

# Sensitivity and specificity of data collection strategies in coding regions

381 European sample reference panel

## Coding variants

**a**

|           | Sens <sub>I</sub> |       |       |       |       |           | Spec <sub>I</sub> |        |       |        |      |
|-----------|-------------------|-------|-------|-------|-------|-----------|-------------------|--------|-------|--------|------|
|           | 0x                | .5x   | 1x    | 2x    | 4x    |           | 0x                | .5x    | 1x    | 2x     | 4x   |
| No Array  | NA                | 81.22 | 87.31 | 91.12 | 97.46 | No Array  | NA                | 99.69  | 99.42 | 100.00 | 100. |
| Affy 100k | 15.23             | 81.98 | 86.29 | 92.39 | 97.21 | Affy 100k | 100.00            | 99.38  | 99.71 | 100.00 | 100. |
| Affy 500k | 58.88             | 84.52 | 87.56 | 92.64 | 96.70 | Affy 500k | 99.57             | 99.70  | 99.71 | 99.73  | 99.  |
| Affy 6    | 73.60             | 88.07 | 90.10 | 92.89 | 97.21 | Affy 6    | 99.66             | 99.71  | 99.72 | 99.73  | 99.  |
| Illum 1M  | 91.88             | 91.88 | 93.65 | 94.67 | 96.95 | Illum 1M  | 100.00            | 100.00 | 99.73 | 100.00 | 100. |
| Omni 2.5  | 93.65             | 94.92 | 94.67 | 95.43 | 97.97 | Omni 2.5  | 100.00            | 100.00 | 99.73 | 100.00 | 100. |

## Noncoding variants

**b**

|           | Sens <sub>I</sub> |       |       |       |       |           | Spec <sub>I</sub> |       |       |       |       |
|-----------|-------------------|-------|-------|-------|-------|-----------|-------------------|-------|-------|-------|-------|
|           | 0x                | .5x   | 1x    | 2x    | 4x    |           | 0x                | .5x   | 1x    | 2x    | 4x    |
| No Array  | NA                | 83.34 | 88.60 | 92.10 | 94.87 | No Array  | NA                | 99.41 | 99.49 | 99.68 | 99.73 |
| Affy 100k | 26.74             | 83.86 | 88.70 | 92.09 | 94.76 | Affy 100k | 97.70             | 99.34 | 99.49 | 99.67 | 99.70 |
| Affy 500k | 70.37             | 86.69 | 89.82 | 92.41 | 94.89 | Affy 500k | 98.64             | 99.38 | 99.48 | 99.61 | 99.69 |
| Affy 6    | 82.00             | 88.58 | 90.67 | 92.87 | 95.14 | Affy 6    | 99.29             | 99.50 | 99.50 | 99.63 | 99.68 |
| Illum 1M  | 88.42             | 90.93 | 91.87 | 93.52 | 95.55 | Illum 1M  | 99.64             | 99.70 | 99.71 | 99.75 | 99.79 |
| Omni 2.5  | 91.38             | 92.50 | 93.13 | 94.38 | 95.93 | Omni 2.5  | 99.75             | 99.78 | 99.75 | 99.80 | 99.84 |