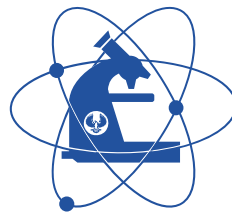


# Cancer Cytogenetics & Microarray

*Sarah Moore*

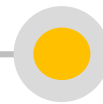
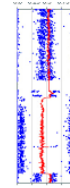
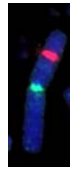


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## KARYOTYPE

Banded  
Karyotype

## FISH

Targeted  
probes

## Microarray

Higher  
resolution



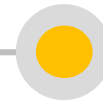
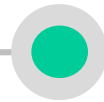
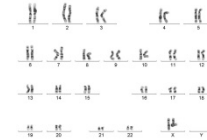
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# Myeloid disease

- Myelodysplastic Syndromes
- Myeloproliferative Neoplasms
- Acute Myeloid Leukaemia
- Other Clonal Haemopoietic disease





**Day 1**

Culture

**Day 2**

Harvest

**Day 3**

G-band

**Day 3**

Analyse/Report

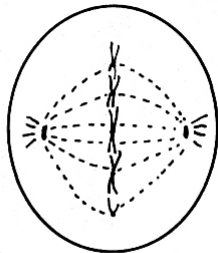


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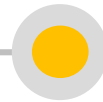
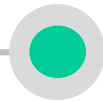
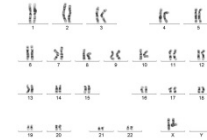
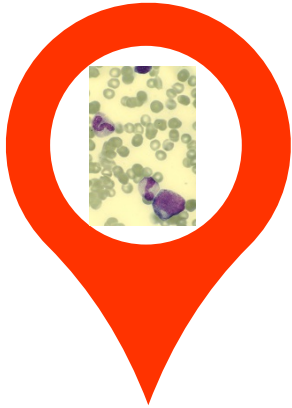
# Harvest

- Colcemid (or colchicine)
- Hypotonic
- Fixation (3:1 methanol:acetic acid)



**METAPHASE**





**Day 1**

Culture

**Day 2**

Harvest

**Day 3**

G-band  
(dark bands  
are gene  
poor)

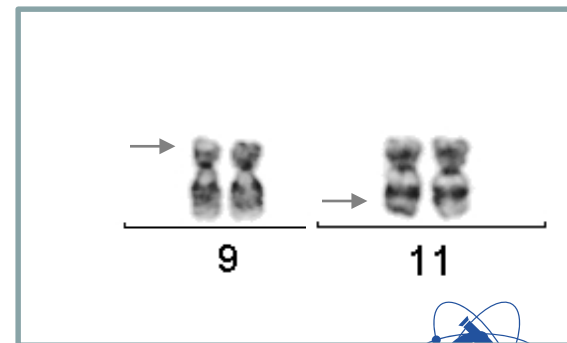
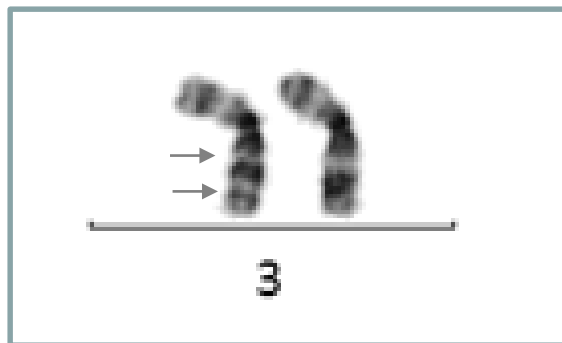
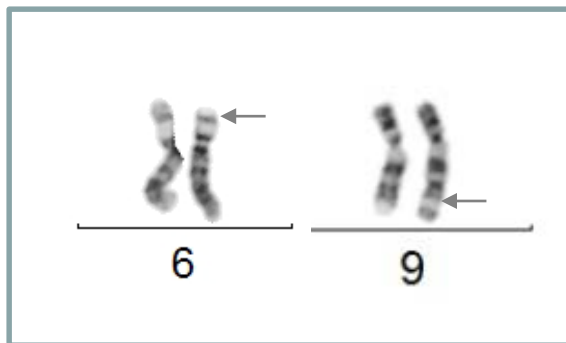
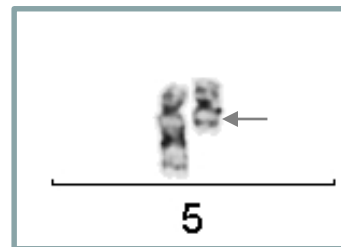
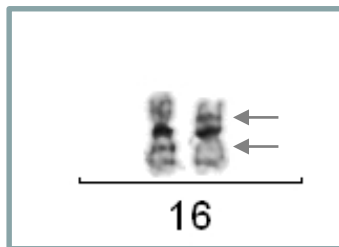
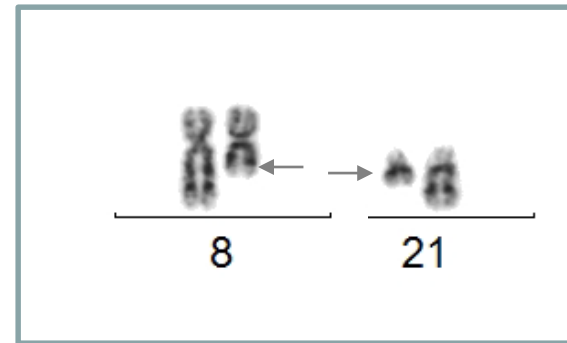
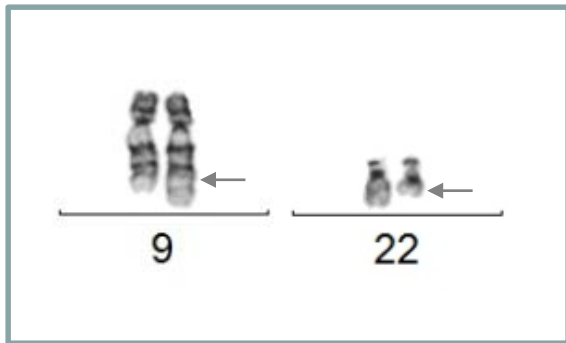
**Day 3**

Analyse/Report



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# Common Myeloid Abnormalities



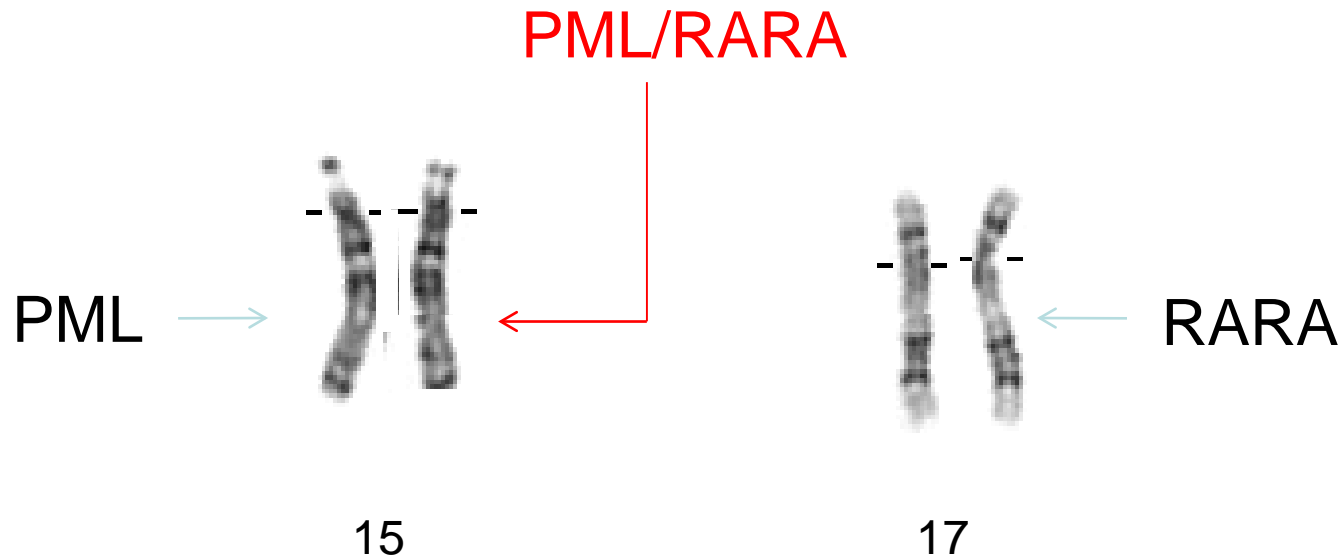
# Subclones & Separate clones

- $45,XY,-7[5]/47,XY,+8[20]$ 
  - Two separate clones
  - May represent divergent clonal evolution
  - May represent the emergence of a second malignancy
- $47,XY,+8[15]/46,XY,-7,+8[5]$ 
  - Represents clonal evolution





# t(15;17) of Acute Promyelocytic Leukaemia



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# Fluorescence In Situ Hybridization

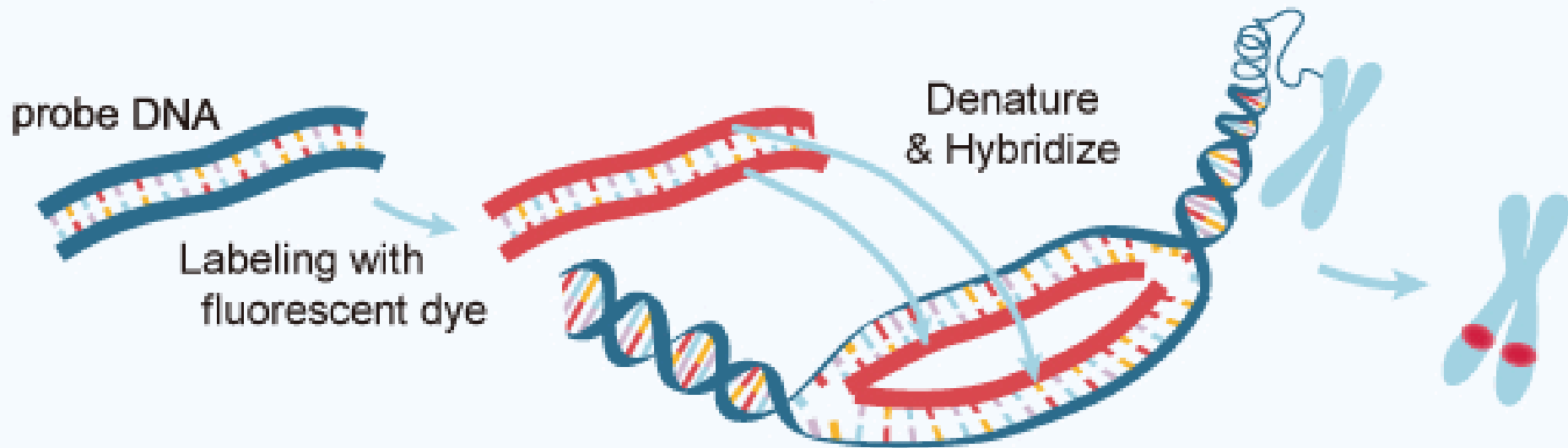


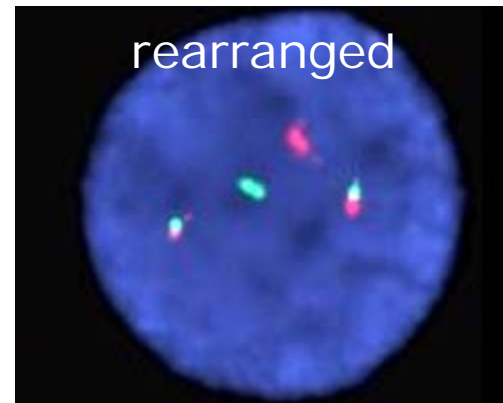
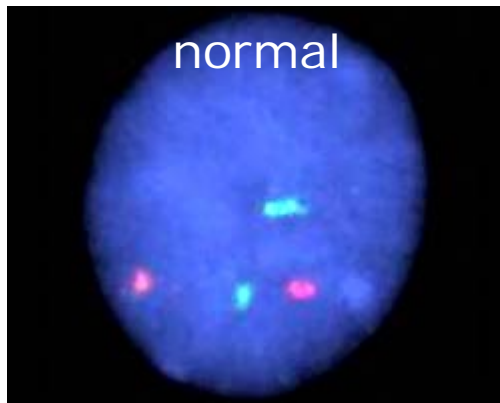
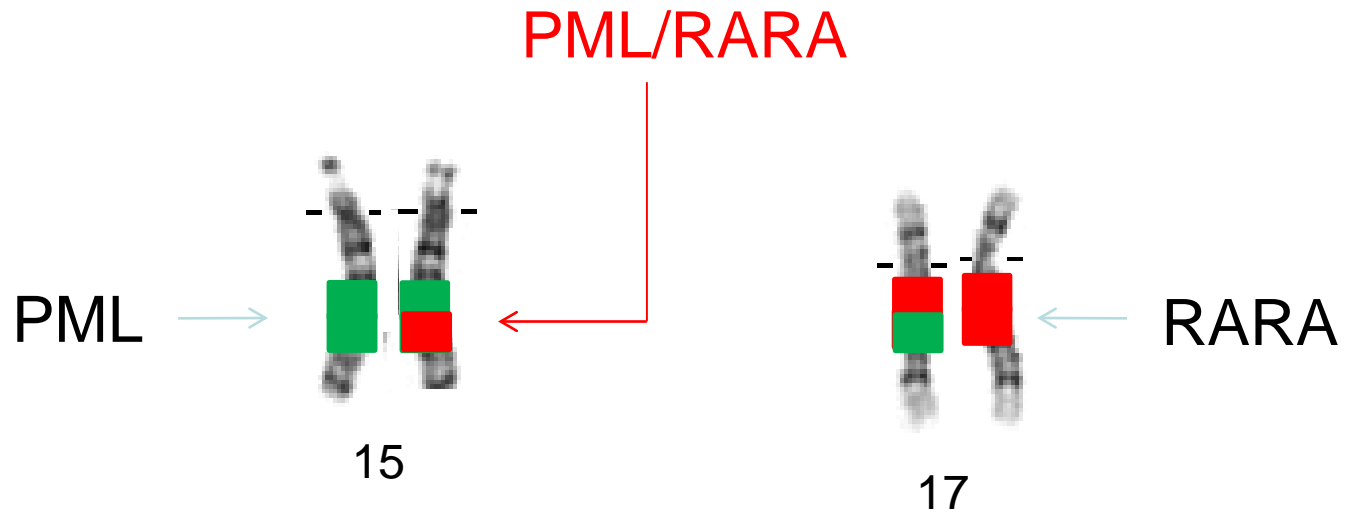
IMAGE FROM [ABNOVA.COM](http://ABNOVA.COM)



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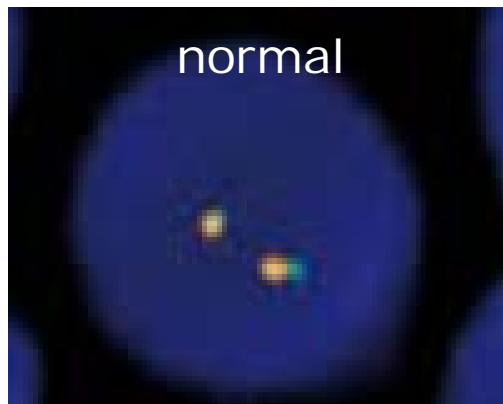
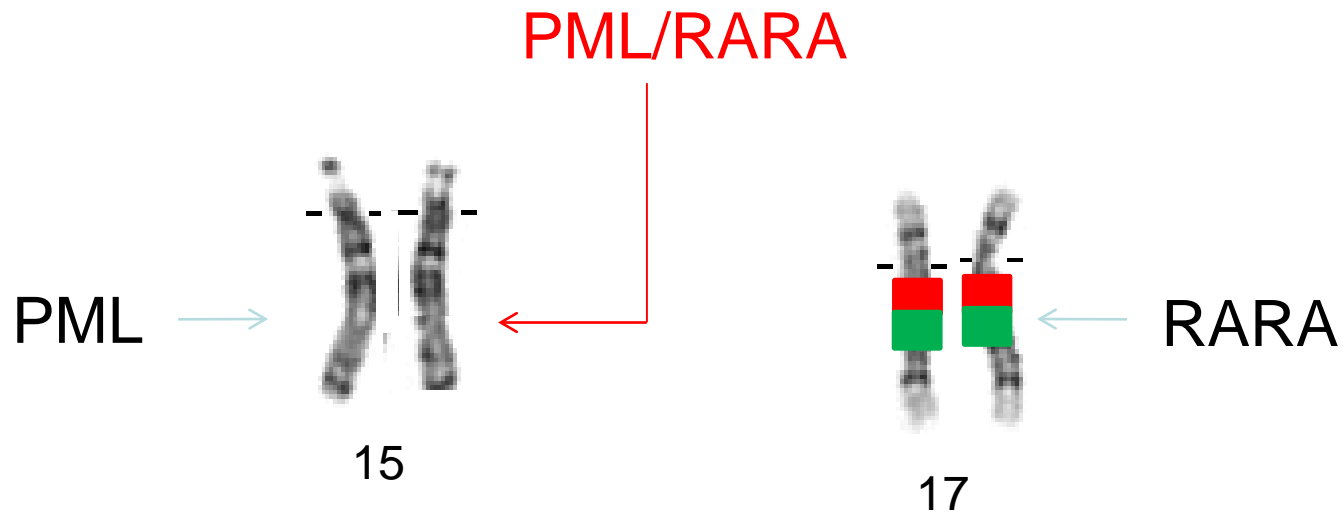
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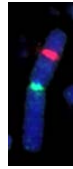
# Dual Fusion FISH of APML



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# RARA breakapart FISH of APML





**Day 1**

Hybridise

**Day 1**

Analyse/Report



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# Acute Lymphoblastic Leukaemia

## Current testing

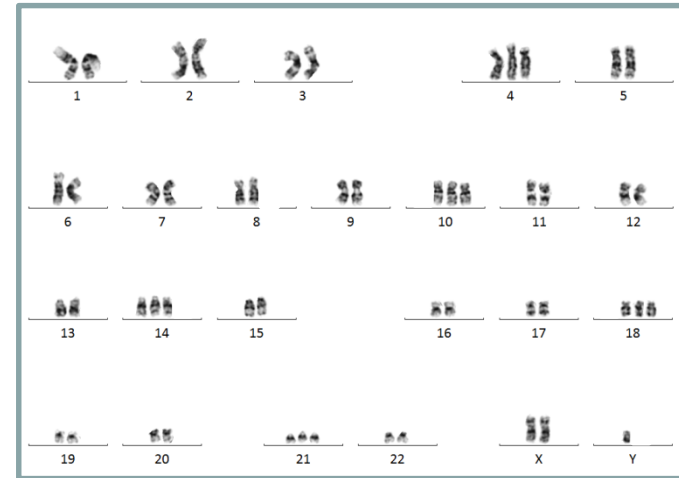
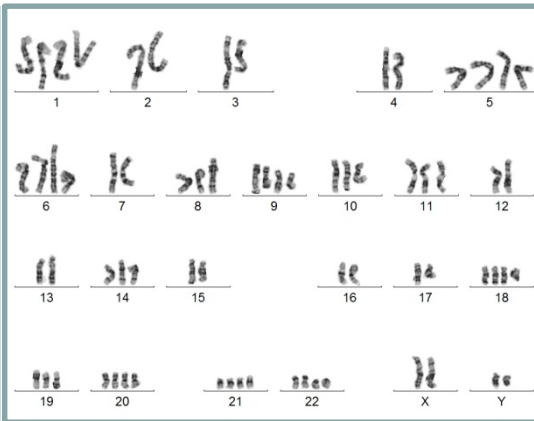
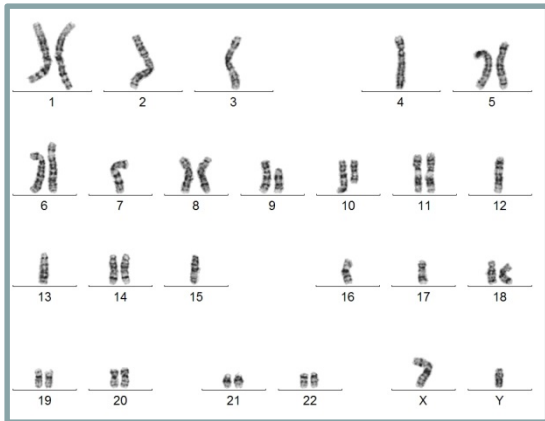
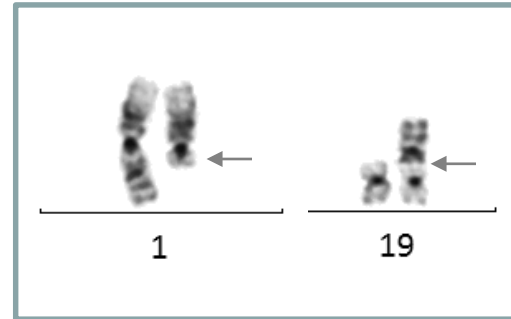
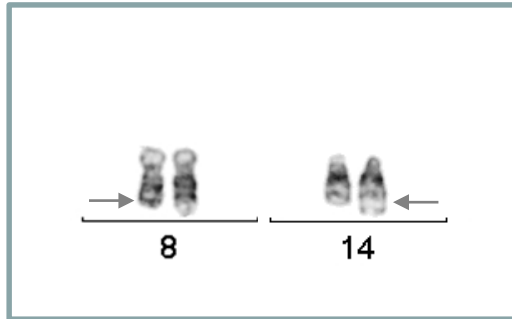
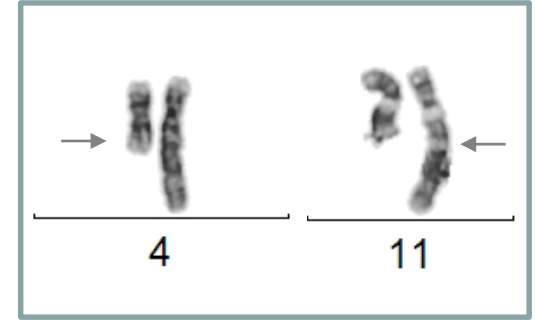
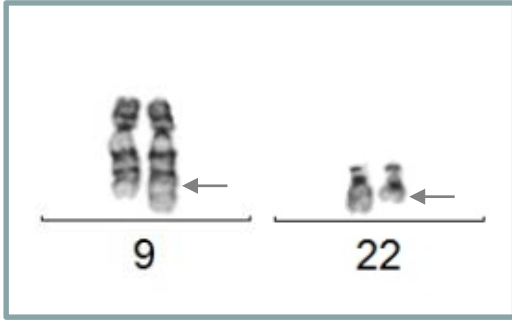
- Karyotype
  - RPMI 1640/FCS
  - MarrowMax
- FISH
  - CEP 4, 10, 17
  - BCR/ABL1
  - ETV6/RUNX1
  - KMT2A
  - CRLF2

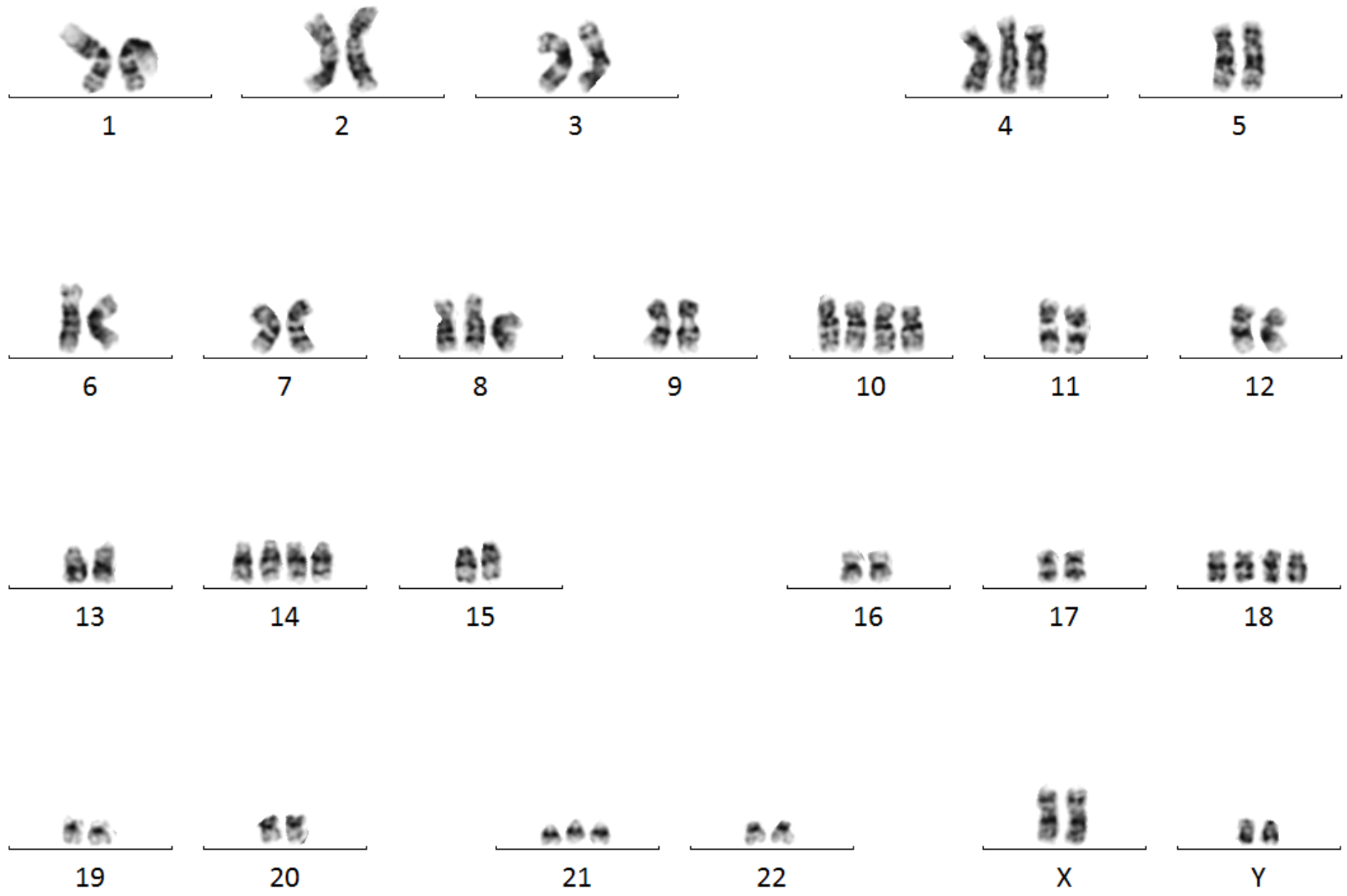
## COG testing

- FISH as previously plus
  - ABL1
  - ABL2
  - PDGFRB



# Karyotype

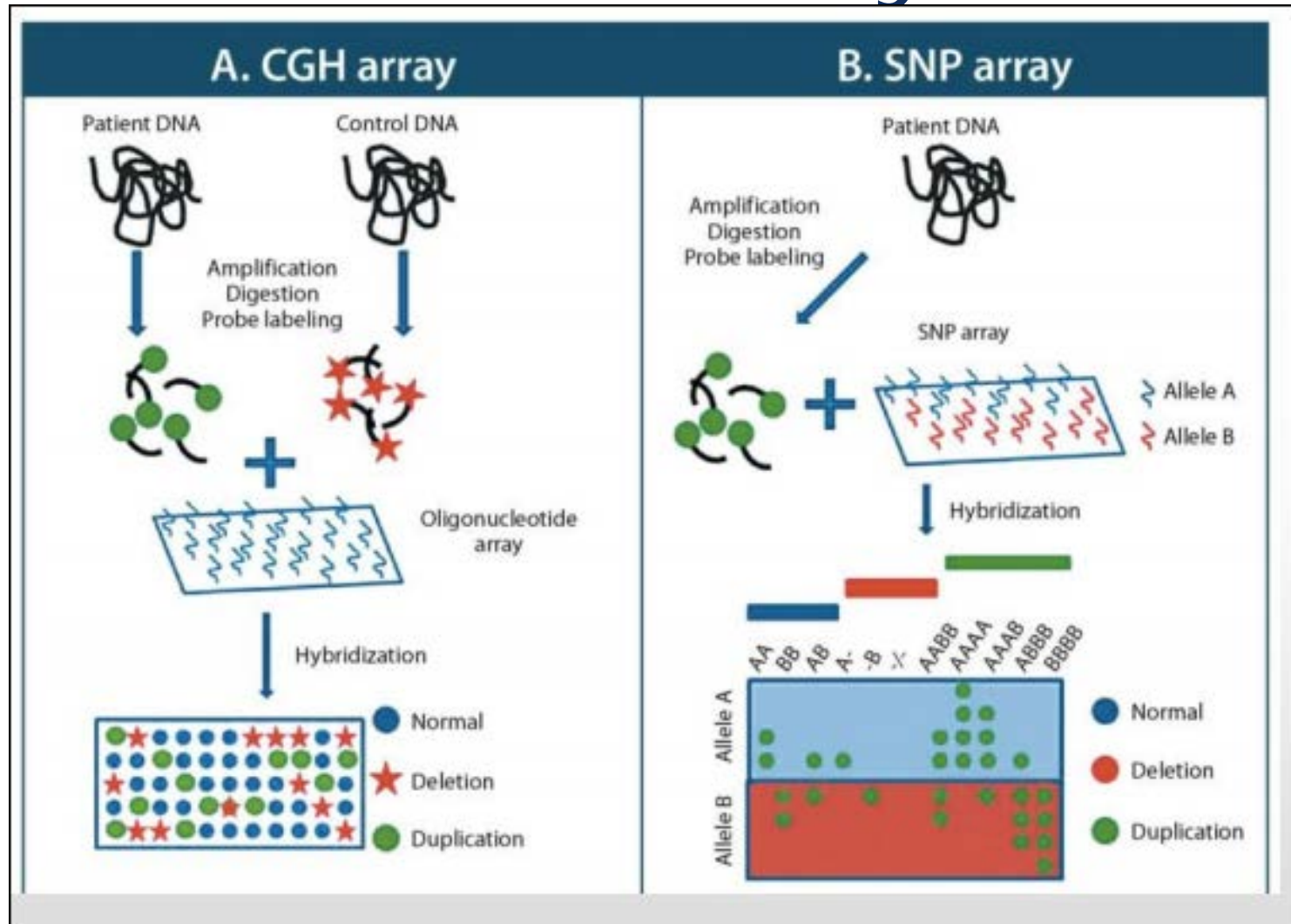




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# Microarray

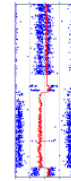
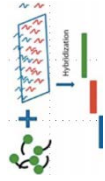


<https://www.seap.es/documents/10157/1582779/L%26L17+009+-+Molecular+Diagnosis+in+Leukemia-Lymphoma+-+Salgado.pdf/8da31673-1474-4c30-bee2-6d9e703f9bb0>



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**Day 1**

Extract DNA

**Day 2-7**

Batch processing/  
hybridisation

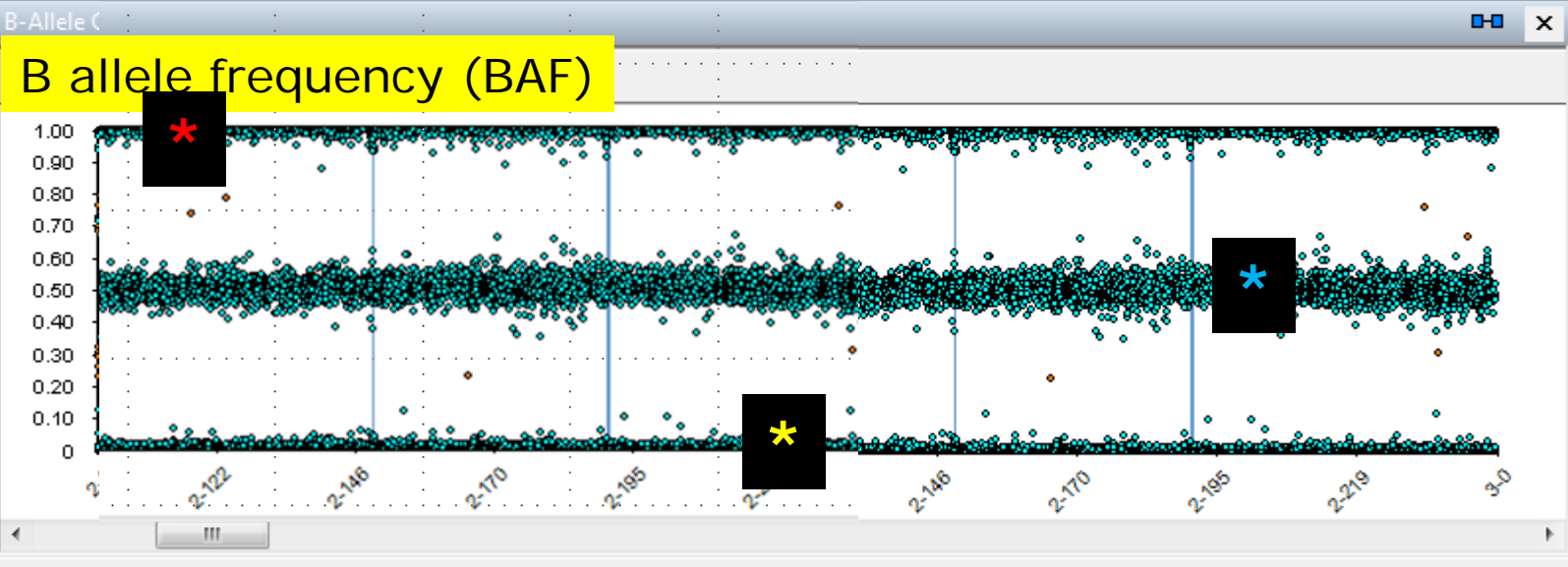
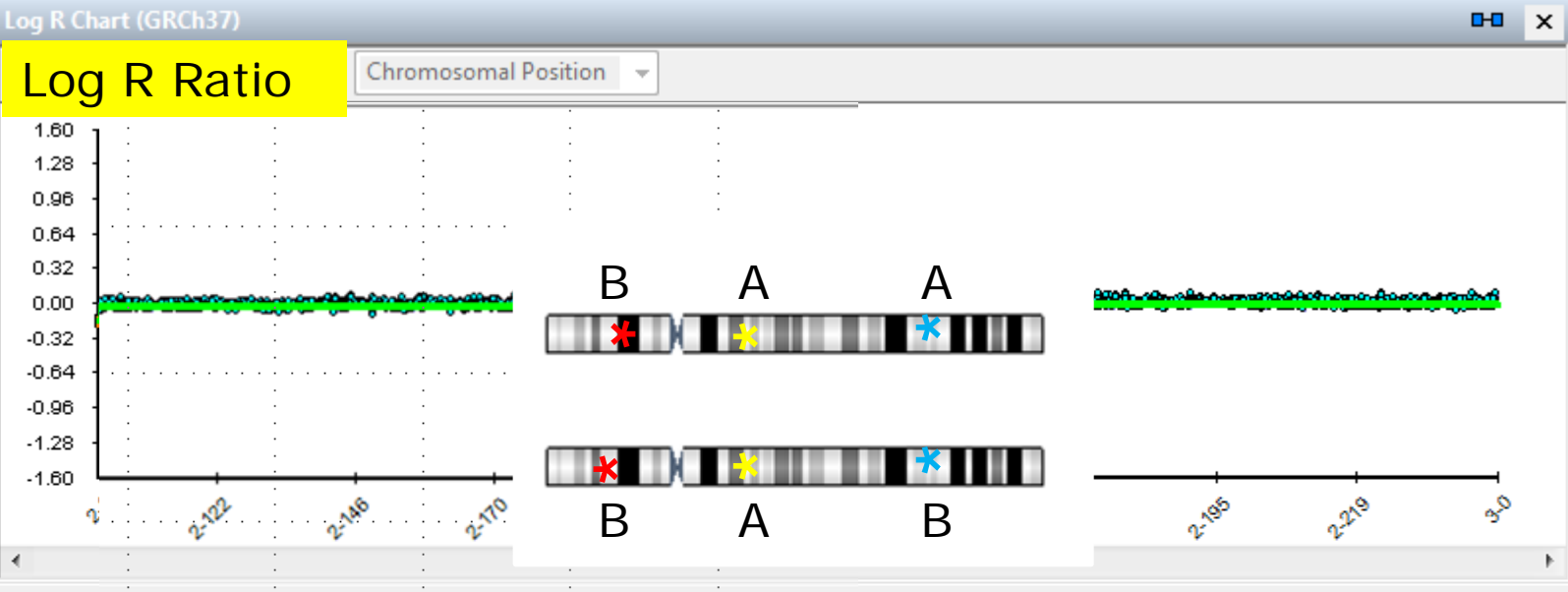
**Day 7**

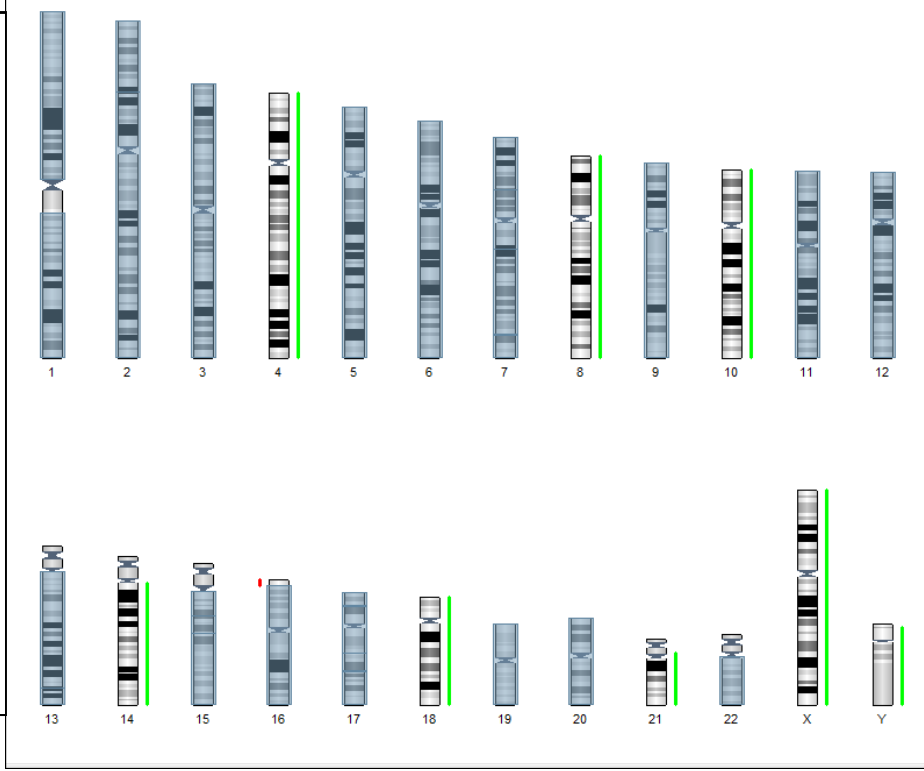
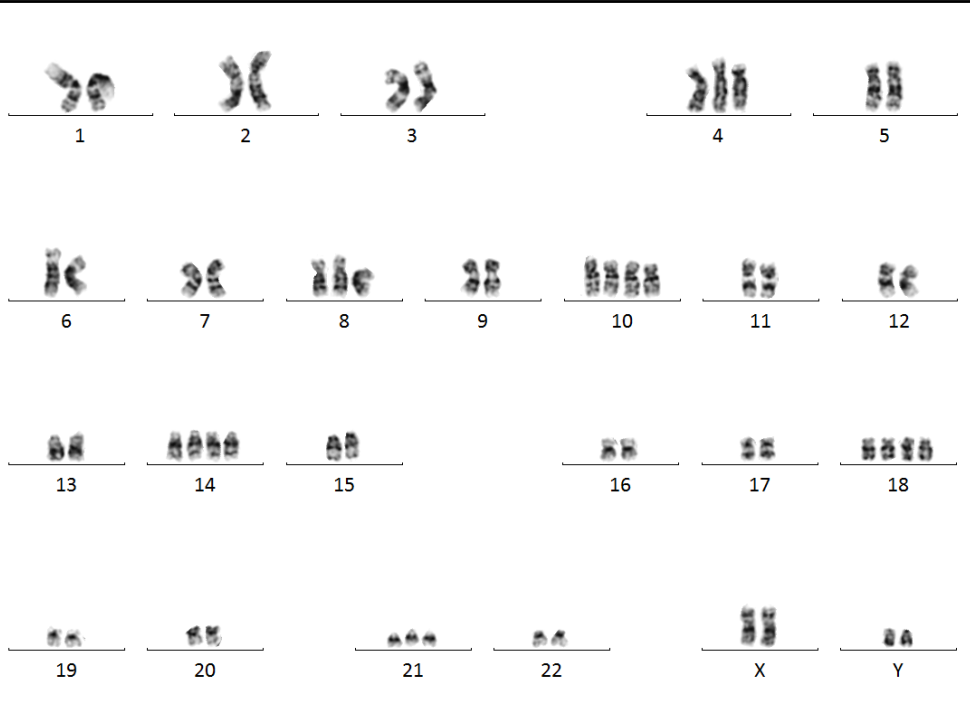
Analyse/Report



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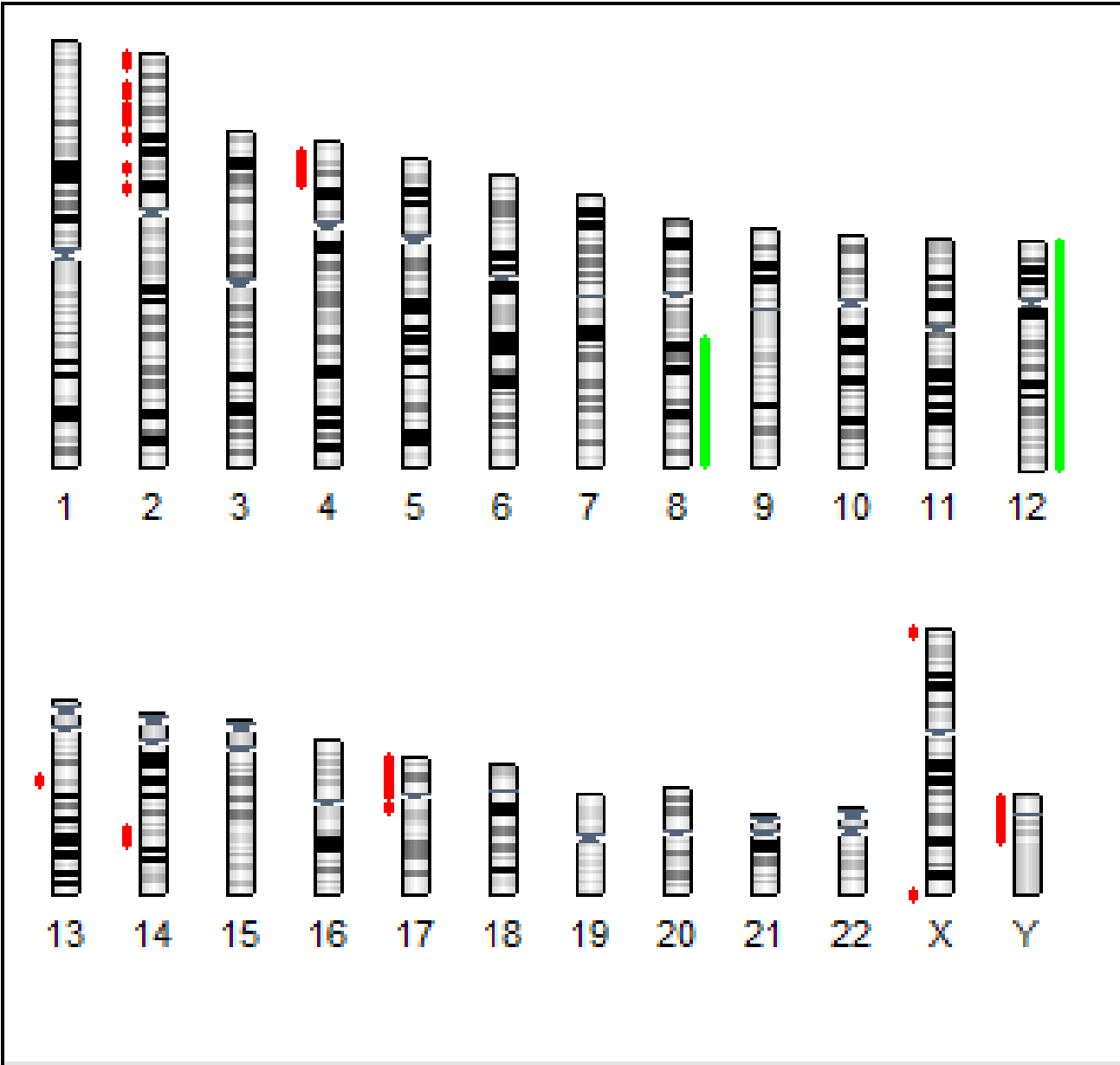
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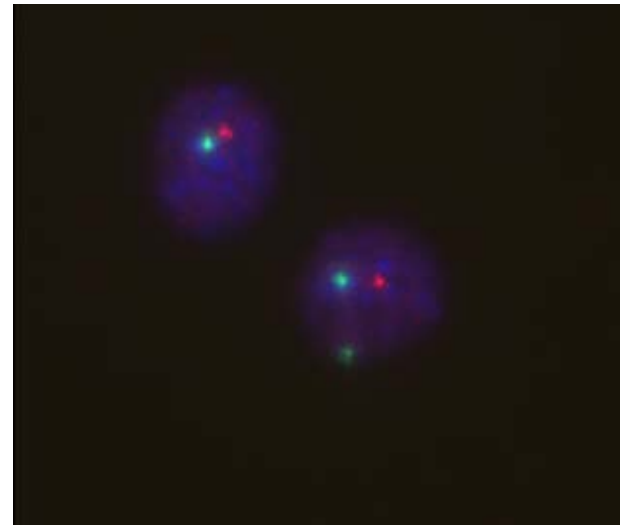
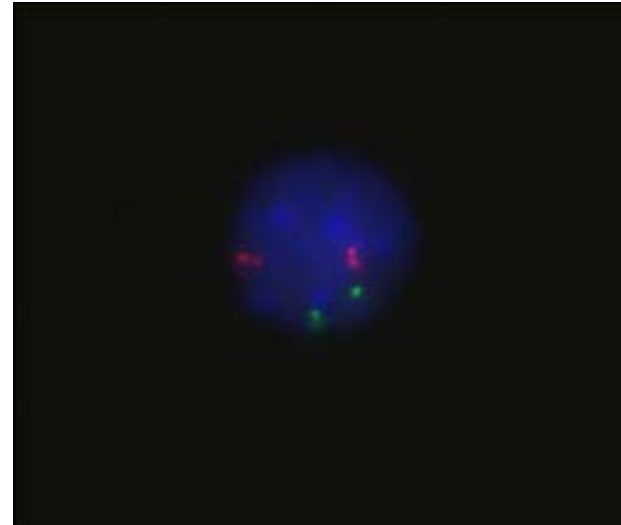
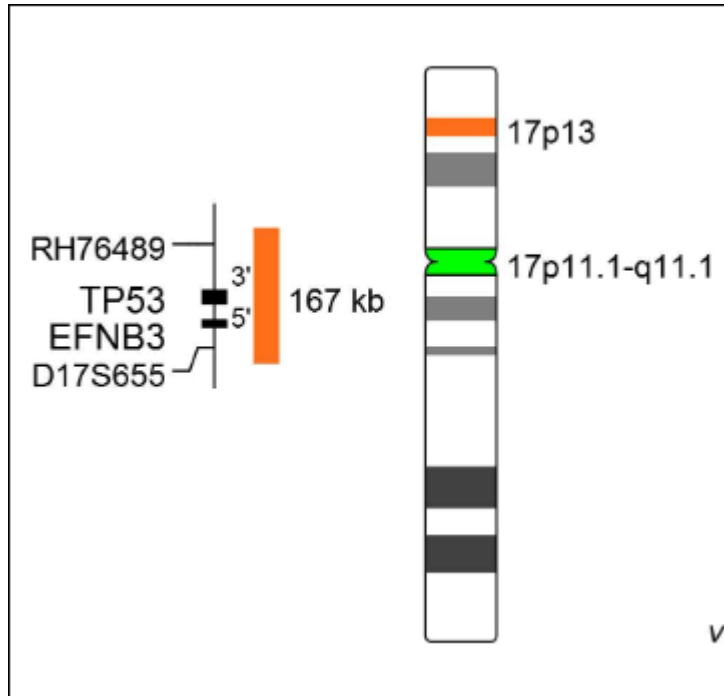
# Chronic Lymphocytic Leukaemia

- Microarray
  - Check that there are at least 30% abnormal lymphocytes
- FISH
  - Greater sensitivity
    - 11q-
    - +12
    - 13q-
    - 17p-





# TP53 FISH



Sensitivity 2%~7% depending on Lab MU



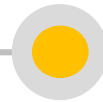
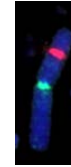
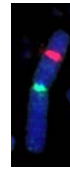
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# Myeloma

- FISH on samples enriched for CD138+
  - 1p/1q
  - TP53
  - CCND1
  - IGH
  - t(4;14)
  - t(14;16)
- Microarray if ?Hyperdiploidy







**Day 1**

Receive sample

**Day 1**

CD138+ enrich

**Day 3**

Hybridise

**Day 4**

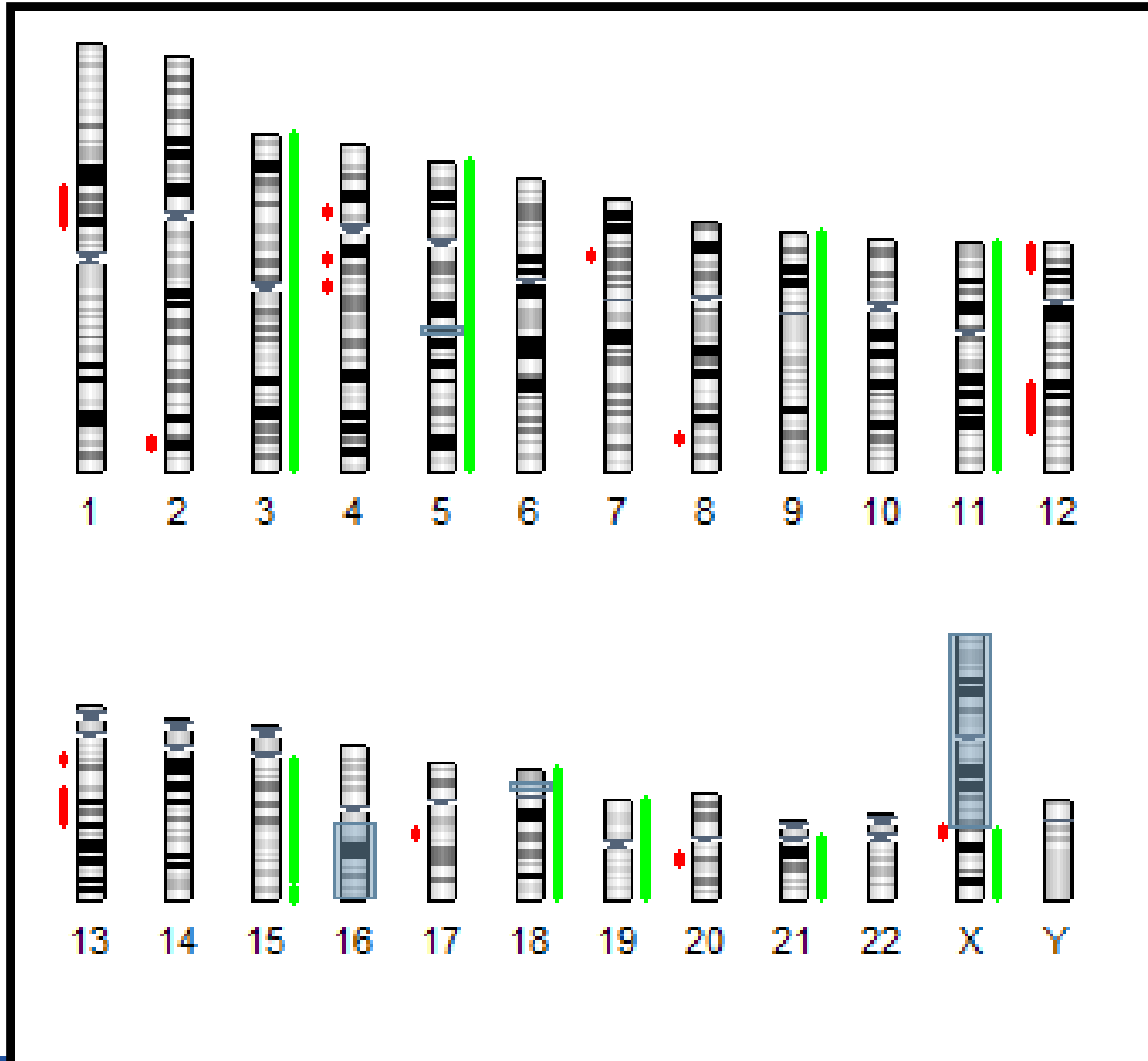
Analyse/Report



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# Microarray



- **Karyotyping** remains the only clinically-available technique that that can clearly delineate independent clones and ascertain clonal evolution definitively
- **FISH** is fast and more sensitive than karyotype or microarray
- **SNP-microarray** is a medium resolution whole genome screen and is the only cytogenetic technique that can identify copy neutral LOH



# In Cancer Cytogenetics...



Horses for courses



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