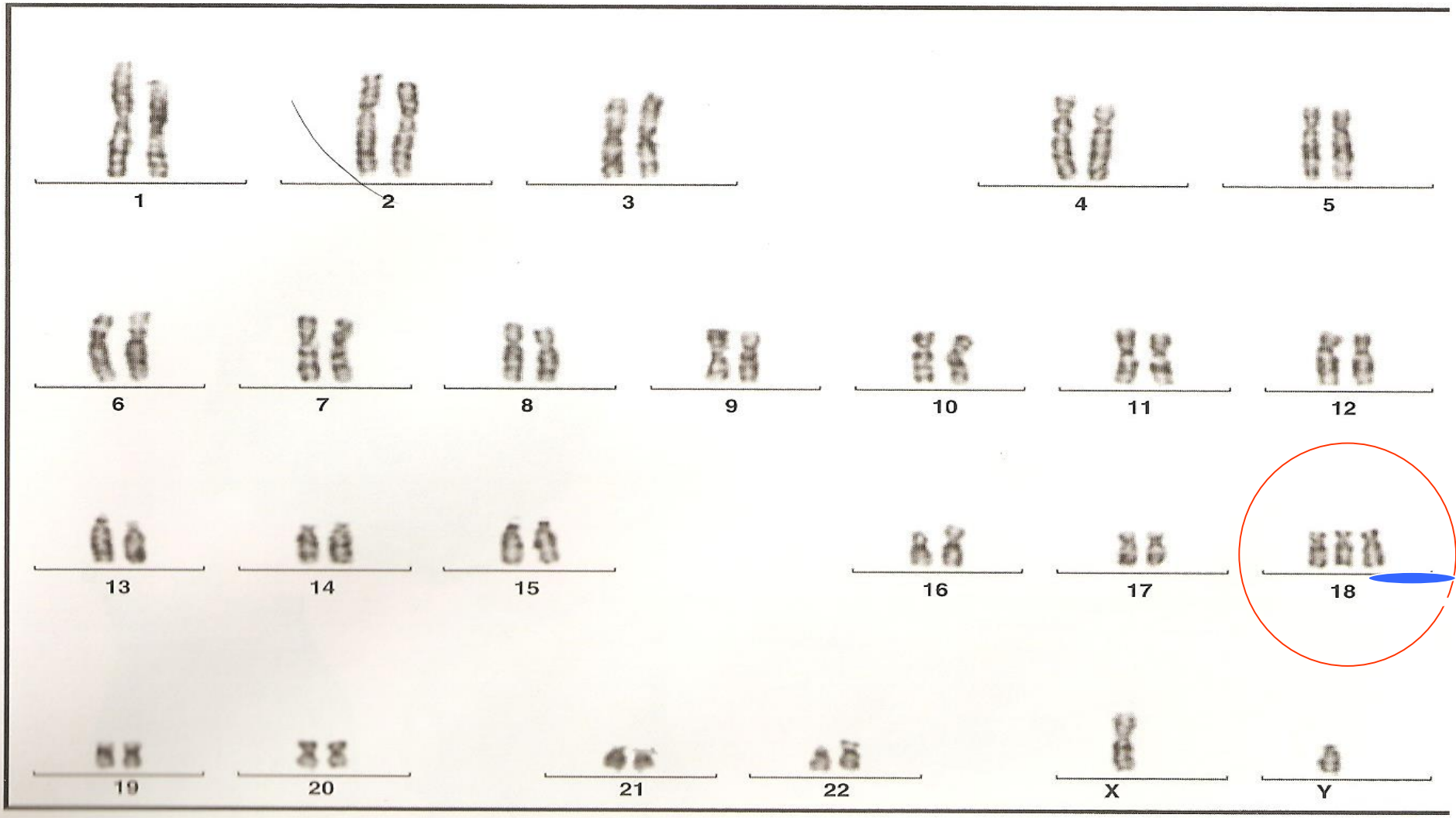
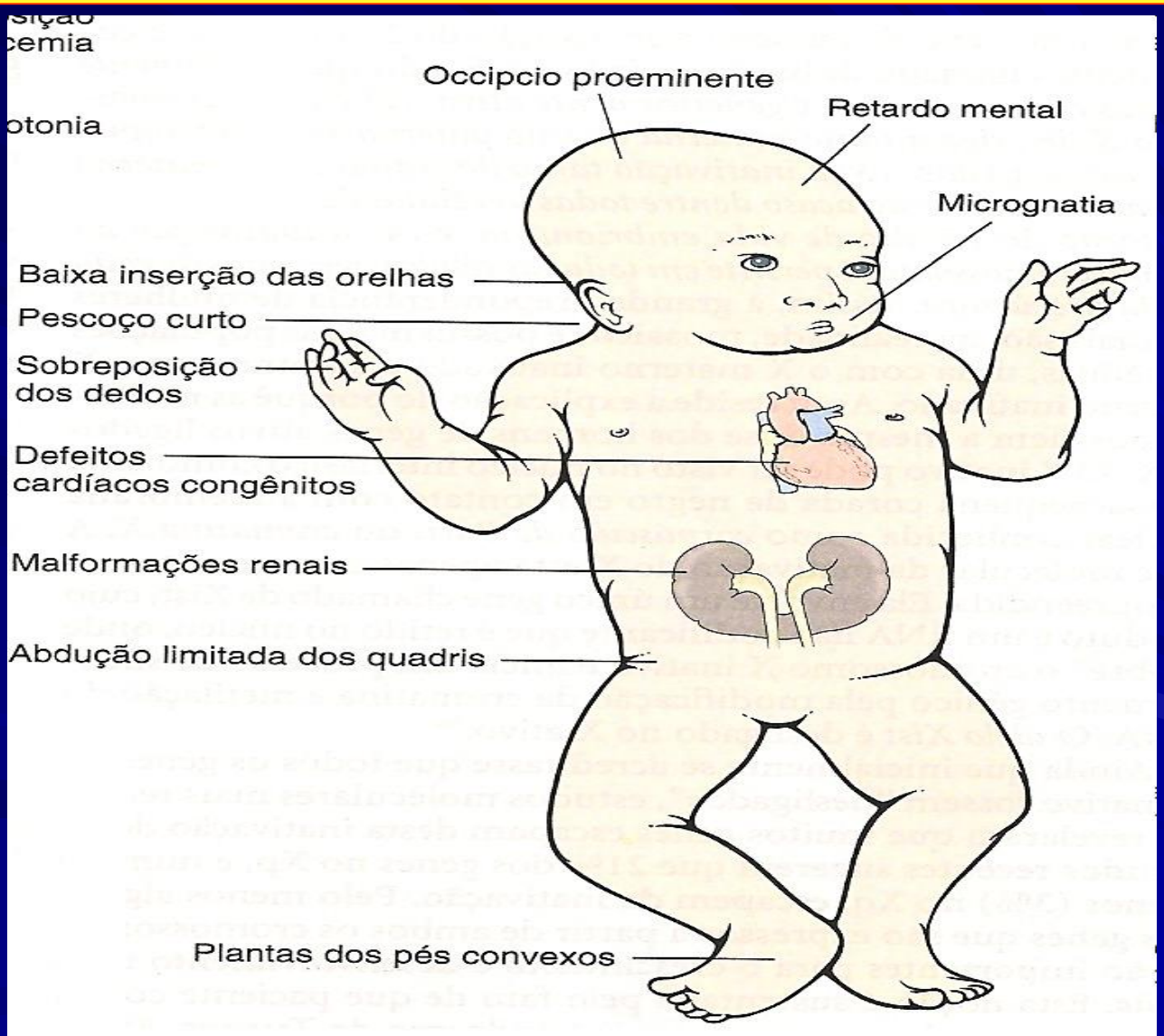
Sindrome de Edwards

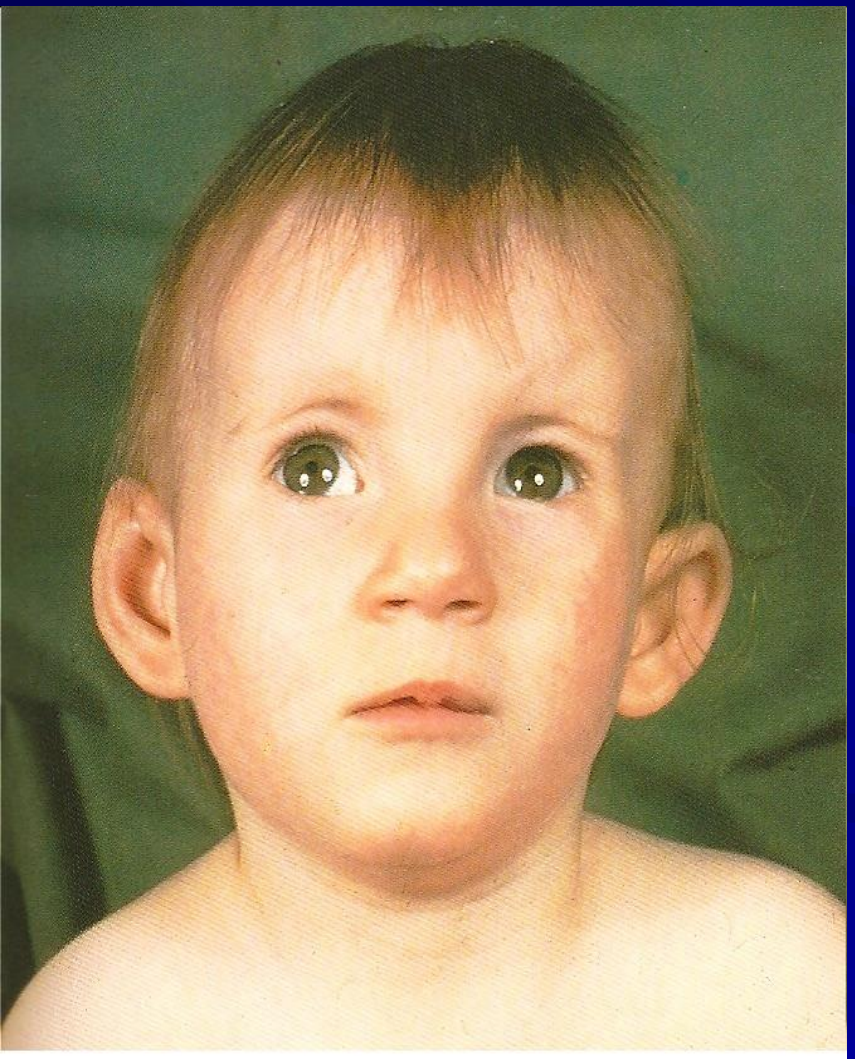
Trissomia do 18 Sind Edwards

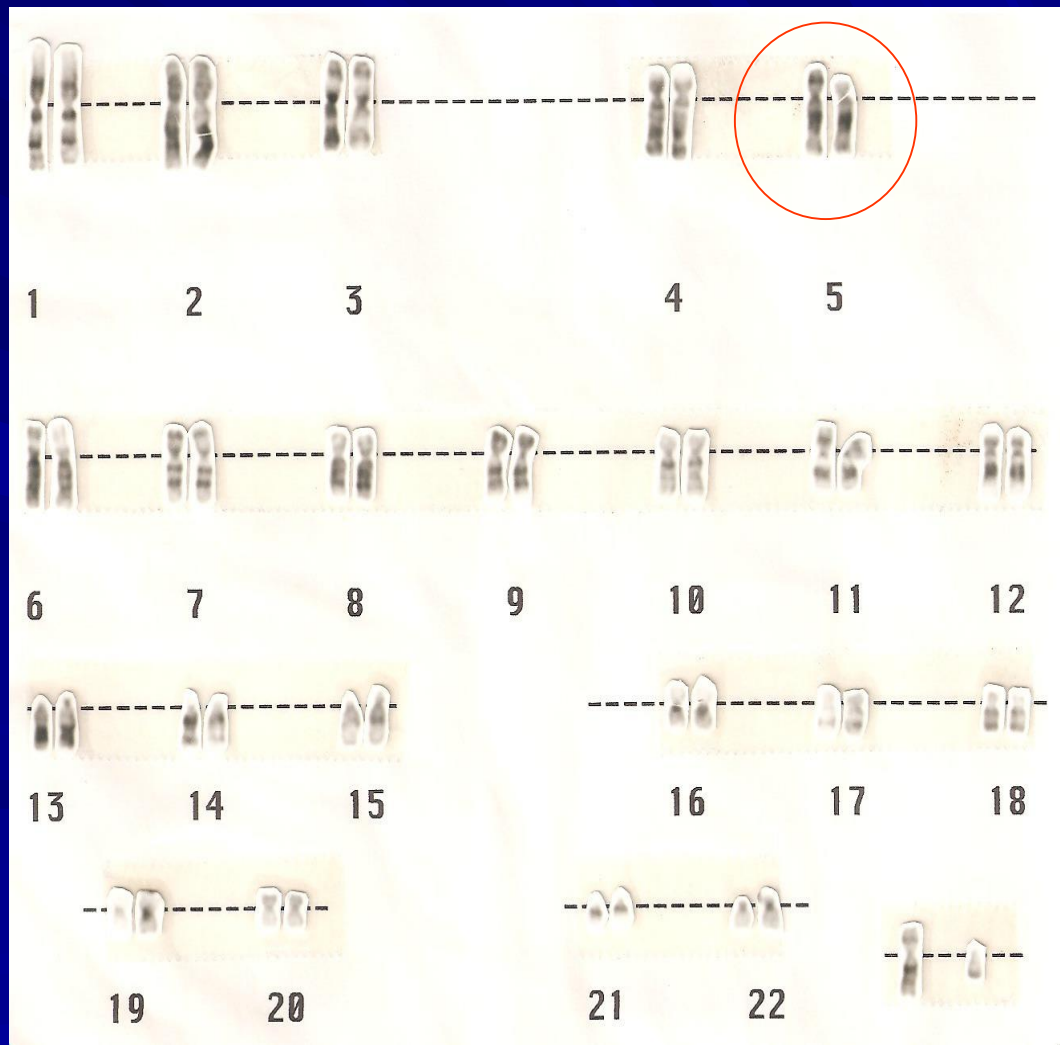


Trissomia do 18 Sínd de Edwards



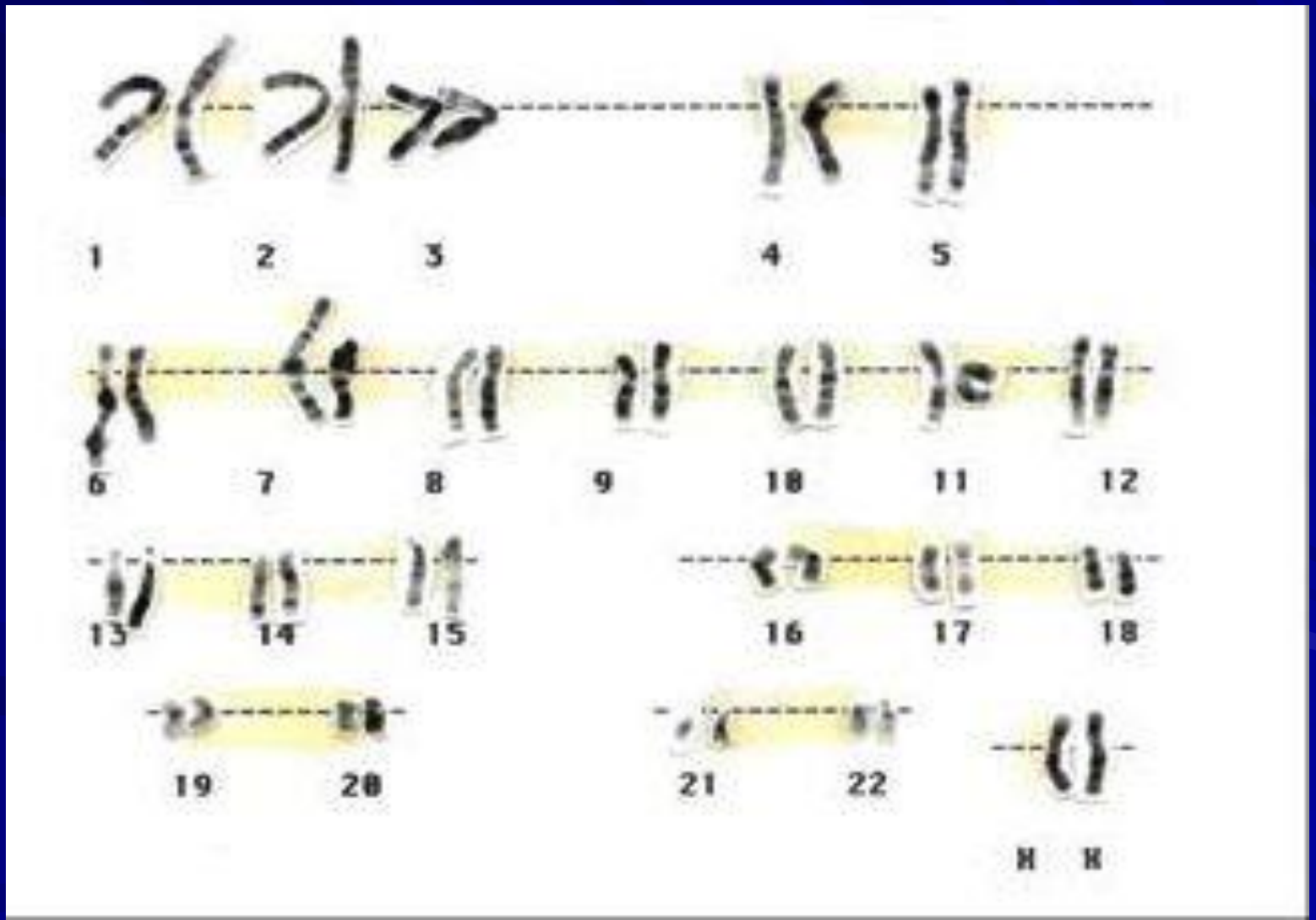
Síndrome de Cri-du-Chat e cariótipo 5p-



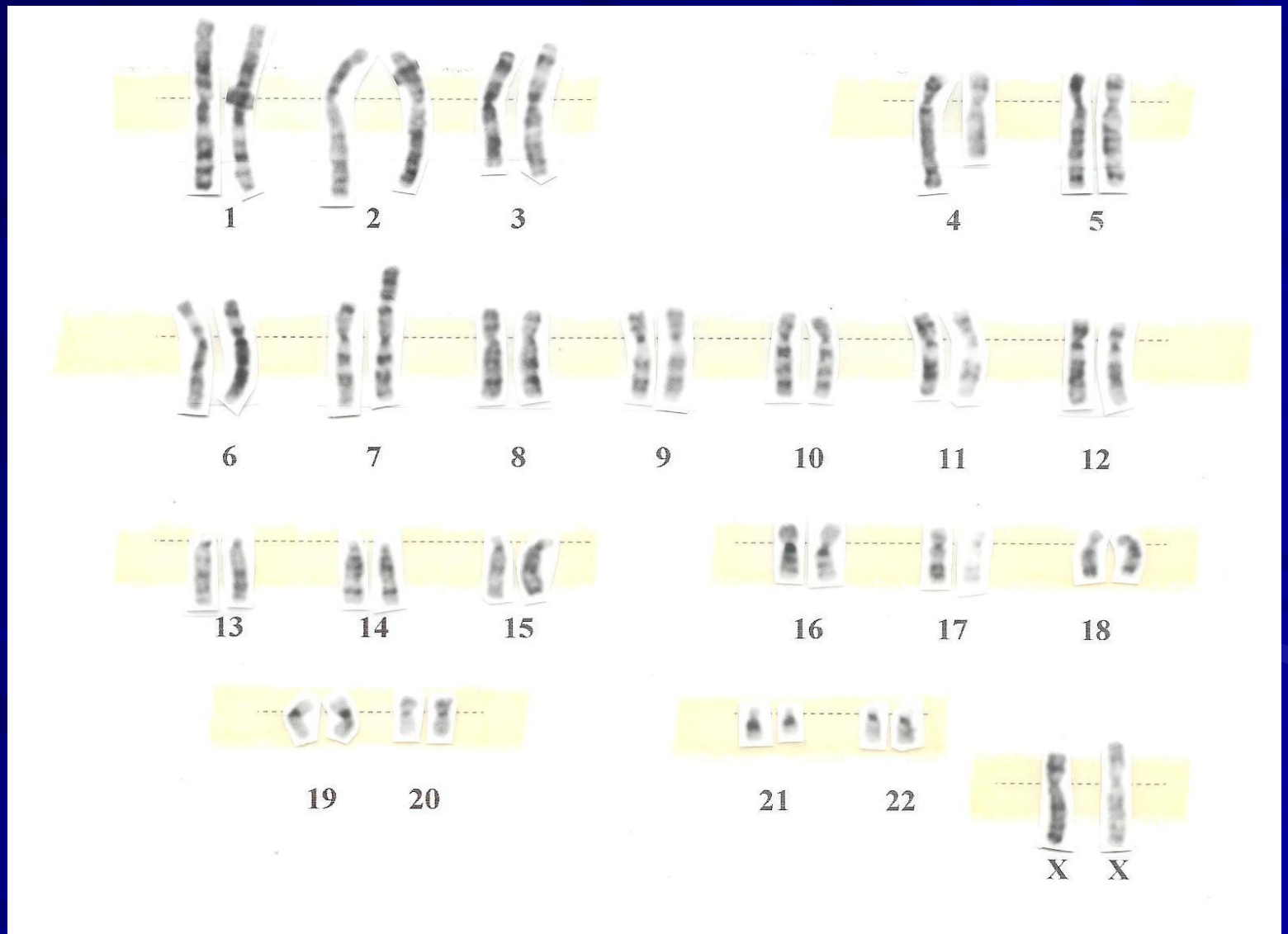


Trissomia parcial 4q Três casos descritos no mundo

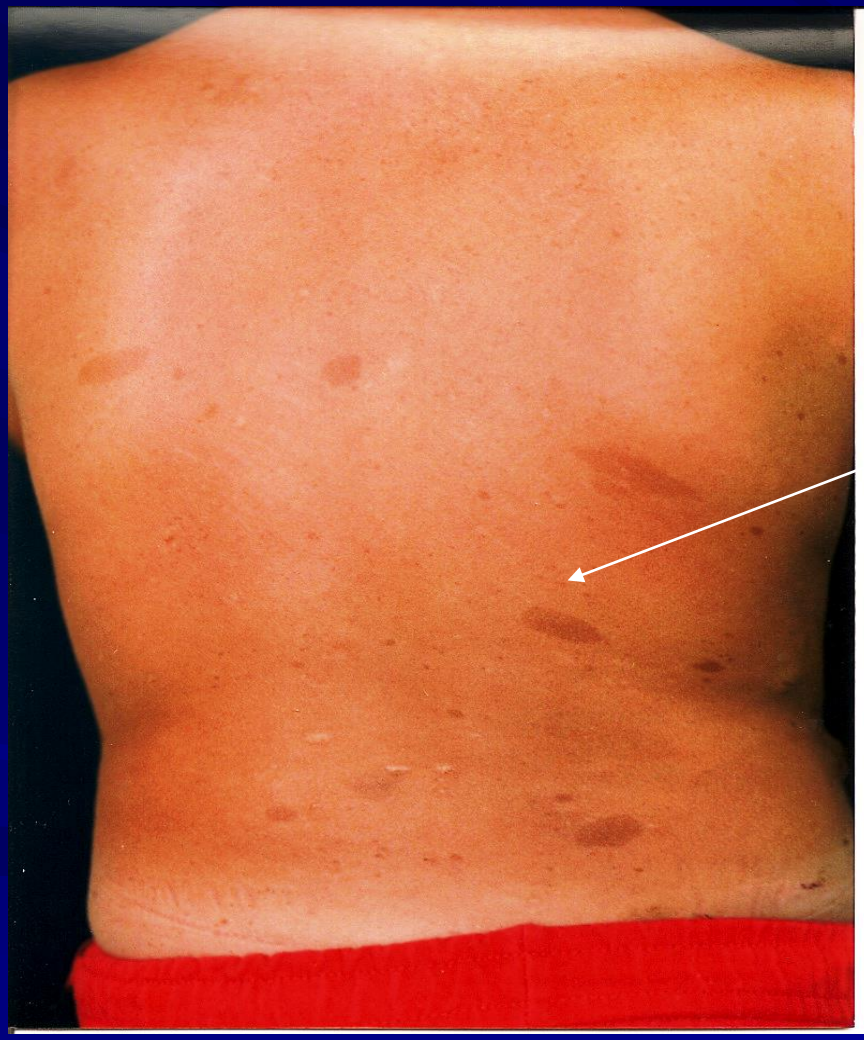
Cariótipo Trissomia parcial 4q



Cariótipo Translocação 4 , 7. mãe



Doença de Von-Recklinghausen



Her.Aut.Dominante

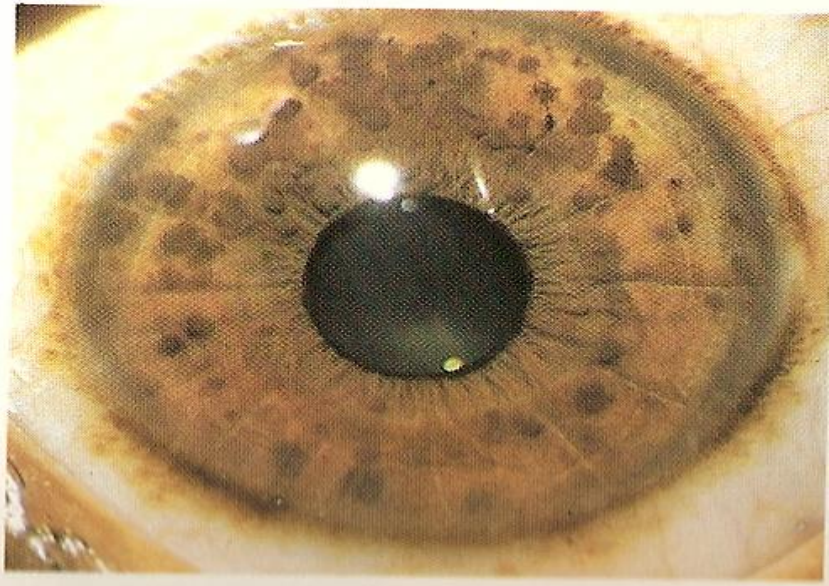
Manchas café com leite

Ambulatório de Genética
Dr Daher-Hemocentro

Neurofibromatóse tipo1

Neurofibromatóse tipo 1

palma. (Cortesia do
Dr. Michael Sherlock).



Síndrome de Seckel

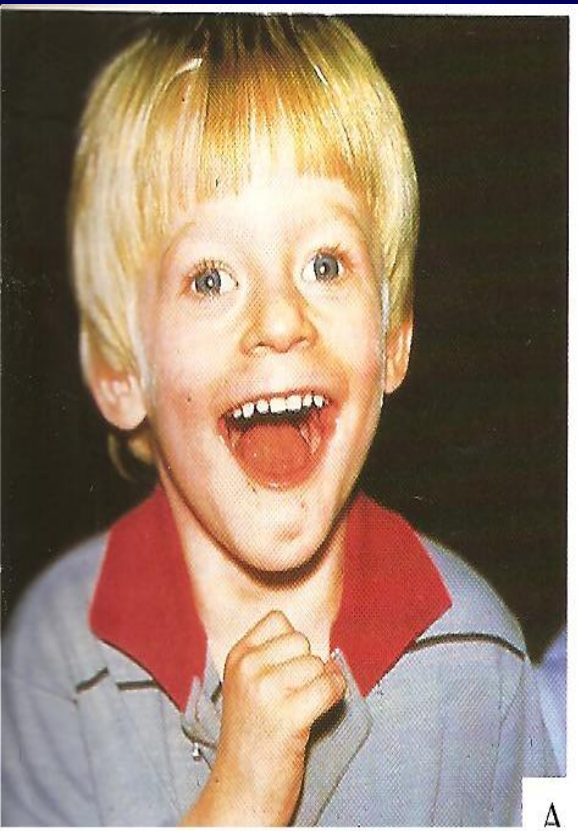
Mesma Idade

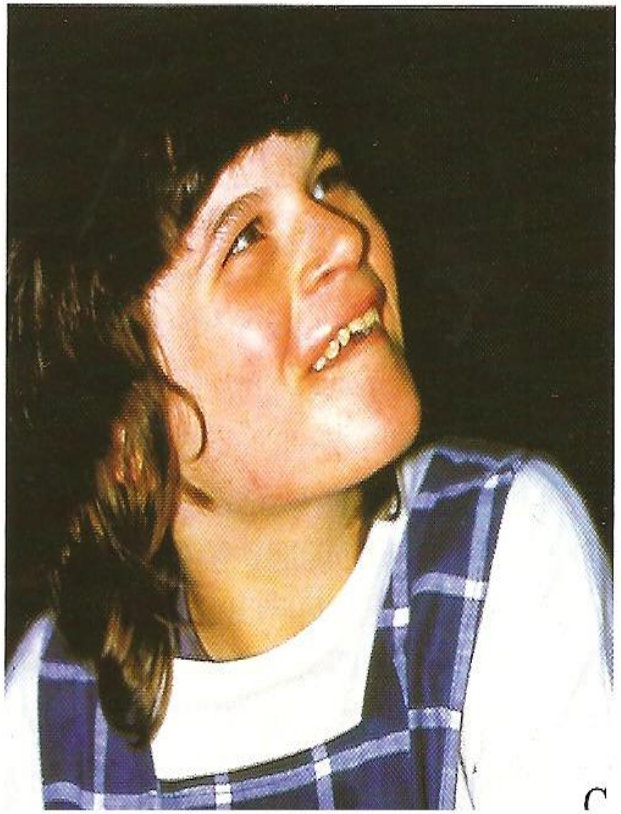
Nanismo extremo

Cabeça de Pássaro

Her. Aut. Recessiva
Mutação 3q, 22-24

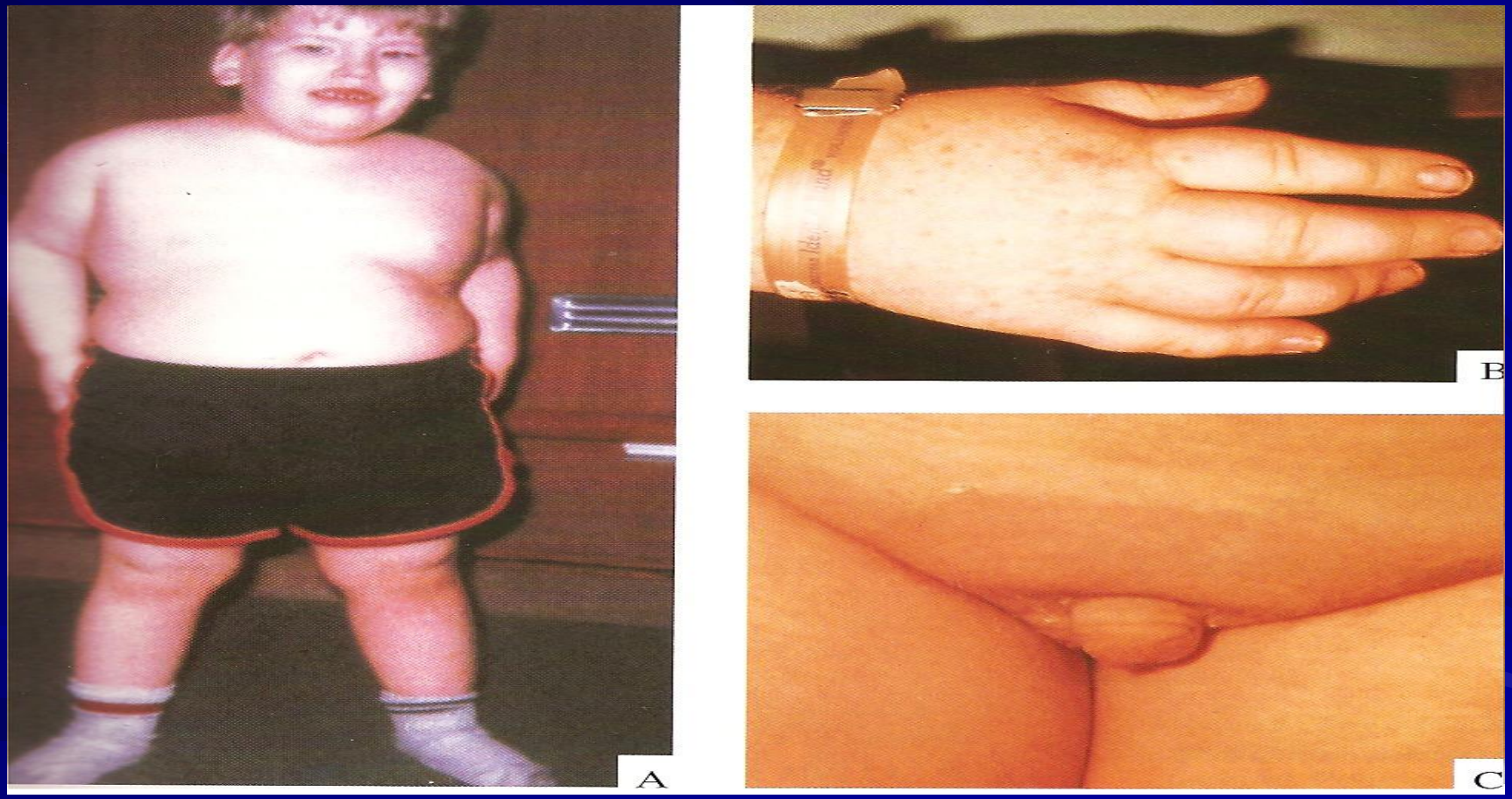
Síndrome de Angelman





Del 15q11-q13

Síndrome de Prader-Willi



Del 15q11-15q13

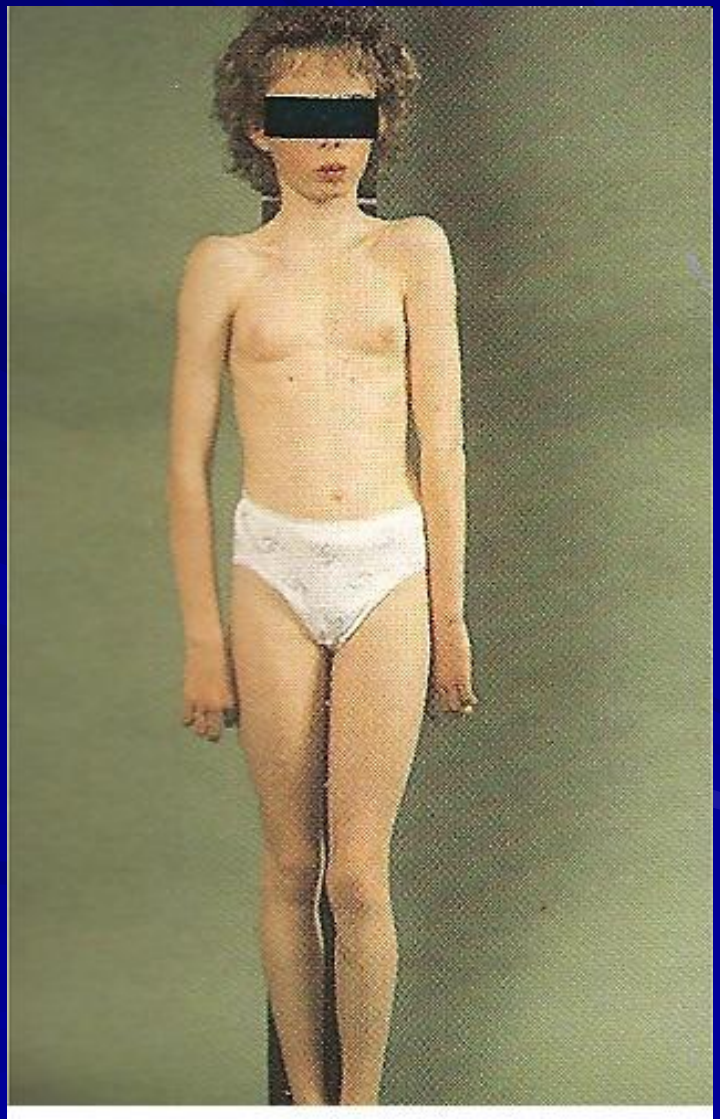
Prader-Willi X Angelman
Dissomia uniparental

1genótipo

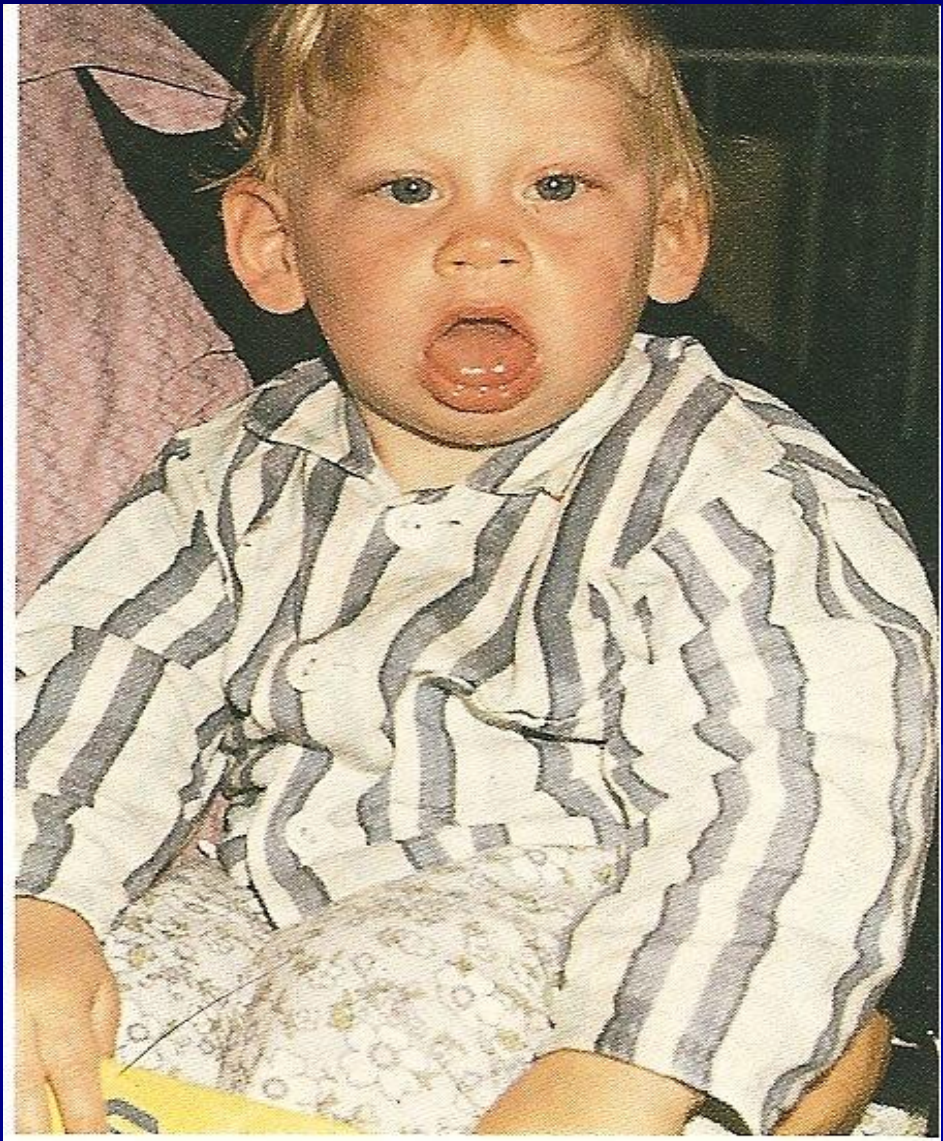
15q11-13

2 fenótipos

Síndrome de Silver Russel



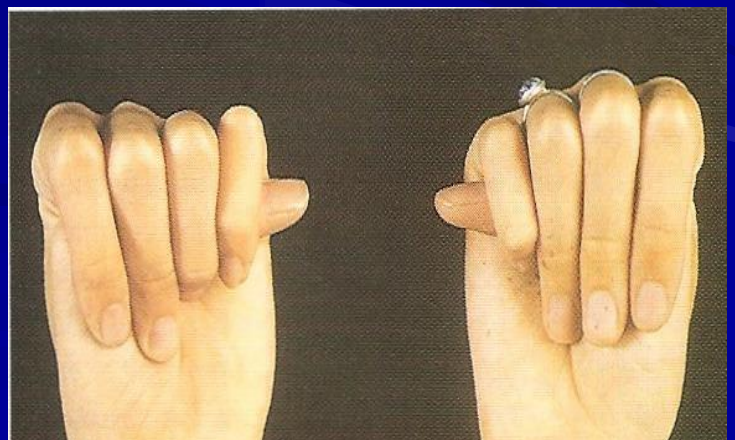
Sínd. de Beckwith-Wiedmann



Progéria:Criança “Velha”

Síndrome de Marfan

Hiper-elasticidade em Marfan



Deficit de Fibrilina