



NGS-A 2019 Next Generation Sequencing

INSTITUTION: Institute of Immunology and Genetics
Kaiserslautern GE 67655

ATTENTION: Frank Glaab

CAP NUMBER: 8086273-01 Kit# 1

KIT INFORMATION:	Kit ID:	Kit Mailed:	Original Evaluation:	Next Mailing Date:
	32618015	3/4/2019	11/5/2019	9/16/2019

COPIED TO: CAP

IMPORTANT NOTES: Code 42 is an acceptable grade for zygosity and variant description if the intended response for that chromosomal position or interval was no variant detected or the position could not be evaluated.

LEGEND: Exception Reason Codes appearing in this evaluation:
[26] = Educational challenge.
[30] = Scientific committee decision.
[42] = No credit assigned due to absence of response.

Reviewed By

Date

The College of American Pathologists recommends that the result of this interlaboratory comparison not be used as a sole criterion for judging the performance of any individual clinical laboratory.



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EVALUATION
ORIGINAL

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Test Method	Specimen	Your Result	Intended Response	Your Grade
Variant Type 1 - 25				
ACADVL chr17 712413etc	NGS-01	NO VARIANT DETECTED	NO VARIANT DETECTED	Good
ALK chr2 30143499	NGS-01	SNV	SNV	Good
ANO5 chr11 22248829etc	NGS-01	DELETION	DELETION	Good
APC chr5 112164561	NGS-01	SNV	SNV	Good
ATM chr11 108121480	NGS-01	NO VARIANT DETECTED	NO VARIANT DETECTED	Good
ATP7B chr13 52524488	NGS-01	SNV	SNV	Good
BBS1 chr11 66282095	NGS-01	SNV	SNV	Good
Variant Type 26 - 50				
BMPR1A chr10 88635779	NGS-01	SNV	SNV	Good
BRCA2 chr13 32944667	NGS-01	SNV	SNV	Good
CACNA1C chr12 2788879	NGS-01	SNV	SNV	Good
CEP290 chr12 88483128	NGS-01	SNV	SNV	Good
COL4A5 chrX 107850076	NGS-01	SNV	SNV	Good
Variant Type 51 - 75				
DES chr2 220285660etc	NGS-01	SNV	SNV	Good
EEF1A2 chr20 62119717	NGS-01	SNV	SNV	Good
FANCL chr2 58388690etc	NGS-01	SNV	SNV	Good
GAA chr17 78082504	NGS-01	SNV	SNV	Good
GATA2 chr3 128205860	NGS-01	SNV	SNV	Good
Variant Type 76 - 100				
GNAS chr20 57478807	NGS-01	SNV	SNV	Good
IL7R chr5 35871190	NGS-01	SNV	SNV	Good
KCNH2 chr7 150648789	NGS-01	SNV	SNV	Good

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Variant Type 101 - 125				
KIF7 chr15 90191824etc	NGS-01	SNV	SNV	Good
LMNA chr1 156105028	NGS-01	SNV	SNV	Good
MBD5 chr2 149240709	NGS-01	NO VARIANT DETECTED	NO VARIANT DETECTED	Good
MCOLN1 chr19 7593589	NGS-01	SNV	SNV	Good
MECP2 chrX 153297831	NGS-01	NO VARIANT DETECTED	NO VARIANT DETECTED	Good
MRE11A chr11 94212048	NGS-01	SNV	SNV	Good
MTM1 chrX 149787580etc	NGS-01	NO VARIANT DETECTED	NO VARIANT DETECTED	Good
MYH6 chr14 23871909	NGS-01	SNV	SNV	Good
Variant Type 126 - 150				
NAGLU chr17 40689455	NGS-01	SNV	SNV	Good
PCDH19 chrX 99597020	NGS-01	NO VARIANT DETECTED	NO VARIANT DETECTED	Good
PEX6 chr6 42933526	NGS-01	SNV	SNV	Good
PLP1 chrX 103041500	NGS-01	NO VARIANT DETECTED	NO VARIANT DETECTED	Good
POLE chr12 133236000	NGS-01	SNV	SNV	Good
PRSS1 chr7 142460335	NGS-01	SNV		See Note [30]
Variant Type 151 - 175				
PTEN chr10 89653840	NGS-01	NO VARIANT DETECTED	NO VARIANT DETECTED	Good
RB1 chr13 48954330	NGS-01	NO VARIANT DETECTED	NO VARIANT DETECTED	Good
RET chr10 43606687	NGS-01	SNV	SNV	Good
RYR2 chr1 237881770	NGS-01	SNV	SNV	Good
SDHD chr11 111958600	NGS-01	NO VARIANT DETECTED	NO VARIANT DETECTED	Good
SERPINA1 chr14 9484etc	NGS-01	SNV	SNV	Good
SMARCB1 chr22 24167580	NGS-01	NO VARIANT DETECTED	NO VARIANT DETECTED	Good

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Test Method	Specimen	Your Result	Intended Response	Your Grade
Variant Type 176 - 200				
SPRED1 chr15 38614525	NGS-01	SNV	SNV	Good
TAZ chrX 153641860	NGS-01	NO VARIANT DETECTED	NO VARIANT DETECTED	Good
TNNC1 chr3 52485788	NGS-01	NO VARIANT DETECTED	NO VARIANT DETECTED	Good
TRPM4 chr19 49704023	NGS-01	SNV	SNV	Good
TSC2 chr16 2138218	NGS-01	SNV	SNV	Good
VPS13B chr8 1004939etc	NGS-01	SNV	SNV	Good
WAS chrX 48547068	NGS-01	NO VARIANT DETECTED	NO VARIANT DETECTED	Good
WRN chr8 30999122	NGS-01	SNV	SNV	Good
WT1 chr11 32421533	NGS-01	SNV	SNV	Good
Zygosity 1 - 25				
ACADVL chr17 712413etc	NGS-01			See Note [42]
ALK chr2 30143499	NGS-01	HOMOZYGOUS	HOMOZYGOUS	Good
ANO5 chr11 22248829etc	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
APC chr5 112164561	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
ATM chr11 108121480	NGS-01			See Note [42]
ATP7B chr13 52524488	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
BBS1 chr11 66282095	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
Zygosity 26 - 50				
BMP1A chr10 88635779	NGS-01	HOMOZYGOUS	HOMOZYGOUS	Good
BRCA2 chr13 32944667	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
CACNA1C chr12 2788879	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
CEP290 chr12 88483128	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
COL4A5 chrX 107850076	NGS-01	HEMIZYGOUS	HEMIZYGOUS	Good

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Zygosity 51 - 75				
DES chr2 220285660etc	NGS-01	HOMOZYGOUS	HOMOZYGOUS	Good
EEF1A2 chr20 62119717	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
FANCL chr2 58388690etc	NGS-01	HOMOZYGOUS	HOMOZYGOUS	Good
GAA chr17 78082504	NGS-01	HOMOZYGOUS	HOMOZYGOUS	Good
GATA2 chr3 128205860	NGS-01	HOMOZYGOUS	HOMOZYGOUS	Good
Zygosity 76 - 100				
GNAS chr20 57478807	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
IL7R chr5 35871190	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
KCNH2 chr7 150648789	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
Zygosity 101 - 125				
KIF7 chr15 90191824etc	NGS-01	HOMOZYGOUS	HOMOZYGOUS	Good
LMNA chr1 156105028	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
MBD5 chr2 149240709	NGS-01			See Note [42]
MCOLN1 chr19 7593589	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
MECP2 chrX 153297831	NGS-01			See Note [42]
MRE11A chr11 94212048	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
MTM1 chrX 149787580etc	NGS-01			See Note [42]
MYH6 chr14 23871909	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
Zygosity 126 - 150				
NAGLU chr17 40689455	NGS-01	HOMOZYGOUS	HOMOZYGOUS	Good
PCDH19 chrX 99597020	NGS-01			See Note [42]
PEX6 chr6 42933526	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
PLP1 chrX 103041500	NGS-01			See Note [42]
POLE chr12 133236000	NGS-01	HOMOZYGOUS	HOMOZYGOUS	Good
PRSS1 chr7 142460335	NGS-01	HETEROZYGOUS		See Note [30]

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Zygoty 151 - 175				
PTEN chr10 89653840	NGS-01			See Note [42]
RB1 chr13 48954330	NGS-01			See Note [42]
RET chr10 43606687	NGS-01	HOMOZYGOUS	HOMOZYGOUS	Good
RYR2 chr1 237881770	NGS-01	HOMOZYGOUS	HOMOZYGOUS	Good
SDHD chr11 111958600	NGS-01			See Note [42]
SERPINA1 chr14 9484etc	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
SMARCB1 chr22 24167580	NGS-01			See Note [42]
Zygoty 176 - 200				
SPRED1 chr15 38614525	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
TAZ chrX 153641860	NGS-01			See Note [42]
TNNC1 chr3 52485788	NGS-01			See Note [42]
TRPM4 chr19 49704023	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
TSC2 chr16 2138218	NGS-01	HOMOZYGOUS	HOMOZYGOUS	Good
VPS13B chr8 1004939etc	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
WAS chrX 48547068	NGS-01			See Note [42]
WRN chr8 30999122	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
WT1 chr11 32421533	NGS-01	HETEROZYGOUS	HETEROZYGOUS	Good
Variant Desc. 1 - 25				
ACADVL chr17 712413etc	NGS-01			See Note [42]
ALK chr2 30143499	NGS-01	c.27C>G, p.Leu9=		See Note [26]
ANO5 chr11 22248829etc	NGS-01	c.364-8delT		See Note [26]
APC chr5 112164561	NGS-01	c.1635G>A, p.Ala545=		See Note [26]
ATM chr11 108121480	NGS-01			See Note [42]
ATP7B chr13 52524488	NGS-01	c.2495A>G, p.Lys832Arg		See Note [26]
BBS1 chr11 66282095	NGS-01	c.378G>A, p.Leu126=		See Note [26]

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Variant Desc. 26 - 50				
BMPR1A chr10 88635779	NGS-01	c.4C>A, p.Pro2Thr		See Note [26]
BRCA2 chr13 32944667	NGS-01	c.8460A>C, p.Val2820=		See Note [26]
CACNA1C chr12 2788879	NGS-01	c.5361G>A, p.Thr1787=		See Note [26]
CEP290 chr12 88483128	NGS-01	c.3710G>A, p.Arg1237His		See Note [26]
COL4A5 chrX 107850076	NGS-01	c.2349G>A, p.Pro783=		See Note [26]
Variant Desc. 51 - 75				
DES chr2 220285660etc	NGS-01	c.1014G>C, p.Leu338=		See Note [26]
EEF1A2 chr20 62119717	NGS-01	c.1326G>A, p.Glu442=		See Note [26]
FANCL chr2 58388690etc	NGS-01	c.981T>C, p.Ser327=		See Note [26]
GAA chr17 78082504	NGS-01	c.1203G>A, p.Gln401=		See Note [26]
GATA2 chr3 128205860	NGS-01	c.15C>G, p.Pro5=		See Note [26]
Variant Desc. 76 - 100				
GNAS chr20 57478807	NGS-01	c.393C>T, p.Ile131=		See Note [26]
IL7R chr5 35871190	NGS-01	c.412G>A, p.Val138Ile		See Note [26]
KCNH2 chr7 150648789	NGS-01	c.1692A>G, p.Leu564=		See Note [26]
Variant Desc. 101-125				
KIF7 chr15 90191824etc	NGS-01	c.1102A>G, p.Thr368Ala		See Note [26]
LMNA chr1 156105028	NGS-01	c.861T>C, p.Ala287=		See Note [26]
MBD5 chr2 149240709	NGS-01			See Note [42]
MCOLN1 chr19 7593589	NGS-01	c.984C>T, p.Asn328=		See Note [26]
MECP2 chrX 153297831	NGS-01			See Note [42]
MRE11A chr11 94212048	NGS-01	c.403-6G>A		See Note [26]
MTM1 chrX 149787580etc	NGS-01			See Note [42]
MYH6 chr14 23871909	NGS-01	c.999C>T, p.Thr333=		See Note [26]

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Variant Desc. 126-150 NAGLU chr17 40689455 PCDH19 chrX 99597020 PEX6 chr6 42933526 PLP1 chrX 103041500 POLE chr12 133236000 PRSS1 chr7 142460335	NGS-01 NGS-01 NGS-01 NGS-01 NGS-01 NGS-01	c.423T>C, p.Ser141= c.2364G>A, p.Val788= c.3156G>A, p.Thr1052= c.508A>G, p.Lys170Glu		See Note [26] See Note [42] See Note [26] See Note [42] See Note [26] See Note [26]
Variant Desc. 151-175 PTEN chr10 89653840 RB1 chr13 48954330 RET chr10 43606687 RYR2 chr1 237881770 SDHD chr11 111958600 SERPINA1 chr14 9484etc SMARCB1 chr22 24167580	NGS-01 NGS-01 NGS-01 NGS-01 NGS-01 NGS-01 NGS-01	c.1296A>G, p.Ala432= c.10503C>T, p.Thr3501= c.840T>C, p.Asp280=		See Note [42] See Note [42] See Note [26] See Note [26] See Note [42] See Note [26] See Note [42]
Variant Desc. 176-200 SPRED1 chr15 38614525 TAZ chrX 153641860 TNNC1 chr3 52485788 TRPM4 chr19 49704023 TSC2 chr16 2138218 VPS13B chr8 1004939etc WAS chrX 48547068 WRN chr8 30999122 WT1 chr11 32421533	NGS-01 NGS-01 NGS-01 NGS-01 NGS-01 NGS-01 NGS-01 NGS-01 NGS-01	c.291G>A, p.Lys97= c.2934T>C, p.Ile978= c.5161-10A>C c.3744A>G, p.Leu1248= c.3138+6C>T c.1059A>G, p.Gln353=		See Note [26] See Note [42] See Note [42] See Note [26] See Note [26] See Note [26] See Note [26] See Note [42] See Note [26] See Note [26]

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