Whole genome and transcriptome analysis of 200 pediatric liver cancer in the JPLT study

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Despite extensive genomic analysis to date, there are few validated genetic abnormalities in hepatoblastoma. To identify biomarkers useful for patient stratification and novel therapeutic targets, we performed whole genome and transcriptome analysis of 200 pediatric cancers using the JPLT cohort. Consistent with previous results using the JPLT2 cohort, CTNNB1 abnormalities were found in 76% of cases and TERT promoter mutations were found in 5%. The median number of exon mutations was 4, with a tumor mutation load per megabase of 0.01-1.0 (median: 0.15). In addition, we identified new hotspot recurrent mutations in transcriptional regulators. We will present a summary of molecular aberrations that can be implemented in future clinical trials.