



ACCEL-AMPLICON™ PLUS LYNCH SYNDROME PANEL

The Lynch Syndrome Panel offers comprehensive coverage of 4 clinically-relevant Lynch Syndrome genes. The panel covers over 600 COSMIC and 7,500 ClinVar mutations and generates targeted libraries compatible with Illumina® sequencing platforms.

- Compatibility with wide range of samples
- Data analysis options including Primerclip and Genialis
- Addition of your own custom content

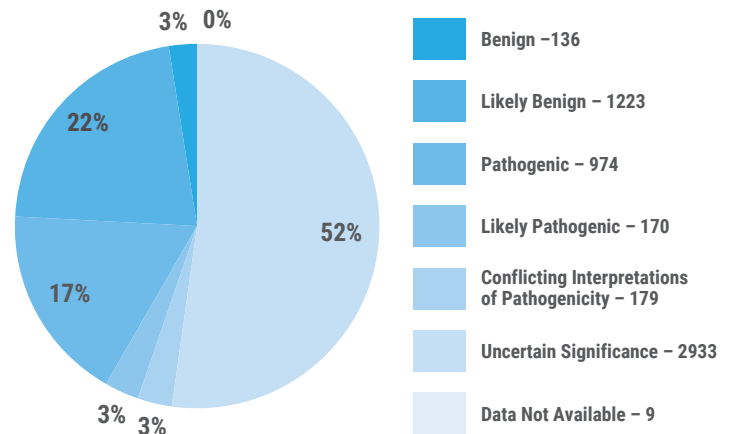


Figure 1. ACMG and AMP-classified variants of clinical significance covered by the Lynch Syndrome Panel

Panel Specifications

Disease Relevance	Lynch Syndrome
Amplicons	180
Average Amplicon Size	140 bp
Number of Genes	4
Gene List	<i>MLH1, MSH2, MSH6, PMS2</i>
Total Target	15 kb
Input Recommended	10 ng amplifiable DNA
Assay Format	Single-tube Multiplex PCR reaction + Dual Indexed Adapters
Time Required	2 hours from DNA to library
Multiplexing on MiSeq v3 at 5000X depth (50M PE reads)	53 samples
Limit of Detection	1% SNV
Sample Compatibility	Cell line, whole blood, cell-free DNA, FFPE

Performance

Achieve Robust Sequencing Performance Over a Wide Range of Samples

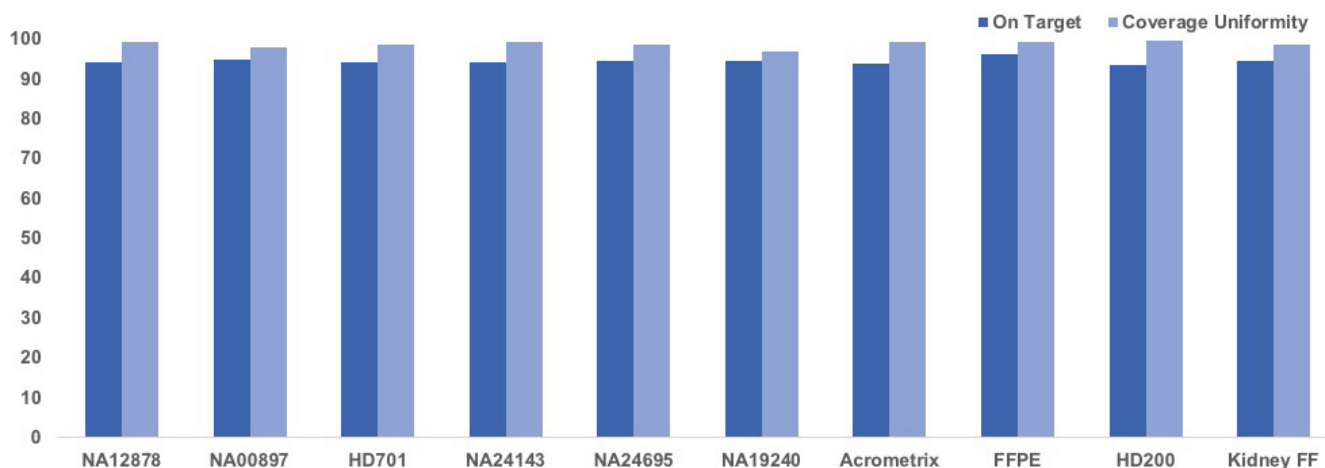


Figure 2. An array of control DNA samples (10 ng input for each) was used to generate libraries with the Accel-Amplicon Plus Lynch Syndrome Panel. The samples include male and female Coriell repository DNAs of different ethnicities, Horizon HD701 quantitative multiplex reference standard, the Acrometrix™ Oncology Hotspot Control, and three formalin-compromised samples, including Horizon HD200 FFPE. Libraries were sequenced on an Illumina MiniSeq instrument and the on target aligned reads and coverage uniformity percentages were plotted.

Detect Low Frequency Variants Accurately from Formalin-Compromised DNA

CHR	POS	Gene	Mutation AA	Mutation Type	Expected Frequency (%)	Average (%)
3	37061883	MLH1	L323M	SNV	10	8.0

Figure 3. The Accel-Amplicon Plus Lynch Syndrome Panel consistently detected the validated MLH1 variant at the expected frequency in replicates from 10 ng of the Horizon Diagnostics Quantitative Multiplex DNA Reference Standard HD200. Variants were called by LoFreq (Genome Institute of Singapore).

Detect Germline and Low Frequency Variants in Key Lynch Syndrome Genes

CHR	POS	Gene	Mutation AA	Mutation Type	Expected Frequency (%)	Average Detected Frequency (%)
2	48030632	MSH6	p.P1082P	SNV	5-15	12.4
2	48030639	MSH6	p.F1088fs*2	DEL	5-15	11.3
2	48030686	MSH6	p.T1100T	SNV	5-15	7.6
2	48030838	MSH6	p.(=)	SNV	50	48.2
3	37067240	MLH1	p.V384D	SNV	5-15	8.6

Figure 4. The Accel-Amplicon Plus Lynch Syndrome Panel consistently detected validated MLH6 variants at the expected frequency from 10 ng of the Oncology Hotspot Control. Variants were called by LoFreq (Genome Institute of Singapore) and GATK HaplotypeCaller (Broad Institute).

Fully customizable. Rapidly create your own panel by building from our pre-designed Accel-Amplicon Plus panels. You can add any target gene including our pre-validated primers or novel targets. We will design, pool and validate your assay for you.



Swift Biosciences, Inc.

674 S. Wagner Road • Ann Arbor, MI 48103 • 734.330.2568 • www.swiftbiosci.com

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