iScience, Volume *25*

Supplemental information

The genomic landscape of pediatric renal cell carcinomas

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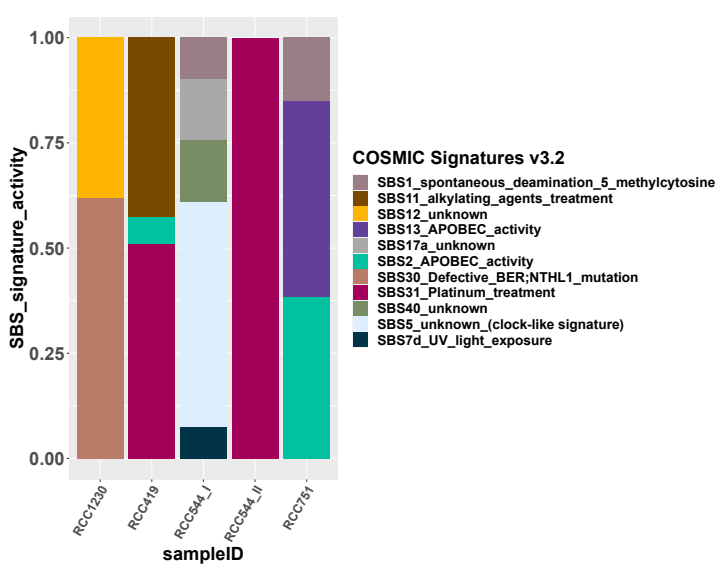
Supplemental Information

Supplementary Figures and Legends

A



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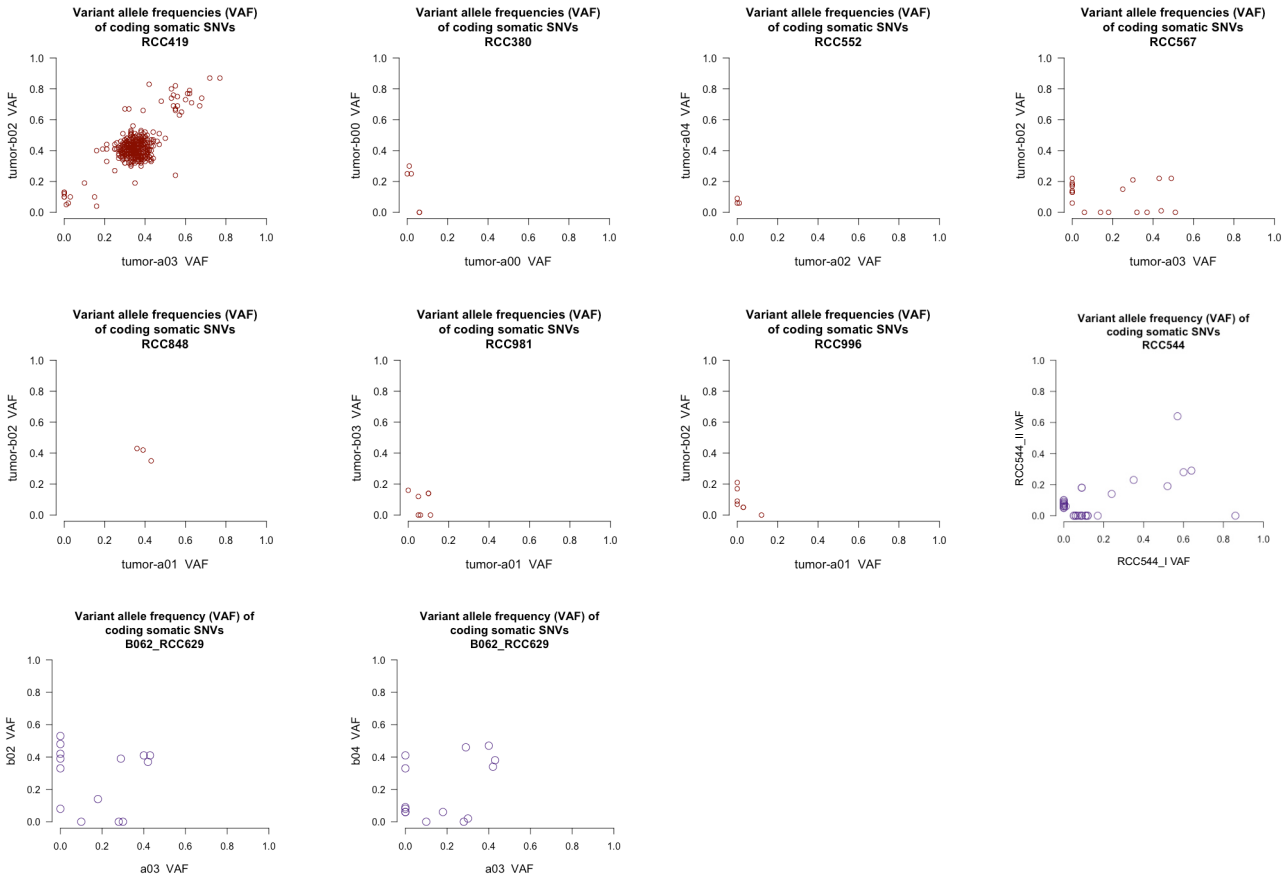
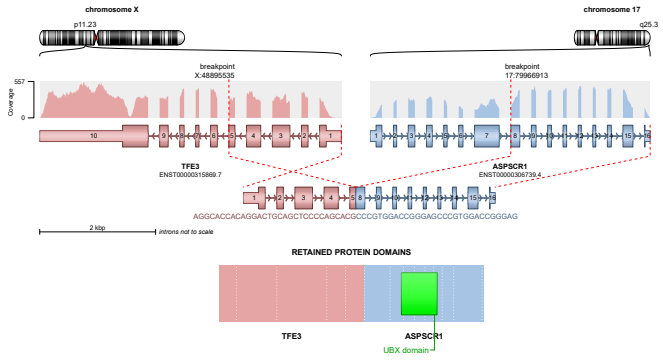
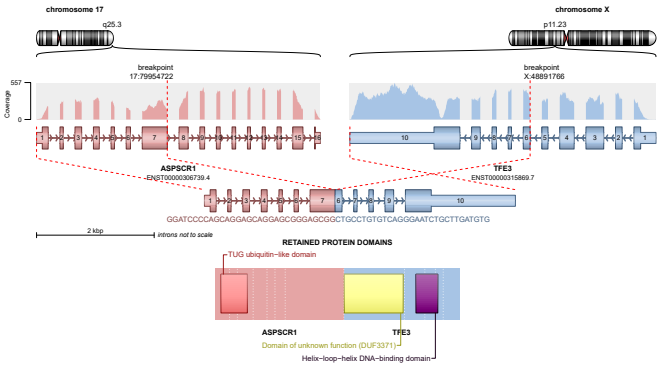


Fig.S1: Mutational signatures and variant allele frequencies (VAF) in pediatric renal cell carcinomas (pRCC), related to Fig.1:

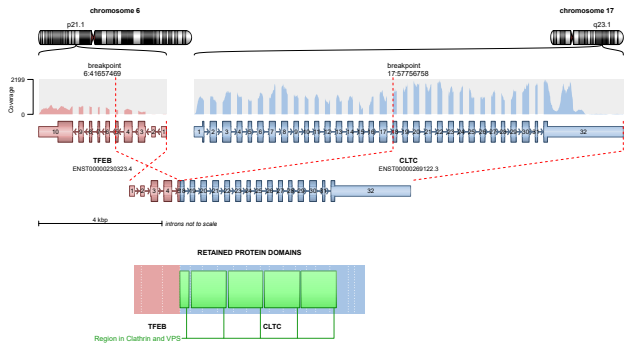
S1a. Mutational signature analysis (COSMIC signatures v3.2) in pediatric RCC samples with higher tumor mutational burden (TMB)
S1b. Variant allele frequencies (VAF) of somatic SNVs in pediatric RCC cases with multiple samples from the same tumor to compare mutation overlap regarding tumor heterogeneity. RCC544_I and RCC544_II are tumor samples taken from different time points of the disease course, while in all other cases multiple samples were taken from different locations of the same tumor piece at the same disease time point.

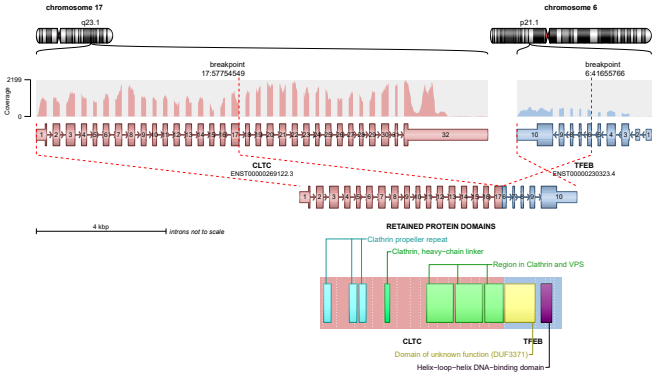
RCC544_II



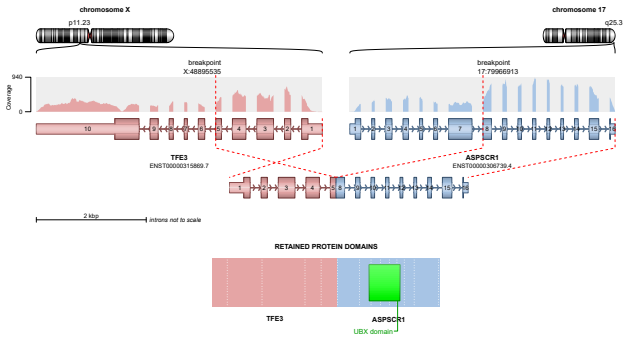


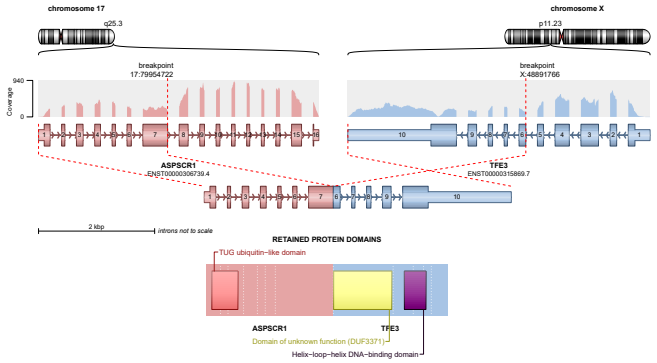
RCC1368



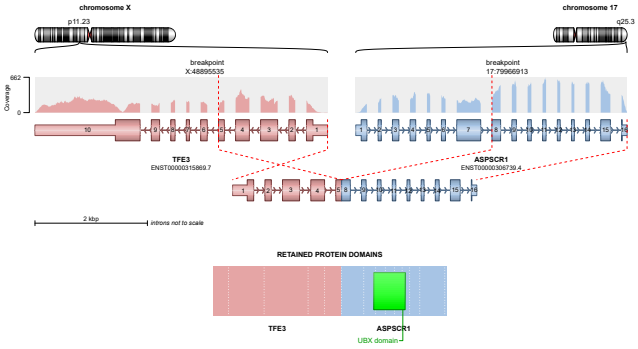


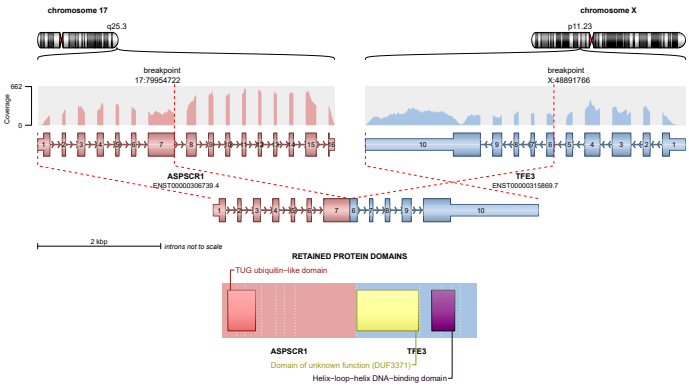
RCC567



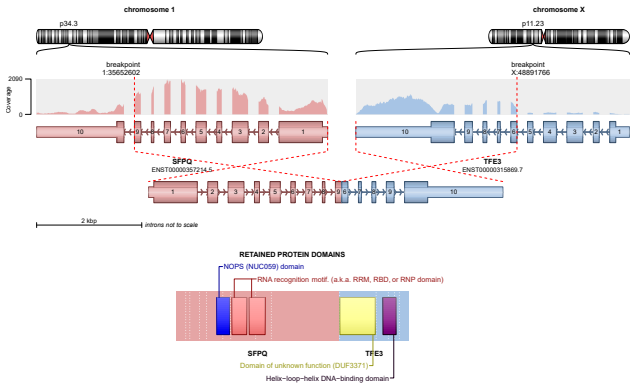


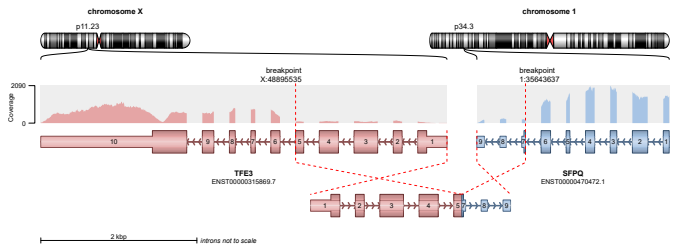
RCC552



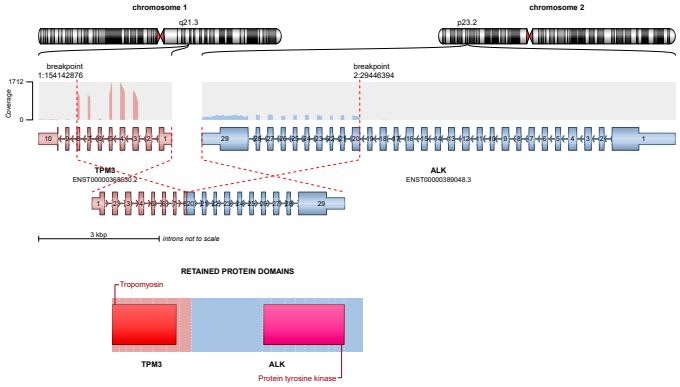


RCC220

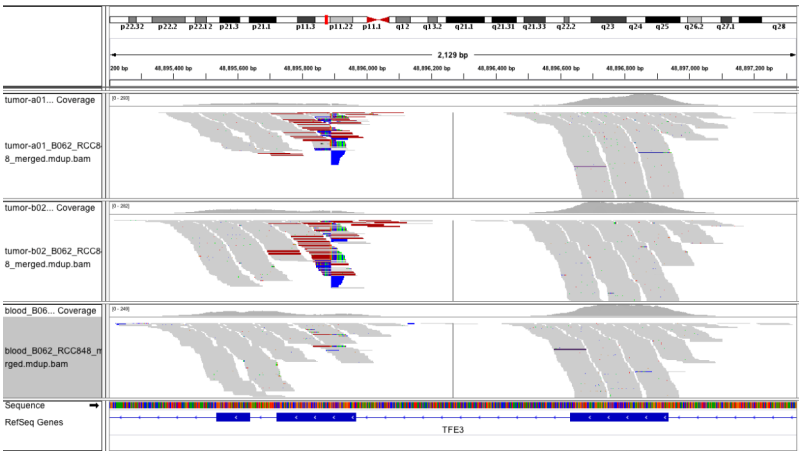




RCC1429



RCC848



RCC1176

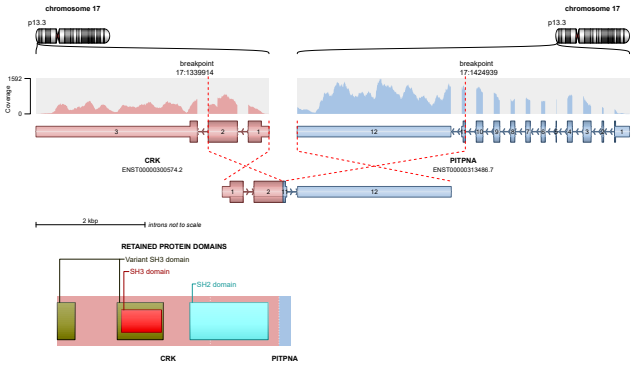


Fig.S2. Fusion genes found in the pediatric RCC cohort, related to Fig.2

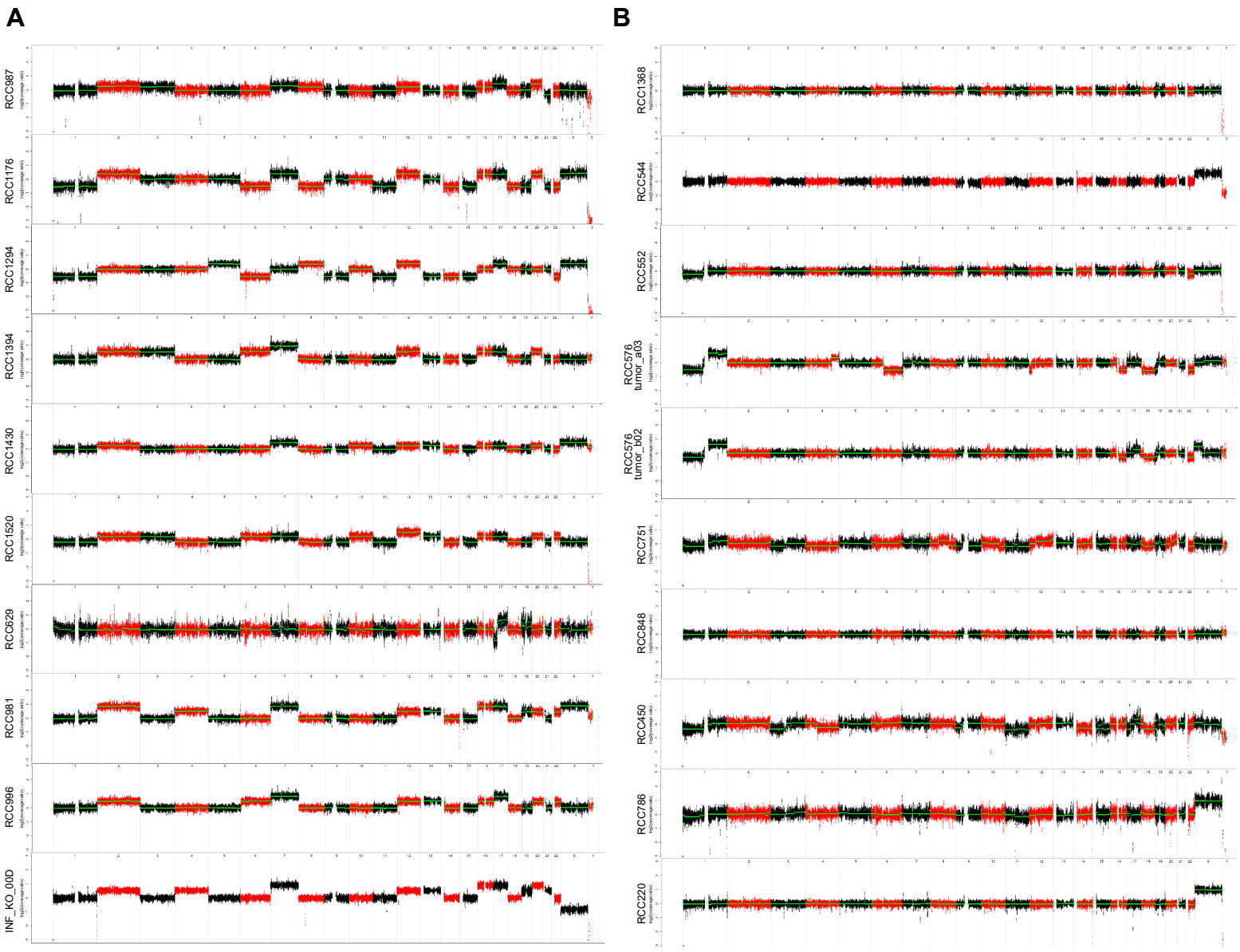
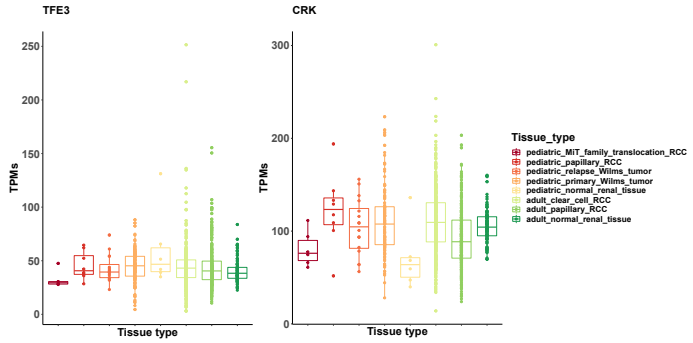


Fig.S3. Tumor genome copy number profiles of the pediatric RCC cohort based on WES data, related to Fig.3

S3a. Copy number profiles of the papillary pediatric RCC samples

S3b. Copy number profiles of the MiT-family translocation pediatric RCC samples; RCC450, RCC786 and RCC220 had no matching blood control and were therefore analysed with the no-control workflow.

A



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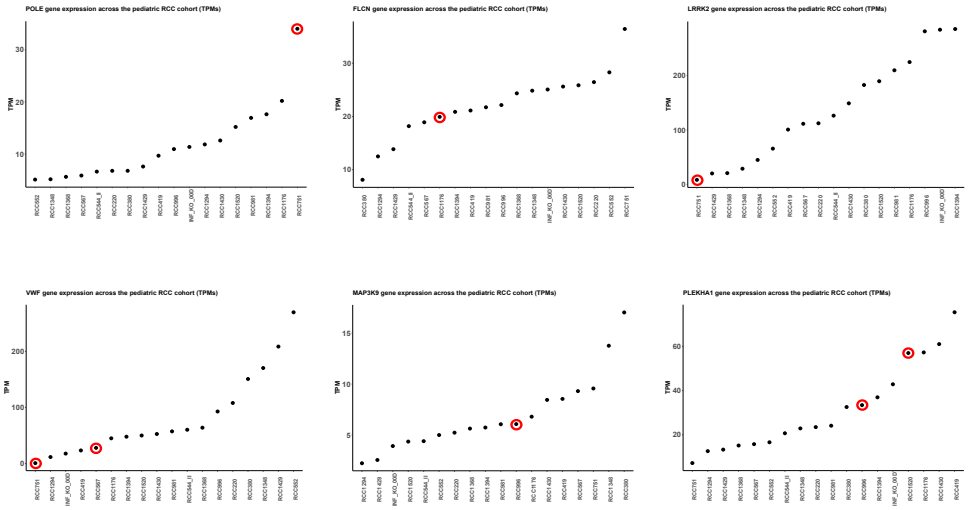

Fig.S4. Gene expression of *TFE3, CRK*, selected germline predisposition genes and selected recurrent somatic mutations in the pediatric RCC cohort, related to Fig.1 and Fig.4

S4a. *TFE3* and *CRK* gene expression in MiT-pRCC and papillary pRCC (ppRCC) compared to Wilms tumor relapses (INFORM), primary Wilms tumors and normal pediatric kidney (TARGET) as well as adult clear cell RCC, adult papillary RCC and adult normal kidney (TCGA)
S4b. Gene expression of the germline predisposition genes *POLE* and *FLCN*, and the recurrently somatic mutated genes *VWF*, *LRRK2*, *MAP3K9* and *PLEKHA1* across the pRCC cohort

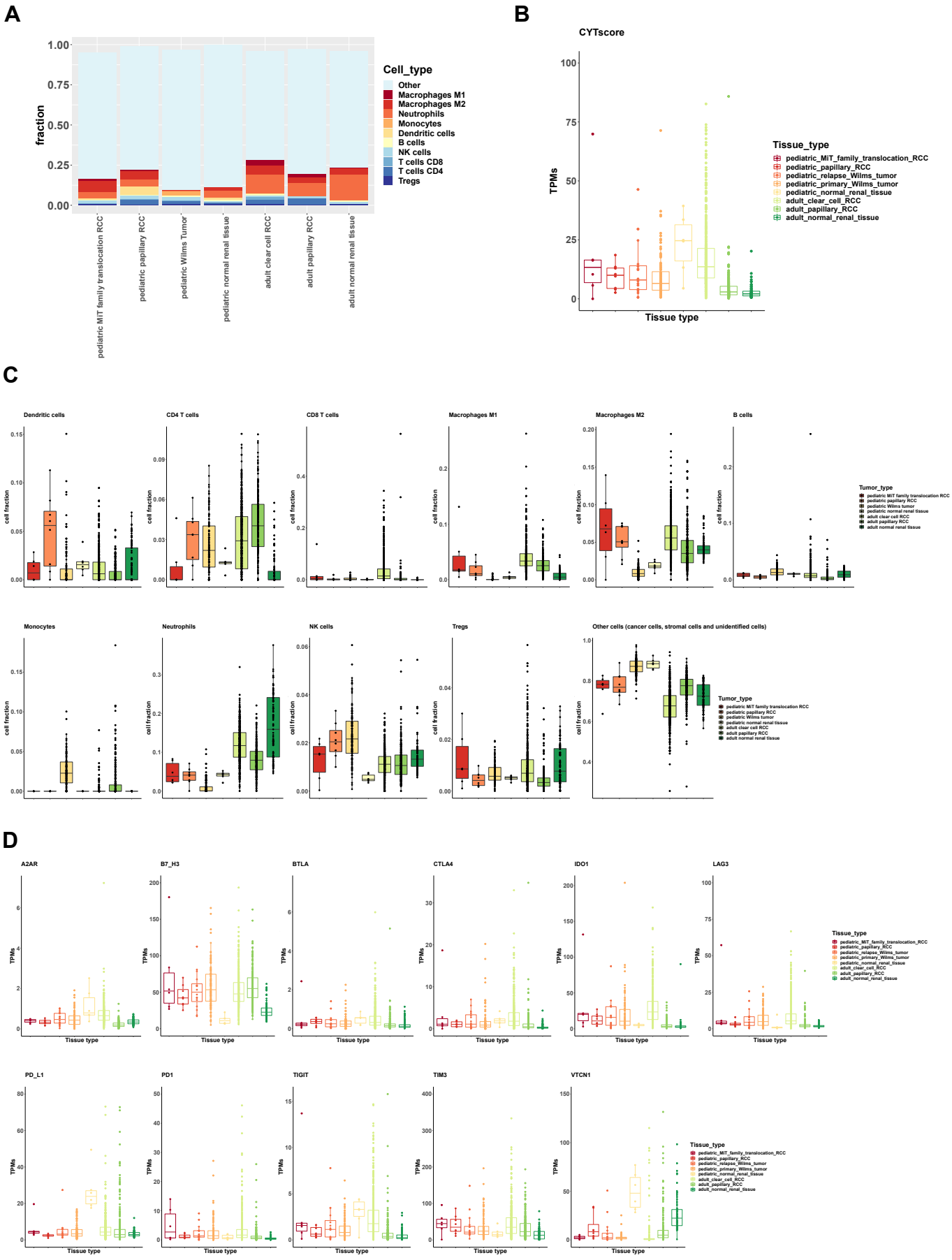

Fig.S5. Tumor immune microenvironment (TME) deconvolution and gene expression of immune checkpoint genes in pediatric RCC (pRCC; n=18) compared to pediatric relapse Wilms tumors (INFORM), pediatric primary Wilms tumors and normal pediatric kidney (TARGET) as well as adult clear cell RCC, adult papillary RCC and adult normal kidney (TCGA), related to Fig.5

S5a. Median value of immune cell fractions in pediatric RCC (pRCC; n=18) compared to pediatric primary Wilms tumors and normal pediatric kidney (TARGET) as well as adult clear cell RCC, adult papillary RCC and adult normal kidney (TCGA)

S5b. Anti-tumoral cytolytic activity (CYTscore) in pediatric RCC (pRCC; n=18) compared to pediatric relapse Wilms tumors (INFORM), pediatric primary Wilms tumors and normal pediatric kidney (TARGET) as well as adult clear cell RCC, adult papillary RCC and adult normal kidney (TCGA)
S5c. Fractions of selected immune cell types in pediatric RCC (pRCC; n=18) compared to pediatric primary Wilms tumors and normal pediatric kidney (TARGET) as well as adult clear cell RCC, adult papillary RCC and adult normal kidney (TCGA)
S5d. Gene expression of selected immune checkpoint genes in pediatric RCC (pRCC; n=18) compared to pediatric relapse Wilms tumors (INFORM), pediatric primary Wilms tumors and normal pediatric kidney (TARGET) as well as adult clear cell RCC, adult papillary RCC and adult normal kidney (TCGA)