Genomics Platform Product List: Descriptions and Deliverables

(P-MCV-0001) Standard 16S Sequencing

Illumina Sequencing Only

(P-SEQ-0001) HiSeq 2x25 paired Lane

(P-SEQ-0002) HiSeq 44 Single Lane

(P-SEQ-0003) HiSeq 2x76 paired Lane

(P-SEQ-0004) HiSeq 2x101 paired Lane

(P-SEQ-0005) MiSeq Short run

(P-SEQ-0006) MiSeq Long Run

(P-SEQ-0007) MiSeq Extra Long Run

Custom Genotyping and Expression

(P-CGE-0001) Fluidigm Custom Expression 48.48

(P-CGE-0002) Fluidigm Custom Expression 96.96

(P-CGE-0003) Fluidigm Custom Genotyping 96.96

(P-CGE-0004) Sequenom Custom Genotyping

Small Target Sequencing

(P-VAL-0001) Fluidigm Custom PCR 48.48

(P-VAL-0002) TSCA (<2500 amplicons)

(P-VAL-0003) Custom Hybrid Selection (<1Mb)

Extraction and Sample Handling

(P-ESH-0003) WGA Add-on

(P-ESH-0004) DNA Extract from blood, fresh frozen tissue, cell pellet, stool, or saliva

(P-ESH-0005) DNA and RNA Extract from fresh frozen tissue or stool (AllPrep)

(P-ESH-0006) DNA Extract from Buffycoat

(P-ESH-0007) DNA Extract from FFPE or slides

(P-ESH-0008) RNA Extract from FFPE

(P-ESH-0009) External Plating into Tubes

(P-ESH-0010) External Plating into Plates

Analysis Only

(P-ANA-0001) Picard Analysis of externally generated data - human exome

Incoming Samples

(P-ESH-0001) Sample Initiation

Product 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) and normalization to a standard Genomics Platform working concentration appropriate for the relevant downstream workflows. We have the ability to process of up to 10,000 samples per week.   All samples entering the Broad Genomics Platform will go through the sample initiation process.

Deliverable: 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.

(P-ESH-0002) Sample Qualification Only (Human Samples)

Product 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 9600 samples per week handled in batches of 1920. 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 genotyping or 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 and genotyping 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.

Deliverable: 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

- Valid funding source

- 25ng of input DNA

Exome Sequencing Analysis

(P-EX-0001) Standard Exome Sequencing

Product Description: The Standard Exome includes sample plating, library preparation, hybrid capture, sequencing, 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. With 5-7Gb PF of data 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

Deliverable: 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) and FFPE samples will be accepted at risk, but success rates are high with 50ng (1ng/uL minimum concentration) or greater.

- Tumor/Normal or Case/Control pairs must be received together if indel co-cleaning is required.

(P-EX-0002) Exome Express

Product 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. Exome Express libraries typically meet or exceed 80% of targets at 20x coverage. The Exome Express product includes sample plating, library preparation, hybrid capture, sequencing, 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.

Deliverable: 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 >500ng of DNA (note extra cost will be applied for extractions). Samples below 250ng (2ng/uL minimum concentration) and FFPE 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

- All samples >0ng will proceed on risk.

(P-EX-0003) Exome Plus

Product Description: Exome Plus includes sample plating, library preparation, hybrid capture, sequencing, 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 25Gb of delivered data 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

Deliverable: 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 50ng (1ng/uL minimum concentration) or greater.

- Tumor/Normal or Case/Control pairs must be received together if indel co-cleaning is required

Whole Genome Sequencing Analysis

(P-WG-0001) Standard Light Coverage Whole Genome Sequencing (6x)

Product Description: Standard Light Coverage Whole Genome Sequencing (6x) includes plating, library preparation, size selection,, sequencing, sample identification QC check, and data storage. The product provides one size selected library (insert size of library is 325bp +/- 20%) and sequence coverage to 20 Gb total aligned. This product is appropriate for the generation of low coverage human and mammalian genomes. We have the ability to process up to 184 samples per week in batches of 92. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org.

Deliverable: 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.

- Tumor/Normal or Case/Control pairs must be received together if indel co-cleaning is required

(P-WG-0002) Standard High Coverage Whole Genome Sequencing (30x)

Product Description: Standard High Coverage Whole Genome Sequencing (30x) includes plating, library preparation, size selection, sequencing, 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 95Gb total aligned. This product is appropriate for the generation of high coverage human and mammalian genomes. We have the ability to process up to 92 samples per week in batches of 92. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org.

Deliverable: 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.

- Tumor/Normal or Case/Control pairs must be received together if indel co-cleaning is required

(P-WG-0003) Standard Deep Coverage Whole Genome Sequencing (60x)

Product Description: Standard Deep Coverage Whole Genome Sequencing (60x) includes plating, library preparation, size selection, sequencing, 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 190 Gb total aligned. This product is appropriate for the generation of deep coverage human and mammalian genomes. We have the ability to process up to 92 samples per week in batches of 92. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org.

Deliverable: 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.

- Tumor/Normal or Case/Control pairs must be received together if indel co-cleaning is required

(P-WG-0004) Non-Human Whole Genome Sequencing

Product Description: Non-Human Whole Genome Sequencing includes plating, library preparation, sequencing, sample identification QC check, and data storage. The product provides a non-size selected library and sequencing to 300Mb (4Mb genome at ~30x coverage).   This is appropriate for microbial and other small genome non-assembly needs such as SNP calling. We are able to process up to 92 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 92 samples.

Deliverable: 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 (2ng/uL minimum concentration). Samples below 250ng will be accepted at risk, but success is expected with 50ng (1ng/uL minimum concentration) or greater.

Whole Genome Array Analysis

(P-WG-0005) Infinium Exome

Product Description: The Infinium Exome Array includes sample plating, array processing, sample identification QC check, primary analysis, and data storage. 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 4,608 samples per week with batches of 12 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 11 samples.

Deliverable: 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

- At least 200ng of DNA (per sample) of input

- Volume: 10ul, Concentration: 20-50ng/ul

- A HapMap control is required and included in the price for every set of samples

(P-WG-0006) Infinium 5M

Product Description: The HumanOmni5-Quad Array includes sample plating, array processing, sample identification QC check, primary analysis, and data storage. 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 1,536 samples per week with batches of 8 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 3 samples.

Deliverable: 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

- At least 360ng of DNA (per sample) of input

- Volume: 18ul, Concentration: 20-50ng/ul

- A HapMap control is required and included in the price for every set of samples

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(P-WG-0007) Infinium 2.5M-8

Product Description: The HumanOmni2.5-8 Array includes sample plating, array processing, sample identification QC check, primary analysis, and data storage. 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 3,072 samples per week with batches of 8 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 7 samples.

Deliverable: 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 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

- At least 200ng of DNA (per sample) of input

- Volume: 10ul, Concentration: 20-50ng/ul

- A HapMap control is required and included in the price for every set of samples

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(P-WG-0008) Infinium Omni-Quad

Product Description: The HumanOmni1-Quad Array includes sample plating, array processing, sample identification QC check, primary analysis, and data storage. The HumanOmni1-Quad Array utilizes the content of the Illumina array to bring a low-cost, quick turnaround solution for genome-wide association studies. On average our genotyping call rates typically exceeding 98%. We are able to process up to 1,536 samples 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 3 samples.

Deliverable: 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. Omni1-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

- At least 200ng of DNA (per sample) of input

- Volume: 10ul, Concentration: 20-50ng/ul

● A HapMap control is required and provided for every set of samples

(P-WG-0009) Infinium Omni Express+ Exome (8)

Product Description: The OmniExpress Exome Array includes sample plating, array processing, sample identification QC check, primary analysis, and data storage. OmniExpressExome 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 3,072 samples per week with batches of 8 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 7 samples.

Deliverable: 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".   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

● At least 200ng of DNA (per sample) of input

● Volume: 10ul, Concentration: 20-50ng/ul

● A HapMap control is required and is included in the price for every set of samples

(P-WG-0010) Infinium Omni Express (12)

Product Description: The OmniExpress Array includes sample plating, array processing, sample identification QC check, primary analysis, and data storage. HumanOmniExpress 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 4,608 samples per week with batches of 12 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 11 samples.

Deliverable: 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 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

● At least 200ng of DNA (per sample) of input

● Volume: 10ul, Concentration: 20-50ng/ul

● A HapMap control is required and included in the price for every set of samples

(P-WG-0011) **Affy 6.0**

Product Description: The Affy 6.0 Array includes sample plating, array processing, sample identification QC check, primary analysis, and data storage. 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 768 samples in 10 days. Processing times vary and depend on current demand. For additional capacity, please contact genomics@broadinstitute.org. Minimum order size is 47 samples.

Deliverable: 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

- At least 500ng of DNA (per sample) of input

- Volume: 10ul, Concentration: 50ng/ul

- A HapMap control is required and is included in the price for every set of samples

RNA Sequencing Analysis

(P-RNA-0001) Standard RNA Sequencing - Low Coverage (15M pairs)

Product Description: Standard 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 non-strand specific Illumina TruSeq Protocol and sequence coverage to 15M Paired reads and 30M total reads. We have the ability to process up to 184 samples per week in batches of 92. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org.

Deliverable: 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.

(P-RNA-0002) Standard RNA Sequencing - High Coverage (50M pairs)

Product Description: Standard 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 non-strand specific Illumina TruSeq Protocol and sequence coverage to 50M Paired reads and 100M total reads. We have the ability to process up to 184 samples per week in batches of 92. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org.

Deliverable: 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.

(P-RNA-0003) Strand Specific RNA Sequencing - Low Coverage (15M pairs)

Product Description: Strand Specific RNA Sequencing includes plating, poly-A selection and strand specific 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 dUTP Protocol and sequence coverage to 15M Paired reads and 30M total reads. We have the ability to process up to 92 samples per week in one batch. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org.

Deliverable: Data delivery will include a de-multiplexed, aggregated Picard BAM file which will be accessed via the BASS file server systemor 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 3ug (minimum concentration 60ng/uL) or greater.

(P-RNA-0004) Strand Specific RNA Sequencing - High Coverage (50M pairs)

Product Description: Strand Specific RNA Sequencing includes plating, poly-A selection and strand specific 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 dUTP Protocol and sequence coverage to 50M Paired reads and 100M total reads. We have the ability to process up to 92 samples per week in one batch. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org.

Deliverable: Data delivery will include a de-multiplexed, aggregated Picard BAM file which will be accessed via the BASS file server system. 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 3ug (minimum concentration 60ng/uL) or greater.

Assembly and Metagenomic Analysis

(P-MCV-0001) Small Genome Illumina Assembly

Product Description: The Small Genome Illumina Assembly product includes plating, library preparation, sequencing, 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. This is appropriate for the assembly and annotation of small (4-6Mb) genomes. We have the ability to process up to 92 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.

Deliverable: 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, Collaborator Participant ID and Collaborator Sample ID

- Minimum input of 1ug or more genomic DNA. Samples below 1ug (minimum concentration 7ng/uL) will be accepted at risk, but success is expected with 250ng (minimum concentration 2ng/uL) or greater.

(P-MCV-0001) Standard 16S Sequencing

Product 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 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. For additional capacity please contact genomics@broadinstitute.org. Minimum order size is 192 samples. Please contact genomics@broadinstitute.org for more details on order size constraints.

Deliverable: 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

Illumina Sequencing Only

(P-SEQ-0001) HiSeq 2x25 paired Lane

Product 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.

Deliverable: Data delivery to a minimum ~100M Paired 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 or concentration requirements will be accepted on risk

(P-SEQ-0002) HiSeq 44 Single Lane

Product 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.

Deliverable: Data delivery to a minimum ~100M 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 or concentration requirements will be accepted on risk

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(P-SEQ-0003) HiSeq 2x76 paired Lane

Product 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.

Deliverable: Data delivery to a minimum ~100M Paired 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 or concentration requirements will be accepted on risk

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(P-SEQ-0004) HiSeq 2x101 paired Lane

Product 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.

Deliverable: Data delivery to a minimum ~100M Paired 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 or concentration requirements will be accepted on risk

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(P-SEQ-0005) MiSeq Short run

Product 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.

Deliverable: Data delivery to a minimum ~5M 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 or concentration requirements will be accepted on risk

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(P-SEQ-0006) **MiSeq Long Run**

Product 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.

Deliverable: Data delivery to a minimum ~ 5M 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 or concentration requirements will be accepted on risk

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(P-SEQ-0007) **MiSeq Extra Long Run**

Product 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.

Deliverable: Data delivery to a minimum ~ 5M 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 or concentration requirements will be accepted on risk

Custom Genotyping and Expression

(P-CGE-0001) Fluidigm Custom Expression 48.48

Product 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. The BioMark HD system can work with many chemistry options such as TaqMan™ Probes, Eva Green® Double-Stranded Binding Dye, Roche Universal Probe Library, & TaqManTM 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. For additional capacity please contact genomics@broadinstitute.org. Minimum order size is 48 samples. Please contact genomics@broadinstitute.org for more details on order size constraints.

Deliverable: 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.

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 letter of non-engagement where needed

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

- Total RNA, Tissue, Lysate, Blood, Slides that yields > 5ng of RNA (note extra cost will be applied for extractions).

(P-CGE-0002) Fluidigm Custom Expression 96.96

Product 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, & TaqManTM 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. For additional capacity please contact genomics@broadinstitute.org. Minimum order size is 48 samples. Please contact genomics@broadinstitute.org for more details on order size constraints.

Deliverable: 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.

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 letter of non-engagement where needed

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

- Total RNA, Tissue, Lysate, Blood, Slides that yields > 5ng of RNA.

(P-CGE-0003) Fluidigm Custom Genotyping 96.96

Product 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 4800 samples per week. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org. Minimum order size is one chip. Please contact genomics@broadinstitute.org for more details on order size constraints.

Deliverable: 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's 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

- At least 25 ng of DNA (per sample) of input

- Volume: 2.5ul, Concentration: 10 ng/ul

(P-CGE-0004) Sequenom Custom Genotyping

Product Description: Mass Spectrometry genotyping technology utilizies 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. We have the ability to process up to 3,840 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 1 sample. Please contact genomics@broadinstitute.org for more details on order size constraints.

Deliverable: 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

- 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

- Sequenom HME: 5ng per pool at 2.5ng/uL 25ul + 2ul per pool

- Sequenom iPLEX: 10ng per pool at 5ng/ul 25ul + 2ul per pool

Small Target Sequencing

(P-VAL-0001) Fluidigm Custom PCR 48.48

Product Description: Fluidigm Custom PCR includes plating, PCR amplification of 48 samples across 48(singleplex) or 480 (multiplex) primer pairs (Primer cost NOT included), 1 MiSeq run at 2x150bases,, and data storage. For any other combinations of samples and primers, please contact the Product Manager or genomics@broadinstitute.org. We have the ability to process up to 96 samples per week in batches of 48. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org.

Deliverable: 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.

(P-VAL-0002) TSCA (<2500 amplicons)

Product 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 or genomics@broadinstitute.org. 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 please contact genomics@broadinstitute.org.

Deliverable: 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.

(P-VAL-0003) Custom Hybrid Selection (<1Mb)

Product Description: Custom Hybrid Selection includes sample plating, library preparation, hybrid capture, sequencing, sample identification QC check, and data storage. 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 or genomics@broadinsitute.org. 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.

Deliverable: 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 (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.

- Tumor/Normal or Case/Control pairs must be received together if indel co-cleaning is required.

Epigenomics Analysis

(P-EPI-0001) RRBS Sequencing

Product Description: RRBS sequencing includes sample plating, MspI restriction enzyme digestion, library preparation and QC, sequencing, and data storage. This product utilizes the "mRRBS" method published by Patrick Boyle et al. Genome Biology (2012) with 10 million reads or greater, RRBS libraries typically meet 10x coverage. 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.

Deliverable: 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.

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

- 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 (20ng/uL minimum concentration) will be considered at risk, but success is possible with 50ng (5ng/uL minimum concentration) or greater.

Extraction and Sample Handling

(P-ESH-0003) WGA Add-on

Product Description: The WGA Add-On 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 768 samples per week handled in batches of 96. Processing times vary and depend on current demand. For additional capacity please contact genomics@broadinstitute.org.

Deliverable: 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

(P-ESH-0004) DNA Extract from blood, fresh frozen tissue, cell pellet, stool, or saliva

Product 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. 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.

Deliverable: High quality DNA (A260/A280 range of 1.7-1.9). DNA is quantified in triplicate using a standardized picogreen assay. Sample yield is 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

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(P-ESH-0005) DNA and RNA Extract from fresh frozen tissue or stool (AllPrep)

Product 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.

Deliverable: High quality DNA (A260/A280 range of 1.7-1.9) with an average length of 15–30 kb. DNA is quantified in triplicate using a standardized picogreen assay. Sample yields is dependent on multiple factors such as: the original material type provided (blood, cells, tissue, etc), amount of material provided, and tissue site. High quality RNA that is quantified via nanodrop along with 260/280, 260/230 purity ratios. The RNA integrity is measured by the RIN value determined by a bioanalyzer.

Input Requirements:

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

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(P-ESH-0006) DNA Extract from Buffycoat

Product Description: This product is appropriate for DNA Extraction from Buffycoat. 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 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.

Deliverable: High quality DNA (A260/A280 range of 1.7-1.9). DNA is quantified in triplicate using a standardized picogreen assay. Sample yields is dependent on multiple factors such as: cell counts, amount of material provided, and quality of buffy coat.
Input Requirements:

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

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(P-ESH-0007) DNA Extract from FFPE or slides

Product 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 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.

Deliverable: High quality DNA (A260/A280 range of 1.7-1.9). DNA is quantified in triplicate using a standardized picogreen assay. Sample yields is 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

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(P-ESH-0008) RNA Extract from FFPE

Product 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 formaldehye 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 10μl. 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.

Deliverable: High quality RNA that is quantified via nanodrop along with 260/280, 260/230 purity ratios. The RNA integrity is measured by the RIN value determined by a bioanalyzer.

Input Requirements:

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

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(P-ESH-0009) External Plating into Tubes

Product 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.

Deliverable: DNA and/or RNA aliquotted 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, address and contact information to the person/institute the samples will be shipped to.

(P-ESH-0010) External Plating into Plates

Product 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.

Deliverable: 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, address and contact information to the person/institute the samples will be shipped to.

Analysis Only

(P-ANA-0001) Picard Analysis of externally generated data - human exome

Product 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.

Deliverable: 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 filesystem

- 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

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(P-ANA-0002) Picard Analysis of externally generated data - human whole genome (4-6x)

Product 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.

Deliverable: 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 filesystem

- 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

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(P-ANA-0003) Picard Analysis of externally generated data - human whole genome (30x)

Product 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.

Deliverable: 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 filesystem

- 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

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(P-ANA-0004) Picard Analysis of externally generated data - human whole genome (>30x)

Product 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.

Deliverable: 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 filesystem

- 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