sequence alignments generated by a Smith-Waterman-based program, SSEACH showed that the average sequence identity was 75%, we found that the mutations at a particular position could be called by the accuracy of 90%, when all the reads covering the corresponding positions were collectively evaluated. We provide the first simple experimental and analytical MinION sequencing procedure, which can be easily followed on site to effectively genotype pathogens of other tropical diseases.

Keywords: MinION; sequencing analysis; sequencers; tropical diseases


AB006. Personalized and precision medicine: are we there yet?

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Abstract: Personalized medicine is determined by an individual's unique clinical, genomic and environmental information. The molecular understanding of diseases allowed development of preventive healthcare strategies and medical treatments at the pre-symptomatic or earliest stage of the disease. To achieve this promise, DNA-based risk assessment, molecular profiling, targeted therapies and dose selection of therapeutic agents were developed to facilitate customization of patient care. Commercially available genomic tests routinely are applied across a wide range of disease states in predictive or prognostic applications. Many clinicians were concerned about the lack of progress in the clinical application of genomic medicine. The development of genomic diagnostic tools such as array comparative genomic hybridization, exome and whole genome sequencing had a vital role to play in the delineation of new Mendelian loci for previously unrecognized syndromes or identification of additional genes or loci contributing to known disease entities. While the costs of these tests had decreased, the interpretation of information of uncertain significance may require increased 'genomic counseling' consultations to allay anxiety. Personalized medicine at present has limited roles in complex disorders or used as a tool lifestyle change decisions as public health or primary care professionals who may not be sufficiently 'genomic-trained'. Genomic health risk assessments and statistical probabilities are difficult for clients to understand and personalized medicine must be integrated into the existing health systems. In addition, clinical workflow with significant changes required in regulatory and reimbursement policies as well as legislative protection related to patient's confidentiality will be required. The difficulties with use of genome-wide association studies in clinical practice, with its limited phenotype-genotype impact must be addressed. Personalized medicine is expected to revolutionize traditional clinical practice; in reality, it will evolve to deliver on the promise of a safer and effective healthcare for the individual patient.

Keywords: Personalized medicine; genomics; genetic counseling; public health

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AB007. Genomic medicine: impact of rare disease research on medicine and health care

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Abstract: Most of rare diseases are genetic and the number of identified pathogenic genes is rapidly increasing since the introduction of next-generation sequencers. To date more than 4,500 Mendelian rare disorders with known molecular basis are listed in the OMIM database. In the past, it was believed by the majority of medical community that research on rare diseases scarcely contributes to medical