Methylation profiling report

Supplier information

Sample identifier:	428-24
Sentrix ID:	208107870102_R03C01_1
Material type:	NA
Gender:	male
Supplier diagnosis:	brain cancer

Automatic prediction		
Array type:	EPICv2	
Material type:	DNA-FFPE	✗
Gender:	unknown	✗

Legend: ✓ Ok | ! Supplier information or prediction not available | ✗ Warning, missmatch of prediction and supplier information

Version 12.8 of the brain classifier results (12.8)

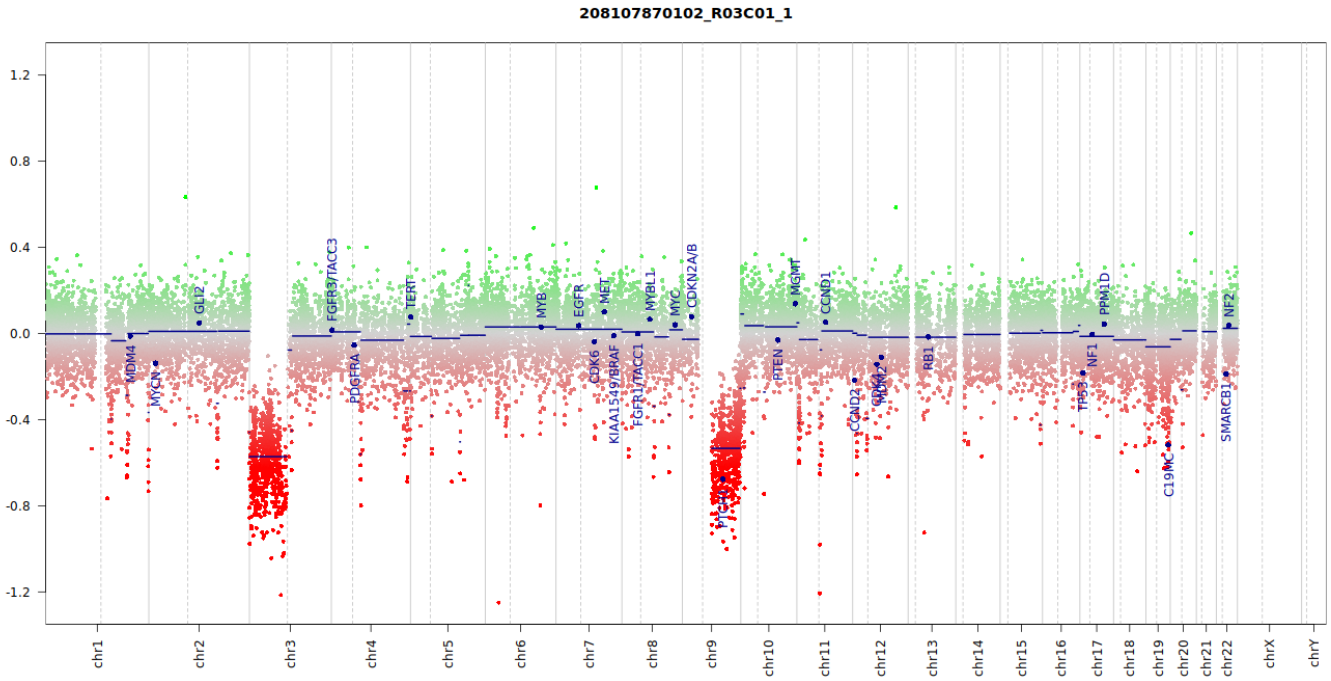
Methylation classes (Highest level >= 0.3, lower levels >= 0.1, all of lowest level)	Calibrated score	Interpretation	
Medulloblastoma	0.99	match	✓
Medulloblastoma, Shh Activated	0.98	match	✓
Medulloblastoma, Shh Activated, Subtype 3	0.54	no match	✗
Mc Medulloblastoma, Shh Activated, Subclass 3 (novel)	0.54	no match	✗
Medulloblastoma, Shh Activated, Subtype 4	0.30	no match	✗
Mc Medulloblastoma, Shh Activated, Subclass 4 (novel)	0.30	no match	✗

Legend: ✓ Match (score >= 0.9) ✗ No match (score < 0.9): possibly still relevant for low tumor content and low DNA quality cases.

Class descriptions

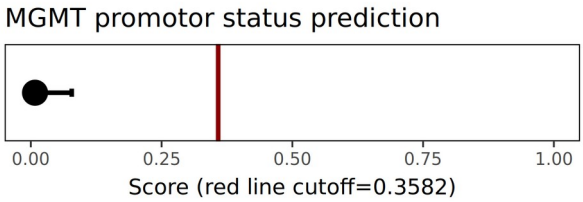
MC Medulloblastoma, SHH-activated, subclass 3 (novel): The "mf Medulloblastoma, SHH activated" comprises the "mc Medulloblastoma, SHH-activated, subtype 1" (mostly infants and SUFU mutant, including germline) , "mc Medulloblastoma, SHH-activated, subtype 2" (9q loss, extensive nodularity), "mc Medulloblastoma, SHH-activated, subtype 3" (non-infant, often TP53 or ELP1 mutant), "mc Medulloblastoma, SHH-activated, subtype 4" (non-infant, mostly U1 and TERT mutant, often PTCH1/SMO altered) and "medulloblastoma, SHH-activated, IDH mutant" and represents Medulloblastoma, molecularly defined, SHH-activated. The molecular classes and subclasses are still being further characterized and in part provisional. Generally, germline or somatic mutations in PTCH1 (~40% of tumours), SMO (~10%) and in SUFU (~10%) are frequent, and amplifications of GLI1 or GLI2 (~10%) and other downstream SHH target genes (MYCN, MYCL, and YAP1; smaller 10%). Common copy-number variations in SHH-activated medulloblastoma include losses of chromosome 9q and 10q (including PTCH1 at 9q22 and SUFU at 10q24). Germline predisposition is often observed in SHH-activated medulloblastoma, thus comprehensive analysis including blood for PTCH1, SUFU, TP53, ELP1, and GPR161, accompanied by genetic counselling may be advised. TP53-wildtype SHH medulloblastomas mostly show desmoplastic/nodular morphology or extensive nodularity, and to rarer extent classic or large cell / anaplastic. Recurrences can transforma to a focal anaplastic morphology. In TP53-mutant SHH medulloblastoma, diffuse anaplasia with partly large-cell phenotype occur in 70%. whereas others show desmoplastic/nodular morphology with focal anaplasia.

Copy number variation profile



Depiction of chromosome 1 to 22 (and X/Y if automatic prediction was successful). Gains/amplifications represent positive, losses negative deviations from the baseline. 29 brain tumor relevant gene regions are highlighted for easier assessment. (see Hovestadt & Zapatka, http://www.bioconductor.org/packages/devel/bioc/html/conumee.html)

MGMT promotor methylation (MGMT-STP27)



Status	Estimated	CI lower	CI upper
unmethylated	0.00801	0.00077	0.07820

(see Bady et al, J Mol Diagn 2016; 18(3):350-61)

Disclaimer

Classification using methylation profiling is a tool for research use only, it is not verified and has not been clinically validated and, therefore, must not be used for diagnostic purposes. This tool is not HIPAA compliant.

Run information

Report: report_website_mnp_brain_v12.8_sample (Version 1.1)

Task version:

Task	Version
idat_preprocess	3.1
idat_qc	4.1
idat_predictBrain	12.8
idat_mgmt	3.1
idat_rs_gender	3.2
idat_cnvp	5.2
report_website_mnp_brain_v12.8_sample	1.1