Case Presentation: Here we present the case of a 51-year-old female with AML with myelodysplastic-related morphologic changes, 13q deletion and t(8;11), where initial fluorescence in situ hybridization (FISH) assays were consistent with the presence of NUP98 and FGFR1 rearrangements, and suggestive of NUP98/FGFR1 fusion. Using a streamlined clinical whole-genome sequencing approach, we resolved the breakpoints of this translocation to intron 4 of NSD3 and intron 12 of NUP98, indicating NUP98/NSD3 rearrangement as the underlying aberration. Furthermore, our approach identified small variants in WT1 and STAG2, as well as an interstitial deletion on the short arm of chromosome 12, which were cryptic in G-banded chromosomes.

Conclusions: NUP98 fusions in acute leukemia are predictive of poor prognosis. The associated fusion partner and the presence of co-occurring mutations, such as WT1, further refine this prognosis with potential clinical implications. Using a clinical whole-genome sequencing analysis, we resolved t(8;11) breakpoints to NSD3 and NUP98, ruling out the involvement of FGFR1 suggested by FISH while also identifying multiple chromosomal and sequence level aberrations.


106. Genotype of Human Papilloma virus in Male Genital Warts in Korean men and review of literature
Jung Joo Moon a, Woo Chul Moon b

a Cellgenemedix, Newark, NJ, USA; b Good Gene Korea, Seoul, South Korea

Purpose: Genital warts are one of the most common sexually transmitted infections and are known to develop due to human papilloma virus (HPV) infection, especially HPV types 6 and 11. However, their prevalence in male genital warts remains poorly defined. HPV vaccine is administered to both sexes and it is important to investigate their expected impact in male anogenital warts.

Methods: We have herein conducted a multi-center, prospective study to analyze HPV type distribution in genital warts of 1000 Korean men by using DNA microarray that can detect 40 types of genital HPV.

Results: 1000 out of 1015 genital warts showed HPV DNA. Out of 1000 HPV-positive samples, 18.8% showed mixed infections and 81.2% showed single infection.

Of the 18 high-risk (16.2%) and 14 low-risk (94.3%; 12 types) HPV types detected, the most common type of HPV type were HPV6 (59.5%), followed by HPV11 (24.3%), HPV16 (6%), HPV91 (5.3%), and HPV40 (3.3%). 94.3% showed low-risk and 16.2% high-risk, and 10.5% both high- and low-risk type HPV types, respectively. 87.2% showed HPV types (HPV6, 11, 16, 18, 31, 33, 45, 52, and 58) covered by the vaccine. Sixteen of the 200 HPV specimens submitted for sequencing showed discrepant results compared to the DNA sequencing.

Conclusions: Male genital warts predominantly show low-risk type HPV (HPV 6 and 11; 82%). However, high-risk HPV is not uncommon. The Gardasil 9 HPV vaccine is expected to provide protection against about >80% of male genital warts. Further HPV typing studies in male genital warts are necessary.


Jung Joo Moon a, Woo Chul Moon b, Sung Woo Moon c

a Cellgenemedix, Newark, NJ, USA; b Good Gene Korea, Seoul, South Korea; c Yonsei University, Seoul, South Korea

Background: We are facing a new era of the COVID-19 pandemic by omicron variant. This raises concern about the accuracy of conventional RTqPCR assays. There is a need for a new RTqPCR assay appropriate for the omicron variant with multiple mutations.

Materials and Methods: We herein have developed 2 types of new RTqPCR assays which detect G142D/de143-145, 452 and 478 of S, 69/70 of S or N, and internal control (IC) to make a new single tube, quadplex RTqPCR assay of COVID-19 (‘GG COVID-19 Omicron and Delta kit’). Analytical performance was analyzed with Twist/BEI controls. Clinical performance analysis was done as a multi-center study in South Korea with specimens from New Jersey, collected between October 2021 and January 2022.

Result: 400 left over nasopharyngeal swabs including 149 SARS-CoV-2 positive samples were collected and tested on January 2022 out of which 148 were due to omicron variant and 1 delta variant, respectively. S142-145 assay was superior to conventional RT-PCR(sensitivity 100% vs 00.0 to 00.0%, mean Ct value 25.0 vs 30.4 to 31.3). 330 samples including 125 SARS-CoV-2 positive samples from October 2021 and also tested, out of which 78 were due to delta and confirmed by sequencing.

Conclusion: GG COVID-19 Omicron and Delta kit showed higher sensitivity than conventional RTqPCR (100% vs 95.4%) and 100% specificity in detecting SARS-CoV-2 and could differentiate all of omicron variants. These results indicate that our GG COVID-19 Omicron and Delta kit may be more sensitive than conventional PCR assays in detecting Omicron and Delta SARS-CoV-2.

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108. A cancer genomics resource built on GA4GH standards
Rahel Paloots, Michael Baudis, Hangjia Zhao, Ziying Yang

University of Zurich, Zurich, Switzerland

Progenetix is a cancer genomics resource that includes genomic profiling data as well as biomedical annotations and provenance data for cancer studies. The main goal of the Progenetix database is to provide easy, open access for research studies and clinical diagnostics. To facilitate sharing of genomic data, Progenetix complies with and contributes to GA4GH and Beacon data standards. Beacon, developed with the support from ELIXIR (the European bioinformatics infrastructure organization), started out as protocol to share genomic variants over federated queries. The current development of Beacon (Beacon v2) enables expanded metadata-rich queries in both public and restricted federated access modes.

The implementation of Beacon v2 API in Progenetix, offers a solution to sharing vast amounts of genomic data securely and effectively. Moreover, it is open-access and well-documented, with an open API for third party use. Currently, Progenetix contains around 130 thousand cancer copy number variant (CNV) profiles from more than 700 different cancer types (NCIt classification), making it the largest resource for cancer CNV profiles. In addition to primary neoplasia samples, Progenetix also includes a set of cancer cell line CNV samples. In order to provide a comprehensive cancer cell line variant database, Progenetix also incorporates known single nucleotide variants of cancer cell lines.

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