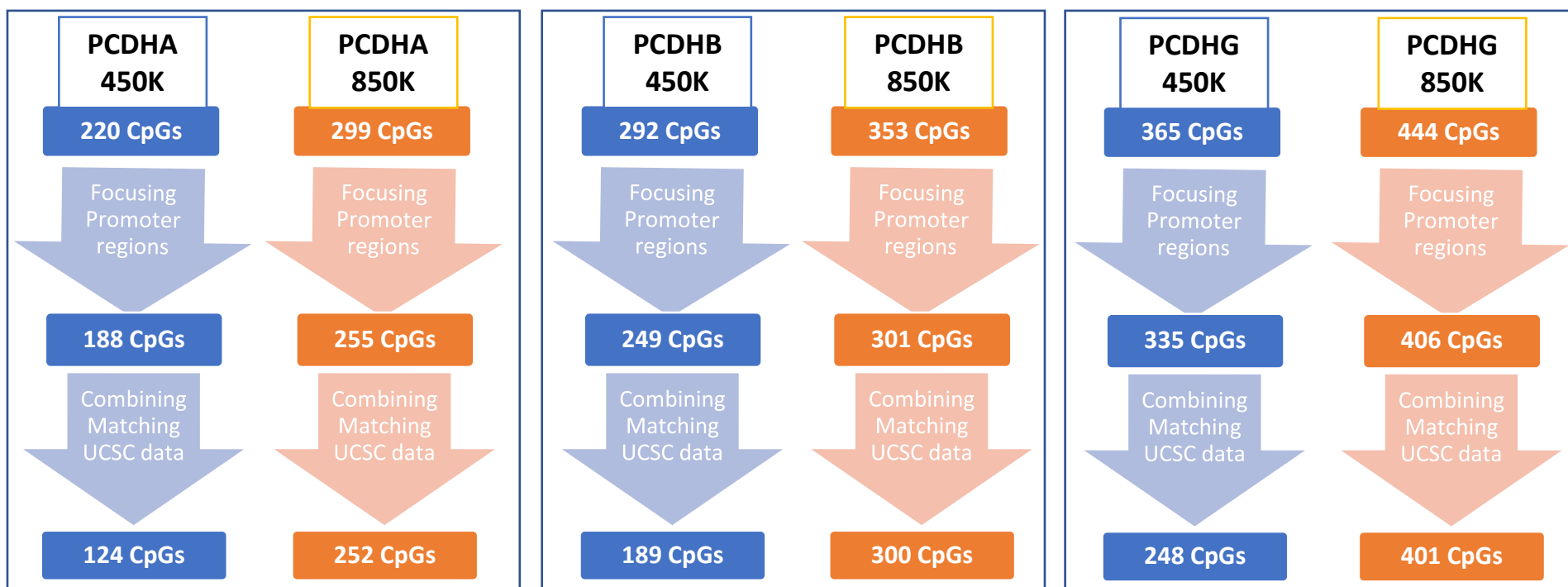


Analysis of Illumina HumanMethylation450K data for cPCDHs

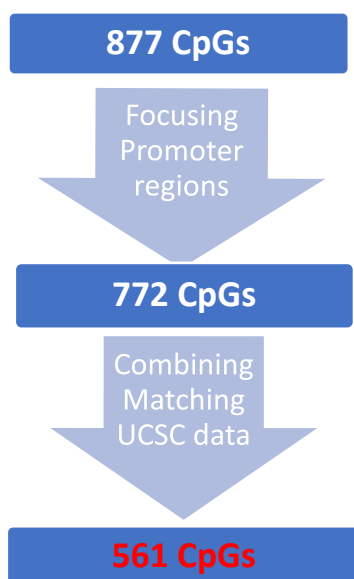
Methylation levels of CpGs (450K) located in the promoter regions PCDH genes for SOTOS, ICF, CHARGE, DUPLICATION7q, Chr16DEL and KABUKI syndromes.

Number of CpGs located in the promoter regions of PCDHs
(450K vs 850K)



Methylation levels of CpGs (450K) located in the promoter regions PCDH genes for SOTOS, ICF, CHARGE, DUPLICATION7q, Chr16DEL and KABUKI syndromes.

- 365 CpGs are in 450K.
- 335 CpGs are in the promoter regions.
- After combining individual beta values from 6 disorders with 772 CpGs from UCSC script, **561 CpGs were detected** in the promoter regions of PCDHs.



Disorder	Number of samples	GEO accession
Sotos	10	GSE74432
Charge	10	GSE97362
Duplication7q11.23	10	GSE66552
Kabuki	10	GSE97362
ICF1	8	GSE95040
ICF2	4	GSE95040
ICF3	2	GSE95040
ICF4	1	GSE95040
Chr16DEL	10	GSE113967
Control	20	GSE74432
Total sample size	85	

- IlluminaHumanMethylation450kanno.ilmn12.hg19 and IlluminaHumanMethylation450kmanifest were used for gene and CpG positions.
- tssRegion=c(-2500, 2500) was used for annotations.
- Beta values for CpGs were obtained from GEO database.

Chromosome 5



140.3 mb

140.5 mb

140.7 mb

140.8 mb

PCDH1A
PCDH1B
PCDH1G

CpG
Islands

0.8

0.6

0.4

0.2

0

<< METHYLATION LEVEL (0-1 beta-value) >>

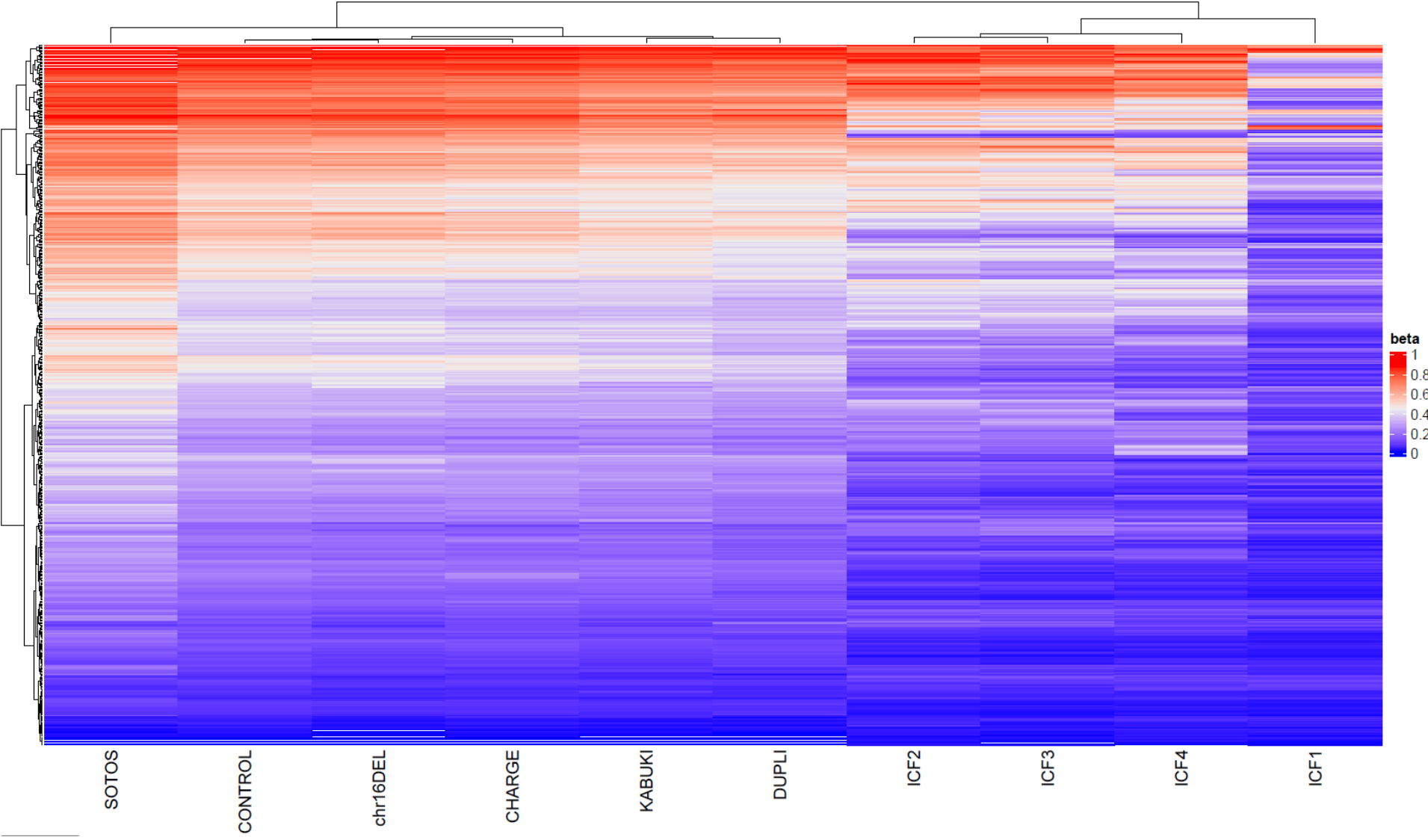
01-SOTOS
05-ICF4
09-chr16DEL

02-ICF1
06-DUPLICATION7q
10-CONTROL

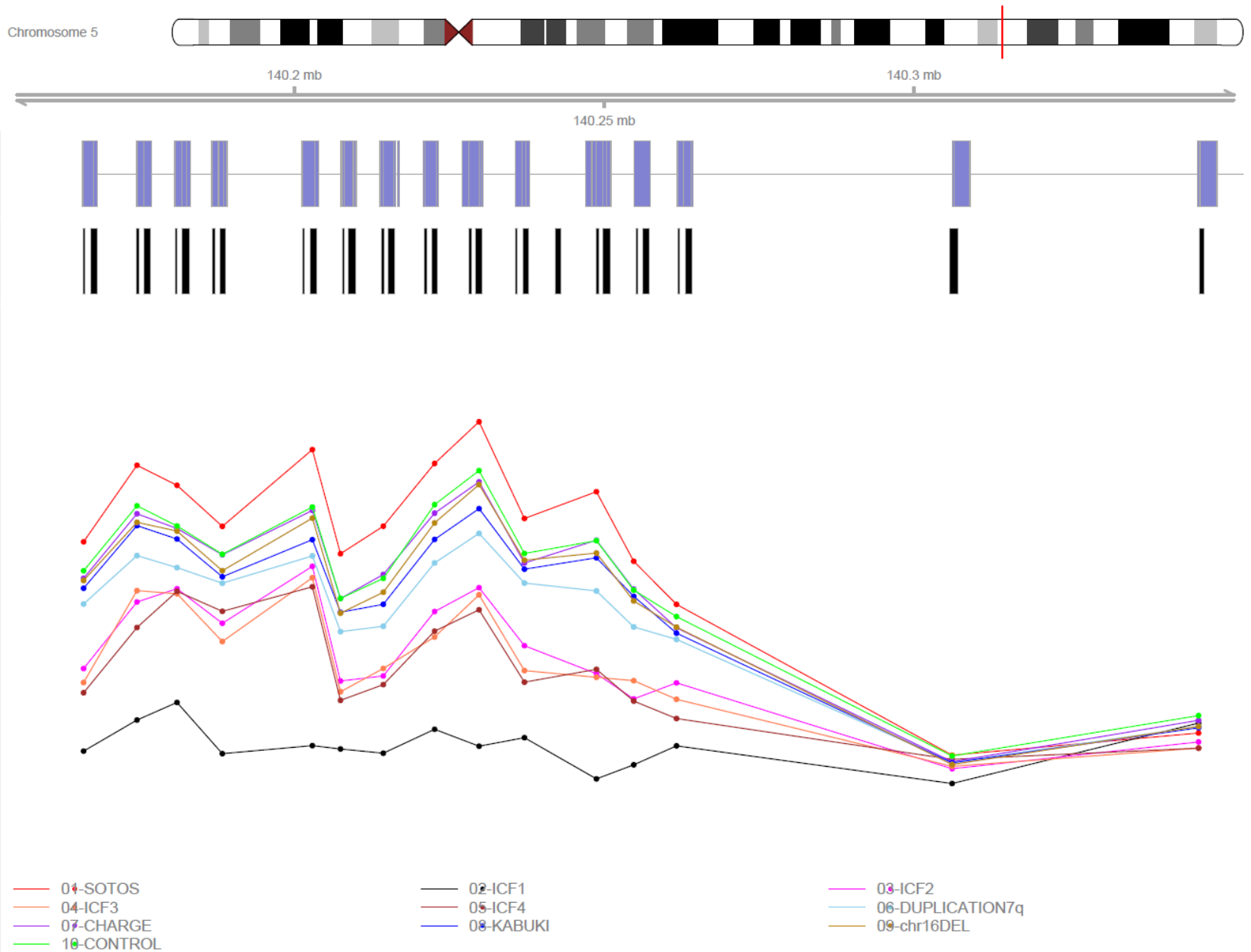
03-ICF2
07-CHARGE

04-ICF3
08-KABUKI

Heatmap according to 561 CpGs located in the promoter regions(2500-TSS+2500) of PCDHAs, PCDHBs and PCDHGs



chr16DEL was added to 450K calculations



Chromosome 5



140.45 mb

140.55 mb

140.5 mb

140.6 mb

PCDHB

CpG
Islands

<< METHYLATION LEVEL (0-1 beta-value) >>

0.8

0.6

0.4

0.2

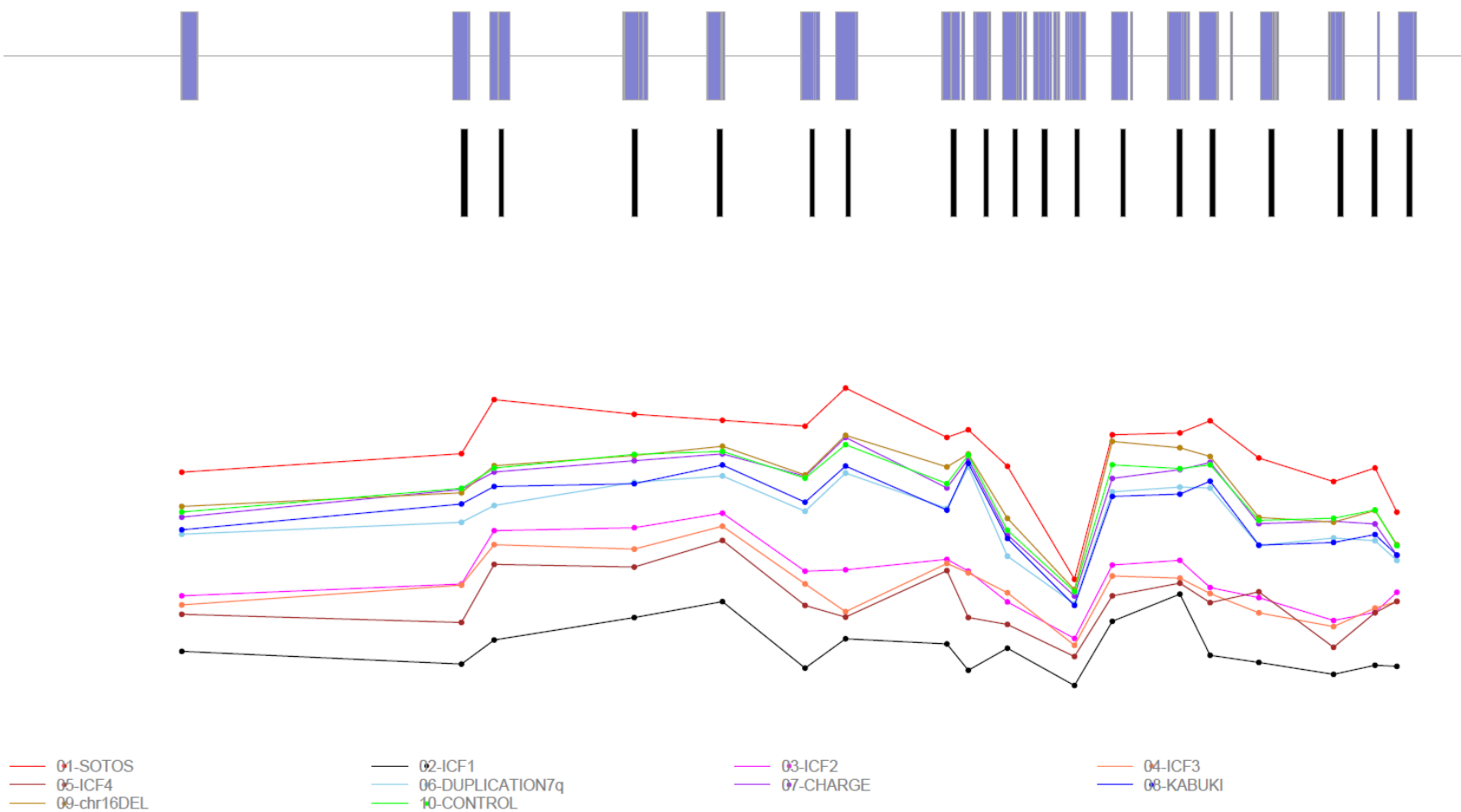
0

01-SOTOS
05-ICF4
09-chr16DEL

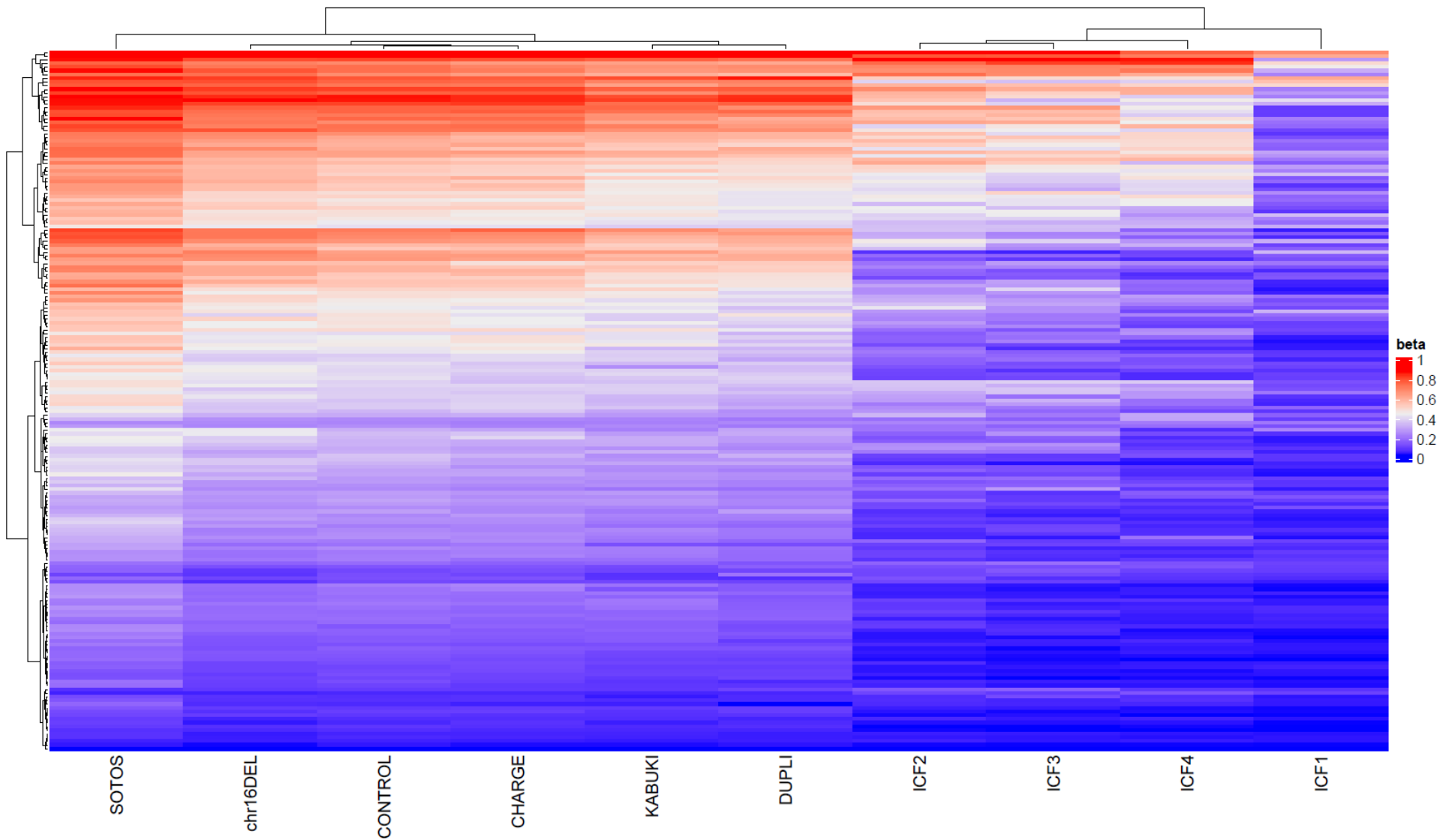
02-ICF1
06-DUPLICATION7q
10-CONTROL

03-ICF2
07-CHARGE

04-ICF3
08-KABUKI



Heatmap according to 189 CpGs located in the promoter regions(2500-TSS+2500) of PCDHBs



Chromosome 5



140.75 mb

140.85 mb

140.8 mb

PCDHg

CpG
islands

<< METHYLATION LEVEL (0 - 1 beta-value) >>

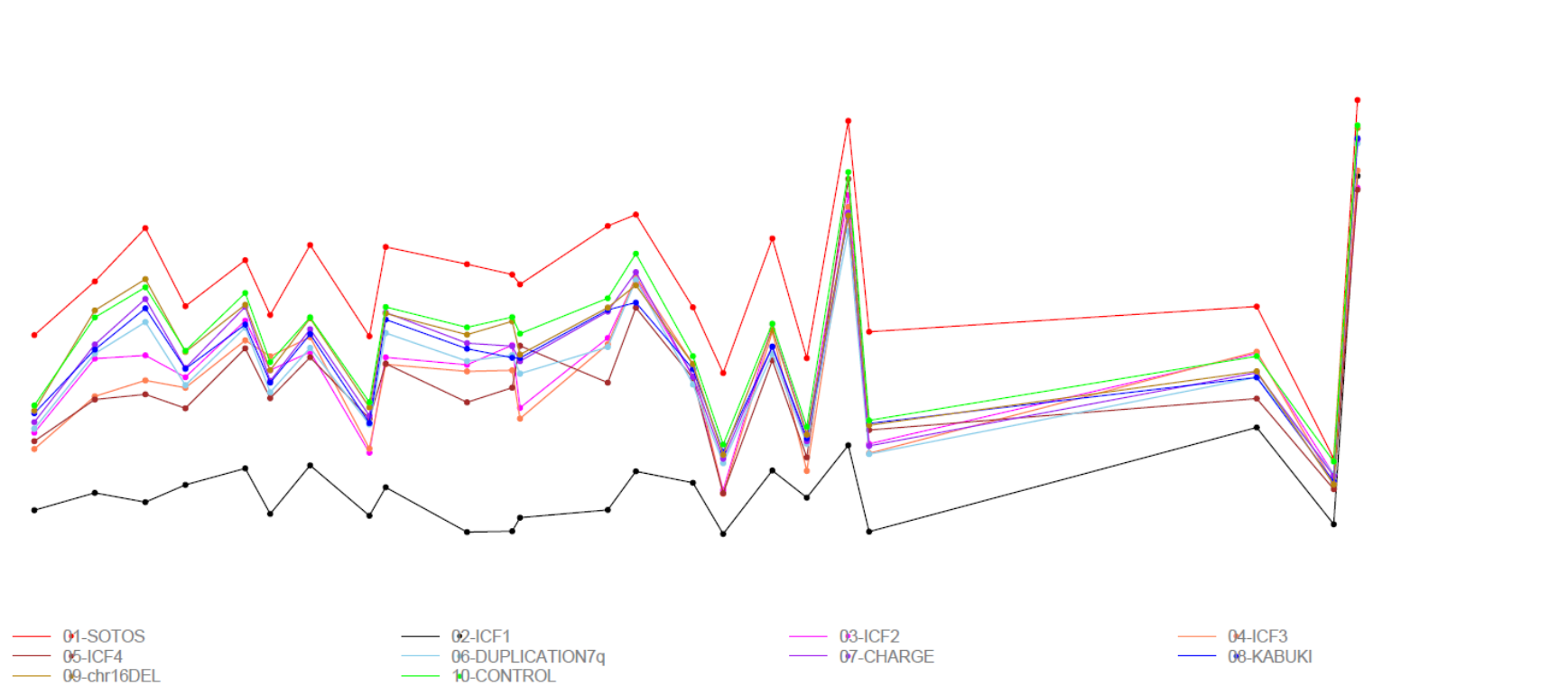
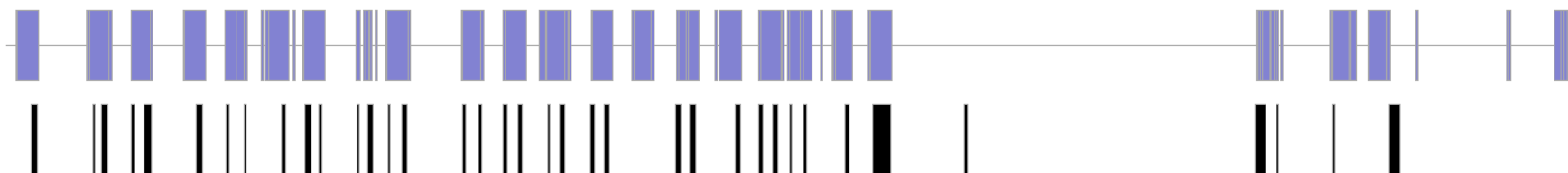
0.8
0.6
0.4
0.2
0

01-SOTOS
05-ICF4
09-chr16DEL

02-ICF1
06-DUPLICATION7q
10-CONTROL

03-ICF2
07-CHARGE

04-ICF3
08-KABUKI



Heatmap according to 248 CpGs located in the promoter regions(2500-TSS+2500) of PCDHGs

