

DATA ANALYSIS

Picard Analysis of externally generated data - Human Exome

Part Number : P-ANA-0001

Product Family Data : Analysis

Availability Date : 11/01/2012

Description

Processing of externally generated Illumina sequencing data through the platform's Picard exome processing pipeline to create an analysis-ready BAM file. Includes any necessary format conversion and merging of data per sample through to creation of a standard Picard BAM file per sample. Includes short indel co-cleaning where appropriate. Data storage is not included. Capacity allows for processing up to 1000 samples' data per week. Volumes above 1000/week can be accommodated on an as-needed basis; please contact picard@broadinstitute.org to discuss any high volume requests.

Deliverables

A Picard BAM file and standard pipeline metrics delivered onto project-owned storage.

Input Requirements

- Illumina sequence data in either BAM or FASTQ format on a Broad file system.
- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed.
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender, Capture Product.

Picard Analysis of externally generated data - Human Whole Genome (4-6x)

Part Number : P-ANA-0002

Product Family : Data Analysis

Availability Date : 11/01/2012

Description

Processing of externally generated Illumina sequencing data through the platform's Picard whole genome processing pipeline to create an analysis-ready BAM file. Includes any necessary format conversion and merging of data per sample through to creation of a standard Picard BAM file per sample. Data storage is not included. Capacity allows for processing up to 1000 samples' data per week. Volumes above 1000/week can be accommodated on an as-needed basis; please contact picard@broadinstitute.org to discuss any high volume requests.

Deliverables

A Picard BAM file and standard pipeline metrics delivered onto project-owned storage.

Input Requirements

- Illumina sequence data in either BAM or FASTQ format on a Broad file system.
- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed.
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender.

Picard Analysis of externally generated data - Human Whole Genome (30x)

Part Number : P-ANA-0003

Product Family : Data Analysis

Availability Date : 11/01/2012

Description

Processing of externally generated Illumina sequencing data through the platform's Picard whole genome processing pipeline to create an analysis-ready BAM file. Includes any necessary format conversion and merging of data per sample through to creation of a standard Picard BAM file per sample. Includes short indel co-cleaning where appropriate. Data storage is not included. Capacity allows for processing up to 200 samples' data per week. Volumes above 200/week can be accommodated on an as-needed basis; please contact picard@broadinstitute.org to discuss any high volume requests.

Deliverables

A Picard BAM file and standard pipeline metrics delivered onto project-owned storage.

Input Requirements

- Illumina sequence data in either BAM or FASTQ format on a Broad file system.
- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed.
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender.

Picard Analysis of externally generated data - human whole genome (>30x)

Part Number : P-ANA-0004

Product Family : Data Analysis

Availability Date : 11/01/2012

Description

Processing of externally generated Illumina sequencing data through the platform's Picard whole genome processing pipeline to create an analysis-ready BAM file. Includes any necessary format conversion and merging of data per sample through to creation of a standard Picard BAM file per sample. Includes short indel co-cleaning where appropriate. Data storage is not included. Capacity allows for processing up to 100 samples' data per week. Volumes above 100/week can be accommodated on an as-needed basis; please contact picard@broadinstitute.org to discuss any high volume requests.

Deliverables

A Picard BAM file and standard pipeline metrics delivered onto project-owned storage.

Input Requirements

- Illumina sequence data in either BAM or FASTQ format on a Broad file system.
- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed.
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender.

EPIGENOMICS**RRBS Sequencing**

Part Number : P-EPI-0001

Product Family : Epigenomics

Availability Date : 11/01/2012

Description

RRBS sequencing includes sample plating, MspI restriction enzyme digestion, library preparation, sequencing, and data storage. This product utilizes the mRRBS method published by Patrick Boyle et al. Genome Biology (2012). Resulting RRBS libraries typically achieve a mean CPG coverage (mean coverage of CpG sites) of 15x. We have the ability to process up to 96 samples per week handled in batches of 96. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org. Minimum order size is 24 samples.

Deliverables

Data delivery will include a de-multiplexed, aggregated Picard BAM file which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage for 5 years is provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Participant ID and Collaborator Sample ID
- Genomic DNA, fresh frozen or FFPE tissue, blood, stool, saliva, slides, cell pellets, or buffy coats that preferably yield >250ng of DNA (note extra cost will be applied for extractions)
- Samples Lab Pico results from within the last year

- Samples below 250ng (5ng/uL minimum concentration) will be considered at risk, but success is possible with 50ng (5ng/uL minimum concentration) or greater.

RRBS Sequencing (low coverage)

Part Number : P-EPI-0002

Product Family : Epigenomics

Availability Date : 02/01/2013

Description

RRBS sequencing includes sample plating, MspI restriction enzyme digestion, library preparation, sequencing, and data storage. This product utilizes the "mRRBS" method published by Patrick Boyle et al. Genome Biology (2012). Resulting RRBS libraries typically achieve a mean CPG coverage (mean coverage of CpG sites) of 4x. We have the ability to process up to 96 samples per week handled in batches of 96. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org.

Minimum order size is 24 samples.

Deliverables

Data delivery will include a de-multiplexed, aggregated Picard BAM file which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage for 5 years is provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Participant ID and Collaborator Sample ID
- Genomic DNA, fresh frozen or FFPE tissue, blood, stool, saliva, slides, cell pellets, or buffy coats that preferably yield >250ng of DNA (note extra cost will be applied for extractions)
- Samples Lab Pico results from within the last year
- Samples below 250ng (5ng/uL minimum concentration) will be considered at risk, but success is possible with 50ng (5ng/uL minimum concentration) or greater.

EXOME

Standard Exome Sequencing v2

Part Number : P-EX-0005

Product Family : Exome

Availability Date : 06/01/2013

Description

The Standard Exome v2 includes sample plating, library preparation, hybrid capture, sequencing (76bp paired reads), sample identification QC check, and data storage. This product utilizes the Agilent Sure-Select Human All Exon v2.0, 44Mb baited target with the Broad in-solution hybrid selection process. Our hybrid selection libraries typically meet or exceed 80% of targets at 20x and a mean target coverage >80x. We have the ability to process up to 1472 samples per week handled in batches of 92. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org

Deliverables

Data delivery will include a de-multiplexed, aggregated Picard BAM file which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage for 5 years is provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender

- Genomic DNA, fresh frozen or FFPE tissue, blood, stool, saliva, slides, cell pellets, or buffy coats that preferably yield >250ng of DNA (note extra cost will be applied for extractions).
- Samples below 250ng (2ng/uL minimum concentration), WGA and FFPE samples will be accepted at risk, but success rates are high with 50ng (1ng/uL minimum concentration) or greater.
- Samples below 40ng are not accepted
- Tumor/Normal or Case/Control pairs must be received together if indel co-cleaning is required.
- Samples Lab Pico results from within the last year

Exome Express v2

Part Number : P-EX-0007

Product Family : Exome

Availability Date : 7/24/2013

Description

The Exome Express pipeline is a uniquely designed workflow optimized for speed and utilizes the Agilent Sure-Select Human All Exon v2.0, 44Mb baited target with the Broad in-solution hybrid selection process. Our Exome Express hybrid selection libraries meet or exceed 80% of targets at 20x The Exome Express product includes sample plating, library preparation, hybrid capture, sequencing (76bp paired reads), sample identification QC check, and data storage. We have the ability to process up to 184 samples per week handled in two batches of 92. For additional capacity please contact genomics@broadinstitute.org

Deliverables

Data delivery will include a de-multiplexed, aggregated BAM file which will be accessed via the BASS file server system or FTP for non-Broad users. Turnaround time from verified* sample receipt to aggregated BAM file generation is 21 calendar days or less. Samples that fail to meet this deliverable will be charged at the Standard Exome rate. Data storage for 5 years is also provided.

*verified sample receipt includes a validation of funding and compliance agreements

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- Genomic DNA, fresh frozen or FFPE tissue, blood, stool, saliva, slides, cell pellets, or buffy coats that preferably yield >250ng of DNA (note extra cost will be applied for extractions). Samples below 250ng (2ng/uL minimum concentration) and non-standard input materials e.g. FFPE, WGA, CTC samples will be considered at risk, but success is expected with 50ng (1ng/uL minimum concentration) or greater.
- Tumor/Normal or Case/Control pairs must be received together if indel co-cleaning is required.
- Samples below 20ng are not accepted.

Exome Plus

Part Number : P-EX-0003

Product Family : Exome

Availability Date : 11/01/2012

Description

Exome Plus includes sample plating, library preparation, hybrid capture, sequencing (76bp paired reads), sample identification QC check, and data storage. The Exome Plus utilizes the Broad-designed expanded human content, manufactured by Agilent, with ~160Mb baited target and the Broad in-solution hybrid selection process. Exome Plus includes the standard exome targets with the following additions - intronic and promoter sequences for known cancer genes, significant targets identified in Cancer GWAS studies, TCGA and the CCLE. Also included are novel exons identified in the 29 mammals comparative study, regulatory motifs from ENSEMBL as well as lincRNA sequence and additional sequence in known areas of copy number alterations. With 1 HiSeq lane, which typically yields 25Gb PF (+/- 5%) or more of delivered data, our hybrid selection libraries typically meet or exceed 80% of targets at 20x (+/- 5%). We have the

ability to process up to 1472 samples per week handled in batches of 92. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org
Minimum order size is 24 samples.

Deliverables

Data delivery will include a de-multiplexed, aggregated Picard BAM file which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage for 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- Genomic DNA, fresh frozen or FFPE tissue, blood, stool, saliva, slides, cell pellets, or buffy coats that preferably yield >250ng of DNA (note extra cost will be applied for extractions). Samples below 250ng (2ng/uL minimum concentration), WGA and FFPE samples will be accepted at risk, but success is expected with 50ng (1ng/uL minimum concentration) or greater.
- Tumor/Normal or Case/Control pairs must be received together if indel co-cleaning is required
- Samples below 250ng are not accepted.
- Samples Lab Pico results from within the last year

MICROBIAL & VIRAL ANALYSIS

Small Genome Illumina Assembly

Part Number : P-MCV-0001

Product Family : Microbial & Viral Analysis

Availability Date : 11/01/2012

Description

The Small Genome Illumina Assembly product includes plating, library preparation, sequencing (101bp paired reads), and data storage. Libraries include 1) a paired end library with 180bp +/- 10% insert size and 2) a 3-5kb jumping library. The two library types are sequenced to a total of 1.2Gb coverage (+/- 5%). This is appropriate for the assembly and annotation of small (4-6Mb) genomes. We have the ability to process up to 95 samples per week in one batch. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org. Minimum order size is 24 samples.

Deliverables

Data delivery will include a de-multiplexed, aggregated Picard BAM file which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage of 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Genus, Species, Strain, Collaborator Participant ID and Collaborator Sample ID
- Minimum input of 1.25ug or more genomic DNA. Samples below 1.25ug (minimum concentration 7ng/uL) will be accepted at risk, but success is expected with 550ng (minimum concentration 5ng/uL) or greater.
- Samples Lab Pico date from within the last year

Standard 16S Sequencing

Part Number : P-MCV-0002

Product Family : Microbial & Viral Analysis

Availability Date : 11/01/2012

Description

The Standard 16S Sequencing product includes plating, library preparation, sequencing and data storage. This provides targeted amplification of a ~250bp region of the microbial 16S gene with tailed primers (515F & 806R). QC, Pooling and 1 MiSeq (175bp paired) run per batch of 192 samples. We have the ability to process up to 192 samples per week in one batch. Processing times vary and depend on current demand. Minimum order size is 192 samples. For additional capacity please contact genomics@broadinstitute.org.

Deliverables

Data delivery will include a de-multiplexed, aggregated, unaligned BAM file which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage of 5 years is also provided

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Sample ID
- 30ul volume-based input (>5ng/uL concentration preferred) is sufficient for one attempt

RNA Sequencing & Expression**Fluidigm Custom Expression 48.48**

Part Number : P-CGE-0001

Product Family : RNA

Availability Date : 11/01/2012

Description

Fluidigm is an open microfluidics system for a variety of applications but can be used specifically for targeted gene expression for 48 to 96 genes or primers (Run cost does NOT include primers). The BioMark HD system can work with many chemistry options such as TaqMan Probes, Eva Green Double-Stranded Binding Dye, Roche Universal Probe Library, & TaqMan™ MegaPlex miRNA Assays. In addition, the Genomics Platform can also process novel, custom assays using Fluidigm's Deltagene assays. We have the ability to process up to 384 samples per week in batches of 48. Processing times vary and depend on current demand. Minimum order size is 48 samples. For additional capacity or order constraints please contact genomics@broadinstitute.org.

Deliverables

Data delivery will include a BML file and a data export file that includes the Cts for each probe across the samples. The data can be accessed via the Data and Analysis Portal or an external site.

Input Requirements

- Primer panel for targets must be provided or can be designed. GAPDH will be included as a control gene. (Run cost does NOT include primers)
- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID
- Total RNA, Tissue, Lysate, Blood, Slides that yields > 5 ng of RNA (note extra cost will be applied for extractions).

Fluidigm Custom Expression 96.96

Part Number : P-CGE-0002

Product Family : RNA

Availability Date : 11/01/2012

Description

Fluidigm is an open microfluidics system for a variety of applications but can be used specifically for targeted gene expression for 96 to 384 genes or primers. The BioMark HD system can work with many chemistry options such as TaqMan Probes, Eva Green Double-Stranded Binding Dye, Roche Universal Probe Library, & TaqMan™ MegaPlex miRNA Assays. In addition, the Genomics Platform can also process novel, custom assays using Fluidigm's Deltagene assays. We have the ability to process up to 384 samples per week 4 batches of 96 or 8 batches of 48. Processing times vary and depend on current demand. Minimum order size is 48 samples. For additional capacity or order constraints please contact genomics@broadinstitute.org.

Deliverables

Data delivery will include a BML file and a data export file that includes the Cts for each probe across the samples. The data can be accessed via the Data and Analysis Portal or an external site.

Input Requirements

- Primer panel for targets must be provided or can be designed. GAPDH will be included as a control gene. (Run cost does not include primers).
- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID
- Total RNA, Tissue, Lysate, Blood, Slides that yields > 5 ng of RNA.

Standard RNA Sequencing - Low Coverage (15M pairs)

Part Number : P-RNA-0001

Product Family : RNA

Availability Date : 11/01/2012

Description

Standard RNA Sequencing includes plating, poly-A selection and cDNA synthesis, library preparation, sequencing (76 bp paired reads), sample identification QC check (when Sample Qualification of a matching DNA sample is chosen), and data storage. The product provides library construction using a non-strand specific Illumina TruSeq Protocol and sequence coverage to 15M Paired reads or 15M Total reads (+/- 5%) (depending on the reference sequence supplied). We have the ability to process up to 190 samples per week in batches of 95. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org

Deliverables

Data delivery will include a de-multiplexed, aggregated Picard BAM file which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage of 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- RNA with a RIN (or equivalent QC metric) score below 7 will be attempted on risk.
- RNA, tissue, blood, stool, or cell pellets that preferably yield >250ng of RNA (note extra cost will be applied for extractions). Samples below 250ng (minimum concentration 5ng/uL) and FFPE samples will be accepted at risk, but success is expected with 100ng (minimum concentration 2ng/uL) or greater.

Standard RNA Sequencing - High Coverage (50M pairs)

Part Number : P-RNA-0002

Standard RNA Sequencing - High Coverage (50M pairs)

Product Family : RNA

Availability Date : 11/01/2012

Description

Standard RNA Sequencing includes plating, poly-A selection and cDNA synthesis, library preparation, sequencing (76 bp paired reads), sample identification QC check (when Sample Qualification of a matching DNA sample is chosen), and data storage. The product provides library construction using a non-strand specific Illumina TruSeq Protocol and sequence coverage to 50M Paired reads or 50M Total reads (+/- 5%) (depending on the reference sequence supplied). We have the ability to process up to 190 samples per week in batches of 95. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org

Deliverables

Data delivery will include a de-multiplexed, aggregated Picard BAM file which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage of 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- RNA with a RIN (or equivalent QC metric) score below 7 will be attempted on risk.
- RNA, tissue, blood, stool, or cell pellets that preferably yield >250ng of RNA (note extra cost will be applied for extractions). Samples below 250ng (minimum concentration 5ng/uL) and FFPE samples will be accepted at risk, but success is expected with 100ng (minimum concentration 2ng/uL)

Strand Specific RNA Sequencing - Low Coverage (15M pairs)

Part Number : P-RNA-0003

Product Family : RNA

Availability Date : 11/01/2012

Description

Strand Specific RNA Sequencing includes plating, poly-A selection and strand specific cDNA synthesis, library preparation, sequencing (101 bp paired reads), sample identification QC check (when Sample Qualification of a matching DNA sample is chosen), and data storage. The product provides library construction using a stranded dUTP Protocol and sequence coverage to 15M Paired reads or 15M Total reads (+/- 5%) (depending on the reference sequence supplied). We have the ability to process up to 95 samples per week in one batch. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org

Deliverables

Data delivery will include a de-multiplexed, aggregated Picard BAM file which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage of 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- RNA with a RIN (or equivalent QC metric) score below 7 will be attempted on risk.
- RNA, tissue, blood, stool, or cell pellets that preferably yield >5ug of RNA (note extra cost will be applied for extractions). Samples below 5ug (minimum concentration 100ng/uL) and FFPE samples will be accepted at risk, but success is expected with 4ug (minimum concentration 80 ng/uL) or greater.

Strand Specific RNA Sequencing - High Coverage (50M pairs)

Part Number : P-RNA-0004

Product Family : RNA

Availability Date : 11/01/2012

Description

Strand Specific RNA Sequencing includes plating, poly-A selection and strand specific cDNA synthesis, library preparation, sequencing (101 bp paired reads), sample identification QC check (when Sample Qualification of a matching DNA sample is chosen), and data storage. The product provides library construction using a stranded dUTP Protocol and sequence coverage to 50M Paired reads or 50M Total reads (+/- 5%) (depending on the reference sequence supplied). We have the ability to process up to 95 samples per week in one batch. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org

Deliverables

Data delivery will include a de-multiplexed, aggregated Picard BAM file which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage of 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed.
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- RNA with a RIN (or equivalent QC metric) score below 7 will be attempted on risk.
- RNA, tissue, blood, stool, or cell pellets that preferably yield >5ug of RNA (note extra cost will be applied for extractions). Samples below 5ug (minimum concentration 100ng/uL) and FFPE samples will be accepted at risk, but success is expected with 4ug (minimum concentration 80ng/uL) or greater.

Tru-Seq Strand Specific RNA Sequencing - Low Coverage (15M pairs)

Part Number : P-RNA-0007

Product Family : RNA

Availability Date : 07/01/2013

Description

Tru-Seq Strand Specific RNA Sequencing includes plating, poly-A selection and cDNA synthesis, library preparation, sequencing, sample identification QC check (when Sample Qualification of a matching DNA sample is chosen), and data storage. The product provides library construction using a strand specific Illumina TruSeq Protocol and sequence coverage to 15M Paired reads or 15M Total reads (+/- 5%) (depending on the reference sequence supplied). We have the ability to process up to 190 samples per week in batches of 95. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org.

Deliverables

Data delivery will include a de-multiplexed, aggregated Picard BAM file which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage of 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- RNA with a RIN (or equivalent QC metric) score below 7 will be attempted on risk.
- RNA, tissue, blood, stool, or cell pellets that preferably yield > 250ng of RNA (note extra cost will be applied for extractions). Samples below 250ng (minimum concentration 5ng/uL) and FFPE samples will be accepted at risk, but success is expected with 100ng (minimum concentration 2ng/uL) or greater.

Tru-Seq Strand Specific RNA Sequencing - High Coverage (50 M pairs)

Part Number : P-RNA-0008

Product Family : RNA

Availability Date : 07/01/2013

Description

Tru-Seq Strand Specific RNA Sequencing includes plating, poly-A selection and cDNA synthesis, library preparation, sequencing, sample identification QC check (when Sample Qualification of a matching DNA sample is chosen), and data storage. The product provides library construction using a strand specific Illumina TruSeq Protocol and sequence coverage to 50M Paired reads or 50M Total reads (+/- 5%) (depending on the reference sequence supplied). We have the ability to process up to 190 samples per week in batches of 95. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org.

Deliverables

Data delivery will include a de-multiplexed, aggregated Picard BAM file which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage of 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- RNA with a RIN (or equivalent QC metric) score below 7 will be attempted on risk.
- RNA, tissue, blood, stool, or cell pellets that preferably yield > 250ng of RNA (note extra cost will be applied for extractions). Samples below 250ng (minimum concentration 5ng/uL) and FFPE samples will be accepted at risk, but success is expected with 100ng (minimum concentration 2ng/uL) or greater.

SAMPLE INITIATION, QUALIFICATION & CELL CULTURE

Sample Initiation

Part Number : P-ESH-0001

Product Family : Sample Initiation, Qualification & Cell Culture

Availability Date : 11/01/2012

Description

Sample Initiation includes the registration of kits/samples and storage upon receipt from collaborator, as well as the upload and linking of corresponding sample information, participant phenotypic data, and sample phenotypic data. Incoming DNA and RNA samples will receive sample quantification (PicoGreen for DNA, BioAnalyzer/Nanodrop for RNA - if requested) and normalization to a standard Genomics Platform working concentration appropriate for the relevant downstream workflows. We have the ability to process up to 10,000 samples per week. All samples entering the Broad Genomics Platform will go through the sample initiation process.

Deliverables

Sample receipt confirmation, sample and phenotypic data upload and sample quantification.

Input Requirements

- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender, Genus and Species where applicable.

Sample Qualification Only (Human Samples)

Part Number : P-ESH-0002

Product Family : Sample Initiation, Qualification & Cell Culture

Availability Date : 11/01/2012

Description

Sample qualification includes plating and Fluidigm Fingerprinting. The Fluidigm fingerprint panel includes 29 SNPs that overlap with the Affy 6.0 array and have multiple proxy SNPs each, 66 SNPs that overlap with Illumina's 1m and 2.5m arrays and have multiple proxy SNPs each, 32 SNPs in transcribed regions of housekeeping genes that are expressed in most cell types and 1 gender determining SNP. We have the ability to process up to 4800 samples per week handled in batches of 960. Processing times vary and depend on current demand. This product is appropriate where a fingerprint is required for sample selection from a new or existing cohort prior to choosing a downstream product. Fingerprints are no

longer required prior to sequencing. Only choose this product when you require a pause after the fingerprint to review the data and choose which samples should go for downstream processing. Note: All downstream sequencing products include a fingerprint which will be compared to the final data output for an identity QC check. We will only fingerprint your samples once.

Deliverables

Fluidigm fingerprint data (.xls format) is available via the Genomics Platform Samples (BSP) file system.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- 25ng of input DNA

WGA

Part Number : P-ESH-0003

Product Name : WGA

Product Family : Sample Initiation, Qualification & Cell Culture

Availability Date : 11/01/2012

Description

The WGA provides Whole Genome Amplification and clean up for DNA samples with low yields. A minimum input of 5-40ng of high molecular weight DNA is required in order to have a successful Whole Genome Amplification. Whole Genome Amplification is performed using the Qiagen REPLI-g WGA Midi Kit. The Whole Genome Amplified material is the "cleaned" to remove primers and random hexomers by using Ultra Filtration. We are able to process up to 384 samples per week handled in batches of 96. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org.

Deliverables

15-25ug of Whole Genome Amplified DNA free of excess primers and hexamers quantified in triplicate via picogreen.

Input Requirements

- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- At least 5ng of high molecular weight DNA

DNA Extract from blood, fresh frozen tissue, cell pellet, stool, or saliva

Part Number : P-ESH-0004

Product Family : Sample Initiation, Qualification & Cell Culture

Availability Date : 11/01/2012

Description

This product is appropriate for extraction from blood (fresh or frozen), cells, and tissue (fresh, snap-frozen), cell pellets, stool, or saliva. DNA is extracted using a column-based DNeasy Kit. The samples are first lysed with Proteinase K. Buffering conditions are adjusted so to provide optimal DNA binding conditions to the DNeasy spin column. Once the lysed sample is added to the column, DNA is selectively bound to the column membrane as contaminants and enzyme inhibitors pass through in the wash steps. The DNA is then eluted off the column with TE buffer and is ready to be quantified via picogreen. Processing times vary and depend on current demand. We are able to process up to 360 samples per week handled in batches of 12. For additional capacity please contact genomics@broadinstitute.org.

Deliverables

DNA quantified in triplicate using a standardized PicoGreen assay. Sample yield and quality are dependent on multiple factors such as: the original material type provided (blood, cells, tissue, etc), amount of material provided, and tissue site and quality.

Input Requirements

- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender

DNA and RNA Extract from fresh frozen tissue or stool (AllPrep)

Part Number : P-ESH-0005

Product Family : Sample Initiation, Qualification & Cell Culture

Availability Date : 11/01/2012

Description

This product is appropriate for co-extraction of DNA and RNA from fresh frozen tissue, blood, or stool, or FFPE. The AllPrep DNA/RNA Mini Kit is designed for purifying both genomic DNA and total RNA. Since there is no need to divide the sample into two for separate purification procedures, maximum yields of DNA and RNA can be achieved. The purified DNA and RNA are eluted separately and ready to use in any downstream application. We are able to process up to 360 samples per week handled in batches of 12. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org.

Deliverables

DNA quantified in triplicate using a standardized PicoGreen assay. Sample yield and quality are dependent on multiple factors such as: the original material type provided (blood, cells, tissue, etc), amount of material provided, and tissue site. RNA quantified via Nanodrop along with 260/280, 260/230 purity ratios. RNA integrity is measured by RIN value as determined by Agilent Bioanalyzer.

Input Requirements

- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
Expected Cycle Time (Days)

DNA Extract from FFPE or Slides

Part Number : P-ESH-0007

Product Family : Sample Initiation, Qualification & Cell Culture

Availability Date : 11/01/2012

Description

This product is appropriate for Extraction of DNA from Formalin-Fixed-Paraffin-Embedded Tissue cores, scrolls, and sections on slides. Once the samples are de-paraffinized, the DNA is extracted using a column-based DNeasy Kit. The samples are first lysed with Proteinase K. Buffering conditions are adjusted so to provide optimal DNA binding conditions to the DNeasy spin column. Once the lysed sample is added to the column, DNA is selectively bound to the column membrane as contaminants and enzyme inhibitors pass through in the wash steps. The DNA is then eluted off the column with TE buffer and is ready to be quantified via picogreen. We are able to process up to 96 samples per week handled in batches of 12. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org.

Deliverables

DNA quantified in triplicate using a standardized PicoGreen assay. Sample yield and quality are dependent on multiple factors such as: the original material type provided (blood, cells, tissue, etc), amount of material provided, and tissue site.

Input Requirements

- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender

RNA Extract from FFPE

Part Number : P-ESH-0008

Product Family : Sample Initiation, Qualification & Cell Culture

Availability Date : 11/01/2012

Description

This product is appropriate for high quality RNA extractions from Formalin-Fixed-Paraffin-Embedded Tissue. RNA is extracted using a column-based RNeasy Kit. Special lysis and incubation conditions reverse

formaldehyde modification of RNA. In addition, the lysis buffer efficiently releases RNA from tissue sections while avoiding further RNA degradation. The kit also uses DNase and DNase Booster Buffer for optimized removal of genomic DNA contamination. RNeasy MinElute spin columns enable purification of total RNA with elution volumes of as low as 10ul. We are able to process up to 360 samples per week handled in batches of 12. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org.

Deliverables

RNA quantified via Nanodrop along with 260/280, 260/230 purity ratios. RNA integrity is measured by RIN value as determined by an Agilent Bioanalyzer.

Input Requirements

- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender

External Plating into Tubes

Part Number : P-ESH-0009

Product Family : Sample Initiation, Qualification & Cell Culture

Availability Date : 11/01/2012

Description

This product is appropriate when you wish to send samples anywhere outside of the Genomics Platform. Samples are cherry-picked in a specified order or randomly picked in no specific order (determined by the collaborator). These samples are either plated at a specific concentration and volume in 2D bar-coded Matrix tubes using a calibrated liquid handler or the entire stock tube is returned to the collaborator. We are able to process up to 1000 samples per week. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org.

Deliverables

DNA and/or RNA aliquoted at a specific concentration and volume into 2D bar-coded Matrix tubes. These tubes are shipped to the collaborator on dry ice or ice packs along with sample maps, sample and phenotypic data that correspond to each sample in the shipment.

Input Requirements

- List of Samples to return, tube type specification, and an address and contact information to the person/institute the samples will be shipped to as well as a FedEx account number to ship the samples.

External Plating into Plates

Part Number : P-ESH-0010

Product Family : Sample Initiation, Qualification & Cell Culture

Availability Date : 11/01/2012

Description

This product is appropriate when you wish to send samples anywhere outside of the Genomics Platform. Samples are cherry-picked in a specified order or randomly picked in no specific order (determined by the collaborator). These samples are either plated at a specific concentration and volume in barcoded 96 well plates using calibrated liquid handling automation. We are able to process up to 1000 samples per week. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org.

Deliverables

DNA and/or RNA aliquotted at a specific concentration and volume into bar-coded 96 well plates (various well sizes available). These plates are tightly sealed with an adhesive plate cover and shipped back to the collaborator on dry ice or ice packs along with sample maps, sample and phenotypic data that correspond to each sample in the shipment.

Input Requirements

List of Samples to return, plate specification, and an address and contact information to the person/institute the samples will be shipped to as well as a FedEx account number to ship the samples.

Viable Cell Line Retrieval

Part Number : P-ESH-0011

Product Family : Sample Initiation, Qualification & Cell Culture

Availability Date : 01/01/2013

Description

Retrieval of existing cell line.

SEQUENCE ONLY

HiSeq 2x25 Paired Lane

Part Number : P-SEQ-0001

Product Family : Sequence Only

Availability Date : 11/01/2012

Description

Includes quantification, denaturation, and sequence coverage for a High Quality Illumina prepared library with low adapter dimer, using the Broad (8base)/Illumina (6base) compatible indices. Low-Quality or Base-biased samples will be accepted on risk please contact the Product Manager or genomics@broadinstitute.org for information regarding these samples

Deliverables

Data delivery to a minimum ~100M Total Raw Reads and will include a de-multiplexed, aggregated, aligned BAM file (when ref seq provided) which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage of 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Sample ID and reference sequence where known.
- Broad (8base) / Illumina (6base) compatible indices
- Library with concentration of 2nM or greater in at least 20ul total volume. Median fragment size (adapter lengths included) must be provided. Libraries below 2nM in 20uL total volume will be accepted on risk, but success is expected with 0.2nM or greater in 20uL.
- Samples not meeting these barcode, concentration and/ or volume requirements will be accepted on risk

HiSeq 44 Single Lane

Part Number : P-SEQ-0002

Product Family : Sequence Only

Availability Date : 11/01/2012

Description

Includes quantification, denaturation, and sequence coverage for a High Quality Illumina prepared library with low adapter dimer, using the Broad (8base)/Illumina (6base) compatible indices. Low-Quality or Base-biased samples will be accepted on risk please contact the Product Manager or genomics@broadinstitute.org for information regarding these samples.

Deliverables

Data delivery to a minimum ~ 100M Total Raw Reads and will include a de-multiplexed, aggregated, aligned BAM file (when ref seq provided) which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage of 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Sample ID and reference sequence where known.

- Broad (8base) / Illumina (6base) compatible indices
- Library with concentration of 2nM or greater in at least 20ul total volume. Median fragment size (adapter lengths included) must be provided. Libraries below 2nM in 20uL total volume will be accepted on risk, but success is expected with 0.2nM or greater in 20uL.
 - Samples not meeting these barcode, concentration and/ or volume requirements will be accepted on risk

HiSeq 2x76 Paired Lane

Part Number : P-SEQ-0003

Product Family : Sequence Only

Availability Date : 11/01/2012

Description

Includes quantification, denaturation, and sequence coverage for a High Quality Illumina prepared library with low adapter dimer, using the Broad (8base)/Illumina (6base) compatible indices. Low-Quality or Base-biased samples will be accepted on risk please contact the Product Manager or genomics@broadinstitute.org for information regarding these samples.

Deliverables

Data delivery to a minimum ~100M Total Raw Reads and will include a de-multiplexed, aggregated, aligned BAM file (when ref seq provided) which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage of 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Sample ID and reference sequence where known.
- Broad (8base) / Illumina (6base) compatible indices
- Library with concentration of 2nM or greater in at least 20ul total volume. Median fragment size (adapter lengths included) must be provided. Libraries below 2nM in 20uL total volume will be accepted on risk, but success is expected with 0.2nM or greater in 20uL.
 - Samples not meeting these barcode, concentration and/ or volume requirements will be accepted on risk

HiSeq 2x101 Paired Lane

Part Number : P-SEQ-0004

Product Family : Sequence Only

Availability Date : 11/01/2012

Description

Includes quantification, denaturation, and sequence coverage for a High Quality Illumina prepared library with low adapter dimer, using the Broad (8base)/Illumina (6base) compatible indices. Low-Quality or Base-biased samples will be accepted on risk please contact the Product Manager or genomics@broadinstitute.org for information regarding these samples.

Deliverables

Data delivery to a minimum ~100M Total Raw Reads and will include a de-multiplexed, aggregated, aligned BAM file (when ref seq provided) which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage of 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Sample ID and reference sequence where known.
- Broad (8base) / Illumina (6base) compatible indices

- Library with concentration of 2nM or greater in at least 20ul total volume. Median fragment size (adapter lengths included) must be provided. Libraries below 2nM in 20uL total volume will be accepted on risk, but success is expected with 0.2nM or greater in 20uL
- Samples not meeting these barcode, concentration and/ or volume requirements will be accepted on risk

MiSeq Short Run

Part Number : P-SEQ-0005

Product Family : Sequence Only

Availability Date : 11/01/2012

Description

Includes quantification, denaturation, and sequence coverage for a High Quality Illumina prepared library with low adapter dimer, using the Broad (8base)/Illumina (6base) compatible indices for up to 50 total cycles. Low-Quality or Base-biased samples will be accepted on risk please contact the Product Manager or genomics@broadinstitute.org for information regarding these samples.

Deliverables

Data delivery to a minimum ~5M Total Raw Reads and will include a de-multiplexed, aggregated, aligned BAM file (when ref seq provided) which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage of 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Sample ID and reference sequence where known.
- Broad (8base) / Illumina (6base) compatible indices
- Library with concentration of 2nM or greater in at least 20ul total volume. Median fragment size (adapter lengths included) must be provided. Libraries below 2nM in 20uL total volume will be accepted on risk, but success is expected with 0.2nM or greater in 20uL.
 - Samples not meeting these barcode, concentration and/ or volume requirements will be accepted on risk

MiSeq Long Run

Part Number : P-SEQ-0006

Product Family : Sequence Only

Availability Date : 11/01/2012

Description

Includes quantification, denaturation, and sequence coverage for a High Quality Illumina prepared library with low adapter dimer, using the Broad (8base)/Illumina (6base) compatible indices for up to 300 total cycles. Low-Quality or Base-biased samples will be accepted on risk please contact the Product Manager or genomics@broadinstitute.org for information regarding these samples.

Deliverables

Data delivery to a minimum ~5M Total Raw Reads and will include a de-multiplexed, aggregated, aligned BAM file (when ref seq provided) which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage of 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Sample ID and reference sequence where known.
- Broad (8base) / Illumina (6base) compatible indices

- Library with concentration of 2nM or greater in at least 20ul total volume. Median fragment size (adapter lengths included) must be provided. Libraries below 2nM in 20uL total volume will be accepted on risk, but success is expected with 0.2nM or greater in 20uL.
 - Samples not meeting these barcode, concentration and/or volume requirements will be accepted on risk

MiSeq Extra Long Run

Part Number : P-SEQ-0007

Product Family : Sequence Only

Availability Date : 11/01/2012

Description

Includes quantification, denaturation, and sequence coverage for a High Quality Illumina prepared library with low adapter dimer, using the Broad (8base)/Illumina (6base) compatible indices for up to 500 total cycles. Low-Quality or Base-biased samples will be accepted on risk please contact the Product Manager or genomics@broadinstitute.org for information regarding these samples.

Deliverables

Data delivery to a minimum ~5M Total Raw Reads and will include a de-multiplexed, aggregated, aligned BAM file (when ref seq provided) which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage of 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Sample ID and reference sequence where known.
- Broad (8base) / Illumina (6base) compatible indices
- Library with concentration of 2nM or greater in at least 20ul total volume. Median fragment size (adapter lengths included) must be provided. Libraries below 2nM in 20uL total volume will be accepted on risk, but success is expected with 0.2nM or greater in 20uL.
- Samples not meeting these barcode, concentration and/ or volume requirements will be accepted on risk

HiSeq 2500 Paired Lane (>50 Cycle)

Part Number : P-SEQ-0008

Product Family : Sequence Only

Availability Date : 01/01/2013

Description

Includes quantification, denaturation, and sequence coverage for a High Quality Illumina prepared library with low adapter dimer, using the Broad (8base)/Illumina (6base) compatible indices. Low-Quality or Base-biased samples will be accepted on risk please contact the Product Manager or genomics@broadinstitute.org for information regarding these samples.

Deliverables

Data delivery to a minimum ~100M Total Raw Reads and will include a de-multiplexed, aggregated, aligned BAM file (when ref seq provided) which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage of 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Sample ID and reference sequence where known.
- Broad (8base) / Illumina (6base) compatible indices

- Library with concentration of 2nM or greater in at least 20ul total volume. Median fragment size (adapter lengths included) must be provided. Libraries below 2nM in 20uL total volume will be accepted on risk, but success is expected with 0.2nM or greater in 20uL.
- Samples not meeting these barcode, concentration and/ or volume requirements will be accepted on risk

HiSeq 2500 Paired Lane (<=50 Cycle)

Part Number : P-SEQ-0009

Product Family : Sequence Only

Availability Date : 03/01/2013

Description

Includes quantification, denaturation, and sequence coverage for a High Quality Illumina prepared library with low adapter dimer, using the Broad (8base)/Illumina (6base) compatible indices. Low-Quality or Base-biased samples will be accepted on risk please contact the Product Manager or genomics@broadinstitute.org for information regarding these samples.

Deliverables

Data delivery to a minimum ~100M Total Raw Reads and will include a de-multiplexed, aggregated, aligned BAM file (when ref seq provided) which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage of 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Sample ID and reference sequence where known.
- Broad (8base) / Illumina (6base) compatible indices
- Library with concentration of 2nM or greater in at least 20ul total volume. Median fragment size (adapter lengths included) must be provided. Libraries below 2nM in 20uL total volume will be accepted on risk, but success is expected with 0.2nM or greater in 20uL.
- Samples not meeting these barcode, concentration and/ or volume requirements will be accepted on risk.

SMALL TARGET SEQUENCING

Fluidigm Access Array Run

Part Number : P-VAL-0005

Product Family : Small Design, Validation & Extension

Availability Date : 06/01/2013

Description

Fluidigm Custom PCR includes plating, PCR amplification of 47 samples across 48 (singleplex) or 480 (multiplex) primer pairs (Primer cost and Sequencing NOT included). For any other combinations of samples and primers, please contact the Product Manager. We have the ability to process up to 188 samples per week in minimum batch sizes of 47 or multiples thereof. For additional capacity or order constraints please contact genomics@broadinstitute.org.

Deliverables:

Pooled, multiplexed library that is ready for sequencing on Illumina MiSeq Instrument.

(Data delivery will include a de-multiplexed, aggregated BAM file which will be accessed via the BASS file server system.)

Input Requirements

- Primer panel for targets must be provided or can be designed for a fee. Run cost does **NOT** include primers.
- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed.
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender.

- Genomic DNA, fresh frozen or FFPE tissue, blood, stool, saliva, slides, cell pellets, or buffy coats that preferably yield >250ng of DNA or greater (note extra cost will be applied for extractions). Samples below 250ng (40ng/uL minimum concentration) and FFPE samples will be accepted at risk, but success rates are high with 25ng (5ng/uL minimum concentration) or greater. Samples Lab Pico results from within the last year.

TSCA (<2500 amplicons)

Part Number : P-VAL-0002

Product Family : Small Design, Validation & Extension

Availability Date : 11/01/2012

Description

TSCA (Illumina TruSeq Custom Amplicon) includes plating, PCR amplification of up to 2500 amplicons, 250 bases in length, (Primer cost NOT included), sequencing at 2x150bases, and data storage. For any other combinations of samples and primers, please contact the Product Manager. This product is best suited for targeted projects with large numbers of samples. We have the ability to process up to 960 samples per week in batches of 96. Processing times vary and depend on current demand. For additional capacity or order constraints please contact genomics@broadinstitute.org.

Deliverables

Data delivery will include a de-multiplexed, aggregated BAM file which will be accessed via the BASS file server system.

Input Requirements

- List of coordinates of interest (Run cost does NOT include primers)
- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- Genomic DNA, fresh frozen or FFPE tissue, blood, stool, saliva, slides, cell pellets, or buffy coats that preferably yield >250ng of DNA (note extra cost will be applied for extractions). Samples below 250ng (10ng/uL minimum concentration) and FFPE samples will be accepted at risk, but success rates are high with 75ng (7.5ng/uL minimum concentration) or greater. Samples Lab Pico results from within the last year.

Custom Hybrid Selection (<1Mb)

Part Number : P-VAL-0003

Product Family : Small Design, Validation & Extension

Availability Date : 11/01/2012

Description

Custom Hybrid Selection includes sample plating, library preparation, hybrid capture, sequencing, sample identification QC check, and data storage (Custom Bait cost NOT included). This product utilizes the Agilent Sure-Select Design tools with the Broad in-solution hybrid selection process. This product is appropriate for target sizes <1Mb. For larger size targets, please contact the Product Manager. We have the ability to process up to 1472 samples per week handled in batches of 95. Processing times vary and depend on current demand. Minimum order size is 24 samples. For additional capacity or order constraints please contact genomics@broadinstitute.org.

Deliverables

Data delivery will include a de-multiplexed, aggregated BAM file which will be accessed via the BASS file server system.

Input Requirements

- List of coordinates of interest (Custom Bait cost NOT included)
- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender

- Genomic DNA, fresh frozen or FFPE tissue, blood, stool, saliva, slides, cell pellets, or buffy coats that preferably yield >250ng of DNA (note extra cost will be applied for extractions). Samples below 250ng (2ng/uL minimum concentration) and FFPE samples will be accepted at risk, but success rates are high with 50ng (1ng/uL minimum concentration) or greater. Samples Lab Pico results from within the last year.
- Tumor/Normal or Case/Control pairs must be received together if indel co-cleaning is required.

Fluidigm Custom Genotyping 96.96

Part Number : P-CGE-0003

Product Family : Small Design, Validation & Extension

Availability Date : 11/01/2012

Description

Fluidigm is an open microfluidics system for a variety of applications but can be used specifically for targeted genotyping for 96 to 192 384 genes variants. The BioMark HD system can work with many chemistry options such as TaqMan Probes, Fluidigm SNPTYPE assays, or LGC Genomic KASP Technology. The Genomics Platform can process previously designed assays as well as novel, custom assays. We have the ability to process up to 3800 samples per week. Processing times vary and depend on current demand. Minimum order size is one chip. For additional capacity or order constraints please contact genomics@broadinstitute.org.

Deliverables

Data delivered will consist of the following raw and called genotype formats: BML file and Plink formatted files and will be made available via the analysis portal. All genotypes will be called using Fluidigm proprietary software. Data storage is also provided.

Input Requirements

- Primer panel for targets must be provided or can be designed. (Run cost does not include primers)
- Funding and compliance requirements must be in place - this includes a valid IRB or a non-engagement letter where needed.
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID.
- One empty is required per plate
- Samples with lab pico results within the last year
- At least 25 ng of DNA (per sample) of input
- Volume: 2.5ul, Concentration: 10 ng/ul

Sequenom Custom Genotyping

Part Number : P-CGE-0004

Product Family : Small Design, Validation & Extension

Availability Date : 11/01/2012

Description

Mass Spectrometry genotyping technology utilizes AssayDesigner v.3.1 software to design PCR and extension primers for low and high multiplex SNP and IN/DEL assays. SNPs are amplified in multiplex PCR reactions consisting of between one SNP (hME reaction) to a maximum 36 loci (iPlex Gold reaction) each (Run cost does NOT include primers). We have the ability to process up to 3,800 samples per week handled in batches of 95. Processing times vary and depend on current demand. For additional capacity or order constraints please contact genomics@broadinstitute.org.

Deliverables

Data delivered will consist of the Plink formatted files and will be made available via the analysis portal. All genotypes will be called using Sequenom SpectroTyper. Data storage is also provided.

Input Requirements

- Primer panel for targets must be provided or can be designed. (Run cost does not include primers).

- Design input file requires 2 columns: SNP_ID and sequence 150 bases on each side of the SNP e.g. ACTCACAGGG[C/T]GCTTGCCGAGGGA
- Funding and compliance requirements must be in place - this includes a valid IRB or a non-engagement letter where needed.
- Minimum sample data including - Collaborator Participant ID, Collaborator Sample ID.
- One empty is required per plate
- Samples with lab pico results within the last year
- Sample Requirements: Sequenom HME - 5ng per pool at 2.5ng/ul, 25ul + 2ul per pool
- Sample Requirements: Sequenom iPLEX - 10ng per pool at 5ng/ul, 20ul + 2ul per pool

WHOLE GENOME SEQUENCING

Standard Light Coverage Whole Genome Sequencing (6x)

Part Number : P-WG-0001

Product Family : Whole Genome

Availability Date : 11/01/2012

Description

Standard Light Coverage Whole Genome Sequencing (6x) includes plating, library preparation, size selection, sequencing (101bp paired reads), sample identification QC check, and data storage. The product provides one size selected library (typical insert size of library is 385bp +/- 20%) and sequence coverage to 20Gb total aligned (+/- 5%). This product is appropriate for the generation of low coverage human and mammalian genomes. We have the ability to process up to 190 samples per week in batches of 95. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org

Deliverables

Data delivery will include a de-multiplexed, aggregated and aligned Picard BAM file which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage for 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- Genomic DNA, fresh frozen or FFPE tissue, blood, stool, saliva, slides, cell pellets, or buffy coats that preferably yield >250ng of DNA (note extra cost will be applied for extractions). Samples below 250ng (2ng/uL minimum concentration) and FFPE samples will be accepted at risk, but success is expected with 50ng (1ng/uL minimum concentration) or greater.
 - Samples Lab Pico results from within the last year
- Tumor/Normal or Case/Control pairs must be received together if indel co-cleaning is required.

Standard High Coverage Whole Genome Sequencing (30x)

Part Number : P-WG-0002

Product Family : Whole Genome

Availability Date : 11/01/2012

Description

Standard High Coverage Whole Genome Sequencing (30x) includes plating, library preparation, size selection, sequencing (101bp paired reads), sample identification QC check, and data storage. The product provides two size selected libraries (typical insert sizes of libraries are 325bp and 355bp, +/- 20%) and sequence coverage to 95Gb total aligned (+/- 5%). This product is appropriate for the generation of high coverage human and mammalian genomes. We have the ability to process up to 95 samples per week in

batches of 95. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org

Deliverables

Data delivery will include a de-multiplexed, aggregated and aligned Picard BAM file which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage for 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- Genomic DNA, fresh frozen or FFPE tissue, blood, stool, saliva, slides, cell pellets, or buffy coats that preferably yield >250ng of DNA (note extra cost will be applied for extractions). Samples below 250ng (2ng/uL minimum concentration) and FFPE samples will be accepted at risk, but success is expected with 75ng (1ng/uL minimum concentration) or greater.
- Samples Lab Pico results from within the last year
- Tumor/Normal or Case/Control pairs must be received together if indel co-cleaning is required

Standard Deep Coverage Whole Genome Sequencing (50x)

Part Number : P-WG-0021

Product Family : Whole Genome

Availability Date : 05/01/2013

Description

Standard Deep Coverage Whole Genome Sequencing (50x) includes plating, library preparation, size selection, sequencing (101bp paired reads), sample identification QC check, and data storage. The product provides two size selected libraries (insert sizes of libraries are 325bp and 355bp, +/- 20%) and sequence coverage to 160Gb total aligned (+/- 5%). This product is appropriate for the generation of deep coverage human and mammalian genomes. We have the ability to process up to 95 samples per week in batches of 95. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org

Deliverables

Data delivery will include a de-multiplexed, aggregated Picard BAM file which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage for 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- Genomic DNA, fresh frozen or FFPE tissue, blood, stool, saliva, slides, cell pellets, or buffy coats that preferably yield >250ng of DNA (note extra cost will be applied for extractions). Samples below 250ng (2ng/uL minimum concentration) and FFPE samples will be accepted at risk, but success is expected with 75ng (1ng/uL minimum concentration) or greater.
- Samples Lab Pico results from within the last year
- Tumor/Normal or Case/Control pairs must be received together if indel co-cleaning is required.

Non-Human Whole Genome Sequencing (4-6Mb Genome)

Part Number : P-WG-0004

Product Family : Whole Genome

Availability Date : 11/01/2012

Description

Non-Human Whole Genome Sequencing includes plating, library preparation, sequencing (101bp paired reads) and data storage. The product provides a non-size selected library and sequencing coverage up to

300Mb (+/- 5%). This is appropriate for microbial and other small genome non-assembly needs such as SNP calling. We are able to process up to 95 samples per week handled in batches of 95. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org. Minimum order size is 24 samples.

Deliverables

Data delivery will include a de-multiplexed, aggregated and aligned (where ref seq is provided) Picard BAM file which will be accessed via the BASS file server system or FTP for non-Broad users. Data storage for 5 years is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or letter of non-engagement where needed (if sample is human derived)
- Minimum Sample data including - Genus, Species, Collaborator Participant ID and Collaborator Sample ID
- 250ng or more of genomic DNA (5ng/uL minimum concentration). Samples below 250ng will be accepted at risk, but success is expected with 50ng (1ng/uL minimum concentration) or greater.
- Samples Lab Pico date from within the last year.

WHOLE GENOME ARRAY

Affy 6.0 Array Processing

Part Number : P-WG-0024

Product Family : Whole Genome

Availability Date : 05/01/2013

Description

The Affy 6.0 Array includes sample plating, array processing, sample, primary analysis, and data storage (ARRAY NOT INCLUDED). The Genome-Wide Human SNP Array 6.0 contains 906,600 SNPs and more than 946,000 probes for the detection of copy number variation on a single genotyping array. The Genome-Wide Human SNP Array 6.0 uses a robust Birdsuite calling pipeline that delivers SNP as well as CNV calls shortly after processing in the laboratory. We are able to process up to 384 arrays in 2 weeks with batches of 4 plates of 96 samples. Processing times vary and depend on current demand. For additional capacity, please contact genomics@broadinstitute.org.

Deliverables

Data can be delivered as a raw data file (.cel) or as a called data set using PLINK formatted files (.bed,.bim,.ped). Examples of information that can be passed along with the called genotypes are files listing sample information, Contrast QC Values, Signature SNP genotypes and CHP and SNP Summary Data. Data storage is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or a non-engagement letter where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- Samples with lab pico results within the last year
- At least 500ng of DNA (per sample) of input: Volume: 10ul, Concentration: 50 ng/ul
- A HapMap control is required and is included in the price for every set of samples
- Non standard input material e.g. WGA, FFPE samples will be considered on risk

Infinium 2.5M-8 Array Processing

Part Number : P-WG-0022

Product Family : Whole Genome

Availability Date : 05/01/2013

Description

The HumanOmni2.5-8 Array Processing Fee includes sample plating, array processing primary analysis, and data storage (ARRAY NOT INCLUDED). The HumanOmni2.5-8 provides content on a 8-sample Infinium array of 2,379,855 fixed genome-wide markers that can interrogate genetic variation >2.5% minor allele frequency (MAF). On average our genotyping call rates typically exceed 98%. We are able to process up to 192 arrays per week with batches of 4 plates of 96 samples a day. Processing times vary and depend on current demand. For additional capacity, please contact genomics@broadinstitute.org. Minimum order size is 47 samples.

Deliverables

Data delivered will consist of the following raw and called genotype formats: .idats, .gtc, .gtc.txt, and Plink formatted files and will be made available via the analysis portal. Omni2.5-8 Array called genotypes are initially clustered with a command-line version of Illumina's GeneTrain calling algorithm, all samples >98% call rate are re-clustered with the genotype birdsuite calling algorithm.-Data storage is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or a non-engagement letter where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- Samples with lab pico results within the last year
- At least 200 ng of DNA (per sample) of input: Volume: 10ul, Concentration: 20-50 ng/ul
- A HapMap control is required and included in the price for every set of samples
- Sample Identification QC Check
- Non standard input materials e.g. WGA, FFPE, Mixed Population Studies will be considered on risk

Infinium Omni Express+ Exome Array Processing

Part Number : P-WG-0023

Product Family : Whole Genome

Availability Date : 05/01/2013

Description

The Omni Express Exome Array includes sample plating, array processing primary analysis, and data storage (ARRAY NOT INCLUDED). Omni Express Exome provides content on a 8-sample Infinium array of >700,000 genome-wide markers as well as >240,000 functional exonic markers. On average our genotyping call rates typically exceed 98%. We are able to process up to 192 arrays per week with batches of 4 plates of 96 samples a day. Processing times vary and depend on current demand. For additional capacity, please contact genomics@broadinstitute.org. Minimum order size is 47 samples.

Deliverables

Data delivered will consist of the following raw and called genotype formats: .idats, .gtc, .gtc.txt, and Plink formatted files and will be made available via the analysis portal. OmniExpressExome array called genotypes are initially clustered with a command-line version of Illumina's GeneTrain calling algorithm before being fed into our in-house Z-caller, all samples >98% call rate are re-clustered with the genotype birdsuite calling algorithm. Data storage is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or a non-engagement letter where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- Samples with lab pico results within the last year
- At least 200 ng of DNA (per sample) of input: Volume: 10ul, Concentration: 20-50 ng/ul
- A HapMap control is required and is included in the price for every set of samples
- Sample identification QC Check
- Non standard input materials e.g. WGA, FFPE, Mixed Populations Studies will be considered on risk

Infinium Exome (High Volume >10,000 sample) Array Processing

Part Number : P-WG-0025

Product Family : Whole Genome

Availability Date : 05/01/2013

Description

The Infinium Exome Array includes sample plating, array processing, primary analysis, and data storage (ARRAY NOT INCLUDED). The Exome chip provides content on a 12-sample Infinium array of >250,000 total functional exonic markers, enabling high-throughput and delivering industry standard robust genotypes. On average our genotyping call rates typically exceed 98%. We have the ability to process up to 192 arrays per week with batches of 6 plates of 96 samples a day. Processing times vary and depend on current demand. For additional capacity, please contact genomics@broadinstitute.org. For instance: 11, 23, 35, 47, 59, 71, 83, 95 Minimum order size is 47 samples.

Deliverables

Data delivered will consist of the following raw and called genotype formats: .idats, .gtc, .gtc.txt, and Plink formatted files and will be made available via the analysis portal. Exome array called genotypes are initially clustered with a command-line version of Illumina's GeneTrain calling algorithm before being fed into our in-house Z-caller. Data storage is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or a non-engagement letter where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- Samples with lab pico results with the last year
- At least 200 ng of DNA (per sample) of input: Volume: 10ul, Concentration: 20-50 ng/ul
- A HapMap control is required and included in the price for every set of samples
- Sample identification QC Check
- Non standard input materials e.g. WGA, FFPE, Mixed Population Studies will be considered on risk

Infinium Exome (Low Volume <10,000 sample) Array Processing

Part Number : P-WG-0029

Product Family : Whole Genome

Availability Date : 05/01/2013

Description

The Infinium Exome Array includes sample plating, array processing, primary analysis, and data storage (ARRAY NOT INCLUDED). The Exome chip provides content on a 12-sample Infinium array of >250,000 total functional exonic markers, enabling high-throughput and delivering industry standard robust genotypes. On average our genotyping call rates typically exceed 98%. We have the ability to process up to 192 arrays per week with batches of 6 plates of 96 samples a day. Processing times vary and depend on current demand. For additional capacity, please contact genomics@broadinstitute.org—Minimum order size is 47 samples.

Deliverables

Data delivered will consist of the following raw and called genotype formats: .idats, .gtc, .gtc.txt, and Plink formatted files and will be made available via the analysis portal. Exome array called genotypes are initially clustered with a command-line version of Illumina's GeneTrain calling algorithm before being fed into our in-house Z-caller. Data storage is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or a non-engagement letter where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender

- Samples with lab pico results within the last year
- At least 200 ng of DNA (per sample) of input: Volume: 10ul, Concentration: 20-50 ng/ul
- A HapMap control is required and included in the price for every set of samples
- Sample identification QC Check
- Non standard input materials e.g. WGA, FFPE, Mixed Population Studies will be considered on risk

Infinium Human Methylation450 Array Processing

Part Number P-WG-0026

Product Family : Whole Genome

Availability Date : 05/01/2013

Description

The Infinium HumanMethylation450 Array includes sample plating, array processing, primary analysis, and data storage (ARRAY NOT INCLUDED). The Infinium HumanMethylation450 BeadChip provides content on a 12-sample Infinium array of >485,000 methylation sites. It covers 99% of RefSeq genes, with an average of 17 CpG sites per gene region distributed across the promoter, 5'UTR, first exon, gene body, and 3'UTR. We have the ability to process up to 192 arrays per week with batches of 6 plates of 96 samples a day. Processing times vary and depend on current demand. For additional capacity, please contact genomics@broadinstitute.org. Minimum order size is 47 samples

Deliverables

Data delivered will consist of the following raw and called genotype formats: .idats, .gtc, .gtc.txt, and Plink formatted files and will be made available via the analysis portal. Data storage is also provided. Input Requirements

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or a non-engagement letter where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- Samples with lab pico results within the last year
- At least 200 ng of DNA (per sample) of input: Volume: 30ul, Concentration: 50 ng/ul
- A HapMap control is required and included in the price for every set of samples

Infinium Omni Express Array Processing

Part Number : P-WG-0028

Product Name : Infinium Omni Express Array Processing

Product Family : Whole Genome

Availability Date : 05/01/2013

Description

The Omni Express Array includes sample plating, array processing, primary analysis, and data storage (ARRAY NOT INCLUDED). Human Omni Express Array provides content on a 12-sample Infinium array of 730,525 fixed genome-wide markers. On average our genotyping call rates typically exceed 98%. We are able to process up to 192 arrays per week with batches of 6 plates of 96 samples a day. Processing times vary and depend on current demand. For additional capacity, please contact genomics@broadinstitute.org. Minimum order size is 47 samples.

Deliverables

Data delivered will consist of the following raw and called genotype formats: .idats, .gtc, .gtc.txt, and Plink formatted files and will be made available via the analysis portal. OmniExpress Array called genotypes are initially clustered with a command-line version of Illumina's GeneTrain calling algorithm, all samples >98 call rate are reclustered with the genotype birdsuite calling algorithm. Data storage is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or a non-engagement letter where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender

- Samples with lab pico results within the last year
- At least 200 ng of DNA (per sample) of input: Volume: 10ul, Concentration: 20-50 ng/ul
- A HapMap control is required and included in the price for every set of samples
- Sample identification QC Check
- Non standard input materials e.g. WGA, FFPE, Mixed Population Studies will be considered on risk

Infinium 5M Array Processing

Part Number : P-WG-0030

Product Family : Whole Genome

Availability Date : 05/01/2013

Description

The HumanOmni5-Quad Array includes sample plating, array processing, primary analysis, and data storage (ARRAY NOT INCLUDED). The Human Omni5-Quad provides content on a 4-sample Infinium array of 4,301,331 fixed genome-wide markers that can interrogate genetic variation as low as 1% minor allele frequency (MAF). On average our genotyping call rates typically exceed 98%. We are able to process up to 192 arrays per week with batches of 4 plates of 48 samples a day. Processing times vary and depend on current demand. For additional capacity, please contact genomics@broadinstitute.org. Minimum order size is 47 samples.

Deliverables

Data delivered will consist of the following raw and called genotype formats: .idats, .gtc, .gtc.txt, and Plink formatted files and will be made available via the analysis portal. Omni5-Quad Array called genotypes are initially clustered with a command-line version of Illumina's GeneTrain calling algorithm before being manually reviewed in GenomeStudio to create a custom clustering file. Data storage is also provided.

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or a non-engagement letter where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- Samples with lab pico results within the last year
- At least 360 ng of DNA (per sample) of input: Volume: 18ul, Concentration: 20-50 ng/ul
- A HapMap control is required and included in the price for every set of samples
- Sample identification QC Check
- Non standard input materials e.g. WGA, FFPE, Mixed Population Studies will be considered on risk

Infinium Human Core Exome Array Processing

Part Number : P-WG-0031

Product Family : Whole Genome

Availability Date : 05/01/2013

Description

The Human Core Exome Array includes sample plating, array processing, primary analysis, and data storage (ARRAY NOT INCLUDED). The Human Core Exome Bead Chip includes all the tag SNPs found on the Human Core Bead Chip, plus over 240,000 markers from Illumina's Human Exome Bead Chip. On average our genotyping call rates typically exceed 98%. We have the ability to process up to 192 arrays per week with batches of 6 plates of 96 samples a day. Processing times vary and depend on current demand. For additional capacity, please contact genomics@broadinstitute.org. Minimum order size is 47 samples.

Deliverables

Data delivered will consist of the following raw and called genotype formats: .idats, .gtc, .gtc.txt, and Plink formatted files and will be made available via the analysis portal. Exome array called genotypes are initially clustered with a command-line version of Illumina's GeneTrain calling algorithm before being fed into our in-house Z-caller. Data storage is also provided. Input Requirements

Input Requirements

- Funding and compliance requirements must be in place - this includes a valid IRB or a non-engagement letter where needed
- Minimum Sample data including - Collaborator Participant ID, Collaborator Sample ID, Gender
- Samples with lab pico results within the last year
- At least 200 ng of DNA (per sample) of input: Volume: 10ul, Concentration: 20-50 ng/ul
- A HapMap control is required and included in the price for every set of samples
- Sample identification QC Check
- Non standard input materials e.g. WGA, FFPE, Mixed Population Studies will be considered on risk