UKAS MEDICAL 9059

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MLH1 Promoter Hypermethylation Status Report

Patient Details		Source Information		Sample Information	
Lab Number:	MP19- XXXX	Requester Ref:		Date Received:	02/01/2019
Surname:	Atient	Surgical No.:	19S 0001 A1	Primary Tumour Site:	Sigmoid Colon
Forename:	Parker	Sample Type:	FFPE Block	Tumour Subtype:	Adenocarcinoma
D.O.B. (D/M/Y)	17/09/1964	Consultant:	Smith	Tissue Sample Site:	Sigmoid Colon
Gender:	Male	Hospital:	Hospital	% Tumour (Whole): (Selected):	>30%

Result

MLH1 Promoter Hypermethylation Detected

Comment:

Evidence of MLH1 promotor hypermethylation was detected in the tumour-derived DNA component of this sample. Within the context of a microsatellite unstable (MSI-H) and/or mismatch repair deficient (dMMR by IHC) primary colorectal tumour, this finding is consistent with localised epigenetic inactivation of MLH1 gene expression. It should be noted that, although these results are indicative of a somatic or acquired process, they do not necessarily exclude Lynch Syndrome in a patient with a strong family history (Amsterdam II Criteria), in which case germline DNA testing may still be considered appropriate.

Approved by:
Signature:
Name: Dr F. Irst **Date:** 08/01/2019
Job Title:
Clinical Scientist ✓
Consultant Histopathologist
BMS (senior)

Checked by:
Signature:
Name: Dr S. Econd **Date:** 08/01/2019
Job Title:
BMS
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Molecular Biology, PhD ✓

This assay was performed using paired tumour and normal FFPE tissue derived DNA, which was bisulphite converted using the EpiTect Fast Bisulfite Kit. The methylation status of the tumour tissue was assessed, relative to the matched normal tissue, by methylation specific high resolution melt curve analysis (MS-HRM), using the MethylDetect MLH1 primer kit and EpiTect HRM PCR master mix on a Rotor-Gene Q 5Plex HRM platform. The MethylDetect MLH1 kit contains primers that amplify a region covering 8 CpG sites in the promotor region proximal to exon 1 of the MLH1 gene (NM_000249.3), along with high-, low-, and non-methylated control materials. MS-HRM data analysis was conducted using the Rotor-Gene Q Software v2.3.1.49.