Novogene

Comprehensive View of Human Whole Genome Sequencing

Novogene is highly experienced in the applications of Whole Genome Sequencing (WGS) for characterising variants. We provide researchers with high quality data in a highly cost-effective manner. Bioinformatic analysis includes, but is not limited to, detection of single nucleotide polymorphisms (SNPs), Insertion/deletion mutants (InDels), structural variants (SVs) and copy number variants (CNVs), with high accuracy and verification rates. Applications of our human WGS service range from disease research to population evolution studies.

Why Novogene?

Extensive experience with over 3000 projects

Industry-leading data quality guarantee

In house pipeline to meet different analysis requirement

Find out more: novogene.com

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Sample requirements

Sample Type	Amount	Volume	Concentration
Genomic DNA	≥ 0.2 μg	≥ 20 μL	≥ 10 ng/μL
Genomic DNA (PCR-free library)	≥ 1.5 μg	≥ 20 μL	≥ 20 ng/μL
FFPE DNA	≥ 0.8 μg	-	-
HMW DNA	≥ 15 μg	≥ 50 μL	≥ 80 ng/μL

Sequencing parameters

Sample Type	Sequencing Platform	Sequencing strategy	Purity	Data Quality
Genomic DNA	Illumina NovoSeq 6000	Pair-end 150	OD260/280 = 1.8-2.0, no genomic DNA degradation, no contamination	Q30 ≥ 85%
Genomic DNA (PCR-free library)				
FFPE DNA			Fragments should be longer than 1500 bp.	
HMW DNA	≥ 15 μg	≥ 50 μL	≥ 80 ng/μL	

Publications using Novogene's expertise

bioRxiv, 2020.
Identification of Avramr1 from Phytophthora infestans using long read and cDNA pathogen-enrichment sequencing (PenSeq)

Animals, 2019.
Age-Dependent Expression of MyHC Isoforms and Lipid Metabolism-Related Genes in the Longissimus Dorsi Muscle of Wild and Domestic Pigs

Applied and Environmental Microbiology, 2019.
l-Rhamnose Metabolism in Clostridium beijerinckii Strain DSM 6423

Journal of Oral Pathology & Medicine, 2018.
Copy number variation: A prognostic marker for young patients with squamous cell carcinoma of the oral tongue

Scientific Reports, 2018.
Csde1 binds transcripts involved in protein homeostasis and controls their expression in an erythroid cell line

PLOS ONE, 2018.
Strap associates with Csde1 and affects expression of select Csde1-bound transcripts

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Case study

An integrated personal and population-based Egyptian genome reference.

Wohlers et al., 2020. Nature Communications. DOI: 10.1038/s41467-020-17964-1.

Research objective:
The de novo assembly of an Egyptian reference genome to assist in the use of precision medicine and disease risk assessment in a non-European population.

Sample collection:
10 healthy individuals that identified as Egyptian up to third generation, recruited when they accompanied patients to Mansoura University Hospital. Medical histories were documented, and all individuals underwent a medical examination and routine laboratory testing.

Sequencing strategy:
A combination of PacBio, 10X Genomics and Illumina pair-end sequencing data with an overall genome coverage of 270x.

Data amount:
808Gb

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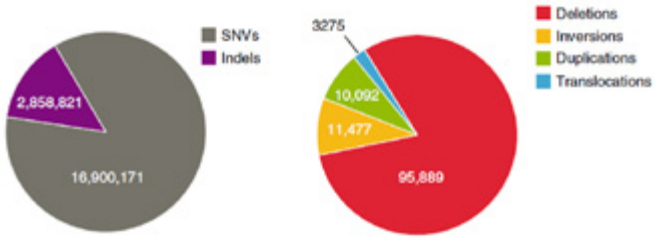

Figure 1
The number of variant types identified in the Egyptian cohort.

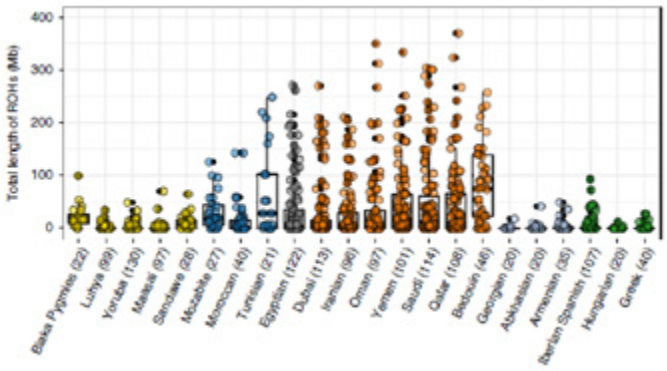

Figure 2
Box plots of ROH for Egyptians and several populations from relevant world regions

Results

A high number of variants (fig. 1) were found in the Egyptian cohort when compared against existing databases that are primarily based on European individuals. 6,599,037 SNVs were identified that were either extremely rare or not detected in any other population.

Egyptians were found to have Middle Eastern, European/Eurasian, North African and East African influence, in descending order. mtDNA analysis showed haplogroups present in Egyptians most often found in Europeans. These findings overall support the idea that Egypt's transcontinental geographic location shaped Egyptians' genetics.

Runs of homozygosity (ROH) were analysed and compared with other populations. Egyptian ROH lengths were most comparable to those of Middle Eastern individuals. Long ROH abundance is typical in the Middle East, due to the common practice of consanguineous marriage.

Conclusions

This study showed how genetic variations in Egyptians can compromise the transferability of disease risk and polygenic scores from European-based genomes. This substantiates the need for a multi-ethnic genome database which will be a valuable future resource for precision medicine.

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