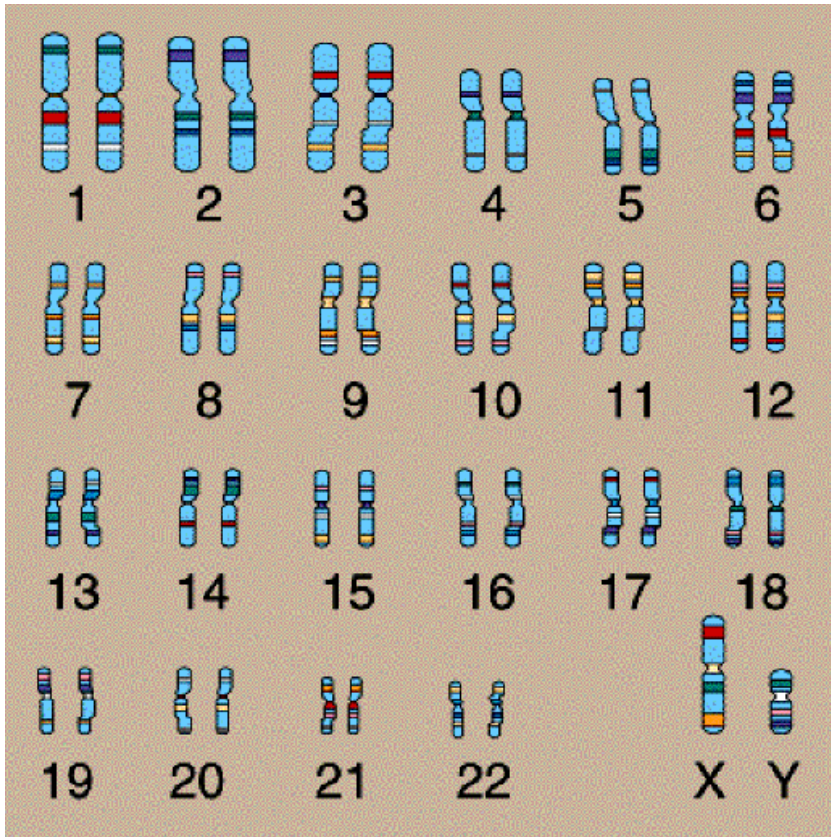


CITOGENÉTICA MOLECULAR

Aplicação em Anatomia Patológica



Cariótipo Humano



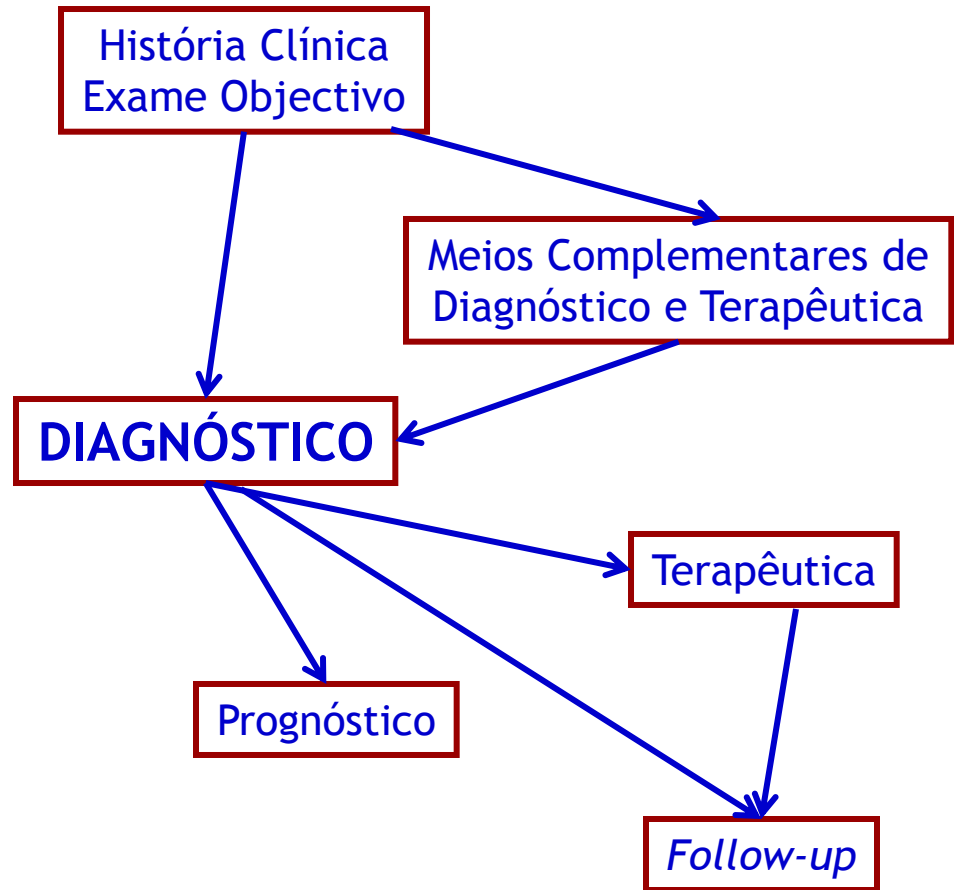
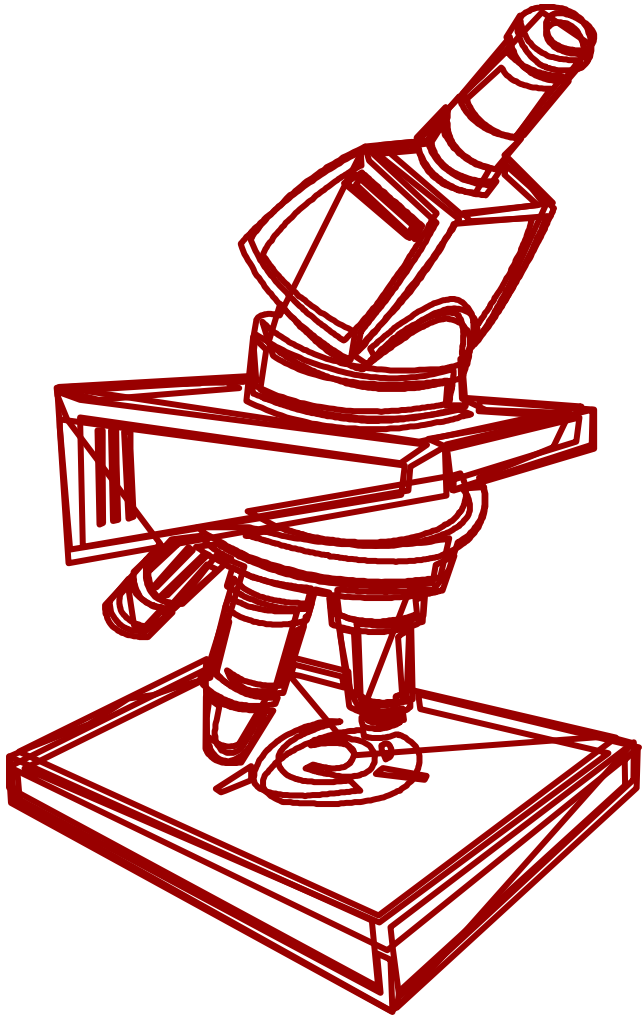
Sofia Loureiro dos Santos

Serviço de Anatomia Patológica

HFF, 25 de Novembro/2010

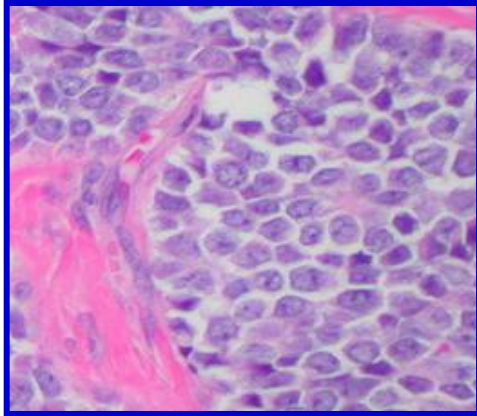


Anatomia Patológica

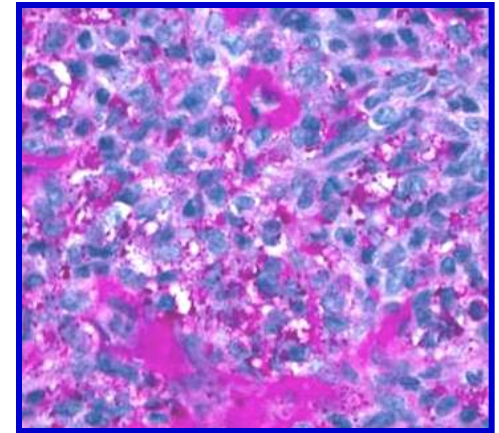


Anatomia Patológica

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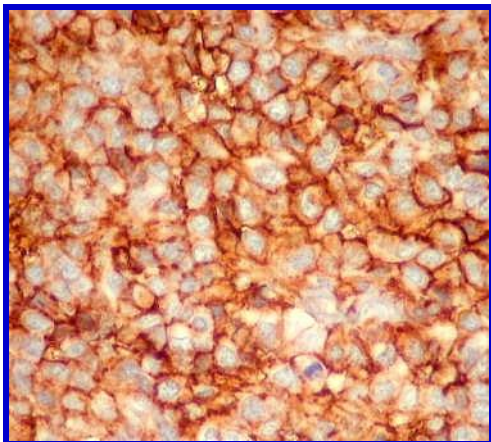


PAS

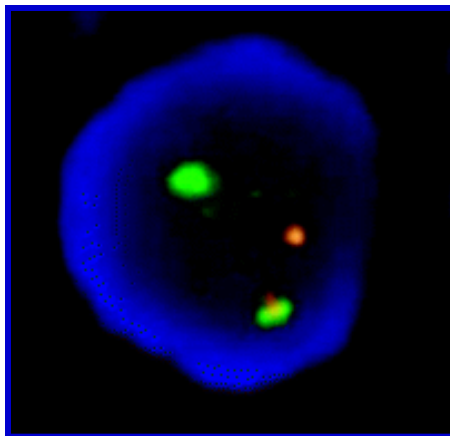


SARCOMA DE EWING

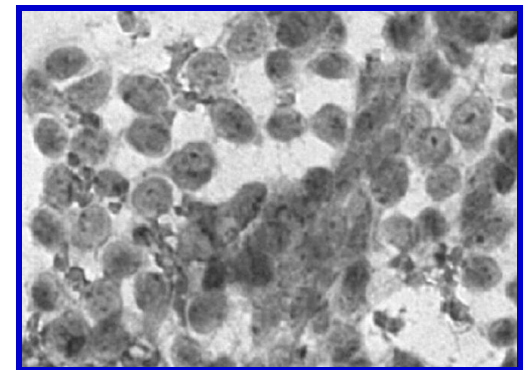
CD99



FISH



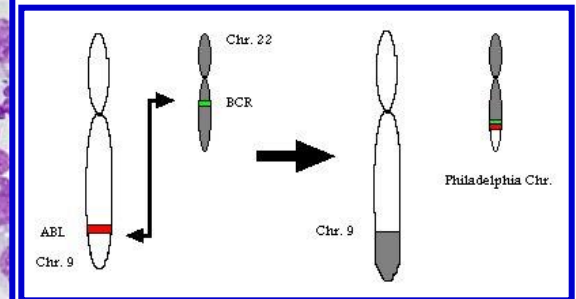
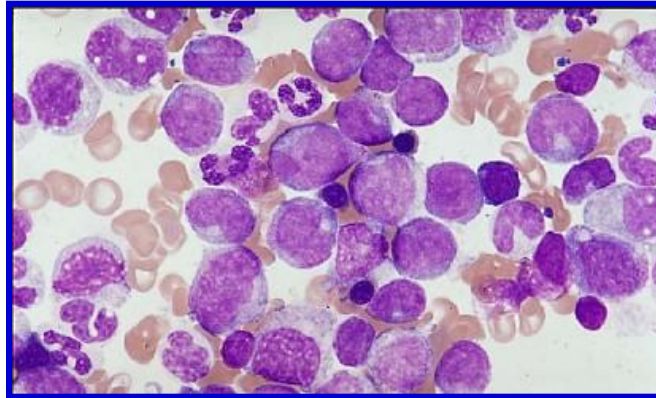
ME



Diagnóstico

Alterações estruturais

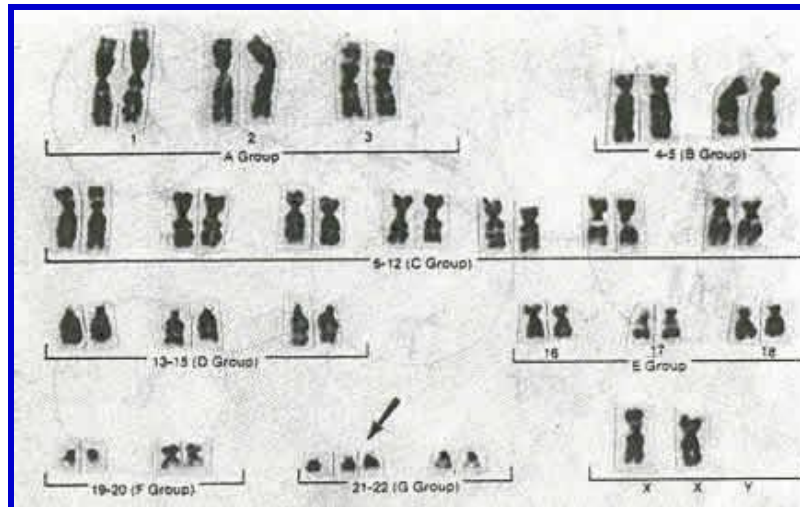
- Deleções
- Inversões
- Inserções
- Translocações
- (...)



t(9;22)
Cromossoma Philadelphia
LMC

Alterações numéricas

- Poliploidia
- Endoreduplicação
- Aneuploidia
- (...)

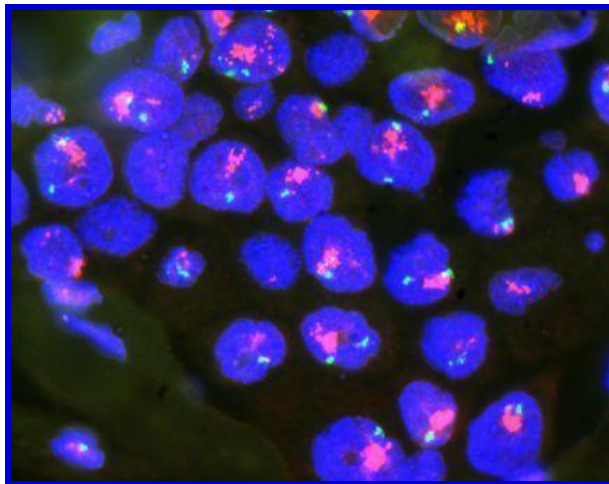
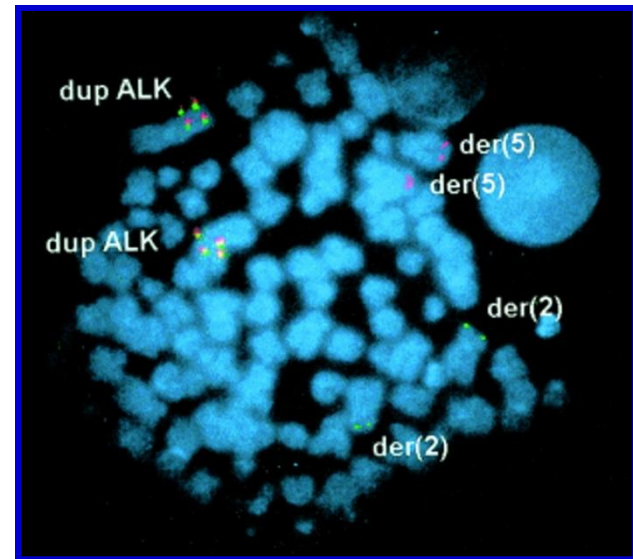


Trissomia 21
Síndrome de Down

Prognóstico e Terapêutica

(A)naplastic (L)ymphoma (K)inase
ALK - t(2;5)(p23;q35)

(Anaplastic Large Cell Lymphoma
ALK (+) melhor prognóstico que ALK (-)

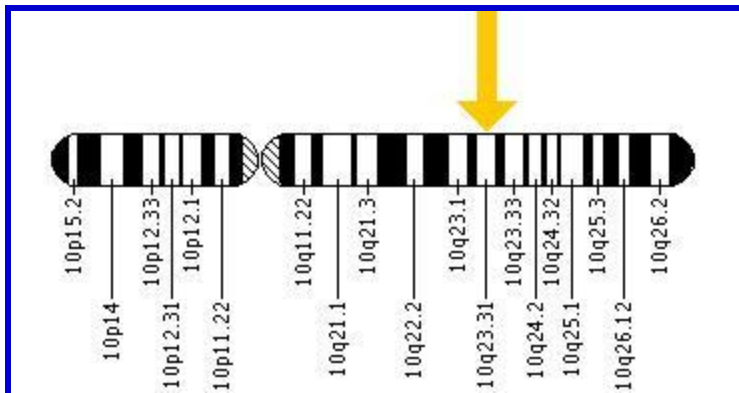
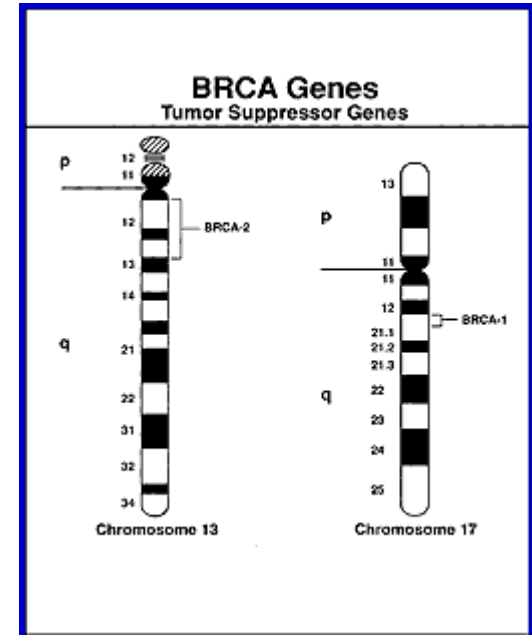


HER2
17q12-21

Terapêutica com Trastuzumab

Risco Familiar e Monitorização Terapêutica

BRCA1 - gene supressor (cromossoma 17)
BRCA2 - gene supressor - (cromossoma 13)
Mutação dos genes nas células germinativas
Risco familiar de carcinoma da mama, ovário, próstata



PTEN - gene supressor (cromossoma 10)
Perda de PTEN - resistência ao Trastuzumab