

Test Description

The MolQ Sanger Validation test confirms the variants obtained from high through put technology using sanger sequencing.

Patient Demographic

Name: Ms Archita Singh
Sex: Female
Date of Birth/Age: 46 years
Disease: Carcinoma Breast

Clinician

Clinician Name: Dr Amit Verma
Medical Facility: Dr AV Institute of Personalized Cancer Therapy and Research
Pathologist: Not Provided

Specimen

Booking ID: 012206070082
Site: NA
Sample Type: Blood
Date of Collection: 07-06-2022
Date of Booking: 07-06-2022

CLINICAL SYNOPSIS

Ms. Archita Singh, presented with clinical indications of Carcinoma Breast (right). The NGS sequencing data analysis has identified variant, c.1340+1G>A in *BRIP1* gene [Test Outside MolQ Laboratory]. The same variation is being validated by Sanger sequencing.

RESULTS

Variant is confirmed to be present by Sanger sequencing.

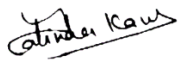
Analysis For: Variation in <i>BRIP1</i> Gene		Gene Name: <i>BRIP1</i> (Intron 9)
S.No.	Variation Detected in NGS	Sanger Validation Result
1.	chr17:59876460C>T; c.1340+1G>A (splice donor variant)	Present (Heterozygous)

*The variant analysis in Sanger sequencing is based on the *BRIP1* gene reference sequence NM_032043.3¹. The intron number and nucleotide numbers will differ based on the reference file chosen and the database used.

CLINICAL CORRELATION AND VARIANT INTERPRETATION

Variant description: A heterozygous variant in intron 9 of the *BRIP1* gene (**chr17:59876460C>T; c.1340+1G>A**) was reported in Ms. Archita Singh by NGS.

The same variation was detected in heterozygous condition in this patient by Sanger sequencing (Figure 1).



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APPENDIX 1: TEST METHODOLOGY

METHOD

Targeted gene Sanger sequencing: Intron 9 of the *BRIP1* gene was PCR-amplified and the product was sequenced using Sanger sequencing. In case of mosaicism in leucocytes, the detection limits of Sanger sequencing for presence of variants are ~20%. The sequences were aligned to available reference sequences NM_032043.3¹ to detect variants using variant analysis software programs.

DISCLAIMER

About 0.44% of total cases are susceptible to allele dropout/dropin phenomenon, which can lead to misdiagnosis².

REFERENCES

- https://www.ncbi.nlm.nih.gov/nucore/NM_032043
- Blais, Jonatan *et al.* Risk of Misdiagnosis Due to Allele Dropout and False-Positive PCR Artifacts in Molecular Diagnostics. *The Journal of Molecular Diagnostics*, Volume 17, Issue 5, 505 – 514.

Figure 1: Sequence chromatogram and alignment to the reference sequence showing the variant in intron 9 of the *BRIP1* gene [chr17:59876460C>T; c.1340+1G>A (splice donor variant)] detected in heterozygous condition in Ms. Archita Singh.

