

Description of Additional Supplementary Files

File Name: Supplementary Data 1

Description: Patient sample characteristics and pathogenic genomic variants detected by standard of care methods along with classification calls by Epimarker

File Name: Supplementary Data 2

Description: Overview of samples in positive and negative cohort

File Name: Supplementary Data 3

Description: Number of genomic variants detected in negative cohort - CNVs (QDNAseq), SVs (Sniffles and cuteSV) and SNVs (Claire3)

File Name: Supplementary Data 4

Description: Overview of samples processed for transcriptome analysis

File Name: Supplementary Data 5

Description: Alignment statistics of samples from transcriptome sequencing

File Name: Supplementary Data 6

Description: Summary statistics of differentially expressed genes

File Name: Supplementary Data 7

Description: Validated pathogenic variants detected in the cohort; assays and the PCR primers used for validation