

REQUESTS ONLY BY CLINICAL GENETICISTS

HGNC approved gene symbol	Transcript	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x	% covered >50x
<i>APC</i> **	NM_000038.5	611731	360	100	100	100	100
<i>AXIN2</i>	NM_004655.3	604025	132	100	100	100	100
<i>CHEK2</i>	NM_0007194.3	604373	85	57	57	57	57
<i>MLH1</i> **	NM_000249.3	120436	144	100	100	100	100
<i>MLH3</i>	NM_001040108.1	604395	313	100	100	100	100
<i>MSH2</i> **	NM_000251.2	609309	163	100	100	100	100
<i>MSH6</i> **	NM_000179.2	600678	340	100	100	100	100
<i>MUTYH</i> **	NM_001128425.1	604933	145	100	100	100	100
<i>NTHL1</i>	NM_002528.5, exon 2			Sanger sequencing			
<i>PMS2</i> ***	NM_000535.5	600259	162	100	100	99	98
<i>POLE</i>	NM_006231.2, exon 9_14	174762		Sanger sequencing			
<i>POLD1</i>	NM_002691.3, exon 8_12	174761		Sanger sequencing			
<i>PTEN</i> ***	NM_000314.4	601728	142	100	100	100	100
<i>STK11</i>	NM_000455.4	602216	125	100	100	100	97

* Colorectal carcinoma

** Core genes

***although the coverage is good, the analysis is less reliable because of the presence of pseudogene(s)

MLPA for *MLH1*, *MSH2*, *EPCAM*, *MSH6*, *PMS2*, *APC*, *MUTYH* and *GREM1* is included in the CRC panel.

NM veranderd, coverage nog checken!!

