J Clin Exp Pathol 2015, 5:3
http://dx.doi.org/10.4172/2161-0681.S1.017

OMICS International
conferenceseries.com

4th International Conference and Exhibition on
Pathology
July 13-15, 2015 New Orleans, USA

Next generation sequencing: A perspective of a clinical diagnostic scientist

Shanxiang Shen
Moffitt Cancer Center, USA

Next generation sequencing (NGS) is a powerful technology and can be used in areas of infectious diseases, genetic disorders, cancer and pharmacogenomics of clinical diagnostics. Due to the complexity of the technology and various scenarios of clinical diagnostics, test development requires special expertise. No matter what platform is used, depth of coverage of each sequencing fragment is the most important information for determining the sequencing quality, calculate variant frequency, estimate copy number variant and predict other structural variant. It is also an important quality control indicator. What to choose from sequencing techniques such as whole genome, whole exome and targeted panel is determined by the application. Bioinformatics plays a more important role in NGS tests than in other clinical tests which involves hardware and software need for storage, procession and communication of test data. NGS tests need to be validated and verified to comply with regulations. The tests need to be validated that meet the requirement of clinical diagnosis of pertained disease or disorder as well be verified that it will provide liable result. Validation and verification not only bench top procedures but also data analysis pipeline. Quality control measurement and material are also need to be established before start service and QC conducted periodically during service. Cost of routine clinical diagnostic operation need to be estimated carefully which should include costs of bioinformatics and data storage. Finally, NGS is powerful but not for everything.

Shanxiang.Shen@moffitt.org

Mutational landscape of aggressive cutaneous squamous cell carcinoma

Jane H Zhou
Tufts Medical Center, USA

Aggressive cutaneous squamous cell carcinoma (cSCC) occurs frequently in the head and neck region resulting in disfiguration and even death. Furthermore, the incidence of cutaneous squamous cell carcinoma is increasing. At present time, little is known about the mutations that drive aggressive cSCC. We performed whole-exome sequencing on 39 cases of aggressive cSCC to identify driver mutations and potential therapeutic targets. Besides the well-known cancer-associated genes TP53, CDKN2A, NOTCH1, AJUBA, HRAS, CASP8, FAT1, KMT2C (MLL3), we identified 3 novel candidate tumor suppressors, NOTCH2, PARD3 and RASA1 of which germ line mutations are known to cause diseases. In addition, we found that KMT2C mutations were associated with poor outcome and increased bone invasion. These findings improve our understanding of aggressive cutaneous squamous cell carcinoma and guide the direction of further study of this disease.

jhzhou211@gmail.com

Notes: