



# Molecular Phenotyping and Genotyping Core (MPGC)

## Joslin Diabetes Center

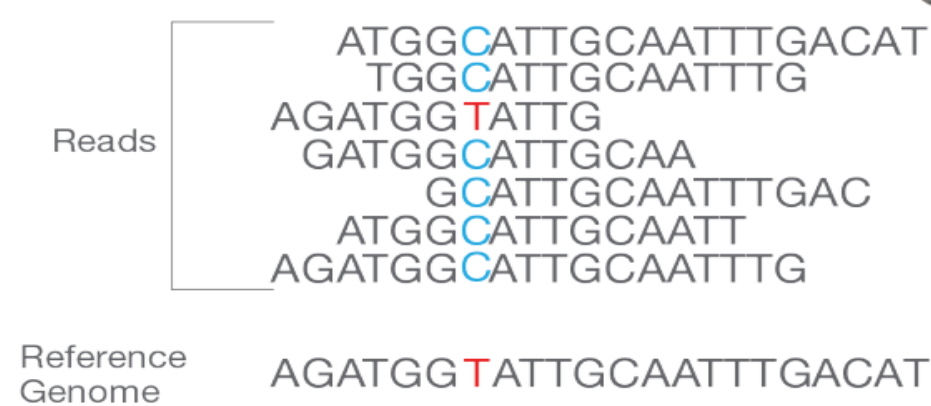
Co-Directors: Alessandro Doria, Mary-Elizabeth Patti



### Diabetes & Complications Genetic Characterization Array

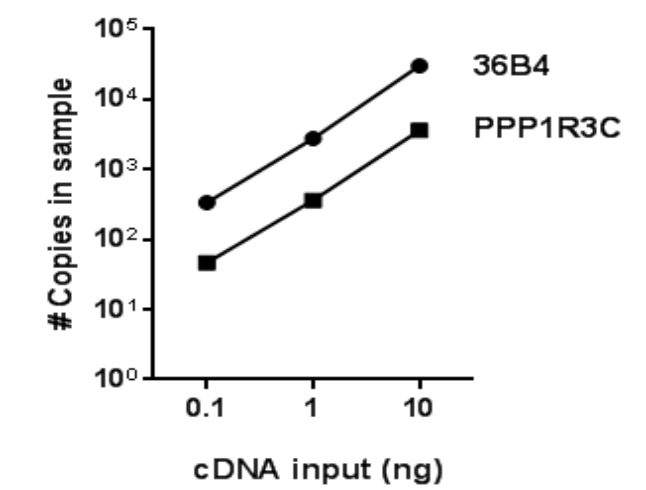
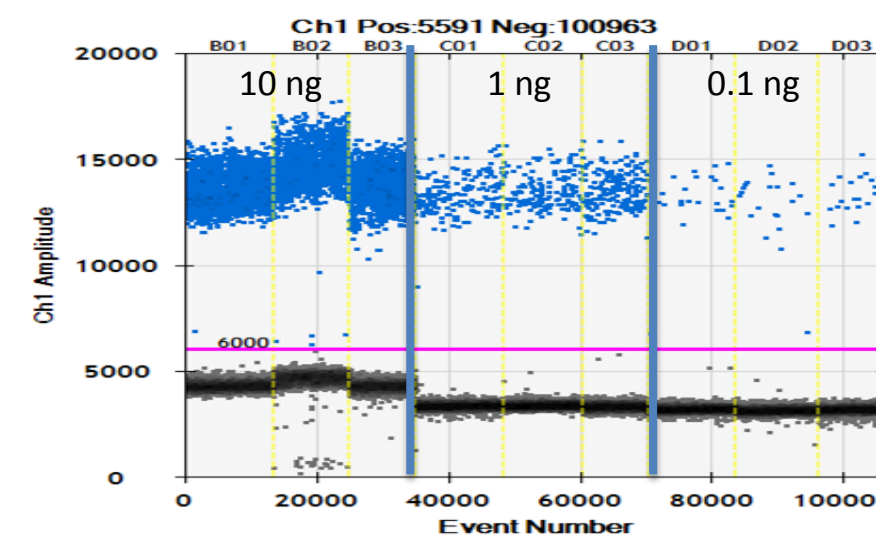
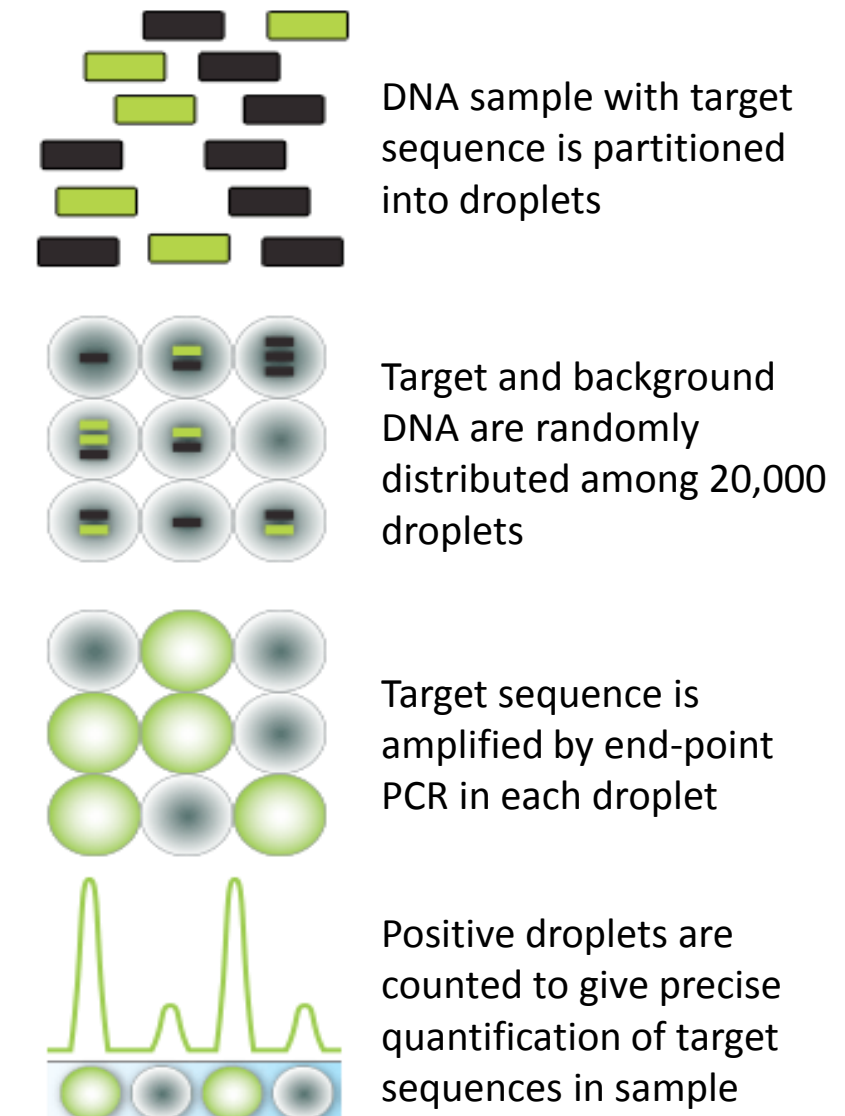
- Utilizing NGS technology to characterize genes and SNPs associated with diabetes and/or complications
- Gene Report
  - Identification of all mutations found in coding regions of 26 genes related to monogenic forms of DM
- SNP Report
  - Genotypes called at each of 571 loci associated with non-HLA T1DM, T2DM, and/or complications of DM

Array SNP Overview	
Phenotype	Number of SNPs
T1DM	85
T2DM	130
T1DM/T2DM	1
Cardiovascular	41
Nephropathy	40
Retinopathy	14
AIMs	260



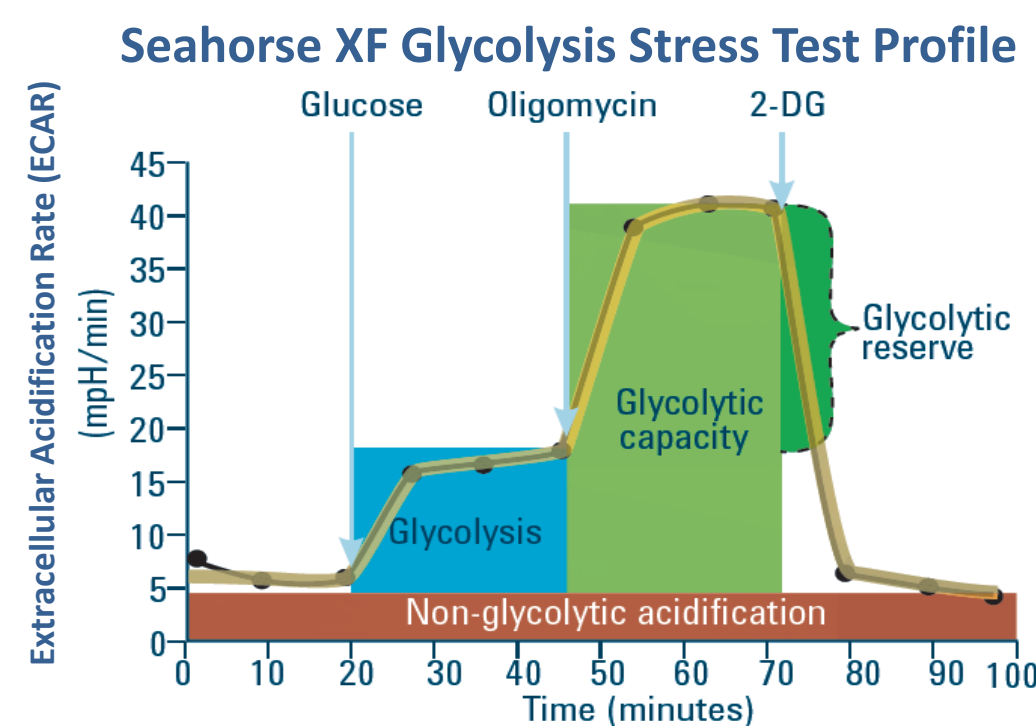
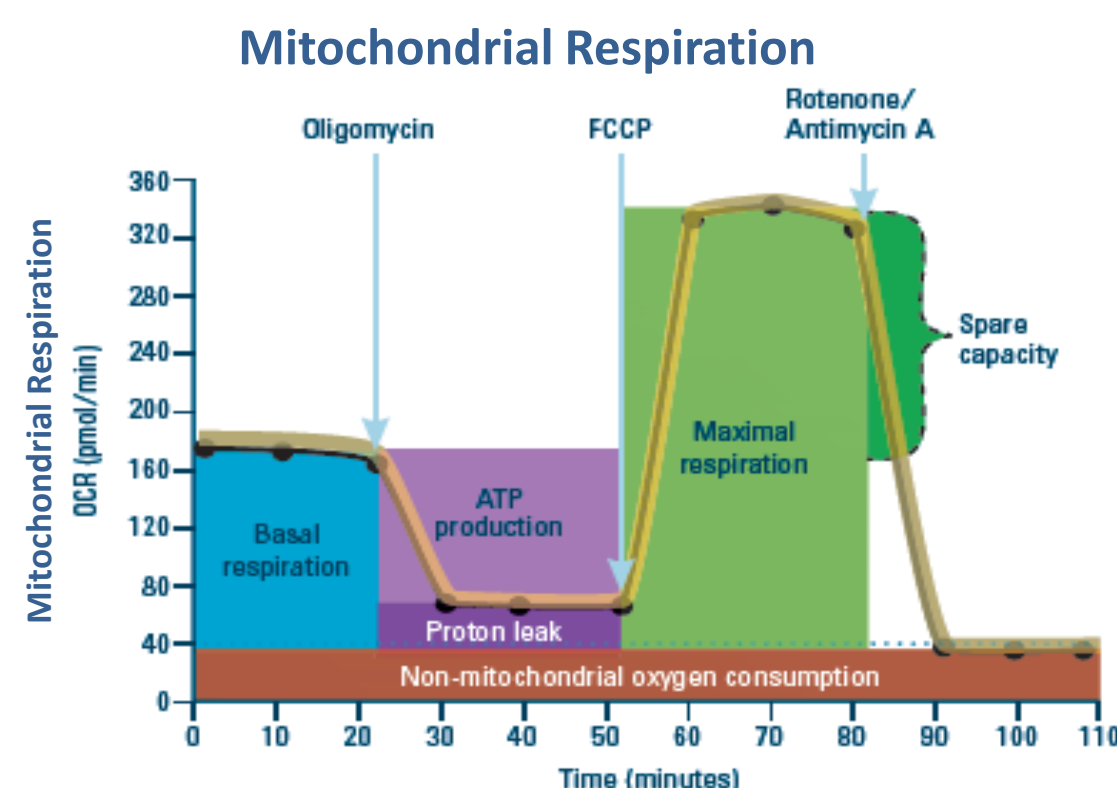
### Bio-Rad QX100 Droplet Digital PCR System

- Bio-Rad QX100 Droplet Digital PCR System
  - Absolute quantification
  - No need for standard curves
  - High sensitivity
  - 5 orders of magnitude
  - Library quantification



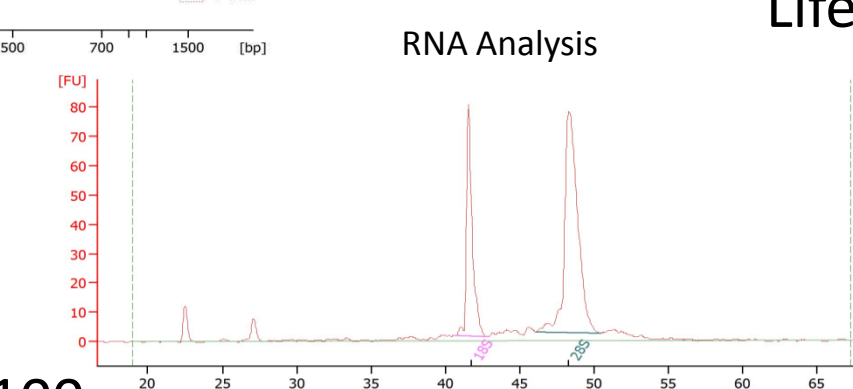
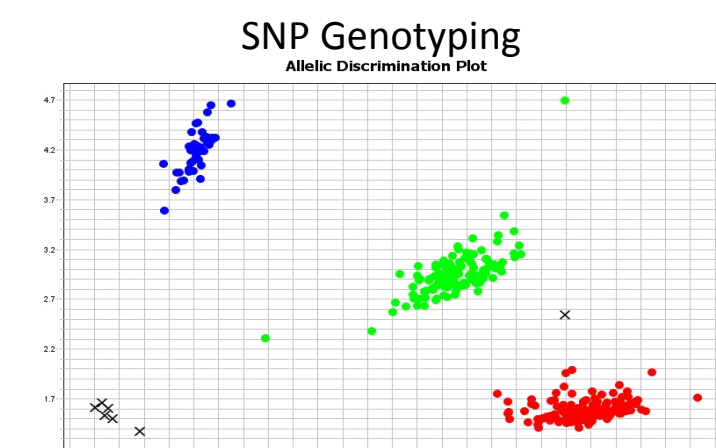
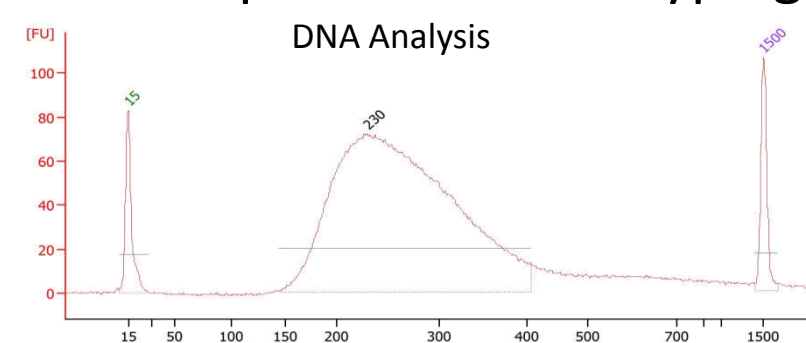
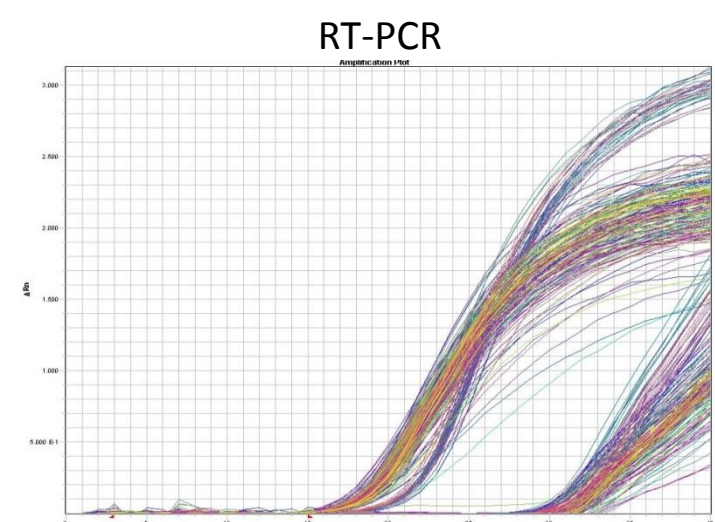
### Seahorse XF24/XF96

- Quantification of cultured cell and tissue explant metabolism through measurement of extracellular acidification and oxygen consumption, assessment of metabolic flexibility
  - Seahorse XF24
    - 24 well format
  - Seahorse XF96
    - 96 well format



### Other Services

- Bioanalyzer Quantification
  - DNA analysis (DNA 1000, HS)
  - RNA analysis (RNA Nano)
- DNA Extraction
- DNA Repository
- DNA Shearing
  - Covaris E220
- PCR Expression Analysis
  - ABI 7900, 7900HT, QS6
- NGS library prep
- Taqman SNP Genotyping



Life Technologies Quantstudio 6

Agilent Bioanalyzer 2100