



Quality Control

Katrina Grasby and Lucia Colodro Conde

Why Do Quality Control?

Poor quality data → false positives / negatives

- To remove genotyping errors
 - Low quality or quantity of DNA
 - Contaminated DNA
 - Chemical or machinery failure
 - Human error
- To ensure data suitable for the analyses
 - Relatedness

From DNA to data



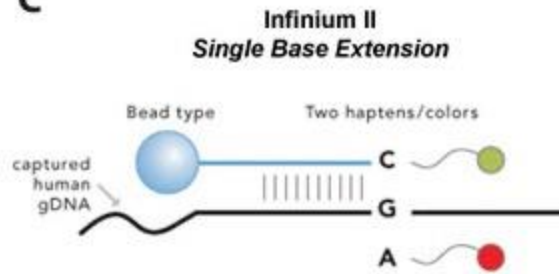
A



B



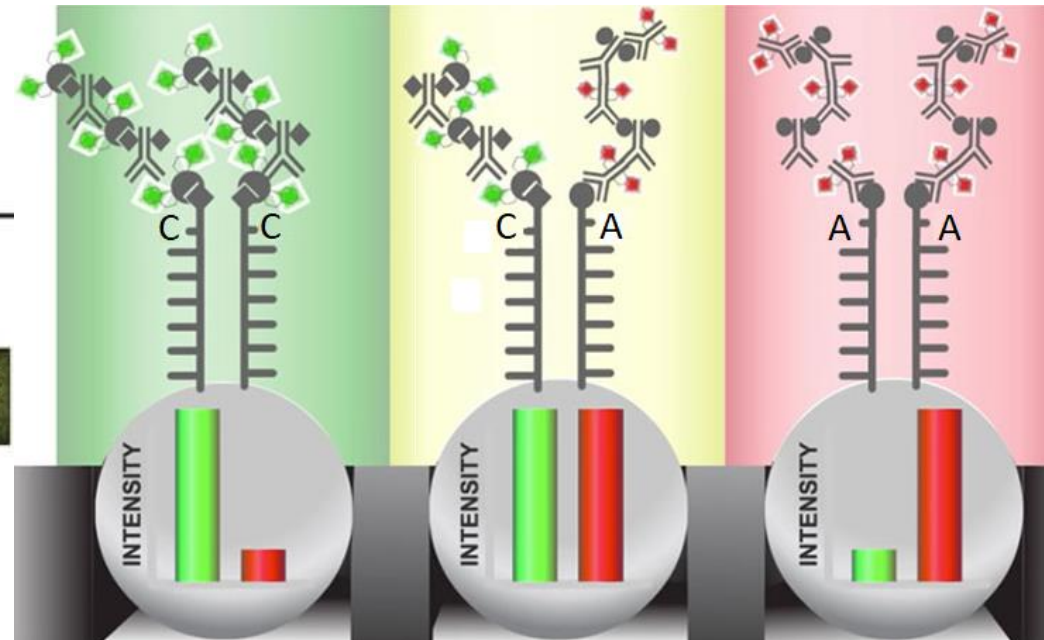
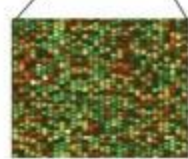
C



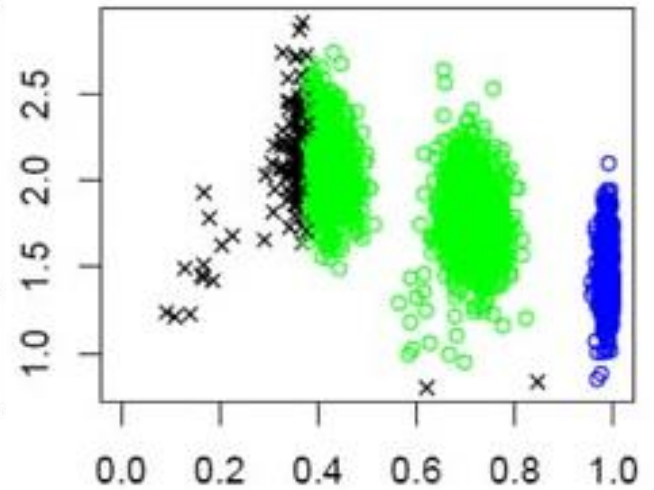
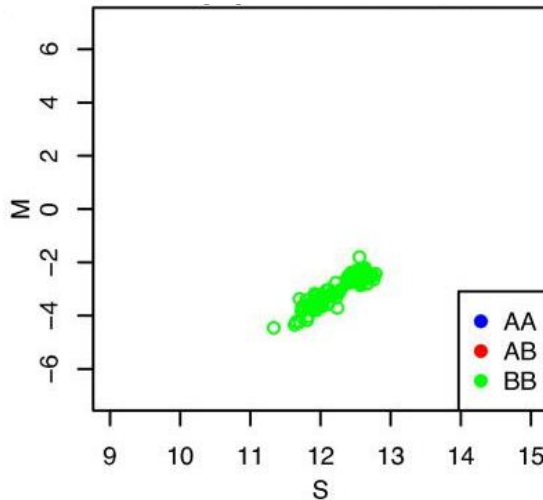
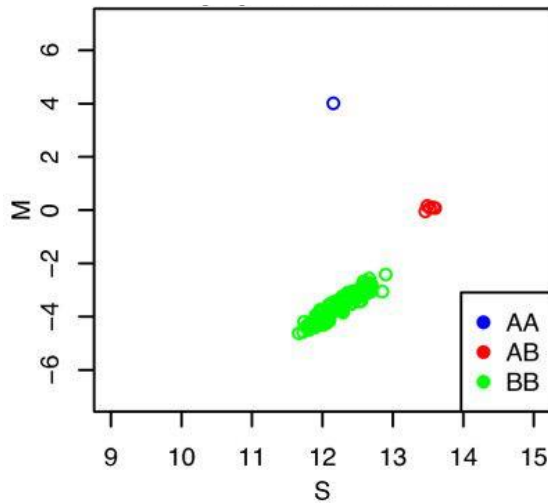
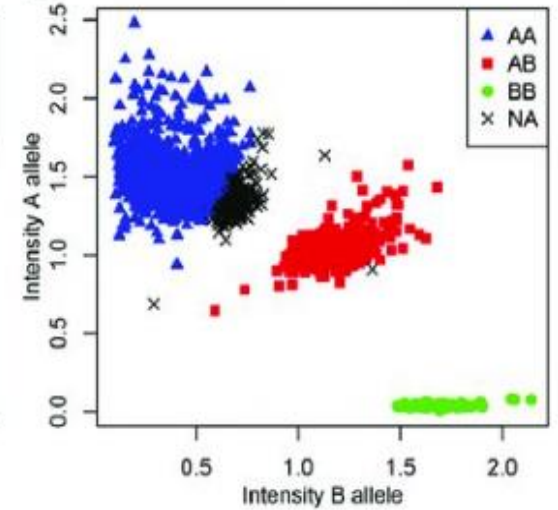
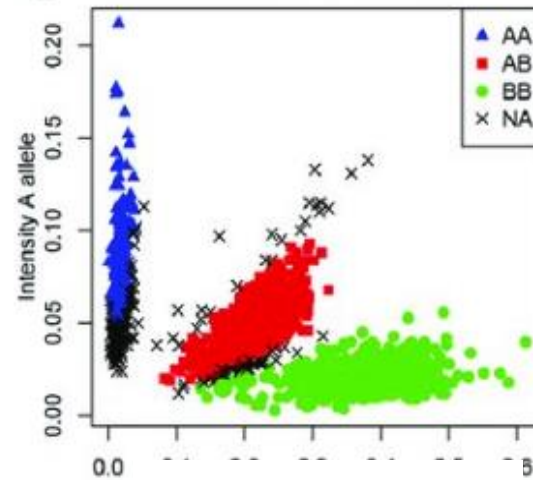
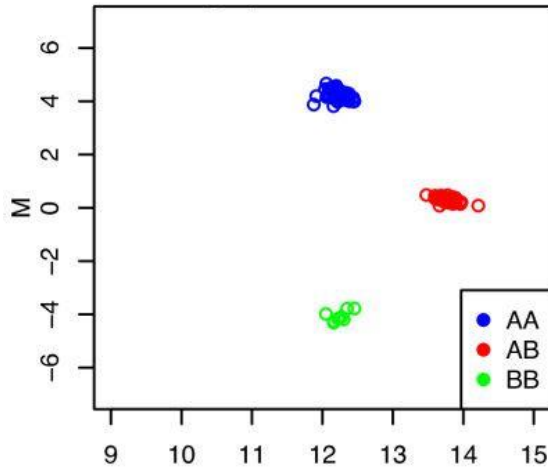
D



E



Genotyping Intensities



Key Steps in QC

1. Check Data (file format, data coding, missing, build)

Check the build

<https://genome.ucsc.edu>

UNIVERSITY OF CALIFORNIA
SANTA CRUZ Genomics
Institute

UCSC

Genome Browser

Home **Genomes** Genome Browser Tools Mirrors Downloads My Data Help About Us

Our tools

- **Genome Browser**
interactively visualize genomic data

Home Genomes **Genome Browser** Tools Mirrors Downloads My Data Help About Us

Browse/Select Species

POPULAR SPECIES

- Human
- Mouse
- Rat
- Fruitfly
- Worm
- Yeast

Find Position

Human Assembly

- ✓ Dec. 2013 (GRCh38/hg38)
- Feb. 2009 (GRCh37/hg19)
- Mar. 2006 (NCBI36/hg18)
- May 2004 (NCBI35/hg17)

GO

Key Steps in QC

1. Check Data (file format, data coding, missing, build)
2. Sex (chr X heterozygosity)

Key Steps in QC

1. Check Data (file format, data coding, missing, build)
2. Sex (chr X heterozygosity)
3. Genotyping Call Rate (SNPs missing individuals)

Key Steps in QC

1. Check Data (file format, data coding, missing, build)
2. Sex (chr X heterozygosity)
3. Genotyping Call Rate (SNPs missing individuals)
4. Hardy-Weinberg Equilibrium

Key Steps in QC

1. Check Data (file format, data coding, missing, build)
2. Sex (chr X heterozygosity)
3. Genotyping Call Rate (SNPs missing individuals)
4. Hardy-Weinberg Equilibrium
5. **Minor Allele Frequency**

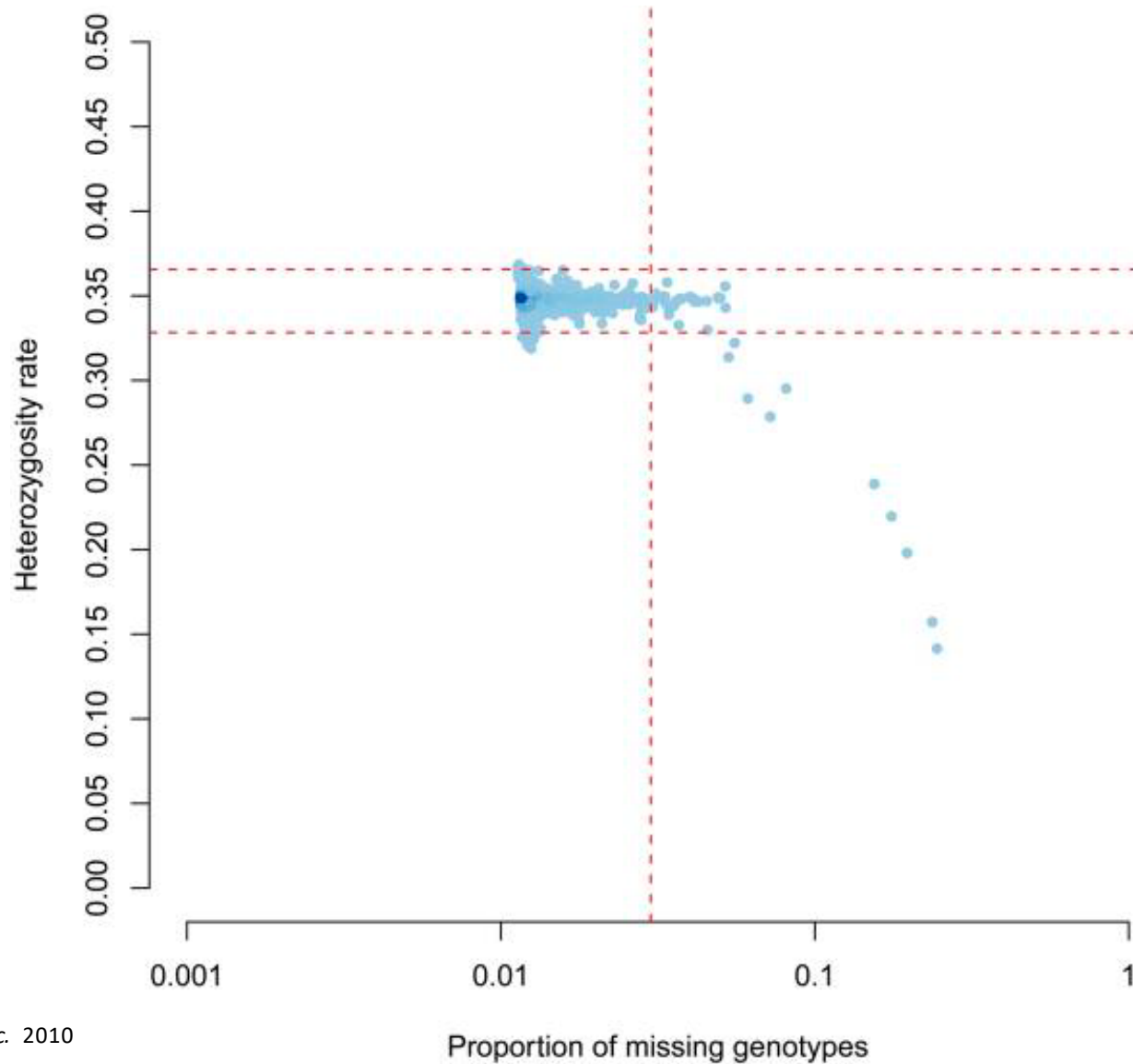
Key Steps in QC

1. Check Data (file format, data coding, missing, build)
2. Sex (chr X heterozygosity)
3. Genotyping Call Rate (SNPs missing individuals)
4. Hardy-Weinberg Equilibrium
5. Minor Allele Frequency
6. **Sample Call Rate (individuals missing genotypes)**

Key Steps in QC

1. Check Data (file format, data coding, missing, build)
2. Sex (chr X heterozygosity)
3. Genotyping Call Rate (SNPs missing individuals)
4. Hardy-Weinberg Equilibrium
5. Minor Allele Frequency
6. Sample Call Rate (individuals missing genotypes)
- 7. Proportion of Heterozygosity**

Heterozygosity vs. Missing



Key Steps in QC

1. Check Data (file format, data coding, missing, build)
2. Sex (chr X heterozygosity)
3. Genotyping Call Rate (SNPs missing individuals)
4. Hardy-Weinberg Equilibrium
5. Minor Allele Frequency
6. Sample Call Rate (individuals missing genotypes)
7. Proportion of Heterozygosity
- 8. Relatedness**

Key Steps in QC

1. Check Data (file format, data coding, missing, build)
2. Sex-check (chr X heterozygosity)
3. Genotyping Call Rate (SNPs missing individuals)
4. Hardy-Weinberg Equilibrium
5. Minor Allele Frequency
6. Sample Call Rate (individuals missing genotypes)
7. Proportion of Heterozygosity
8. Relatedness
- 9. Population Structure / Stratification**

Key Steps in QC

1. Check Data (file format, data coding, missing, build)
2. Sex-check (chr X heterozygosity)
3. Genotyping Call Rate (SNPs missing individuals)
4. Hardy-Weinberg Equilibrium
5. Minor Allele Frequency
6. Sample Call Rate (individuals missing genotypes)
7. Proportion of Heterozygosity
8. Relatedness
9. Population Structure / Stratification