



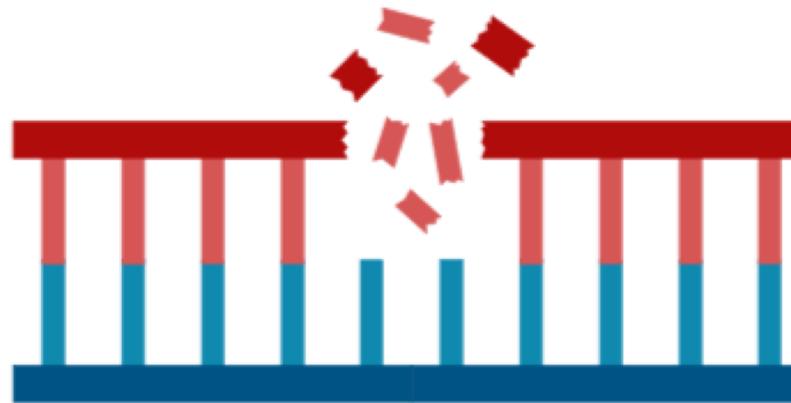
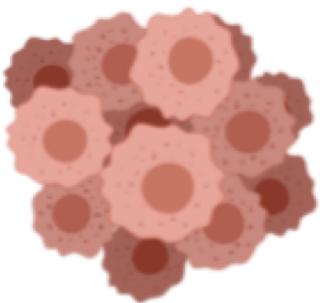
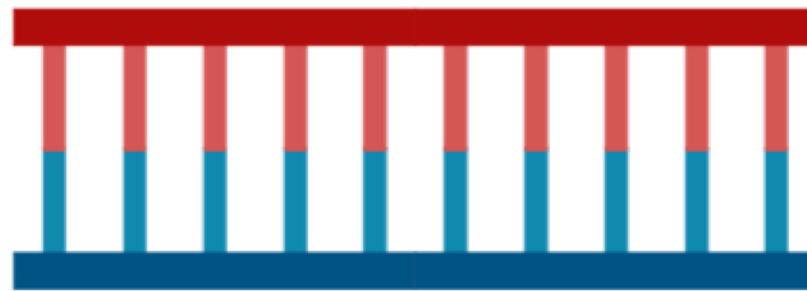
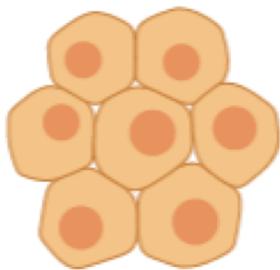
PCAWG
PanCancer Analysis
OF WHOLE GENOMES

**Justin Whalley
GMS Journal Club**

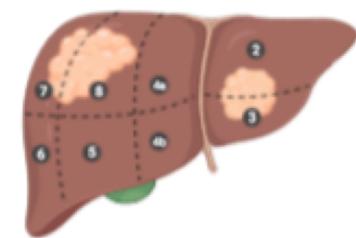
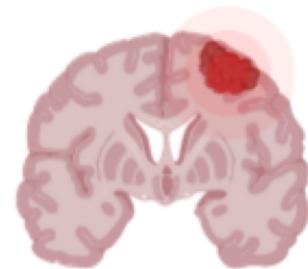
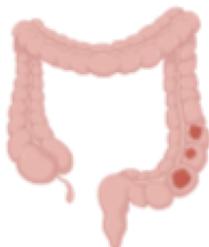
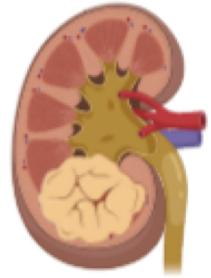
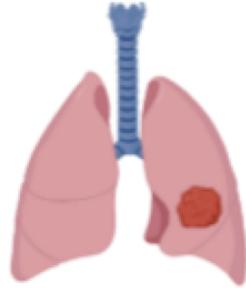
28th February 2020

Whole genome, cancer sequences

Sequence tumour sample and normal sample from the same patient.



Pan-Cancer



nature

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Pan-cancer analysis of whole genomes

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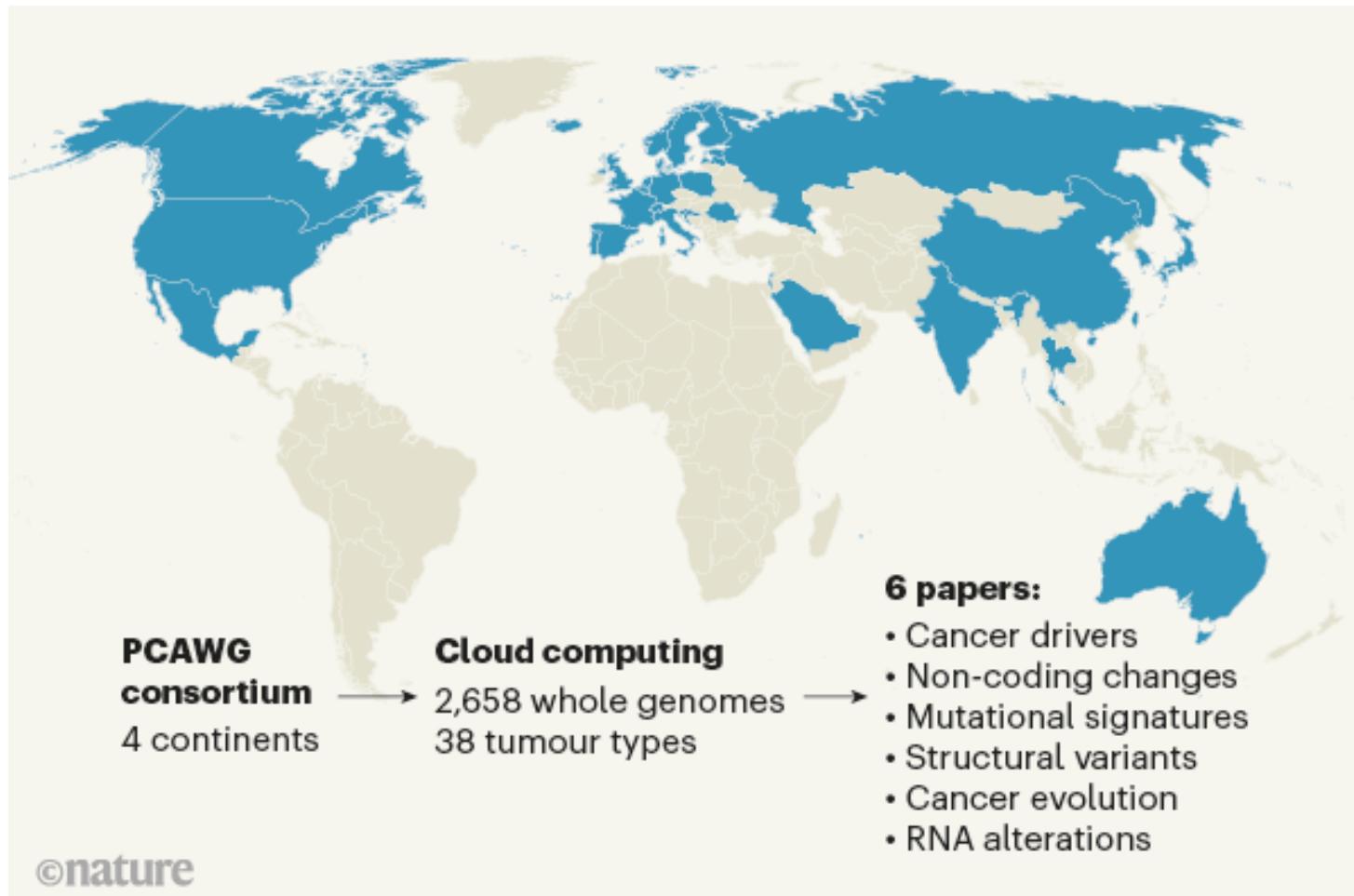
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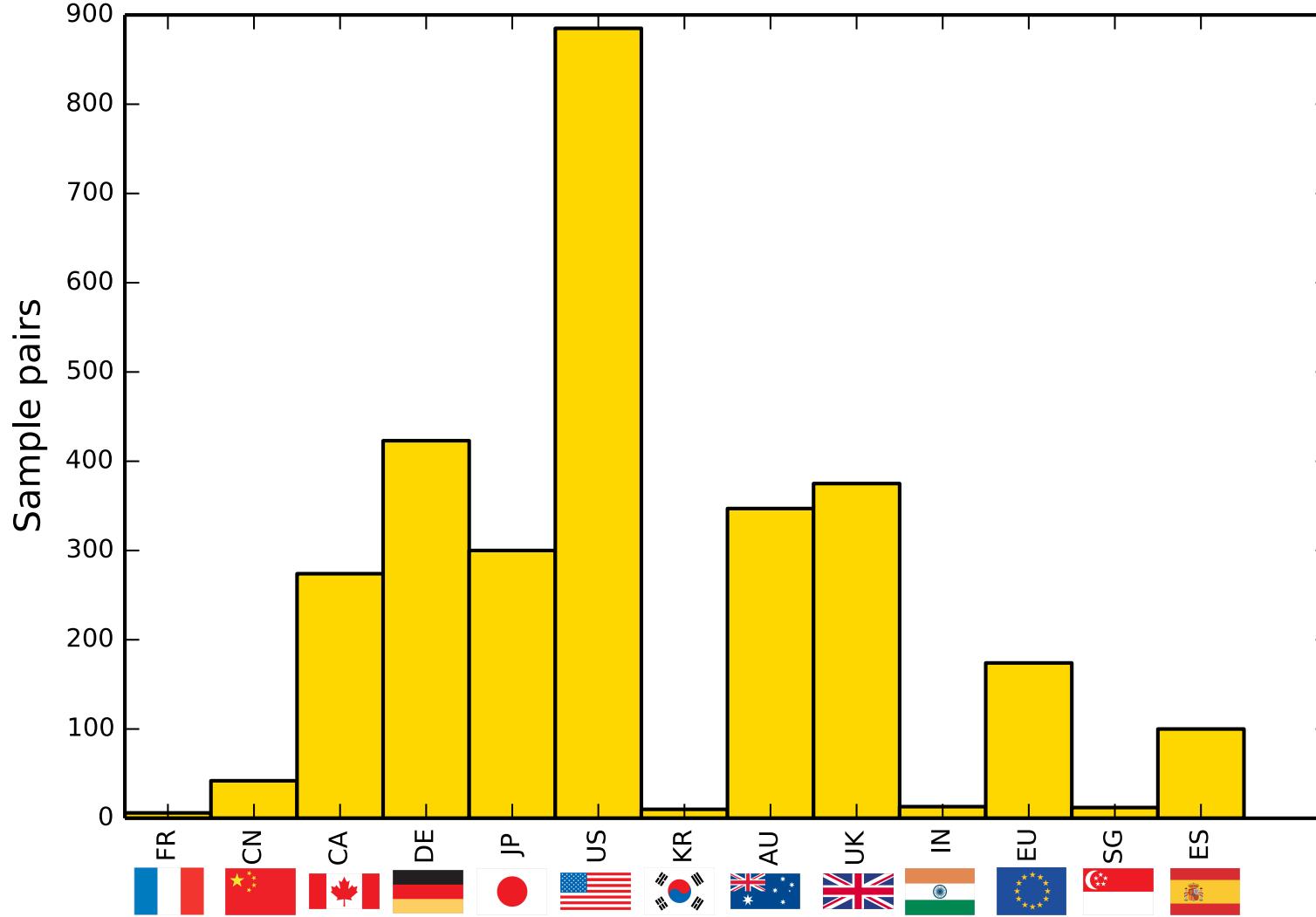


Challenges of PCAWG



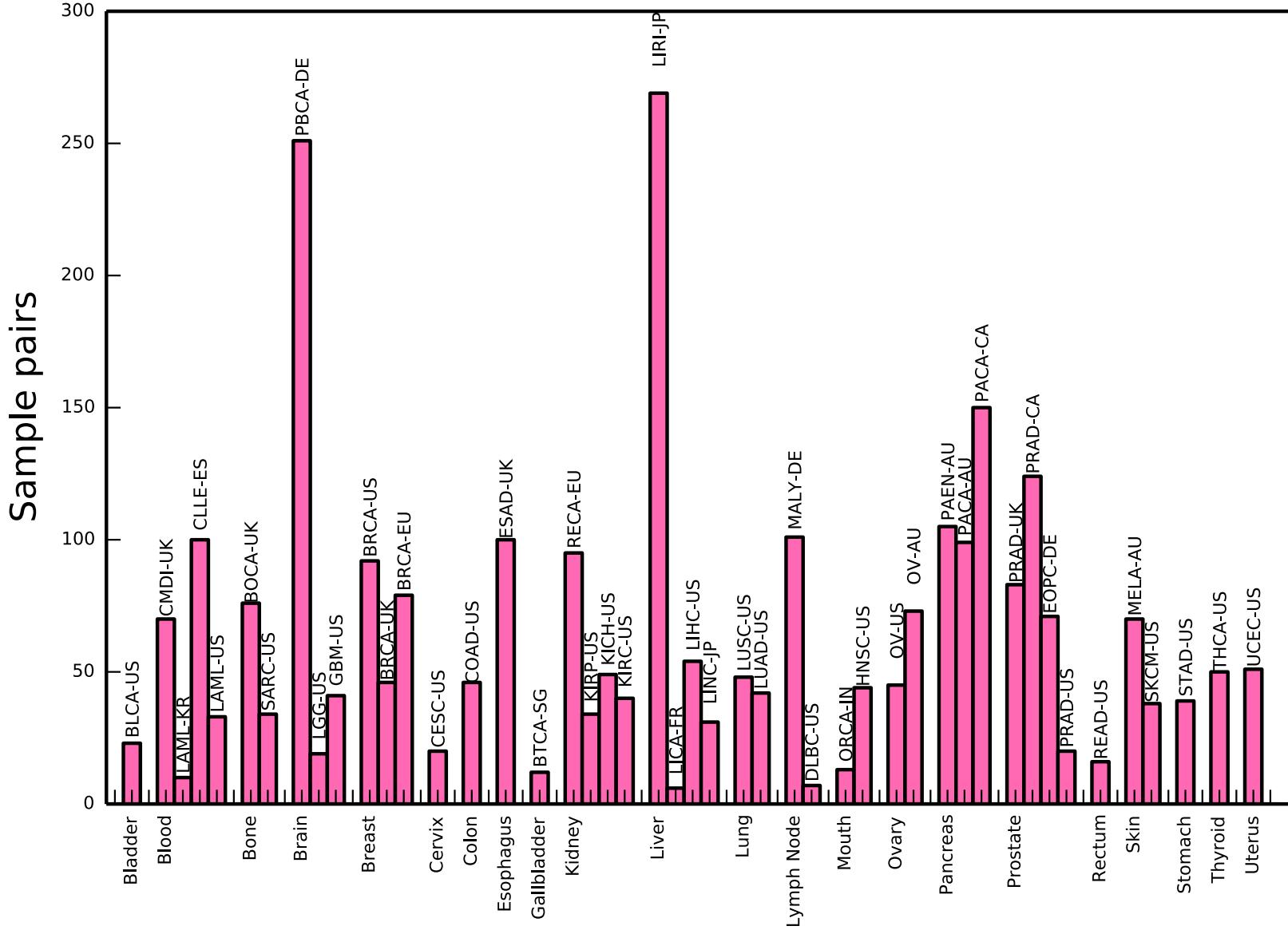
PCAWG Sequencing Centres

2591 Cancer Genomes (normal-tumour sample pairs)



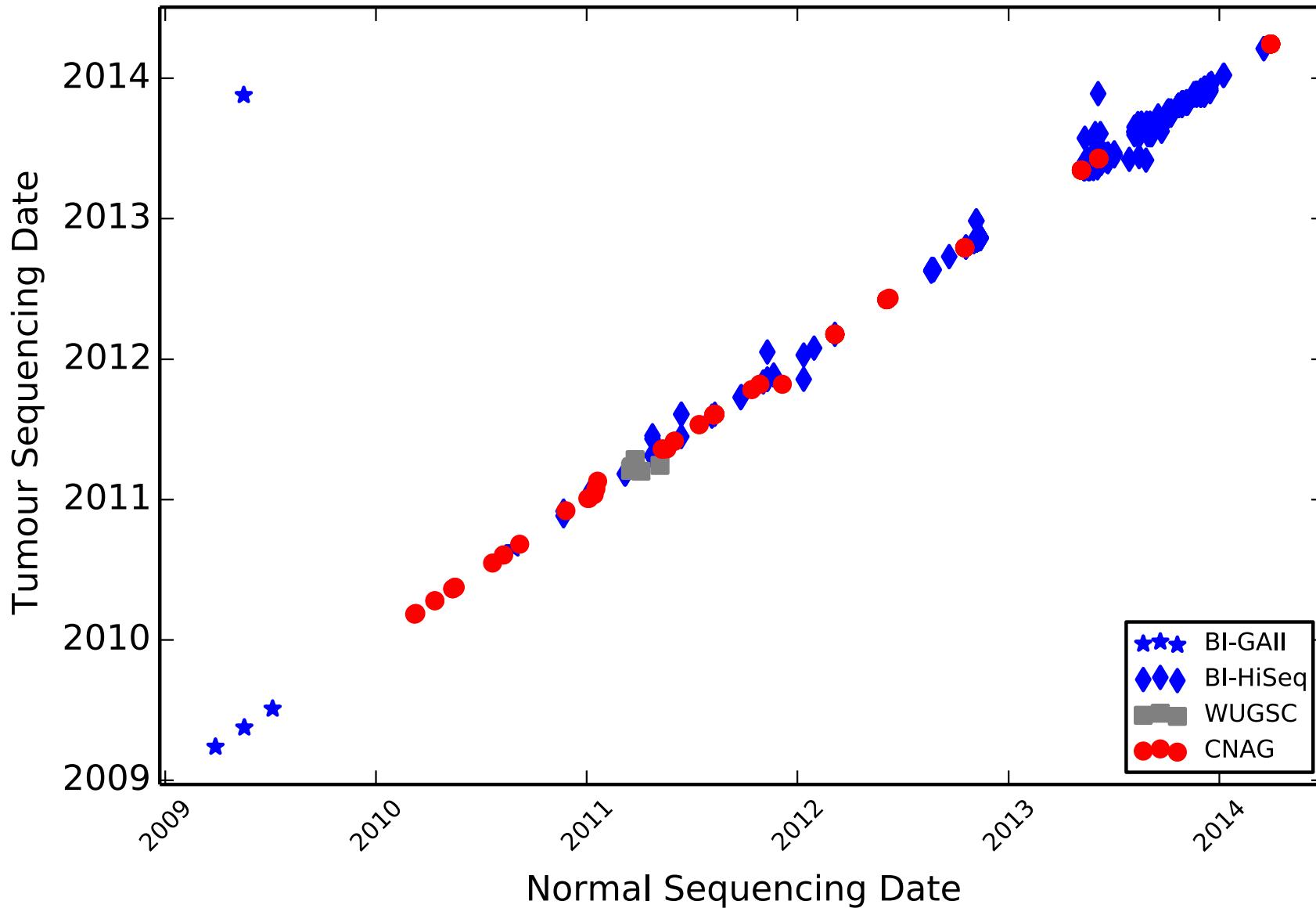
PCAWG Projects

2591 Cancer Genomes (normal-tumour sample pairs)



PCAWG Sequencing dates

Sequencing dates for CNAG, Broad, Wash.U.



PCAWG-QC working group



cnag



dkfz.



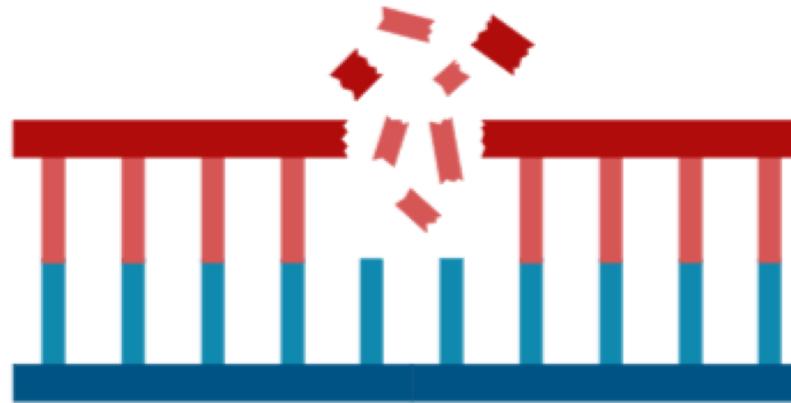
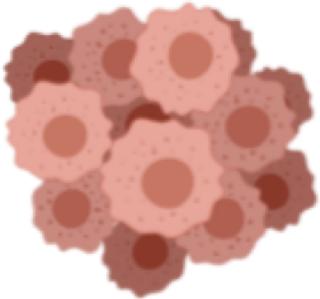
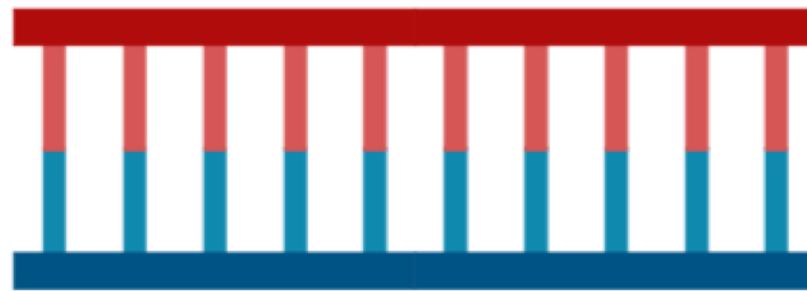
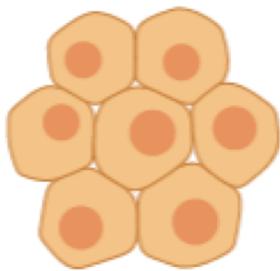
UCSF

University of California
San Francisco



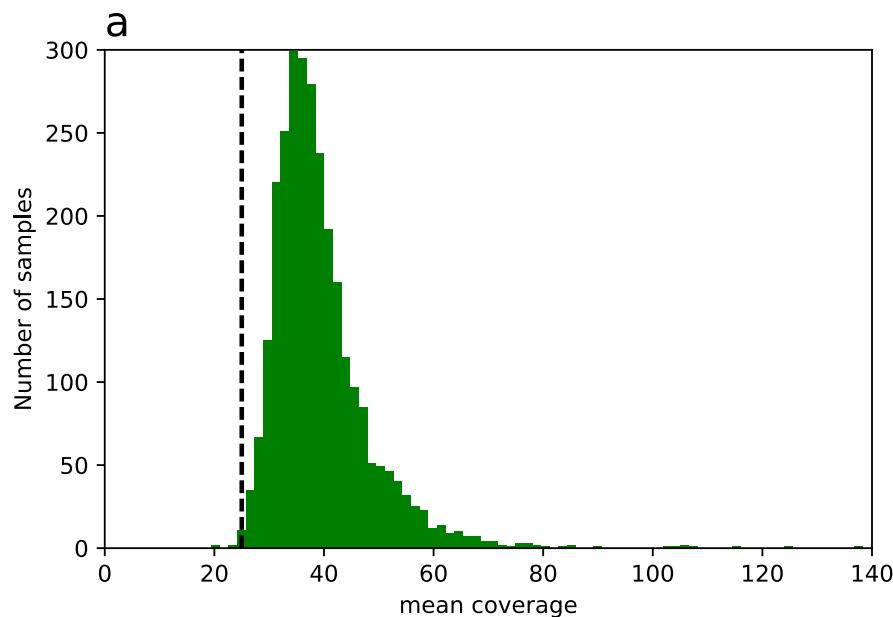
Whole genome, cancer sequences

Sequence tumour sample and normal sample from the same patient.

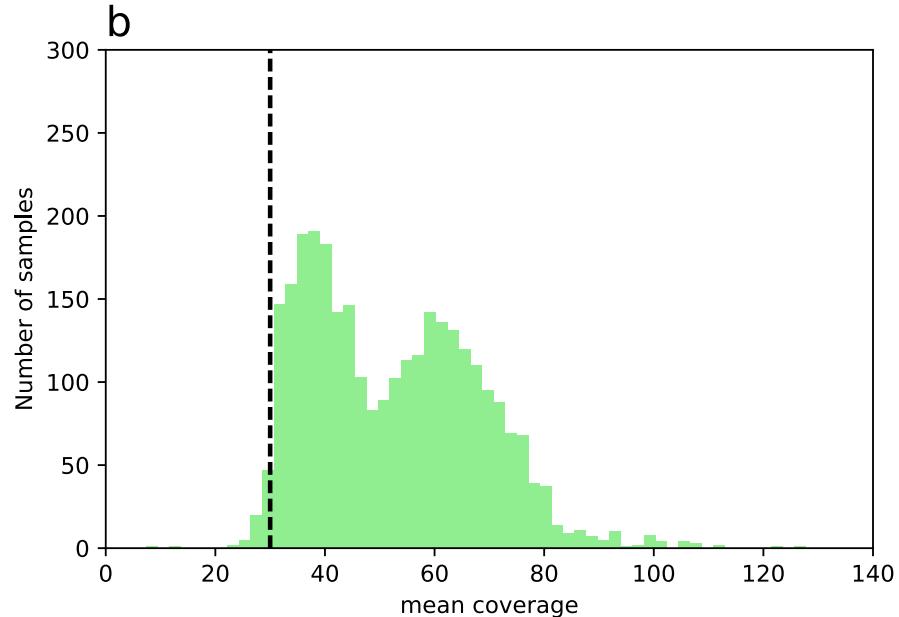


1) Mean coverage

- Excluding duplicates and low quality.
- The coverage should be more than 25X for normal and more than 30X for tumour samples.



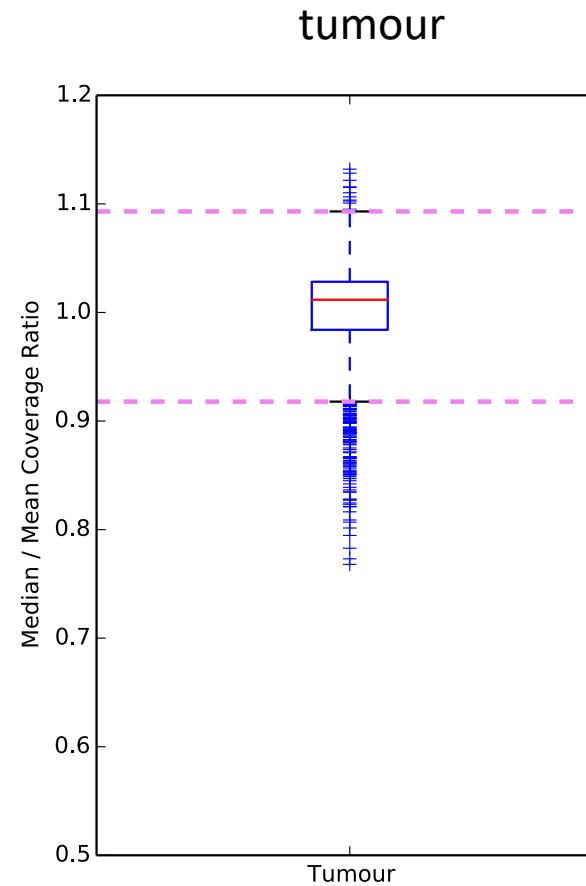
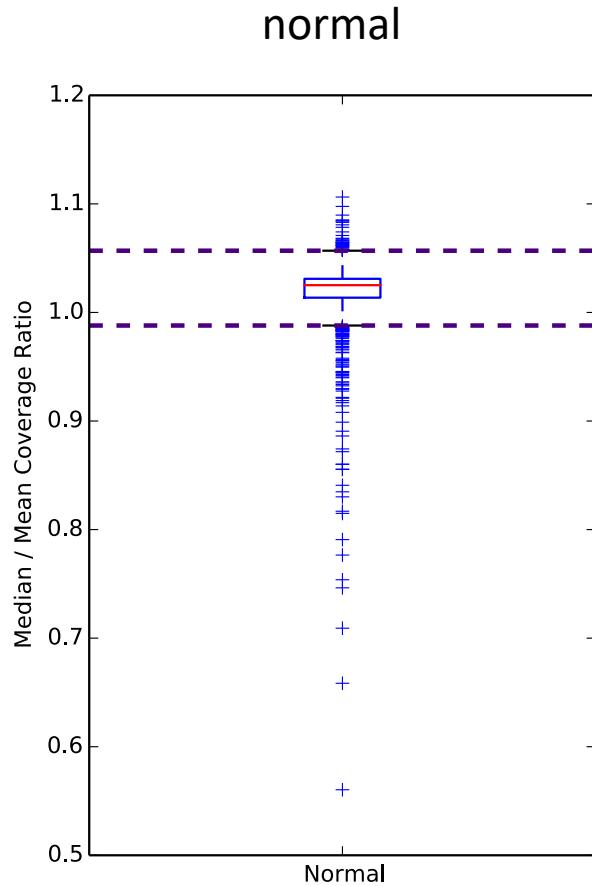
For the normal samples, 0.4% are below the threshold



For the tumour samples, 2.2% are below the threshold

2a) Evenness of coverage

- First measure the Median_coverage/Mean_coverage ratio



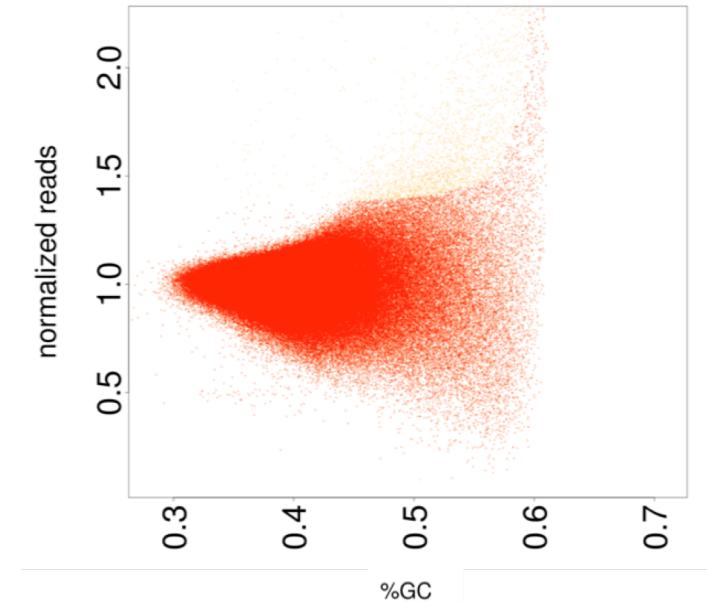
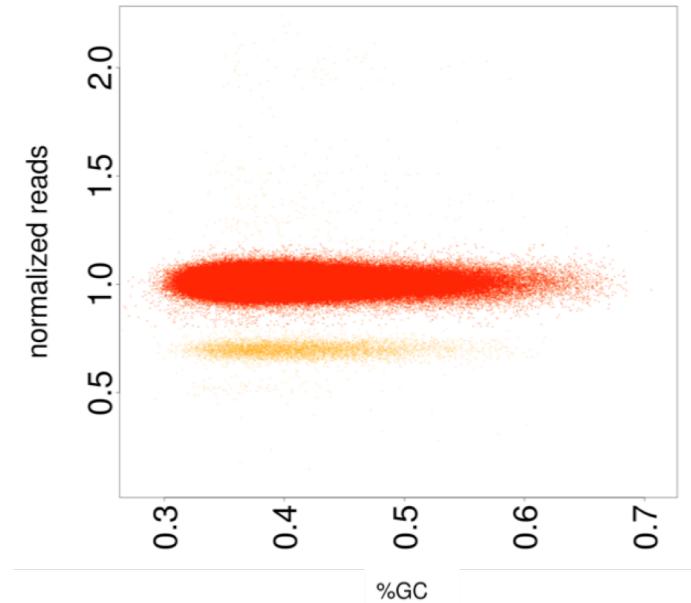
Samples which are evenly covered should be within these bounds:

for the normal samples: 0.99 – 1.06

for the tumour samples: 0.92 – 1.09

2b) Evenness of coverage

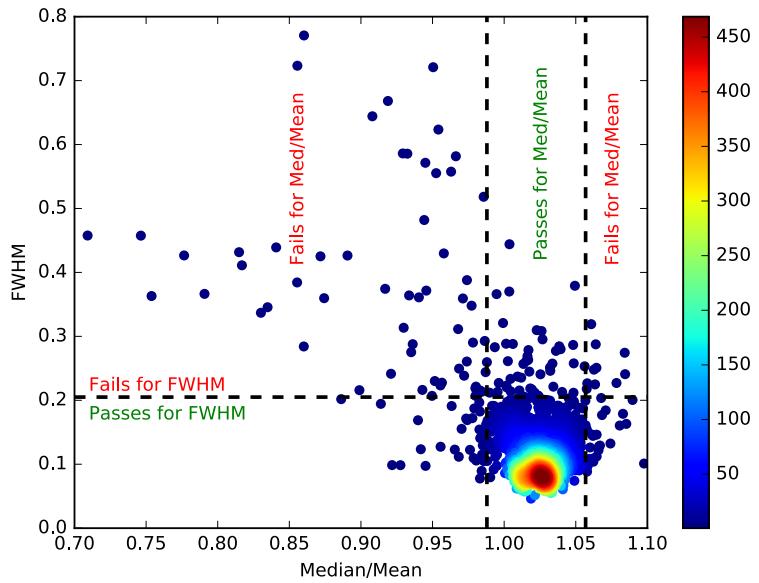
- Second measure the full width at half maximum (FWHM) of the normalised coverage after correction for GC-coverage.



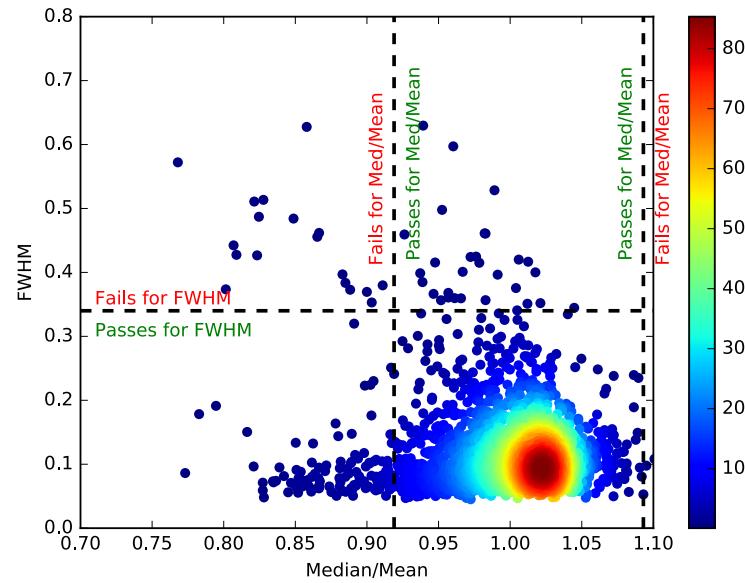
Samples with an even coverage should have FWHM values below are 0.205 for the normal samples and the 0.34 for tumour samples.

2) Evenness of coverage

For a sample to have evenness of coverage, it needs to be within the thresholds set by both of our measures.



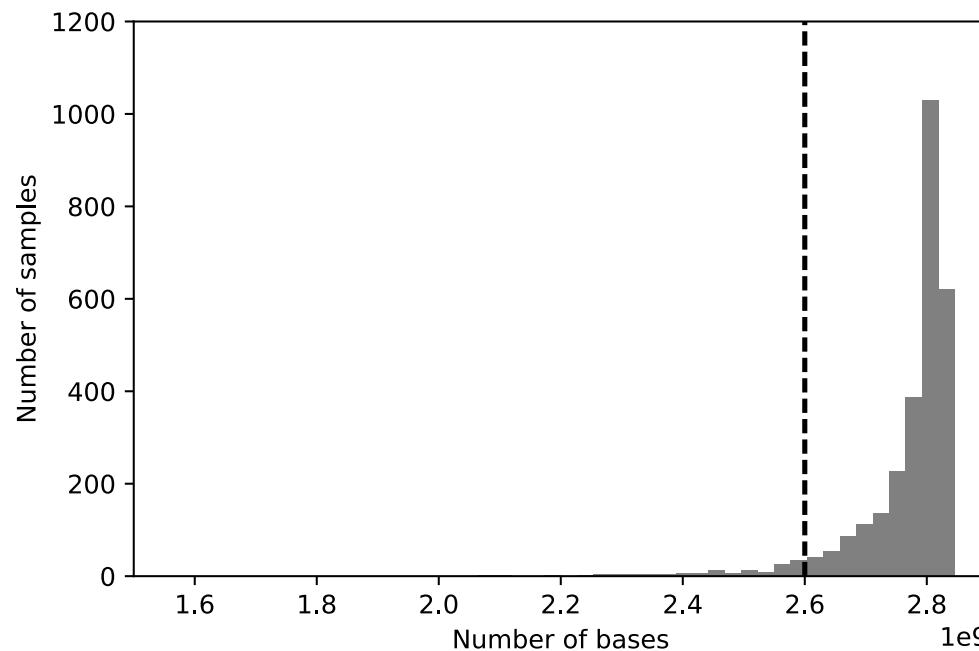
For the normal samples, 6.3% are outside the thresholds



For the tumour samples, 5.8% are outside the thresholds

3) Number of bases covered sufficiently for mutation calling.

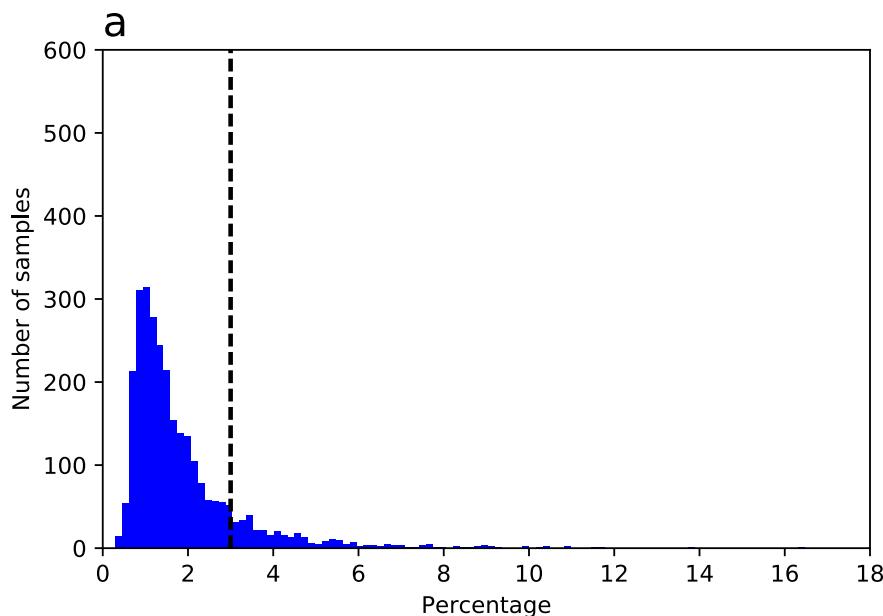
The total number of bases with sufficient coverage in both the normal and tumour samples to call somatic mutations should be greater than 2.6 gigabases.



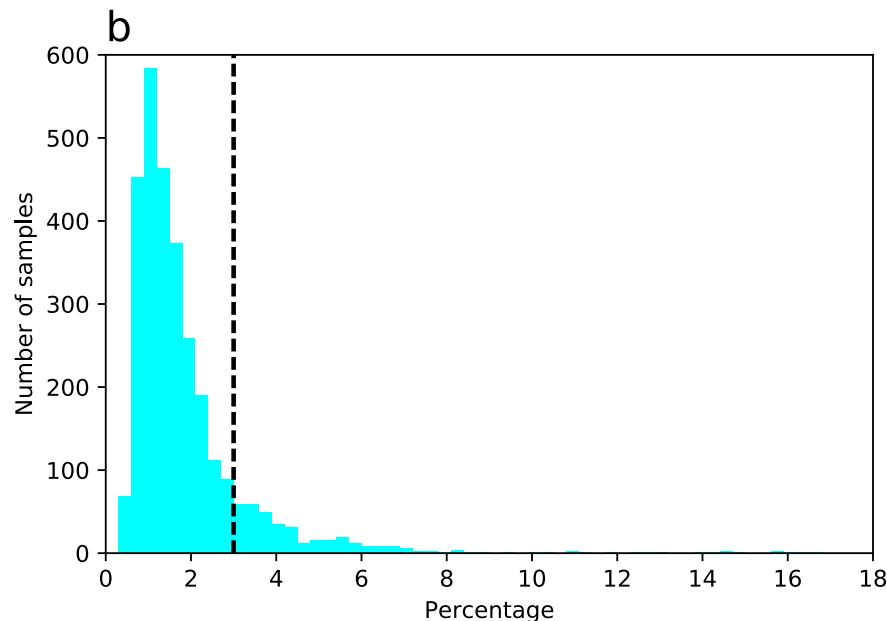
For the normal-tumour sample pairs,
6.0% are below the threshold

4) Paired reads mapping to a different chromosomes

Normal and tumour samples should have no more than 3% of paired reads mapping to different chromosomes.



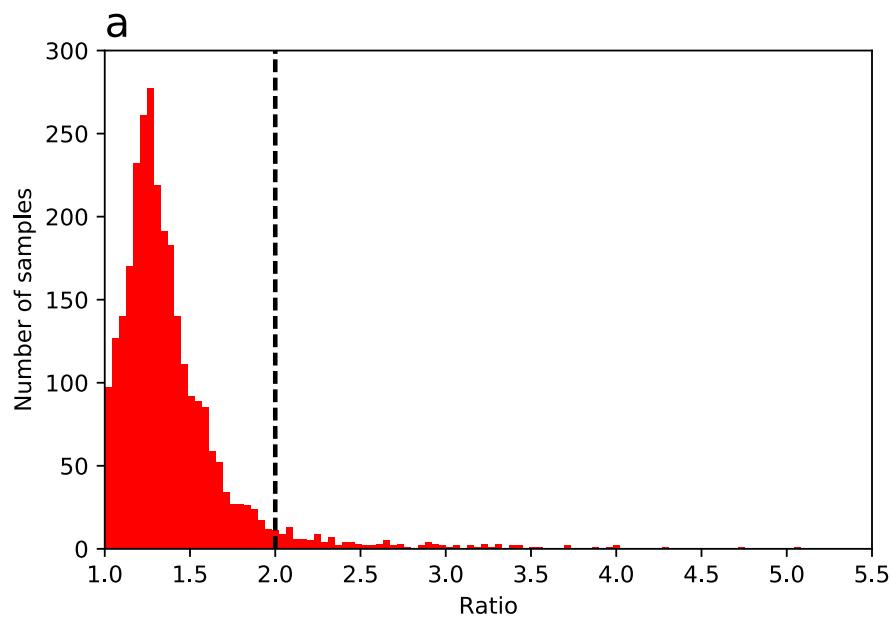
For the normal samples, 15.5% are above the threshold



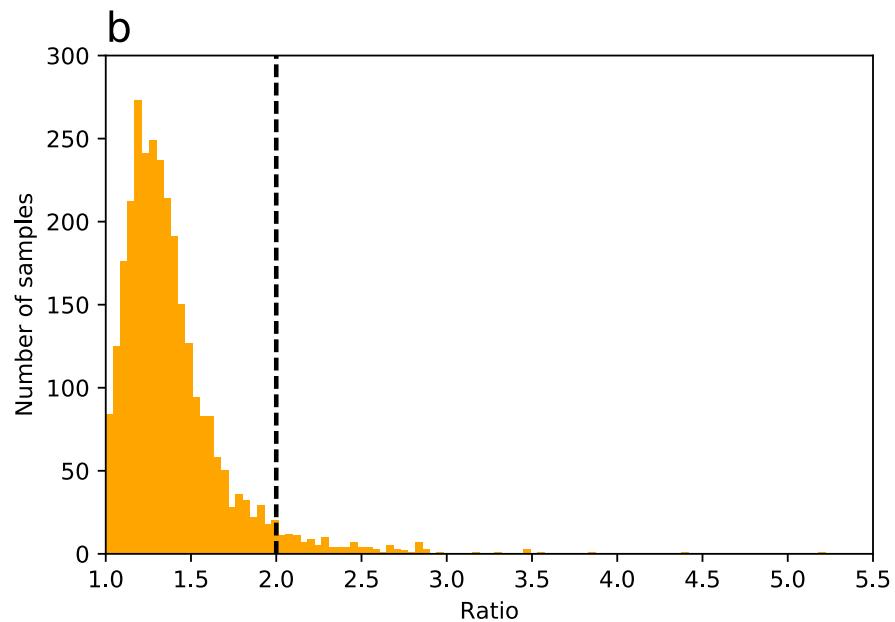
For the tumour samples, 13.0% are above the threshold

5) Ratio of difference in edits between paired reads

The ratio of the sum of edits for the paired reads should not be greater than 2 for both normal and tumour samples.

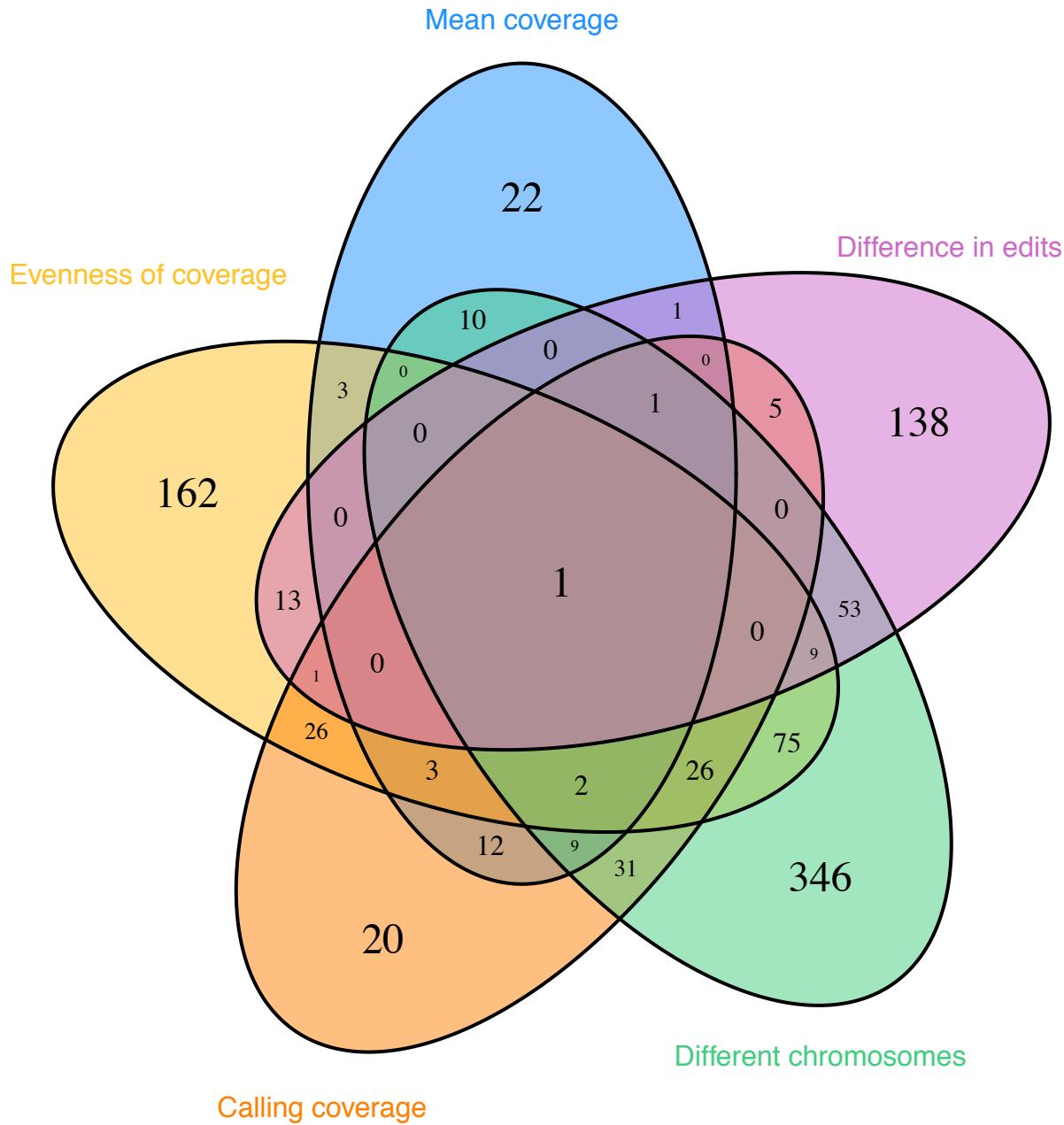


For the normal samples, 4.7% are above the threshold

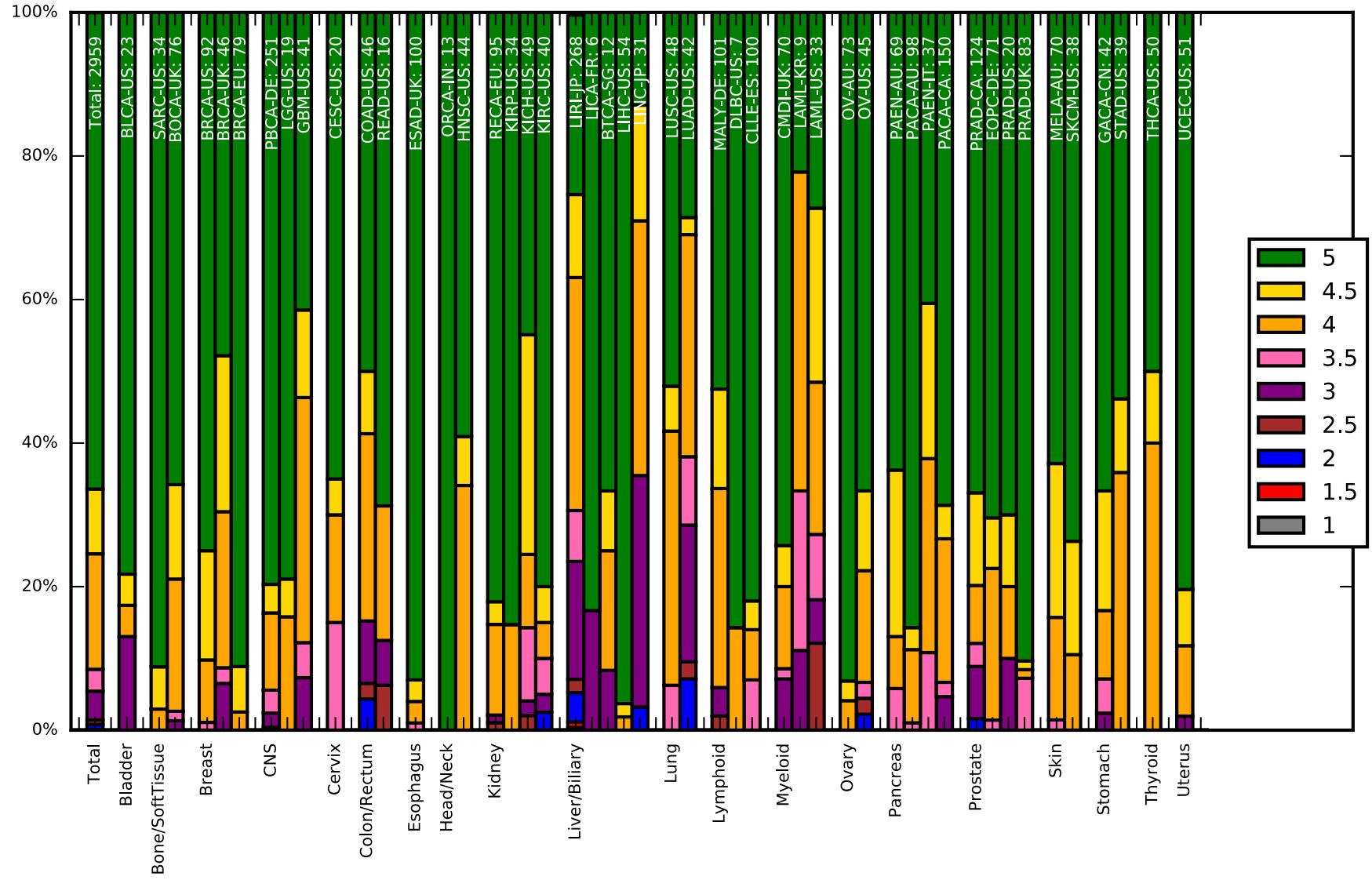


For the tumour samples, 4.5% are above the threshold

Are the QC measures non-redundant?



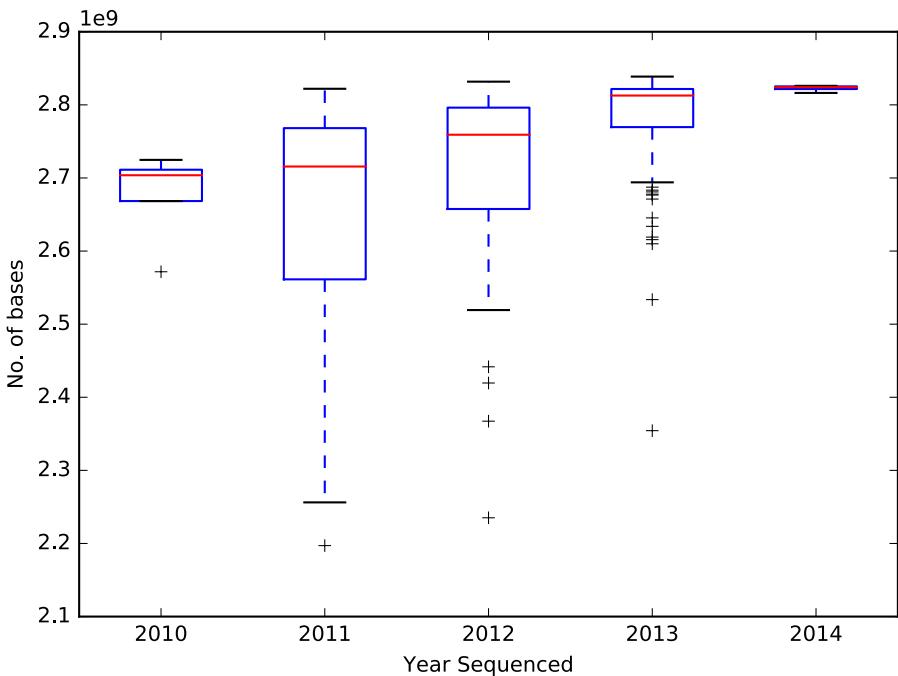
Star rating summarised for 2959 samples



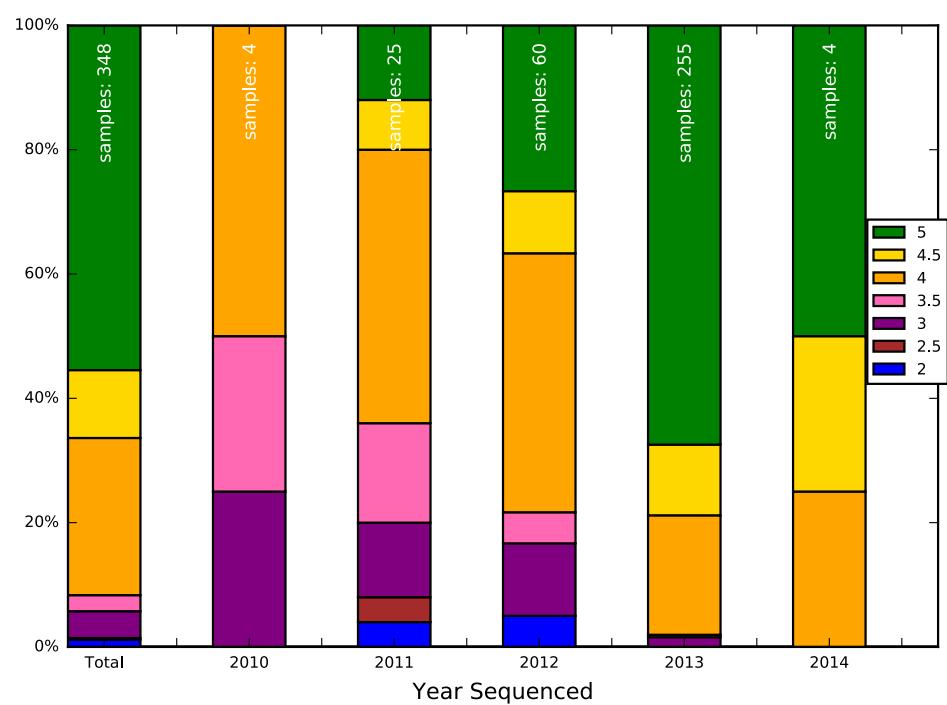
QC measures vs historical data

Samples sequenced at the Broad Institute

a



b

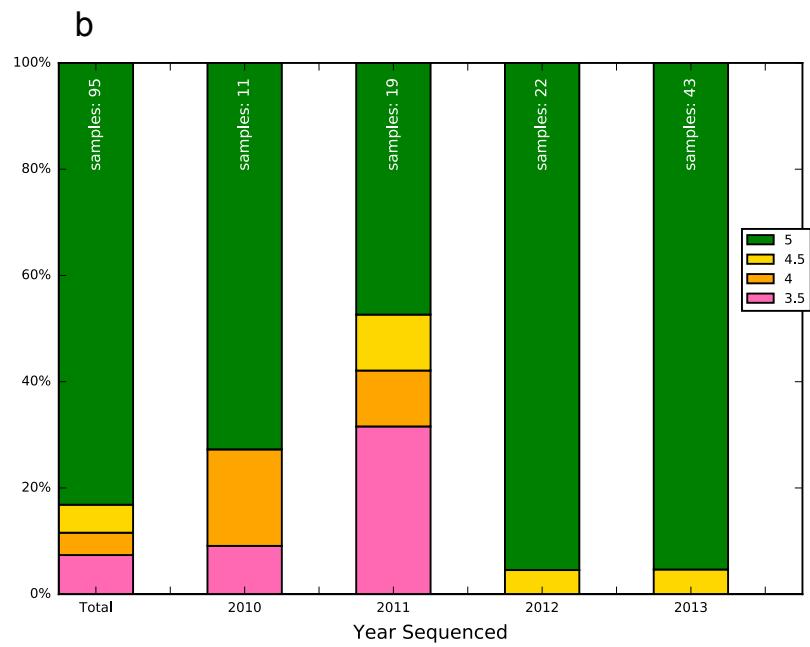
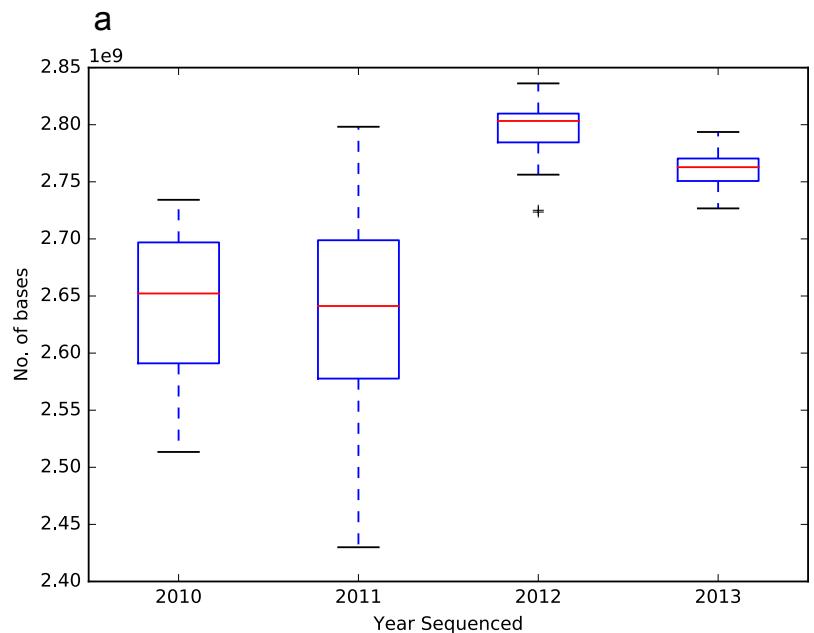


Somatic Calling Coverage

Star Rating

QC measures vs historical data

Samples sequenced at CNAG

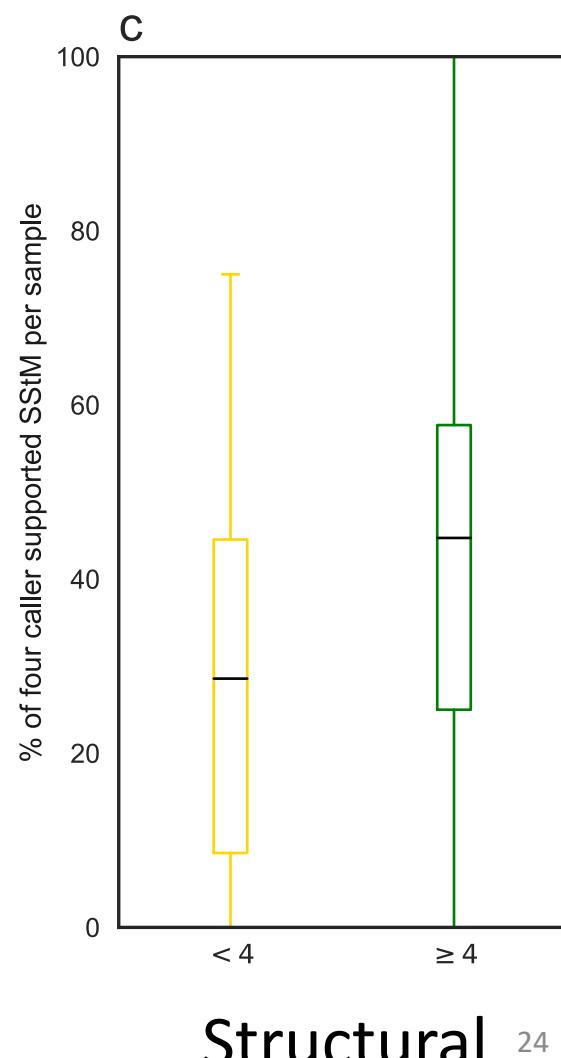
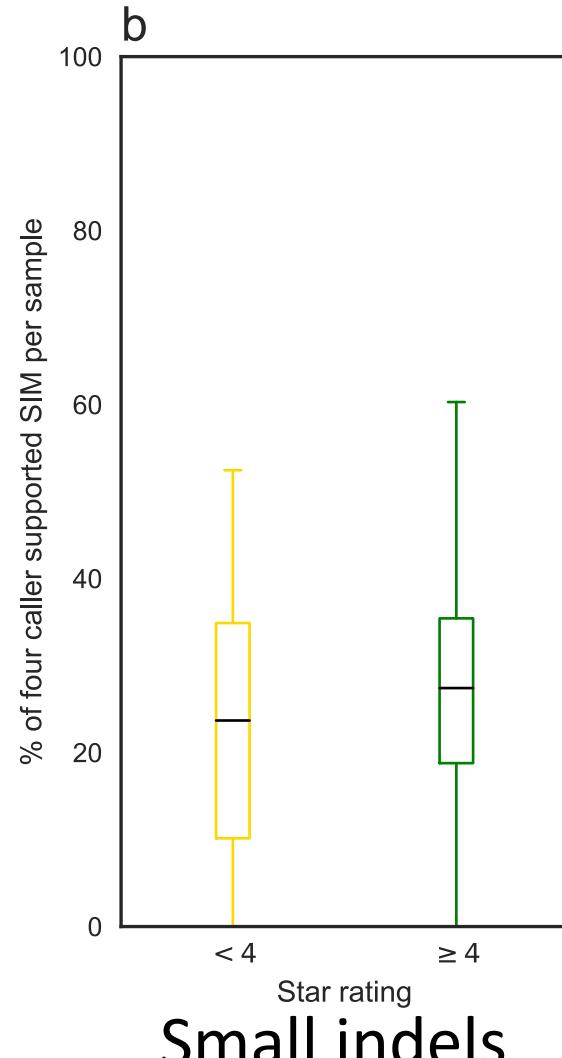
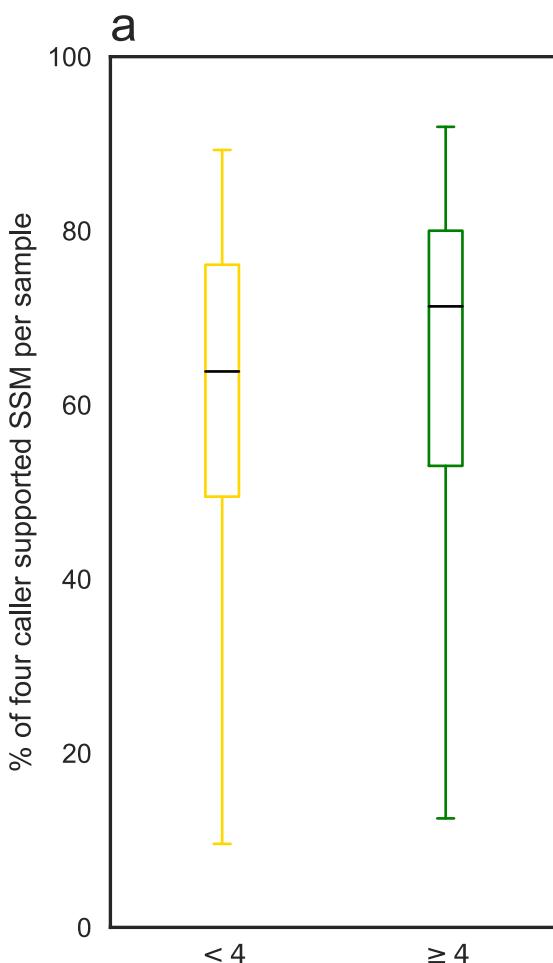


Somatic Calling Coverage

Star Rating

QC measures vs downstream analyses

Star rating vs % of somatic mutation callers agreeing?



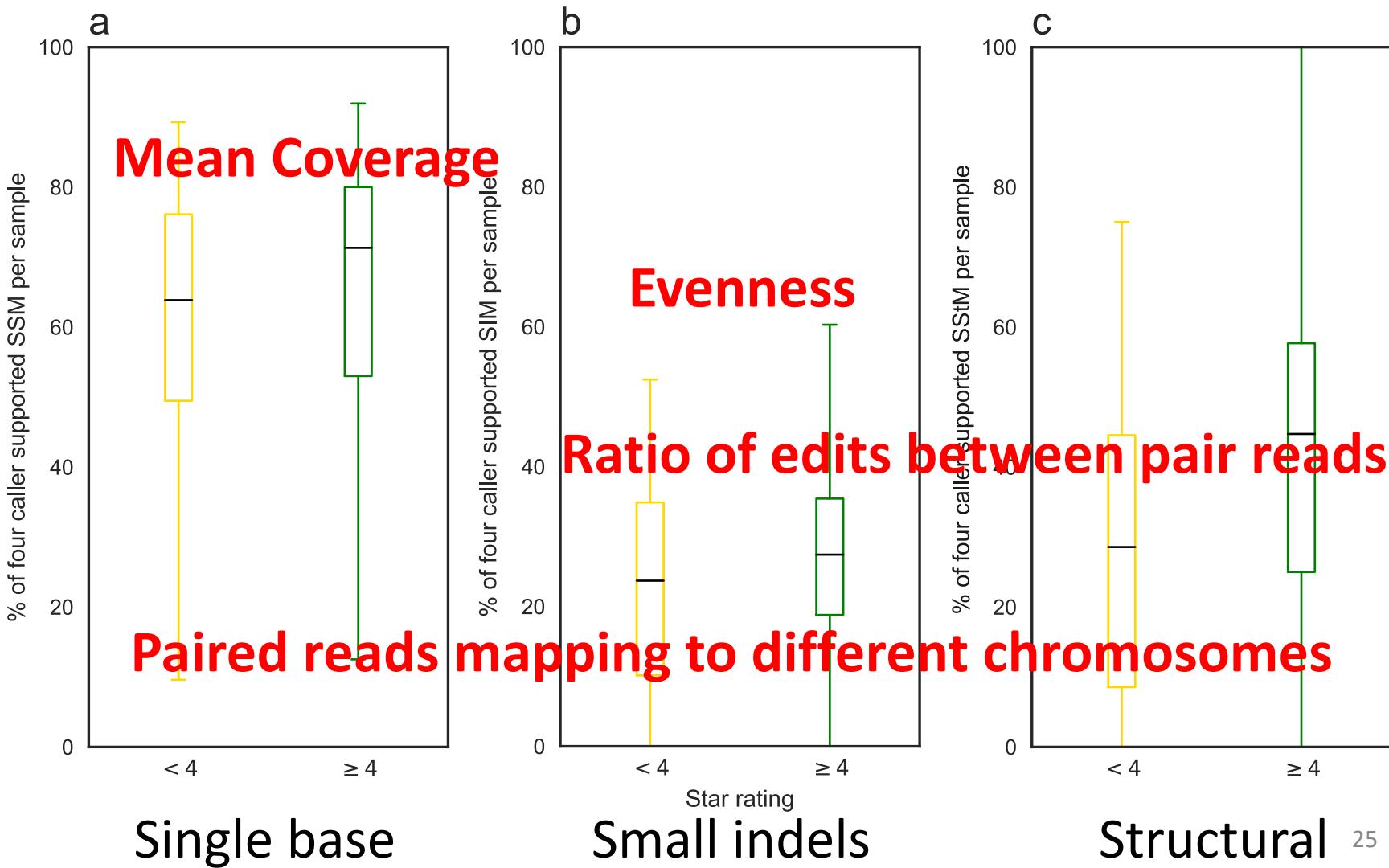
Single base

Small indels

Structural

QC measures vs downstream analyses

Star rating vs % of somatic mutation callers agreeing?



PCAWG-QC Working Group

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Sanger

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PCAWG-QC Working Group



x 22

PCAWG-QC Working Group



x 22



International
Cancer Genome
Consortium

DRAFT AGENDA
PanCancer Analysis of Whole Genomes (PCAWG)
Face-to-Face (F2F)
April 7-8, 2016 – Barcelona, Spain

PCAWG-QC towards publication



Cold
Spring
Harbor
Laboratory



Framework for quality assessment of whole genome, cancer sequences

Justin P. Whalley, Ivo Buchhalter, Esther Rheinbay, Keiran M. Raine, Kortine Kleinheinz, Miranda D. Stobbe, Johannes Werner, Sergi Beltran, Marta Gut, Daniel Huebschmann, Barbara Hutter, Dimitri Livitz, Marc Perry, Mara Rosenberg, Gordon Saksena, Jean-Rémi Trotta, Roland Eils, Jan Korbel, Daniela S. Gerhard, Peter Campbell, Gad Getz, Matthias Schlesner, Ivo G. Gut

doi: <https://doi.org/10.1101/140921>

Posted September 19, 2017.

PCAWG-QC towards publication

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Pan-cancer analysis of whole genomes

The ICGC/TCGA Pan-Cancer Analysis of Whole Genomes Consortium

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PCAWG-QC towards publication



Framework for quality assessment of whole genome, cancer sequences

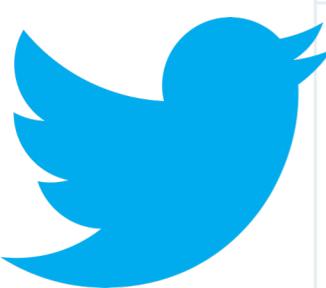
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PCAWG
PanCancer Analysis
OF WHOLE GENOMES



Nuria Lopez-Bigas

@nlbigas

▼

Glad #PCWAG is over. My lessons learned

- Collaborations and data sharing necessary to advance cancer research
- Centralised control kills creativity and can easily lead to abuse of power
- Cancer genomes are amazing
- Preprints mitigate science publishing problems and delays

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Pan-cancer analysis of whole genomes

[The ICGC/TCGA Pan-Cancer Analysis of Whole Genomes Consortium](#)

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