

Test Description

Lynch syndrome is a hereditary cancer arising from loss of function mutations in DNA mismatch repair genes, such as *MLH1*, *MSH2*, *MSH3*, *MSH6*, *PMS2*, and *EPCAM*. MolQ Lynch Syndrome panel includes next-generation sequencing of these genes for mutations and large deletions/duplications.

Patient Demographic

Name: Mr. Naresh Chadha
Sex: Male
Date of Birth/Age: 55 years
Disease: Healthy Individual

Clinician

Clinician Name: Dr Amit Verma
Medical Facility: Max Hospital
Pathologist: Not Provided

Specimen

Booking ID: 011906270369
Site: NA
Sample Type: Blood
Date of Collection: 27-06-2019
Date of Booking: 27-06-2019

CLINICAL SYNOPSIS

The index patient, Ms. Seema Sachdeva is a case of moderately differentiated endometrioid adenocarcinoma. She was found to harbor a heterozygous variation in *MLH1* gene. Her sibling is being evaluated for the same variation.

RESULTS

The same likely pathogenic variation was detected in heterozygous condition in the asymptomatic sibling of the index patient, Mr. Naresh Chadha.

Gene	Location	Variation reported in the index patient	Zygoty	Clinical condition of family member	Classification	Variation reported in family member
<i>MLH1</i> (ENST00000231790.2) ¹	Exon 3	Chr3:37042544G>T (HET); c.306G>T (p.Glu102Asp)	Heterozygous	Asymptomatic	Likely Pathogenic	Present

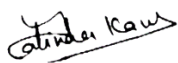
CLINICAL CORRELATION AND VARIANT INTERPRETATION

Variant description: A heterozygous missense variation in exon 3 of the *MLH1* gene (chr3:37042544G>T; c.306G>T) that results in the amino acid substitution of Aspartic Acid for Glutamic Acid at codon 102 (p.Glu102Asp) was detected in the index patient, Ms. Seema Sachdeva (Sample ID: 218474) by NGS and was further validated by Sanger sequencing.

The same likely pathogenic variation was detected in heterozygous condition in the asymptomatic sibling of the index patient, Mr. Naresh Chadha (Figure 1). Incomplete penetrance and variable age of cancer development has been reported for Lynch Syndrome².

RECOMMENDATIONS

Careful correlation with clinical and investigational findings is recommended for the sibling of index case.



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