family support and lastly to heed the advice of medical professionals. However, concerns with regards to cost and the perceived lack of ability to cope with the genetic results deterred participants from taking up the test. Additionally, the lack of information about genetic tests was cited by participants as a factor that hindered them from making a decision of whether to take the test.

S. Sun: None.

E-P15.06

Comparison of seven contemporary pharmacogenetic assays with the PharmGKB database

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Genotyping of pharmacogenetic (PGx) genes can provide informed personalized drug therapy and can prevent common adverse drug reactions. Today, there are several tests commercially available to genotype individual Single Nucleotide Polymorphisms (SNPs) in PGx genes. In this study seven modern SNP genotyping assays were compared with the Pharmacogenomics Knowledge Base (PharmGKB) database to determine what proportion of the currently known pharmacogenetic effects is measured by these assays. These seven assays are the "Ion AmpliSeq™ Pharmacogenomics Panel", the "VeraCode® ADME Core Panel", the "iPLEX® PGx Pro Panel", "DMET™ Plus", "PharmcoScan™", "Living DNA", and "23andMe". The PharmGKB database contains 3474 clinical annotations, which describe a variant-drug interaction. 94% of PharmGKB's clinical annotations can be determined with SNP assays. The other part can be determined with haplotypes and copy number variations. Of PharmGKB's clinical annotations, 76%, 68%, and 44% can be determined by PharmacoScan™, Living DNA, and 23andMe respectively. The other described assays, are designed to test only specific subset of PGx SNPs.

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E-P16 Omics/Bioinformatics

E-P16.01

Alu elements as source of microRNA sites in the human genome

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Alu sequences are the most abundant short interspersed repeated elements in the human genome. These sequences are the most abundant mobile elements in the human genome, representing 10.6% of nuclear DNA. The aim of the study was to carry out a full-genomic bioinformational search for microRNA motifs localized in human Alu sequences. Genomic sequences were extracted from the NCBI database using the E-utilities suite of utilities. The micro-RNA sequences were taken from the mirRBase database, release 21. The search was performed using the miRanda and GLAM2Scan software suite. About 80-90% of the miR-619 miR-619, miR-5585, miR-5095, and miR-5096 microRNA binding sites were detected within the Alu elements. The presence of binding sites for certain microRNAs within Alu elements also explains the high conservatism of the distances between sites. The sequences miR-1273g and miR-1285-1,2 were found inside the Alu elements, but lying on the opposite chain. The pre-Mir-1273g sequences were detected within the Alu-elements, which makes the latter a prospective source of the mature miR-1273g. miR-5196 and miR-466 were not found inside the Alu elements, but they are also widely represented in the human genome. The study was supported by the Ministry of education and science of the RF, project № 6.6762.2017.

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E-P16.02

Discovering potential causative mutations in human coding and noncoding genome with the interactive software BasePlayer

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Introduction: Next-generation sequencing (NGS) is routinely applied in life sciences and clinical practice, where interpretation of the resulting massive genomic data has become a critical challenge. The genome-wide mutation analyses enabled by NGS have had a revolutionary impact in revealing the predisposing and driving DNA alterations behind a multitude of disorders. The workflow to identify causative mutations commonly involves phases such as quality filtering, case-control comparison, genome annotation and visual validation, which require multiple