CÖRE DIAGNOSTICS™

Case ID	: 24010006043
Patient Name	: Mr. R.H.S.R PREMARATNE
Age/DOB/Sex	: 55 Years / / Male
Hospital Name	: Aegle Omics (Private) Limited, Colombo
Physician Name	: Dr. Wasantha Rathnayake
Regn Date	: 11-Jul-2024 10:29
Collection On	: 08-Jul-2024 00:00
Reported On	: 19-Jul-2024 20:33
Process AT	: CORE-Gurugram
Ref no	:
Sample Type	: FFPE Block
Report Status	: Final

MC-2256

UNIQUE PATIENT ID: 99685

TEST NAME

IDH 1 & 2 Mutation Analysis

SPECIMEN INFORMATION

Received 02 paraffin blocks labelled as GJ4727(A & B).

CLINICAL HISTORY

Astrocytoma gemistocytic type (CNS WHO Grade 2/3)

METHODOLOGY

PCR + Sequencing

RESULT

Mutant

DIAGNOSIS

MOLECULAR TEST	INTERPRETATION	Molecular Mutation Tested	Result
IDH1 Mutation			
IDH1 Mutation Exon 4 **Mutant**	**Mutant**	IDH1 132	**Mutant**
IDH2 Mutation			
IDH2 Mutation Exon 4 Wild Type	Wild Type	IDH2 172	Wild Type

COMMENTS

1. There is presence of IDH1 codon 132 (R132H) mutation in the given specimen.
2. A positive test indicates the presence of an IDH1 or IDH2 mutation and supports a diagnosis of grade II or III astrocytoma, oligodendroglioma, oligoastrocytoma or secondary glioblastoma. It is important to note that IDH1 and IDH2 mutations have been found in a variety of other tumors.
3. IDH1 codon 132 and IDH2 codon 172 mutations have been identified in more than 70% of brain tumors diagnosed as grade II and III astrocytoma, oligodendroglioma, oligoastrocytoma and secondary glioblastomas. These mutations are rarely found in other brain tumors and non-brain tumors. The ordering physician is responsible for the diagnosis and management of disease and decisions based on the data provided.
4. False-negative results may occur in specimens when tumor cells comprise <40% of the cell population. Tumor cells are routinely enriched by macrodissection to avoid false-ne

COMMENTS

Assay Description And Methodology:

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COLLEGE of AMERICAN PATHOLOGISTS
AU-ID-2010258

Dr. Shivani Sharma
DCP, DNB, DipRCPath.
Reg. No. 1906

Dr. Rahul Katara
Ph.D.

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Formalin-fixed, paraffin embedded tumor tissue sections are deparaffinized and DNA is extracted using the DNeasy Blood and Tissue Kit (Qiagen, Valencia, CA). Mutated IDH oncogenes are detected using Sequencing based PCR kit. Mutation of (IDH-1) appears to be a very strong prognostic factor in diffuse gliomas and mutations of the related (IDH-2) gene were also detected in astrocytic and oligodendroglial gliomas lacking (IDH-1) mutations.

Intended Use:

This test is intended to be used and be interpreted in conjunction with all other available clinical and laboratory information. Diagnosis of glioma; patients with glioma or AML for whom prognosis/risk stratification is sought.

The test is validated for use with CRC FFPE tissue specimens that contain at least 40% tumor area, or that can be enriched to that tumor content in the course of a histological specimen review. The test has not been validated on other specimen types or other human malignancies.

Disclaimer:

This test is performed using an in-house developed and validated test. The assay is designed to perform the reactions at the specified analytical sensitivity given that the template DNA is not heavily fragmented and does not contain materials that could inhibit the amplification reaction.

REFERENCES

1. Dieffenbach CW, and GS Dveksler 2003 PCR Primer: A Laboratory Manual. Cold Spring Harbor, New York: ColdSpring Harbor Laboratory Press.

2. Innis MA, DH Gelfand, JJ Sninsky, and TJ White (eds.) 1990 PCR Protocols: A Guide to Methods and Applications. San Diego, California:Academic Press.

3. McPherson MJ, SG Moller, R Beynon, and C Howe 2000 PCR: Basics from Background to Bench. Heidelberg:Springer-Verlag.

4. Parsons DW, Jones S, Zhang X, et al. An integrated genomic analysis of human glioblastoma multiforme. Science. 2008;321:1807–1812.

5. Balss J, Meyer J, Mueller W, et al. Analysis of the IDH1 codon 132 mutation in brain tumors. Acta Neuropathol. 2008;116:597–602.

6. Bleeker FE, Lamba S, Leenstra S, et al. IDH1 mutations at residue p.R132 (IDH1(R132)) occur frequently in high-grade gliomas but not in other solid tumors. Hum Mutat. 2009;30:7–11.

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Scan to Connect

If you have any questions about this report or would like to have a conversation about the test results, please feel free to reach out to us at

+91 88828 99999 or **info@corediagnostics.in**

CONDITIONS OF REPORTING

1. The tests are carried out in the lab with the presumption that the specimen belongs to the patient named or identified in the bill/test request form.
2. The test results relate specifically to the sample received in the lab and are presumed to have been generated and transported per specific instructions given by the physicians/laboratory.
3. The reported results are for information and are subject to confirmation and interpretation by the referring doctor.
4. Some tests are referred to other laboratories to provide a wider test menu to the customer.
5. CORE Diagnostics Pvt. Ltd. shall in no event be liable for accidental damage, loss, or destruction of specimen, which is not attributable to any direct and mala fide act or omission of CORE Diagnostics Pvt. Ltd. or its employees. Liability of CORE Diagnostics Pvt. Ltd. for deficiency of services, or other errors and omissions shall be limited to fee paid by the patient for the relevant laboratory services.

CŌRE Diagnostics (Central Reference Lab) - Gurugram
406, Udyog Vihar, Phase III, Gurugram, Haryana - 122016

CŌRE Diagnostics Satellite Lab - New Delhi
C-13, 1st Floor, Green Park Extension, New Delhi - 110016
67, Hargobind Enclave, New Delhi - 110092
H64, Block H, Bali Nagar, New Delhi - 110015

CŌRE Diagnostics Satellite Lab - Bhubaneswar
Plot No. - 249, Near Police Academy, AIIMS Nagar, Patrapada, Bhubaneswar - 751019

CŌRE Diagnostics Satellite Lab - Bangalore
1st Floor, KMK Tower, 142 KH Road, Bangalore - 560027

CŌRE Diagnostics Satellite Lab - Lucknow
J.S. Tower, Plot No. - K-702, Sector K, Ashiyana, Near Raj Luxmi Sweets, Lucknow, Uttar Pradesh - 226012

CŌRE Diagnostics Satellite Lab - Guwahati
Ground Floor, Honuram Boro Path, Shubham Velocity, GS Road, Dispur, Kamrup Metropolitan, Guwahati, Assam - 781005

CŌRE Diagnostics Satellite Lab -Hyderabad
S. No.155, Mahalakshmi Trade Center, 2nd Floor, Thokatta -Village, Diamond Point Road, Bowenpally, Secunderabad,Telangana-500009