

Description of Additional Supplementary Files

File Name: Supplementary Data 1

Description: Cohort details, methylation array quality control and filtering numbers.

File Name: Supplementary Data 2

Description: Detailed variant table for rare DMRs and epesignatures.

File Name: Supplementary Data 3

Description: List of all rare outlier DMRs for the autosomes and chrX.

File Name: Supplementary Data 4

Description: Comparison of CpG coverage for DMRs validated by targeted EM-seq.

File Name: Supplementary Data 5

Description: Variant table from trio sequencing analysis for the individual with a X;13 translocation.

File Name: Supplementary Data 6

Description: X-inactivation assay for mother with BCLAF3 hypermethylation.

File Name: Supplementary Data 7

Description: Probe information for CHD2 450K and 850K epesignatures.

File Name: Supplementary Data 8

Description: CHD2 DMR master list.

File Name: Supplementary Data 9

Description: List of randomly generated genomic coordinates for comparison of functional annotations with CHD2 DMRs.

File Name: Supplementary Data 10

Description: Enrichment values of CHD2 epesignature probes and DMRs.