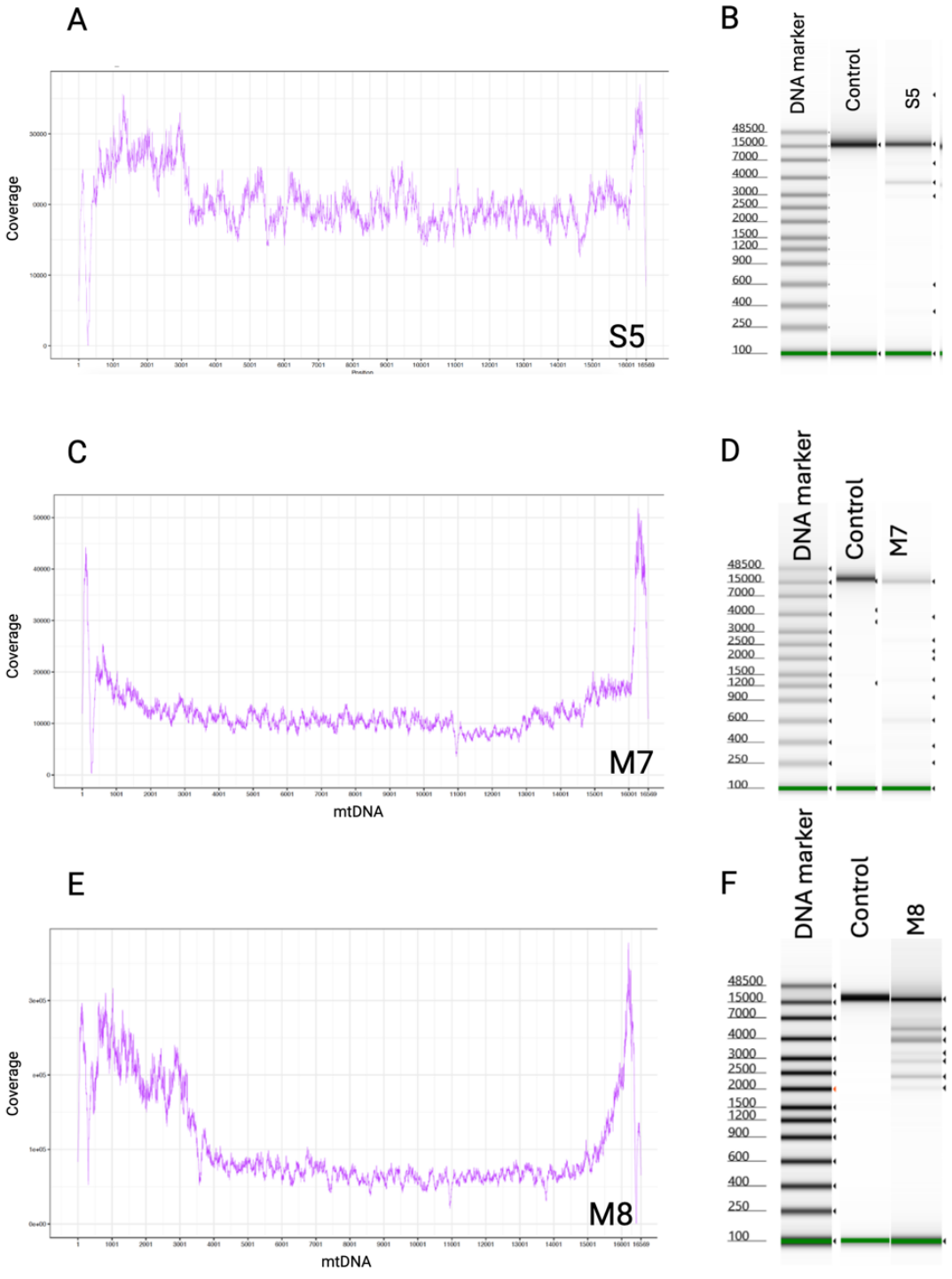


Supplementary Figure 1: MMD detected by long-range PCR and NGS.

The NGS coverage profile showing the NGS read depth (y-axis) across the mtDNA position (x-axis) indicates the presence of large-scale mtDNA deletions (A,C, E). Long-range PCR gel images: The presence of single deletion band (B) and multiple deletion bands supports the detection of SLSMD and MMD by NGS. These gel images have been cropped to remove unrelated samples that were part of the same gel but are not relevant to this experiment. No other edits were made to the gel images.

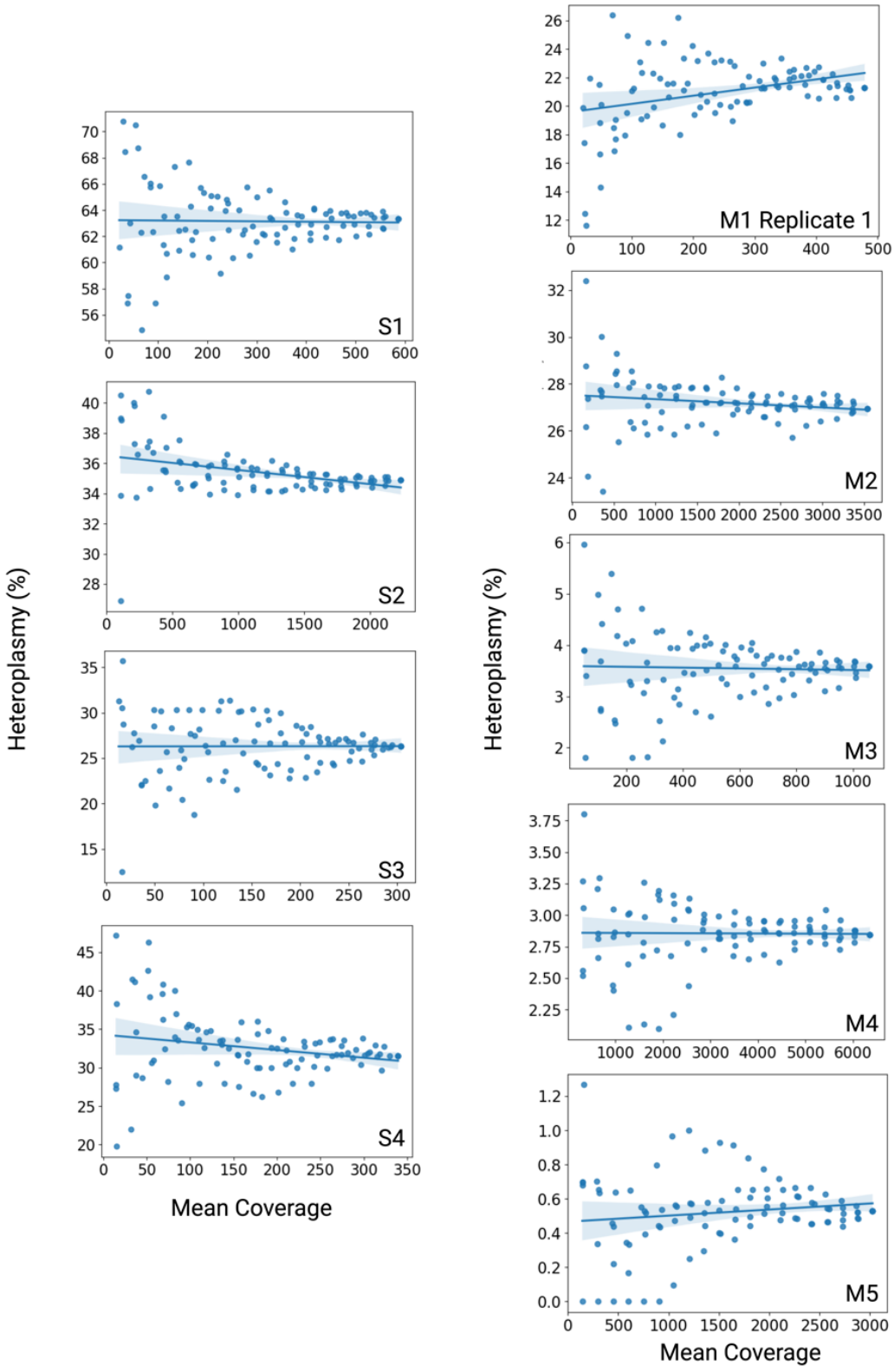
Supplementary Table 1: Reproducibility of SNV detection by LRS

SNVs detected in M1		SNVs detected in M7	
M1 Replicate 1	M1 Replicate 2	M7 Replicate 1	M7 Replicate 2
m.263A>G	m.263A>G	m.263A>G	m.263A>G
m.750A>G	m.750A>G	m.750A>G	m.750A>G
m.1438A>G	m.1438A>G	m.1393G>A	m.1393G>A
m.3992C>T	m.3992C>T	m.1438A>G	m.1438A>G
m.4024A>G	m.4024A>G	m.1719G>A	m.1719G>A
m.4769A>G	m.4769A>G	m.4769A>G	m.4769A>G
m.5004T>C	m.5004T>C	m.4793A>G	m.4793A>G
m.8269G>A	m.8269G>A	m.8860A>G	m.8860A>G
m.8860A>G	m.8860A>G	m.10931T>C	m.10931T>C
m.9123G>A	m.9123G>A	m.15326A>G	m.15326A>G
m.10044A>G	m.10044A>G	m.16261C>T	m.16261C>T
m.14365C>T	m.14365C>T	m.16519T>C	m.16519T>C
m.14582A>G	m.14582A>G		
m.15326A>G	m.15326A>G		

Supplementary Data: SNV and Indel Comparison for All Samples and Heteroplasmy Levels

This spreadsheet contains a comparison of all SNVs and indels called from SRS and LRS workflows described in this study. Variants from LRS were called using LoFreq variant caller. Samples are organized in tabs. “N/A” in columns with SRS data denote

sites where no data was available because the SNVs occurred within the primer set 2 used for LR-PCR amplification.


Heteroplasmy (%)
S1
S2
S3
S4
Mean Coverage
M1 Replicate 1
M2
M3
M4
M5
Heteroplasmy (%)
Mean Coverage

Supplementary Figure 2: Effects of sequencing coverage on deletion heteroplasmy quantification**.**

Datasets of varying mean coverage were simulated for each sample by randomly subsampling the long-read sequencing reads. Deletion heteroplasmy (y-axis) was calculated for each dataset and is plotted here against the mean coverage (x-axis).

Supplementary Table 2: Haplogroup and coverarage analysis

Sample ID	Tissue Type	Haplogroup	Mean nDNA Coverage	Mean mtDNA coverage	Mean Coverage at ND4	Mean Coverage at RNR2
S1	Blood	A2ae	16.33	404.3	219.49	582.03
S2	Muscle	H5c2	1.47	2078.8	1757.85	2330.24
S3	Blood	B4a1a1m1	12.36	265.9	195.81	316.84
S4	Blood	L2c1	16.93	319.3	295.59	344.87
S5	Muscle	H1c3a	0.25	316.8	315.04	307.88
M1 Replicate 1	Muscle	H4a1a	0.22	451.2	411.87	487.52
M1 Replicate 2			0.16	346.2	302.38	414.11
M2	Muscle	K1a4a1a2a	4.80	3266.4	2865.71	3526.36
M3	Muscle	H1as2	1.74	1047.9	1086.67	1104.97
M4	Cardiac Muscle	A2w	0.45	6134.4	6067.22	6299.55
M5	Muscle	H2a2b	1.23	3009.9	3032.07	3032.39
M6	Muscle	J2b1a	2.11	1461.1	1432.25	1549.59

M7 Replicate 1			0.18	343.5	334.76	351.83
M7 Replicate 2	Muscle	H7a1b	0.06	233.8	231.92	252.33
M8	Muscle	T2c1	0.50	822.6	809.07	870.98
Control 1	Muscle	H1c3a	2.22	5058.9	4888.75	5314.47
Control 2	Muscle	T2a1b1a1	6.96	5538.1	5485.23	5925.83
Control 3	Muscle	H5a1g	9.32	19276.7	18428.62	21127.69
Control 4	Blood	A2ah	10.47	183.6	190.96	180.38

Supplementary Table 3: Estimated mtDNA content based on mean coverage of mtDNA and nDNA

	Mean coverage		
Blood Sample	nDNA	mtDNA	mtDNA content
S1	16.33	404.3	49.5
S3	12.36	265.9	43.0
S4	16.93	319.3	37.7
Control 4	10.47	183.6	35.1
Mean	**14.02**	**293.28**	**41.8**

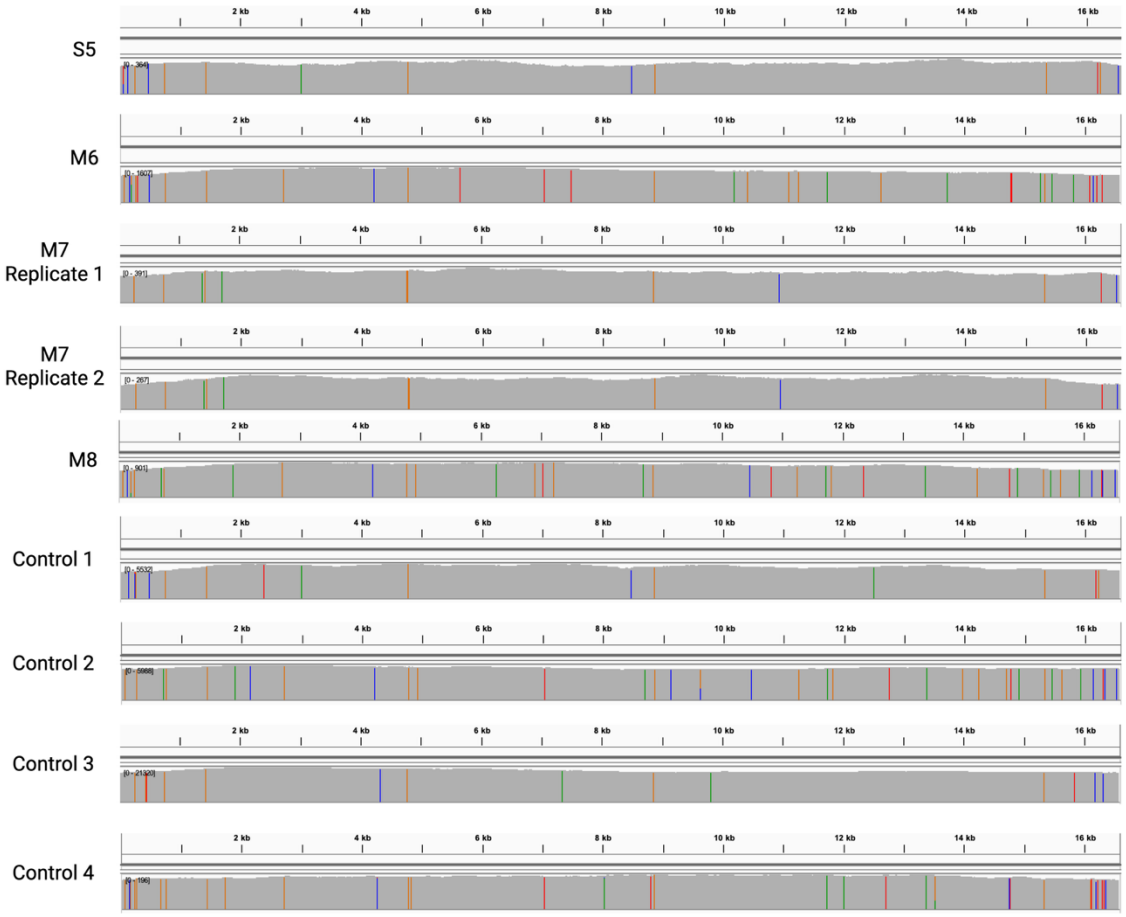
	Mean coverage		
Muscle Sample	nDNA	mtDNA	mtDNA content
S2	1.47	2078.8	2828.3
S5	0.25	316.8	2534.4
M1 Replicate 1	0.22	451.2	4101.8
M1 Replicate 2	0.16	346.2	4327.5
M2	4.8	3266.4	1361.0
M3	1.74	1047.9	1204.5
M4	0.45	6134.4	27264.0

M5	1.23	3009.9	4894.1
M6	2.11	1461.1	1384.9
M7 Replicate 1	0.18	343.5	3816.7
M7 Replicate 2	0.06	233.8	7793.3
M8	0.5	822.6	3290.4
Control 1	2.22	5058.9	4557.6
Control 2	6.96	5538.1	1591.4
Control 3	9.32	19276.7	4136.6
Mean	**2.11**	**3292.42**	**3118.8**

mtDNA content=(mtDNA/nDNA)x2

Supplementary Figure 3: Sequencing coverage for long read sequencing data for samples with 0% heteroplasmy.

Long Read sequencing coverage profiles from IGV for samples with 0% heteroplasmy. All samples with 0% heteroplasmy show relatively even coverage throughout the mitogenome with no sharp dips in coverage.



Supplementary Figure 4: MultiQC report for PacBio LRS data

multiqc

A modular tool to aggregate results from bioinformatics analyses across many samples into a single report.

Report generated on 2025-08-18, 14:27 EDT based on data in:
`/mnt/isilon/rajagopalan_lab/projects/Mito/data/chrMalignments`

Summarize report | Copy report prompt

General Statistics

Copy table | Configure columns | Scatter plot | Violin plot | Export as CSV... | Showing 39/39 rows and 7/17 columns. | Copy Prompt | Summarize table

Sample Name	≥ 30X	Median	Mean Cov.	Dups	GC	Median len	Seqs
23U-GD-00784_chrM.isoseqrealigned_windowsize100	100.0 %	344 X	339.5 X				
Control1_chrM				56.2 %	44.0 %	1 749 bp	0.0 M
Control1_chrM_windowsize100	100.0 %	999 X	5 058.9 X				
Control2_chrM				46.6 %	44.0 %	4 249 bp	0.0 M
Control2_chrM_windowsize100	100.0 %	999 X	5 538.1 X				
Control3_chrM				70.7 %	44.0 %	4 499 bp	0.1 M
Control3_chrM_windowsize100	100.0 %	999 X	19 276.7 X				
Control4_chrM				27.5 %	44.0 %	8 749 bp	0.0 M
Control4_chrM_windowsize100	100.0 %	186 X	183.6 X				
M1_Rep1_chrM				15.1 %	44.0 %	1 749 bp	0.0 M
M1_Rep1_chrM_windowsize100	100.0 %	446 X	445.2 X				
M1_Rep2_chrM				15.0 %	44.0 %	2 749 bp	0.0 M
M1_Rep2_chrM_windowsize100	100.0 %	332 X	346.2 X				
M2_chrM				46.0 %	44.0 %	8 249 bp	0.0 M
M2_chrM_windowsize100	100.0 %	999 X	3 266.4 X				
M3_chrM				27.6 %	44.0 %	4 749 bp	0.0 M
M3_chrM_windowsize100	100.0 %	999 X	1 047.9 X				
M4_chrM				53.9 %	44.0 %	1 749 bp	0.1 M
M4_chrM_windowsize100	100.0 %	999 X	6 134.4 X				
M5_chrM				30.4 %	44.0 %	5 749 bp	0.0 M
M5_chrM_windowsize100	100.0 %	999 X	3 009.9 X				
M6_chrM				28.3 %	44.0 %	5 249 bp	0.0 M
M6_chrM_windowsize100	100.0 %	999 X	1 461.1 X				
M7_Rep1_chrM				15.6 %	44.0 %	1 249 bp	0.0 M
M7_Rep1_chrM_windowsize100	100.0 %	342 X	340.1 X				
M7_Rep2_chrM				12.0 %	44.0 %	1 749 bp	0.0 M
M7_Rep2_chrM_windowsize100	100.0 %	236 X	233.8 X				
M8_chrM				23.7 %	44.0 %	3 749 bp	0.0 M
M8_chrM_windowsize100	100.0 %	844 X	822.6 X				
S1_chrM				41.8 %	44.0 %	9 499 bp	0.0 M
S1_chrM_windowsize100	100.0 %	352 X	404.3 X				
S2_chrM				31.8 %	44.0 %	6 249 bp	0.0 M
S2_chrM_windowsize100	100.0 %	999 X	2 078.8 X				
S3_chrM				24.0 %	44.0 %	4 249 bp	0.0 M
S3_chrM_windowsize100	100.0 %	269 X	265.9 X				
S4_chrM				35.4 %	44.0 %	11 499 bp	0.0 M
S4_chrM_windowsize100	100.0 %	316 X	319.3 X				

Sample Name	≥ 30X	Median	Mean Cov.	Dups	GC	Median len	Seqs
S5_chrM				11.4 %	44.0 %	1 249 bp	0.0 M
S5_chrM_windowsize100	100.0 %	319 X	316.8 X				

Mosdepth

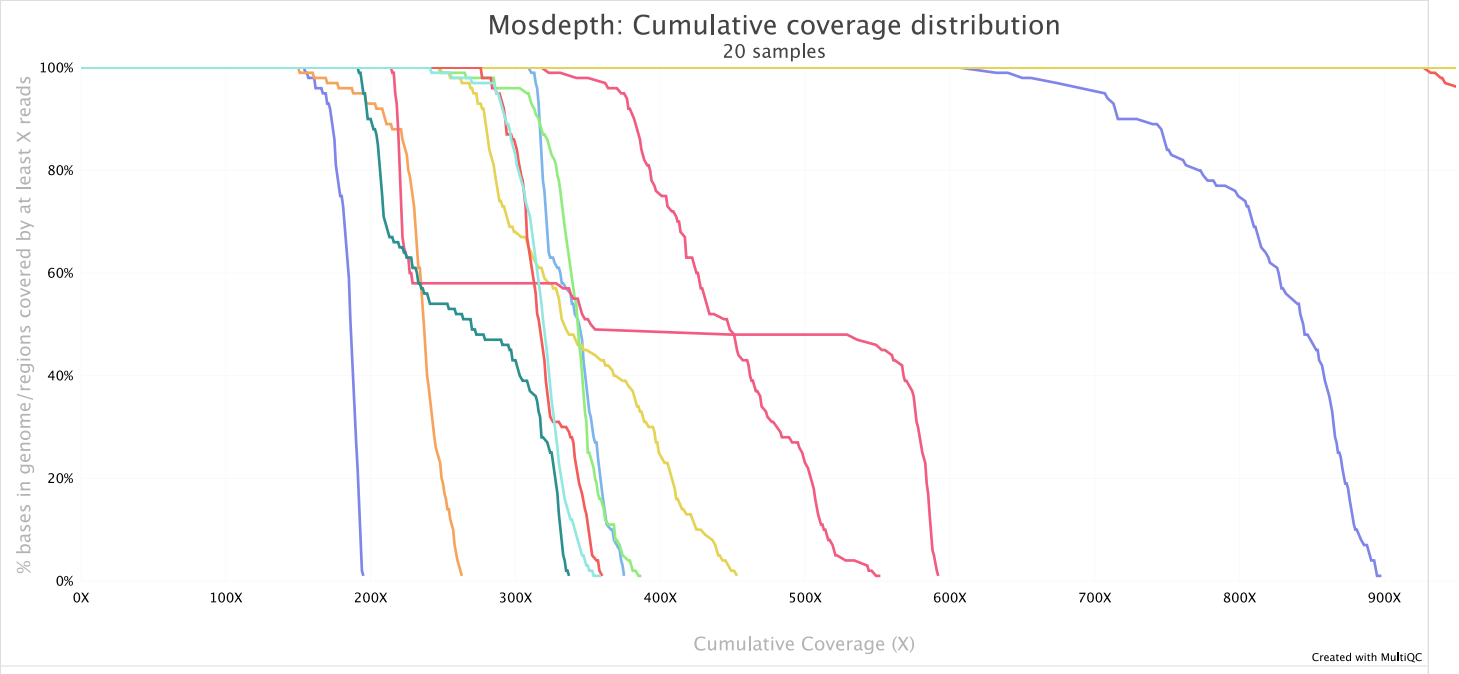
Fast BAM/CRAM depth calculation for WGS, exome, or targeted sequencing. *URL: https://github.com/brentp/mosdepth DOI: 10.1093/bioinformatics/btx699*

Cumulative coverage distribution

Help

Proportion of bases in the reference genome with, at least, a given depth of coverage. Note that for 20 samples, a BED file was provided, so the data was calculated across those regions. For 19 samples, it's calculated across the entire genome length. 19 samples have both global and region reports, and we are showing the data for regions

Copy prompt Summarize plot Export...

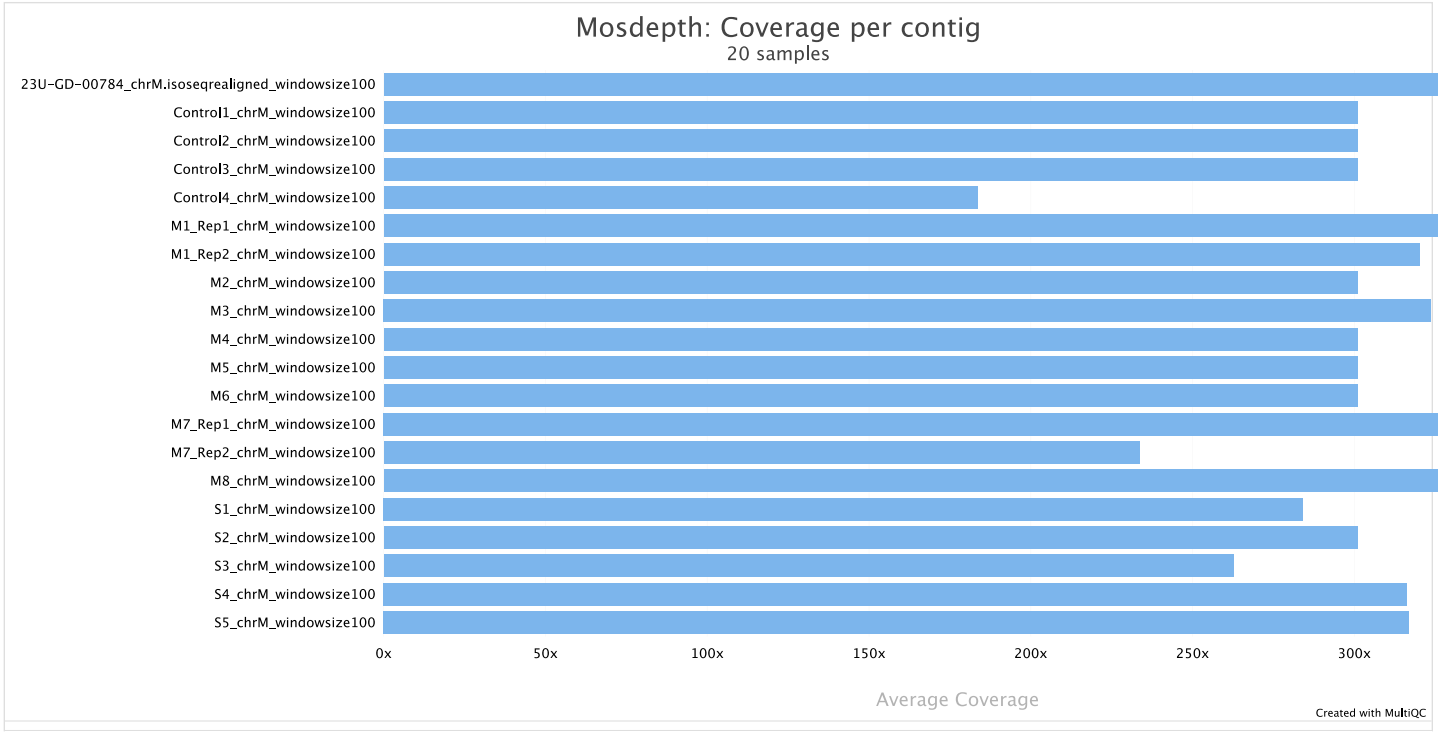


Average coverage per contig

Average coverage per contig or chromosome

Percentages

Copy prompt | Summarize plot | Export...



FastQC *Version:* `0.12.1`

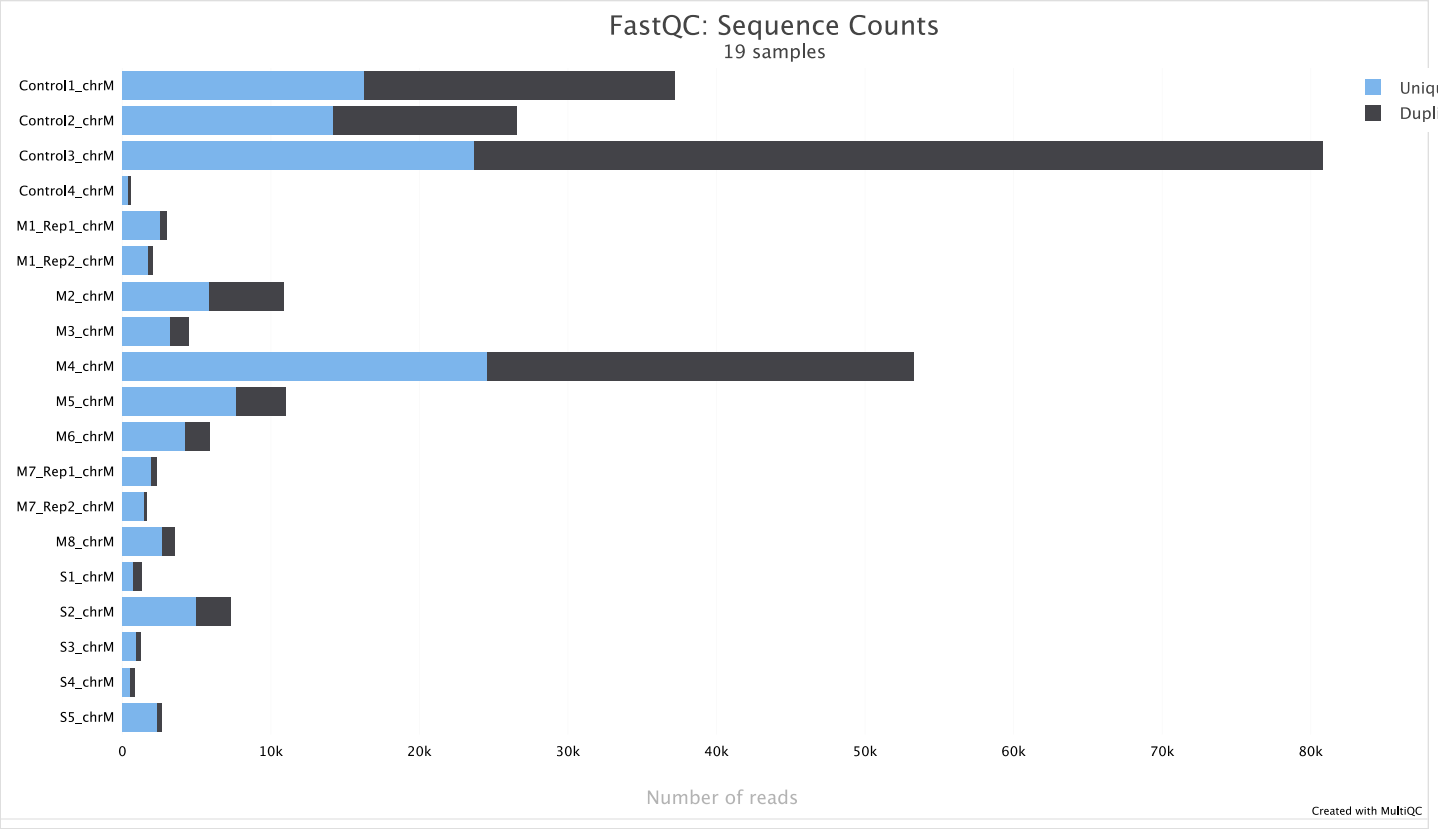
Quality control tool for high throughput sequencing data. *URL: http://www.bioinformatics.babraham.ac.uk/projects/fastqc*

Sequence Counts

Help

Sequence counts for each sample. Duplicate read counts are an estimate only.

Percentages | Copy prompt | Summarize plot | Export...

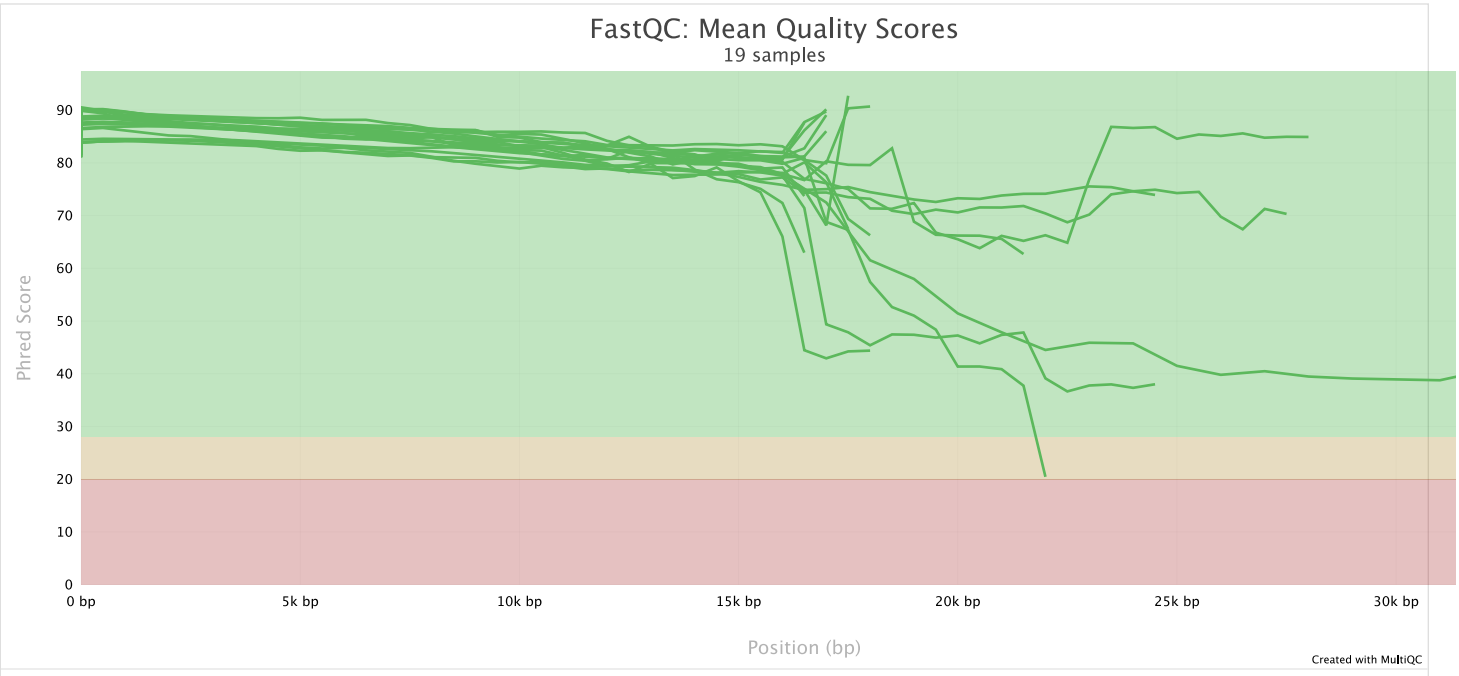


Sequence Quality Histograms 19

Help

The mean quality value across each base position in the read.

Copy prompt | Summarize plot | Export...

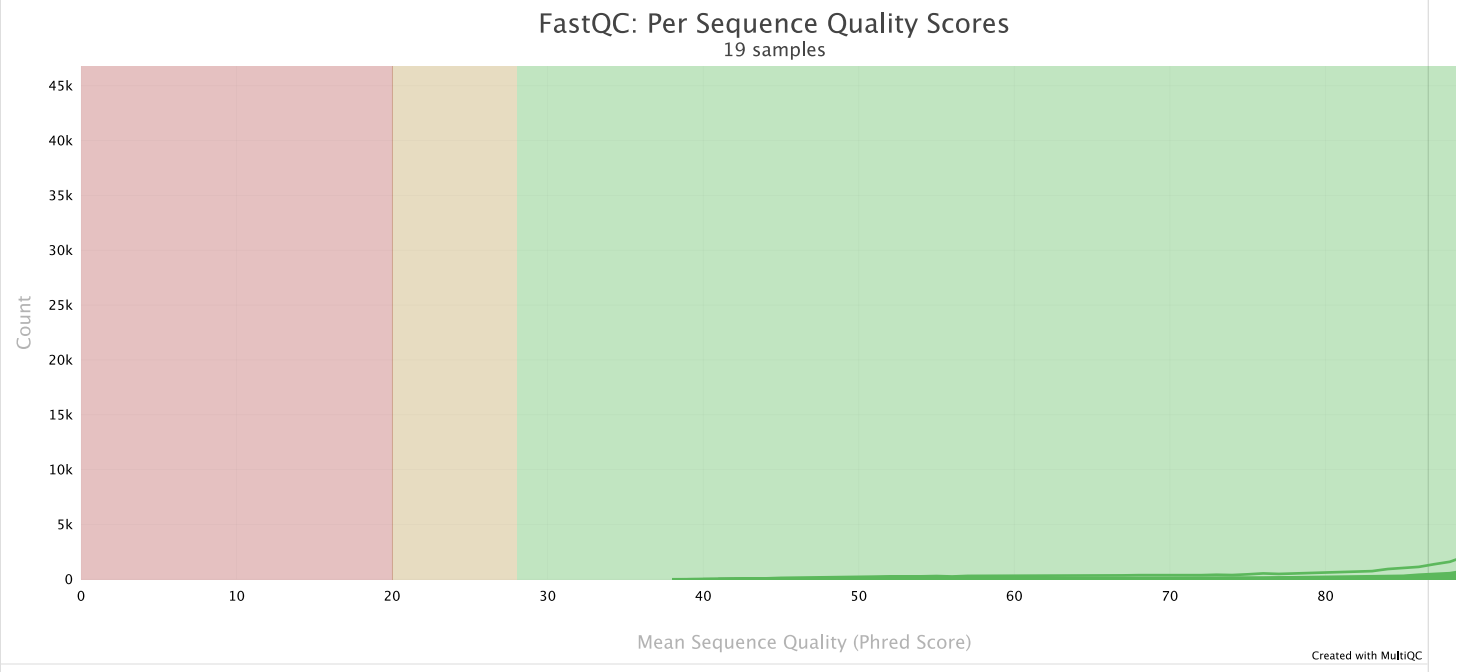


Per Sequence Quality Scores 19

Help

The number of reads with average quality scores. Shows if a subset of reads has poor quality.

Copy prompt | Summarize plot | Export...



Per Base Sequence Content 8

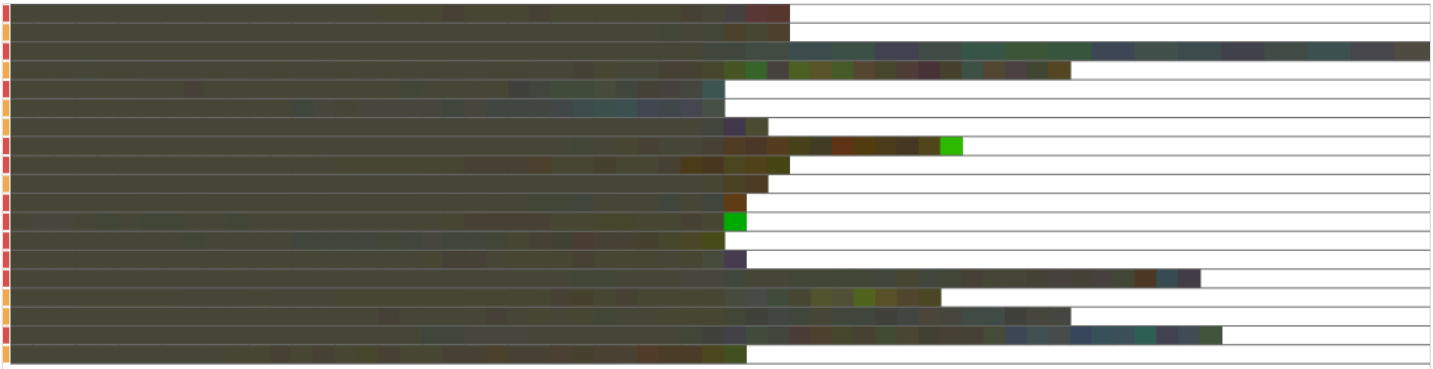
Help

The proportion of each base position for which each of the four normal DNA bases has been called.

Click a sample row to see a line plot for that dataset.

Rollover for sample name

Position: - %T: - %C: - %A: - %G: -



Per Sequence GC Content

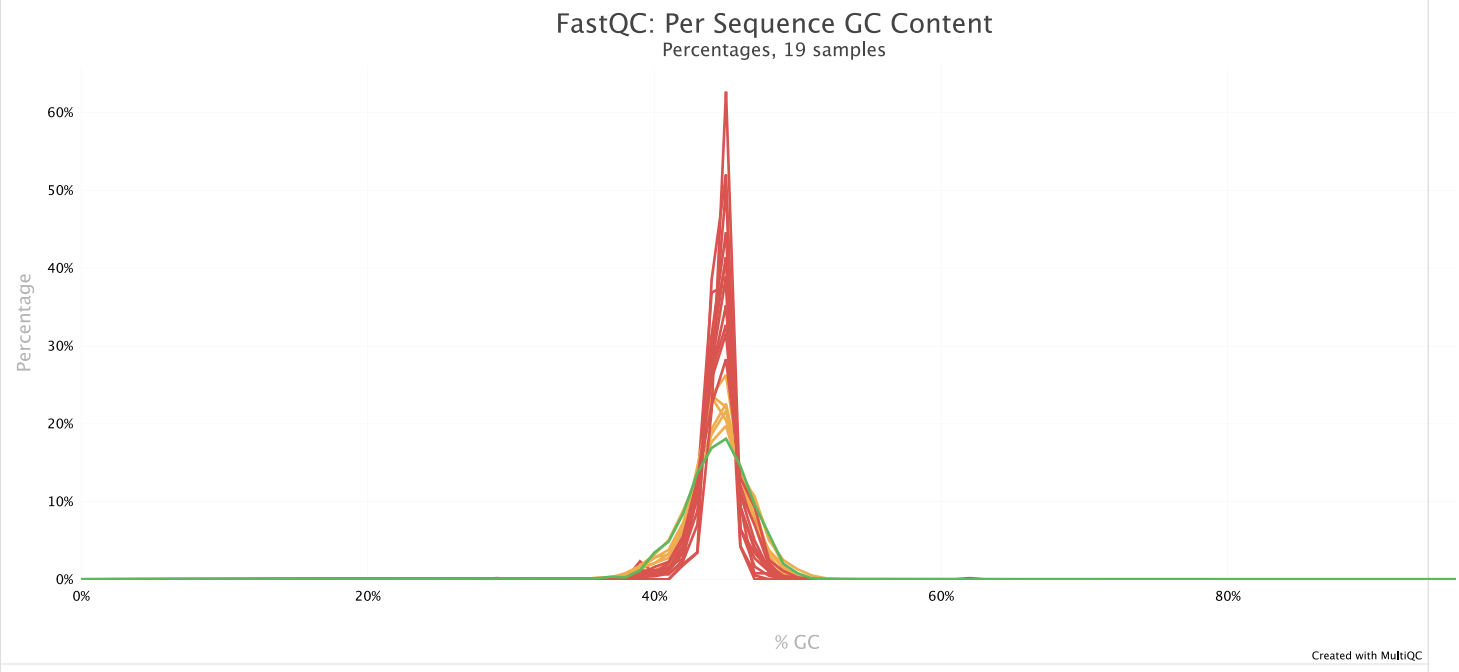
1 6

Help

The average GC content of reads. Normal random library typically have a roughly normal distribution of GC content.

Percentages | Counts

Copy prompt | Summarize plot | Export...



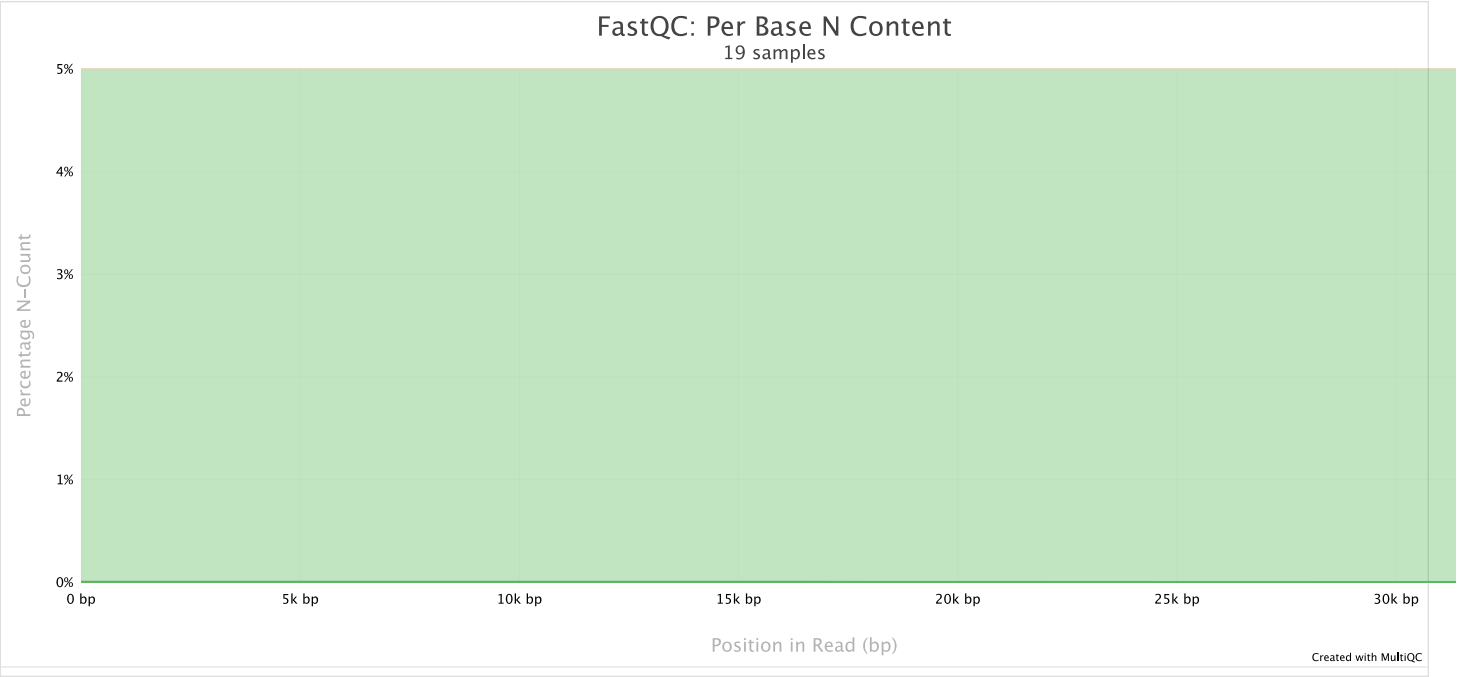
Per Base N Content

19

Help

The percentage of base calls at each position for which an `N` was called.

Copy prompt | Summarize plot | Export...

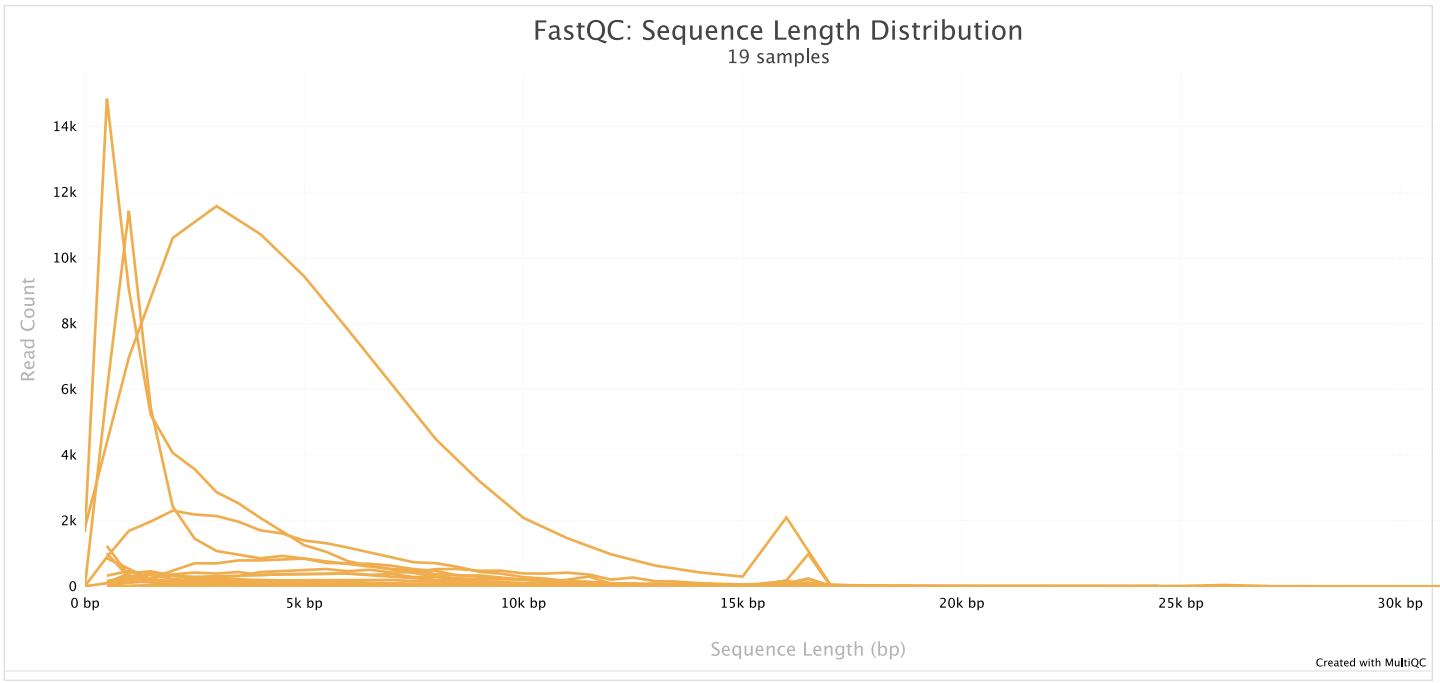


Sequence Length Distribution

19

The distribution of fragment sizes (read lengths) found. See the FastQC help

Copy prompt | Summarize plot | Export...



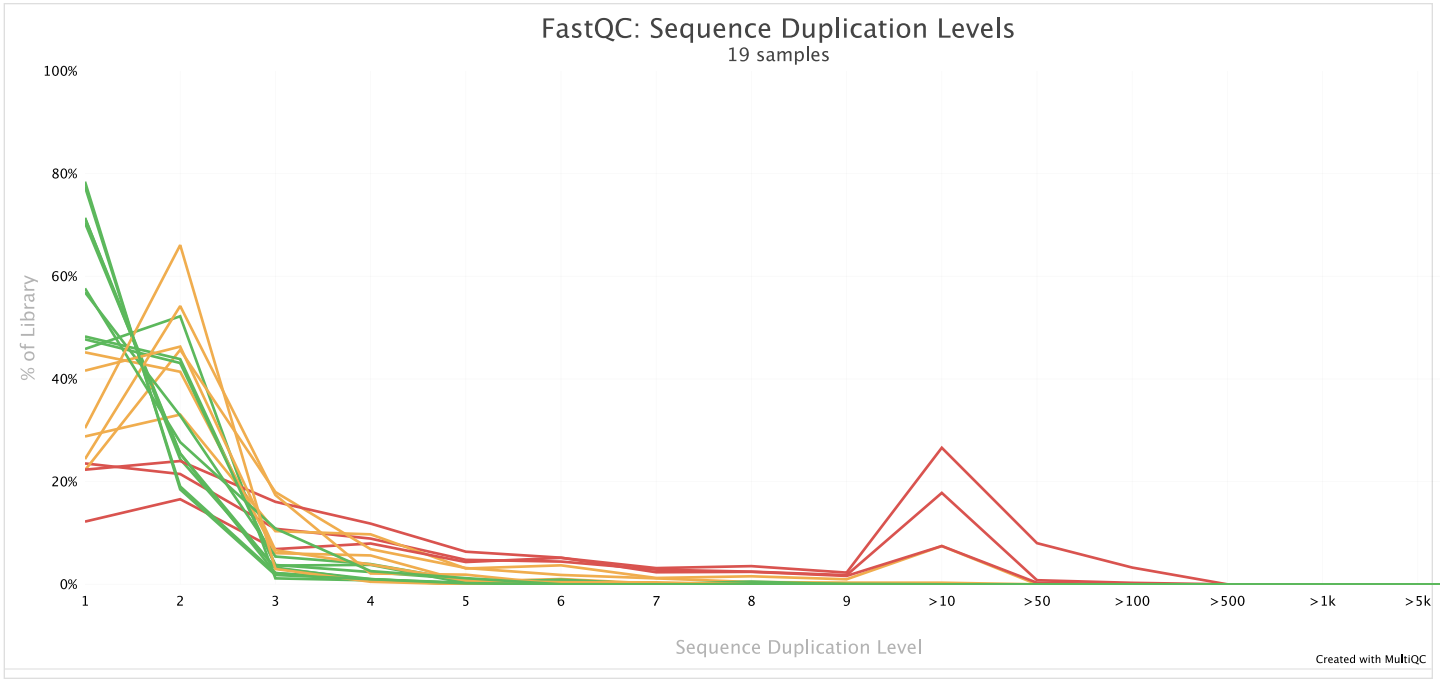
Sequence Duplication Levels

10 6

Help

The relative level of duplication found for every sequence.

Copy prompt | Summarize plot | Export...



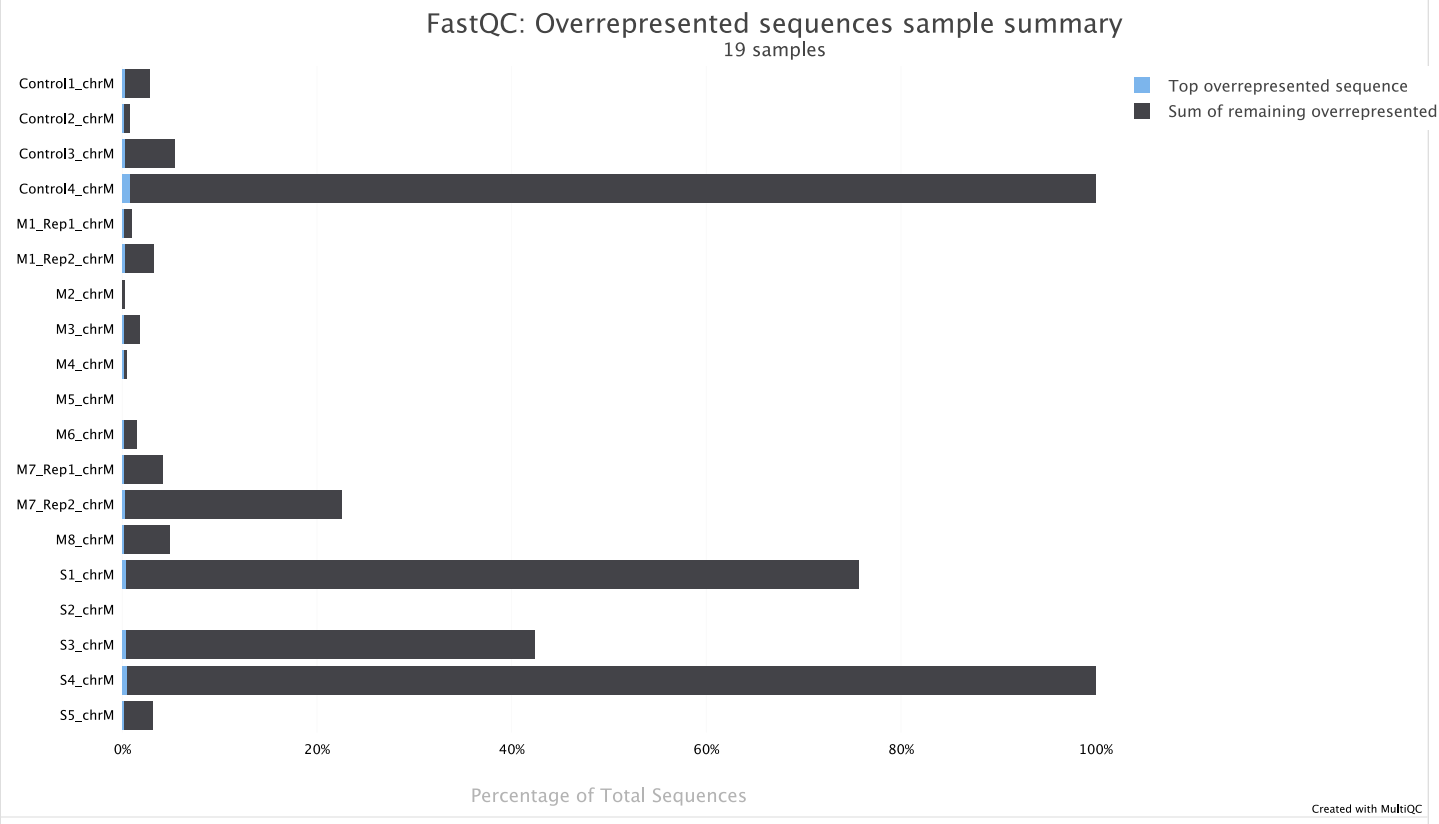
Overrepresented sequences by sample

2 17

Help

The total amount of overrepresented sequences found in each library.

Copy prompt | Summarize plot | Export...



Top overrepresented sequences

Top overrepresented sequences across all samples. The table shows 20 most overrepresented sequences across all samples, ranked by the number of samples they occur in.

Copy table | Configure columns | Scatter plot | Violin plot | Export as CSV... | Showing 20/20 rows and 3/3 columns. | Copy Prompt | Summarize table

Overrepresented sequence	Reports	Occurrences	% of all reads
GGTGCTATAGGGTAAATACGGGCCCTATTTCAAAGATTTTTAGGGGAATT		333	0.1298 %
GGTAGAGGGGGTGCTATAGGGTAAATACGGGCCCTATTTCAAAGATTTTT		299	0.1166 %
GGTGTGGCTAGGCTAAGCGTTTTGAGCTTGCATTGCTGCGTGCTTGATGCT		246	0.0959 %
CCTAGGAATCACCTCCCATTCCGATAAAATCACCTTCCACCCTTACTACA		215	0.0838 %
GGTTTGGTGGAAATTTTTTGTTATGATGTCTGTGTGGAAAGCGGCTGTGC		194	0.0756 %
GGTGTGATAGGTGGCACGGAGAATTTTGGATTCTCAGGGATGGGTTCGAT		186	0.0725 %
GGTGATCTAAAACACTCTTTACGCCGGCTTCTATTGACTTGGGTTAATCG		181	0.0706 %
CCTGAAGCTTCACCGGCGCAGTCATTCTCATAATCGCCCACGGGCTTACA		170	0.0663 %
GGTAGACTGTTCAACCTGTTCCTGCTCCGGCCTCCACTATAGCAGATGCG		164	0.0639 %
GGTGAGGTAAAATGGCTGAGTGAAGCATTGGACTGTAAATCTAAAGACAG		144	0.0561 %
GGTTCATAGTAGAAGAGCGATGGTGAGAGCTAAGGTCGGGGCGGTGATGT		144	0.0561 %
GGTAGGGGCTAGGCTGGAGTGGTAAAAGGCTCAGAAAAATCCTGCGAAGA		129	0.0503 %
GGTGTTAGTCATGTTAGCTTGTTTCAGGTGCGAGATAGTAGTAGGGTCGT		128	0.0499 %
CCTAACAACCCCCCTCCTAATACTAACTACCTGACTCCTACCCCTCACAA		113	0.0441 %
GGTCTTAGCTTTGGCTCTCCTTGCAAAGTTATTTCTAGTTAATTCATTAT		106	0.0413 %
AGTCTCAGCCCTACTCCACTCAAGCACTATAGTTGTAGCAGGAATCTTCT		100	0.0390 %
AAAATACCAAATGCATGGAGAGCTCCCGTGAGTGGTTAATAGGGTGATAG		67	0.0261 %
GGTAAAAGGAGGGCAATTTCTAGATCAAATAATAAGAAGGTAATAGCTAC		53	0.0207 %
GGTGGAGACCTAATTGGGCTGATTTGCCTGCTGCTGCTAGGAGGAGGCCT		50	0.0195 %
CATCCAACATCTCCGCATGATGAAACTTCGGCTCACTCCTTGGCGCCTGC		36	0.0140 %

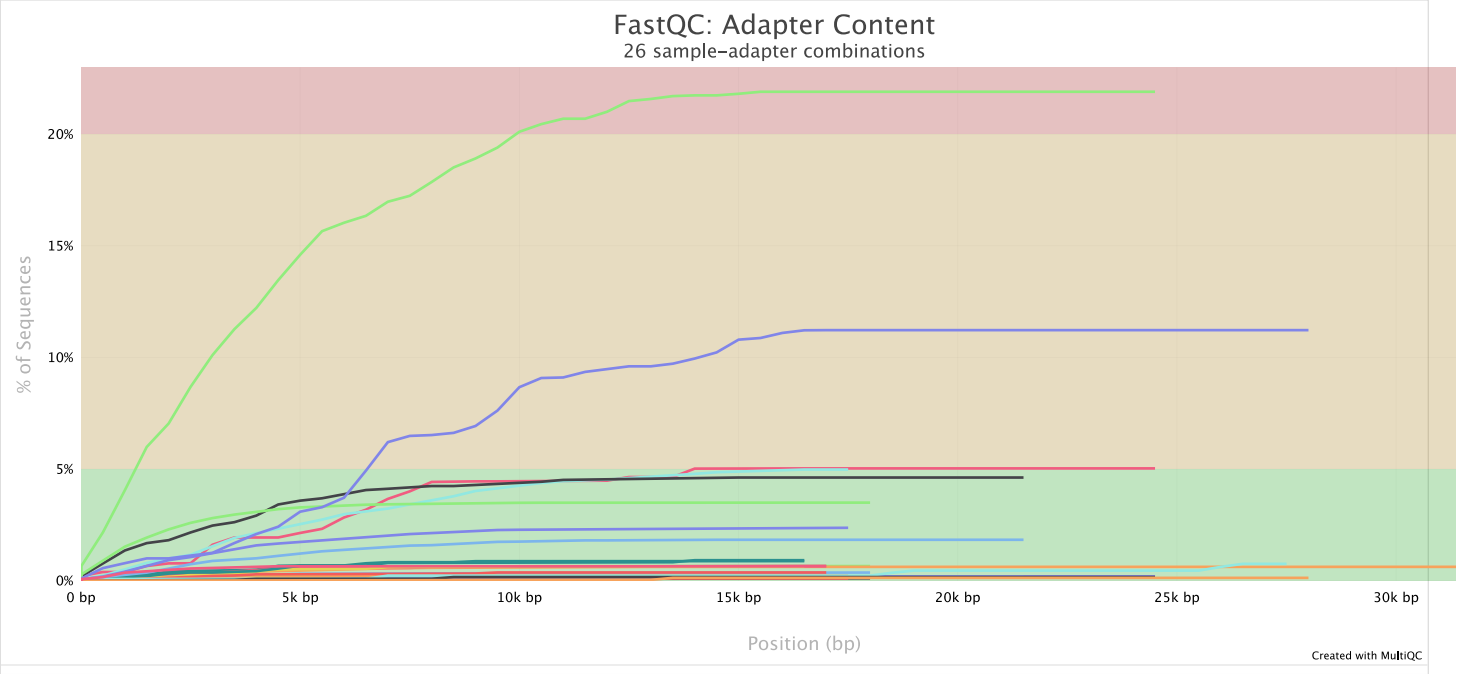
Adapter Content

16 1

Help

The cumulative percentage count of the proportion of your library which has seen each of the adapter sequences at each position.

Copy prompt | Summarize plot | Export...



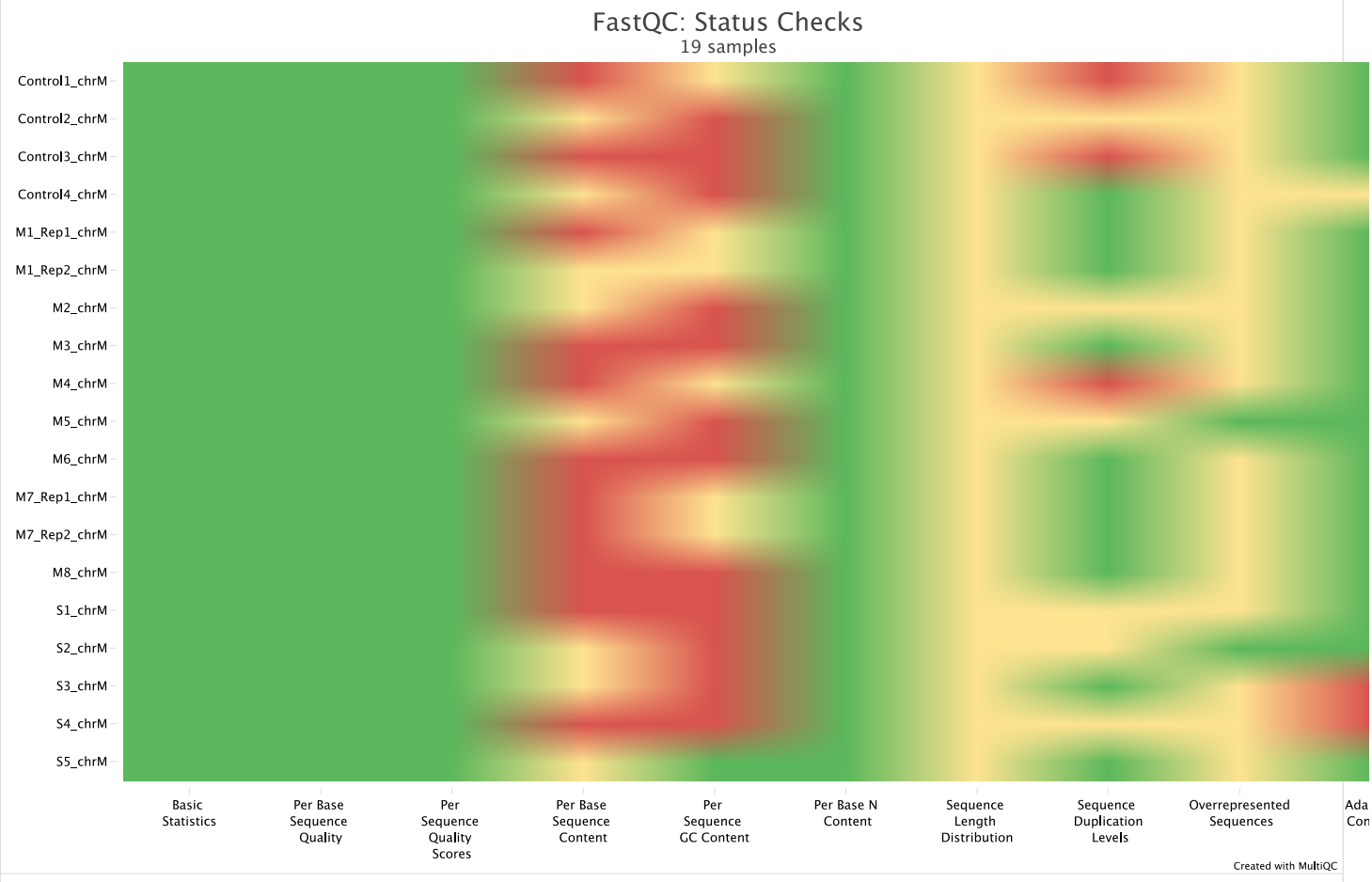
Status Checks

Help

Status for each FastQC section showing whether results seem entirely normal (green), slightly abnormal (orange) or very unusual (red).

Sorted by sample | Clustered

Copy prompt | Summarize plot | Export...



Software Versions

Software Versions lists versions of software tools extracted from file contents.

Copy table

Software	Version
FastQC	0.12.1

Supplementary Figure 5: MultiQC report for SRS data

multiqc

A modular tool to aggregate results from bioinformatics analyses across many samples into a single report.

Report generated on 2025-08-18, 18:53 based on data in:
`/mnt/isilon/rajagopalan_lab/projects/Mito/data/SRS_chrMalignments`

Summarize report | Copy report prompt

General Statistics

Copy table | Configure columns | Scatter plot | Violin plot | Export as CSV... | Showing $^{30}/_{30}$ rows and $^{7}/_{17}$ columns. | Copy Prompt | Summarize table

Sample Name	Dups	GC	Median len	Seqs	≥ 30X	Median	Mean Cov.
Control1_MITO	95.1 %	44.0 %	92 bp	2.2 M			
Control1_MITO_windowsize100					100.0 %	511 X	12 695.1 X
Control2_MITO	95.3 %	44.0 %	122 bp	2.8 M			
Control2_MITO_windowsize100					100.0 %	511 X	19 373.5 X
Control3_MITO	95.2 %	44.0 %	127 bp	3.2 M			
Control3_MITO_windowsize100					100.0 %	511 X	22 281.0 X
Control4_MITO	93.2 %	44.0 %	92 bp	15.5 M			
Control4_MITO_windowsize100					100.0 %	511 X	89 601.2 X
M1_MITO	95.9 %	44.0 %	127 bp	8.9 M	100.0 %	511 X	62 844.7 X
M2_MITO	94.6 %	44.0 %	117 bp	4.4 M			
M2_MITO_windowsize100					100.0 %	511 X	30 003.1 X
M3_MITO	94.9 %	44.0 %	117 bp	1.9 M			
M3_MITO_windowsize100					100.0 %	511 X	12 887.7 X
M4_MITO	93.3 %	44.0 %	142 bp	1.4 M			
M4_MITO_windowsize100					100.0 %	511 X	10 058.0 X
M5_MITO	95.2 %	44.0 %	142 bp	1.7 M			
M5_MITO_windowsize100					100.0 %	511 X	12 873.8 X
M6_MITO	95.0 %	44.0 %	92 bp	2.0 M			
M6_MITO_windowsize100					100.0 %	511 X	11 122.8 X
M7_MITO	93.8 %	44.0 %	112 bp	1.7 M			
M7_MITO_windowsize100					100.0 %	511 X	11 125.2 X
M8_MITO	97.0 %	44.0 %	107 bp	16.6 M			
M8_MITO_windowsize100					100.0 %	511 X	104 954.2 X
S1_MITO	92.5 %	44.0 %	117 bp	5.9 M	100.0 %	511 X	40 940.8 X
S2_MITO	95.6 %	44.0 %	122 bp	2.2 M	100.0 %	511 X	15 434.1 X
S3_MITO	97.3 %	44.0 %	107 bp	11.6 M			
S3_MITO_windowsize100					100.0 %	511 X	74 584.1 X
S4_MITO	96.9 %	44.0 %	107 bp	12.5 M	100.0 %	511 X	80 728.3 X
S5_MITO	92.2 %	44.0 %	127 bp	2.3 M			
S5_MITO_windowsize100					100.0 %	511 X	15 650.3 X

Mosdepth

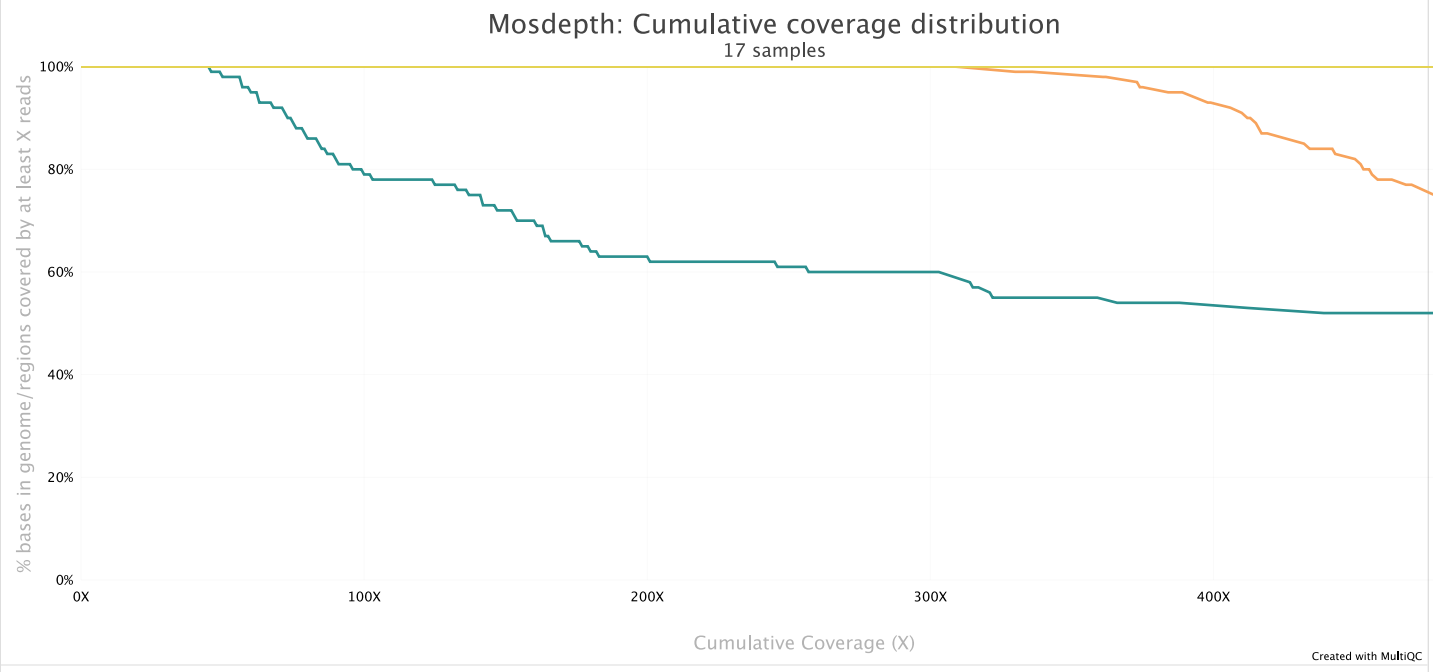
Fast BAM/CRAM depth calculation for WGS, exome, or targeted sequencing. *URL: https://github.com/brentp/mosdepth DOI: 10.1093/bioinformatics/btx699*

Cumulative coverage distribution

Help

Proportion of bases in the reference genome with, at least, a given depth of coverage. Note that for 17 samples, a BED file was provided, so the data was calculated across those regions. For 17 samples, it's calculated across the entire genome length. 17 samples have both global and region reports, and we are showing the data for regions

Copy prompt | Summarize plot | Export...

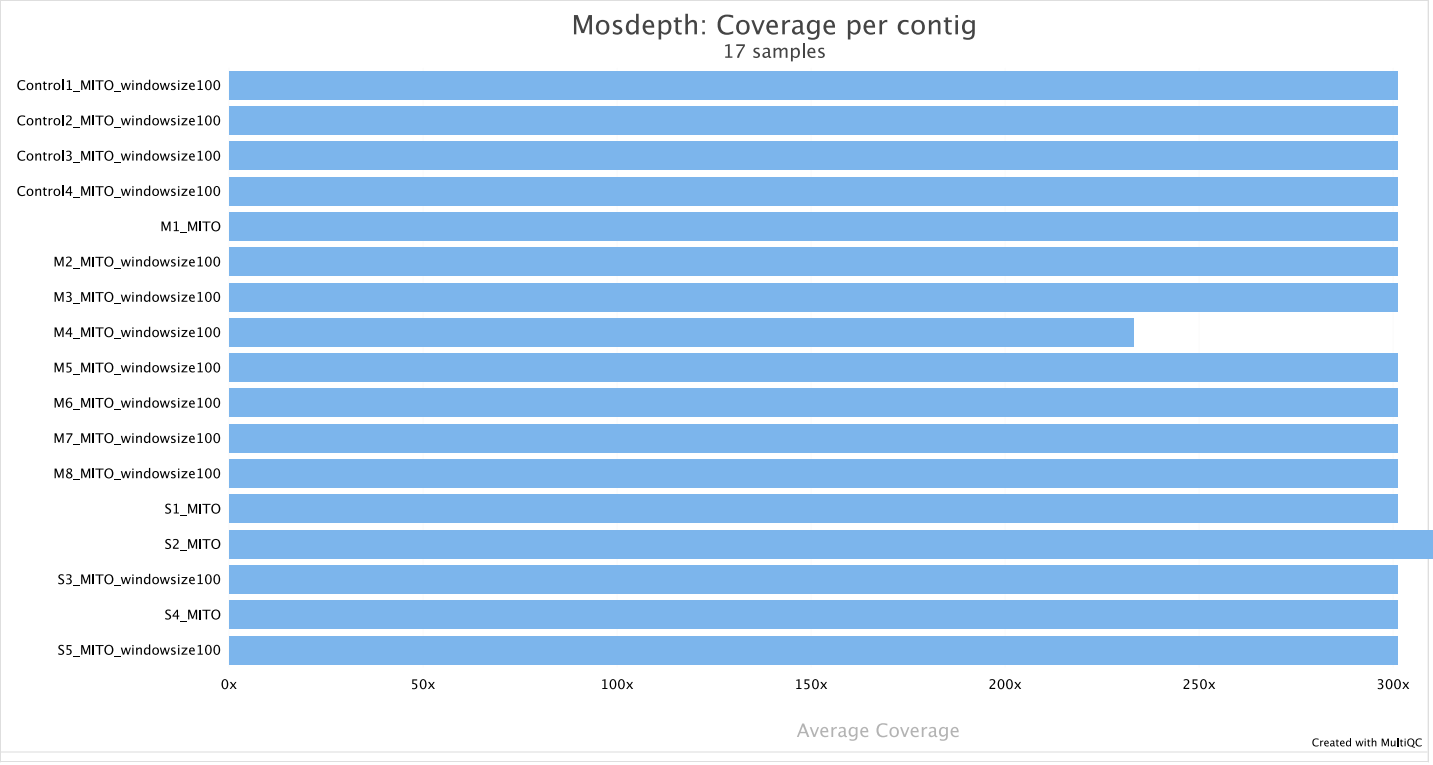


Average coverage per contig

Average coverage per contig or chromosome

Percentages

Copy prompt | Summarize plot | Export...



FastQC *Version:* `0.12.1`

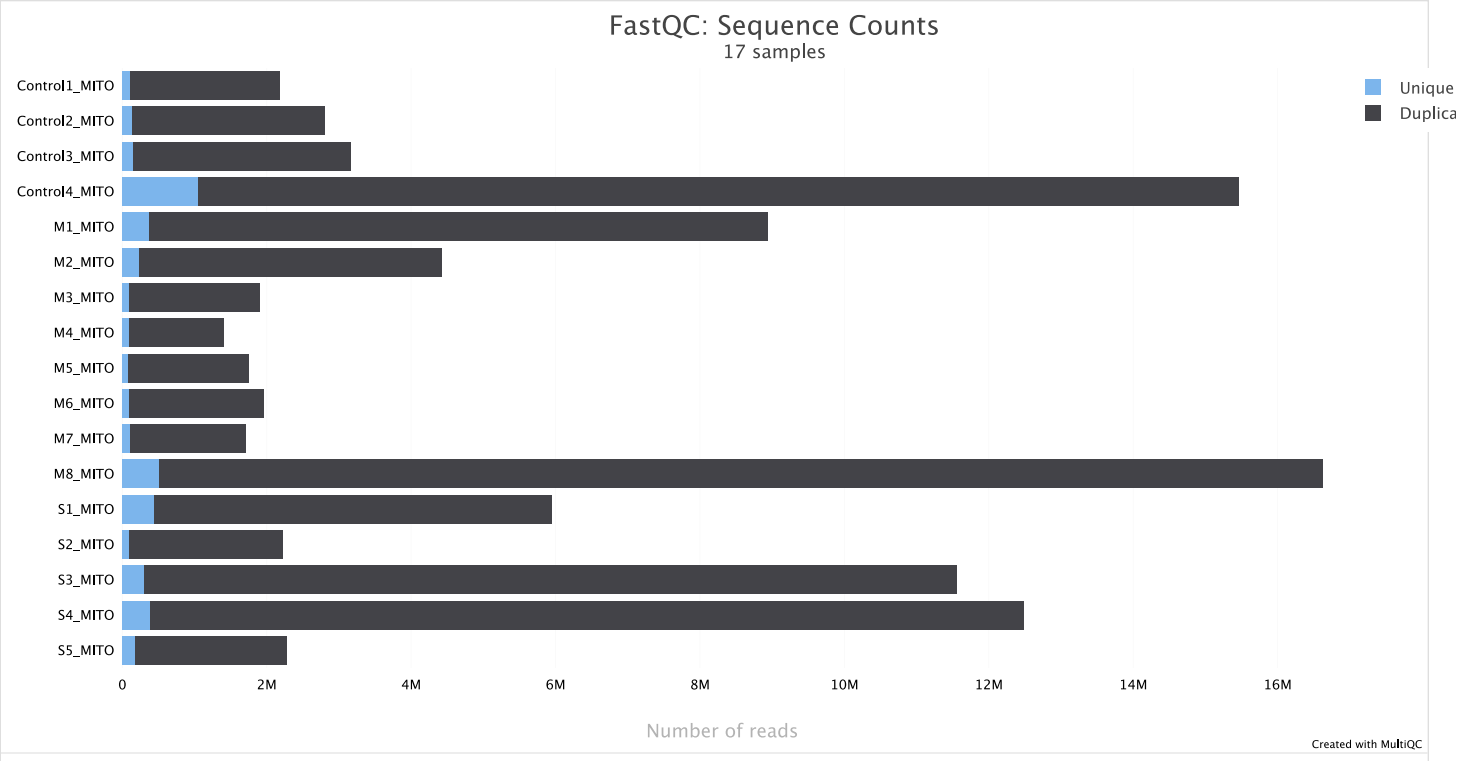
Quality control tool for high throughput sequencing data. *URL: http://www.bioinformatics.babraham.ac.uk/projects/fastqc*

Sequence Counts

Help

Sequence counts for each sample. Duplicate read counts are an estimate only.

Percentages | Copy prompt | Summarize plot | Export...



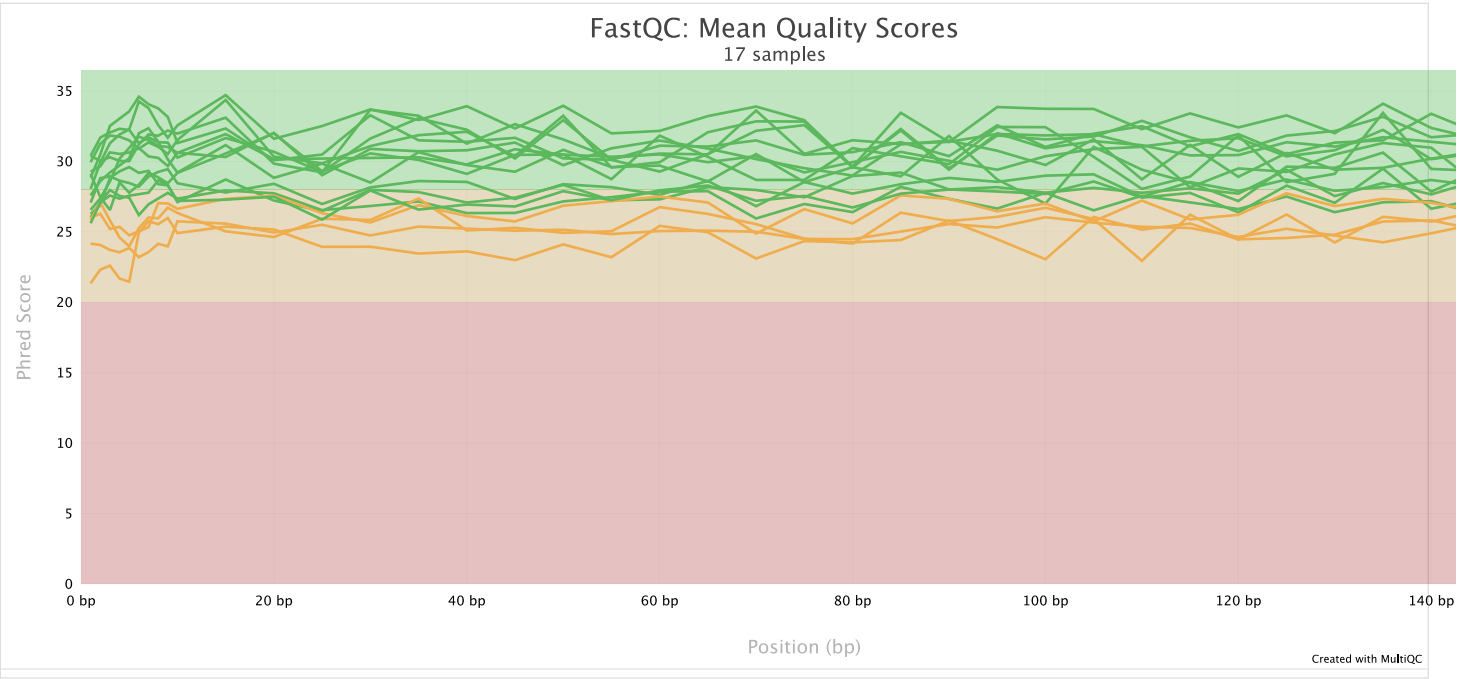
Sequence Quality Histograms

13 4

Help

The mean quality value across each base position in the read.

Copy prompt | Summarize plot | Export...



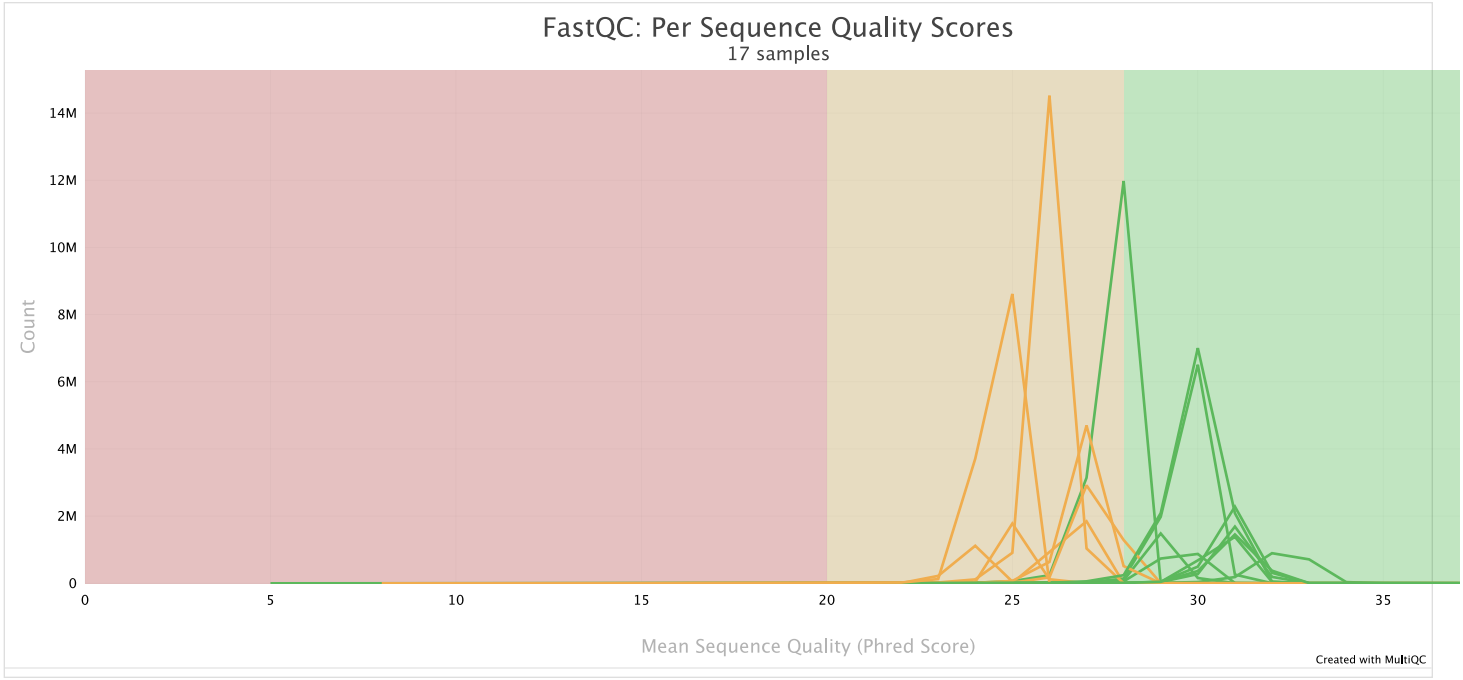
Per Sequence Quality Scores

10 7

Help

The number of reads with average quality scores. Shows if a subset of reads has poor quality.

Copy prompt Summarize plot Export...



Per Base Sequence Content

1

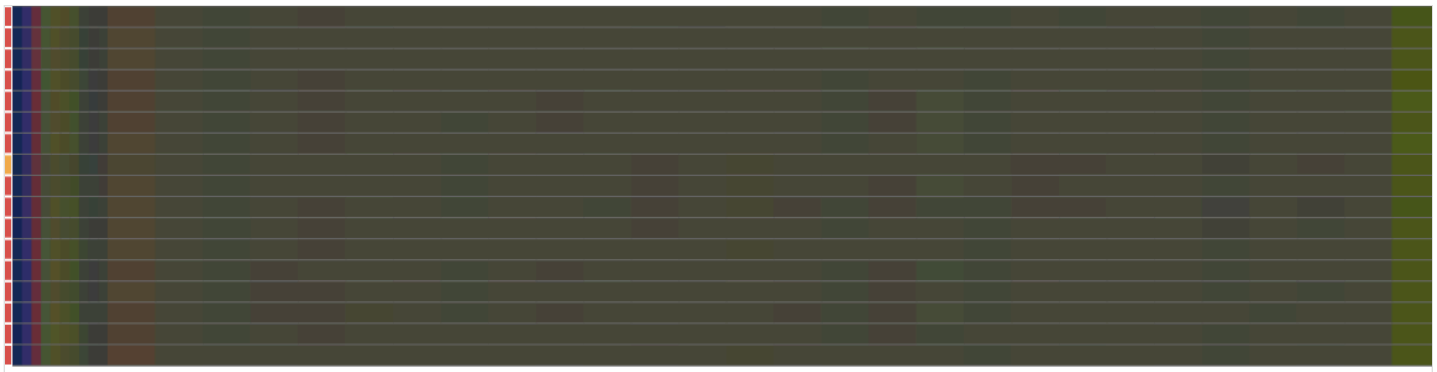
Help

The proportion of each base position for which each of the four normal DNA bases has been called.

Click a sample row to see a line plot for that dataset.

Rollover for sample name

Position: - %T: - %C: - %A: - %G: -



Per Sequence GC Content

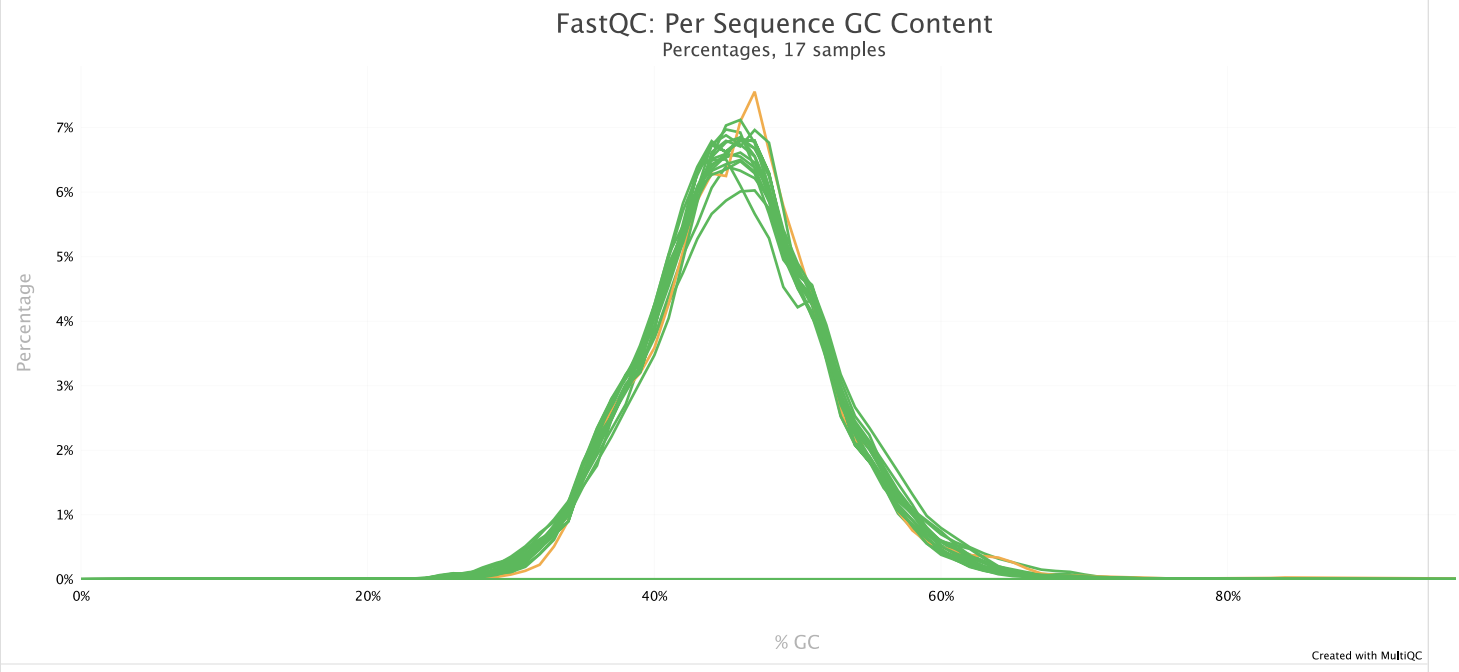
16 1

Help

The average GC content of reads. Normal random library typically have a roughly normal distribution of GC content.

Percentages | Counts

Copy prompt | Summarize plot | Export...



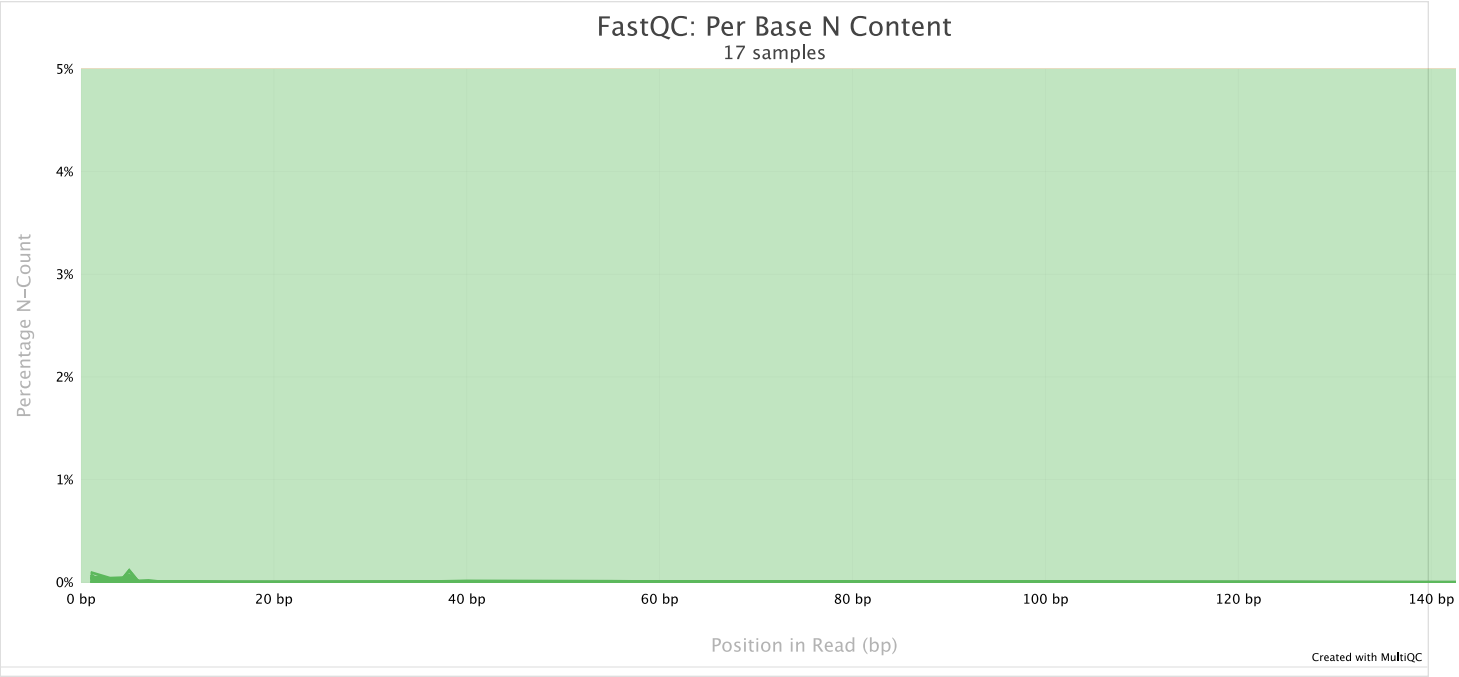
Per Base N Content

17

Help

The percentage of base calls at each position for which an N was called.

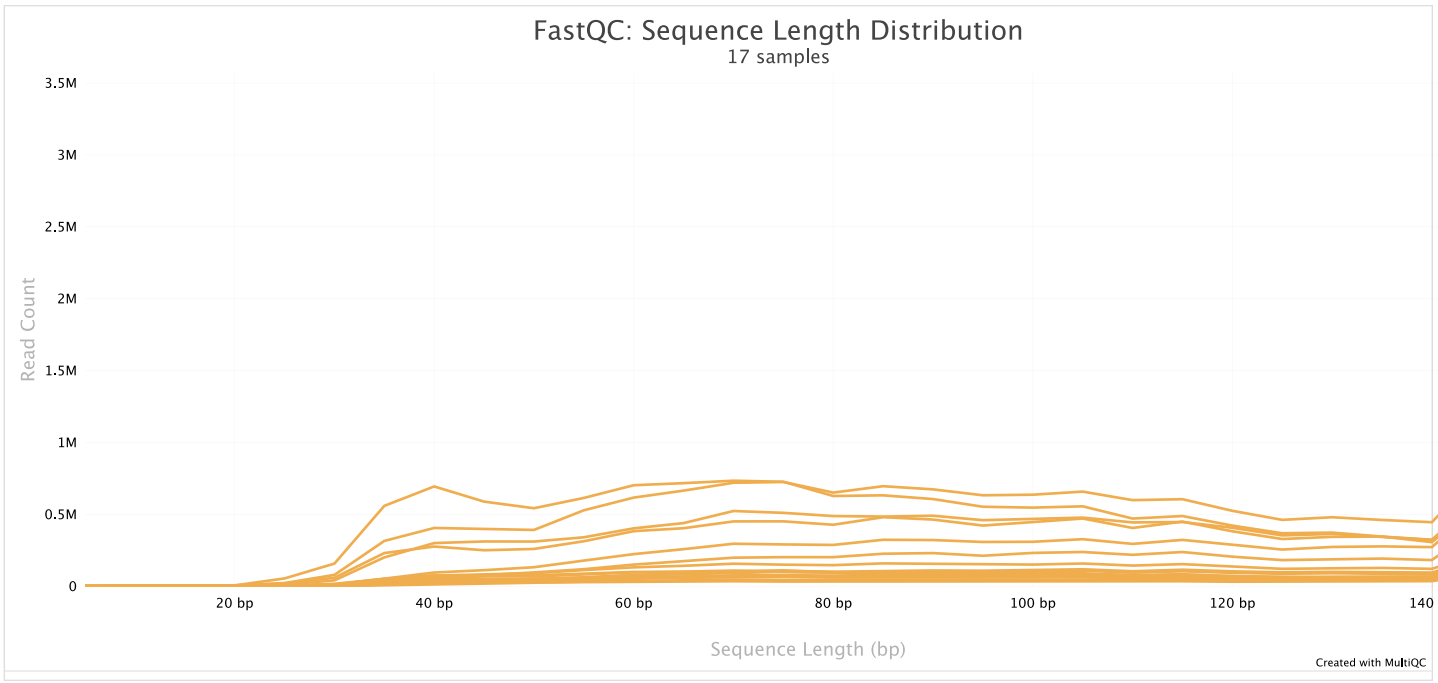
Copy prompt | Summarize plot | Export...



Sequence Length Distribution 17

The distribution of fragment sizes (read lengths) found. See the FastQC help

Copy prompt | Summarize plot | Export...

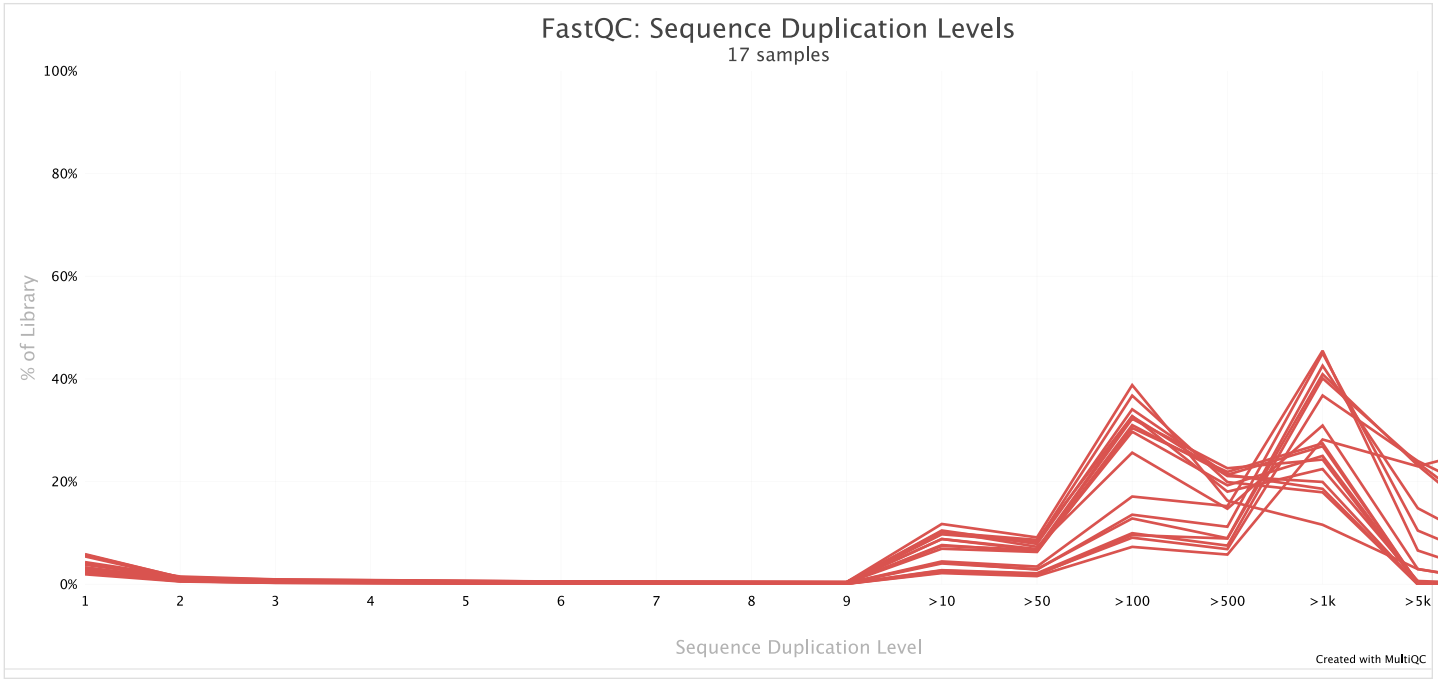


Sequence Duplication Levels

Help

The relative level of duplication found for every sequence.

Copy prompt | Summarize plot | Export...



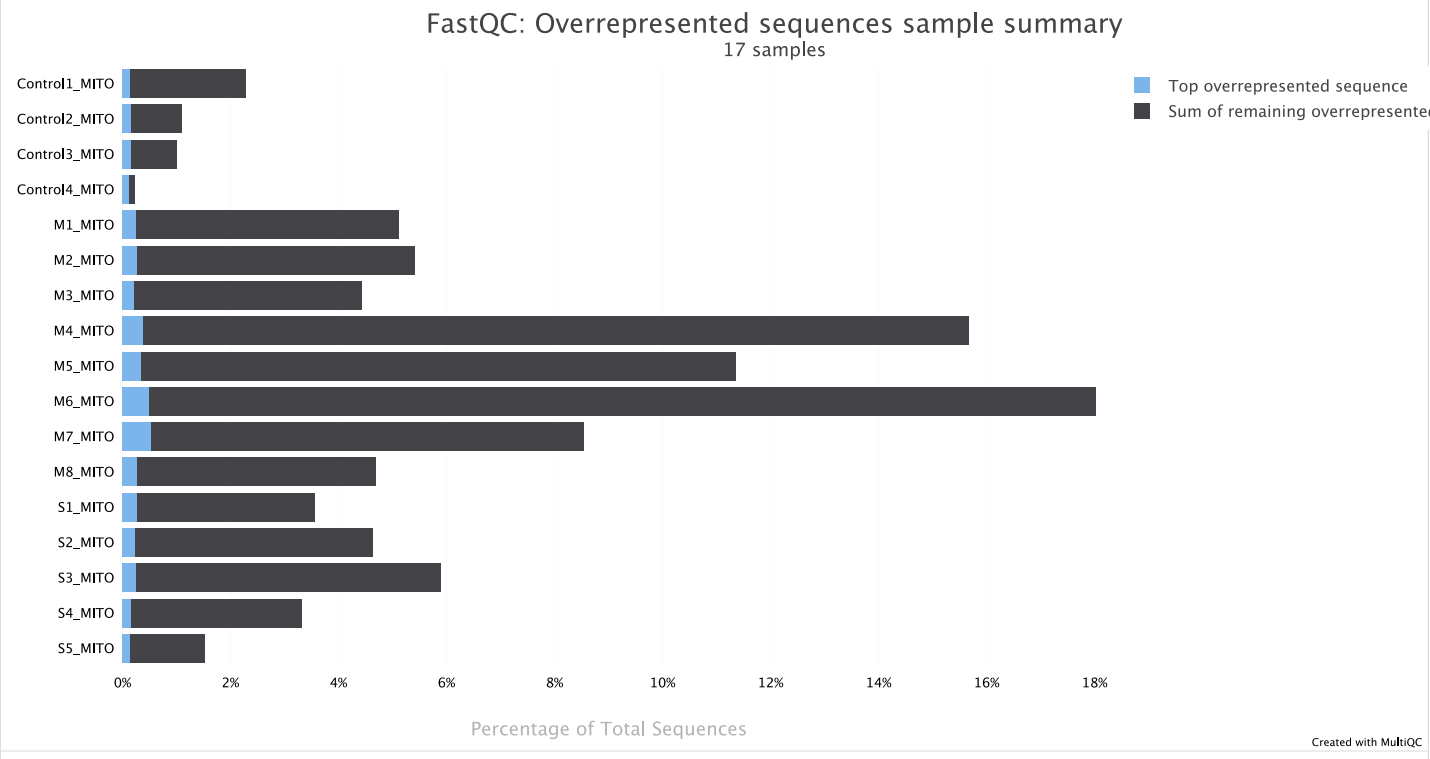
Overrepresented sequences by sample

17

Help

The total amount of overrepresented sequences found in each library.

Copy prompt | Summarize plot | Export...



Top overrepresented sequences

Top overrepresented sequences across all samples. The table shows 20 most overrepresented sequences across all samples, ranked by the number of samples they occur in.

Copy table | Configure columns | Scatter plot | Violin plot | Export as CSV... | Showing $^{24}/_{24}$ rows and $^{3}/_{3}$ columns. | Copy Prompt | Summarize table

Overrepresented sequence	Reports	Occurrences	% of all reads
GTTCTTGGGTGGGTGTGGGTATAATACTAAGTTGAGATGATATCATTTAC		183 782	0.1899 %
CCTTCGACCCTTAAGTTTCATAAGGGCTATCGTAGTTTTCTGGGGTAGAA		166 411	0.1719 %
CTTTCGTAGTCTATTTTGTGTCAACTGGAGTTTTTTACAACTCAGGTGAG		135 846	0.1403 %
CCTCAAAGCAATACACTGAAAATGTTTAGACGGGCTCACATCACCCCATA		131 480	0.1358 %
CCTTTAATAGCGGCTGCACCATCGGGATGTCCTGATCCAACATCGAGGTC		116 053	0.1199 %
CCTGGATTACTCCGGTCTGAACTCAGATCACGTAGGACTTTAATCGTTGA		109 033	0.1126 %
GATGAAGGCTACAAAGTAAGCGCAAGTACCCACGTAAAGACGTTAGGTCA		107 467	0.1110 %
CCTCAAGTATACTTCAAAGGACATTTAACTAAAACCCCTACGCATTTATA		106 687	0.1102 %
CCCTGTACGAAAGGACAAGAGAAATAAGGCCTACTTCACAAAGCGCCTTC		102 042	0.1054 %
GTTTGGGTCTTAGCTATTGTGTGTTCAGATATGTTAAAGCCACTTTCGTA		98 922	0.1022 %
CCAATTAAGAAAGCGTTCAAGCTCAACACCCACTACCTAAAAAATCCCAA		93 843	0.0969 %
CCCAAGAACAGGGTTTGTTAAGATGGCAGAGCCCGGTAATCGCATAAAAC		83 853	0.0866 %
GCTATATTATGCTTGGTTATAATTTTTCATCTTTCCCTTGCGGTACTATA		80 164	0.0828 %
CCTCTAGAGGGATATGAAGCACCGCCAAGGTCCTTTGAGTTTTAAGCTGTG		74 225	0.0767 %
GGTTAGAAGTAGGGTCTTGGTGACAAAATATGTTGTGTAGAGTTCAGGGG		67 787	0.0700 %
CCTCTAGCCTAGCCGTTTACTCAATCCTCCTGATCAGGGTGAGCATCAAAC		67 199	0.0694 %
GTTAGTTGGGGGGTGACTGTTAAAAGTGCCATACCGCCAAAAGATAAAATT		55 531	0.0574 %
GCGTAAAGAGTGTTTTAGATCACCCCCTTCCCCAATAAAGCTAAAACTCAC		53 794	0.0556 %
CCTTAACCTCTACTTCTACCTACGCCTAATCTACTCCACCTCAATCACAC		33 022	0.0341 %
GCTATCACCAGGCTCGGTAGGTTTGTCGCCCTCTACCTATAAATCTTCCCA		29 004	0.0300 %
GATAAAGTATAGGCGATAGAAATTGAAACCTGGCGCAATAGATATAGTAC		9 089	0.1529 %
ACTTCTAACCTCCCTGTTCTTATGAATTCGAACAGCATACCCCCGATTCC		7 189	0.1209 %
CCTATGAAAAAACTTCCTACCACTCACCCTAGCATTACTTATATGATATG		6 854	0.1153 %
AGGGTAACTTGTTCCGTTGGTCAAGTTATTGGATCAATTGAGTATAGTAG		6 594	0.1109 %

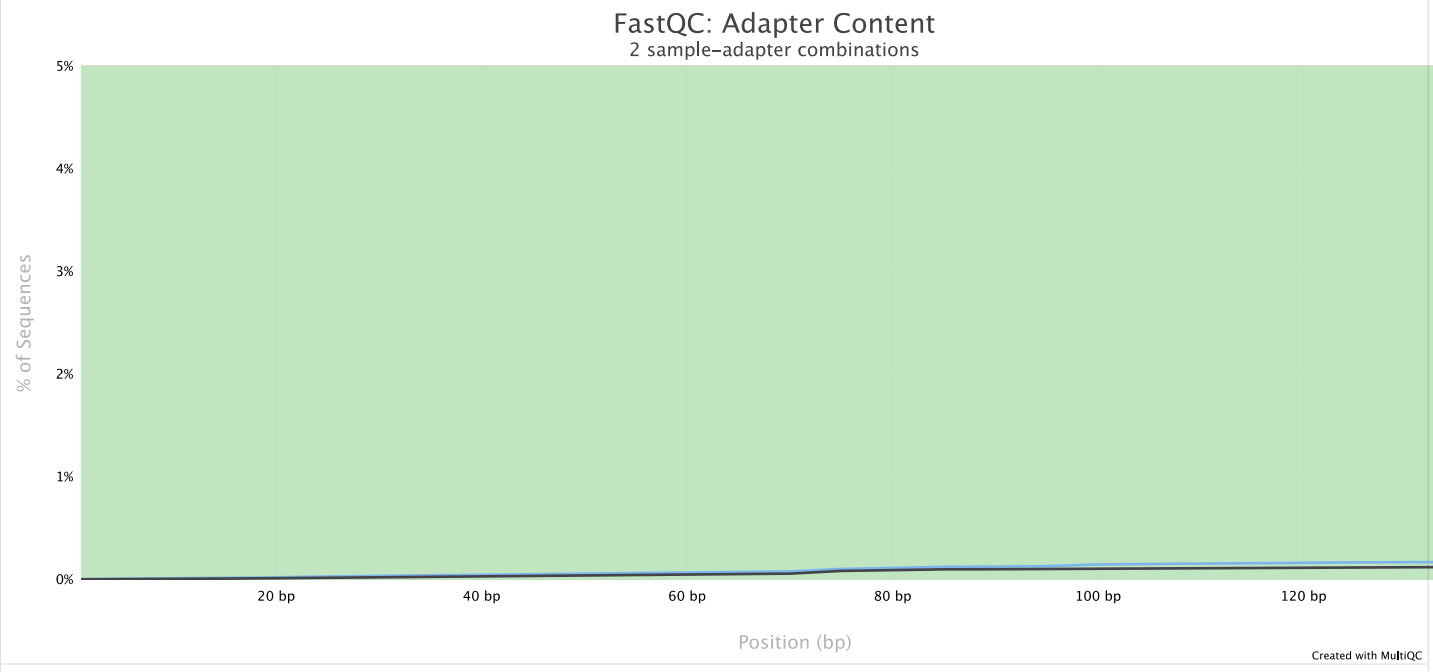
Adapter Content

17

Help

The cumulative percentage count of the proportion of your library which has seen each of the adapter sequences at each position.

Copy prompt | Summarize plot | Export...



Status Checks

Help

Status for each FastQC section showing whether results seem entirely normal (green), slightly abnormal (orange) or very unusual (red).

Sorted by sample | Clustered

Copy prompt | Summarize plot | Export...

Software Versions

Software Versions lists versions of software tools extracted from file contents.

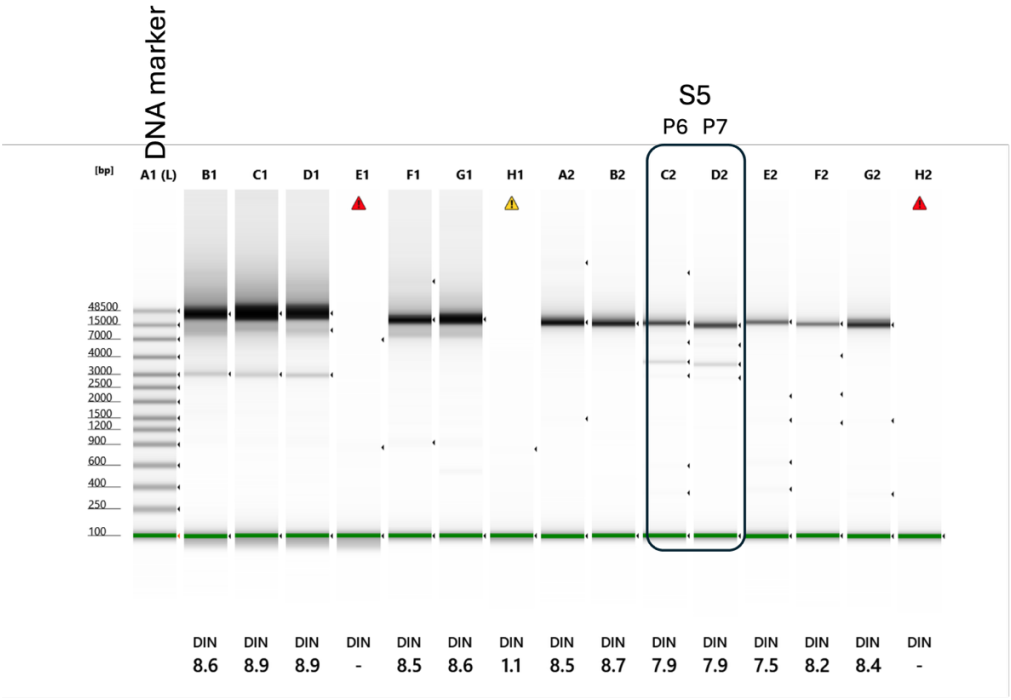
Copy table

Software	Version
FastQC	0.12.1

MultiQC v1.30 - Written by Phil Ewels, available on GitHub.
This report uses Plotly, jQuery, jQuery UI, Bootstrap and FileSaver.js.

Supplementary Figure 6: Original, Uncropped Gel image for Sample S5

This figure shows the original, uncropped gel image containing sample S5. This SLSMD sample is shown in lanes C2 and D2. The labels P6 and P7 above each lane show the two independent primer sets used for this sample as part of the clinical testing protocol. Both lanes show larger bands of the whole mitogenome at approximately 16kbp and smaller bands between 3kbp and 4kbp showing mitogenome fragments containing deletions. This confirms the presence of a very low heteroplasmy deletion in sample S5

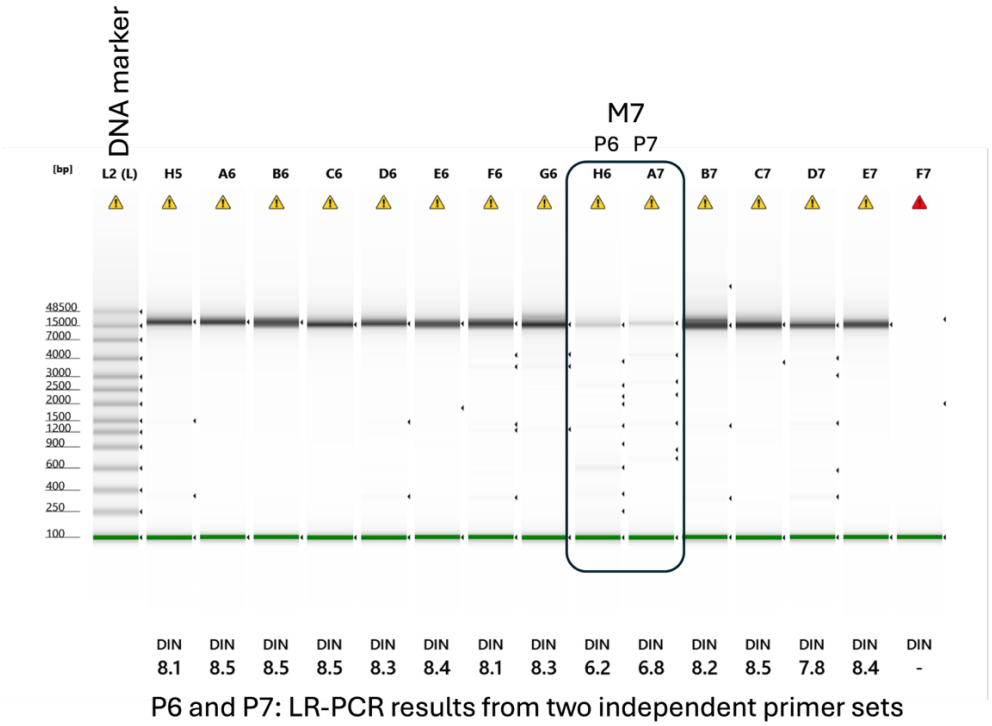


P6 and P7: LR-PCR results from two independent primer sets

Supplementary Figure 7: Original, Uncropped Gel image for Sample M7

This figure shows the original, uncropped gel image containing sample M7. This MMD sample is shown in lanes H6 and A7. The labels P6 and P7 above each lane show the two independent primer sets used for this sample as part of the clinical testing protocol. Both

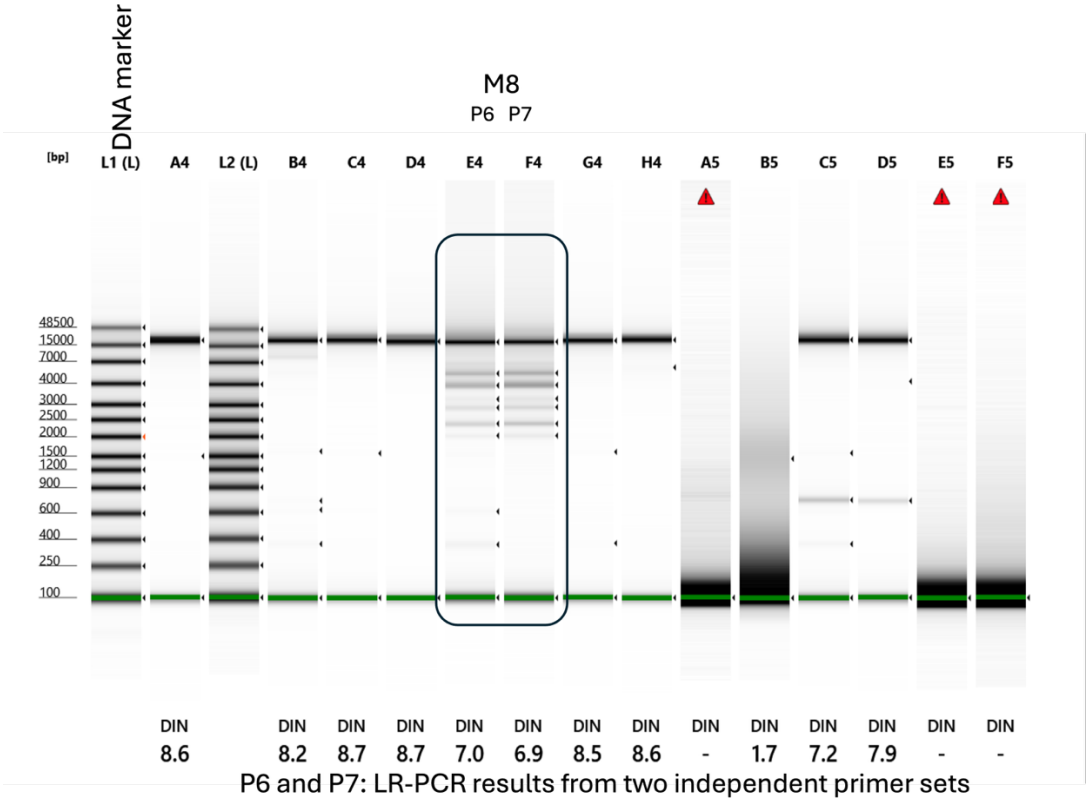
lanes show bands of the whole mitogenome at approximately 16kbp as well as several fainter bands at of varying molecular size showing mitogenome fragments containing deletions. This confirms the presence of a very low heteroplasmy deletions in sample M7



P6 and P7: LR-PCR results from two independent primer sets

Supplementary Figure 8: Original, Uncropped Gel image for Sample M8

This figure shows the original, uncropped gel image containing sample M8. This MMD sample is shown in lanes E4 and F4. The labels P6 and P7 above each lane show the two independent primer sets used for this sample as part of the clinical testing protocol. Both lanes show bands of the whole mitogenome at approximately 16kbp as well as several fainter bands at of varying molecular size showing mitogenome fragments containing deletions. This confirms the presence of a very low heteroplasmy deletions in sample M8



P6 and P7: LR-PCR results from two independent primer sets