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Identifying gene copy number variants associated with colorectal adenoma recurrence

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Background

Colorectal cancer is the third leading cancer cause and represents the final stage of a progressive, multi-step, carcinogenic process of evolution through an adenoma stage [1]. Removing colorectal adenomas at colonoscopy significantly decreases cancer risk [2,3]. One-third of colorectal cancer occurs in familial clusters and increases risk to family members [4]; however, most causative genetic factors are unknown. Here we seek genetic factors associated with metachronous adenoma occurrence, hypothesizing that genetic risk factors have been missed because association studies have sought risk-associated single nucleotide polymorphisms, while ignoring structural variation causing gene copy number changes. We used the Database of Genomic Variants [5] to identify gene copy number variation (CNV) in candidate genes from the vitamin D, polyamine, and selenium pathways (Table 1). We re-analyzed Illlumina genotyping data (Figure 1), and experimentally determined candidate gene copy number status for individuals from two interventional trials using MLPA and TaqMan assays. CNV genotypes are compared between individuals who did and did not develop metachronous adenoma to identify associated variants.

Table 1 Population frequency of CNV in Vitamin D pathway genes [5]

Gene	#CNV / #samples studied	Estimate of average
RXRA	28/95; 3/485; 7/2026	10%
GC	2/112	1.2%
CASR	22/2026; 1/90	8%

No reported CNV: VDR

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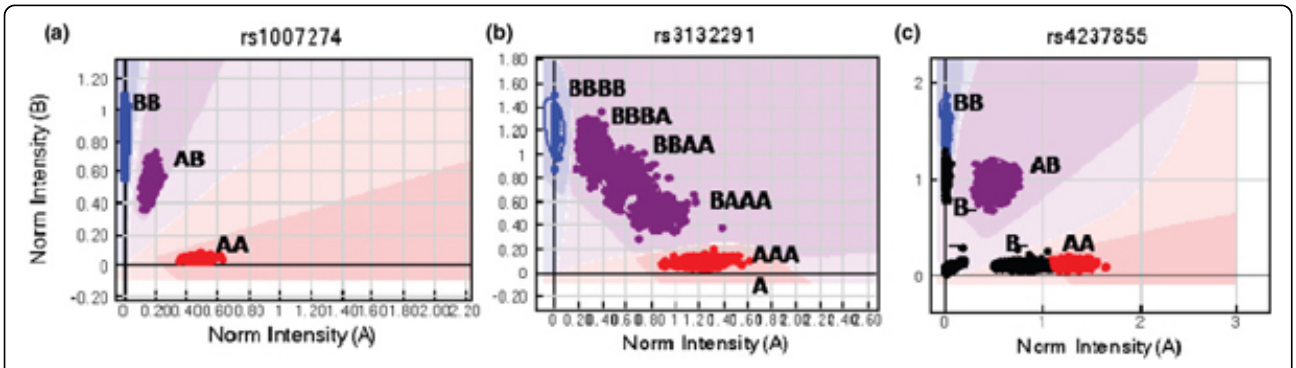

Figure 1 Illumina genotyping data suggesting of CNV in Vitamin D pathway genes.

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