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Expression profiling of *TP53, BLM, DIS3L2, GPC3, NSD1, PAX6* and *AMER1* genes in Wilms' tumor cases

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Wilms' tumor has been linked to a number of genes, but WT1 has been reported to be directly linked to the growth of this embryonic tumor. Due to mutations in additional genes occurring in conjunction with WT1, Wilms' tumor is linked to a variety of disorders that manifest as syndromic conditions, e.g., Li-Fraumeni syndrome, Bloom syndrome, Perlman syndrome, Simpson-Golabi-Behemel syndrome, Sotos syndrome, WAGR syndrome and X-chromosome syndrome, which are linked to oncogenes (OGs) and tumor suppressor genes (TGs), i.e., *TP53, BLM, DIS3L2, GPC3, NSD1, PAX6*, and *AMER1*, respectively. The study demonstrated the mRNA expression levels of the *TP53, BLM, DIS3L2, GPC3, NSD1, PAX6* and *AMER1* genes by code-set chemistry in 24 Wilms' tumor cases in comparison to their internal controls (adjacent to tumor tissues). Capture-and-reporter probe-based expression was carried out using NanoString technology. All the genes of interest were found to be significantly up- and downregulated according to the fold change expression study results. The mRNA expression of TP53 in 95.84%, BLM in 83.34%, DIS32 in 62.50%, NSD1 in 62.50%, AMER1 in 58.33%, GPC3 and PAX6 in 50% of Wilms' tumor cases were significantly upregulated. Hence, this study established that the *NSD1, DIS3L2, AMER1, TP53, BLM, PAX6*, and *GPC3* genes play roles in the development of these embryonic tumors and can be used as biological markers for Wilms' tumors. However, a larger sample size is needed to validate the above data.

Keywords: Wilms' tumor (WT), Li-Fraumeni syndrome, Bloom syndrome, Perlman syndrome, Simpson-Golabi-Behemel syndrome, Sotos syndrome, WAGR syndrome