia)**
- 2. Fechamento incompleto (fendas)**





- 1. Morfogênese incompleta (agenesia, hipoplasia)**
- 2. Fechamento incompleto (fendas)**
- 3. Separação incompleta (sindactilia)**







1. **Morfogênese incompleta (agenesia, hipoplasia)**
2. **Fechamento incompleto (fendas)**
3. **Separação incompleta (coração, sindactilia)**
4. **Excesso (polidactilia, apêndice pré-auricular)**





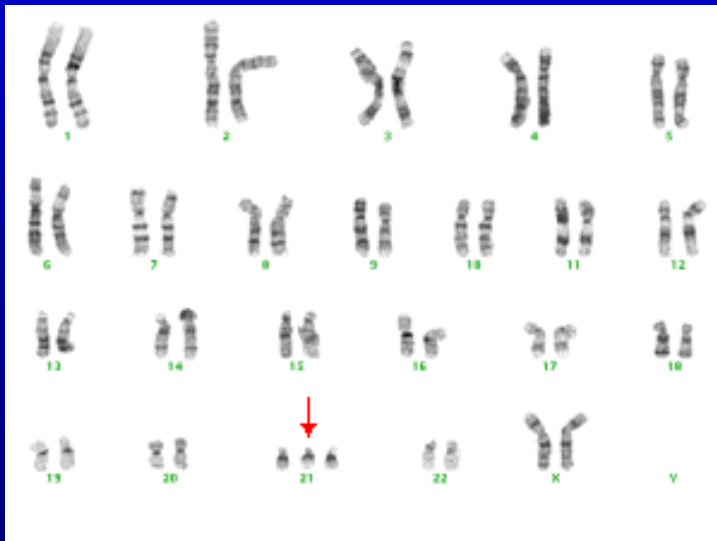


Normal



Malformação

Defeito morfológico de um órgão, parte de um órgão ou de uma área do corpo, resultante de um processo de desenvolvimento **intrinsecamente** anormal.



**Síndrome de  
Down**



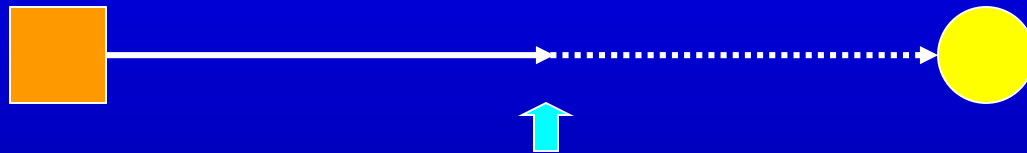
## Anemia de Fanconi (monogênica - AR)



**Normal**



**Malformação**



**Disrupção**

**Defeito devido à interferência externa com o desenvolvimento normal (necrose).**

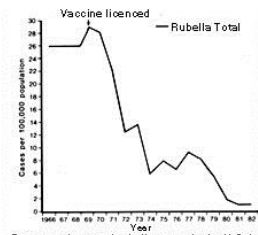
# Talidomida



# Rubéola



Child with congenital rubella syndrome (South 1966).



Decrease in rate of rubella cases in the U.S. between 1966 and 1982. A complementary reduction in congenital rubella syndrome was also observed (South 1986).

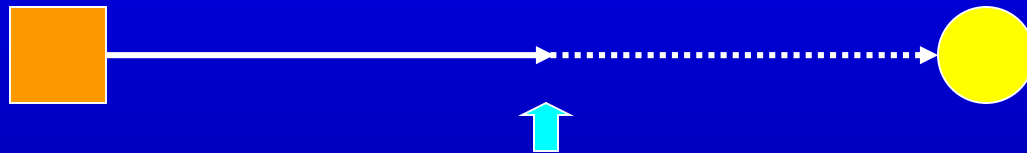




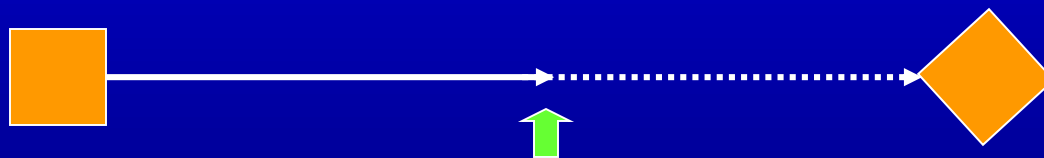
**Normal**



**Malformação**



**Disrupção**



**Deformidade**

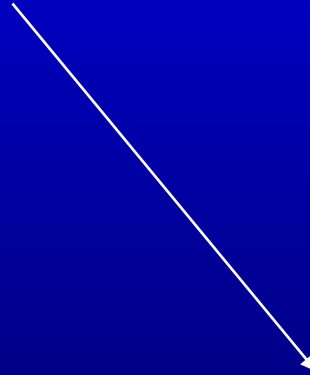
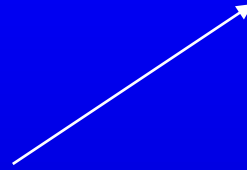
**Forma ou posição anormal de uma parte do corpo,  
causada por força mecânica.**

# Sequência de Potter

**Agenesia Renal**



**Líquido Amniótico**



**Fator mecânico**

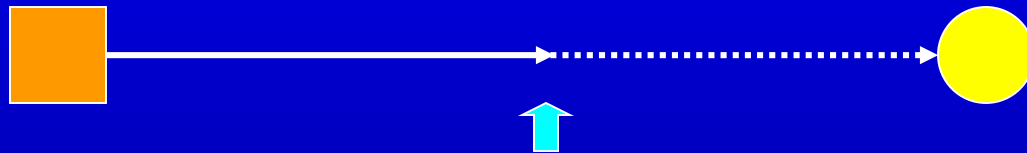




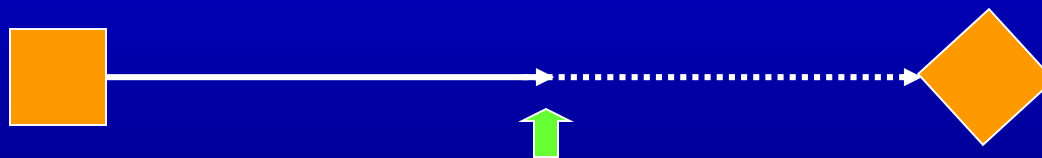
**Normal**



**Malformação**



**Disrupção**



**Deformidade**



**Displasia**

**Desorganização tecidual.**

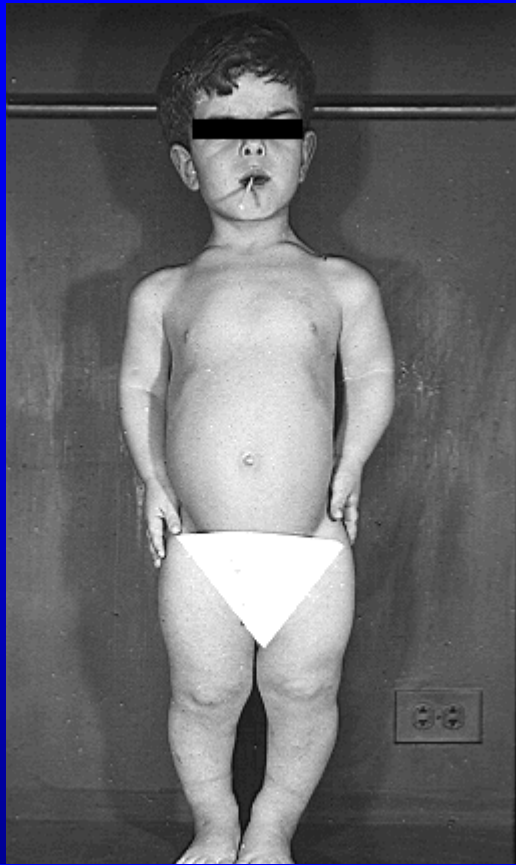
# Osteogênese Imperfeita



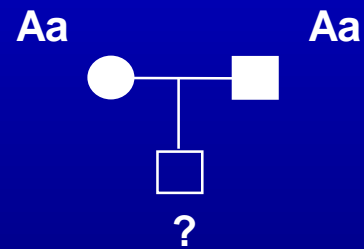
# Osteogênese Imperfeita



**Displasia óssea**



**Acondroplasia**



AA    Aa    Aa    aa

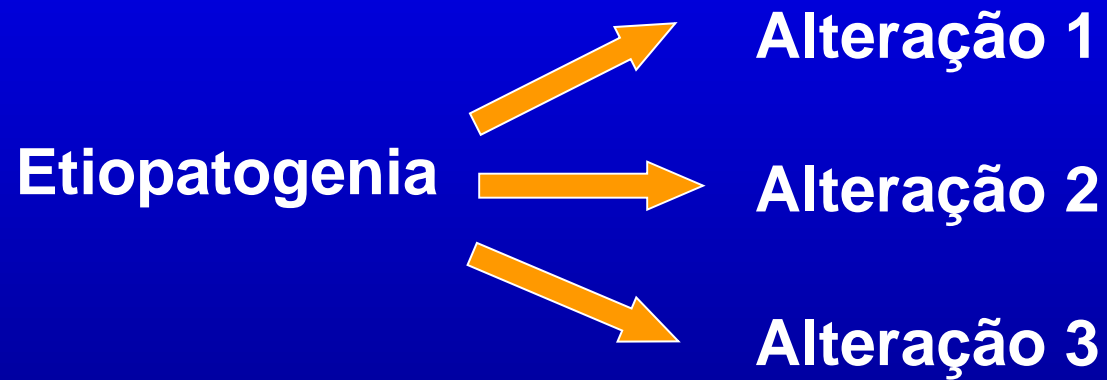


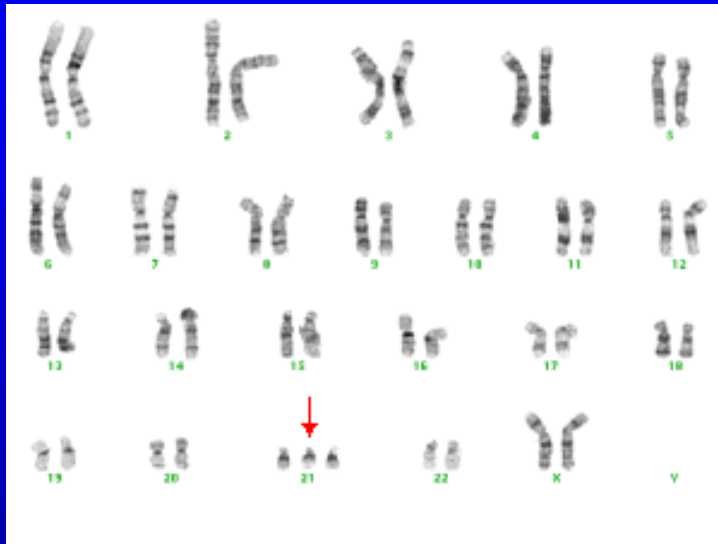


# Displasia ectodérmica



# Síndrome

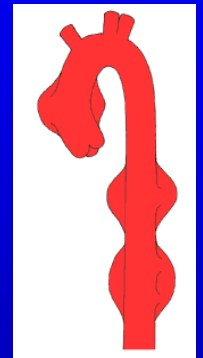


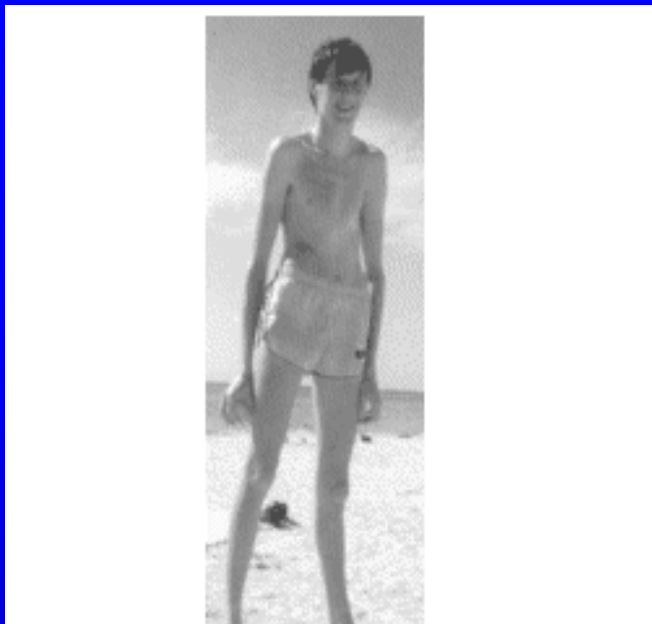


**Síndrome de Down**

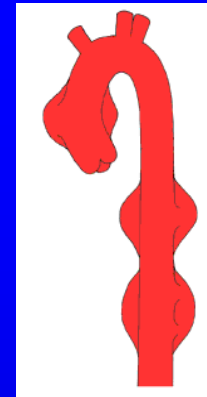
# Síndrome de Marfan

Gene →





**Alta estatura**



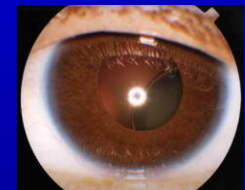
**Dilatação de aorta**



**Aracnodactilia**



**Frouxidão ligamentar**



**Miopia  
Luxação do  
cristalino**

# Síndrome Alcoólica Fetal

Álcool →



[medgen.genetics.utah.edu](http://medgen.genetics.utah.edu)

Print Clear Selected  Include Synonyms  Group by Family Help

- 3A syndrome<sup>2</sup>
- 3C syndrome<sup>2</sup>
- 3M syndrome\*
- 3-hydroxyisobutyric aciduria
- 3-Methylglutaconic aciduria type I
- 3-methylglutaconic aciduria type II<sup>2</sup>
- 3-methylglutaconic aciduria, severe or type III
- 5-oxoprolinuria<sup>2</sup>
- Aagaes - recurrent cholestasis; lymphoedema
- Aarskog syndrome
- Aarskog-Scott syndrome<sup>2</sup>
- Aase - triphalangeal thumb; congenital anaemia
- Aase-Smith - hydrocephalus; cleft palate; joint contractures
- Abbassioun (1986) - acromegaly; pituitary tumours
- Abboud (1985) - diabetes; ketoglutarate dehydrogenase deficiency; deafness
- ABCD syndrome<sup>2</sup>
- Abdallat (1980) - neurocutaneous syndrome
- Abetalipoproteinaemia (Dassen-Kornzweig syndrome)
- Ablepharon-ichthyosis
- Ablepharon-macrostomia
- Abruzzo (1977) - cleft palate; coloboma; radial synostosis

Go to:  Syndrome Details Keyword Search Search on Features

### Search Syndromes on Features

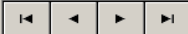
Criterion 1  Mandatory

Criterion 2  Mandatory

Criterion 3  Mandatory

Criterion 4  Mandatory

- BUILD
- STATURE
- CRANIUM
- HAIR
- FOREHEAD
- EARS
- EYES, GLOBES
  - Eyes, general abnormalities (including spacing)
    - Asymmetric eyes
    - Cyclopia
    - Deep-set eyes
    - Dystopia canthorum (telecanthus)
    - Hypertelorism
    - Hypotelorism
    - Prominent eyes/proptosis
  - Anterior chamber, general abnormalities
  - Conjunctiva, general abnormalities
  - Cornea, general abnormalities
  - Globes, general abnormalities
  - Iris, general abnormalities
  - Lens, general abnormalities
  - Macula, general abnormalities
  - Optic disc and nerve, general abnormalities
  - Pupil, general abnormalities



### Marfan syndrome

Location: 15q21

McKusick: 154700

Synonyms:

Inheritance:

MFS\*

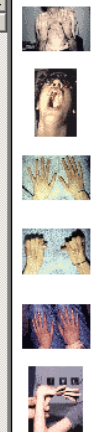
Autosomal dominant

#### Abstract

6 Photos

Patients with this well known syndrome have a combination of dolichostenomelia, arachnodactyly, pectus deformities of the chest, mitral or aortic regurgitation, ectopia lentis and mild joint laxity. Other evidence of a generalised connective tissue disorder may be present such as scoliosis and skin striae. A dilated aortic root can usually be demonstrated by echocardiography and aortic aneurysms can ensue. De Paepe et al., (1996) discuss the diagnostic criteria. Average life expectancy is halved. 95% of deaths are due to a cardiovascular cause. Shores et al., (1994) studied the effect of beta-adrenergic blockade and concluded that this slowed the rate of aortic dilatation and reduced the development of complications from aortic rupture in some patients with Marfan syndrome. There have been about 10 case reports (van den Berg et al., 1996) of cerebral aneurysms in Marfan syndrome, but the association is doubted by some (van den Berg et al., 1996).

The condition has been shown to be caused by mutations of the fibrillin gene on chromosome 15 (see Tsipouras et al., (1992) for review). Gray et al., (1994) estimated a prevalence of 1 in 14,000 in Scotland. Twenty-seven percent of cases appeared to be new mutations. Most mutations are unique to individual families. Intragenic markers can be used for predictive testing (Rantamaki et al., 1994; Pereira et al., 1994), however care must be taken because of possible genetic heterogeneity (for example see Boileau et al., 1993). Diagnosis by assessment of fibrillin immunofluorescence on skin biopsies or fibroblast cultures is still technically difficult and the accuracy is not certain (Schaefer and Godfrey, 1995). Dietz and Pyeritz (1995) provide a good review of mutations in the fibrillin gene in Marfan syndrome.



Abstract Features References

All Syndromes Selected Syndromes All References Selected References





### Marfan syndrome

Location: 15q21

McKusick: 154700

Synonyms:

Inheritance:

MFS*
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Autosomal dominant
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### Features

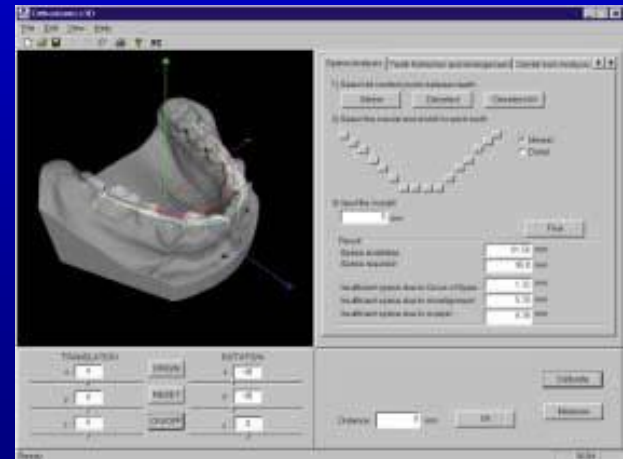
6 Photos

- Tall stature, disproportionate/dolichostenomelia
- Dolichocephaly/scaphocephaly
- Dislocation of lens
- Blue sclera
- Myopia
- Thin/long face
- Prominent mandible
- High palate
- Scoliosis
- Pectus carinatum
- Pectus excavatum
- Aortic incompetence
- Mitral incompetence
- Inguinal hernia
- Hyperextensibility at elbow
- Hyperextensibility at wrist
- Arachnodactyly
- Hyper-mobile/extensible fingers
- Hyper-extensible knees
- Flat arches of feet
- Long toes
- Dissection of the aorta
- Joint laxity
- Deficient adipose tissue or fat/lipodystrophy
- Skin striae/stretch marks



Abstract Features References

All Syndromes Selected Syndromes All References Selected References

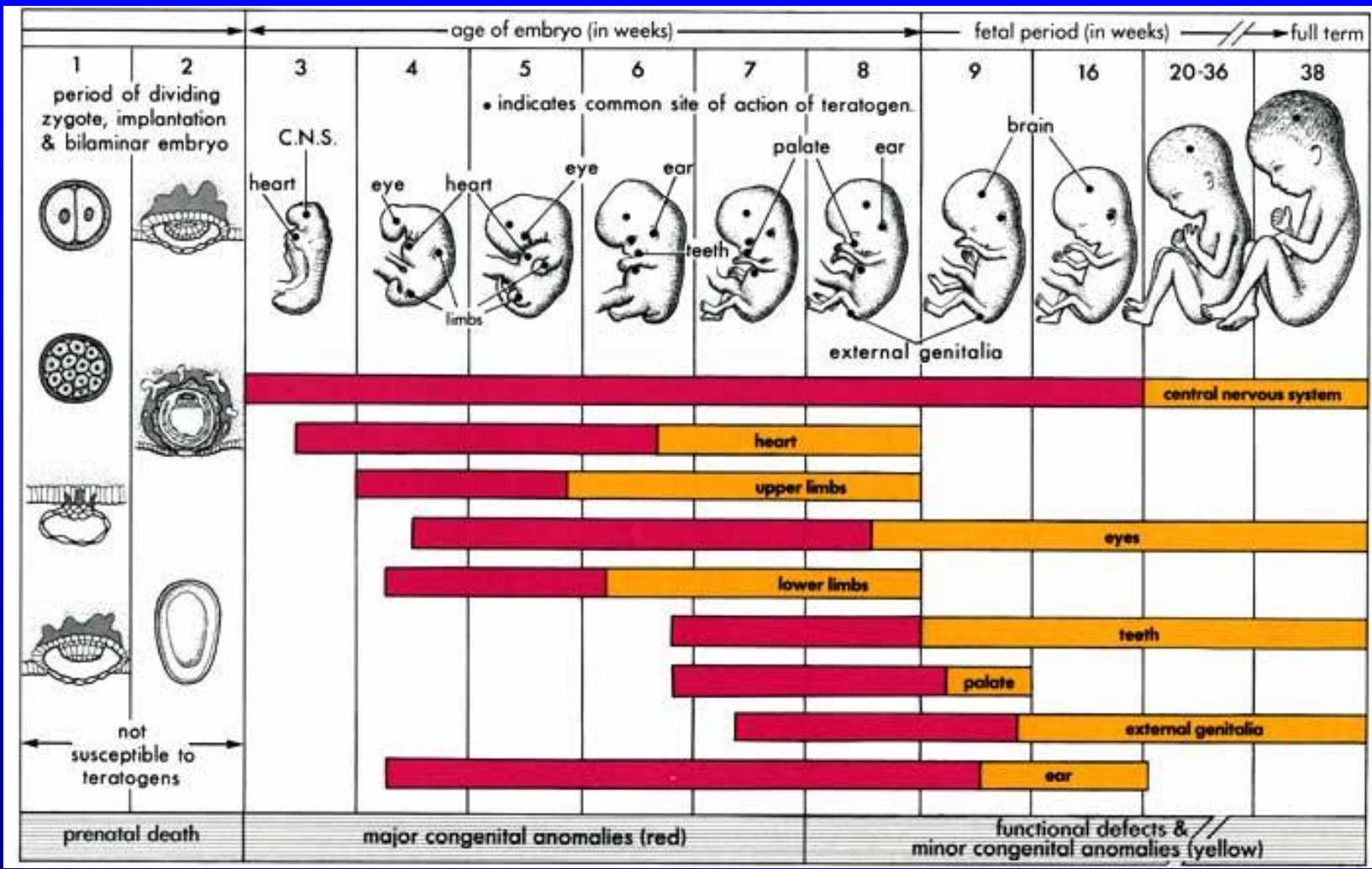




Ambientais  
(teratógenos)



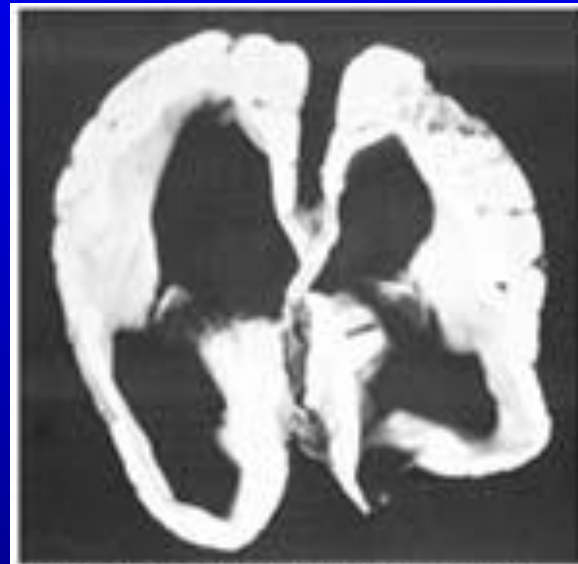
**Defeito devido à interferência externa com o desenvolvimento normal (necrose).**



# AMBIENTAIS

- AGENTES INFECCIOSOS :  
Toxoplasmosis, Sífilis, Citomegalovirus,  
Rubéola

# Ambientais



Infant's brain damaged due to toxoplasmosis induced hydrocephalus (Larsen, 1986)

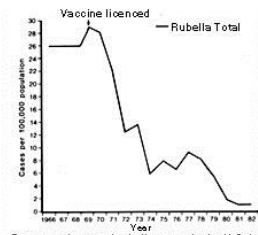
## Toxoplasmose

# AMBIENTAIS

## Rubéola



Child with congenital rubella syndrome (South 1986).



Decrease in rate of rubella cases in the U.S. between 1966 and 1982. A complementary reduction in congenital rubella syndrome was also observed (South 1986).



# AMBIENTAIS

- FATORES MATERNOS :  
Hipertermia (febre, sauna),  
Diabetes



**Regressão caudal  
(diabetes)**

# AMBIENTAIS

- AGENTES FÍSICO-QUÍMICOS :  
Radiação, Talidomida, Hidantoína,  
Aminoptertina (MTX), Misoprostol  
(Cytotec), Álcool



# AMBIENTAIS

**Talidomida**



# Ambientais



Aminopterina (MTX)

# Ambientais



**Hidantoína**

# Ambientais



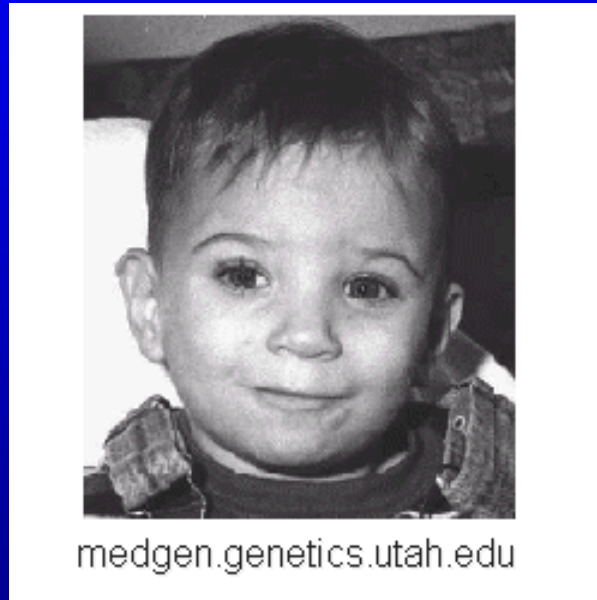
**Warfarin**

# Ambientais



**Misoprostol (Cytotec)**

# Ambientais

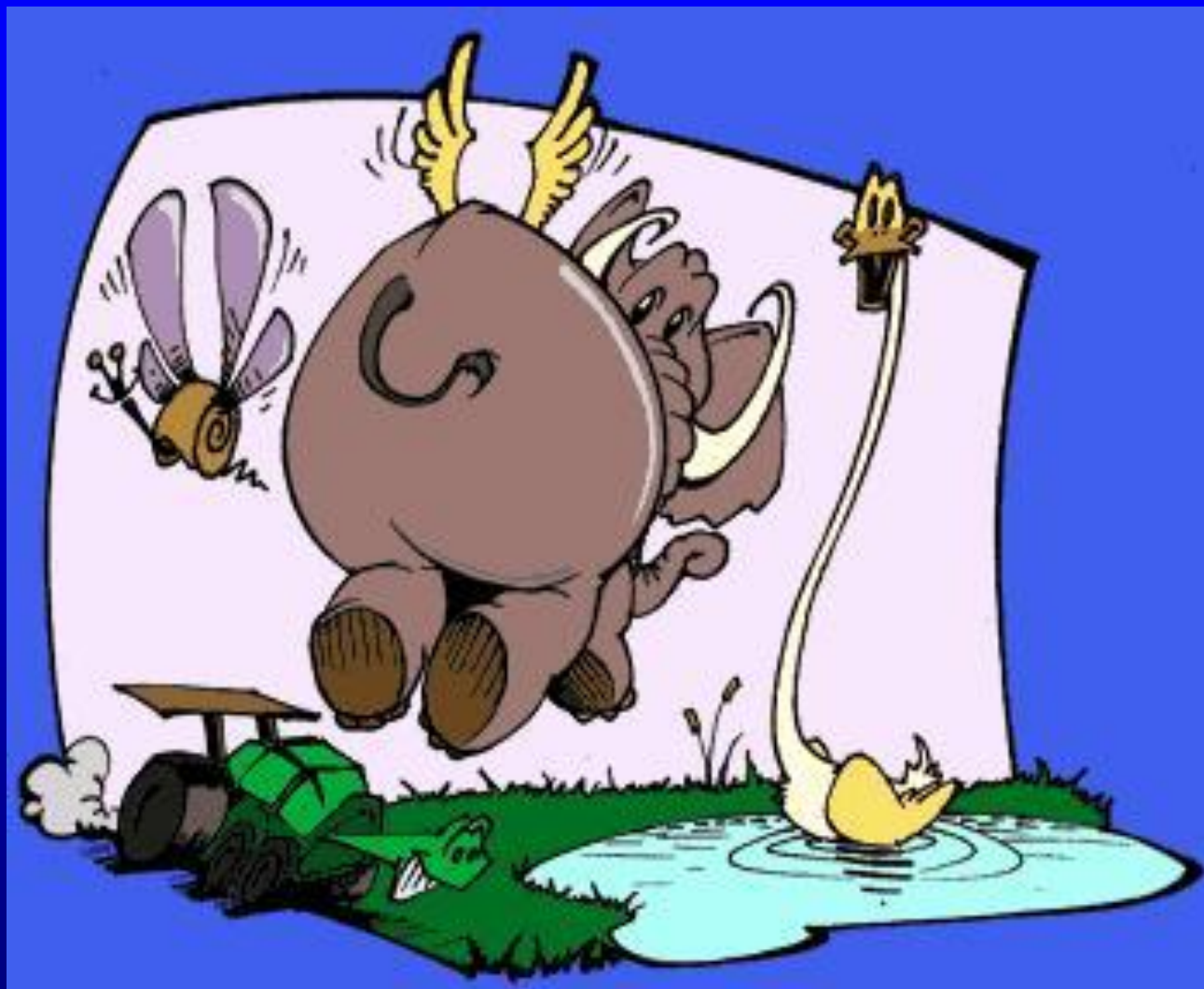


**Síndrome Alcoólica Fetal**



# SAF





**Alterações “maiores”**

**Alterações “menores”**

**3 ou + menores, procurar maior**

# Sinofre



# Epicanto



# Thompson e Thompson – Genética Médica

1. Aspectos Genéticos do Desenvolvimento  
(Genética do Desenvolvimento na Prática Clínica)