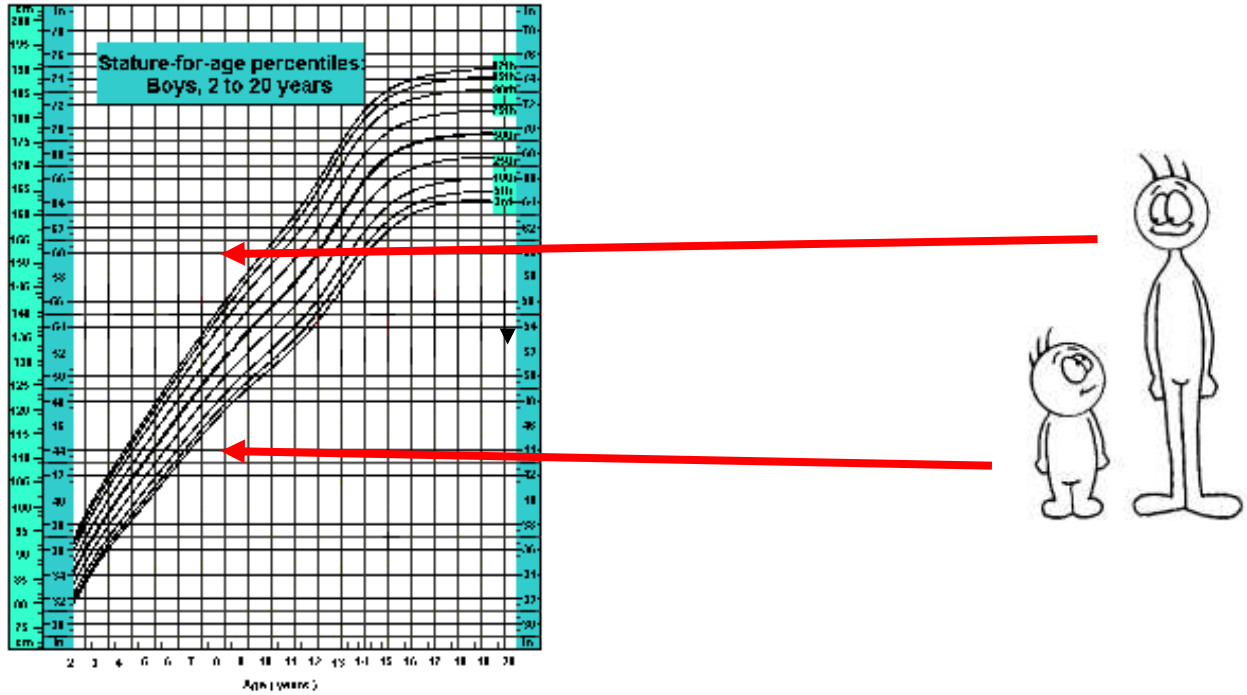
Crescimento



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

Crescimento "Normal"

Estatura Normal

Variação populacional

Distribuição normal

Baixa Estatura

Curvas de crescimento

Avaliação familiar/parental

História nutricional

Doença crônica

Atraso constitucional?

IO/IC

Períodos de Crescimento

- 1. Vida Intrauterina
- 2. Infância
- 3. Adolescência
- 4. Vida Adulta

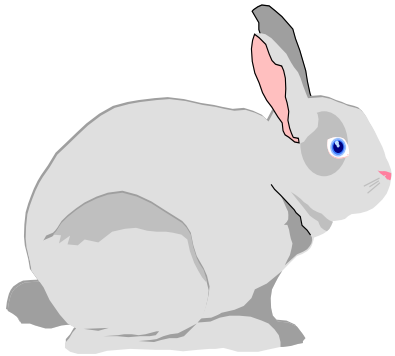
1. Vida Intrauterina

- Maior influência materna no crescimento: o tamanho de um RN a termo está correlacionado ao tamanho da mãe

Fator uterino

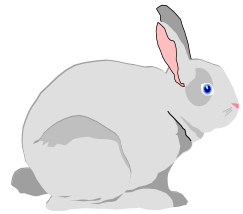
Fator placentário

Influência materna

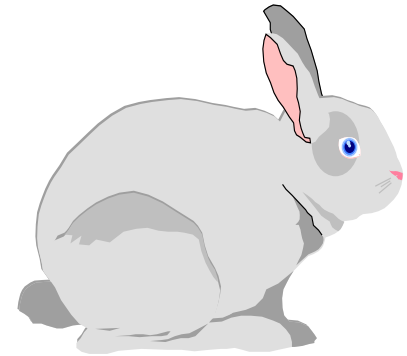


pai

x

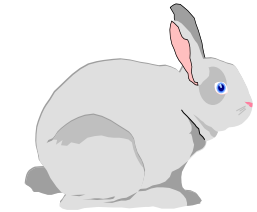


mãe



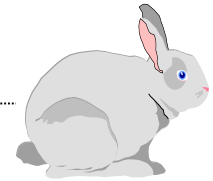
mãe

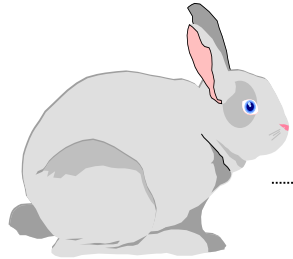
x

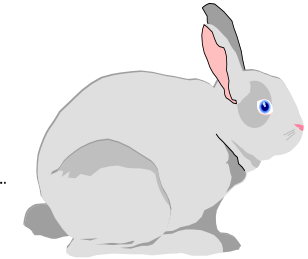


pai

ao nascimento







Influência do patrimônio genético, o tamanho é reflexo do tamanho médio dos pais.

2. Infância

- Período de mudança na taxa de crescimento: a criança tem um crescimento correlacionado ao tamanho médio dos pais

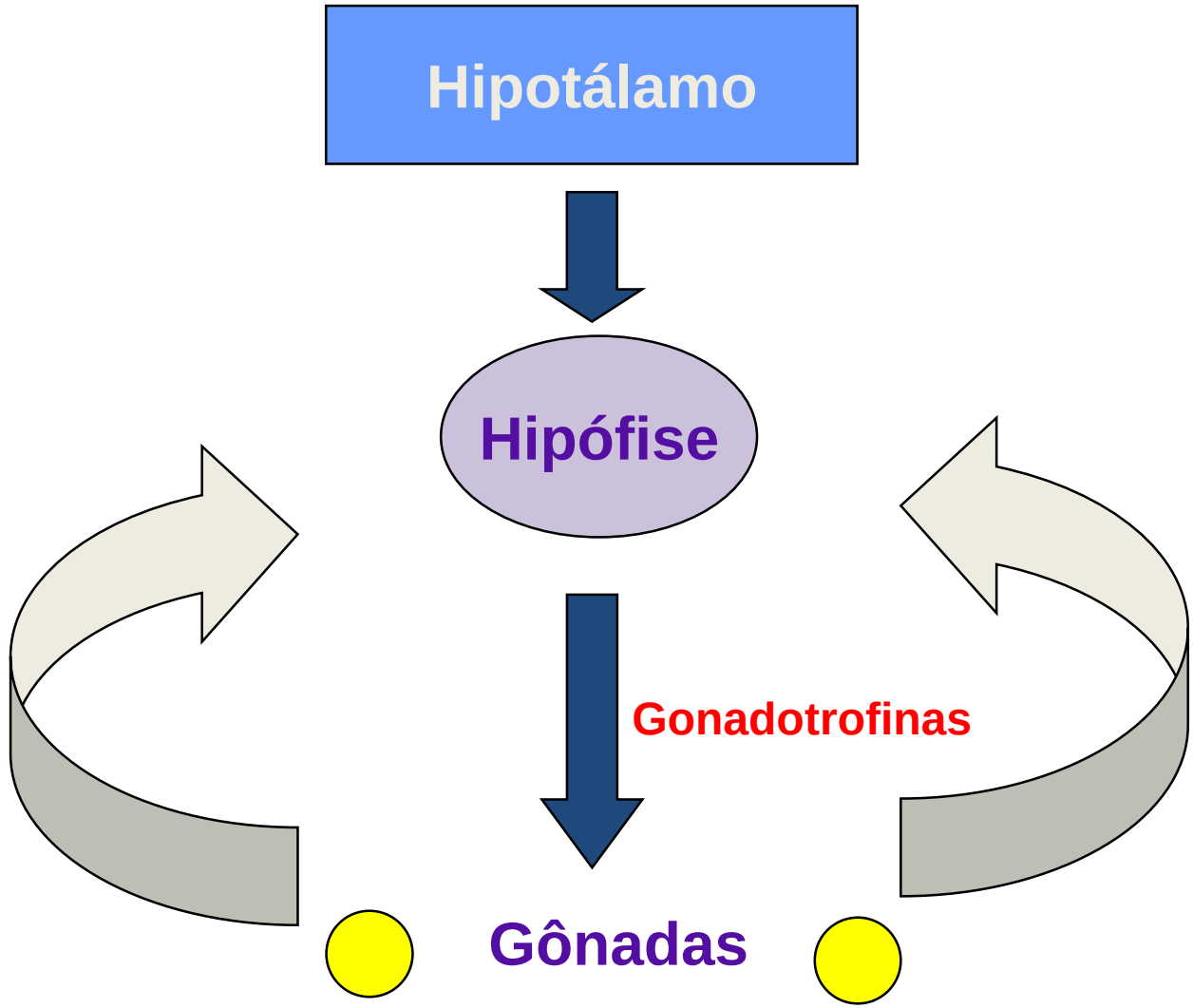
- Catch up - alcançam um novo canal de crescimento

- 2/3 mudam o canal de crescimento (12 aos 18meses)

- Velocidade de crescimento entre homens e mulheres igual. Nível de testosterona cai muito.

3. Adolescência

- Período do crescimento induzido por hormônios


Hipotálamo
Hipófise
Gonadotrofinas
Gônadas

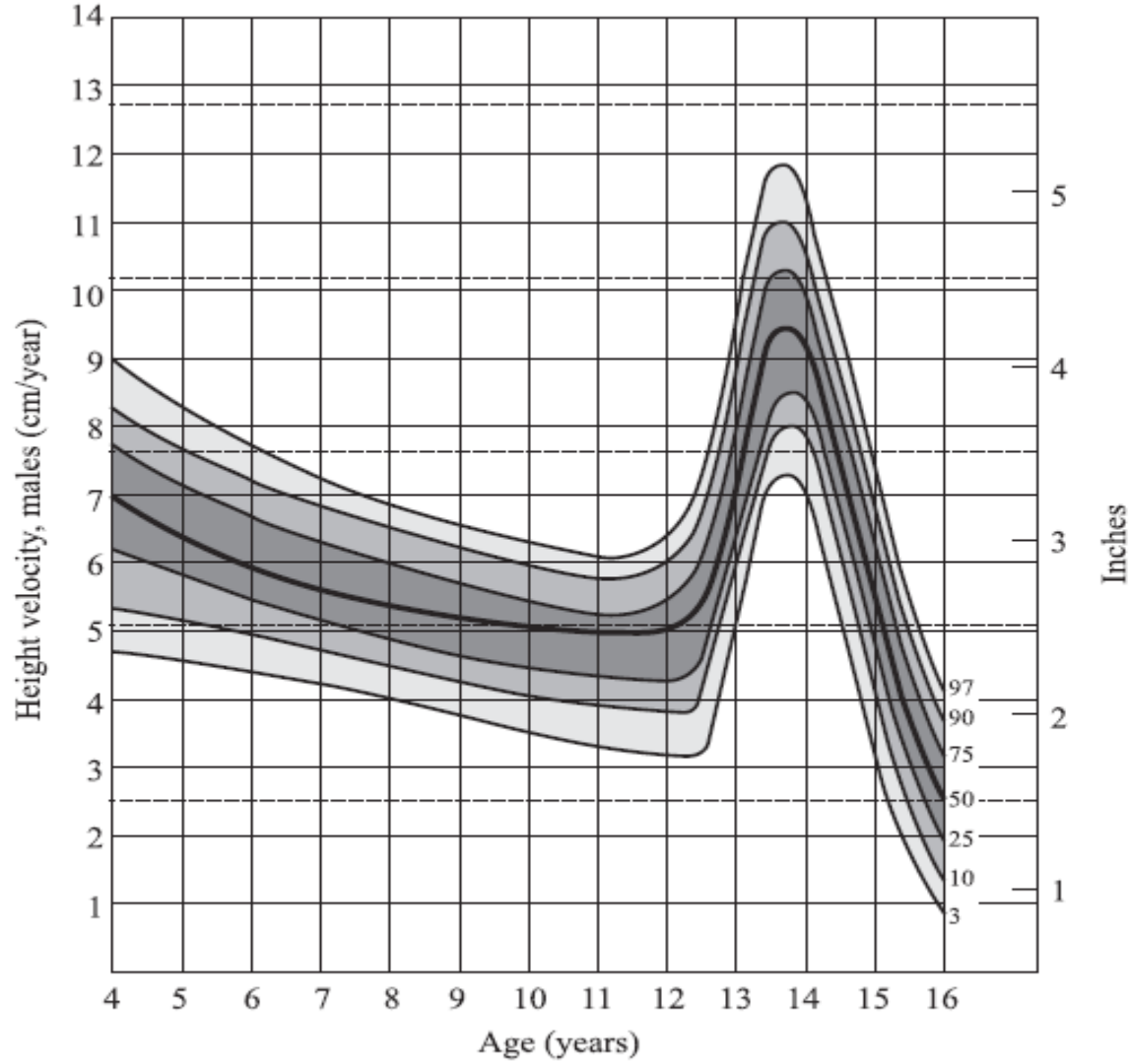


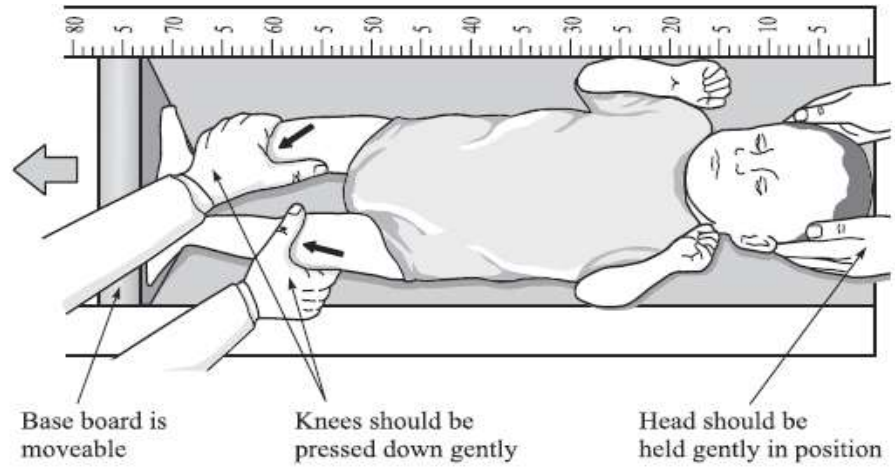
Figure 4.24 Height velocity, males. From Tanner and Whitehouse (1976), by permission.

4. Vida Adulta

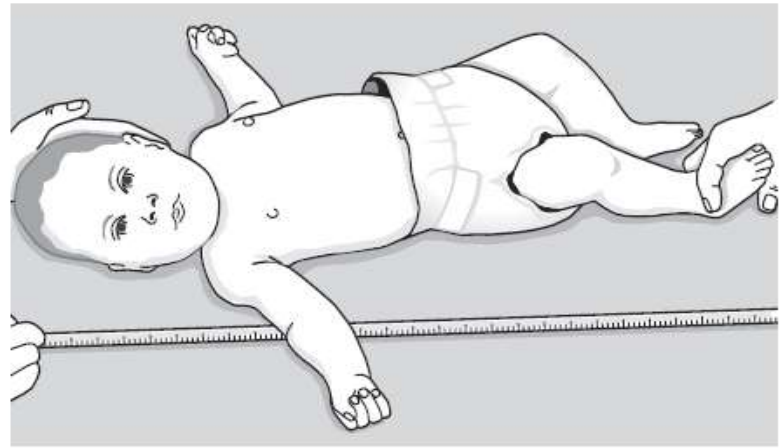
- Período da deterioração: as epífeses dos membros estão ossificadas ± 16 anos - mulheres e ± 18 anos - homens. Com cerca de 40 anos há aumento do tecido adiposo e decréscimo da estatura.

- Crânio, face e metacarpo crescem até ± 60 anos!

- Orelhas crescem até ± 80 anos!



(a)

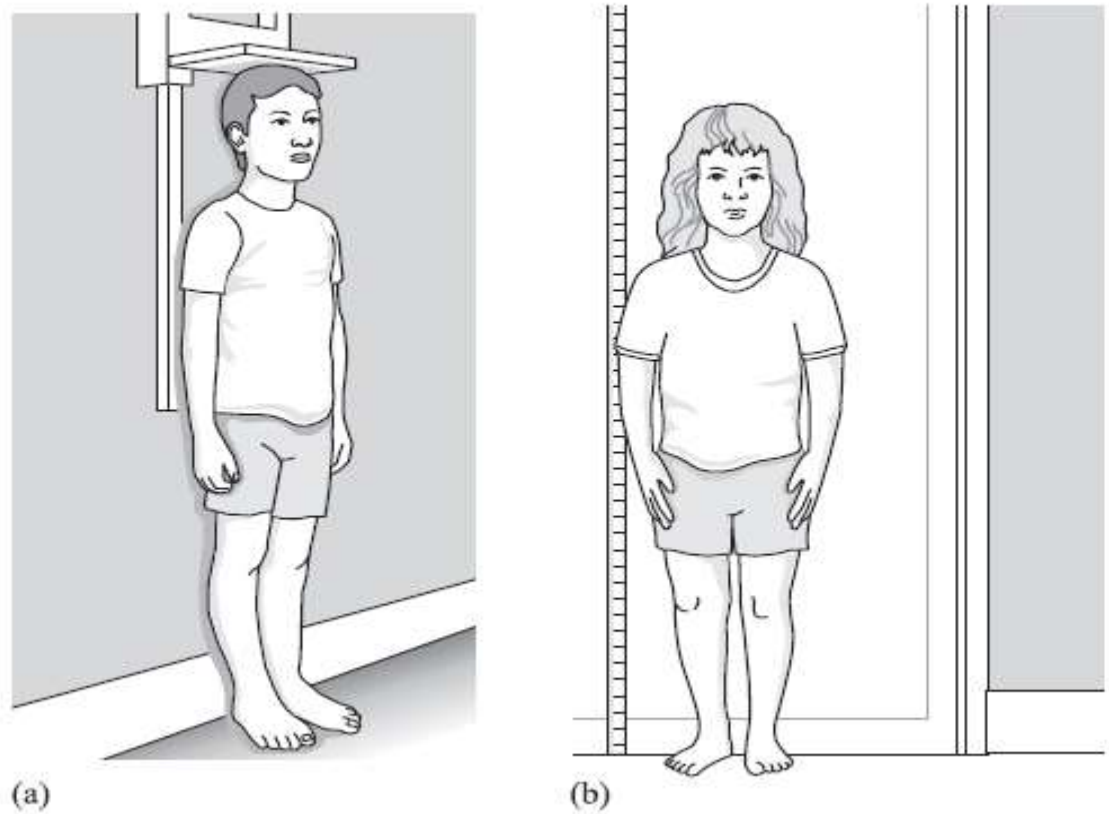


(b)

Figure 4.1 Measuring a child less than two years old with a measuring table (a) or tape-measure (b).

RN a 2a

Figure 4.5 Measuring standard height with a stadiometer (a) or a tape-measure (b).



> 2a

Proporção corpórea

- Proporção cabeça/corpo: o cérebro já tem 2/3 do seu tamanho final ao nascimento

- Cérebro: 12% do corpo ao nasto

  3% aos 16 anos

Figure 3.1 Body proportions during human development.

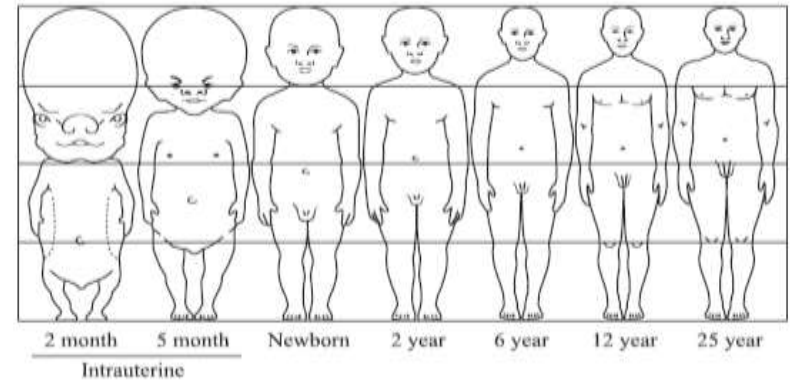
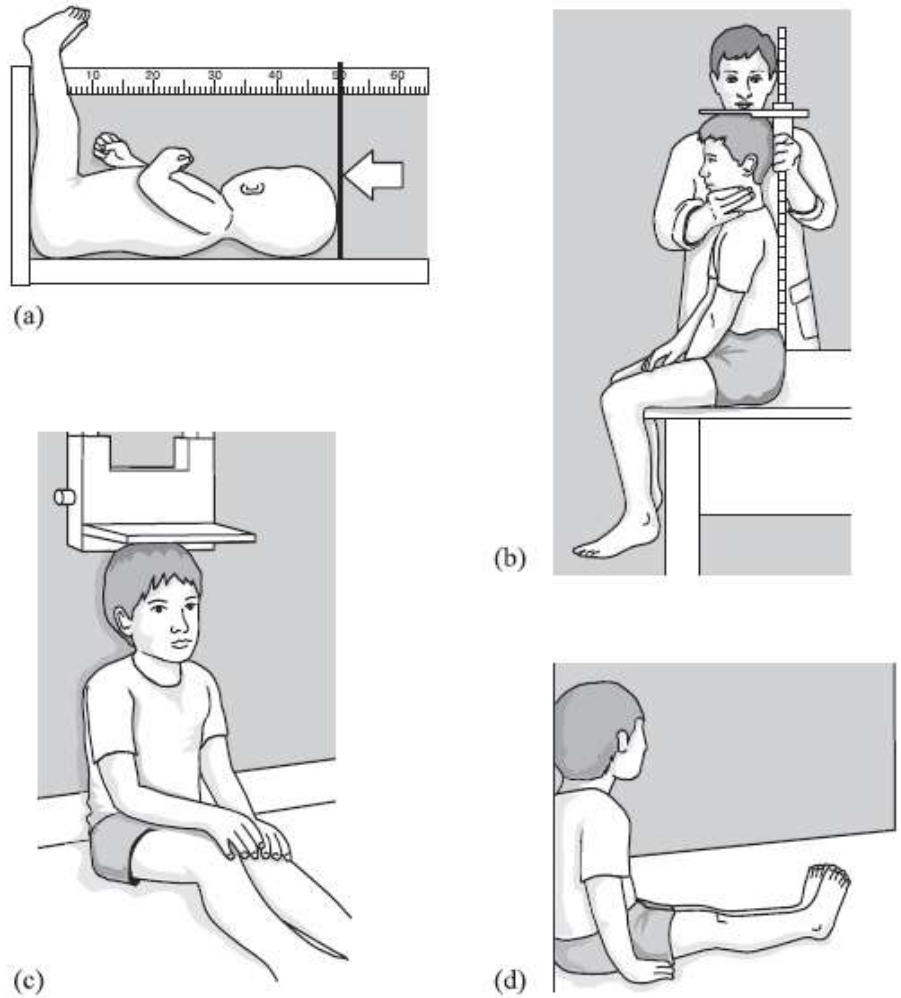
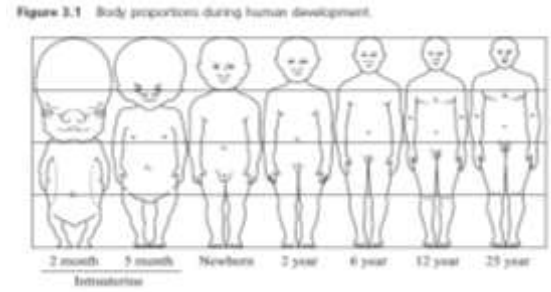


Figure 4.15 Measuring crown–rump length (a) and sitting height (b–d).





Relação SS/SI:

1,7 ao nascimento

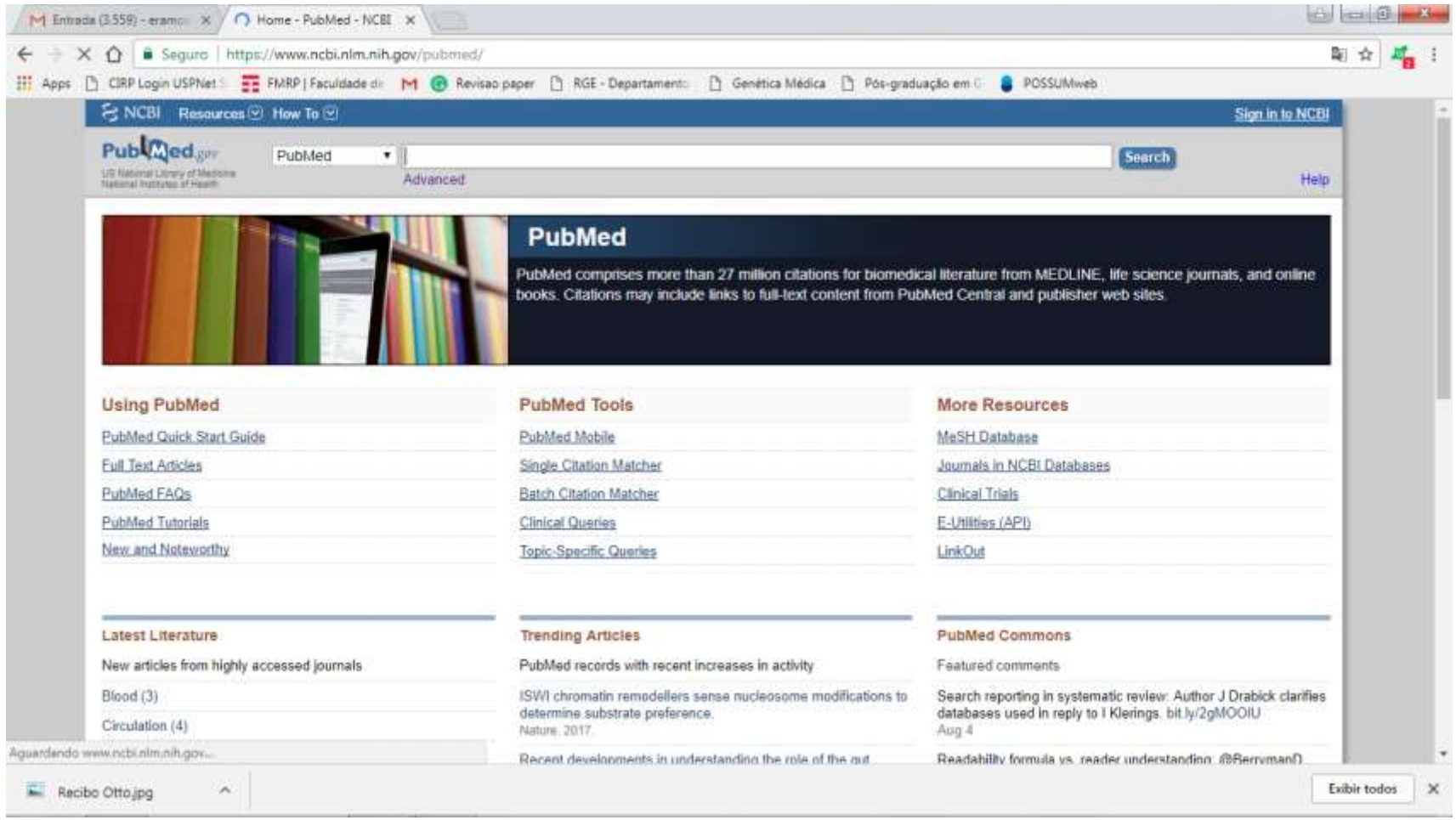
1,0 aos 10 anos

0,9 aos 14 anos

Envergadura

~800 síndromes genéticas com baixa estatura

~50 síndromes com alta estatura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Nature. 2017
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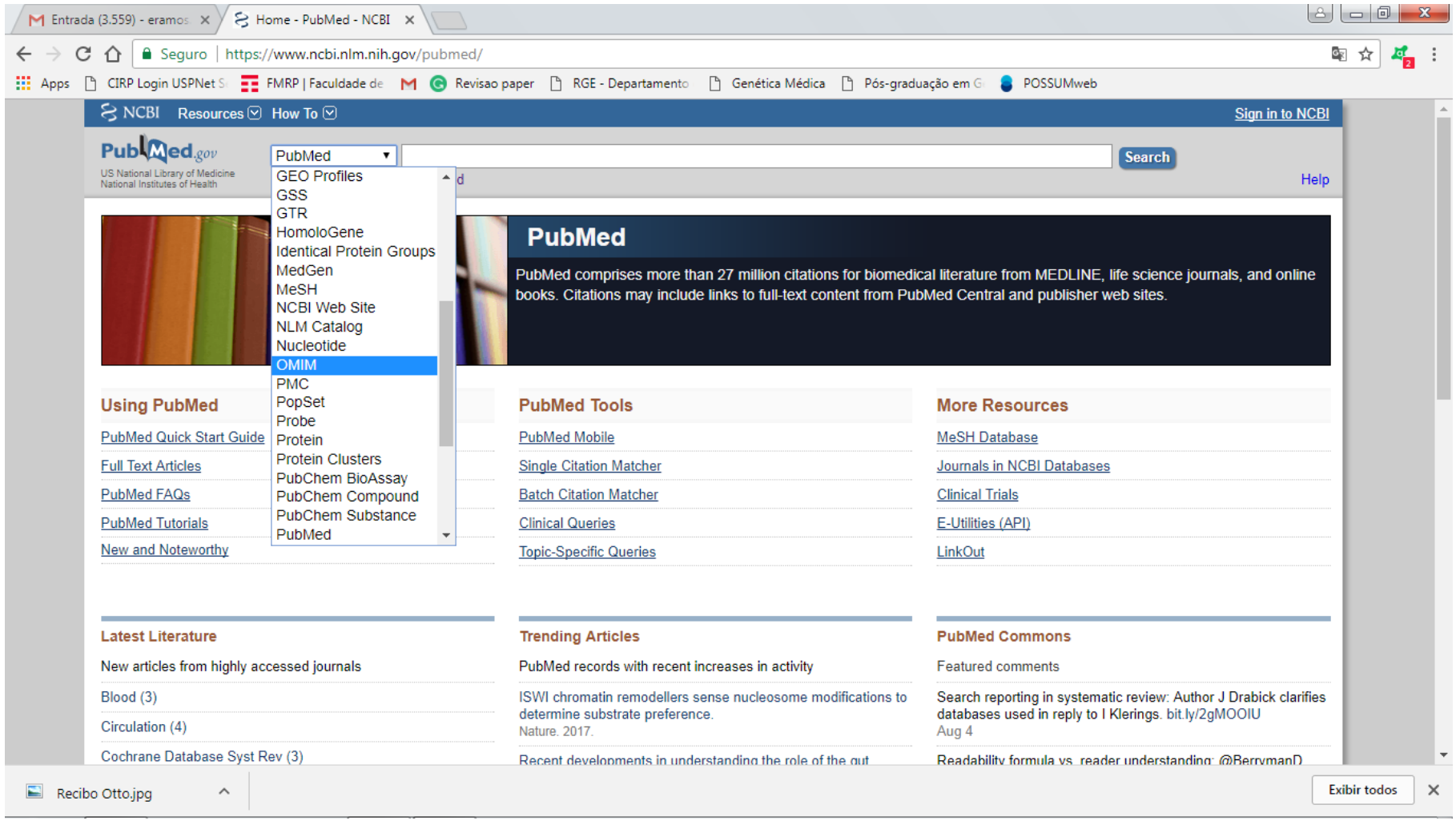
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Cochrane Database Syst Rev (3)
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ISWI chromatin remodellers sense nucleosome modifications to determine substrate preference.
Nature. 2017.
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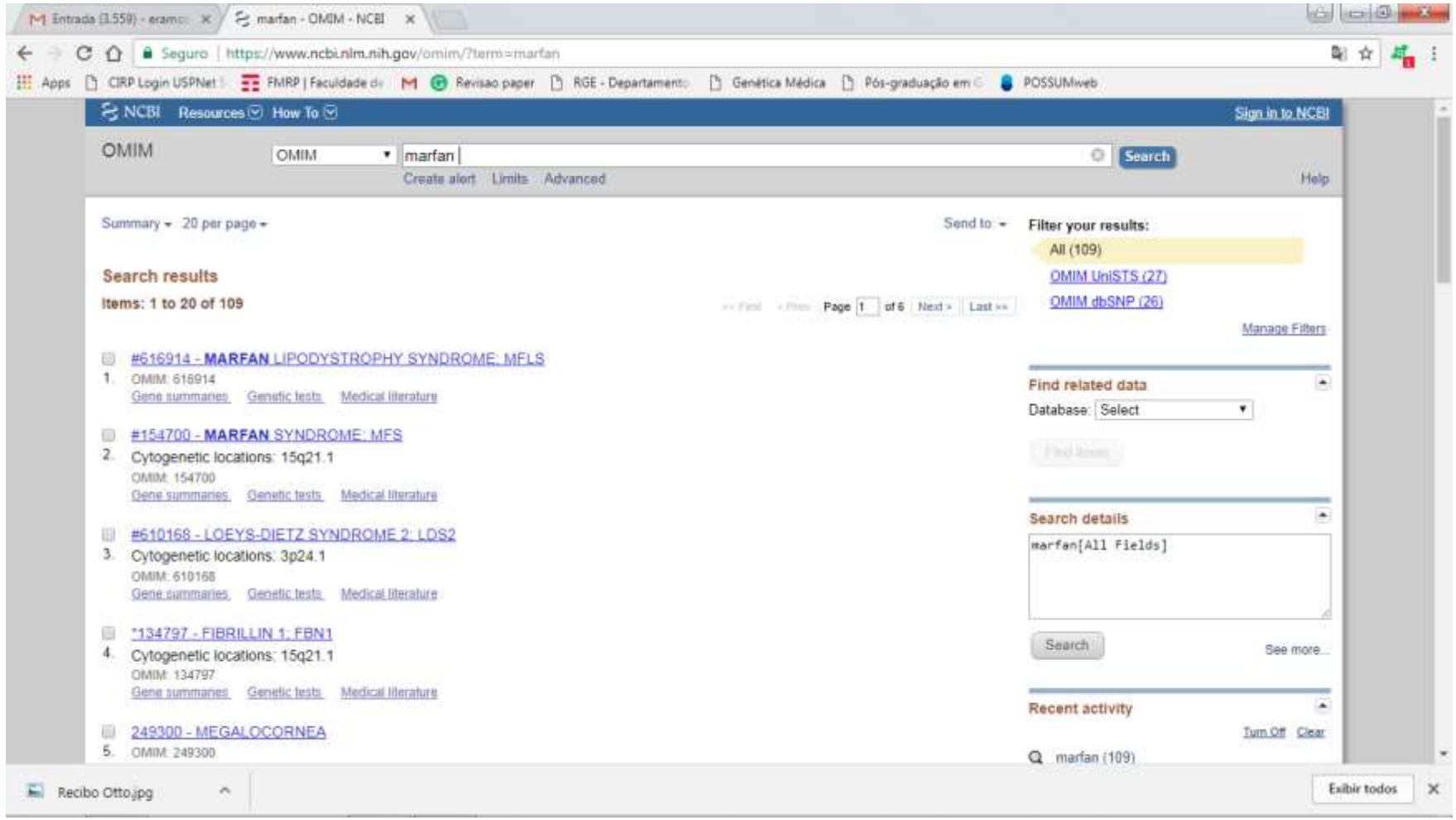
marfan - OMIM - NCBI
https://www.ncbi.nlm.nih.gov/omim/?term=marfan
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Sign in to NCBI
OMIM
OMIM
marfan
Search
Create alert Limits Advanced
Help
Summary 20 per page
Send to
Filter your results:
All (109)
OMIM UniSTS (27)
OMIM dbSNP (26)
Manage Filters
Search results
Items: 1 to 20 of 109
Page 1 of 6 Next Last
#616914 - MARFAN LIPODYSTROPHY SYNDROME; MFLS
1. OMIM: 616914
Gene summaries Genetic tests Medical literature
#154700 - MARFAN SYNDROME; MFS
2. Cytogenetic locations: 15q21.1
OMIM: 154700
Gene summaries Genetic tests Medical literature
#610168 - LOEYS-DIETZ SYNDROME 2; LDS2
3. Cytogenetic locations: 3p24.1
OMIM: 610168
Gene summaries Genetic tests Medical literature
*134797 - FIBRILLIN 1; FBN1
4. Cytogenetic locations: 15q21.1
OMIM: 134797
Gene summaries Genetic tests Medical literature
249300 - MEGALOCORNEA
5. OMIM: 249300
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Search details
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Search
See more...
Recent activity
Turn Off Clear
marfan (109)
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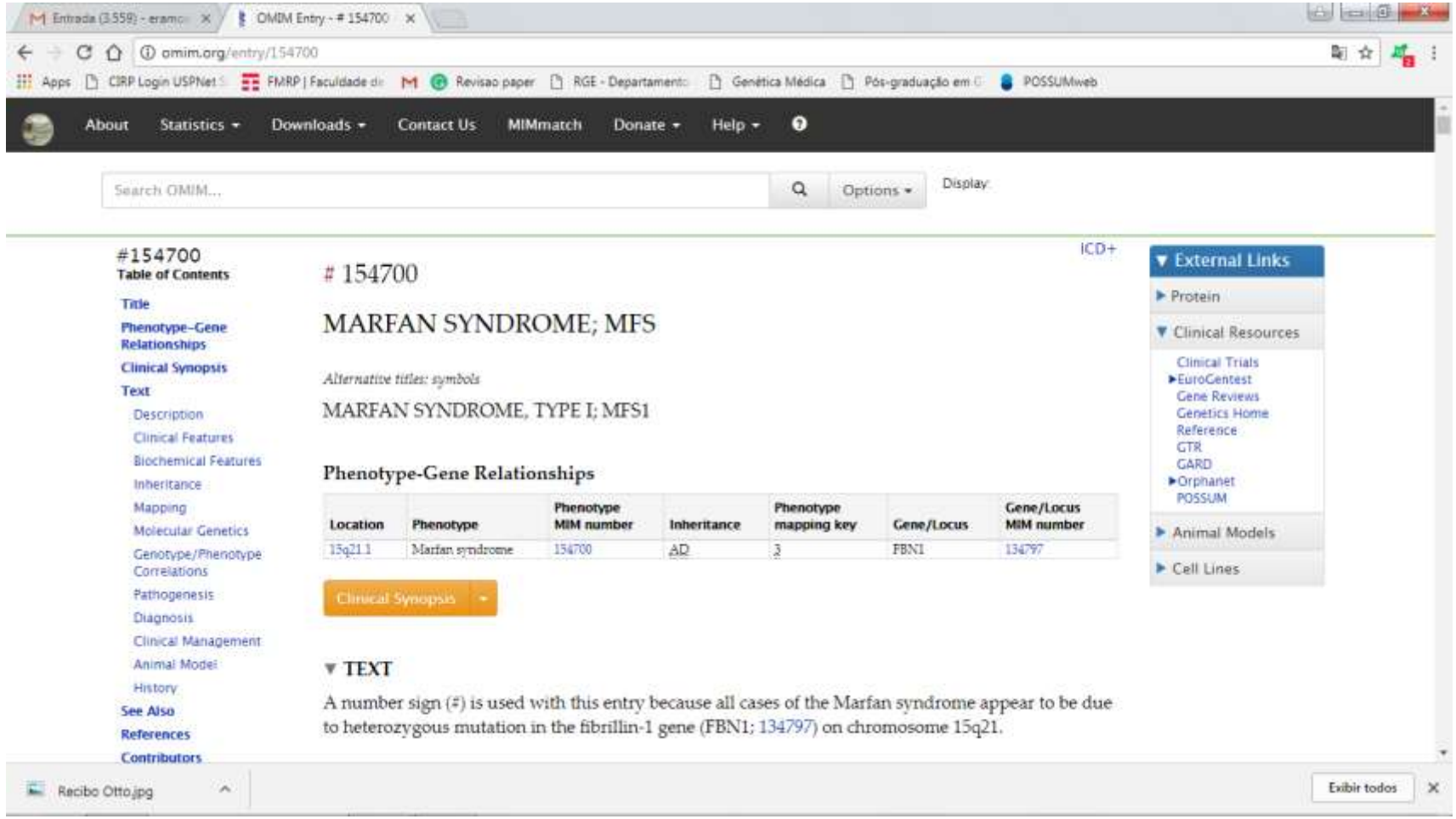
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Clinical Synopsis
Text
Description
Clinical Features
Biochemical Features
Inheritance
Mapping
Molecular Genetics
Genotype/Phenotype Correlations
Pathogenesis
Diagnosis
Clinical Management
Animal Model
History
See Also
References
Contributors
ICD+
154700
MARFAN SYNDROME; MFS
Alternative titles; symbols
MARFAN SYNDROME, TYPE I; MFS1
Phenotype-Gene Relationships
Location
Phenotype
Phenotype MIM number
Inheritance
Phenotype mapping key
Gene/Locus
Gene/Locus MIM number
15q21.1
Marfan syndrome
154700
AD
3
FBN1
134797
Clinical Synopsis
TEXT
A number sign (#) is used with this entry because all cases of the Marfan syndrome appear to be due to heterozygous mutation in the fibrillin-1 gene (FBN1; 134797) on chromosome 15q21.
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Clinical Trials
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Gene Reviews
Genetics Home Reference
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GARD
Orphanet
POSSUM
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Cell Lines
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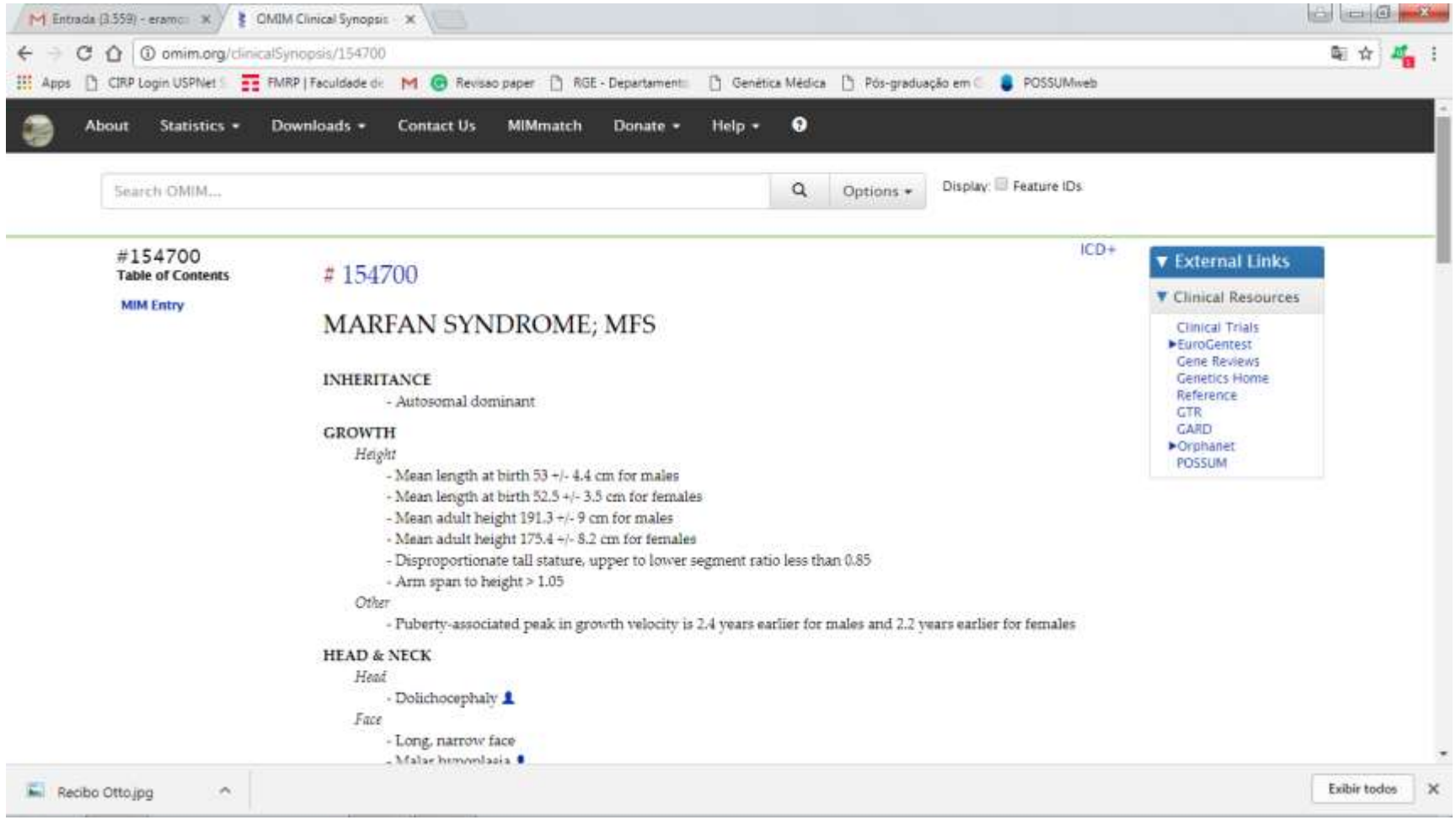
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Table of Contents
MIM Entry
ICD+
154700
MARFAN SYNDROME; MFS
INHERITANCE
- Autosomal dominant
GROWTH
Height
- Mean length at birth 53 +/- 4.4 cm for males
- Mean length at birth 52.5 +/- 3.5 cm for females
- Mean adult height 191.3 +/- 9 cm for males
- Mean adult height 175.4 +/- 8.2 cm for females
- Disproportionate tall stature, upper to lower segment ratio less than 0.85
- Arm span to height > 1.05
Other
- Puberty-associated peak in growth velocity is 2.4 years earlier for males and 2.2 years earlier for females
HEAD & NECK
Head
- Dolichocephaly
Face
- Long, narrow face
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Classificação de Baixa Estatura

BAIXA ESTATURA

VARIANTES NORMAIS
. Baixa Estatura Familial
. Atraso de Crescimento Constitucional

DESPORPORCIONADA
. Displasias Esqueléticas
. Doenças Ósseas Metabólicas

PATOLÓGICAS

PROPORCIONADA

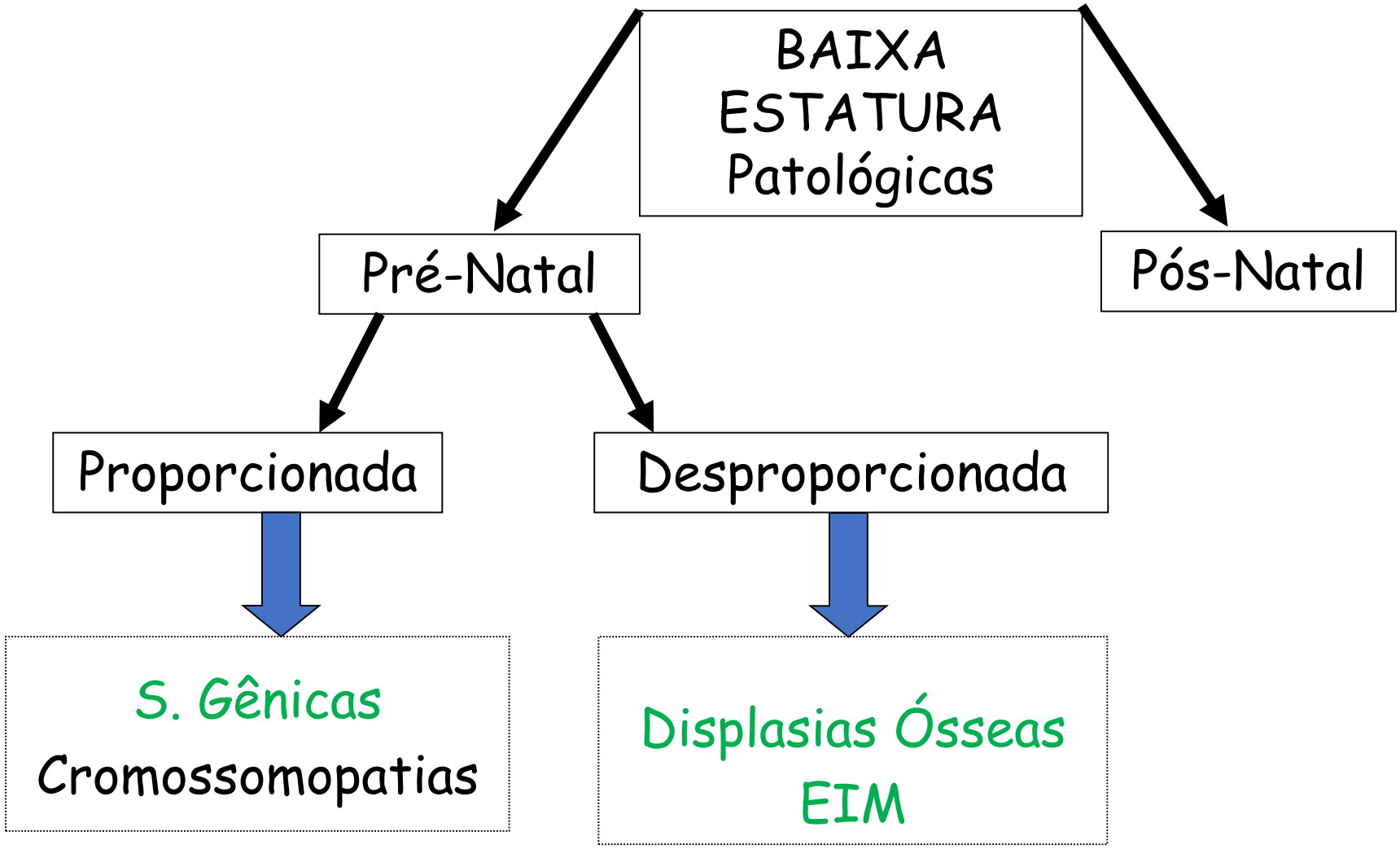
PRÉ-NATAL
. Retardo de Crescimento Intrauterino
- Constrição Fetal
- Disfunção Placentária
- Infecções
- Teratógenos

. Síndromes Dismórficas
. Cromossomopatias

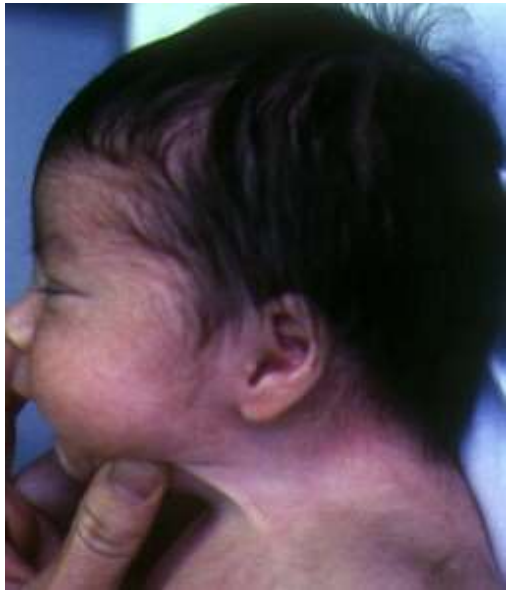
PÓS-NATAL
. Desnutrição
. Doenças Crônicas
- Gastrointestinal
- Cardiopulmonar
- Infecções
- Renal
- Hematológica
- Neurológica

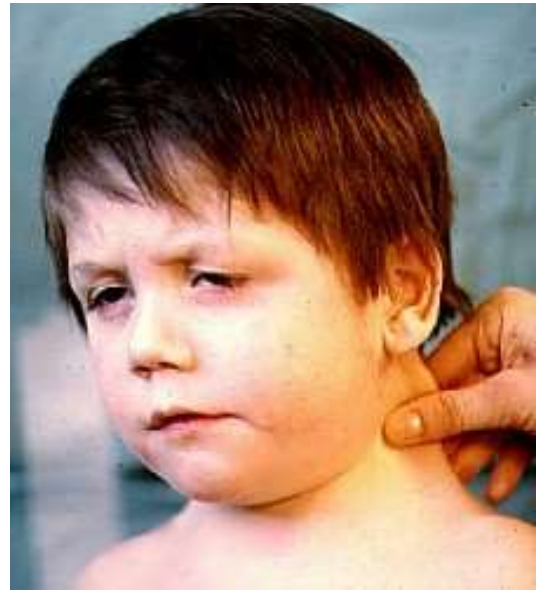
. Drogas
. Nanismo Psicossocial
. Distúrbios Endócrinos

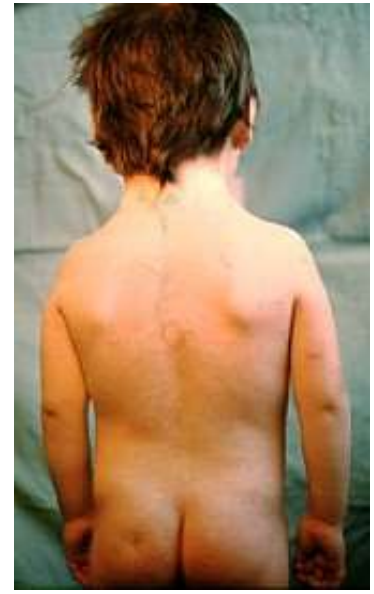
Abordagem clínica - baixa estatura

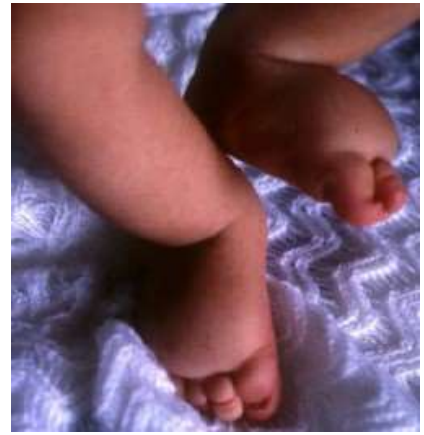


1. Síndrome de Noonan

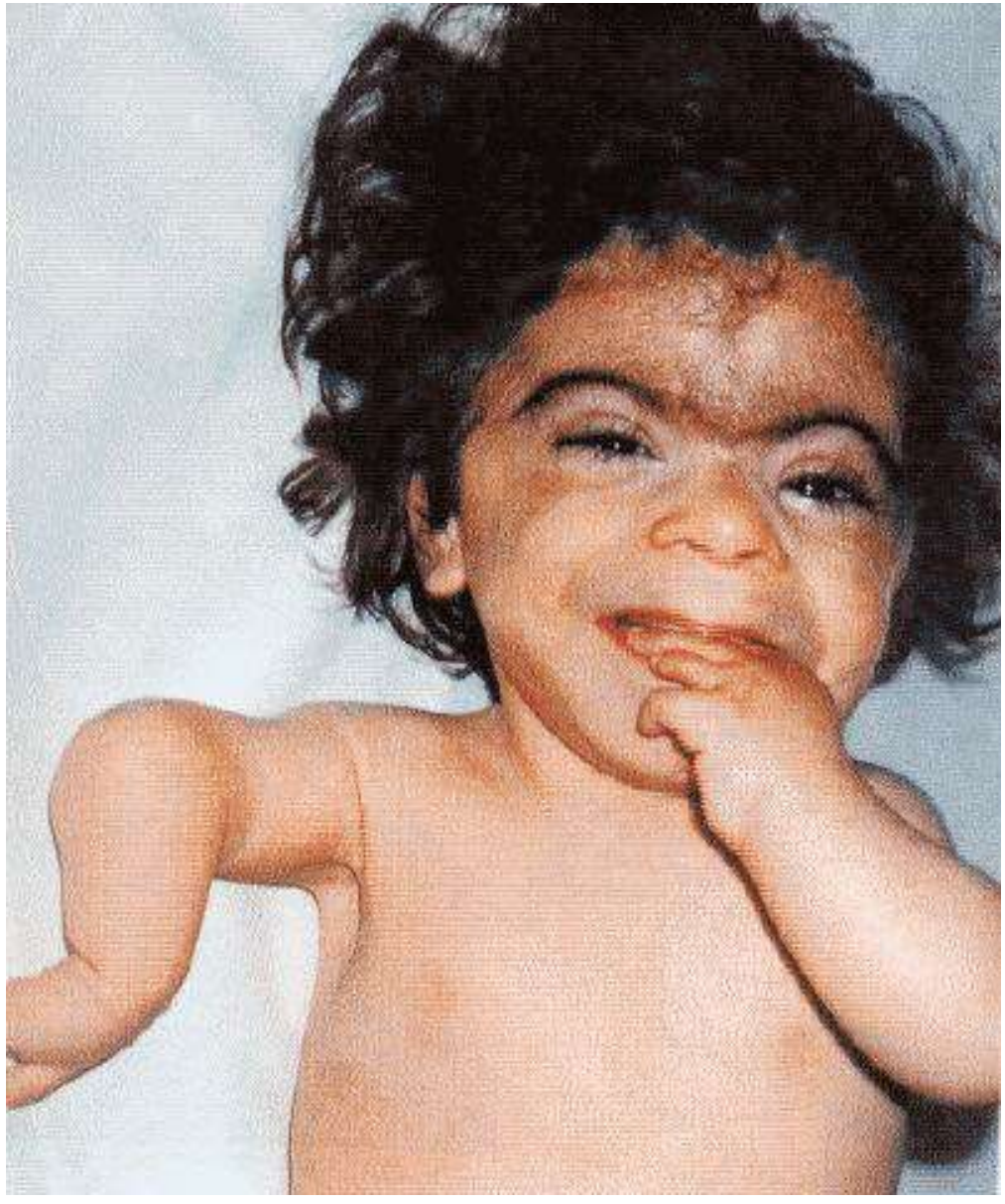


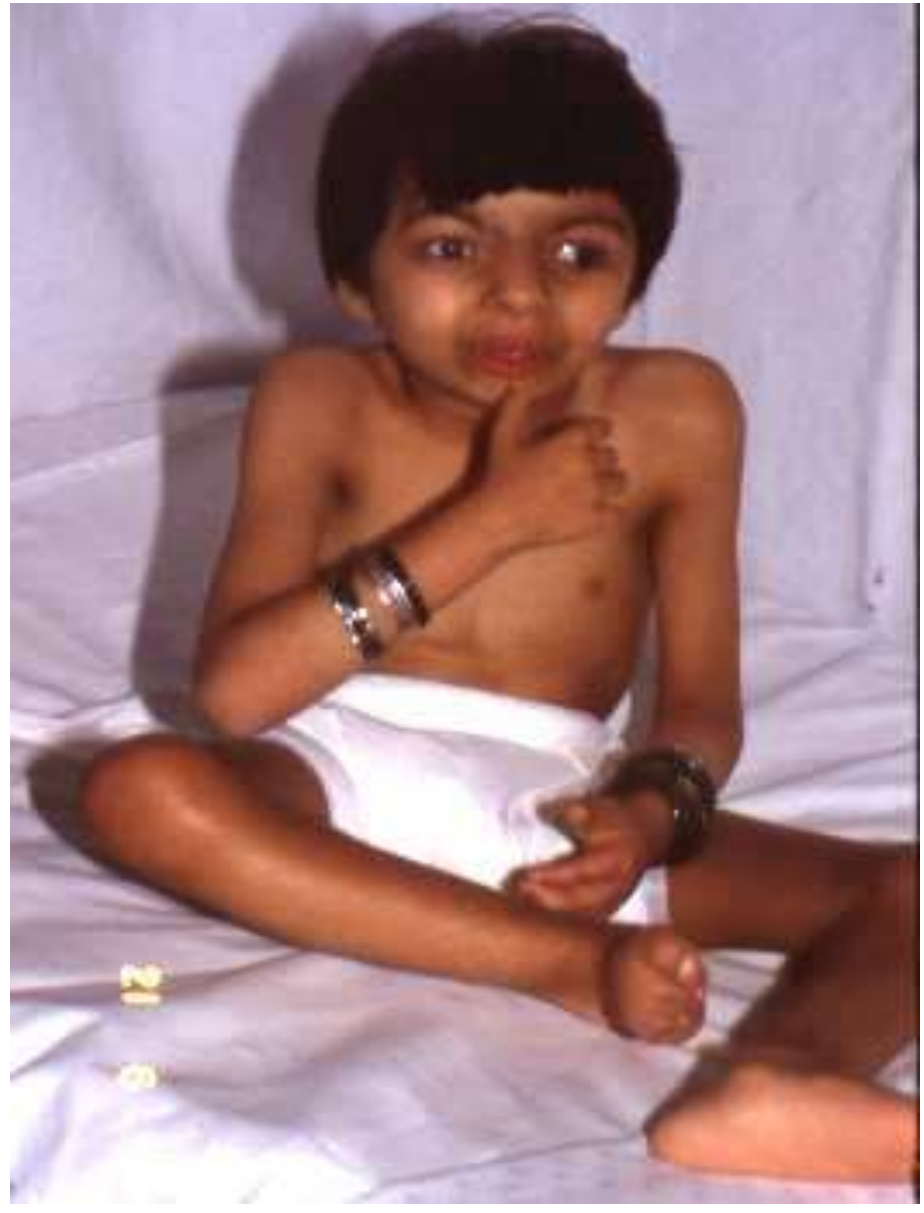






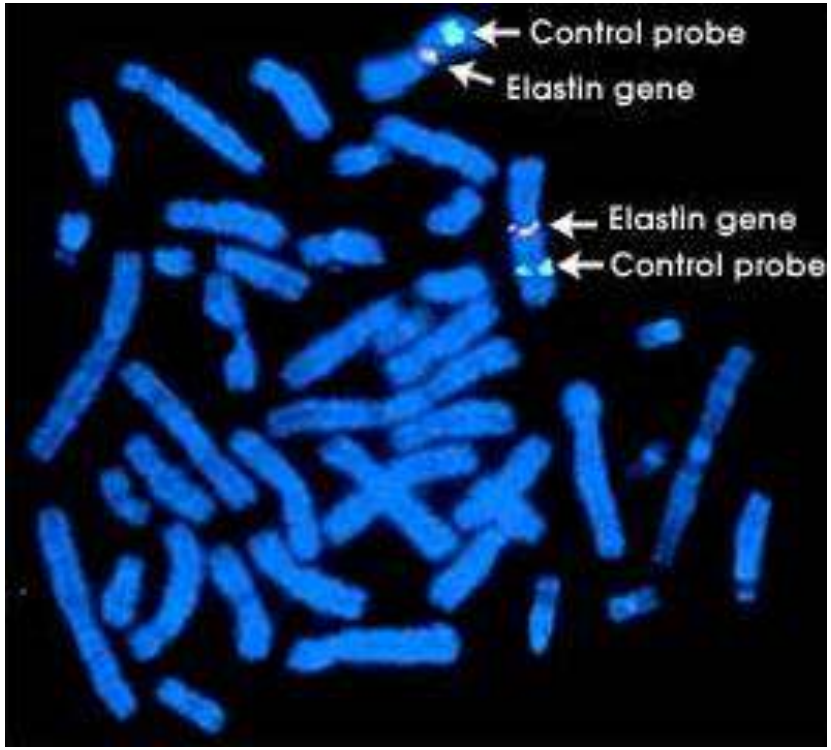
2. S. Cornélia de Lange





4. S. de Williams

3. S. de Williams



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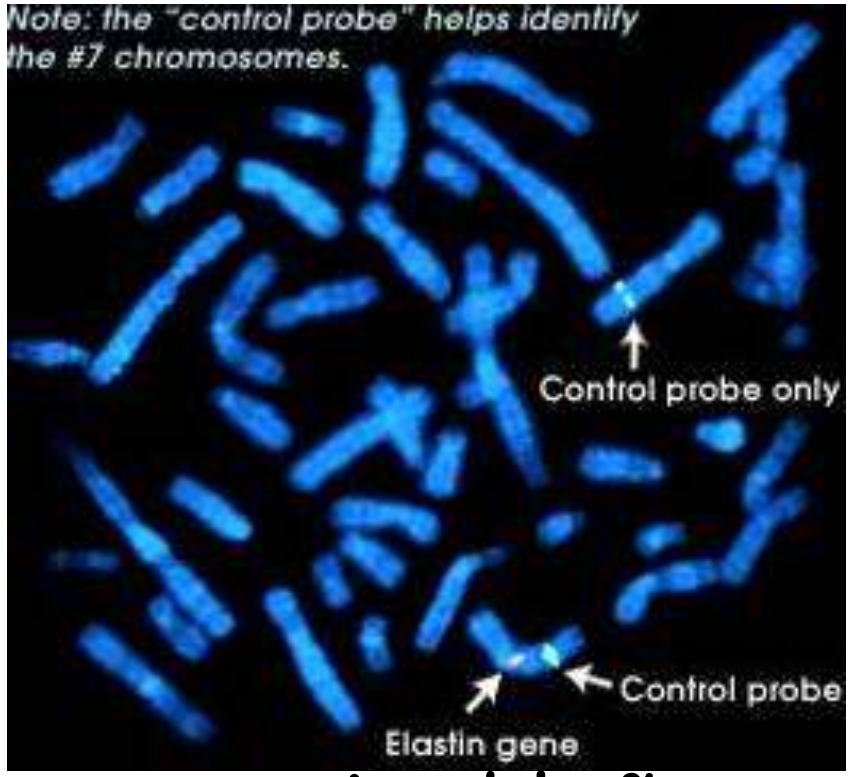
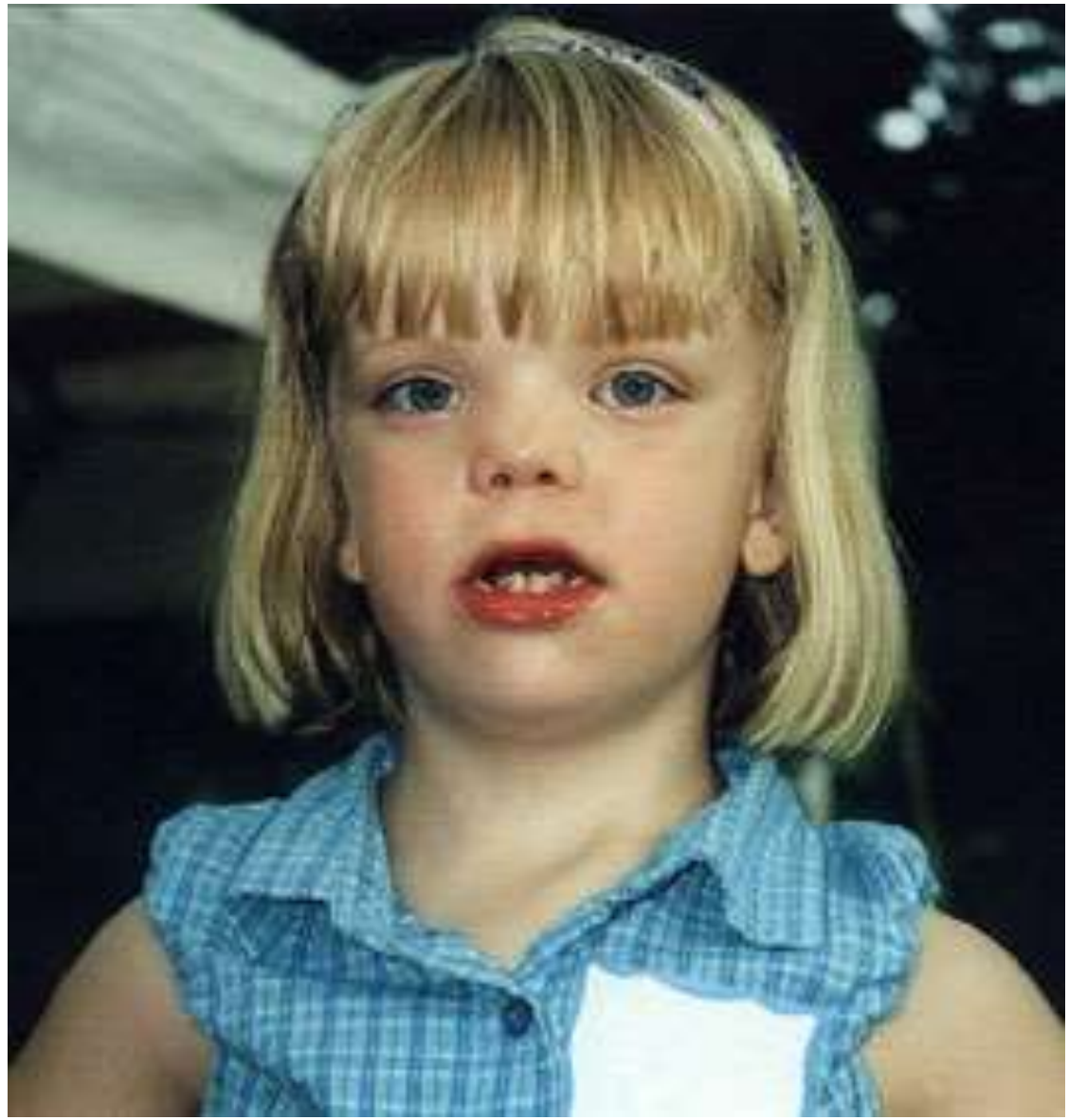


microdeleção

4. S. de Robinow

4. S. de Robinow



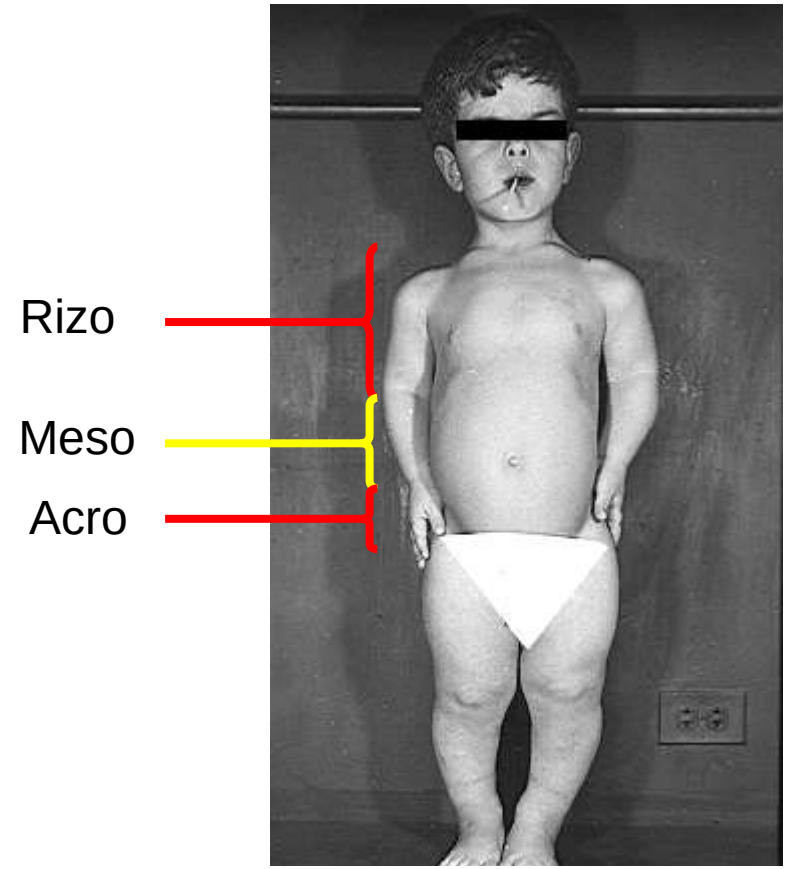
Displasias Ósseas

- Alterações no comprimento, modelagem e densidade óssea que levam à baixa estatura.

RIZOMÉLICO: úmero/fêmur

MESOMÉLICO: rádio-ulna/tíbia-fíbula

ACROMÉLICO: mãos/pés



Acondroplasia

Displasias Ósseas

1. Acondroplasia
2. Hipocondroplasia
3. Displasia Tanatofórica
4. Osteogênese Imperfeita

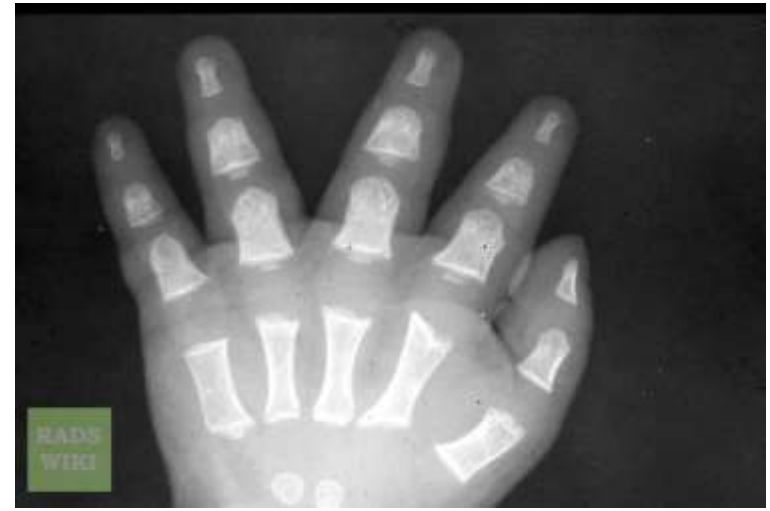
1. Acondroplasia

- Displasia óssea mais frequente, prevalência de 1,5:10.000
- Baixa estatura desproporcionada, com encurtamento rizomélico
- Macrocefalia e hidrocefalia
- Nariz em cela
- Mão em tridente

1. Acondroplasia

- Autossômica dominante, gene *FGFR3*, localizado em 4p16.3
- Formas homozigotas levam a um fenótipo mais acentuado, imcompatível com a vida
- Há compressão do tronco cerebral devido a estreitamento do forame magno⇒aumento da incidência de morte súbita

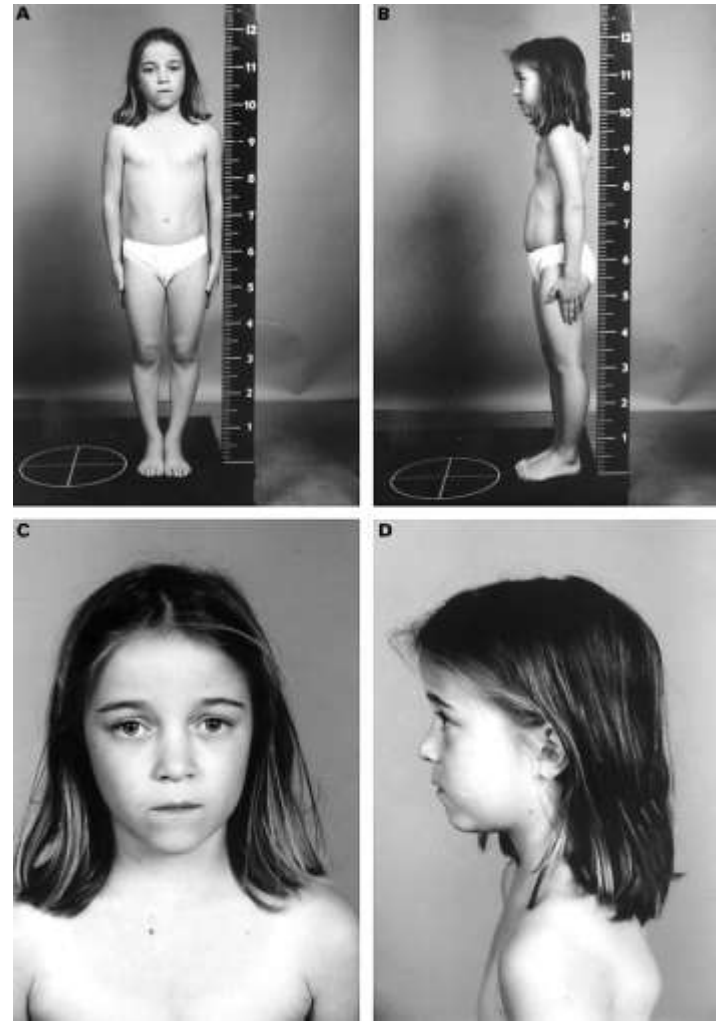

#DomingoShow


RADS
WIKI

2. Hipocondroplasia

- Baixa estatura desproporcionada menos severa que a Acondroplasia
- Encurtamento rizomélico
- Crânio e face normais
- Autossômica dominante, mesmo locus da Acondroplasia

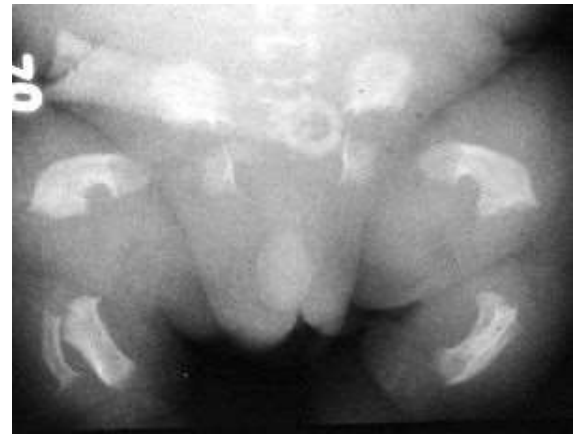
2. Hipocondroplasia

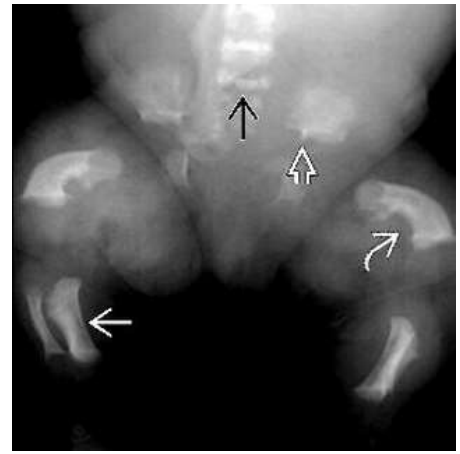


3. Displasia Tanatofórica

- Mais grave de todas as displasias ósseas
- Sempre letal, óbito normalmente intraútero
- Micromelia extrema, macrocefalia, tórax estreito e abdome proeminente
- Todos casos são autossômicos dominantes e representam mutações novas

3. Displasia Tanatofórica





Fêmur - "telefone"

4. Osteogênese Imperfeita

- Principais manifestações clínicas: fragilidade óssea, escleras azuladas, surdez e dentinogênese imperfeita
- Baseado nestas características clínicas podem ser subdivididas em 4 tipos:

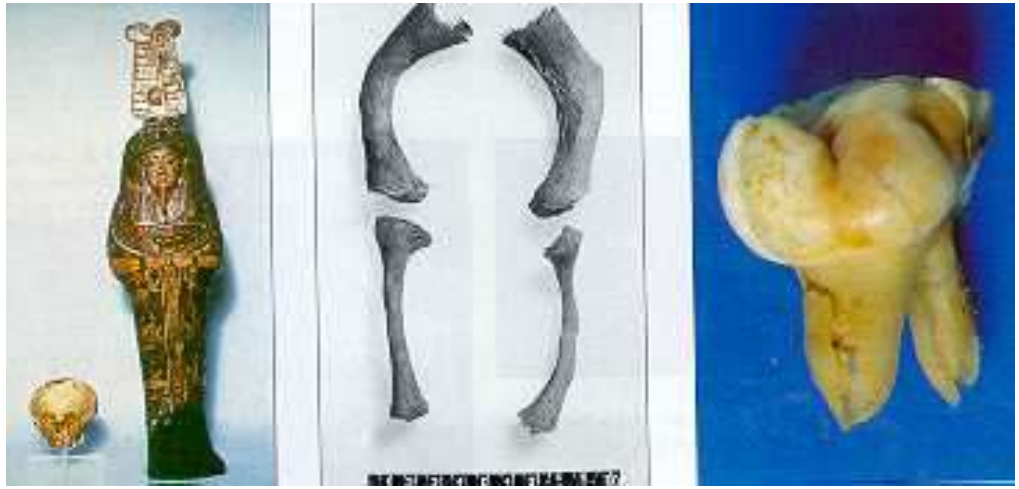
  OI tipo 1 – Forma branda, fragilidade óssea moderada,AD

  OI tipo 2 – 10% dos casos, letal, AD

  OI tipo 3 – Rara, grave, AD e AR

  OI tipo 4 – Rara, expressão variável, AD

Osteogênese Imperfeita



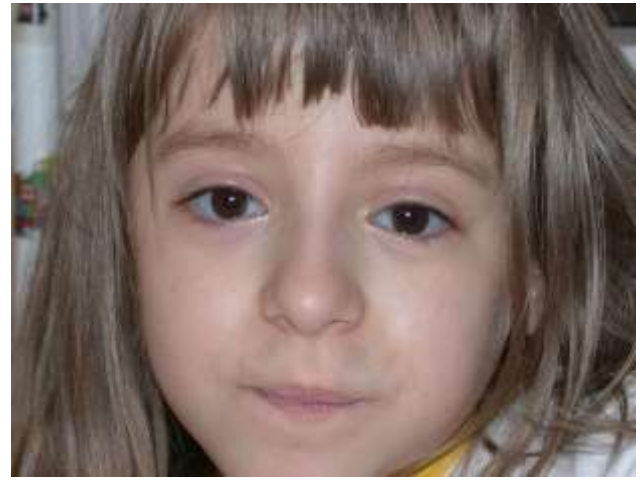
ABOi

ASSOCIAÇÃO BRASILEIRA DE OSTEOGENESIS IMPERFECTA

Fundada em 11 de dezembro de 1999

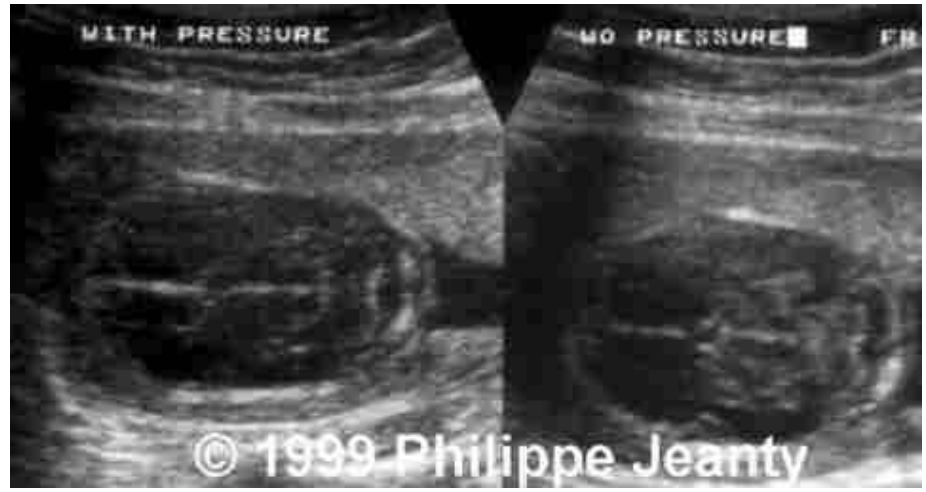
http://www.aboi.org.br

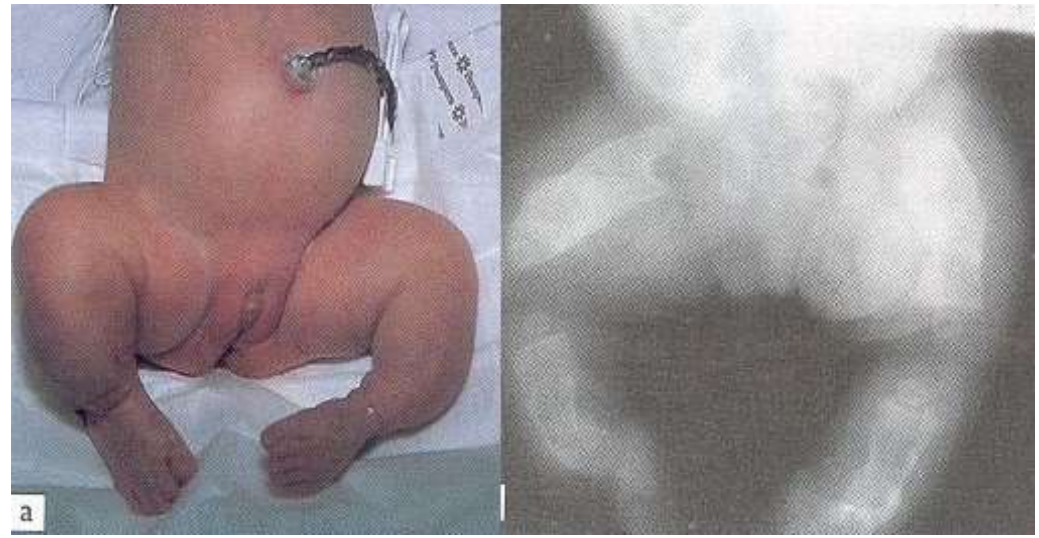
http://www.oif.org/



Esclera azulada

(toda a vida)


WITH PRESSURE
NO PRESSURE
© 1999 Philippe Jeanty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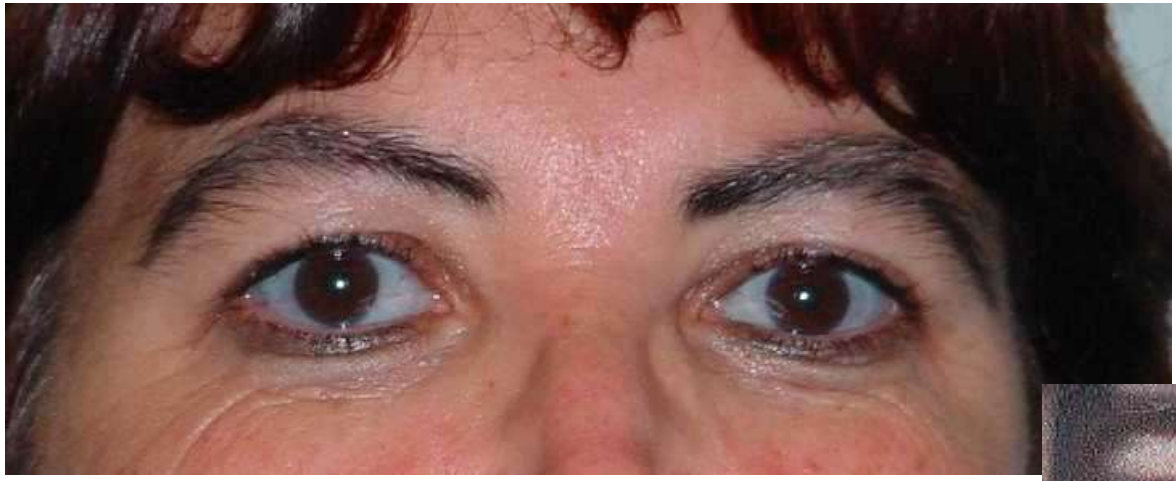
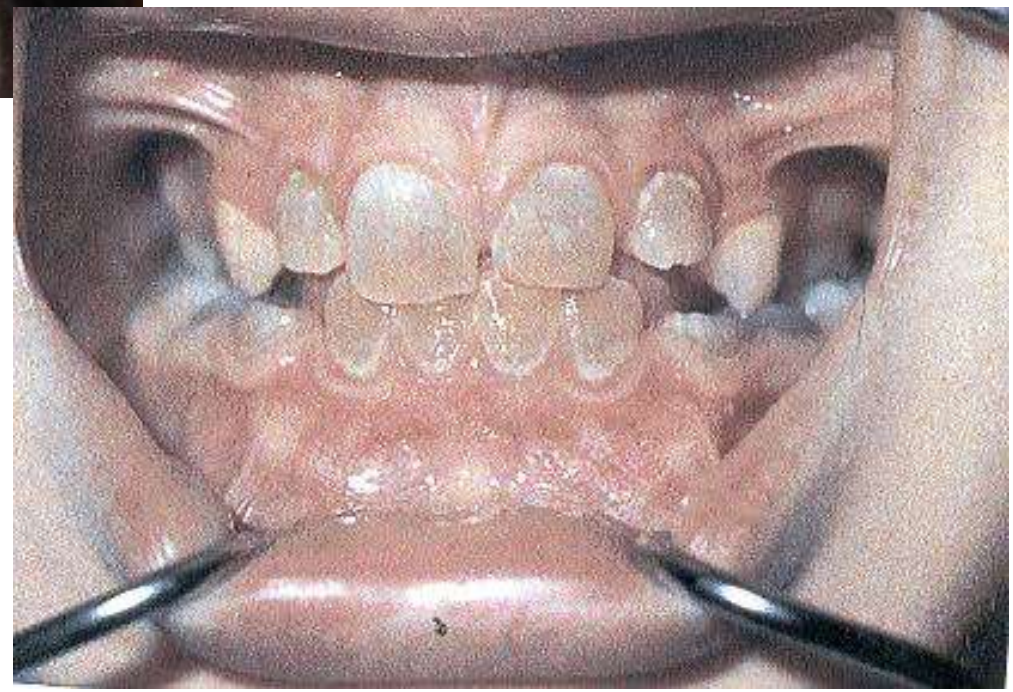


Esclera azulada

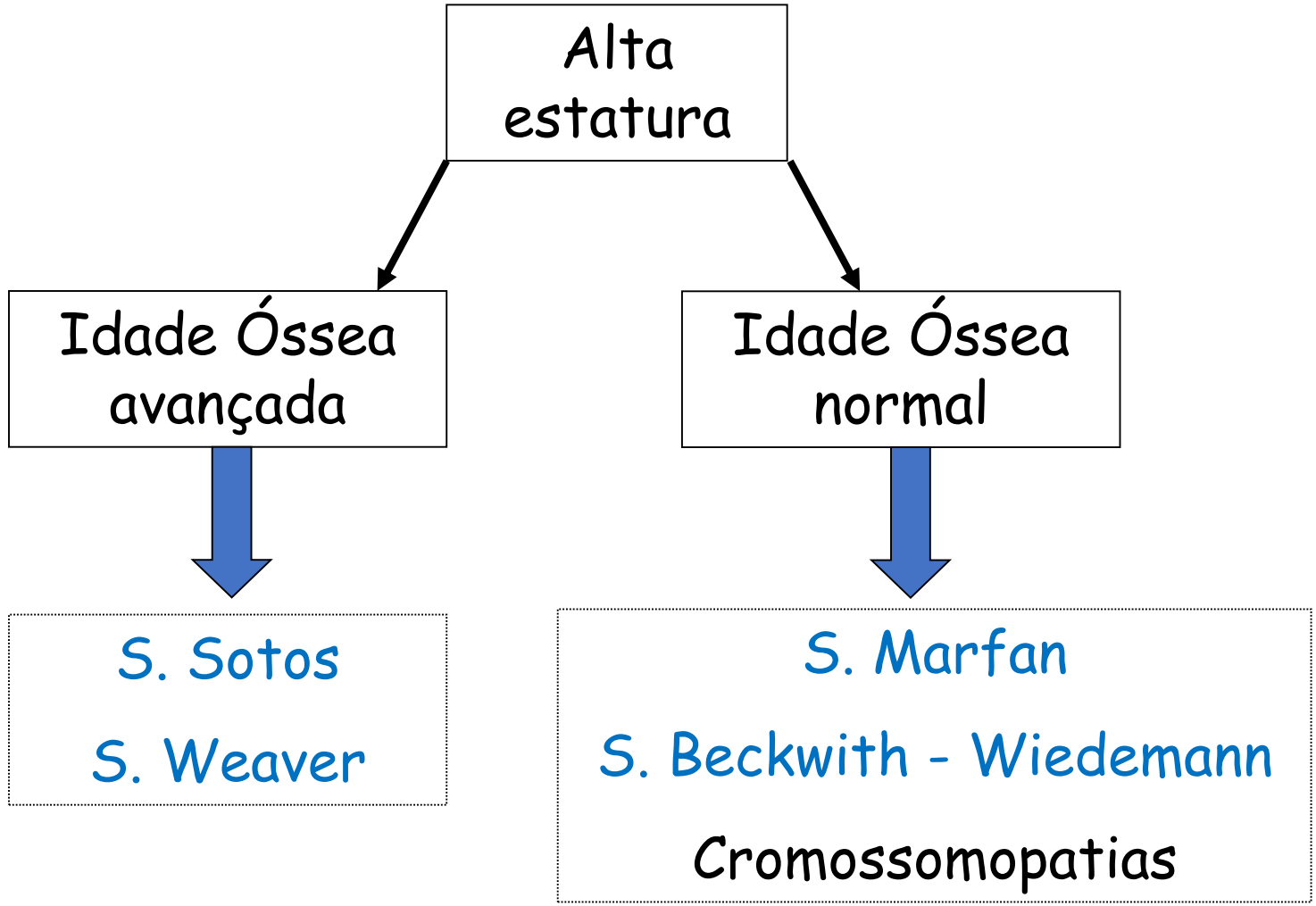


Dentinogênese
Imperfeita

Hipercrescimento

- RNs >4kg
- Excluir mãe diabéticas (5% dos RNs)

Abordagem clínica - alta estatura (macrossomia)



S. Sotos

- S. Gigantismo Cerebral

- Alta estatura pré-natal, com velocidade de crescimento excessivo nos primeiros 4 anos de vida

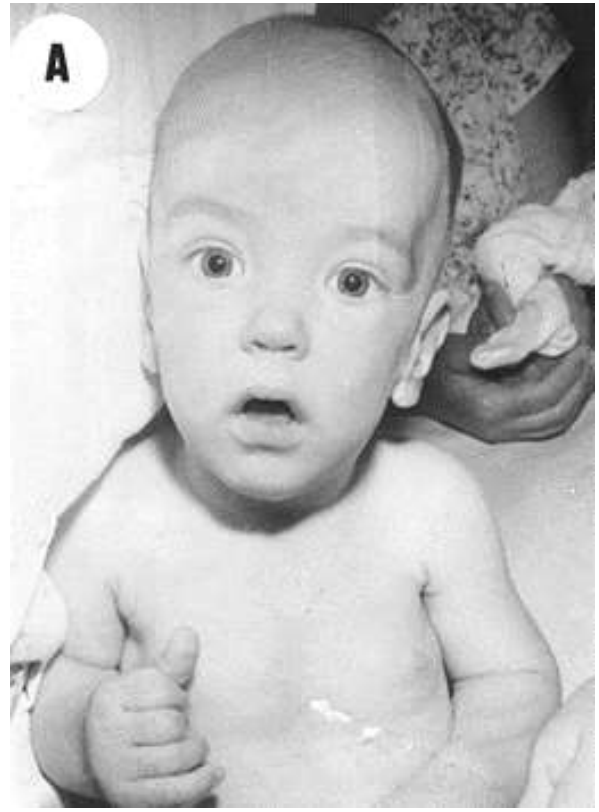
- IO avançada

- Deficit intelectual leve a moderado em 85%,

  restante - inteligência normal

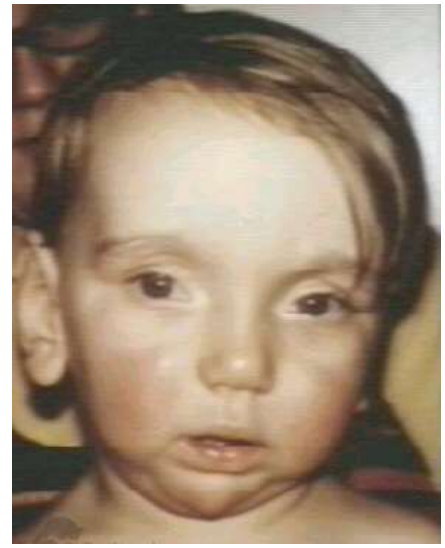
- Macrocefalia, fenda palpebral oblíqua para baixo, queixo proeminente

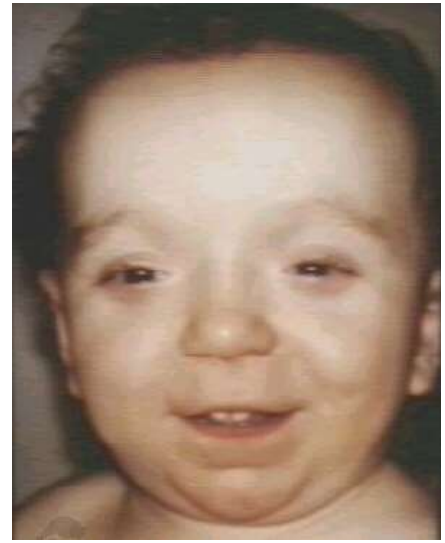
- Ventriculomegalia e alterações estruturais do SNC



S. Weaver

- Alta estatura pré-natal
- Idade óssea avançada
- Deficit intelectual moderado a grave em 80%, restantes - inteligência normal
- Macrocefalia, micrognatia importante
- Camptodactila em mãos, Hipertonia
- Casos esporádicos

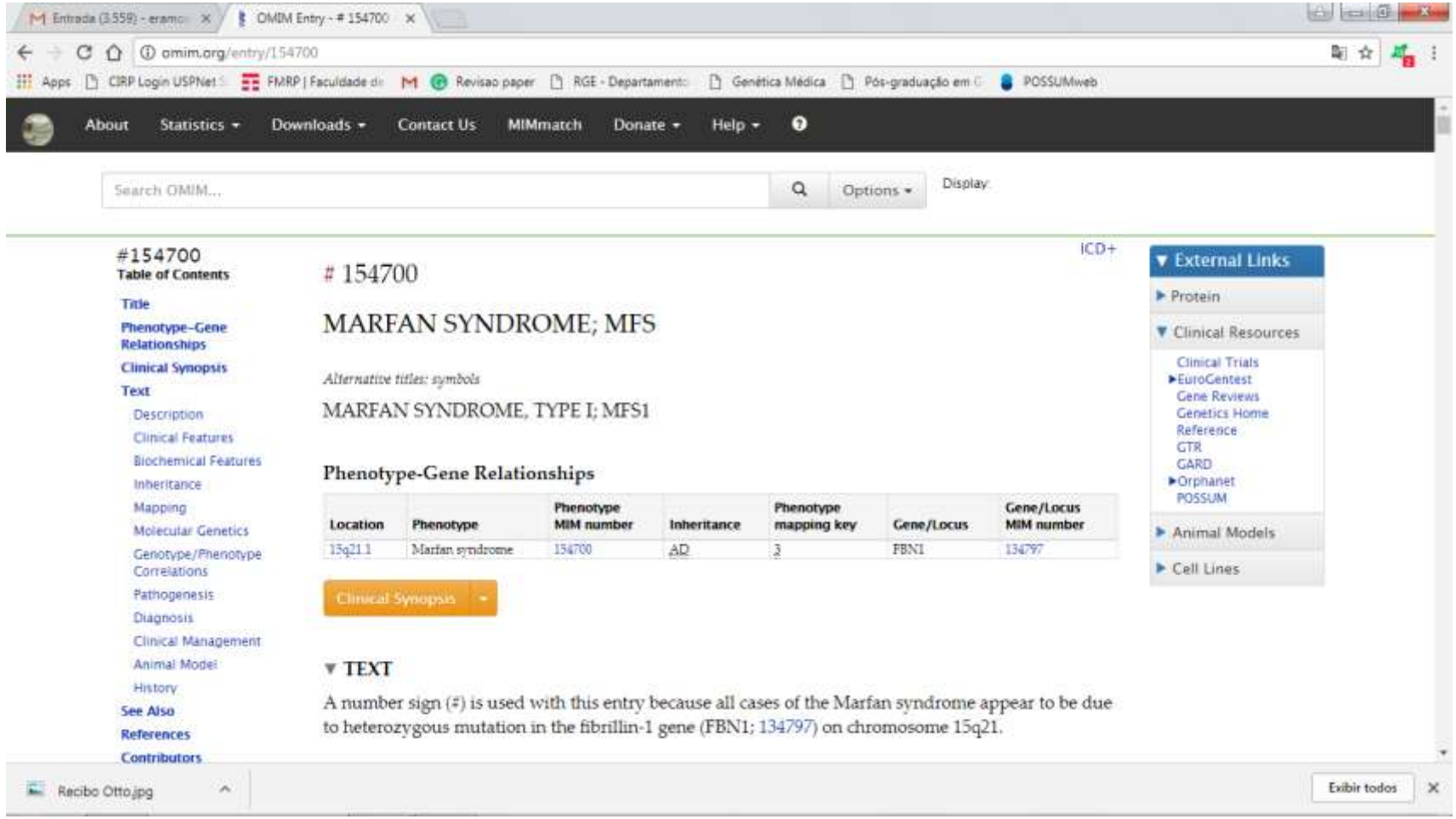


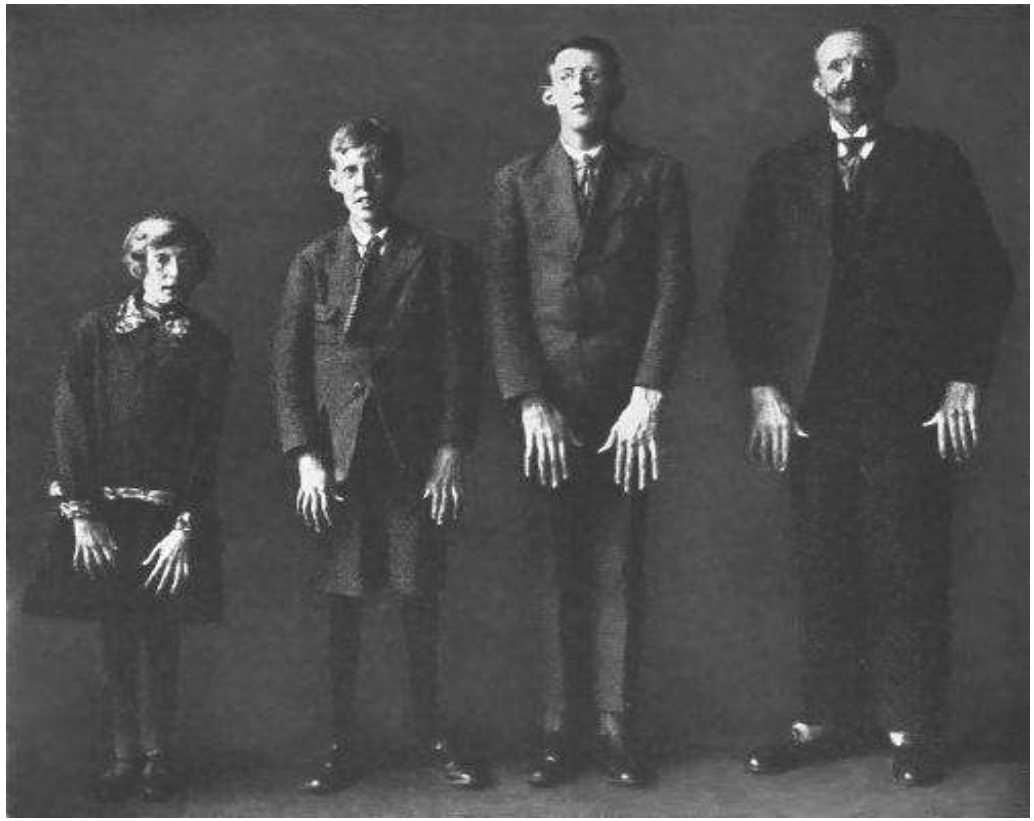


S. Marfan

- 1:10.000 pessoas

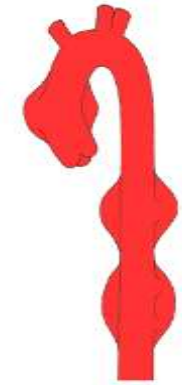
- Longelíneo com alta estatura
- Autossômica dominante


Entrada (3.559) - eramo
OMIM Entry - # 154700
omim.org/entry/154700
Apps
CIRP Login USPNet
FMRP | Faculdade de
Revisao paper
RGE - Departament
Genética Médica
Pós-graduação em G
POSSUMweb
About
Statistics
Downloads
Contact Us
MIMmatch
Donate
Help
Search OMIM...
Options
Display
#154700
Table of Contents
Title
Phenotype-Gene Relationships
Clinical Synopsis
Text
Description
Clinical Features
Biochemical Features
Inheritance
Mapping
Molecular Genetics
Genotype/Phenotype Correlations
Pathogenesis
Diagnosis
Clinical Management
Animal Model
History
See Also
References
Contributors
ICD+
154700
MARFAN SYNDROME; MFS
Alternative titles; symbols
MARFAN SYNDROME, TYPE I; MFS1
Phenotype-Gene Relationships
Location
Phenotype
Phenotype MIM number
Inheritance
Phenotype mapping key
Gene/Locus
Gene/Locus MIM number
15q21.1
Marfan syndrome
154700
AD
3
FBN1
134797
Clinical Synopsis
TEXT
A number sign (#) is used with this entry because all cases of the Marfan syndrome appear to be due to heterozygous mutation in the fibrillin-1 gene (FBN1; 134797) on chromosome 15q21.
External Links
Protein
Clinical Resources
Clinical Trials
EuroGentest
Gene Reviews
Genetics Home Reference
GTR
GARD
Orphanet
POSSUM
Animal Models
Cell Lines
Recibo Otto.jpg
Exibir todos



S. Marfan
(AD)

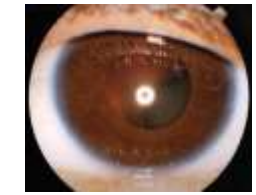
Alta estatura



Dilatação de aorta

Aracnodactilia

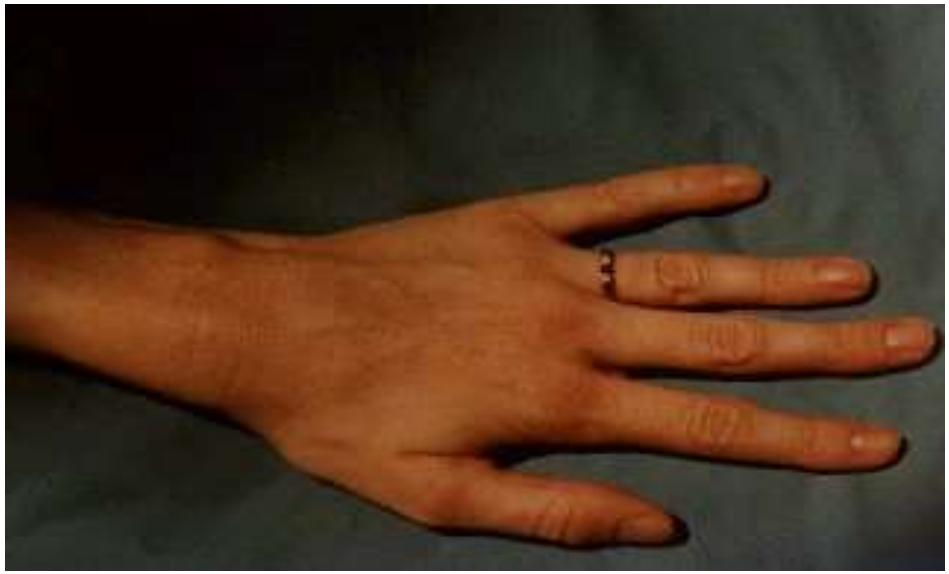
Frouxidão ligamentar

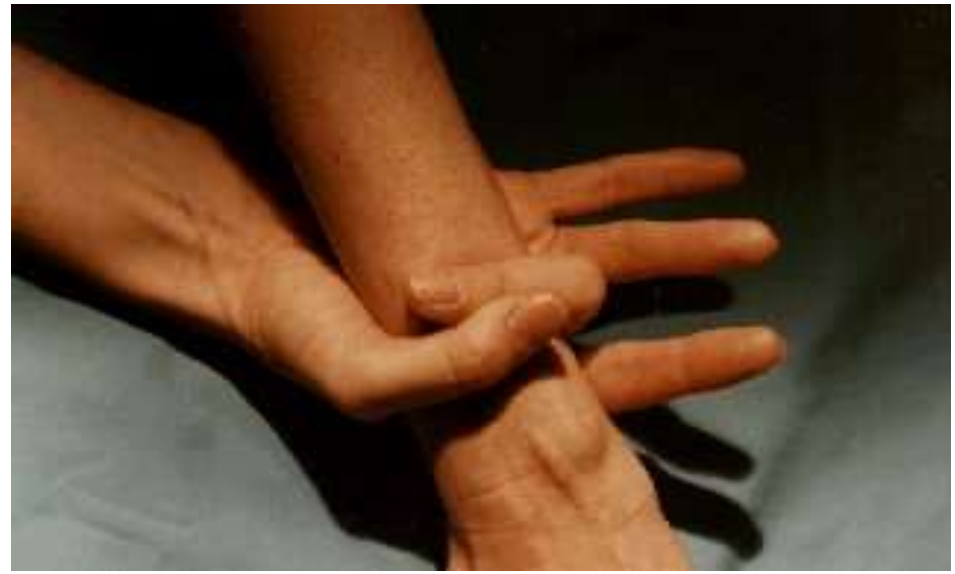


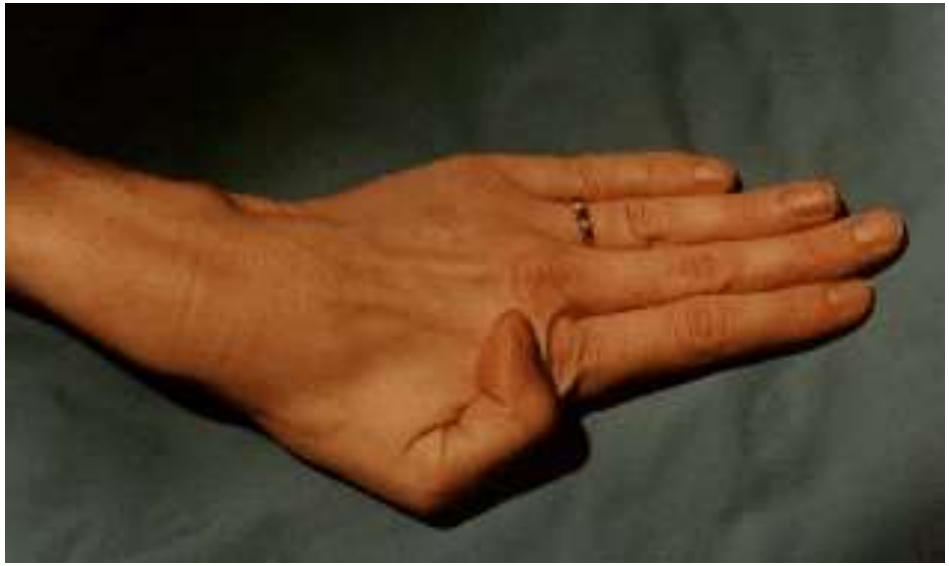
Miopia
Luxação do cristalino

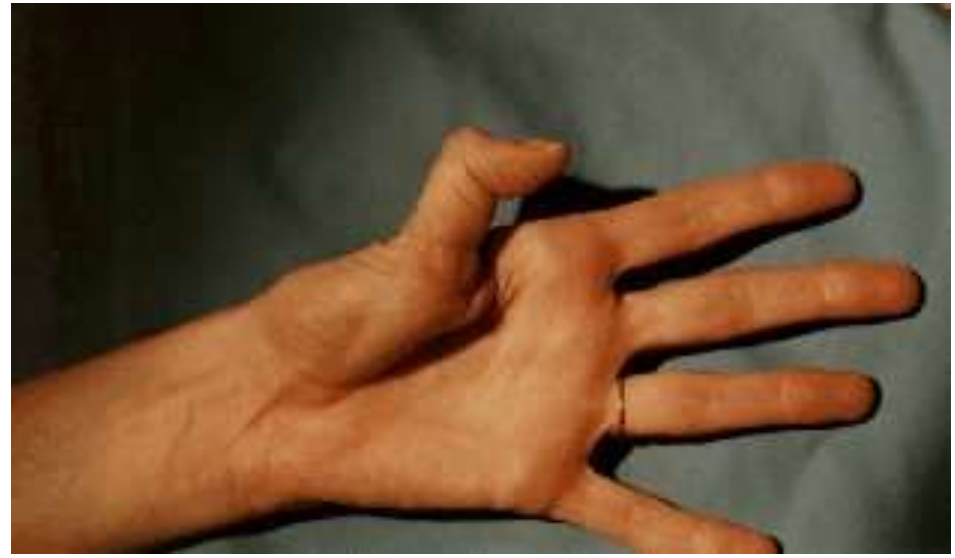

MOVERS

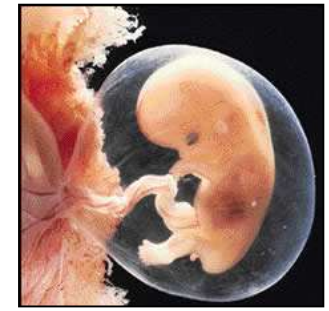
S. Marfan

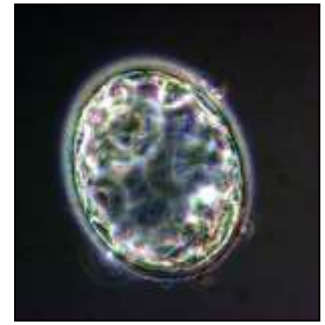








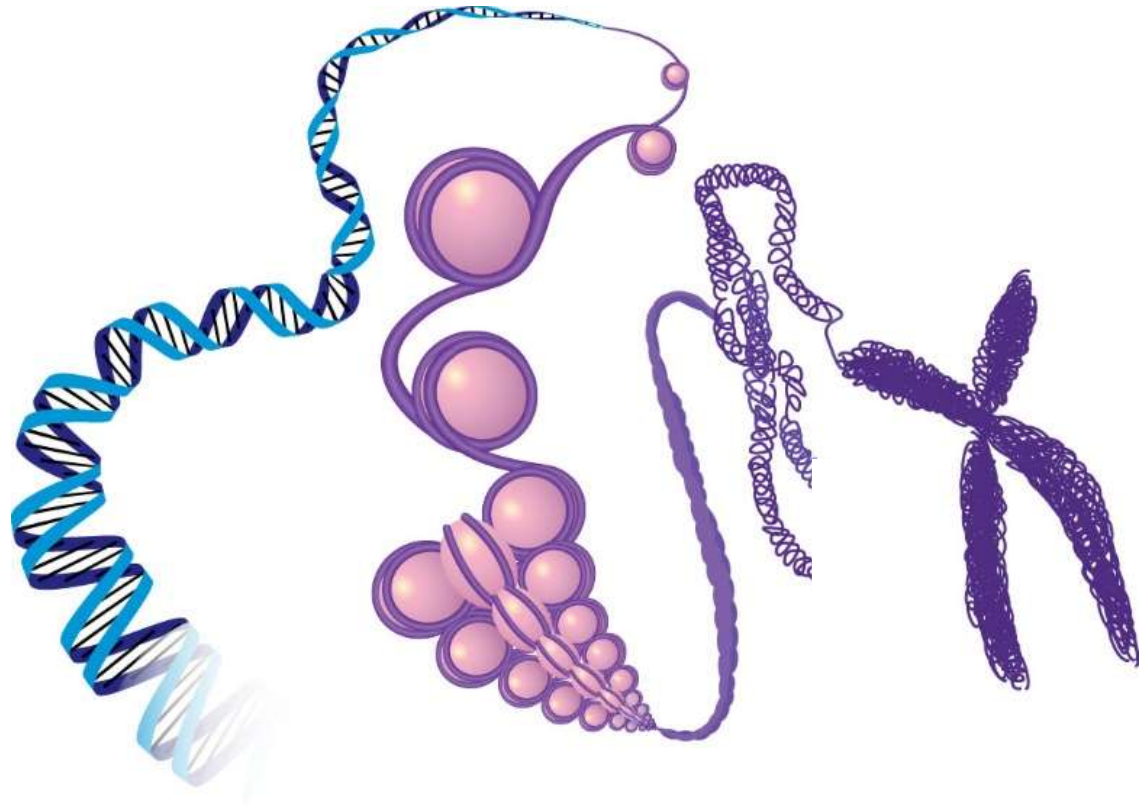




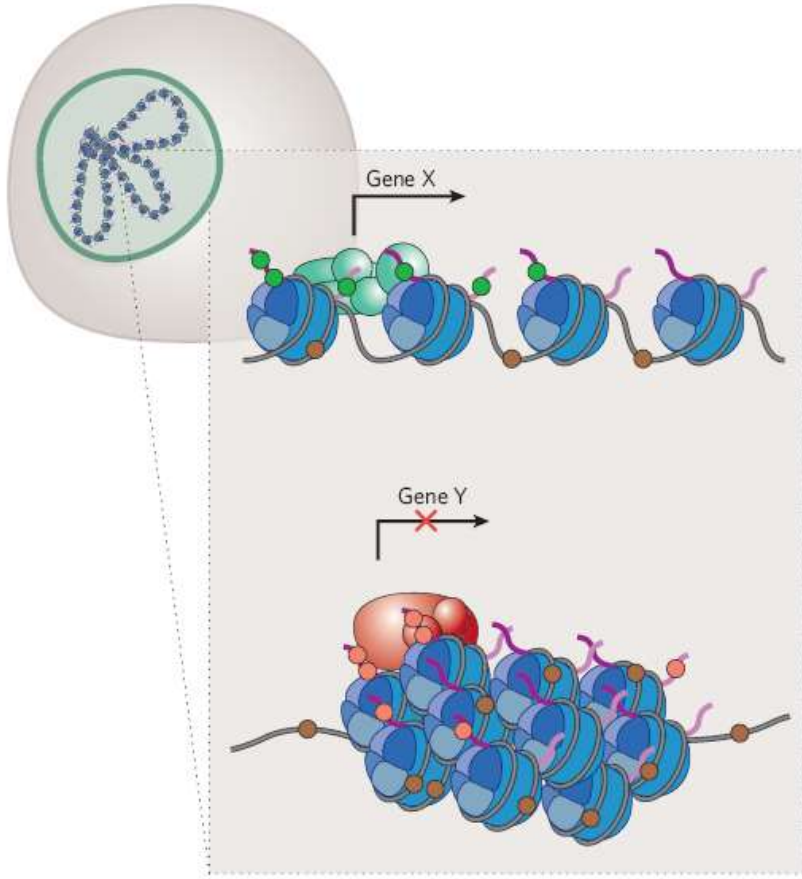
Epigenetics

Tissue (daughter cells)
Development

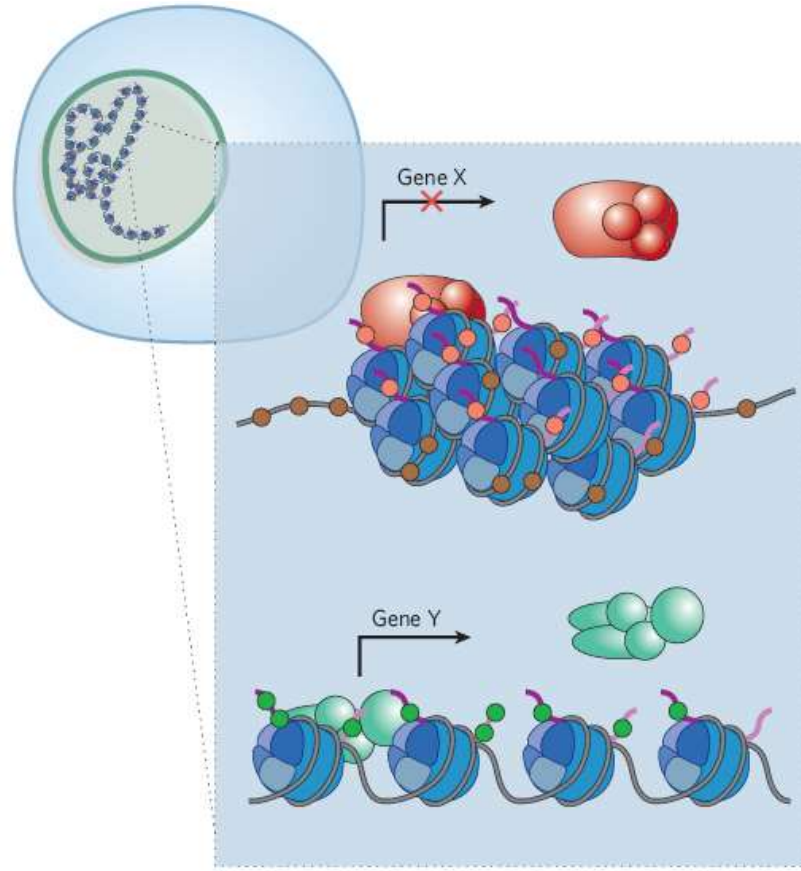
Epigenetics



“Epimutations”



Normal



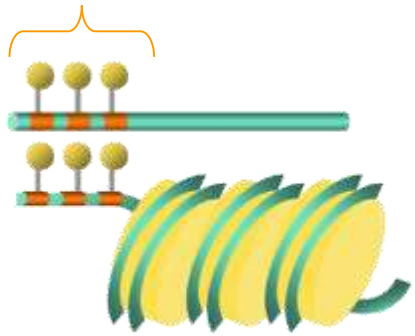
“Epimutation”

Queen Worker

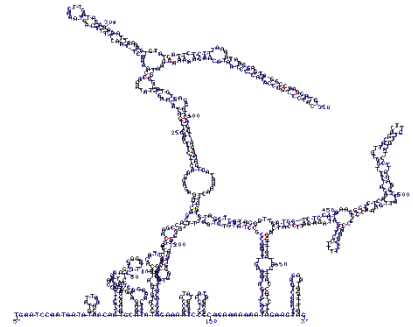
Royal jelly

Dutch famine

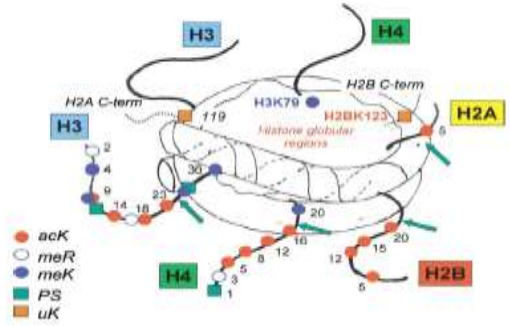
Epigenetics



DNA methylation

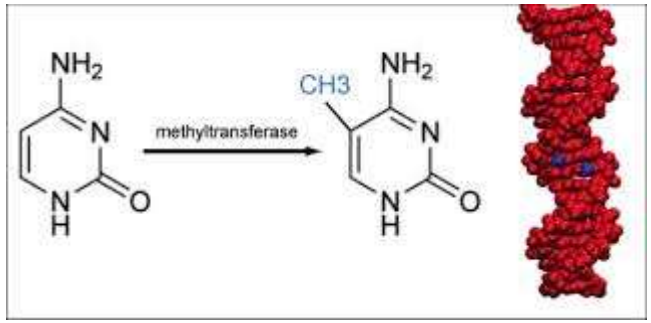


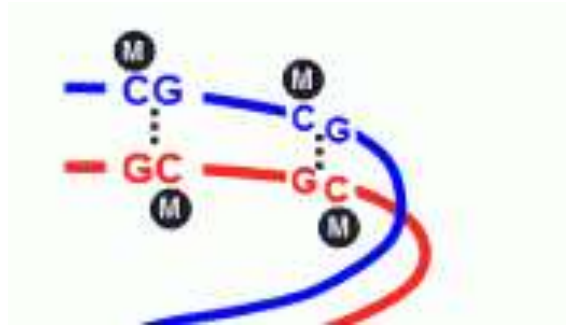
ncRNAs



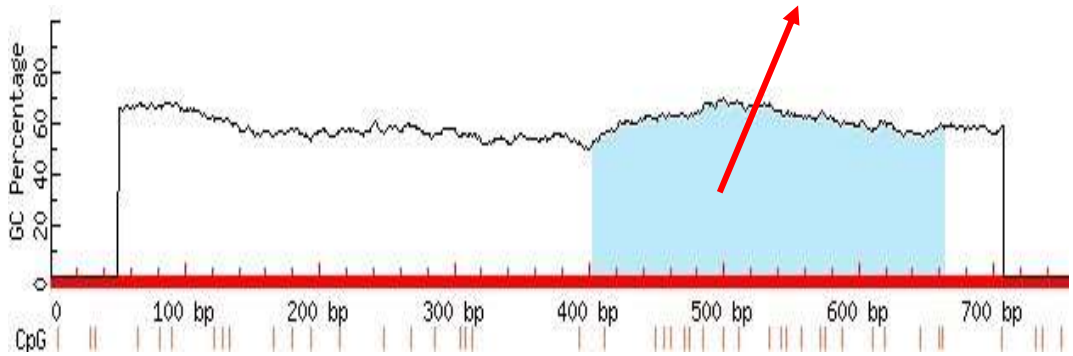
Histone modifications

DNA methylation



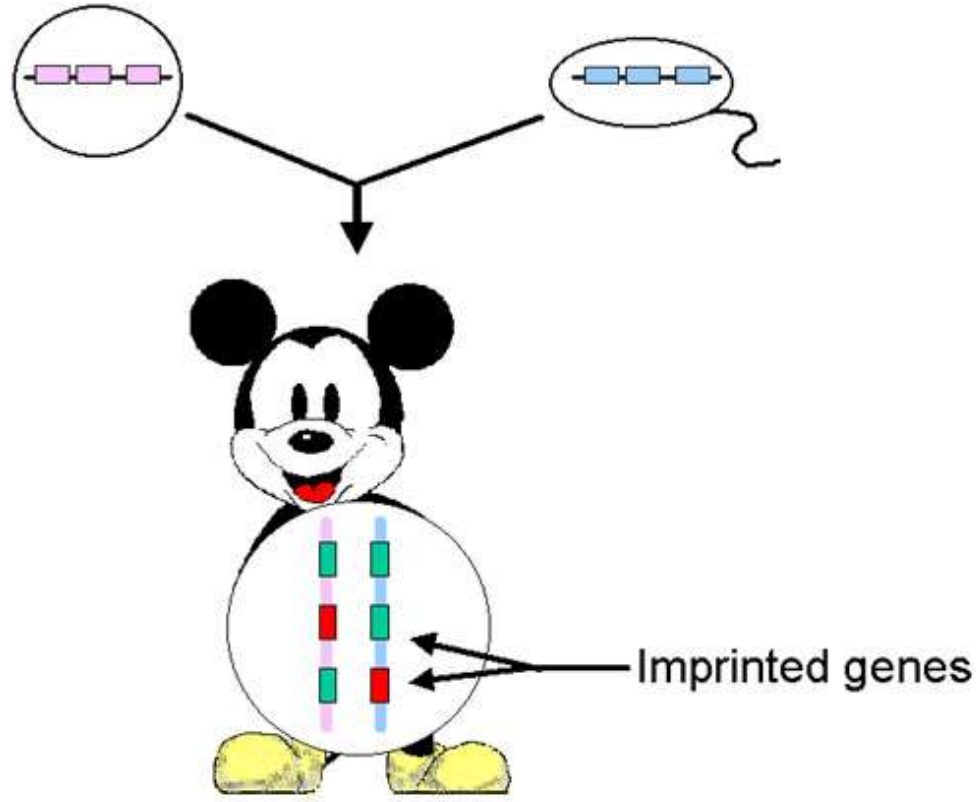


CpG Island



Epigenetics

Genomic Imprinting

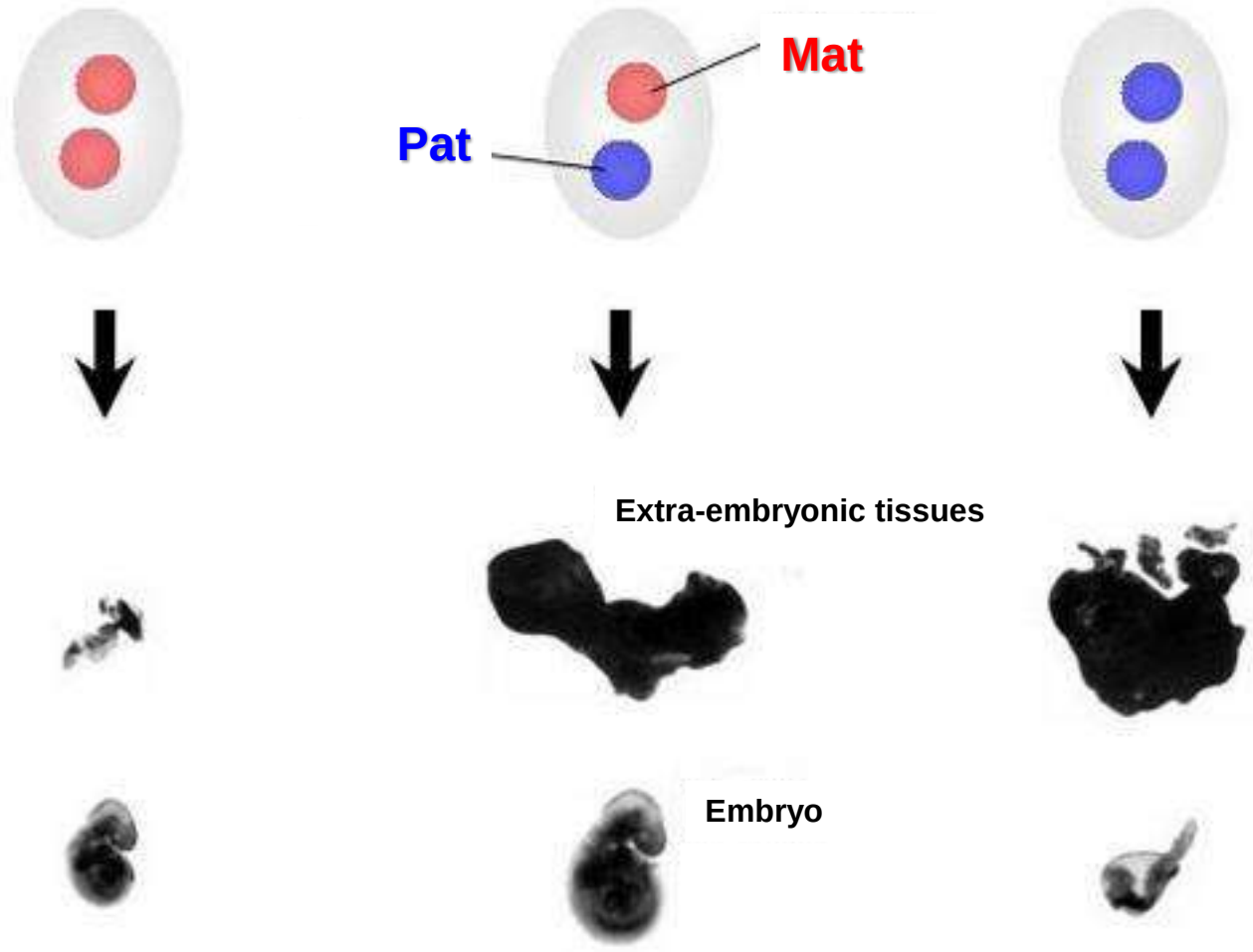


♂ \ ♀	donkey	horse
		
donkey	donkey	hinny
		
horse	mule	horse
		

Images credit: http://www.imeha.org/

Mula e Bardoto

Uniparental embryos (Parthenogenetic)



Surani et al., 1984; McGrath & Solter, 1984

Como cientistas chineses conseguiram que ratos do mesmo sexo se reproduzissem

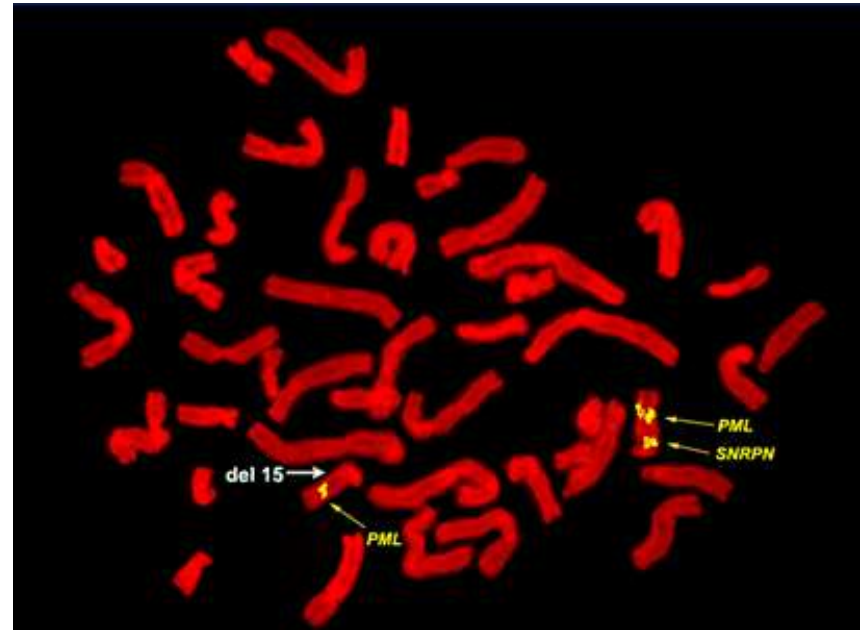
Edison Veiga
De Milão para a BBC News Brasil

11 outubro 2018

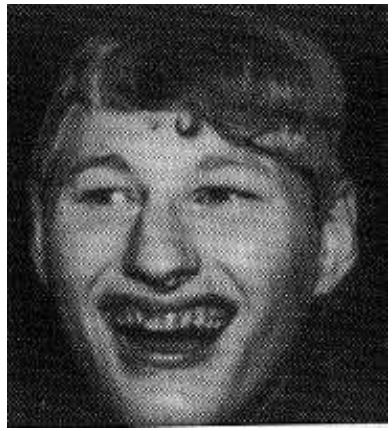
Compartilhar

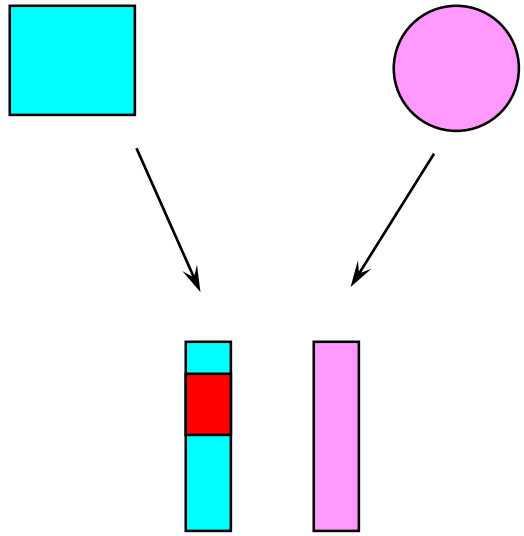
Pesquisadores chineses recorreram a células-tronco embrionárias - e à manipulação genética - para realizar o estudo em laboratório

Prader-Willi syndrome

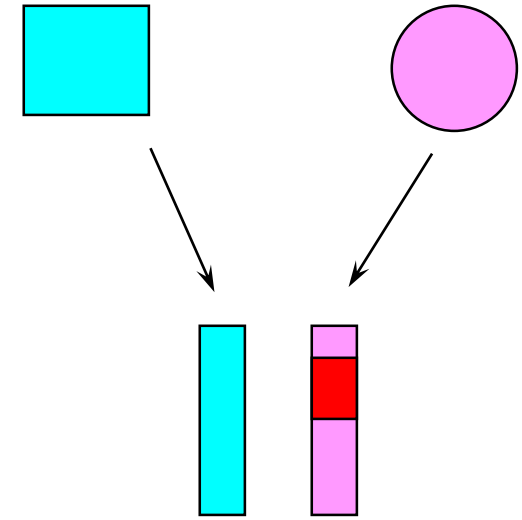


Angelman syndrome (AS)





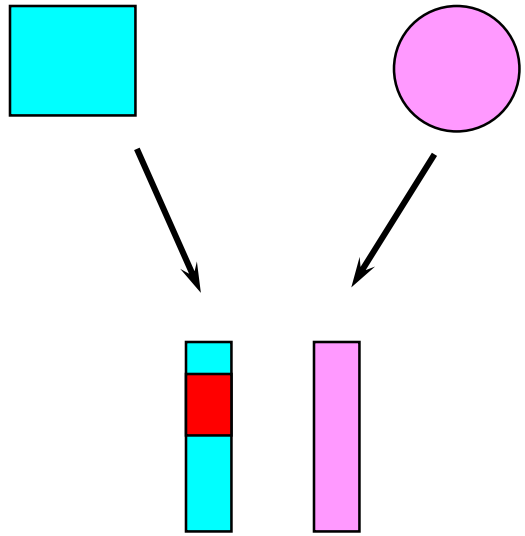
Chromosome 15
15q11-15q13



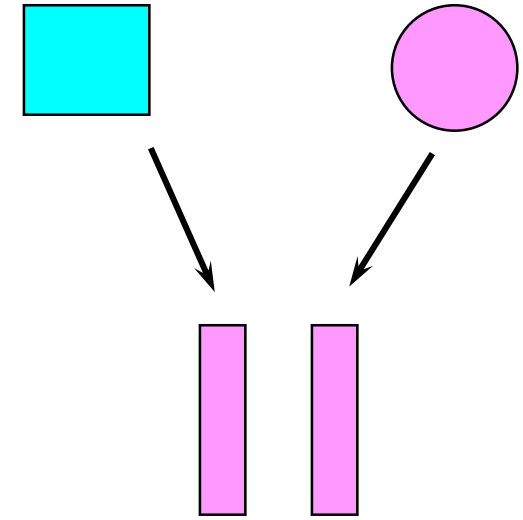
PWS

AS

Prader-Willi syndrome



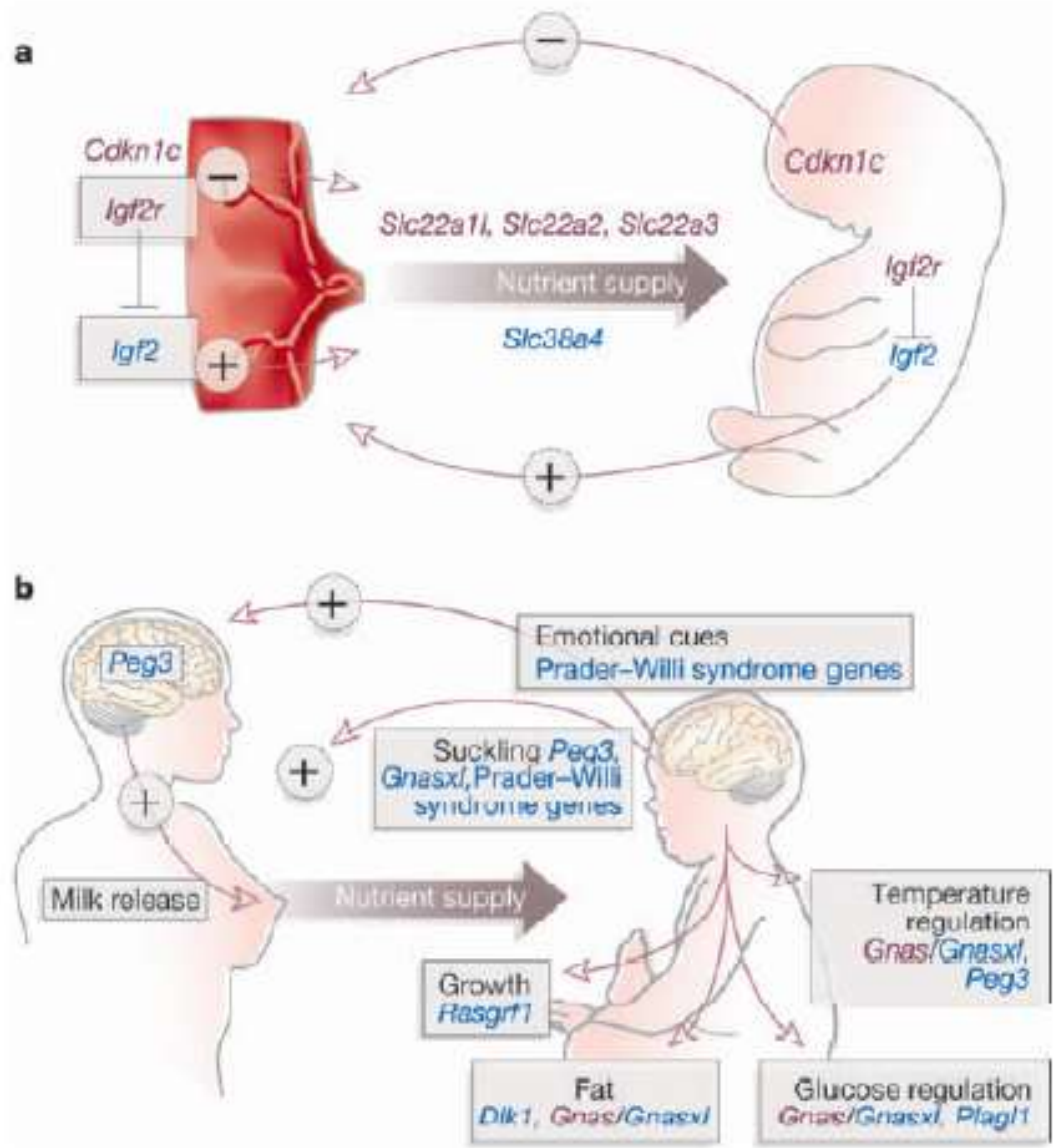
Chromosome 15



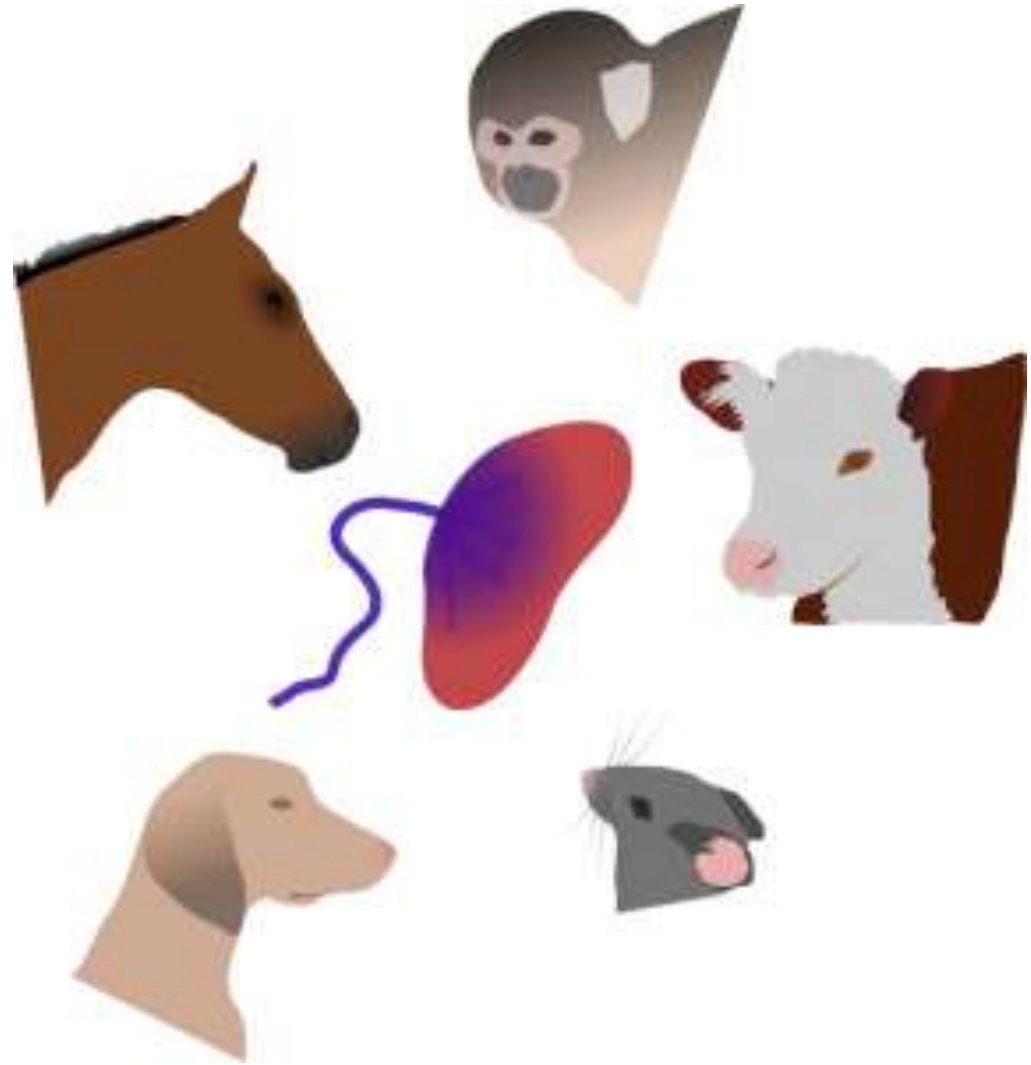
Uniparental disomy (UPD)

- Proliferação celular, crescimento (placenta, embrião), processos neurológicos e no comportamento

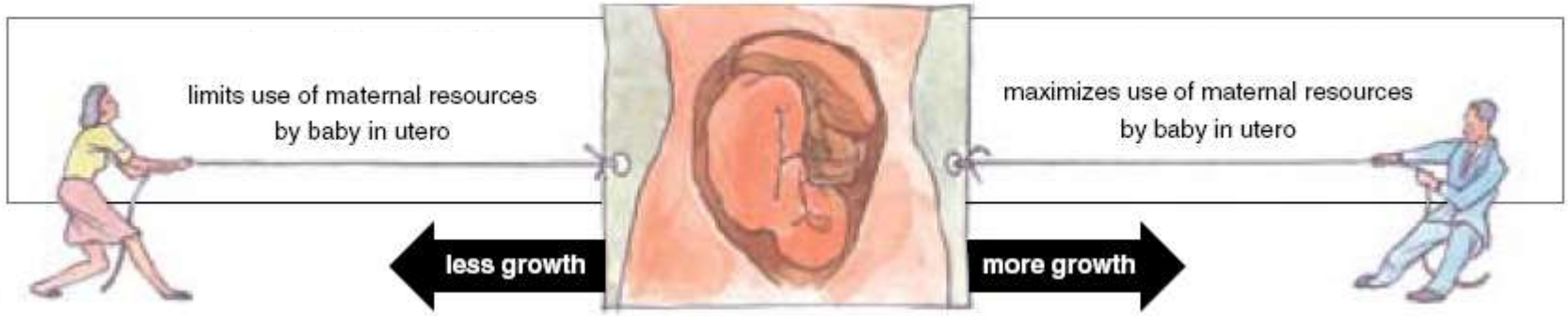
- Ação do ambiente



Genomic Imprinting



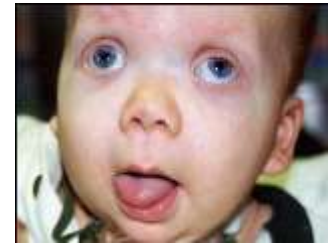
Teoria do Conflito

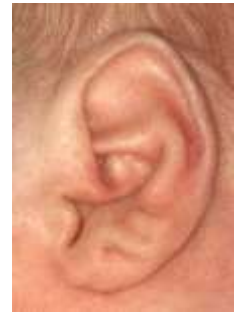


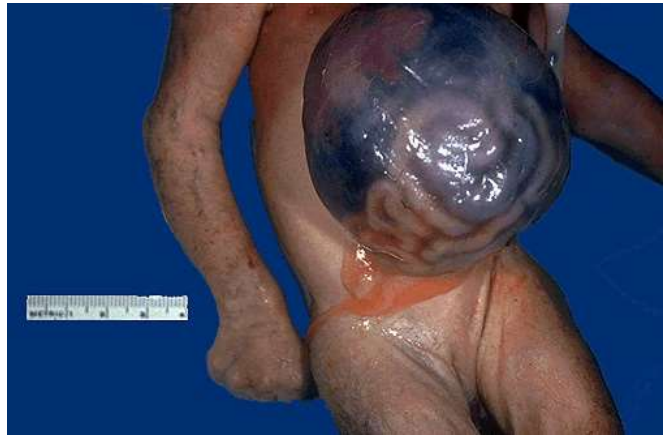
(Jirtle & Weidman, 2007)

Beckwith-Wiedemann syndrome (BWS)

- Overgrowth
  - Macrosomy
  - Visceromegalia – kidney, liver
  - Hemihyperplasia – 12.5%
- Abdominal wall defects (omphalocele, umbilical hernia, *diastasis recti*)
- Macroglossia
- Neoplasias: ~ 7.5% (40% with hemihyperplasia***)
- Ear lobe creases
- Hypoglycemia





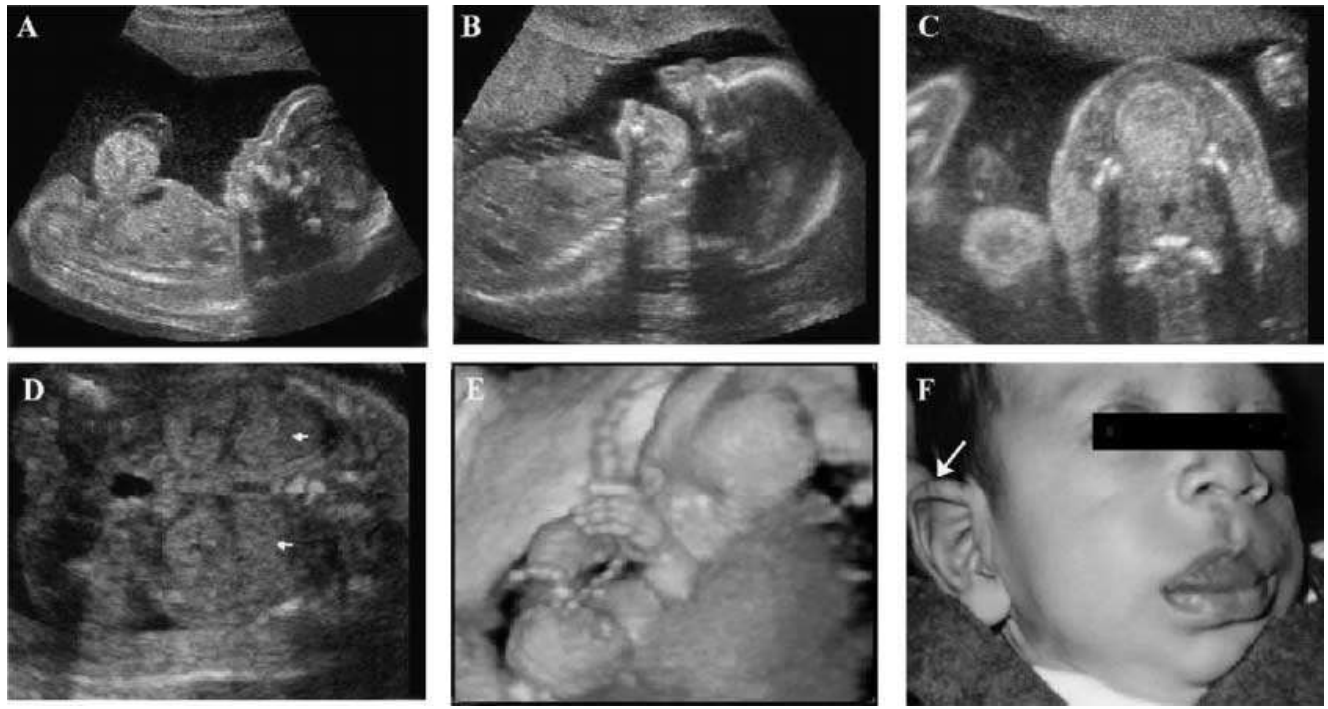


Onfalocele

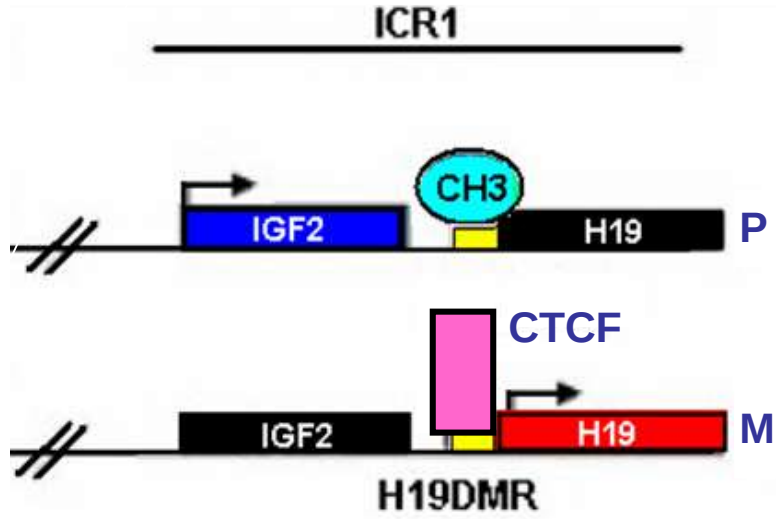
American Journal of Medical Genetics Part A 143A:625–629 (2007)

Methylation Pattern at the KvDMR in a Child With Beckwith–Wiedemann Syndrome Conceived by ICSI

M.V. Gomes,[1*] C.C. Gomes,[2] W. Pinto Jr,[3] and E.S. Ramos[1,4]



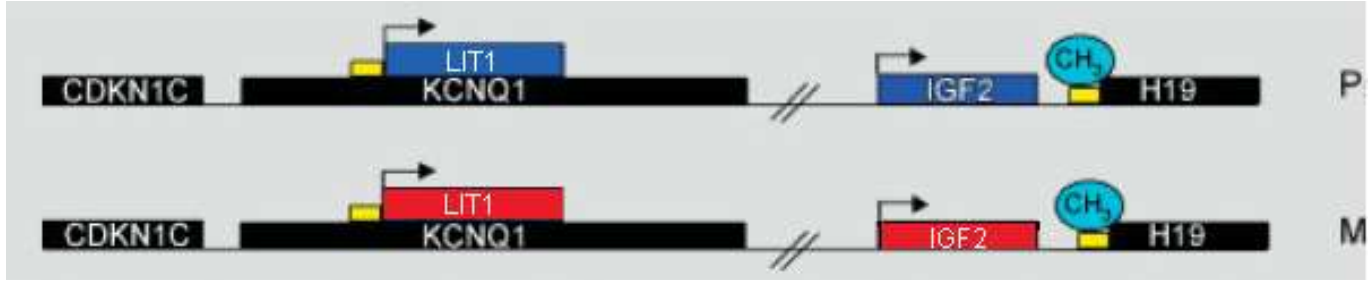
- **11p15.5**



ICR = Imprinting Control Region

DMR = Differntially Methylated Region

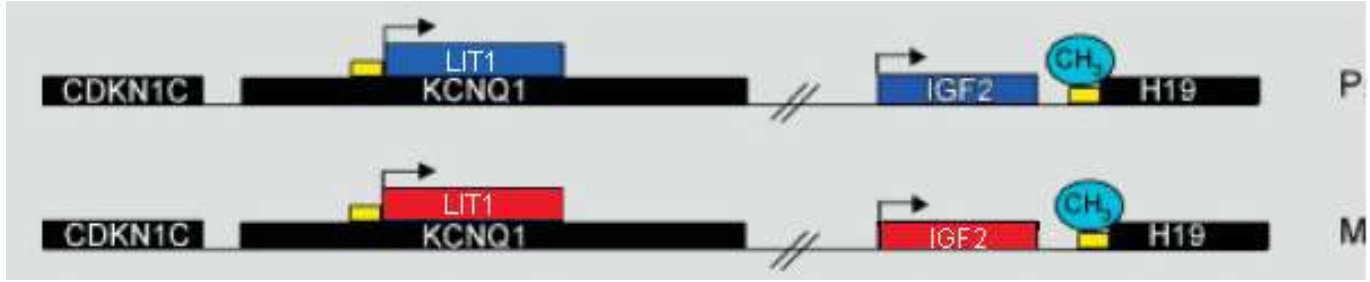
– Síndrome de Beckwith-Wiedemann



Síndrome de Silver-Russell

- Baixa estatura, 5º dedo curto/clinodactilia, assimetria.

– Síndrome de Beckwith-Wiedemann



- Síndrome de Silver-Russell