

KIAA1549-BRAF Fusion

Background

A focal tandem duplication on chromosome **7q34** can cause the fusion of genes **KIAA1549** and **BRAF** and a constitutive activation of the mitogen-activated protein kinase (MAPK) pathway (PMID: 18974108). **Increases in copy number values in this region are a hallmark of such a structural variant** (PMID: 33336421). More complex global structural variants, however, may yield false negatives/positives. Regional PCR or RNAseq would provide definitive confirmation (PMID: 33336421).

Algorithm

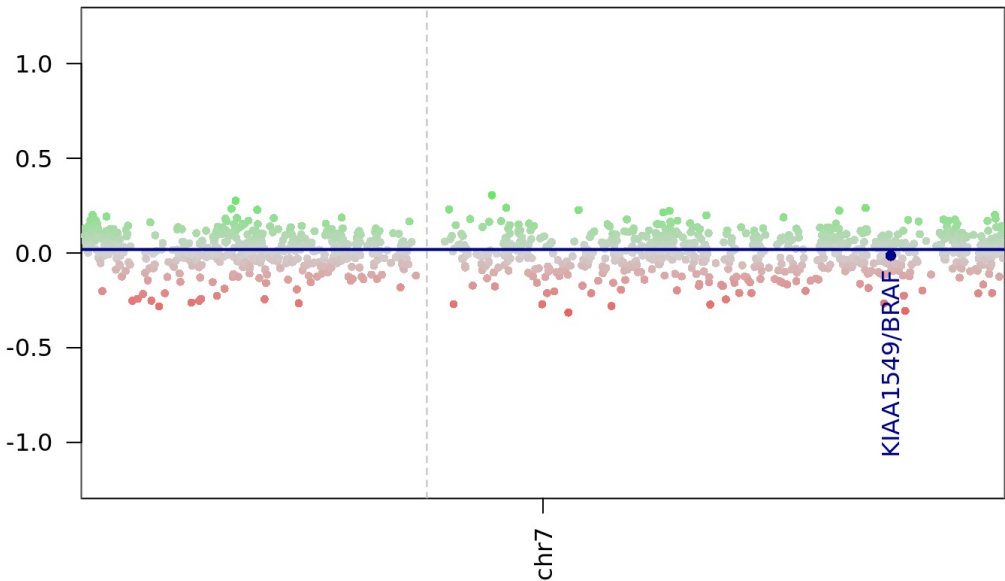
Increases in CNV in 7p34 relative to flanking regions 7p33,7p35 is indicative of a KIAA1549-BRAF fusion. Testing statistically significant differences in these bands extends the methods of Stichel et al, 2021 (PMID: 33336421).

Copy number estimates, binning and segmentations are derived from an implementation of the **conumee** package by the DKFZ mnp_v12b6 classifier package. Binned probe CNPs are used to estimate band effects.

Note: As per Stichel et al, 2021 (PMID: 33336421), a highly segmented profile for chromosome 7 (>10 segments) would yield an unreliable estimate of local increases in copy number values across bands 7q33; 7q34; and 7q35.

Results

NH24-2554 208491430040_R02C01



Main effect of Band

Characteristic	q33, N = 44	q34, N = 28	q35, N = 11	p-value
cnv	-0.01 (-0.06, 0.05)	-0.01 (-0.05, 0.04)	0.01 (-0.04, 0.06)	0.8

¹ Median (IQR)

² One-way ANOVA

Shapiro Normality Test p.value = 0.876

CNPs at bands 7q33, 7q34 and 7q35 are NOT consistent with a KIAA1549-BRAF fusion event.