Anomalias Congênitas: Dismorfologia

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esramos@fmrp.usp.br

Congênito

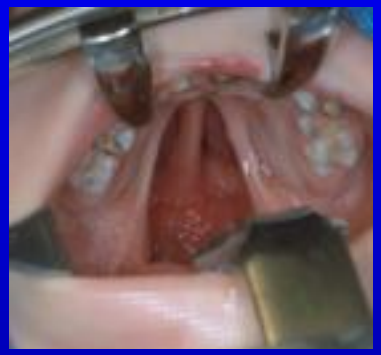
≠

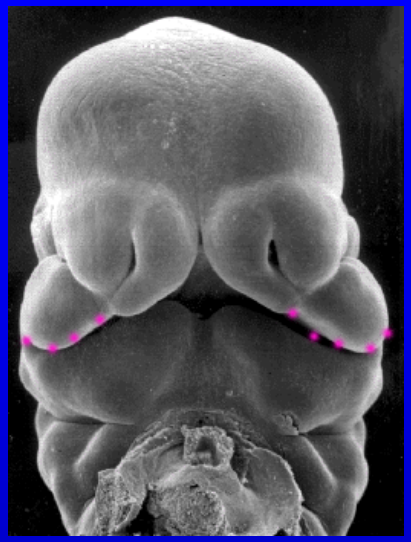
Genético

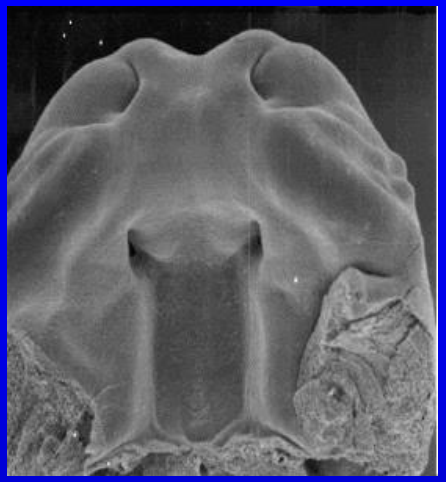
Defeitos congênitos

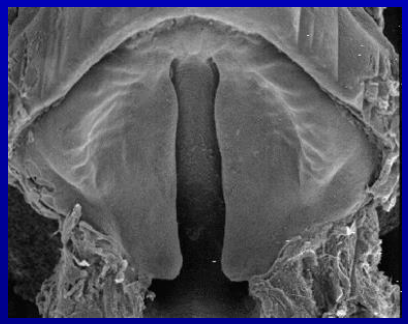
1. **Morfogênese incompleta (agenesia, hipoplasia)**

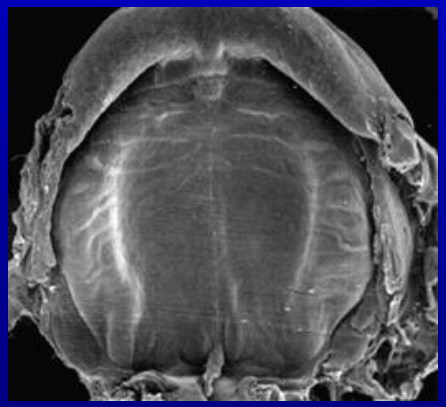
1. **Morfogênese incompleta (agenesia, hipoplasia)**
2. **Fechamento incompleto (fendas)**

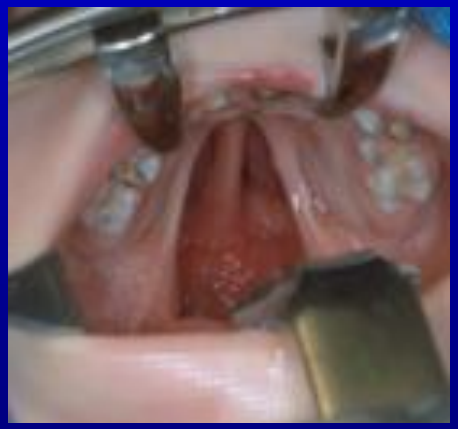




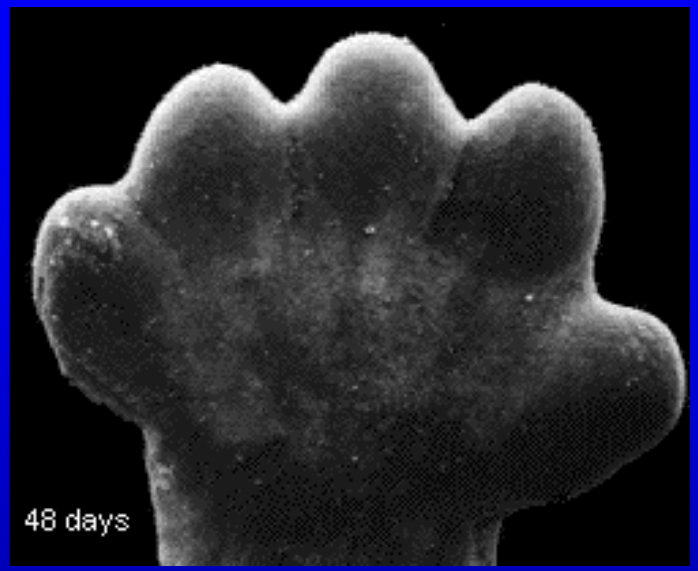




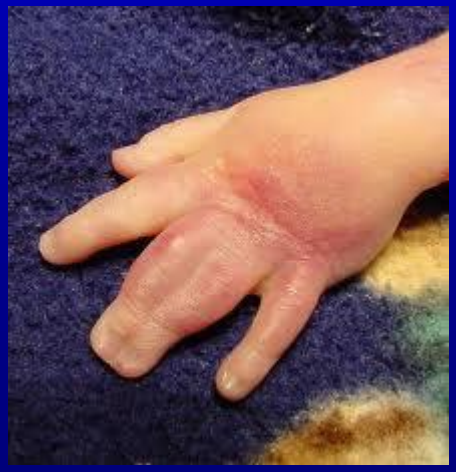




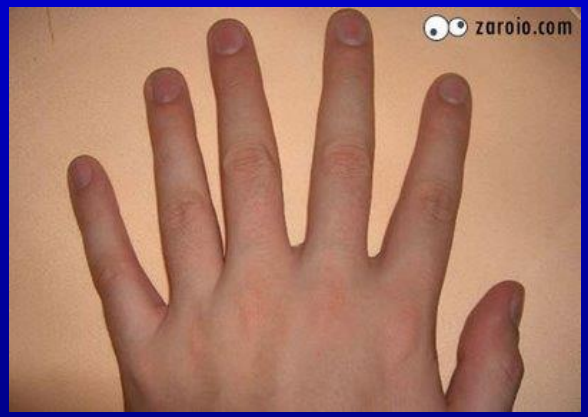
1. **Morfogênese incompleta (agenesia, hipoplasia)**
2. **Fechamento incompleto (fendas)**
3. **Separação incompleta (sindactilia)**

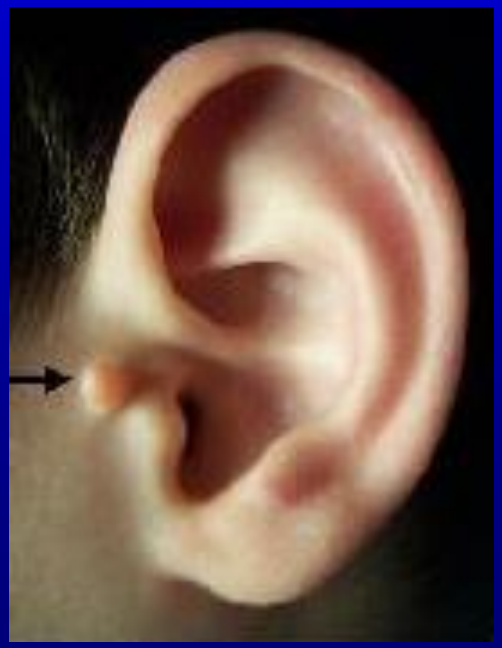

48 days


51 days



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

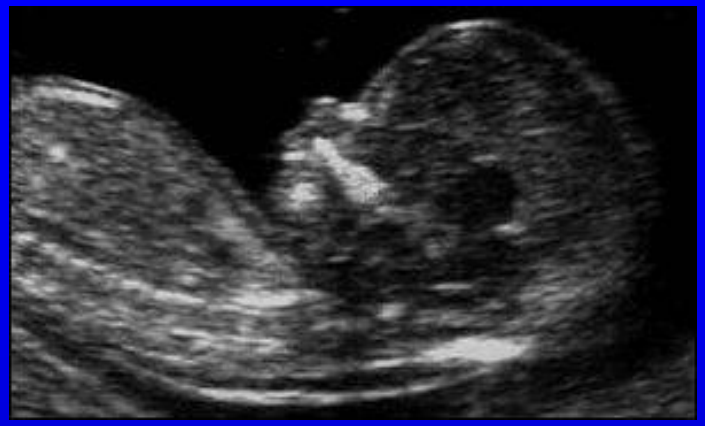


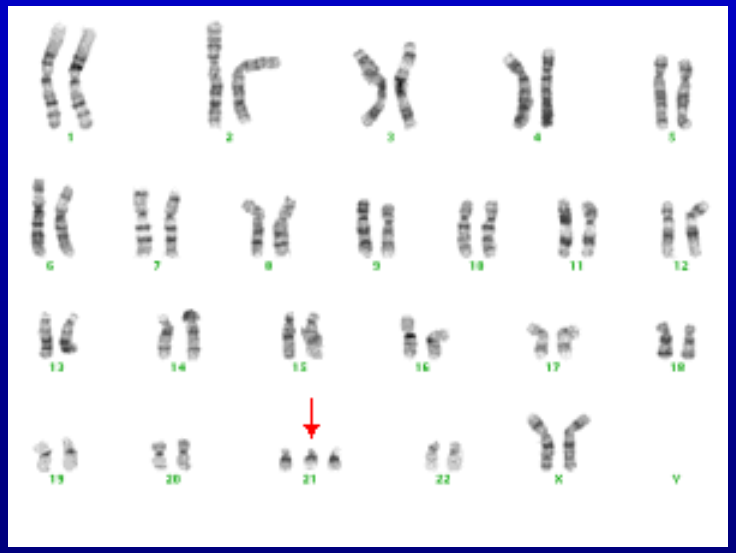


Normal

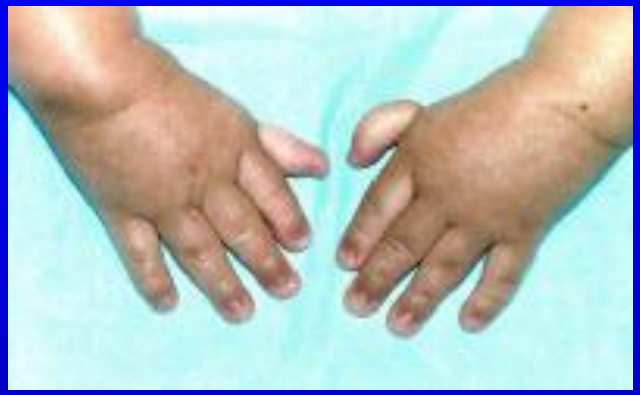
Malformação

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

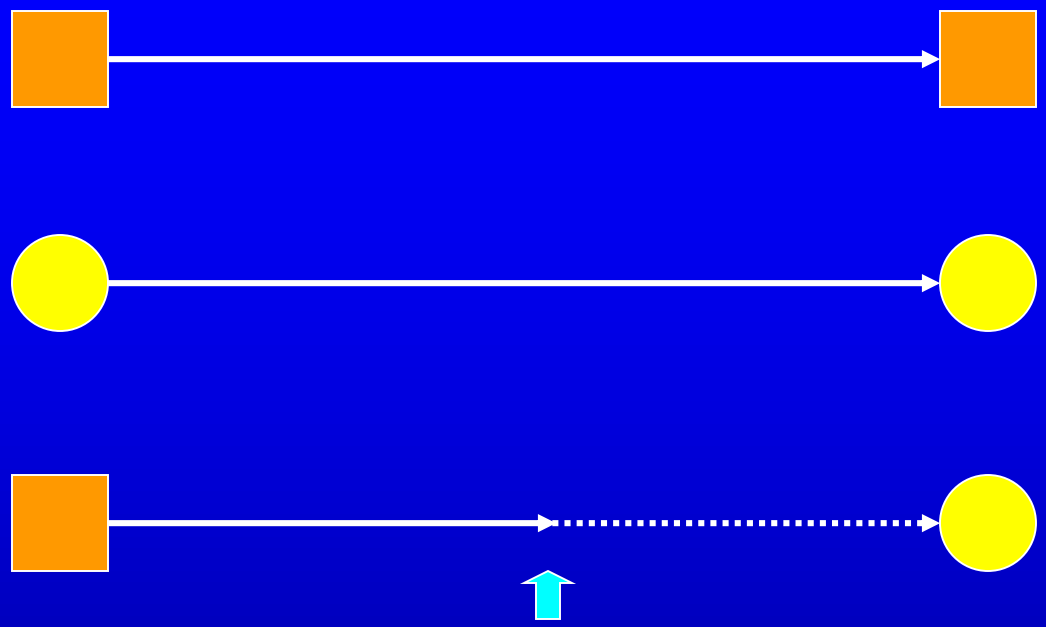




Síndrome de Down

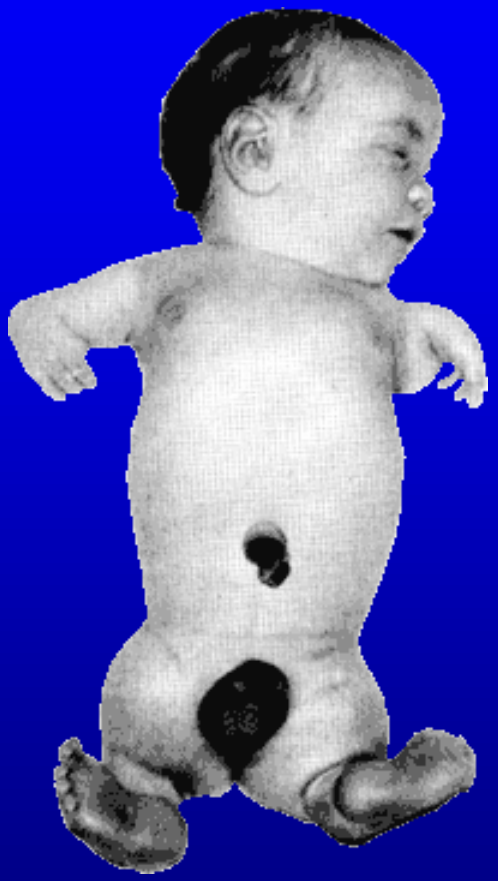


Anemia de Fanconi (monogênica - AR)

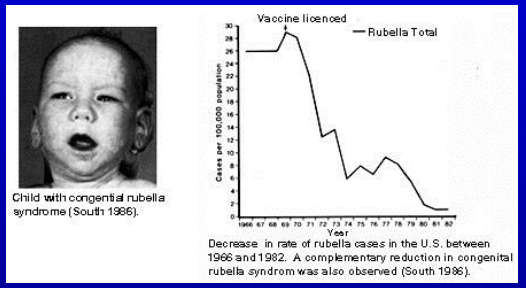


Defeito devido à interferência externa com o desenvolvimento normal (necrose).

Talidomida

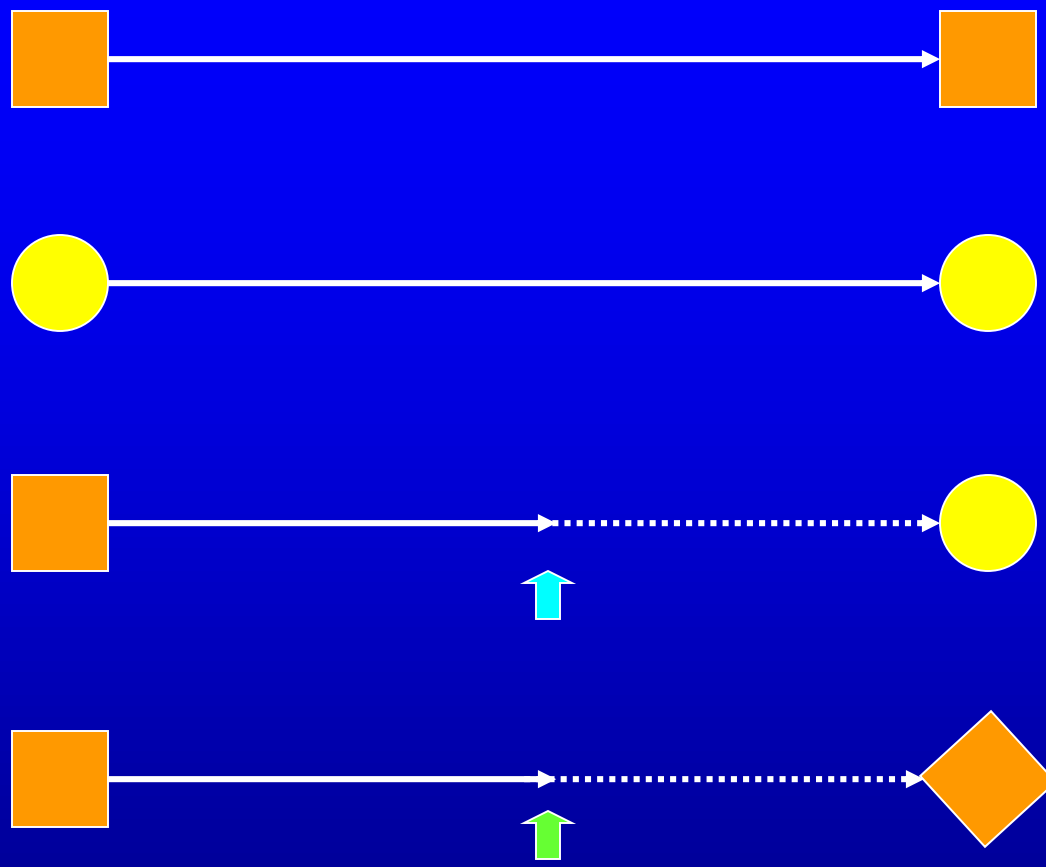


Rubéola



Child with congential rubella syndrome (South 1986).

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



Normal

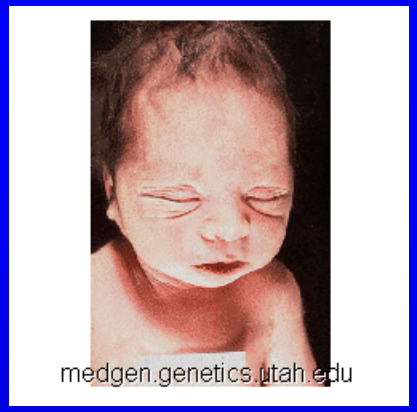
Malformação

Disrupção

Deformidade

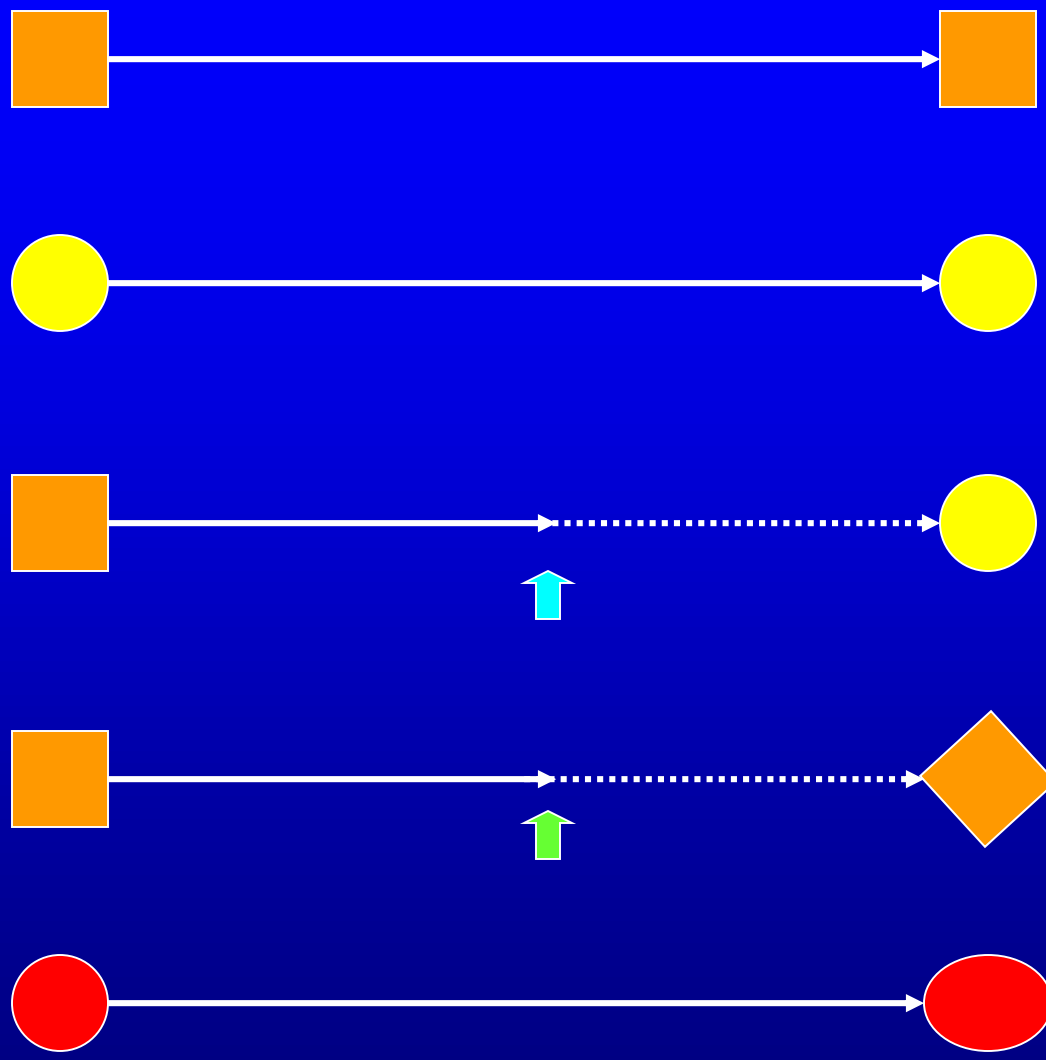
Forma ou posição anormal de uma parte do corpo, causada por força mecânica.

Sequência de Potter



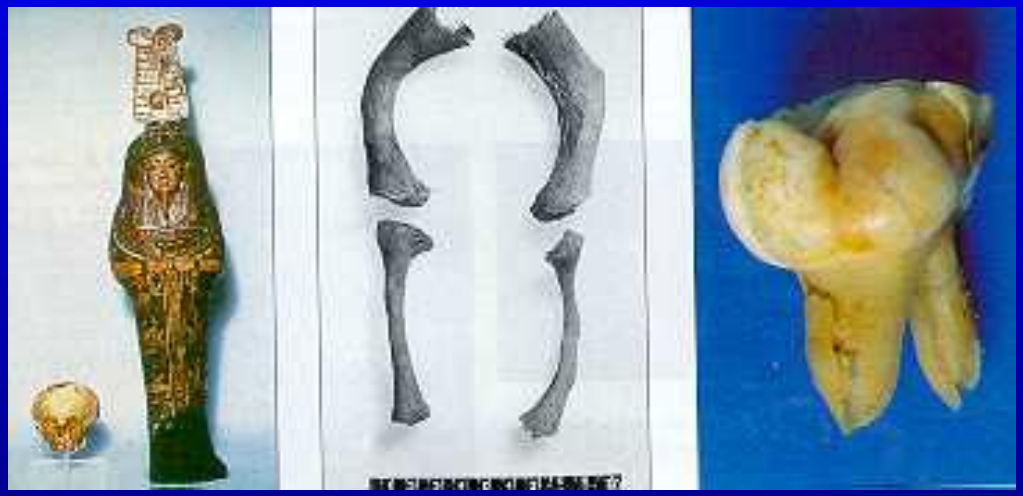
Agenesia Renal → **Líquido Amniótico** ↓

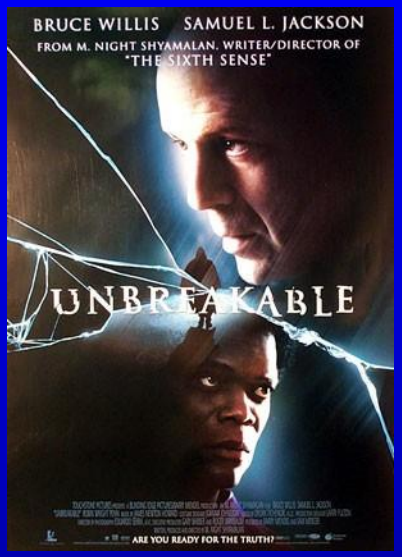
Fator mecânico



Desorganização tecidual.

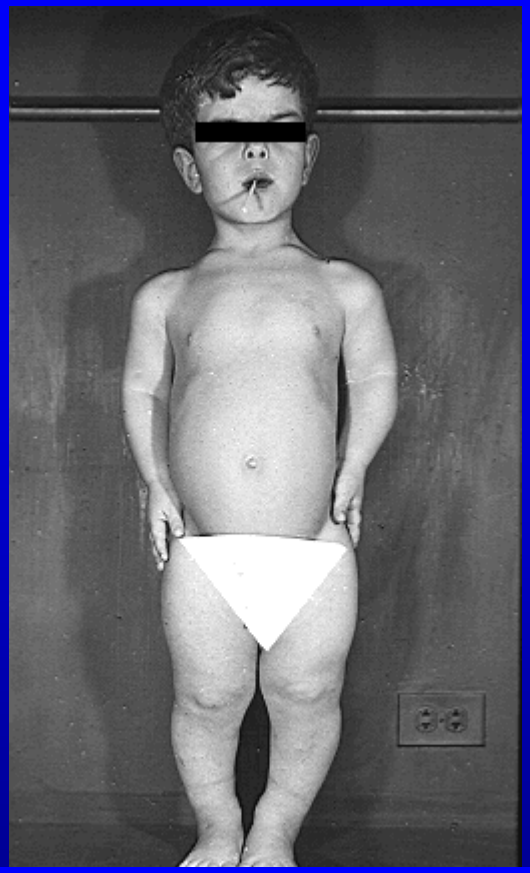
Osteogênese Imperfeita



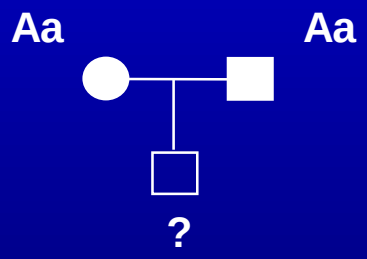


Osteogênese Imperfeita

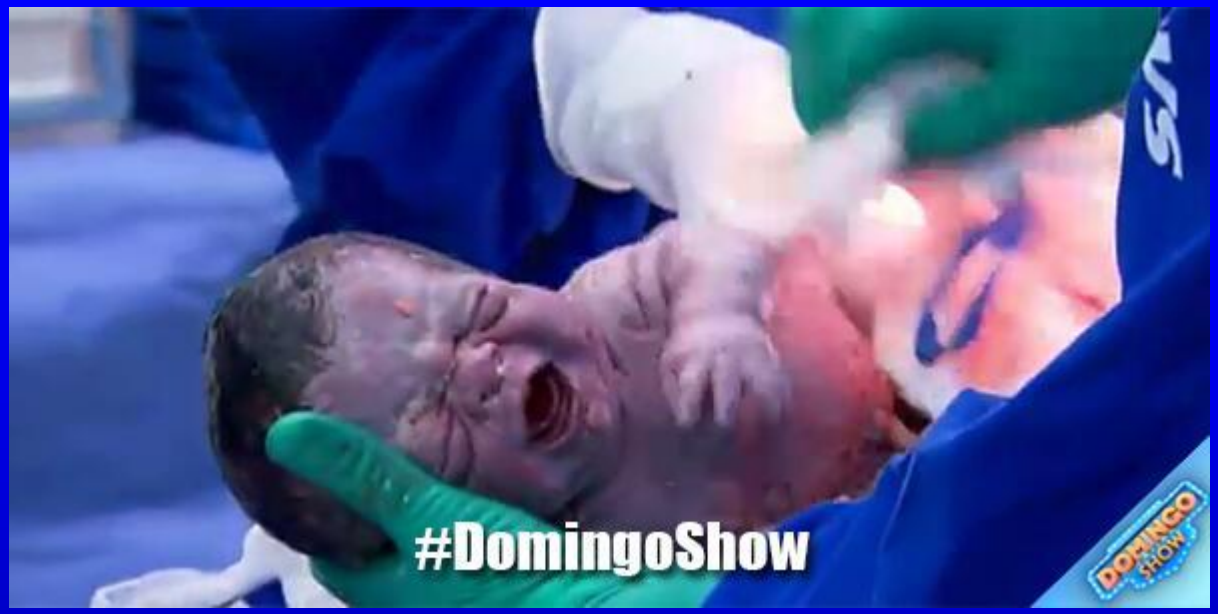
Displasia óssea



Acondroplasia



AA Aa Aa aa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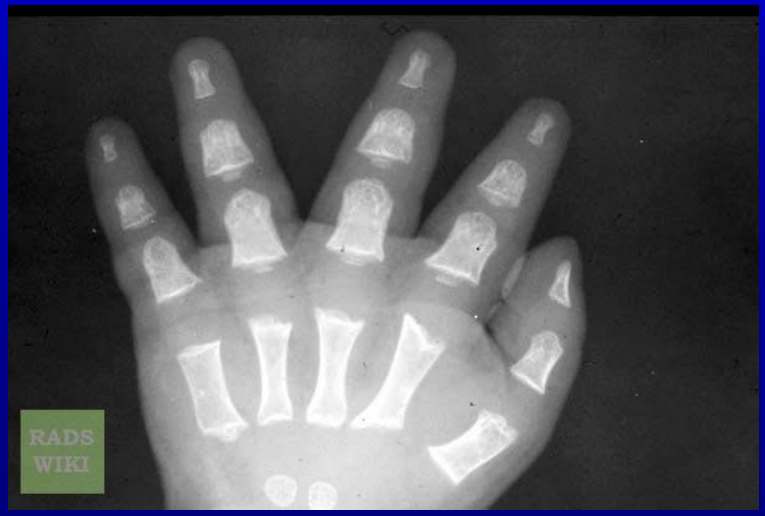
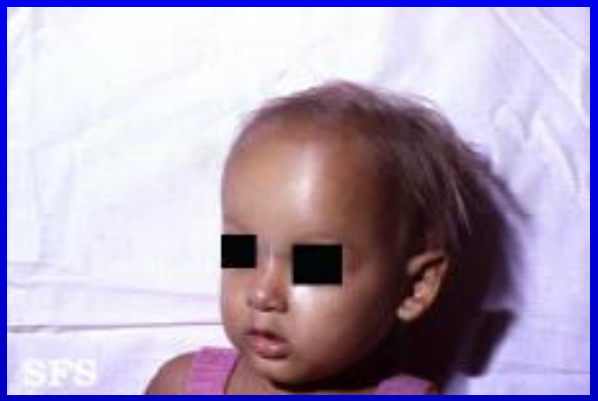
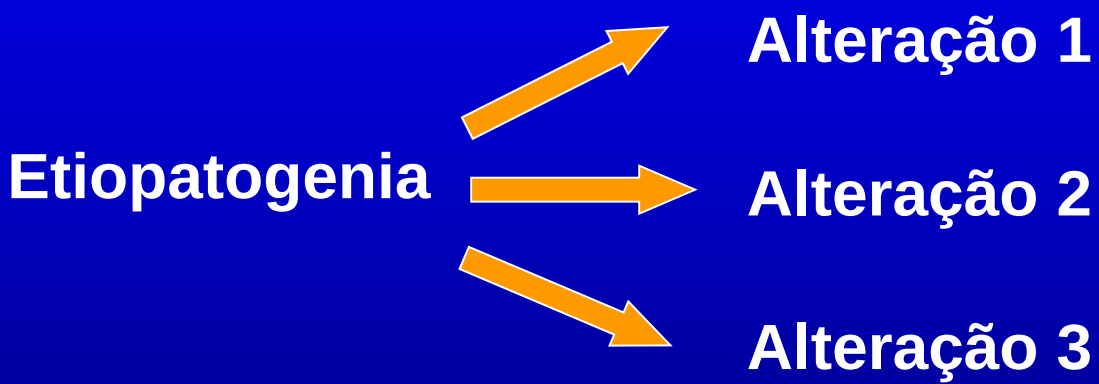
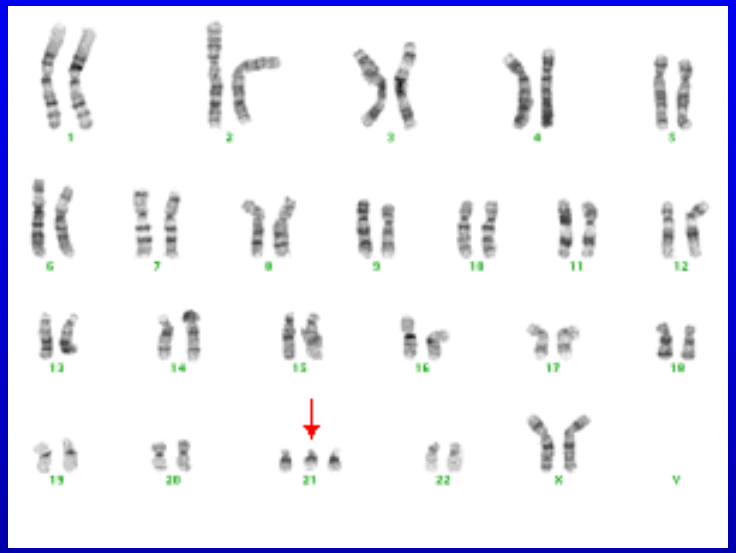

RADS
WIKI



Displasia ectodérmica

Síndrome

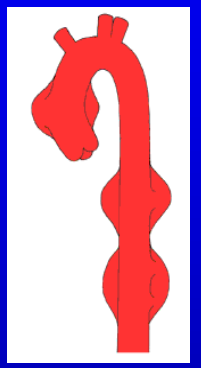


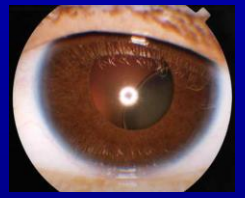


Síndrome de Down

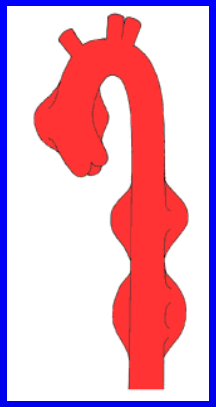
Síndrome de Marfan

Gene →





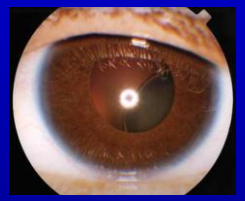
Alta estatura



Dilatação de aorta

Aracnodactilia

Frouxidão ligamentar

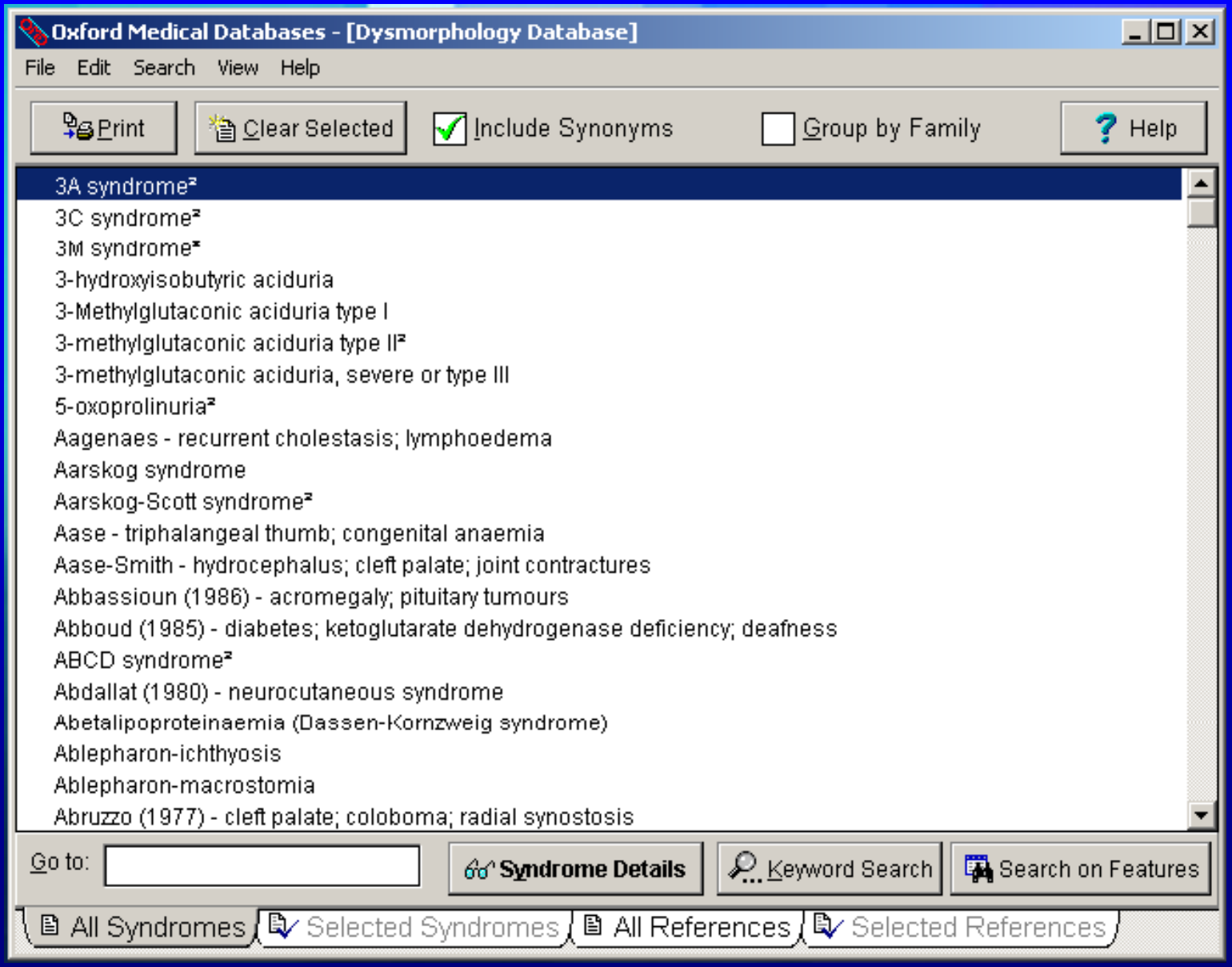


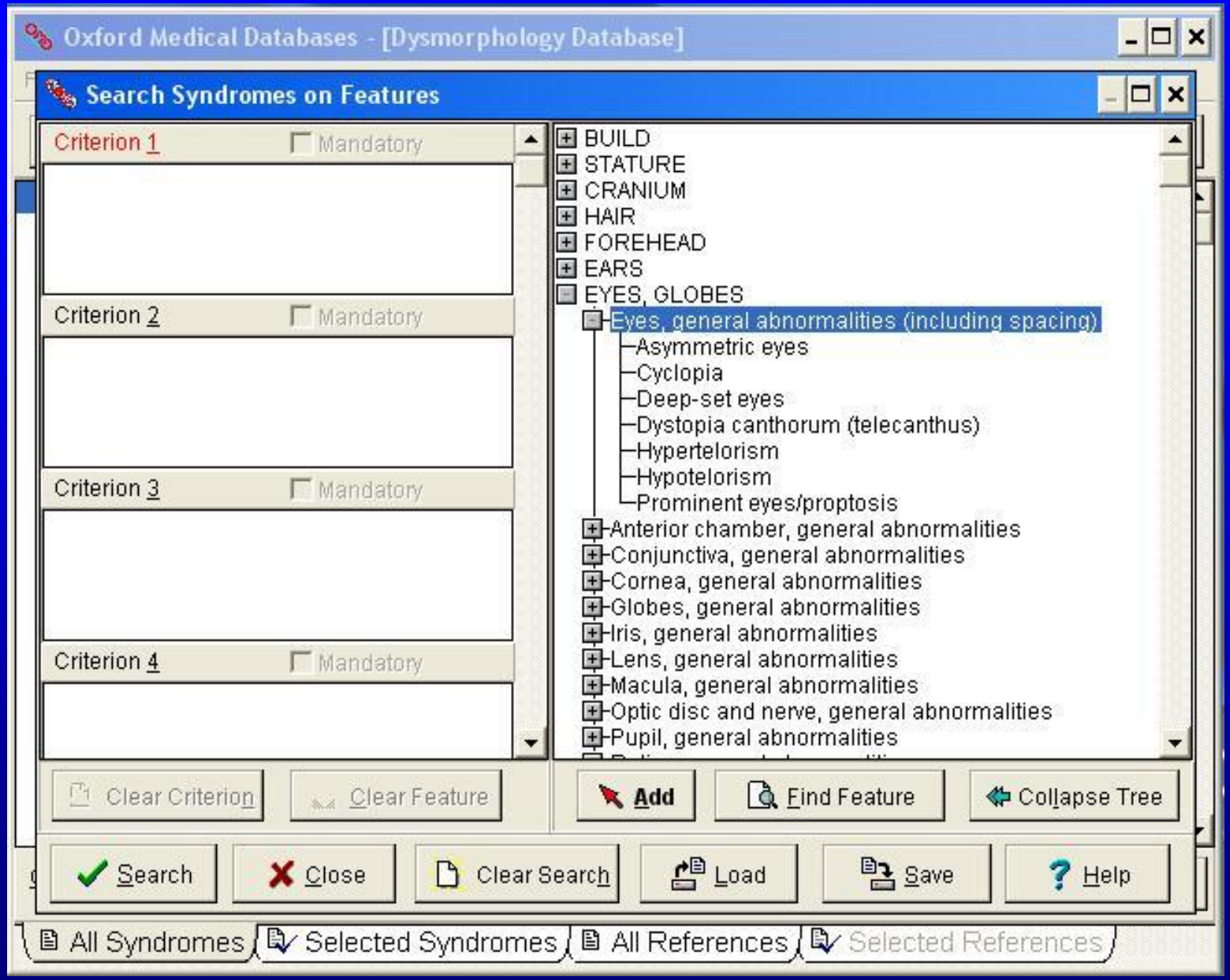
Miopia
Luxação do cristalino

Síndrome Alcoólica Fetal

Álcool ⟶

medgen.genetics.utah.edu


Oxford Medical Databases - [Dysmorphology Database]
File Edit Search View Help
Print
Clear Selected
Include Synonyms
Group by Family
Help
3A syndrome²
3C syndrome²
3M syndrome²
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
All Syndromes
Selected Syndromes
All References
Selected References


Oxford Medical Databases - [Dysmorphology Database]
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
Clear Criterion
Clear Feature
Add
Find Feature
Collapse Tree
Search
Close
Clear Search
Load
Save
Help
All Syndromes
Selected Syndromes
All References
Selected References

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Marfan syndrome

Location: 15q21
McKusick: 154700
Synonyms:
Inheritance:

Synonyms	Inheritance
MFS²	Autosomal dominant

Abstract

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 diagnositc 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.

6 Photos






Abstract | Features | References

All Syndromes | Selected Syndromes | All References | Selected References

Iniciar | Entrada - esramo... | PubMed Home - ... | Aulas | 4 Microsoft Offi... | Genetica_Human... | Oxford Medical ... | imagem - Paint | Norton | 17:13

Oxford Medical Databases - [Dysmorphology Database]

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Marfan syndrome

Location: 15q21
McKusick: 154700

Synonyms:
- MFS²

Inheritance:
- Autosomal dominant

Features

- 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

6 Photos

Abstract | Features | References

All Syndromes | Selected Syndromes | All References | Selected References

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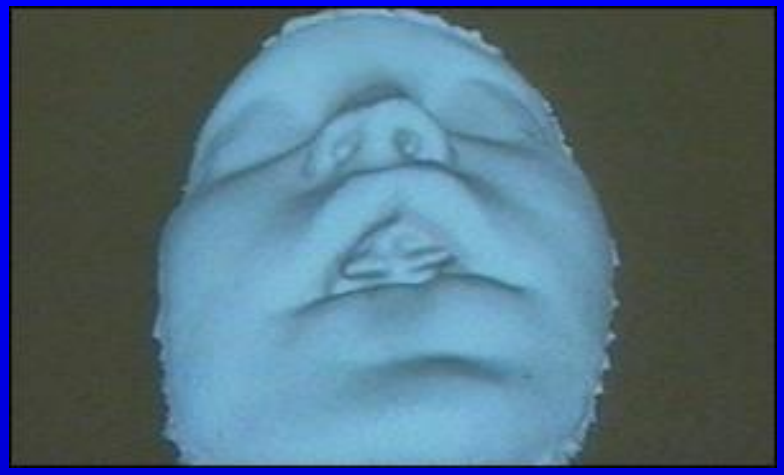
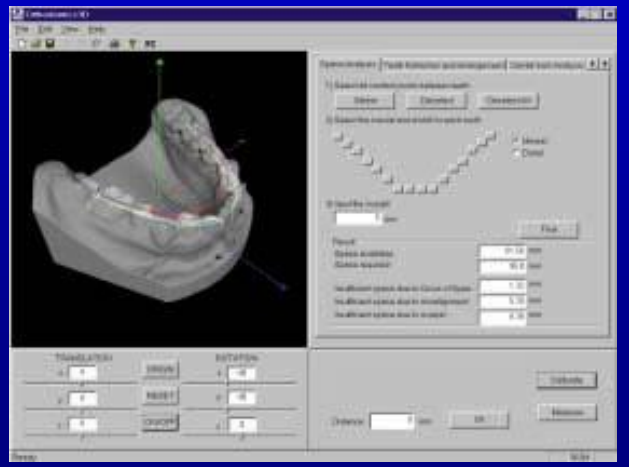
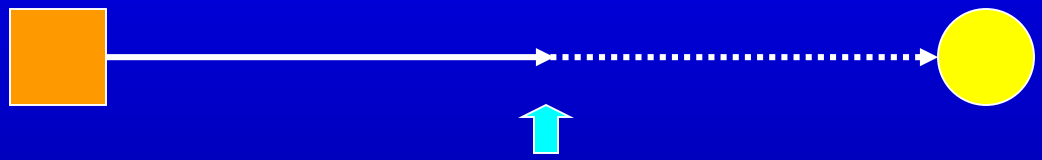




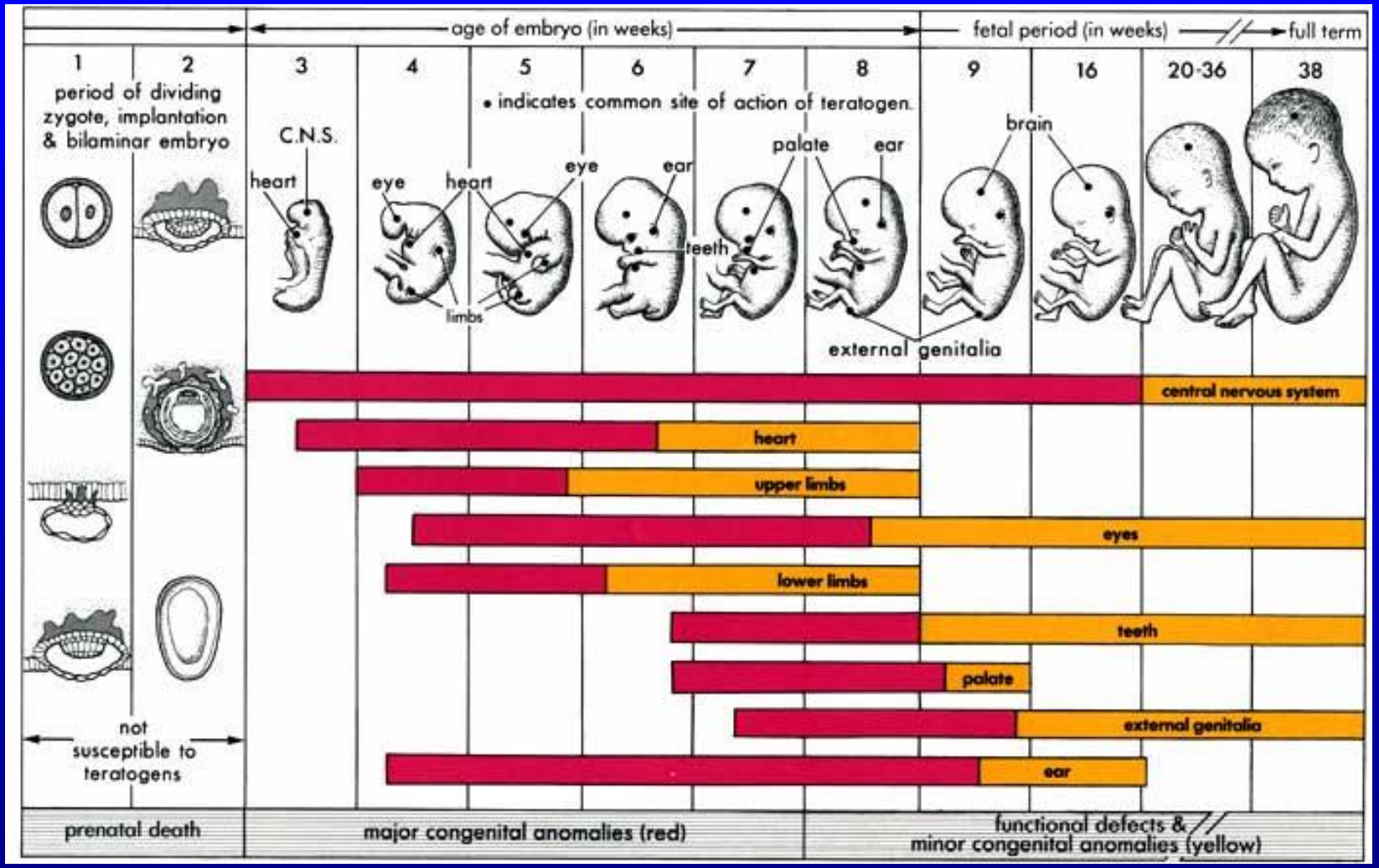
Ambientais
(teratógenos)

Normal



Disrupção

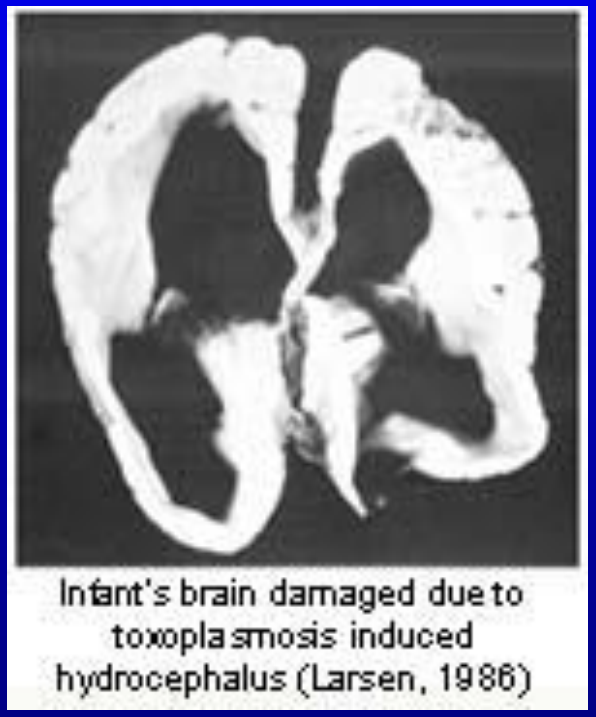
Defeito devido à interferência externa com o desenvolvimento normal (necrose).


age of embryo (in weeks)
fetal period (in weeks)
full term
1
2
3
4
5
6
7
8
9
16
20-36
38
period of dividing zygote, implantation & bilaminar embryo
• indicates common site of action of teratogen.
C.N.S.
heart
eye
heart
eye
ear
teeth
palate
ear
brain
limbs
external genitalia
central nervous system
heart
upper limbs
eyes
lower limbs
teeth
palate
external genitalia
ear
not susceptible to teratogens
prenatal death
major congenital anomalies (red)
functional defects & minor congenital anomalies (yellow)

AMBIENTAIS

- AGENTES INFECCIOSOS : Toxoplasmose, Sífilis, Citomegalovírus, Rubéola

Ambientais

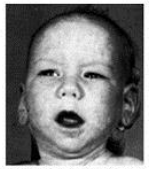


Infant's brain damaged due to toxoplasmosis induced hydrocephalus (Larsen, 1986)

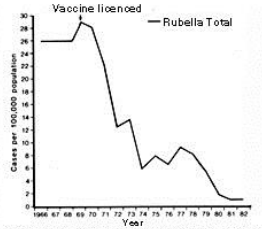
Toxoplasmose

AMBIENTAIS

Rubéola



Child with congential rubella syndrome (South 1986).



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

AMBIENTAIS

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

Regressão caudal

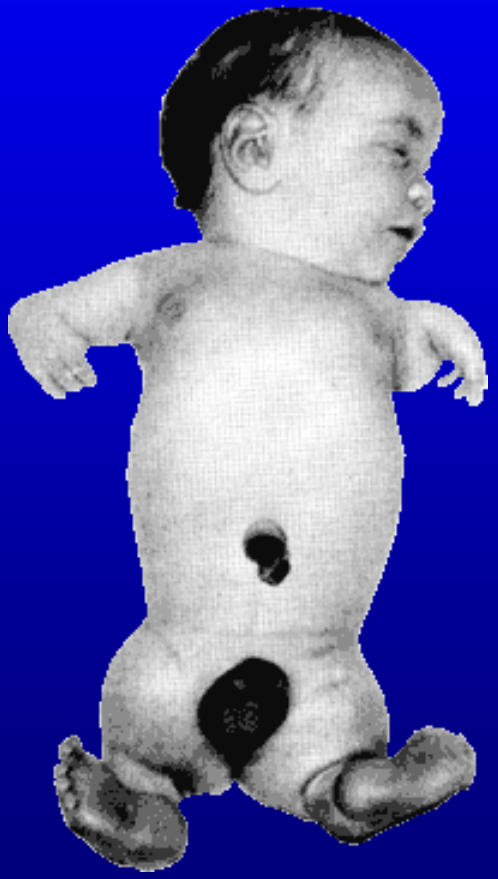
(diabetes)

AMBIENTAIS

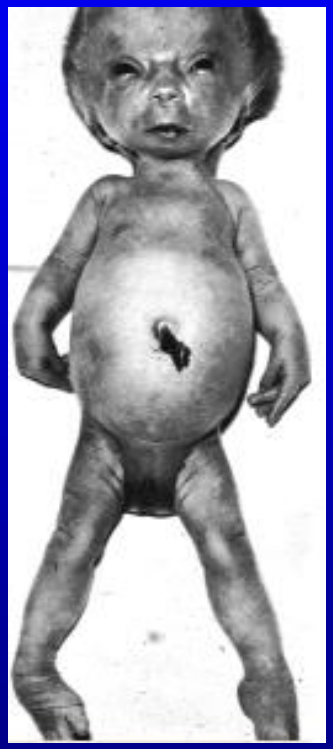
•AGENTES FÍSICO-QUÍMICOS : Radiação, Talidomida, Hidantoína, Aminopterina (MTX), Misoprostol (Cytotec), Álcool

AMBIENTAIS

Talidomida

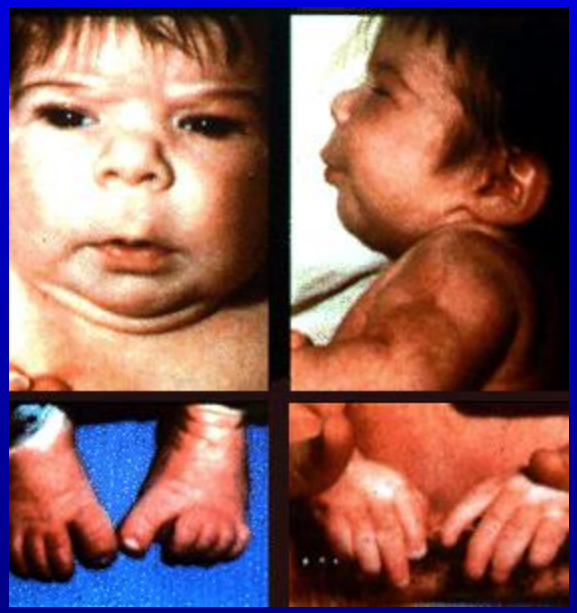


Ambientais



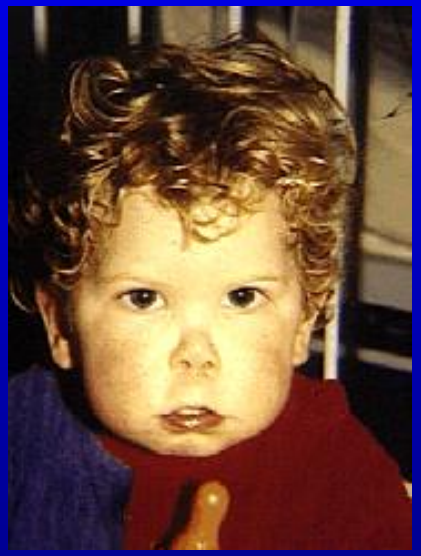
Aminopterina (MTX)

Ambientais



Hidantoína

Ambientais



Warfarin

Ambientais

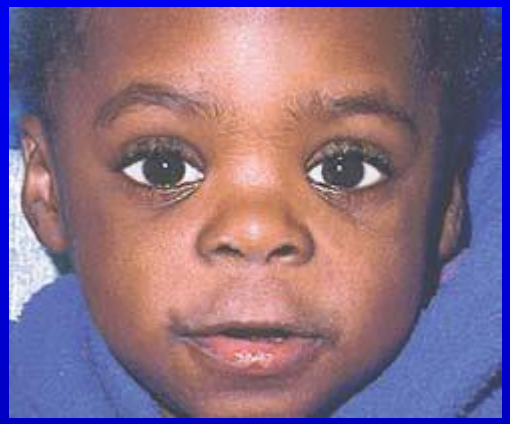
Misoprostol (Cytotec)

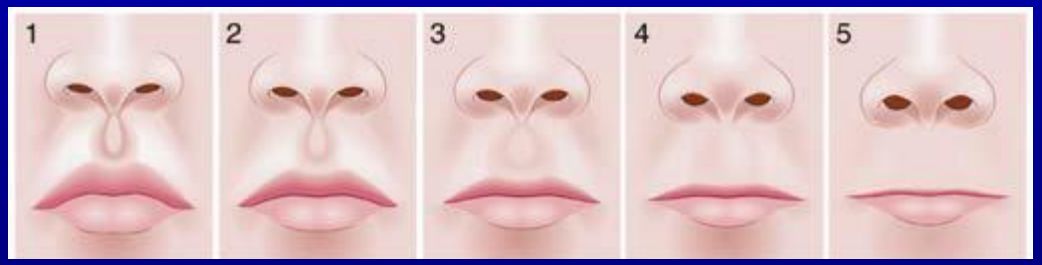
Ambientais

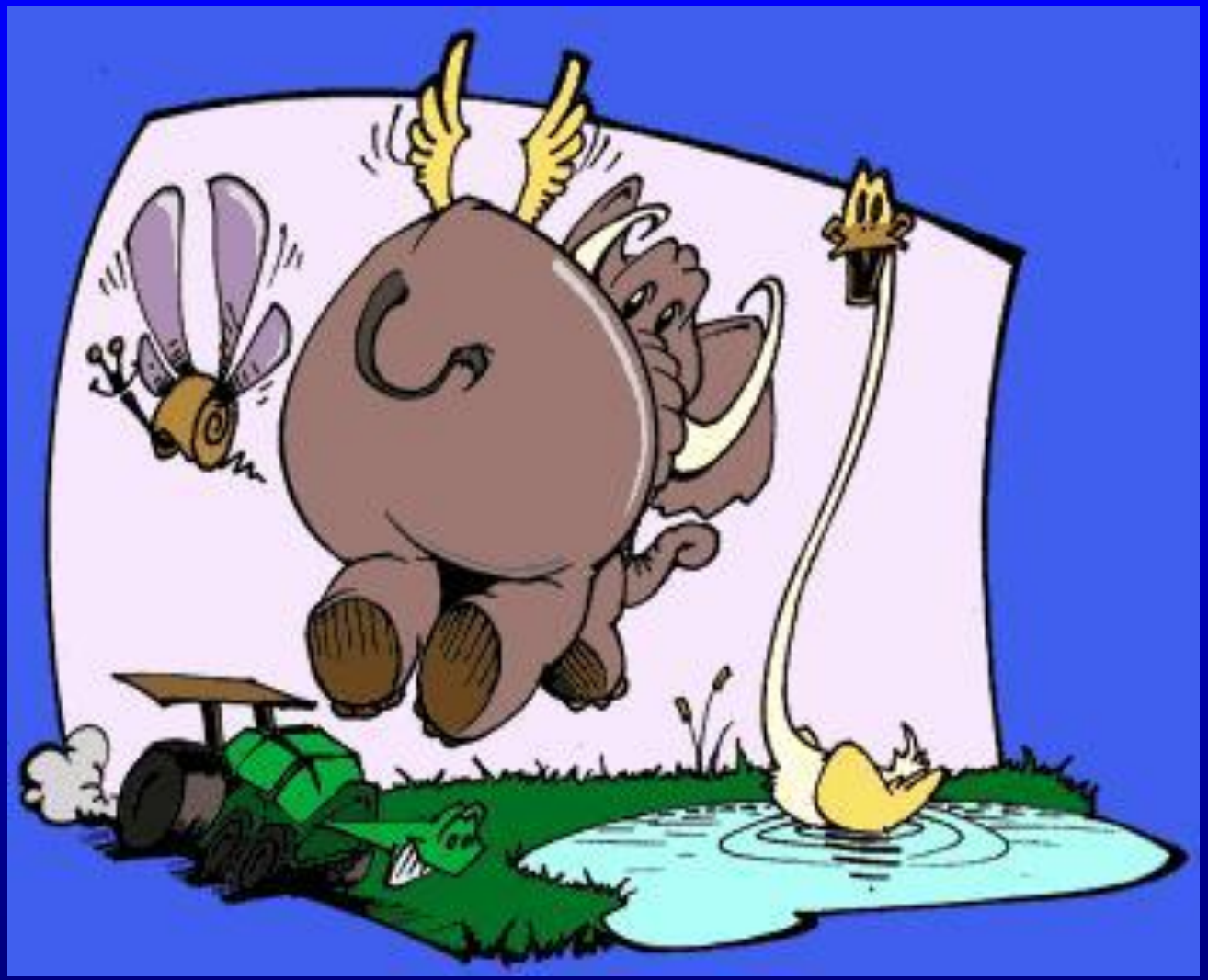
medgen.genetics.utah.edu

Síndrome Alcoólica Fetal

SAF





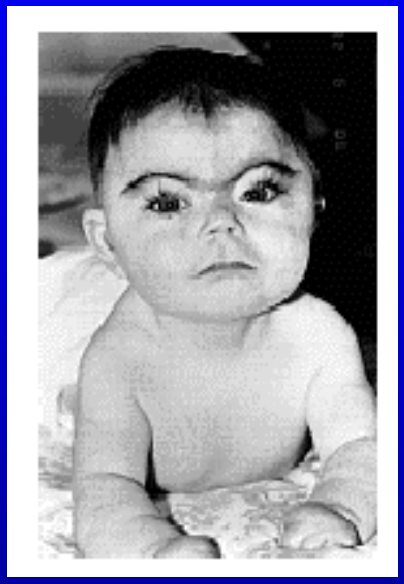


Alterações “maiores”

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)