An optimized workflow to improve reliability of detection of KIAA1549:BRAF fusions from RNA sequencing data

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Online Resource Fig. 1  

a Relative frequency of detected and missed KIAA1549:BRAF fusions in samples with known KIAA1549:BRAF 15:9 or 16:9 fusions. Fisher's exact test on the underlying absolute values.  
b Amplitude of the fusion-characteristic 7q34 gain extracted from methylation array-derived copy number data as an indicator of tumor cell content in samples with a missed or detected KIAA1549:BRAF fusion. Mean ± SD. Unpaired t test.  
c Expression of BRAF in samples with a missed or detected KIAA1549:BRAF fusion. Mean ± SD. Unpaired t test.  
d Estimated library size of samples with a missed or detected KIAA1549:BRAF fusion as calculated by RNA-SeQC (https://github.com/broadinstitute/rnaseqc). Mean ± SD. Unpaired t test.  
e Relative frequency of detected and missed KIAA1549:BRAF fusions in the presented cohort (polyA capture library preparation protocol) compared to an older cohort (RiboZero library preparation protocol). Fisher's exact test on the underlying absolute values.  
f Samples ranked by their KIAA1549 expression with the highest-ranked sample having the highest expression. Mean ± SD. Kruskal-Wallis test followed by Dunn's multiple comparisons test.  
g Samples ranked by their ESTIMATE immune score with the highest-ranked sample having the lowest immune score. Mean ± SD. Kruskal-Wallis test followed by Dunn's multiple comparisons test.  
h Number of split reads identified by Arriba in the standard and optimized workflow for all samples that were initially reported. Paired t test. For all panels: *p < 0.05, **p < 0.01, ***p < 0.001, ****p < 0.0001, n.s.: not significant.
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Online Resource Fig. 2 Typical copy number plot of a PA with a KIAA1549:BRAF fusion. The characteristic focal tandem duplication at 7q34 is magnified. The gain amplitude is measured between gain and baseline.

Online Resource Table 1 Overview of fusion detection results from RNA-Seq data of the presented cohort of fresh-frozen pediatric PA tumor samples. The higher total read counts after re-sequencing are marked in bold. X = detected