



KASP exome sample tracking: Easy tracking of your human DNA samples throughout your NGS workflow

Background

Whole exome sequencing provides a comparatively cost-effective means to sequence protein coding regions within genomes using target enrichment strategies.

The high investment cost of next-generation sequencing instruments means that many laboratories outsource library preparation, sequencing and informatics, ultimately passing custody of precious samples into the hands of others. This can lead to the possibility of expensive preparation mistakes and the return of incorrect data. Simple sample verification can be performed using genotype profiling. A highly discriminatory reference genotype profile can be obtained from the original genome sample, that can then be compared with the subsequent library and data.

The KASP exome sample tracking panel was developed by University of Southampton and LGC in using KASP™ to track sample provenance throughout the stages of changing sample stewardship. The panel uses 24 SNPs (single-nucleotide polymorphisms) within protein coding regions of the human genome that are commonly targeted and enriched by Agilent™ SureSelect Human All Exon V4, Illumina TruSeq Exome Enrichment™ and Nimblegen SeqCap EZ Human Exome Library V3.0™ kits.

*For further information and when referencing this panel please use the following citation: **Pengelly et al. Genome Medicine 2013, 5:89 <http://genome-medicine.com/content/5/9/89>***

Orthogonal genotyping

The KASP exome sample tracking panel uses 24 SNPs across the 22 autosomes. Each assay targets a protein coding region of the human genome to provide a genotype profile for your sample. In Table 1, the target and NCBI SNP database reference numbers are given. A more detailed explanation of the panel and selection of the SNPs can be found in the above reference.

Technical requirements

KASP requires a minimum of 10 ng of DNA per data point from genomic starting material. For our genotyping service please provide samples at a minimum concentration of 10 ng / µL, higher concentrations of up to 80 ng / µL are also tolerated; samples will be diluted by LGC prior to genotyping. Providing 50 µL of both genomic and enriched samples will enable us to perform any repeats if needed. For more information please contact the technical support team: tech.support@lgcgenomics.com

Availability

The KASP exome sample tracking panel is easy to set up in any laboratory with a qPCR instrument. Alternatively the panel is also available as a convenient service in any of our global genotyping facilities.

How to order

To request pricing and to order please contact your local sales representative, our customer support team, or complete our online enquiry form www.lgcgenomics.com/contact_us.

Table 1: KASP exome sample tracking panel targets

| Chromosome | Position† | dbSNP rsID | Gene | Allele FAM | Allele HEX |
|------------|-----------|------------|-----------------|------------|------------|
| 1 | 179520506 | rs1410592 | <i>NPHS2</i> | A | G |
| 1 | 67861520 | rs2229546 | <i>IL12RB2</i> | A | C |
| 2 | 169789016 | rs497692 | <i>ABCB11</i> | A | G |
| 2 | 227896976 | rs10203363 | <i>COL4A4</i> | C | T |
| 3 | 4403767 | rs2819561 | <i>SUMF1</i> | C | T |
| 4 | 5749904 | rs4688963 | <i>EVC</i> | A | G |
| 5 | 82834630 | rs309557 | <i>VCAN</i> | A | G |
| 6 | 146755140 | rs2942 | <i>GRM1</i> | A | G |
| 7 | 48450157 | rs17548783 | <i>ABCA13</i> | C | T |
| 8 | 94935937 | rs4735258 | <i>PDP1</i> | C | T |
| 9 | 100190780 | rs1381532 | <i>TDRD7</i> | C | T |
| 10 | 100219314 | rs10883099 | <i>HPSE2</i> | A | G |
| 11 | 16133413 | rs4617548 | <i>SOX6</i> | A | G |
| 12 | 993930 | rs7300444 | <i>WNK1</i> | C | T |
| 13 | 39433606 | rs9532292 | <i>FREM2</i> | A | G |
| 14 | 50769717 | rs2297995 | <i>L2HGDH</i> | A | G |
| 15 | 34528948 | rs4577050 | <i>SLC12A6</i> | A | G |
| 16 | 70303580 | rs2070203 | <i>AARS</i> | C | T |
| 17 | 71197748 | rs1037256 | <i>COG1</i> | A | G |
| 18 | 21413869 | rs9962023 | <i>LAMA3</i> | C | T |
| 19 | 10267077 | rs2228611 | <i>DNMT1</i> | A | G |
| 20 | 6100088 | rs10373 | <i>FERMT1</i> | C | T |
| 21 | 44323590 | rs4148973 | <i>NDUFV3</i> | G | T |
| 22 | 21141300 | rs4675 | <i>SERPIND1</i> | C | T |

Human reference assembly GRCh37 (hg19)

Key words: KASP, GWAS, TaqMan, Alzheimer's disease, Mx3000P, genotyping.

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