# Sample Results Report - BP02-04 - NA01535

Coffalyser digitalMLPA

## General Information

| Software information | Executed | Program version |
|---|---|---|
| Configuration | n/a | 2022-10-20 12:34 |
| Data conversion | 2023-04-18 14:48 | 2021-10-19 09:58 |
| Fragment analysis | 2023-04-18 14:49 | 2021-10-19 09:58 |
| Comparative analysis | 2023-04-18 14:49 | 2021-10-19 09:58 |
| Extended analysis | n/a | n/a |
| Reporting | 2023-04-18 14:50 | 2021-10-19 09:58 |

| Sample | |
|---|---|
| Sample name (type) | **BP02-04 - NA01535 (Undefined)** |
| Barcode - well (lot) | BP02-04 - D01 (03-008-yymmdd) |
| SNP code | 12011.02110.20221.01022.10111.11221.21121.1112 v1 |
| Gender | Female |
| Estimated DNA quantity | 29 ng |
| Sample track probes | (none) |

| Experiment settings | |
|---|---|
| Experiment name (type) | Demoset D001-C1 (digitalMLPA) |
| Product name | D001-C1-1221 [default sheet] |
| Product detection | Identity detected |
| Digital signatures | All signatures valid |
| Probe catalogue | Default, Version 2022-10-20 12:34 |
| Source files | D:\Demosets\D001-C1\Demoset_D001-C1 final.fastq |

| Analysis details | |
|---|---|
| Normalisation method | Individual reference probe normalisation [default] |
| Coffa file location | C:\D001 Demoset\20230418_143831\Demoset D001-C1\BP02-04 - NA01535.Coffa |
| Coffa file id | ecde31fc-c348-4c54-9a04-4790bf03c2ea |
| Reference samples | REF-Male-1, REF-Male-2, REF-Male-3, REF-Male-4, REF-Male-5 |

| Reads | Items | Reads |
|---|---|---|
| Recognised | 722 | 1.912.642 |
| Unrecognised | n/a | 43.567 |
| Median total reads | n/a | 2.880 |
| Median distinct reads | n/a | 1.902 |

| Probes | Expected | Detected | Unexpected |
|---|---|---|---|
| Test probes | 579 | 579 | 0 |
| Reference probes | 290 | 290 | 0 |
| Y-control probes | 0 | 0 | n/a |
| Mutation probes | 0-5 | 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 |

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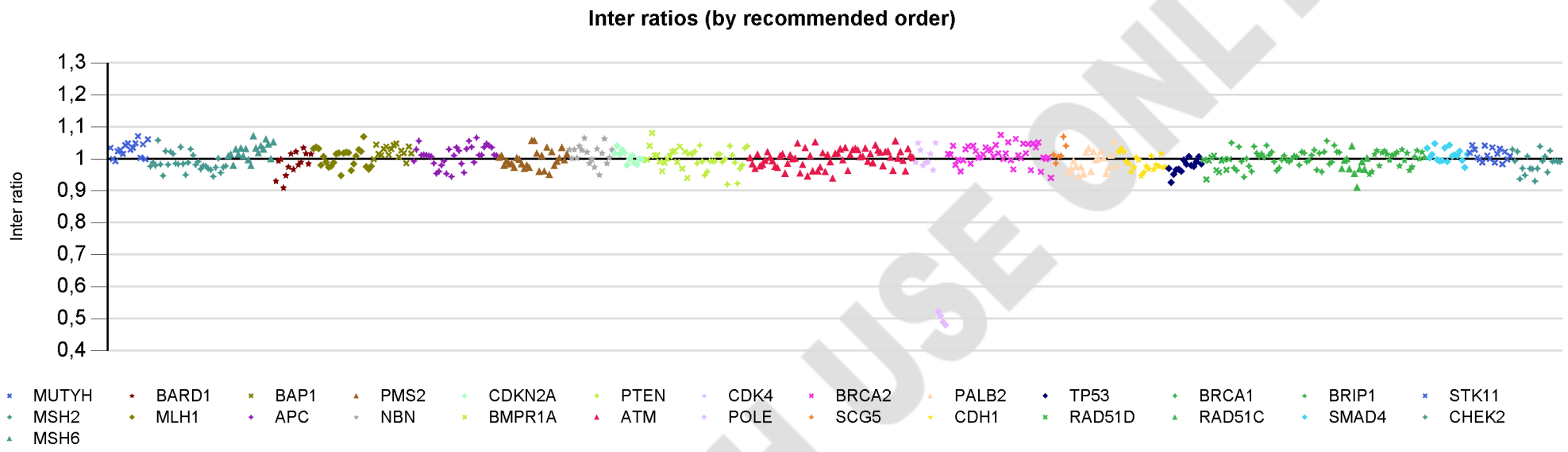
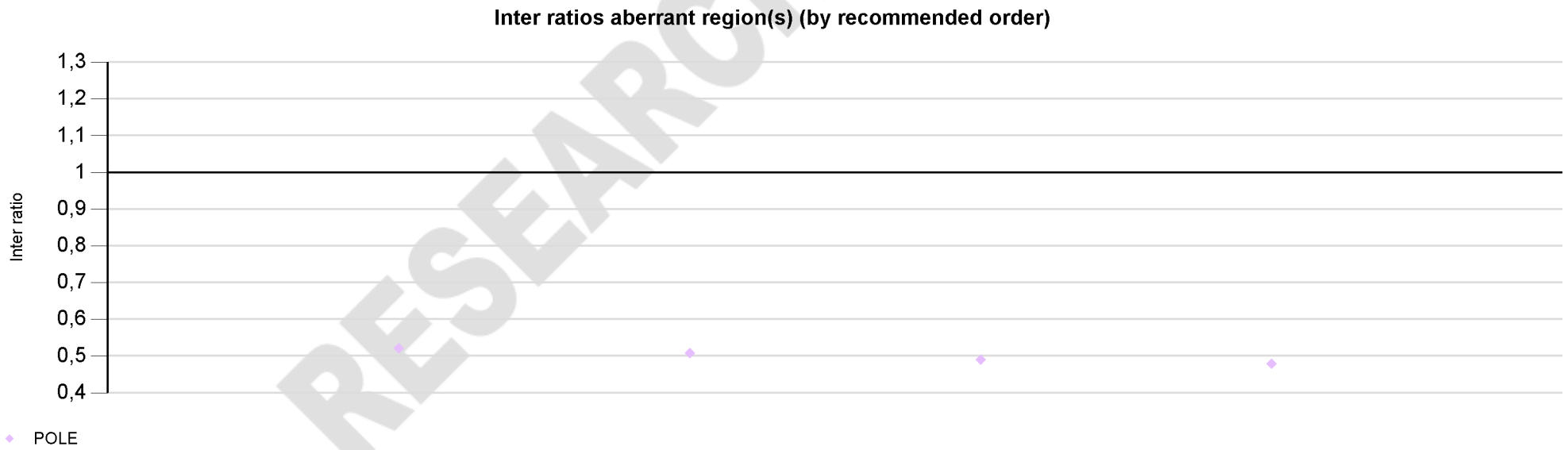
## Result Summary

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

### Chart(s)

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## Mutation-specific probe(s)

| Gene | Mutation | Probe | Mutation status | Additional information |
|---|---|---|---|---|
| MSH2 | 10MBinv | S010444 | Absent | MSH2 10 Mb inversion (PMID24114314); This probe consists of three oligos and has two ligation sites. |
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| MITF | 952G>A | S010555 | Absent | Mutation also known as p.E318K (rs149617956). |
| PMS2 | SVA-ins | S010670 | Absent | PMS2 SVA element insertion (PMID22461402). |
| CHEK2 | 1100delC | S010117 | Absent | Mutation also known as rs555607708; This probe consists of three oligos and has two ligation sites. |

## 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 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| POLE | 12q24.33 | 54,9 Kb | 0,50 | 0,02 | 4 | S019716 | CN | 12-132.626 | 2 | POLE | 46 | 0,52 | TooLow | Decreased2StdDev | False |
| | | | | | | S019402 | CN | 12-132.673 | 2 | POLE | 15 | 0,51 | TooLow | Decreased2StdDev | False |
| | | | | | | S010663 | CN/WT | 12-132.674 | 2 | POLE | 13 | 0,49 | TooLow | Decreased2StdDev | False |
| | | | | | | S019406 | CN | 12-132.681 | 2 | POLE | 02 | 0,48 | TooLow | Decreased2StdDev | False |

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

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