

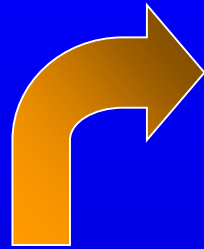
Anomalias Congênitas: Dismorfologia

Profa. Dra. Ester Silveira Ramos
esramos@fmrp.usp.br

Congênito

≠

Genético



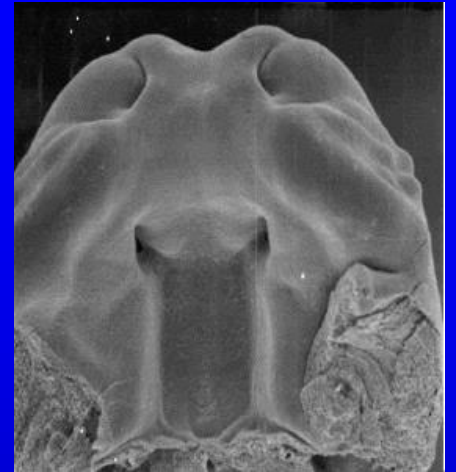
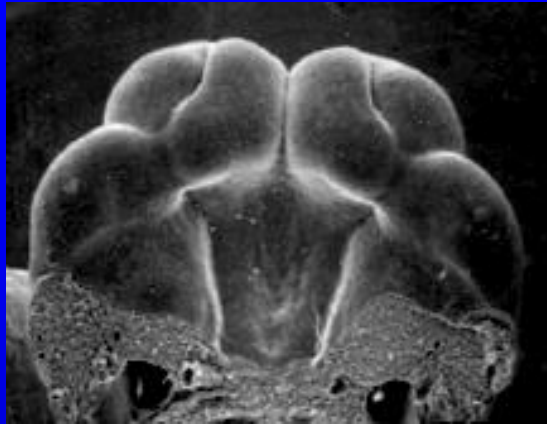
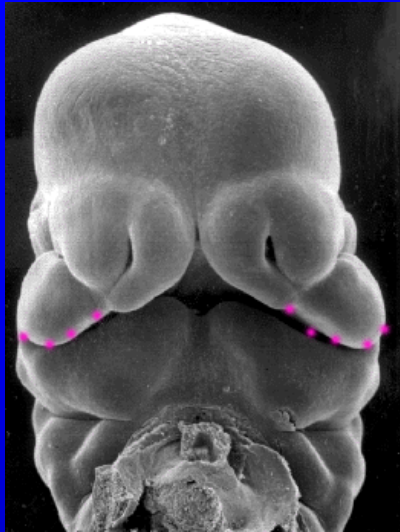
**Defeitos
congênitos**

1. Morfogênese incompleta (agenesia, hipoplasia)



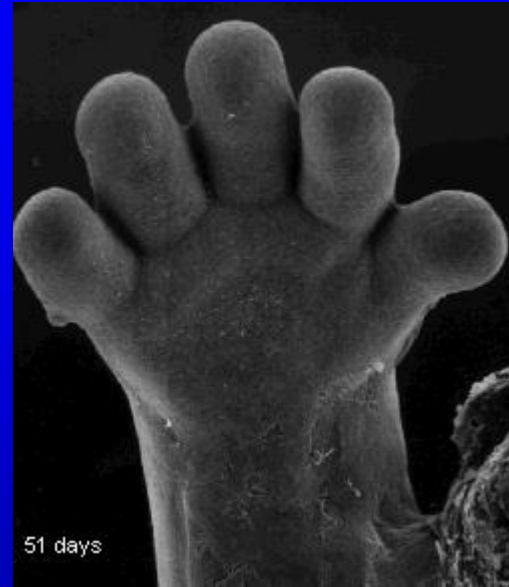
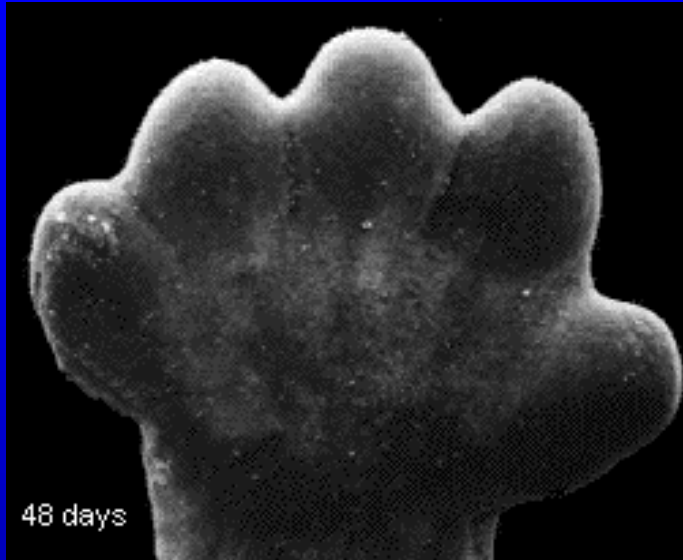
- 1. Morfogênese incompleta (agenesia, hipoplasia)**
- 2. Fechamento incompleto (fendas)**





- 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)**





19/6

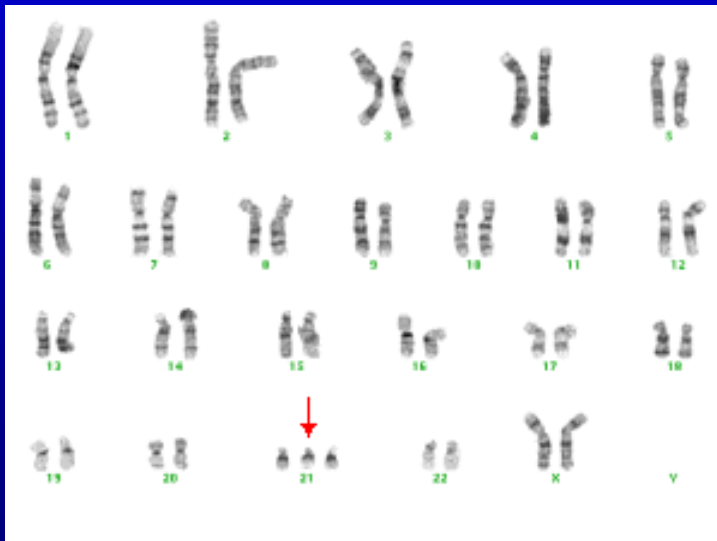


Normal



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**



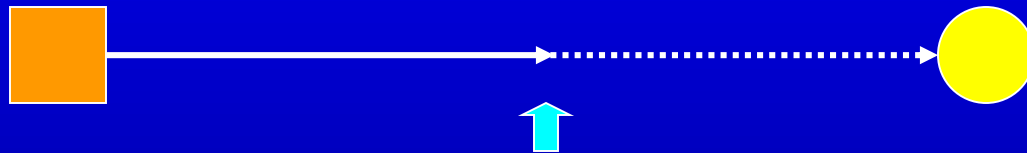
Anemia de Fanconi (monogênica - AR)



Normal



Malformação



Disrupção

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

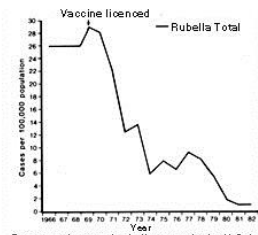
Talidomida



Rubéola



Child with congenital rubella syndrome (South 1966).



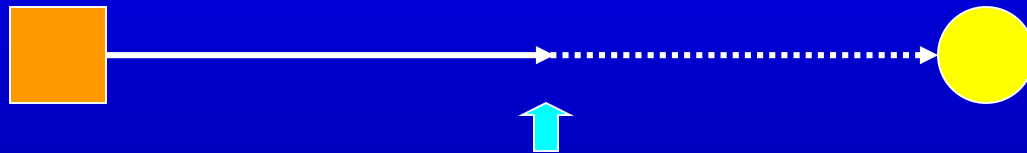
Decrease in rate of rubella cases in the U.S. between 1966 and 1982. A complementary reduction in congenital rubella syndrome was also observed (South 1986).



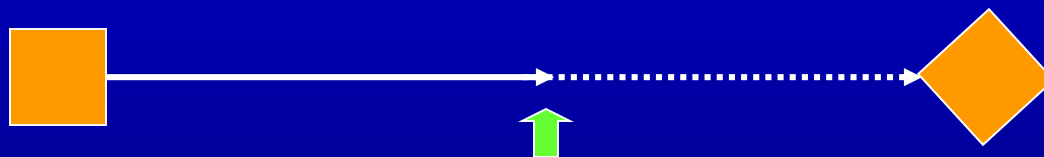
Normal



Malformação



Disrupção



Deformidade

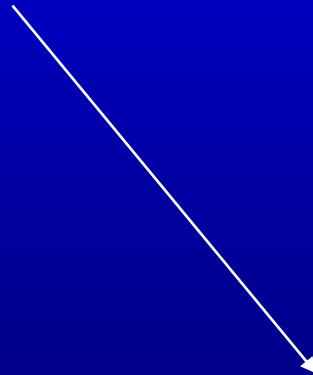
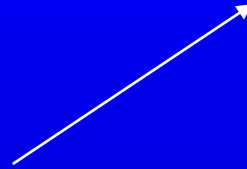
**Forma ou posição anormal de uma parte do corpo,
causada por força mecânica.**

Sequência de Potter

Agenesia Renal



Líquido Amniótico



Fator mecânico

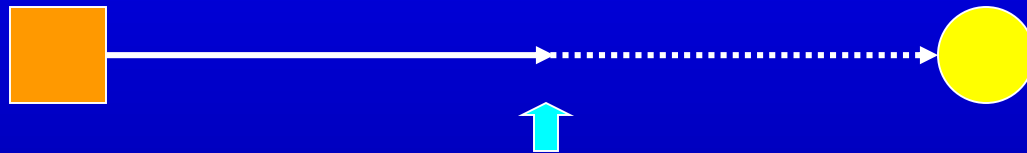




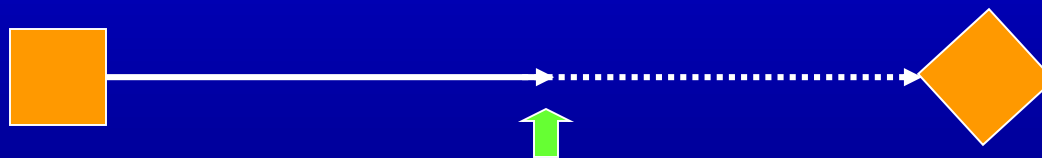
Normal



Malformação



Disrupção



Deformidade



Displasia

Desorganização tecidual.

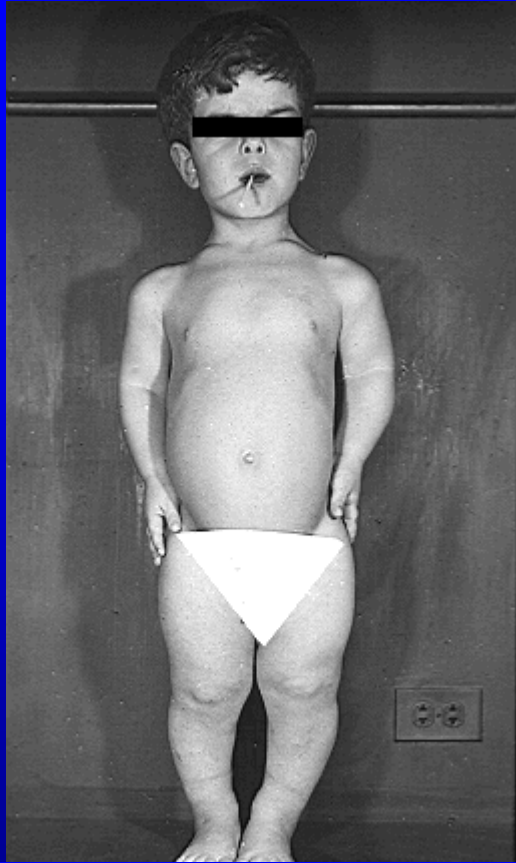
Osteogênese Imperfeita



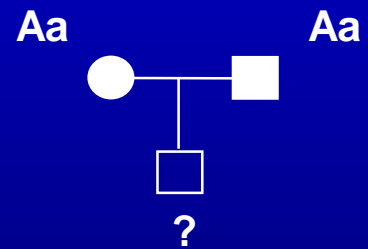
Osteogênese Imperfeita



Displasia óssea



Acondroplasia



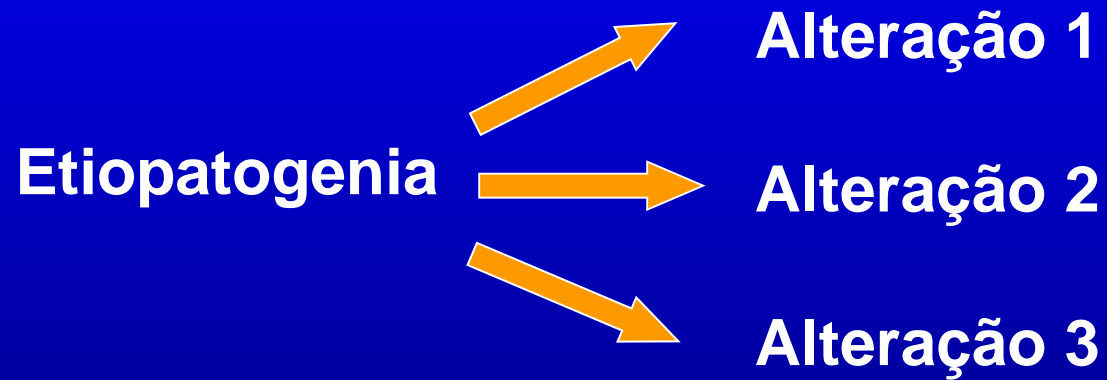
AA Aa Aa aa

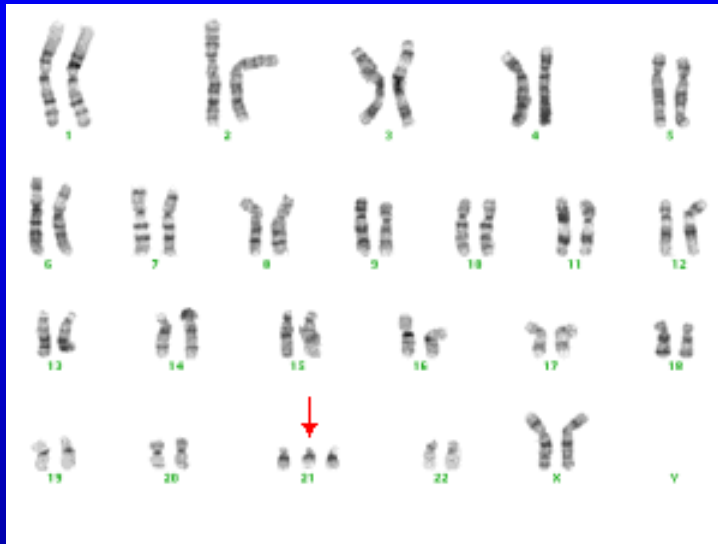




Displasia ectodérmica

Síndrome

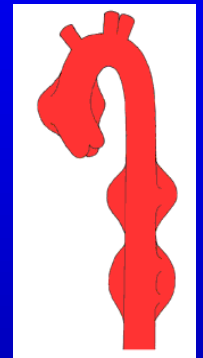


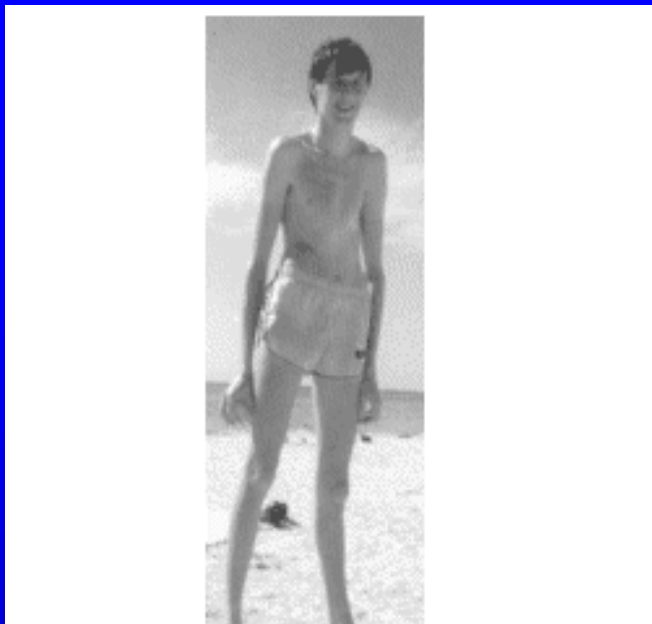


Síndrome de Down

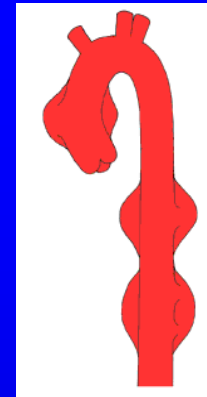
Síndrome de Marfan

Gene →





Alta estatura



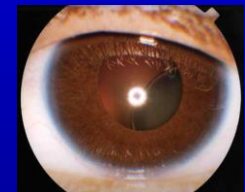
Dilatação de aorta



Aracnodactilia



Frouxidão ligamentar



**Miopia
Luxação do
cristalino**

Síndrome Alcoólica Fetal

Álcool →



medgen.genetics.utah.edu

Print Clear Selected Include Synonyms Group by Family Help

- 3A syndrome²
- 3C syndrome²
- 3M syndrome*
- 3-hydroxyisobutyric aciduria
- 3-Methylglutaconic aciduria type I
- 3-methylglutaconic aciduria type II²
- 3-methylglutaconic aciduria, severe or type III
- 5-oxoprolinuria²
- Aagenaes - recurrent cholestasis; lymphoedema
- Aarskog syndrome
- Aarskog-Scott syndrome²
- 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 syndrome²
- 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

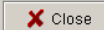
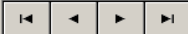
Criterion 1 Mandatory

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*

Autosomal dominant

Features

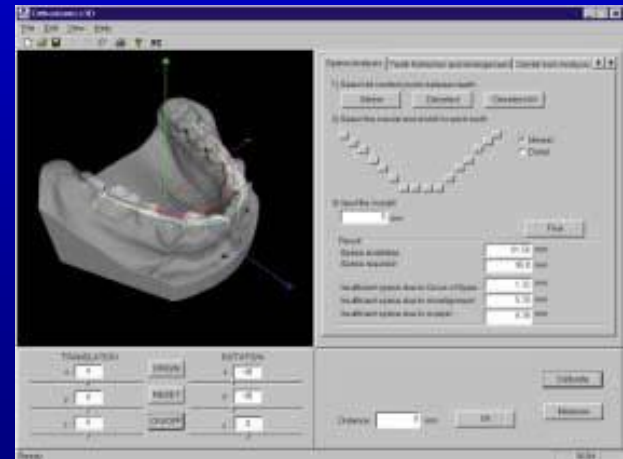
6 Photos


- 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

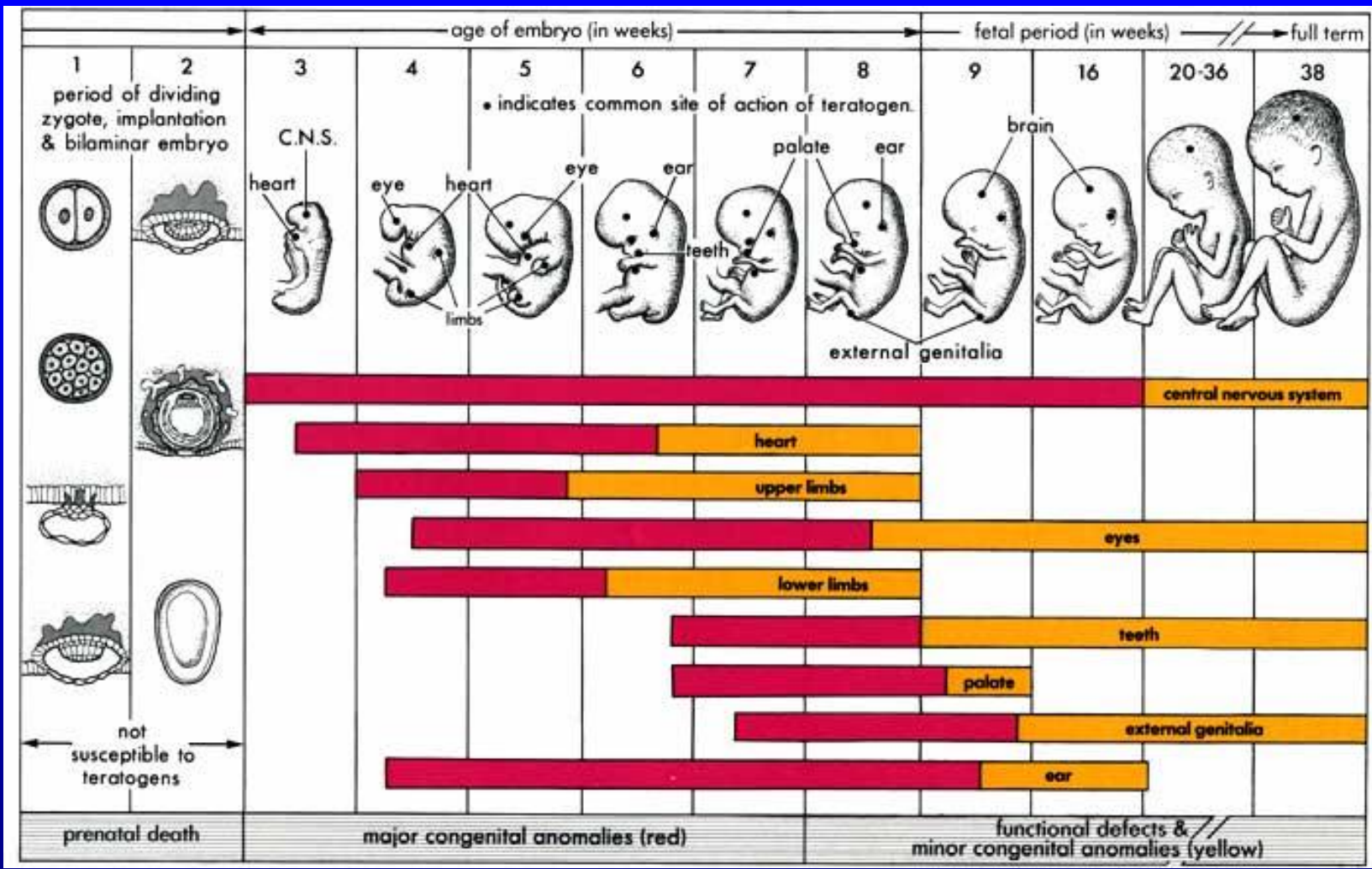




Ambientais
(teratógenos)



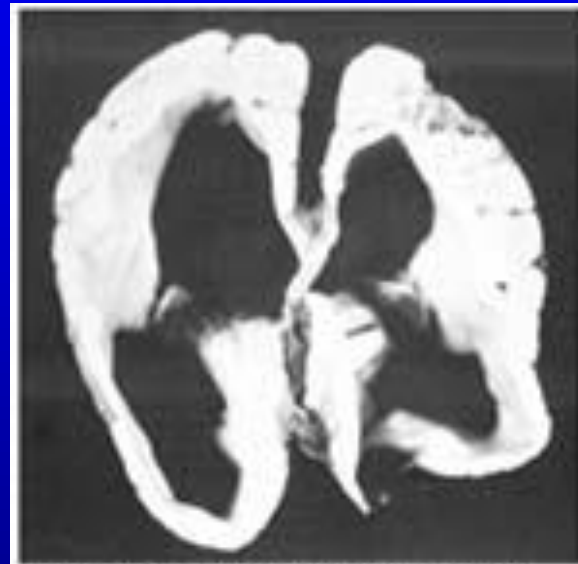
Defeito devido à interferência externa com o desenvolvimento normal (necrose).



AMBIENTAIS

- AGENTES INFECCIOSOS :
Toxoplasmosse, Sífilis, Citomegalovírus,
Rubéola

Ambientais



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

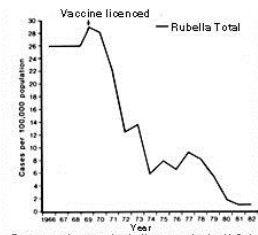
Toxoplasmosose

AMBIENTAIS

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).

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



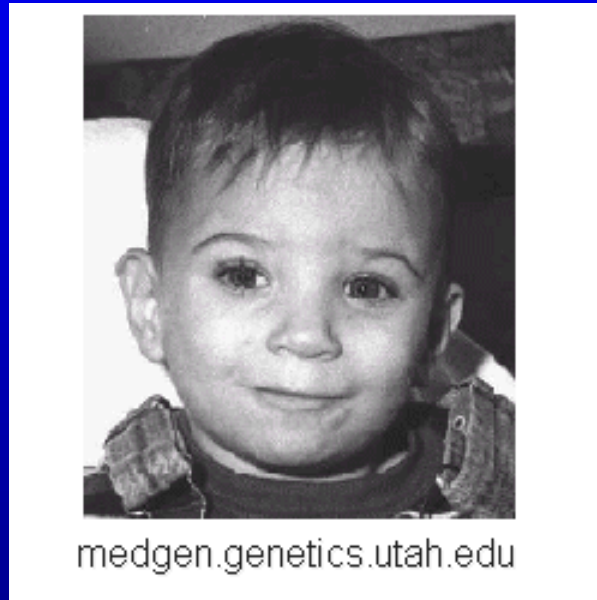
Warfarin

Ambientais



Misoprostol (Cytotec)

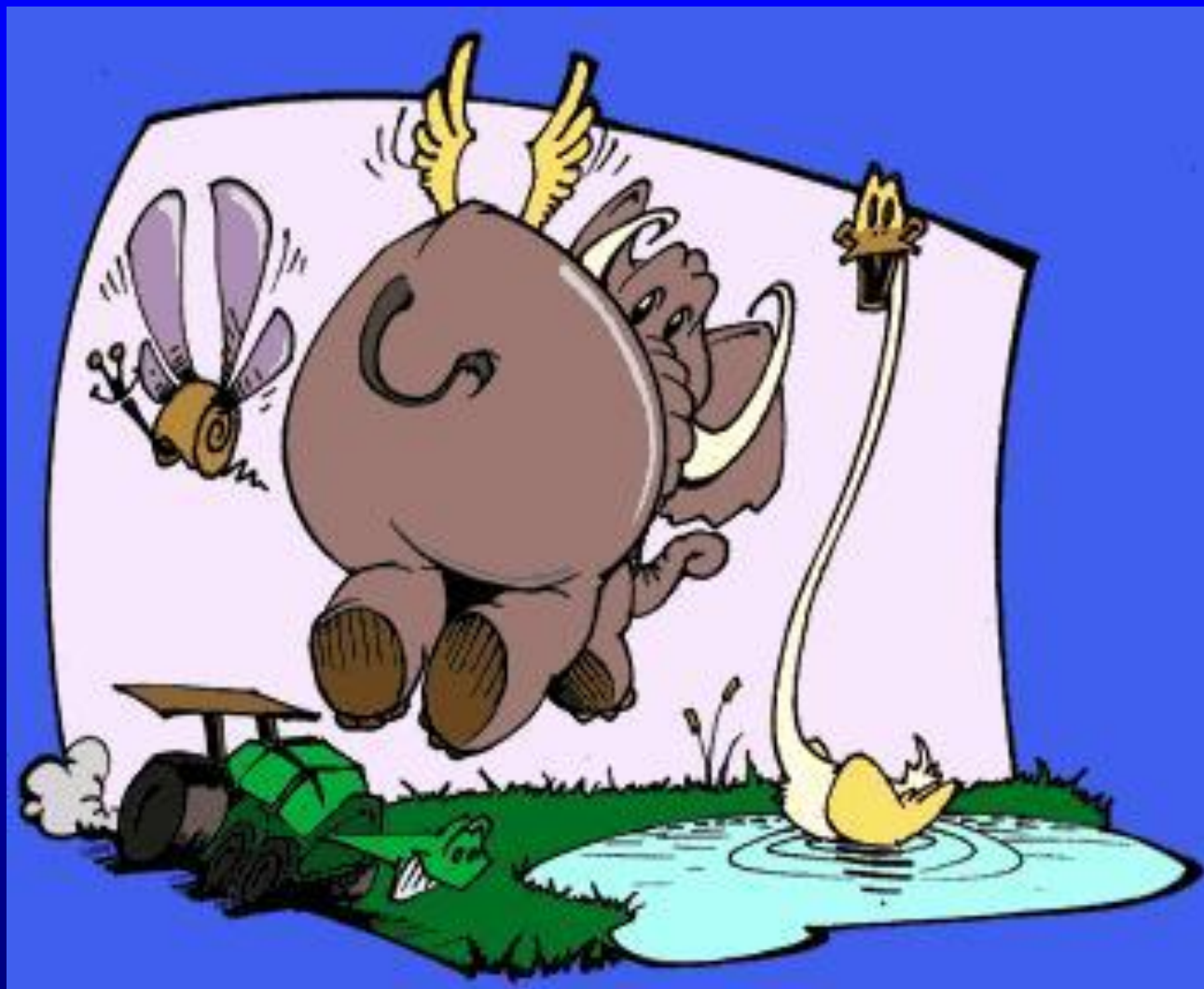
Ambientais



Síndrome Alcoólica Fetal

SAF



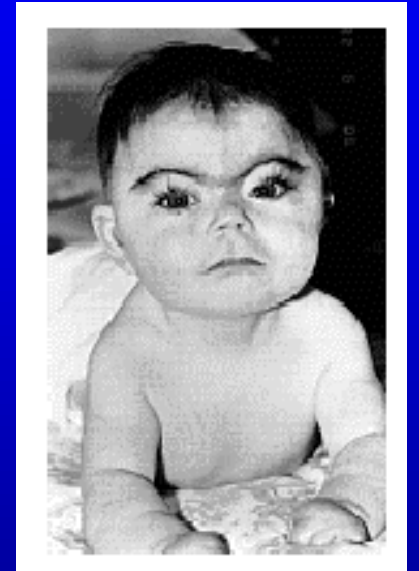


Alterações “maiores”

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)