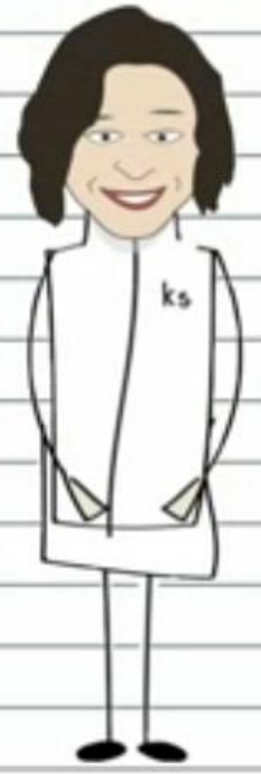
Week Three

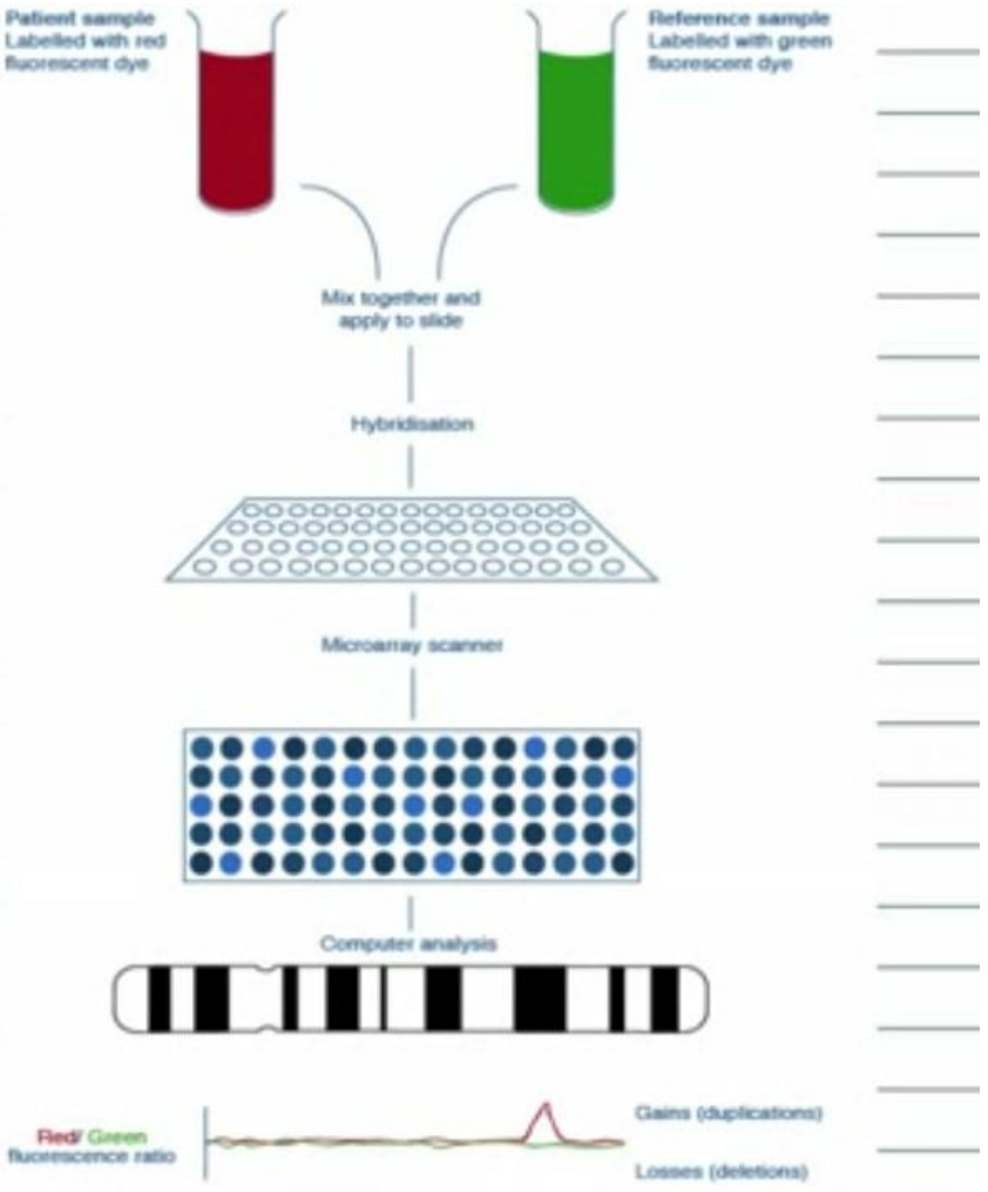
Pulling it all together



This week we have learnt about .....

1. Powerful new next generation sequencing technologies which can interrogate the entire genome cheaply and quickly

2. Array CGH, which examines the chromosomes at high resolution to detect dosage abnormalities.



3. The challenges associated with genomic technologies including:

- big data
- incidental findings
- the "needle in the haystack"
- sequence and copy number variants of uncertain significance

3. And the need to interpret reports with caution, sometimes in conjunction with specialist input

GENETIC TEST REPORT

Referrer Details:
Referrers Name: Dr Findout
Referrers Department: Oncology
Referrers Hospital: St Best Hospital

Patient Details:
Hospital ID: 12345678
NHS ID: 123 456 7891
Laboratory ID: 1234/15

Sample details:
SampleType: DNA
SampleTaken: 30/01/2015
SampleReceived: 03/02/2015

Referral reason: Serous ovarian cancer aged 55
Test: Sequencing and MLPA of *BRCA1* and *BRCA2* genes

Patient Name: Mary Smith	DOB: 31/01/60

Result:

NO PATHOGENIC MUTATION DETECTED
Unclassified variant identified
BRCA1 c.4994T>A (p.Val1665Glu)

Clinical Implications:
- A variant has been identified which requires additional expert evaluation to establish likelihood of relationship to the clinical phenotype
- This family should be referred to Genetics for further investigation
- The classification of this variant may change following evaluation

Reported by: Genetics Lab
Date: 18/03/2015

Authorised by: Second checker

Notes:

3.

Innocent until proven guilty

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Notes:

Key words

Variant of uncertain significance

Copy number variant

Next generation sequencing

Array CGH

Genomic technologies

Incidental findings

Big data

Come back for Week 4 -

"Genomic data in clinical practice"