



NORMENT

Norwegian Centre for
Mental Disorders Research

From DNA to genotypes on TSD

Norment & FHI GWAS seminar

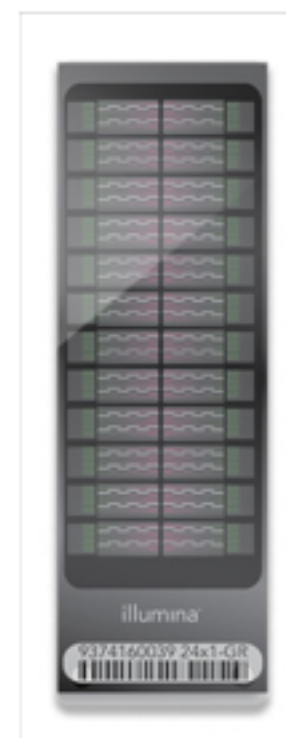
Lavinia Athanasiu, Dec 11th, 2018

Outline

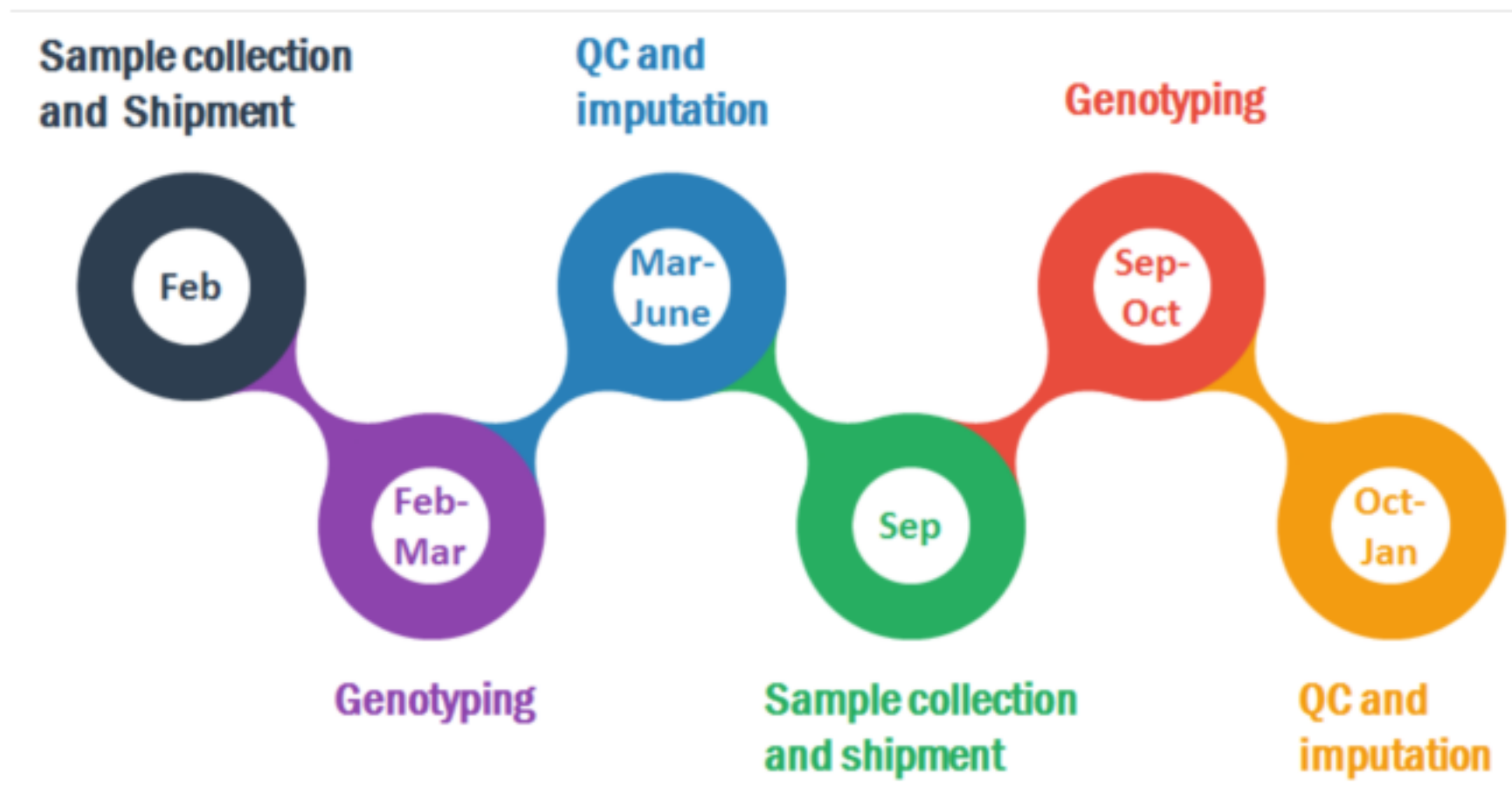
- Logistics genotyping
- DNA
- Genotyping and workflow
- Clustering & QC in Genome Studio
- Export from Genome Studio
- Import TSD
- Overview samples

Genotyping

- Performed at deCODE Genetics, Reykjavik, Iceland
- Illumina platform
 - Infinium OmniExpress chip (- 2017)
 - 24 samples per chip
 - 710.000 fixed markers (up to 30.000 custom)
 - Includes SNPs, CNVs, germline variant, structural variants
- Infinium Global Screening Array (2017 -)
 - 24 samples per chip
 - 640.000 fixed markers (up to 50.000 custom)
 - Combines multi-ethnic genome-wide content
 - Curated clinical research variants
 - Quality control markers for precision medicine research.



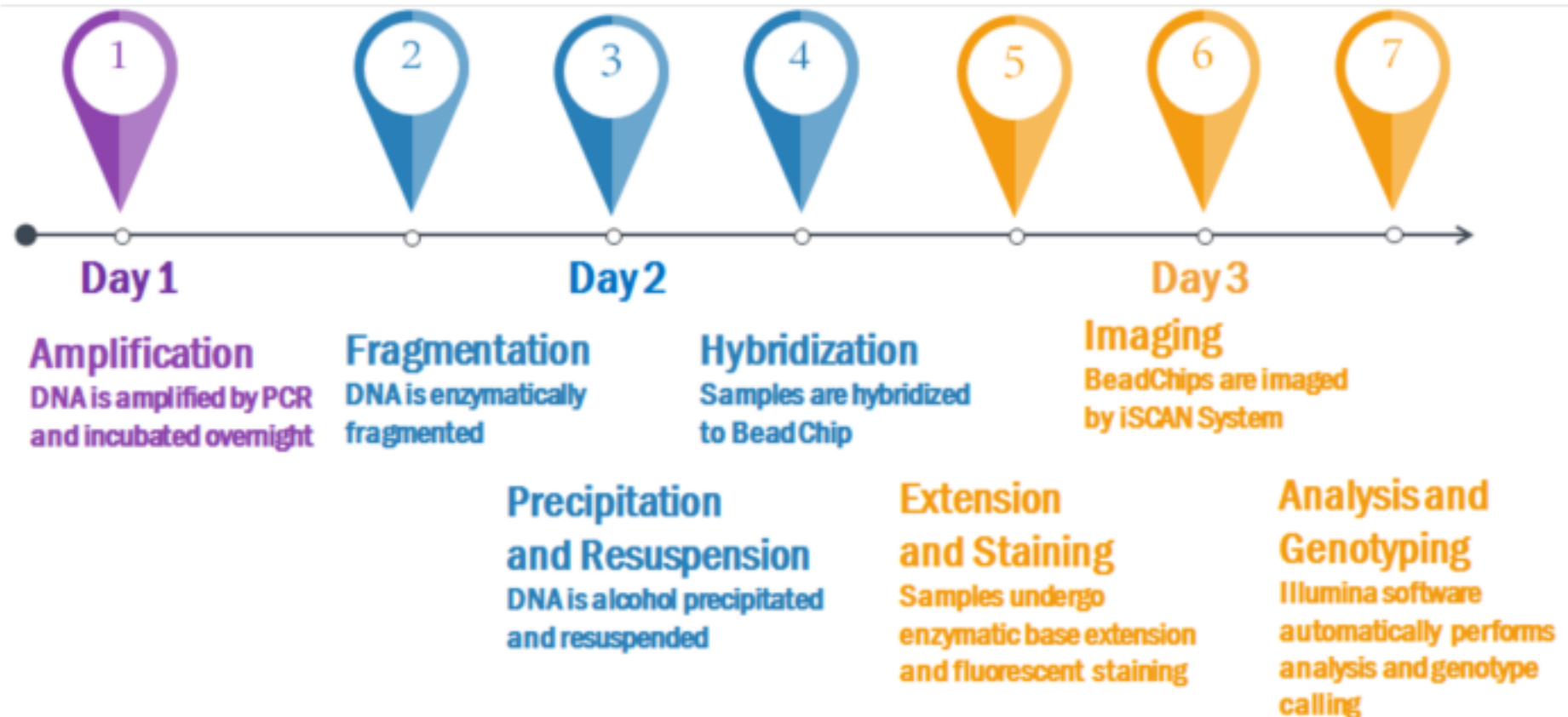
Logistics genotyping



DNA

- Stored in the biobank at Norwegian Institute of public health
 - Directly shipped to deCODE
- Collaborators: ship DNA to NORMENT from other biobanks
- Requirements
 - 200 ng
 - Recommended DNA sample: 20 μ l of 50 ng/ μ l
 - Overview file with ID, age, gender, phenotype
- Shipment on dry ice
 - World Courier

Genotyping workflow

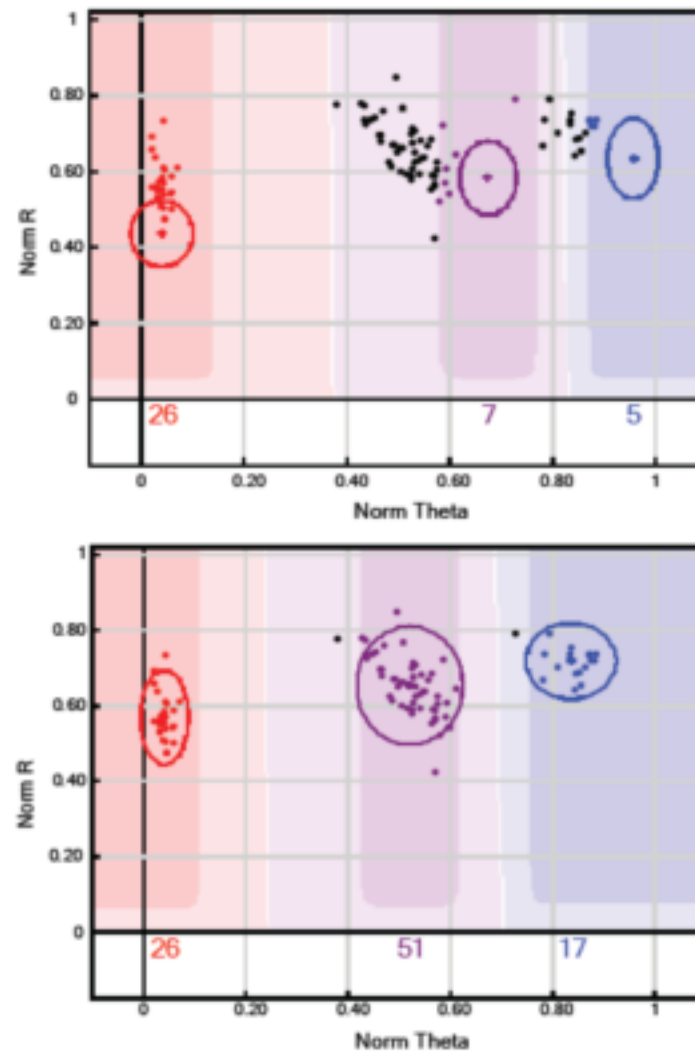


Clustering & QC in Genome Studio

- Genome Studio automatic clustering algorithms
 - Good for 99% of SNPs
 - Other 1 % needs to be manually reviewed



Figure 3: Example of a Successfully Reclustered SNP



QC measures in Genome Studio

- Identifying poorly performing samples
- QC of haploid SNPs
- QC of autosomal SNPs
- Final quality thresholds
- Exporting data (plink format)

References:

Guo, Yan, Jing He, Shilin Zhao, Hui Wu, Xue Zhong, Quanhui Sheng, David C. Samuels, Yu Shyr, and Jirong Long. "Illumina human exome genotyping array clustering and quality control." *nature protocols* 9, no. 11 (2014): 2643-2662.

Illumina (2016). Infinium Genotyping Data Analysis. [Technical Data Sheet]. Retrieved 18 January, 2016 from http://www.illumina.com/Documents/products/technotes/technote_infinium_genotyping_data_analysis.pdf

Overview samples

- Norment wiki with link:

https://docs.google.com/spreadsheets/d/1DAc4nXQmLNjB4yedTZC0hB_EgtMW90n9MM8K9IY7cng/edit?usp=sharing

- Genotypes
 - stored on TSD project p33
 - /tsd/p33/data/durable/vault/genetics/rawdata/genotypes/
- Database on TSD project p33
 - Clinical variables, overview QC measures, phenotypes etc.



NORMENT

Norwegian Centre for
Mental Disorders Research

Thank you!

Oslo University Hospital HF
Division of Mental Health and Addiction
Psychosis Research Unit/TOP
Ullevål Hospital, building 49
P.O. Box 4956 Nydalen
N-0424 Oslo
Norway