is a new and recently developed non-invasive cancer diagnostic technique. This technique includes collection of blood or urine sample and diagnosis of cancer based on analysing molecular bits or cancer cells that are released from tumour tissue in to the blood or urine system. Circulating cell-free DNA (cfDNA) fragments is one those molecular bits that are released into the bloodstream after the rapid apoptosis or necrosis of the tumour cells in the cancer patients.

**Material and methods** Our goal is to do the comprehensive study between distinct types of glioma cancer tumours and cfDNA of the respective patients to elucidate the scope of cfDNA in liquid biopsy technique for glioma diagnosis.

**Results and discussions** We have successfully detected glioma specific mutations such as IDH1 and 2, PDGFRA, NOTCH1, PIK3R1 etc., on the cfDNA isolated from the plasma of glioma patients and could relate this mutations to the different tumour grades of glioma. We are also studying the dynamics of these mutations in response to the glioma drug treatment by collecting blood samples at different time intervals.

**Conclusion** This study may help in developing liquid biopsy technique for glioma tumour diagnosis and in its prognosis for monitoring the glioma treatment by non-invasive approach, and will eventually help physicians to decide the right treatment on right time and will bypass the existing ‘wait-and-see’ approach of treatment monitoring.

**THE ROLE OF SPARCL1 IN UPPER URINARY TRACT CANCER OF TAIWAN**

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**Introduction** Upper urinary tract urothelial carcinoma (UTUC) in Taiwan is a relatively high prevalent cancer and locally advanced UTUC often carries a poor prognosis. This study is to analyse role of SPARCL1 in UTUC of Taiwan and analyse if advanced UTUC often carries a poor prognosis. This study is to analyse role of SPARCL1 in UTUC of Taiwan and analyse if advanced UTUC often carries a poor prognosis. This study is to analyse role of SPARCL1 in UTUC of Taiwan and analyse if advanced UTUC often carries a poor prognosis.

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**Conclusion** This study may help in developing liquid biopsy technique for glioma tumour diagnosis and in its prognosis for monitoring the glioma treatment by non-invasive approach, and will eventually help physicians to decide the right treatment on right time and will bypass the existing ‘wait-and-see’ approach of treatment monitoring.

**GERMLINE DETERMINANTS OF THE SOMATIC MUTATION LANDSCAPE IN 2642 CANCER GENOMES**

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**Introduction** Cancers develop through somatic mutations; however, germline genetic variation contributes to cancer risk via diverse mechanisms including by modulating mutational processes.

**Material and methods** Within the Pan Cancer Analysis of Whole Genomes (PCAWG) project, we discovered and phased 88 million single nucleotide variants, short insertions/deletions, and large structural variants in whole genomes from 2642 cancer patients, and employed this resource to investigate germline determinants of somatic mutation across 39 cancer types.

**Results and discussions** We describe over 100 germline L1 retrotransposons mediating somatic retrotransposition activity in cancer. Furthermore, rare damaging germline mutations in genes involved in DNA repair, DNA replication, and cell cycle associate with a variety of somatic mutation processes. We implicate mutations in the DNA glycosylase MBD4 with an elevated rate of C>T mutations at CpG dinucleotides, resulting in the genetic modulation of a widespread mutational process. Genome-wide association analysis reveals common genetic variation within the APOBEC3 gene cluster modulating mutations attributed to APOBEC cytidine deaminases in multiple cancer types. Analysis of somatic structural variation additionally exposed complex rearrangement patterns including duplications and template insertion cycles in BRCA1-deficient cancers.

**Conclusion** Our study underscores the notable impact rare and common germline variants have on cancer mutational landscapes.

**CHROMATIN ACCESSIBILITY PROFILING IDENTIFIES AN UNDERLYING HNF4A-GATA6 REGULATORY MODULE IN OESOPHAGEAL ADENOCARCINOMA**

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**Poster Presentation: Cancer Genomics, Epigenetics and Genomic Instability**