Inherited Disease Panel
Gene panel CRC*, version 2, 1-1-2019

Leids Universitair Medisch Centrum

HGNC approved gene symbol	Transcript	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x	% covered >50x
*APC***	NM_001127511	611731	360	100	100	100	100
AXIN2	NM_004655.3	604025	132	100	100	100	100
EPCAM	NM_002354, exon 9			MLPA			
GREM1	NM_013372.6, upstream			MLPA			
*MLH1***	NM_000249	120436	144	100	100	100	100
*MSH2***	NM_000251	609309	163	100	100	100	100
*MSH6***	NM_000179	600678	340	100	100	100	100
*MUTYH***	NM_001048171	604933	145	100	100	100	100
NTHL1	NM_002528, exon 2			Sanger sequencing			
*PMS2****	NM_000535	600259	162	100	100	99	98
POLE	NM_006231, exon 9_14	174762		Sanger sequencing			
POLD1	NM_002691, exon 8_12	174761		Sanger sequencing			
*PTEN*****	NM_000314	601728	142	100	100	100	100
STK11	NM_000455	602216	125	100	100	100	97

* Colorectal carcinoma
** Core genes
*** variants apparently in *PMS2* exons 12_15 need further analysis and material to locate the variant to PMS2 or pseudogene PMS2CL.
****although the coverage is good, the analysis is less reliable because of the presence of pseudogenes.
MLPA for *MLH1, MSH2, EPCAM, MSH6, PMS2, APC, MUTYH and GREM1* is included in the CRC panel.