# Sample Results Report - BP02-17 - NA07981

Coffalyser digitalMLPA

## General Information

| Software information | Executed | Program version |
|---|---|---|
| Configuration | n/a | 2022-10-20 12:34 |
| Data conversion | 2023-04-19 06:52 | 2021-10-19 09:58 |
| Fragment analysis | 2023-04-19 06:53 | 2021-10-19 09:58 |
| Comparative analysis | 2023-04-19 06:53 | 2021-10-19 09:58 |
| Extended analysis | n/a | n/a |
| Reporting | 2023-04-19 06:53 | 2021-10-19 09:58 |

| Sample | |
|---|---|
| Sample name (type) | **BP02-17 - NA07981 (Undefined)** |
| Barcode - well (lot) | BP02-17 - A03 (03-008-yymmdd) |
| SNP code | 12002.01200.20210.01011.01112.11122.11120.1210 v1 |
| Gender | Male |
| Estimated DNA quantity | 8 ng |
| Sample track probes | (none) |

| Experiment settings | |
|---|---|
| Experiment name (type) | D006 Demoset (digitalMLPA) |
| Product name | D006-D006-A1 tube AB4B0D 13-10-2022 [default sheet] |
| Product detection | Identity detected |
| Digital signatures | All signatures valid |
| Probe catalogue | Default, Version 2022-10-20 12:34 |
| Source files | C:\D006-A1 Demoset\Demoset_D006-A1 final.fastq |

| Analysis details | |
|---|---|
| Normalisation method | Individual reference probe normalisation [default] |
| Coffa file location | C:\D006-A1 Demoset\20230419_064513\D006 Demoset\BP02-17 - NA07981.Coffa |
| Coffa file id | 09195273-f659-4619-ab3c-84b16fa22202 |
| Reference samples | REF-Male-1, REF-Male-2, REF-Male-3, REF-Male-4, REF-Male-5 |

| Reads | Items | Reads |
|---|---|---|
| Recognised | 660 | 1.527.256 |
| Unrecognised | n/a | 86.190 |
| Median total reads | n/a | 2.496 |
| Median distinct reads | n/a | 1.030 |

| Probes | Expected | Detected | Unexpected |
|---|---|---|---|
| Test probes | 508 | 508 | 0 |
| Reference probes | 81 | 81 | 0 |
| Y-control probes | 3 | 3 | n/a |
| Mutation probes | 0-1 | 0 | n/a |

## Quality Details

**Quality Checks: Passed**

| Quality | | |
|---|---|---|
| **Category** | **Description** | **Classification** |
| Reaction analysis | Read depth | Ok |
| | Sequence quality | Ok |
| | Unrecognised reads | Ok |
| | Sample DNA denaturation | Ok |
| | Hybridisation Tm | Ok |
| | Hybridisation completeness | Ok |
| | Ligation start temperature | Ok |
| | Ligase activity | Ok |
| | Ligase inactivation | Ok |
| | Polymerase activity | Ok |
| Comparative analysis | Sample DNA denaturation | Ok |
| | Sample DNA depurination | Ok |
| | Sample DNA fragment length | Ok |
| | Hybridisation Tm | Ok |
| | Hybridisation completeness | Ok |
| | Ligase activity | Ok |
| | Polymerase activity | Ok |
| | Reference probe quality | Ok |
| | Reference sample quality | Ok |

RESEARCH ONLY

## Result Summary

**Note:** Please refer to the 'Probe Information File' for additional information that may be crucial for correct result interpretation.

### Chart(s)

RESEARCH USE ONLY

## Mutation-specific probe(s)

| Gene | Mutation | Probe | Mutation status | Additional information |
|---|---|---|---|---|
| BRAF | V600E | S011487 | Absent | p.V600E = c.1799T>A |

## Aberrant region(s) table

| Region | Chromo-somal band (hg38) | Region size | Region ratio | Stdev | Probes in region | Probe number | Probe function | Mapview (hg38) in kb | Normal copy number | Gene | Exon | Inter ratio | Within normal range | Different from reference population | Ambi-guous |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 12P | 12p13.33 - 12p11.22 | 28,6 Mb | 2,00 | 0,06 | 27 | S013063 | CN | 12-000.884 | 2 | WNK1 | | 1,93 | TooHigh | Increased2StdDev | False |
| | | | | | | S013116 | CN | 12-001.847 | 2 | CACNA2D4 | | 2,04 | TooHigh | Increased2StdDev | False |
| | | | | | | S011505 | CN | 12-003.084 | 2 | TSPAN9 | | 2,00 | TooHigh | Increased2StdDev | False |
| | | | | | | S011432 | CN | 12-006.385 | 2 | LTBR | 4 | 2,08 | TooHigh | Increased2StdDev | False |
| | | | | | | S011431 | CN | 12-006.390 | 2 | LTBR | 9 | 2,02 | TooHigh | Increased2StdDev | False |
| | | | | | | S011503 | CN | 12-006.450 | 2 | CD27 | | 2,04 | TooHigh | Increased2StdDev | False |
| | | | | | | S011428 | CN | 12-006.465 | 2 | VAMP1 | | 2,00 | TooHigh | Increased2StdDev | False |
| | | | | | | S011429 | CN | 12-006.514 | 2 | NCAPD2 | 8 | 1,89 | TooHigh | Increased2StdDev | False |
| | | | | | | S011427 | CN | 12-006.528 | 2 | NCAPD2 | 23 | 1,90 | TooHigh | Increased2StdDev | False |
| | | | | | | S011430 | CN | 12-006.582 | 2 | CHD4 | 30 | 2,07 | TooHigh | Increased2StdDev | False |
| | | | | | | S011504 | CN | 12-006.606 | 2 | CHD4 | 2 | 1,95 | TooHigh | Increased2StdDev | False |
| | | | | | | S011799 | CN | 12-011.839 | 2 | ETV6 | 3 | 2,09 | TooHigh | Increased2StdDev | False |
| | | | | | | S011801 | CN | 12-011.884 | 2 | ETV6 | 6 | 1,95 | TooHigh | Increased2StdDev | False |
| | | | | | | S011502 | CN | 12-011.894 | 2 | ETV6 | 8 | 1,99 | TooHigh | Increased2StdDev | False |
| | | | | | | S020146 | CN | 12-012.080 | 2 | BCL2L14 | | 1,98 | TooHigh | Increased2StdDev | False |
| | | | | | | S020143 | CN | 12-012.181 | 2 | LRP6 | | 1,97 | TooHigh | Increased2StdDev | False |
| | | | | | | S020145 | CN | 12-012.436 | 2 | BORCS5 | | 2,04 | TooHigh | Increased2StdDev | False |
| | | | | | | S018334 | CN | 12-012.719 | 2 | CDKN1B | 2 | 2,00 | TooHigh | Increased2StdDev | False |
| | | | | | | S011501 | CN | 12-012.721 | 2 | CDKN1B | 3 | 1,89 | TooHigh | Increased2StdDev | False |
| | | | | | | S019999 | CN | 12-012.941 | 2 | GPRC5D | 4 | 2,00 | TooHigh | Increased2StdDev | False |
| | | | | | | S020003 | CN | 12-012.942 | 2 | GPRC5D | 3 | 1,95 | TooHigh | Increased2StdDev | False |
| | | | | | | S019996 | CN | 12-012.950 | 2 | GPRC5D | 2 | 2,04 | TooHigh | Increased2StdDev | False |
| | | | | | | S011086 | CN | 12-019.463 | 2 | AEBP2 | | 1,92 | TooHigh | Increased2StdDev | False |
| | | | | | | S019994 | CN | 12-027.697 | 2 | REP15 | | 2,09 | TooHigh | Increased2StdDev | False |
| | | | | | | S020118 | CN | 12-027.767 | 2 | MANSC4 | | 2,02 | TooHigh | Increased2StdDev | False |
| | | | | | | S019990 | CN | 12-027.963 | 2 | PTHLH | | 2,00 | TooHigh | Increased2StdDev | False |
| | | | | | | S011085 | CN | 12-029.516 | 2 | TMTC1 | | 2,02 | TooHigh | Increased2StdDev | False |

RESEARCH USE ONLY

| | Analysis performed | Final approval |
|---|---|---|
| **Quality Checks: Passed** | Date:<br>Signature: | Date:<br>Signature: |

RESEARCH USE ON