Pharmacogenomics Next Generation Sequencing Run Quality Control Metrics

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1 Summary

Table 1: Summary of QC check results.

Barcode ID	Sample Name	Depth QC Check	Uniformity QC Check	Coverage QC Check	Overall QC Checkpoint
IonCode_0121	Sample 1	PASS	ACCEPTABLE	PASS	PASS
IonCode_0122	Sample 2	PASS	ACCEPTABLE	PASS	PASS
IonCode_0123	Sample 3	PASS	FAIL	PASS	FAIL
IonCode_0124	Sample 4	PASS	ACCEPTABLE	PASS	PASS
IonCode_0125	Sample 5	PASS	ACCEPTABLE	PASS	PASS
IonCode_0126	Sample 6	PASS	ACCEPTABLE	PASS	PASS
IonCode_0127	Sample 7	PASS	ACCEPTABLE	PASS	PASS
IonCode_0128	Sample 8	PASS	FAIL	PASS	FAIL
IonCode_0129	Sample 9	PASS	ACCEPTABLE	PASS	PASS
IonCode_0130	Sample 10	PASS	FAIL	PASS	FAIL
IonCode_0131	Sample 11	PASS	FAIL	FAIL	FAIL
IonCode_0132	Sample 12	PASS	ACCEPTABLE	PASS	PASS
IonCode_0133	Sample 13	PASS	ACCEPTABLE	PASS	PASS
IonCode_0134	Sample 14	PASS	ACCEPTABLE	PASS	PASS

Contents

2 Base Depth

2.1 Mean Depth

The mean depth for each sample is shown in ***table 2***.

Table 2: Mean Depth Results.

Barcode ID	Sample Name	Mean Depth	Depth QC Check
IonCode_0121	Sample 1	10763	PASS
IonCode_0122	Sample 2	13867	PASS
IonCode_0123	Sample 3	9712	PASS
IonCode_0124	Sample 4	6887	PASS
IonCode_0125	Sample 5	7129	PASS
IonCode_0126	Sample 6	7081	PASS
IonCode_0127	Sample 7	8287	PASS
IonCode_0128	Sample 8	11103	PASS
IonCode_0129	Sample 9	7582	PASS
IonCode_0130	Sample 10	8432	PASS
IonCode_0131	Sample 11	8082	PASS
IonCode_0132	Sample 12	8402	PASS
IonCode_0133	Sample 13	6474	PASS
IonCode_0134	Sample 14	7504	PASS

2.2 Regions with <100% Bases Read 500X (by Sample)

The distribution of the fraction of bases read 500X in a given amplicon is shown in ***figure 1***.

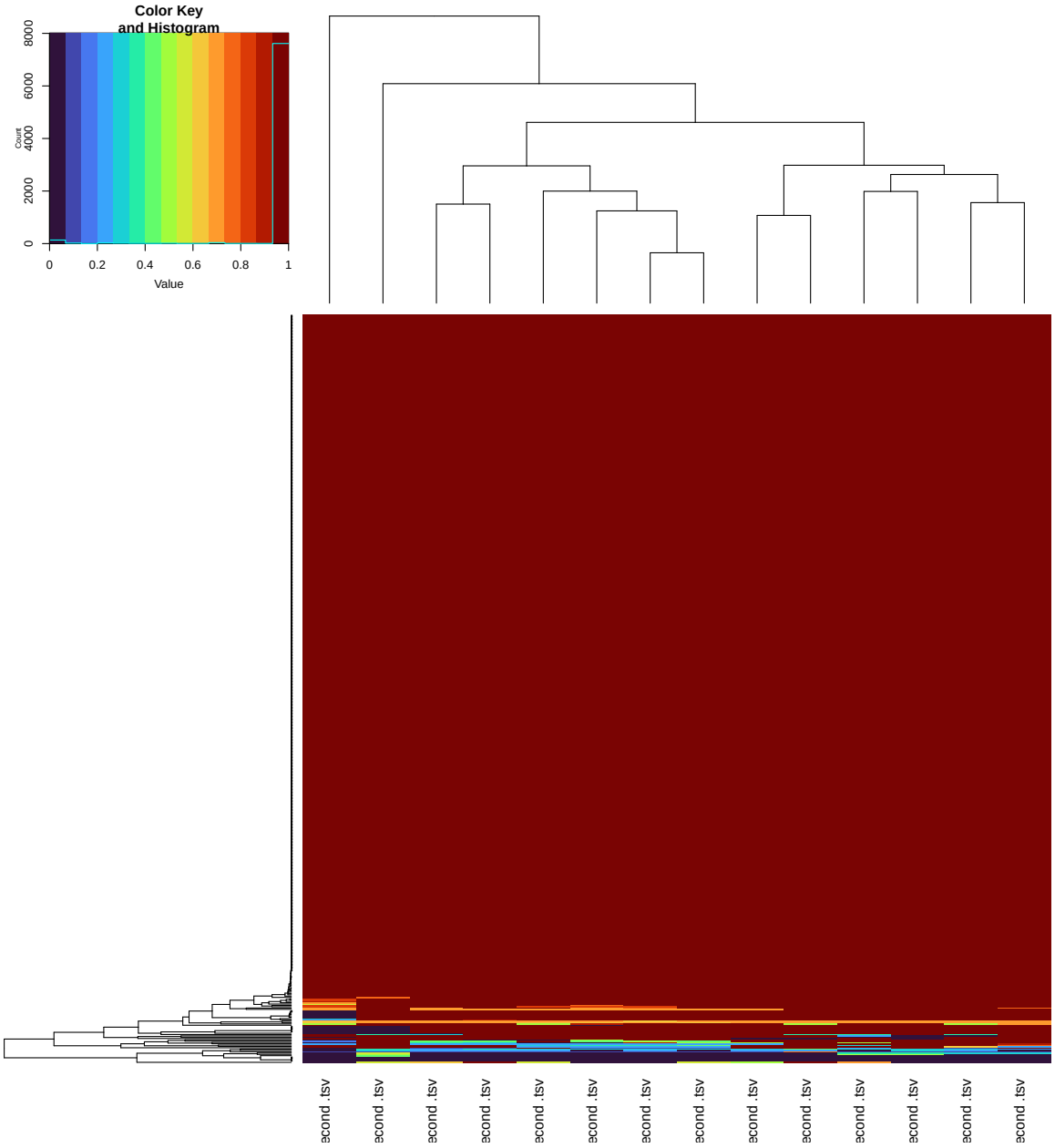

Figure 1: Heatmap showing the distribution of the fraction of bases read 500X in a given amplicon. Rows represent amplicons and columns represent samples

The following regions do not meet the criteria of having 100% of bases read 500X (by sample). The total number of unique regions that do not meet this criteria across samples is 69 regions.

```
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_01_140723_1108_day second .tsv`
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_01_140723_1108_day second .tsv`$region_id
##  [1] "AMPL3925991" "AMPL3926426" "AMPL3926093" "AMPL3926201" "AMPL3926248"
##  [6] "AMPL3926466" "AMPL3926510" "AMPL2649381" "AMPL3925990" "AMPL3926044"
## [11] "AMPL3926172" "AMPL3091682" "AMPL3926529" "AMPL3926313" "AMPL3926668"
```

```
## [16] "AMPL3926182" "AMPL3926491" "AMPL3925971" "AMPL3926501" "AMPL3926165"
## [21] "AMPL3926019" "AMPL2654393" "AMPL2686855"
##
##
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_02_140723_1108_day second .tsv`
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_02_140723_1108_day second .tsv`$region_id
##  [1] "AMPL3925991" "AMPL3926277" "AMPL3925943" "AMPL3926515" "AMPL3926625"
##  [6] "AMPL3926052" "AMPL3926072" "AMPL3926201" "AMPL3926093" "AMPL3926510"
## [11] "AMPL3926426" "AMPL3926248" "AMPL2649381" "AMPL3925990" "AMPL3091682"
## [16] "AMPL3926313" "AMPL3926095" "AMPL3926172" "AMPL3926466" "AMPL3926166"
## [21] "AMPL3926668" "AMPL3926491" "AMPL3925971" "AMPL3926036" "AMPL3926019"
## [26] "AMPL3926448" "AMPL2654393"
##
##
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_03_140723_1108_day second .tsv`
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_03_140723_1108_day second .tsv`$region_id
##  [1] "AMPL3926668" "AMPL3925991" "AMPL3926093" "AMPL3697894" "AMPL3926201"
##  [6] "AMPL3926510" "AMPL3926426" "AMPL3925990" "AMPL3925968" "AMPL3926313"
## [11] "AMPL3926248" "AMPL3926172" "AMPL2649381" "AMPL3922006" "AMPL3926095"
## [16] "AMPL3091682" "AMPL3926466" "AMPL3926295" "AMPL3926491" "AMPL3926165"
## [21] "AMPL3925971" "AMPL3926546" "AMPL3926019" "AMPL3926204" "AMPL3926556"
## [26] "AMPL3926448" "AMPL2654393"
##
##
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_04_140723_1108_day second .tsv`
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_04_140723_1108_day second .tsv`$region_id
##  [1] "AMPL3925991" "AMPL3926093" "AMPL3926201" "AMPL3926313" "AMPL3926466"
##  [6] "AMPL3926510" "AMPL3926426" "AMPL3925990" "AMPL2649381" "AMPL3091682"
## [11] "AMPL3926248" "AMPL3926476" "AMPL3926044" "AMPL3925912" "AMPL3926668"
## [16] "AMPL3926172" "AMPL3925971" "AMPL3926491" "AMPL3926247" "AMPL3926573"
## [21] "AMPL3926367" "AMPL3926165" "AMPL3926019" "AMPL3926339" "AMPL3925922"
## [26] "AMPL3926501" "AMPL2686855" "AMPL2654393"
##
##
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_05_140723_1108_day second .tsv`
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_05_140723_1108_day second .tsv`$region_id
##  [1] "AMPL3925991" "AMPL3926668" "AMPL3926093" "AMPL3926201" "AMPL3926466"
##  [6] "AMPL3926426" "AMPL3926510" "AMPL2649381" "AMPL3926248" "AMPL3091682"
## [11] "AMPL3925990" "AMPL3698522" "AMPL3926517" "AMPL3926172" "AMPL3925912"
## [16] "AMPL3926313" "AMPL3926491" "AMPL3925971" "AMPL3926386" "AMPL3926501"
## [21] "AMPL3926165" "AMPL3926019" "AMPL2683406" "AMPL3926155" "AMPL3926002"
## [26] "AMPL3926367" "AMPL3926339" "AMPL3925922" "AMPL2654393" "AMPL2686855"
## [31] "AMPL3926591"
##
##
```

```
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_06_140723_1108_day second .tsv`
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_06_140723_1108_day second .tsv`$region_id
##  [1] "AMPL3926668" "AMPL3925991" "AMPL3926093" "AMPL3926201" "AMPL3926466"
##  [6] "AMPL3926426" "AMPL3926248" "AMPL3926510" "AMPL3091682" "AMPL3926313"
## [11] "AMPL3925990" "AMPL3926172" "AMPL2649381" "AMPL3926517" "AMPL3926044"
## [16] "AMPL3926386" "AMPL3925971" "AMPL3926501" "AMPL3926491" "AMPL3926165"
## [21] "AMPL3926367" "AMPL2683406" "AMPL3926019" "AMPL3925922" "AMPL3926339"
## [26] "AMPL2654393" "AMPL2686855" "AMPL3926591"
##
##
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_07_140723_1108_day second .tsv`
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_07_140723_1108_day second .tsv`$region_id
##  [1] "AMPL3926668" "AMPL3925991" "AMPL3926093" "AMPL3926201" "AMPL3926510"
##  [6] "AMPL3926426" "AMPL3925990" "AMPL3091682" "AMPL3926313" "AMPL2649381"
## [11] "AMPL3926466" "AMPL3926172" "AMPL3926491" "AMPL3925971" "AMPL3926165"
## [16] "AMPL3926155" "AMPL2683406" "AMPL3926367" "AMPL3926019" "AMPL3926339"
## [21] "AMPL2686855" "AMPL2654393"
##
##
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_08_140723_1108_day second .tsv`
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_08_140723_1108_day second .tsv`$region_id
##  [1] "AMPL3926313" "AMPL3925991" "AMPL3926512" "AMPL3697894" "AMPL3926093"
##  [6] "AMPL3926201" "AMPL3926426" "AMPL3926510" "AMPL3925968" "AMPL3547871"
## [11] "AMPL3925990" "AMPL2649381" "AMPL3091682" "AMPL3926248" "AMPL3926466"
## [16] "AMPL3926172" "AMPL3926668" "AMPL3926491" "AMPL3925971" "AMPL2683406"
## [21] "AMPL3926165" "AMPL3926019" "AMPL2654393"
##
##
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_09_140723_1108_day second .tsv`
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_09_140723_1108_day second .tsv`$region_id
##  [1] "AMPL3926512" "AMPL3925991" "AMPL3926201" "AMPL3926510" "AMPL3091682"
##  [6] "AMPL3925990" "AMPL2649381" "AMPL3926426" "AMPL3926093" "AMPL3926466"
## [11] "AMPL3926248" "AMPL3926668" "AMPL3926172" "AMPL3926095" "AMPL3926313"
## [16] "AMPL3925971" "AMPL3926491" "AMPL3926632" "AMPL3926002" "AMPL3926339"
## [21] "AMPL3926165" "AMPL3926019" "AMPL3926448" "AMPL2686855" "AMPL2654393"
##
##
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_10_140723_1108_day second .tsv`
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_10_140723_1108_day second .tsv`$region_id
##  [1] "AMPL3925991" "AMPL3926313" "AMPL3926093" "AMPL3926201" "AMPL3926426"
##  [6] "AMPL3926510" "AMPL3926248" "AMPL3925990" "AMPL3925968" "AMPL2649381"
## [11] "AMPL3091682" "AMPL3922006" "AMPL3926172" "AMPL3926095" "AMPL3926466"
## [16] "AMPL3926386" "AMPL3926668" "AMPL3925914" "AMPL3926491" "AMPL3926339"
## [21] "AMPL3926036" "AMPL2683406" "AMPL3925971" "AMPL3926019" "AMPL3926591"
```

```
## [26] "AMPL3926448" "AMPL2654393"
##
##
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_11_140723_1108_day second .tsv`
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_11_140723_1108_day second .tsv`$region_id
##  [1] "AMPL3697894" "AMPL3926277" "AMPL3926172" "AMPL3925991" "AMPL3926625"
##  [6] "AMPL3926052" "AMPL3925943" "AMPL3926093" "AMPL3926515" "AMPL3926248"
## [11] "AMPL3926072" "AMPL3926201" "AMPL3926426" "AMPL3925990" "AMPL2649381"
## [16] "AMPL3926044" "AMPL3926529" "AMPL3926313" "AMPL3926466" "AMPL3926088"
## [21] "AMPL3926668" "AMPL3091682" "AMPL3926510" "AMPL3698522" "AMPL3926386"
## [26] "AMPL3926491" "AMPL3219749" "AMPL3925912" "AMPL3922006" "AMPL3698542"
## [31] "AMPL3926349" "AMPL2654412" "AMPL3926354" "AMPL3926579" "AMPL2683245"
## [36] "AMPL3926539" "AMPL3926470" "AMPL3925971" "AMPL2683406" "AMPL3926546"
## [41] "AMPL3926019" "AMPL3926036" "AMPL3926367" "AMPL3926501" "AMPL3926204"
## [46] "AMPL3925922" "AMPL3926591" "AMPL3926556" "AMPL2686855" "AMPL2654393"
##
##
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_12_140723_1108_day second .tsv`
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_12_140723_1108_day second .tsv`$region_id
##  [1] "AMPL3925991" "AMPL3926625" "AMPL3926426" "AMPL3926201" "AMPL3926248"
##  [6] "AMPL3926510" "AMPL3926093" "AMPL3926466" "AMPL2649381" "AMPL3091682"
## [11] "AMPL3926095" "AMPL3925990" "AMPL3926313" "AMPL3926172" "AMPL3926668"
## [16] "AMPL3926491" "AMPL3925971" "AMPL3926501" "AMPL3926339" "AMPL3926367"
## [21] "AMPL3926019" "AMPL3926573" "AMPL3926448" "AMPL2686855" "AMPL2654393"
##
##
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_13_140723_1108_day second .tsv`
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_13_140723_1108_day second .tsv`$region_id
##  [1] "AMPL3925991" "AMPL3926093" "AMPL3926201" "AMPL3926466" "AMPL3926510"
##  [6] "AMPL3926426" "AMPL2649381" "AMPL3091682" "AMPL3926044" "AMPL3925990"
## [11] "AMPL3926172" "AMPL3926248" "AMPL3925912" "AMPL3926668" "AMPL3926491"
## [16] "AMPL3926313" "AMPL3925971" "AMPL3926165" "AMPL3926002" "AMPL3926367"
## [21] "AMPL3926019" "AMPL3925922" "AMPL3926339" "AMPL2686855" "AMPL2654393"
## [26] "AMPL3925962"
##
##
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_14_140723_1108_day second .tsv`
## $`experiment_data_140723_1108_day second /depth_140723_1108_day second
↪  /depth_14_140723_1108_day second .tsv`$region_id
##  [1] "AMPL3926512" "AMPL3925991" "AMPL3926093" "AMPL3926201" "AMPL3926426"
##  [6] "AMPL3926510" "AMPL3926466" "AMPL3091682" "AMPL2649381" "AMPL3925990"
## [11] "AMPL3926313" "AMPL3926172" "AMPL3925912" "AMPL3926668" "AMPL3926248"
## [16] "AMPL3926491" "AMPL3925971" "AMPL3926165" "AMPL3926019" "AMPL2686855"
## [21] "AMPL2654393"
```

2.3 Intersect Between Regions with <100% Bases Read 500X in at least Half of the Samples

The following 69 regions have <100% bases read 500X in at least one sample.

```
##  [1] "AMPL3925991" "AMPL3926426" "AMPL3926093" "AMPL3926201" "AMPL3926248"
##  [6] "AMPL3926466" "AMPL3926510" "AMPL2649381" "AMPL3925990" "AMPL3926044"
## [11] "AMPL3926172" "AMPL3091682" "AMPL3926529" "AMPL3926313" "AMPL3926668"
## [16] "AMPL3926182" "AMPL3926491" "AMPL3925971" "AMPL3926501" "AMPL3926165"
## [21] "AMPL3926019" "AMPL2654393" "AMPL2686855" "AMPL3926277" "AMPL3925943"
## [26] "AMPL3926515" "AMPL3926625" "AMPL3926052" "AMPL3926072" "AMPL3926095"
## [31] "AMPL3926166" "AMPL3926036" "AMPL3926448" "AMPL3697894" "AMPL3925968"
## [36] "AMPL3922006" "AMPL3926295" "AMPL3926546" "AMPL3926204" "AMPL3926556"
## [41] "AMPL3926476" "AMPL3925912" "AMPL3926247" "AMPL3926573" "AMPL3926367"
## [46] "AMPL3926339" "AMPL3925922" "AMPL3698522" "AMPL3926517" "AMPL3926386"
## [51] "AMPL2683406" "AMPL3926155" "AMPL3926002" "AMPL3926591" "AMPL3926512"
## [56] "AMPL3547871" "AMPL3926632" "AMPL3925914" "AMPL3926088" "AMPL3219749"
## [61] "AMPL3698542" "AMPL3926349" "AMPL2654412" "AMPL3926354" "AMPL3926579"
## [66] "AMPL2683245" "AMPL3926539" "AMPL3926470" "AMPL3925962"
```

The following 20 regions have <100% bases read 500X in at least half of the samples.

```
##  [1] "AMPL2649381" "AMPL2654393" "AMPL2686855" "AMPL3091682" "AMPL3925971"
##  [6] "AMPL3925990" "AMPL3925991" "AMPL3926019" "AMPL3926093" "AMPL3926165"
## [11] "AMPL3926172" "AMPL3926201" "AMPL3926248" "AMPL3926313" "AMPL3926339"
## [16] "AMPL3926426" "AMPL3926466" "AMPL3926491" "AMPL3926510" "AMPL3926668"
```

3 Amplicon Coverage

3.1 Coverage Frequency

The fraction of amplicons in each sample that are not covered at least 500X is shown in ***table 3***.

Table 3: Coverage Frequency Results. 'Freq' is the fraction of amplicons that are not covered at least 500X.

Barcode ID	Sample Name	Freq	Coverage QC Check
IonCode_0121	Sample 1	0.0176991	PASS
IonCode_0122	Sample 2	0.0194690	PASS
IonCode_0123	Sample 3	0.0141593	PASS
IonCode_0124	Sample 4	0.0176991	PASS
IonCode_0125	Sample 5	0.0194690	PASS
IonCode_0126	Sample 6	0.0230088	PASS
IonCode_0127	Sample 7	0.0159292	PASS
IonCode_0128	Sample 8	0.0159292	PASS
IonCode_0129	Sample 9	0.0088496	PASS
IonCode_0130	Sample 10	0.0159292	PASS
IonCode_0131	Sample 11	0.0513274	FAIL
IonCode_0132	Sample 12	0.0176991	PASS
IonCode_0133	Sample 13	0.0176991	PASS
IonCode_0134	Sample 14	0.0176991	PASS

The distribution of the coverage of amplicons is shown in ***figure 2***.

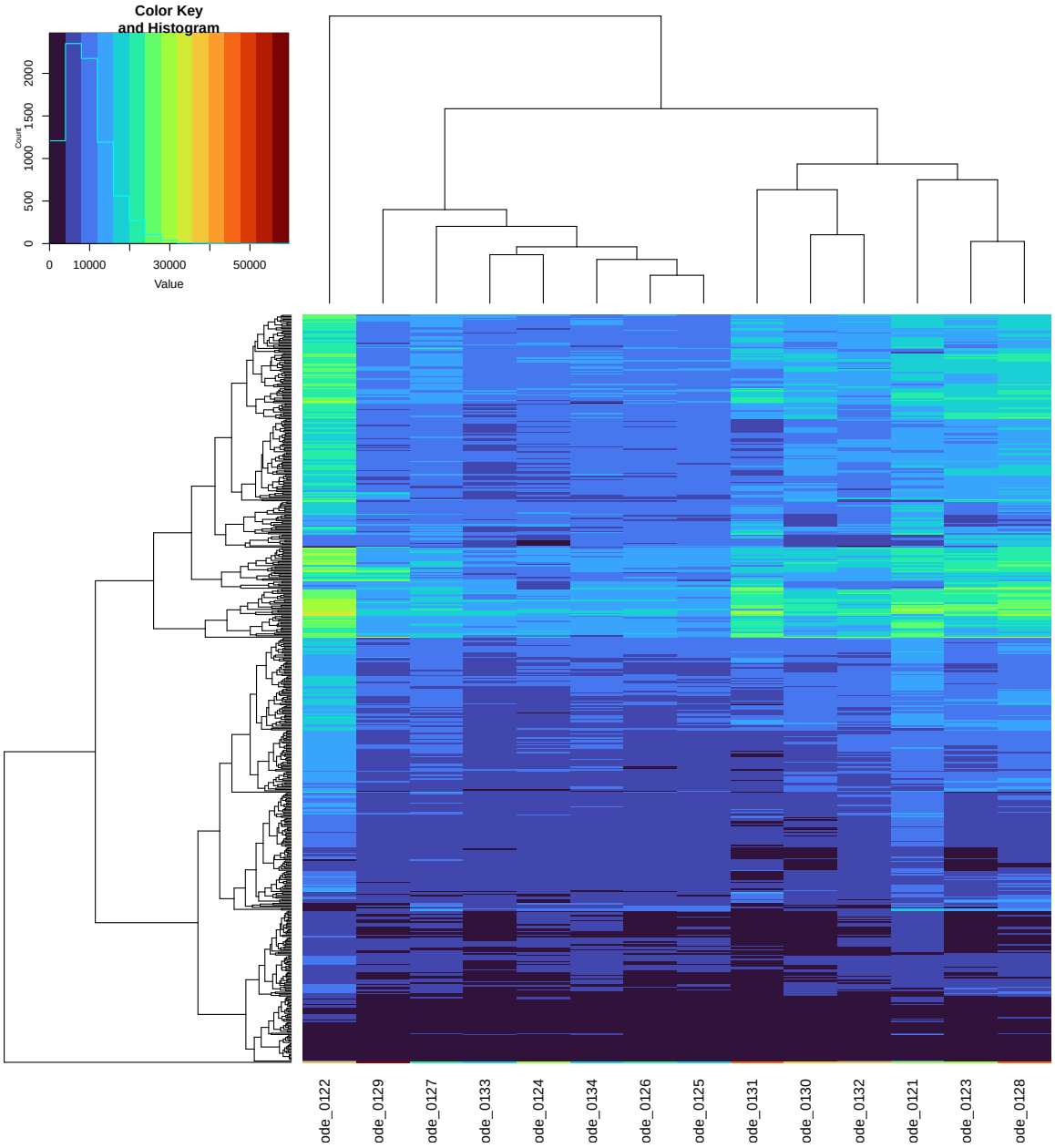

Figure 2: Heatmap showing the distribution of the coverage of each amplicon. Rows represent amplicons and columns represent samples

3.2 Intersect Between Regions covered <500X in at least Half of the Samples

The following amplicons were covered less than 500X in at least half of the samples.

```
## $Target
## [1] "AMPL3925990" "AMPL3926201" "AMPL3091682" "AMPL2649381" "AMPL3925991"
## [6] "AMPL3926093" "AMPL3926466" "AMPL3926426" "AMPL3926510"
```

4 Uniformity

The uniformity results for each sample are shown in ***table 4***.

Table 4: Uniformity Results.

Barcode ID	Sample Name	Uniformity	Uniformity QC Check
IonCode_0121	Sample 1	93.57%	ACCEPTABLE
IonCode_0122	Sample 2	93.14%	ACCEPTABLE
IonCode_0123	Sample 3	91.87%	FAIL
IonCode_0124	Sample 4	94.17%	ACCEPTABLE
IonCode_0125	Sample 5	94.25%	ACCEPTABLE
IonCode_0126	Sample 6	93.69%	ACCEPTABLE
IonCode_0127	Sample 7	95.28%	ACCEPTABLE
IonCode_0128	Sample 8	92.74%	FAIL
IonCode_0129	Sample 9	94.45%	ACCEPTABLE
IonCode_0130	Sample 10	91.83%	FAIL
IonCode_0131	Sample 11	86.14%	FAIL
IonCode_0132	Sample 12	93.63%	ACCEPTABLE
IonCode_0133	Sample 13	94.20%	ACCEPTABLE
IonCode_0134	Sample 14	94.26%	ACCEPTABLE