



Supplementary Figure S5. GPS detected allele-specific methylation and verification.

(A) Schematic of workflow for allele-specific methylation identification. SNVs were identified from Read2 of GPS with VarScan (v2.3.9). Raw reads were assigned into different alleles to each heterozygous SNV. Methylation was determined by corresponding Read1. Bar represents the methylation level of a cytosine site. Allele specific methylation was verified by Sanger sequencing from bisulfite treated DNA. Each column of circle represents a cytosine corresponding to the bar. The solid circle (●) and the open circle (○) represent methylated and unmethylated cytosine, respectively. (B) A/G allele of SNP rs16958999 located in the intron of *CCDC97* gene. Allele-specific methylation is in the downstream of third exon of *CCDC97*, which was validated by bisulfite sequencing on the right. Allele-specific methylation locus is enriched with POLR2A, BCL3 binding and DNase I hypersensitive sites. (C) C/T allele of SNP rs72701741 located in *TOP1MT* gene, where allele-specific methylation is detected and validated by bisulfite sequencing PCR with MAX binding and DNase I hypersensitive sites.