

# EpiGe-App

## EpiGe Medulloblastoma Subgroup Prediction (v1.0)

Basic Information	
Sample ID:	Sample_test
Uploaded file:	EpiGe_samples_test.txt
Size file:	0.8 Mb
Date:	Mar 02, 2023, 18:33:18

## Medulloblastoma Subgroup Assignment

### Medulloblastoma, non-WNT/non-SHH

Medulloblastoma, non-WNT/non-SHH is an embryonal tumour of the cerebellum without a molecular signature associated with activation of the WNT or SHH signalling pathway. Non-WNT/non-SHH are classified as group 3 and group 4 tumours and comprise eight molecular subgroups. Group 3 tumours occur during infancy and childhood, and account for about 25% of medulloblastomas (~40% of cases in infants). Group 4 medulloblastomas occurs across all age groups, accounting for 40% of cases. Approximately 30-40% of patients of both subgroups, present metastasis disease at the moment of diagnosis. Histologically most cases fall into the classical and large cell/anaplastic groups. Group 3 and group 4 are characterized by partly overlapping genetic features, making their distinction in some cases difficult. Consequently, group 3 and group 4 have been described as a single subgroup designated non-WNT/non-SHH. *MYC* or *MYCN* amplification, isochromosome 17q (found in >50%) and GFI1/1B activation by enhancer hijacking are recurrent features in both subgroups. *MYC* amplification is a common genetic alteration seen in group 3 (17%) medulloblastomas, and is associated with a poor outcome. *MYCN* amplification is the most frequent alteration identified in group 4 medulloblastomas. Metastatic disease at presentation of the disease, is associated with poor outcome in group 4 medulloblastomas.

### Disclaimer

EpiGe-App is a research tool that is intended only for scientific purposes: the EpiGe-App has not been validated clinically and was not designed to be used for diagnostic purpose nor to replace the services of a licensed, trained physician or health professional or to be a substitute for medical advice.

## Cytosine Methylation Status Predictor

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**Table of reference patterns**

Subgroup	cg18849583	cg01268345	cg10333416	cg12925355	cg25542041	cg02227036
non-WNT/non-SHH	1	0	0	1	0	1
SHH	0	1	1	0	0	1
WNT	0	1	0	1	1	0

**Table of probabilities of being methylated obtained by logistic regression**

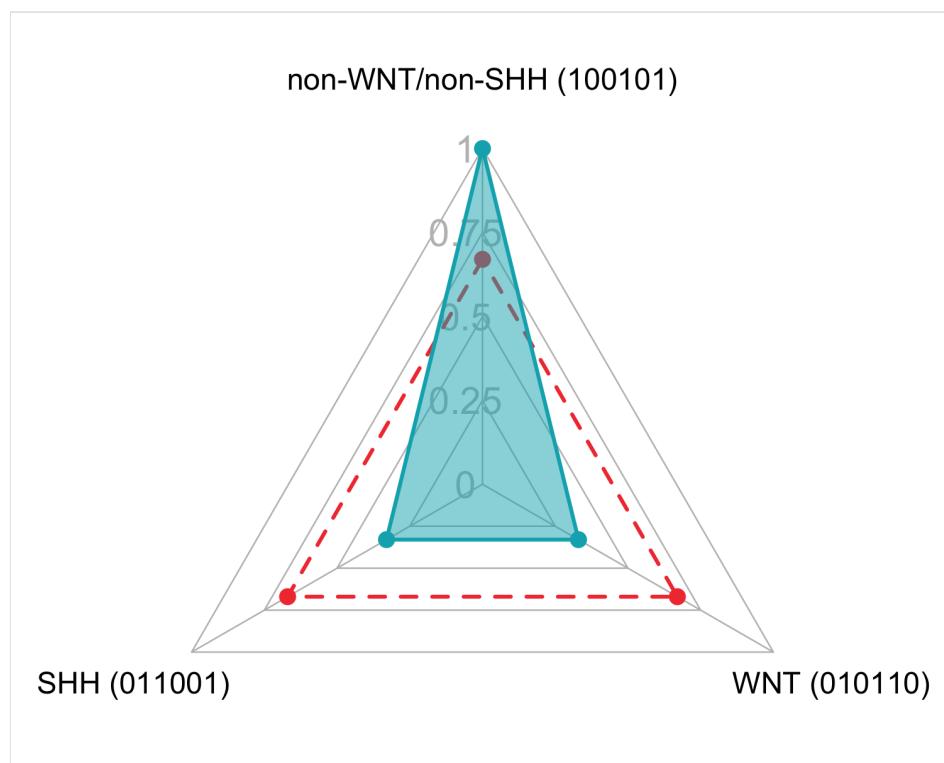
CpG	Status	Probability of being methylated
cg18849583	Methylated	0.96
cg01268345	Unmethylated	0.01
cg10333416	Unmethylated	0.00
cg12925355	Methylated	0.73
cg25542041	Unmethylated	0.01
cg02227036	Methylated	0.59

**Table of sample pattern obtained by logistic regression**

Sample_ID	cg18849583	cg01268345	cg10333416	cg12925355	cg25542041	cg02227036
Sample	1	0	0	1	0	1

## Medulloblastoma Subgroup Classification by Hamming Distance

### Radar plot with Hamming distances



### Table of scores

Molecular subgroup	Score for WNT	Score for SHH	Score for non-WNT/non-SHH
non-WNT/non-SHH	0.33	0.33	1.0

## Quality Control Summary

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CheckPoint	Result	Possible Solution
<b>Text file complete</b>	The txt file contains all the information.	
<b>All CpG</b>	All 6 CpG are included.	
<b>Dye calibration</b>	All dyes are calibrated.	
<b>No template controls</b>	No amplification was detected in any no template control.	
<b>SD &lt; 0.5</b>	Some pairs of replicates have a standard deviation greater than 0.5.	See the technical report for more information.

## Technical report

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### Calibration information

ROX is expired	ROX performed	FAM is expired	FAM performed	VIC is expired	VIC performed
No	01-25-2022	No	01-25-2022	No	01-25-2022

### No template control amplification

Well Position	SNP Assay Name	Allele1 Ct	Allele2 Ct
A1	cg25542041	Undetermined	Undetermined
A2	cg02227036	Undetermined	Undetermined
A3	cg10333416	Undetermined	Undetermined
A4	cg12925355	Undetermined	Undetermined
A5	cg18849583	Undetermined	Undetermined
A6	cg01268345	Undetermined	Undetermined
B1	cg25542041	Undetermined	Undetermined
B2	cg02227036	Undetermined	Undetermined
B3	cg10333416	Undetermined	Undetermined
B4	cg12925355	Undetermined	Undetermined
B5	cg18849583	Undetermined	Undetermined
B6	cg01268345	Undetermined	Undetermined

No amplification detected.

### SD < 0.5

SNP Assay Name	Allele1 Ct Std	Allele2 Ct Std
cg18849583	0.02	0.00
cg01268345	0.12	0.14
cg10333416	0.00	0.28
cg12925355	0.72	0.92
cg25542041	0.00	0.86
cg02227036	0.00	0.00

Some pairs of replicates have a standard deviation greater than 0.5.

## Run information

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#	Task	Version
1	EpiGe_qc	1.0
2	EpiGe_logisticRegression	1.0
3	EpiGe_hammingDistance	1.0
4	EpiGe_report	1.0

## References

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1. [Louis DN, Perry A, Wesseling P, Brat DJ, Cree IA, Figarella-Branger D, Hawkins C, Ng HK, Pfister SM, Reifenberger G, Soffietti R, von Deimling A, Ellison DW. The 2021 WHO Classification of Tumors of the Central Nervous System: a summary. Neuro Oncol. 2021 Aug 2;23\(8\):1231-1251. doi: 10.1093/neuonc/noab106. PMID: 34185076; PMCID: PMC8328013.](#)
2. [WHO Classification of Tumours. Central Nervous System Tumours/Fifth Edition, IARC 2021.](#)
3. [Gómez S, Garrido-García A, García-Gerique L, Lemos I, Suñol M, de Torres C, Kulis M, Pérez-Jaume S, Carcaboso ÁM, Luu B, Kieran MW, Jabado N, Kozlenkov A, Dracheva S, Ramaswamy V, Hovestadt V, Johann P, Jones DTW, Pfister SM, Morales La Madrid A, Cruz O, Taylor MD, Martin-Subero JI, Mora J, Lavarino C. A Novel Method for Rapid Molecular Subgrouping of Medulloblastoma. Clin Cancer Res. 2018 Mar 15;24\(6\):1355-1363. doi: 10.1158/1078-0432.CCR-17-2243. Epub 2018 Jan 19. PMID: 29351917.](#)

## Involved parties

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## Funding

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SJD HOSPITAL  
ASSOCIATIONS OF  
PATIENTS' FAMILIES



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