DesignStudio™ Microarray Assay Designer Release Notes

v.2.1.0

March 2024

Introduction

DesignStudio Microarray Assay Designer is a web-based software tool that allows customers to design custom and semi-custom Infinium™ BeadChips. Highlights include:

Powerful Custom Genotyping Arrays

Design custom panels to support tailored research applications. Choose from multiple BeadChip formats and supported species to suit the project objectives.

Sophisticated Design Tools

Supports successful probe design for single-nucleotide polymorphisms (SNPs) and insertions/deletions (indels) by providing predicted success and Infinium validation status.

These Release Notes detail the key features and changes to software components for the release of DesignStudio Microarray Assay Designer v.2.1.0. As a web application, the new release affects all customers. For information on how to use the system, see the DesignStudio Microarray Assay Designer Online Help. DesignStudio is an online tool for creating custom and semi-custom Infinium BeadChips, including features such as:

- Login using MyIllumina account
- Supports creation of custom and semi-custom Infinium BeadChips in the following assay formats:
  - XT
  - HTS
- Allows design of semi-custom content based on popular catalog array backbones
- Designs one to four species on a single array for XT BeadChips
- Designs BeadChips for human and agricultural species
- View project status
- Informs on product criteria, including:
  - Minimum and maximum number of markers per chip
  - Minimum number of chips that need to be ordered
  - Manufacturing conversion rate
- Provides standard inputs for custom designs
- Generates designs using:
  - rsIDs from dbSNP (human only)
  - Genomic coordinates (human only)
  - Gene names
  - Existing Illumina IDs (ILMN IDS)
  - Flanking sequences and alleles
  - Previous score file
- Alerts to potential issues with submitted design
- Outputs list of input targets that were unsuccessful
- Output file containing the sequence of each design and predicted success

RELEASE v.2.1.0 HIGHLIGHTS

- Genome reference database updated to align with the latest version (dbSNP Build 155).
- Ability to replicate targets providing higher likelihood that high important content will be successful in the final design.
- Conversion of multi-allelic targets to bi-allelic targets to optimize BeadChip space for low minor allele frequency variants known by the reference database.
- Enabled design for MNV and Infinium I indel targets.
- Minor defect repairs.

NEW FEATURES IN DETAIL

- Reference database updates for human as species.
    - Updated from dbSNP Build 147 to dbSNP Build 155
    - rsIDs entered in Identity file type for the input target file will be updated to the sequence aligned with dbSNP Build 155. These updates may lead to changes in the sequence associated with rsIDs previously entered.
    - Implemented banner management functionality to provide users advanced noticed of upcoming dbSNP updates.
- Replication of targets to be included in the final design.
    - Replication option available as column in the Identity and Sequence file types for the input target file.
    - Valid input options of 1 to 3 replicates.
    - Replication counts toward the total Attempted Bead Types (ABT) included in the final design.
- Conversion of multi-allelic targets to bi-allelic targets for human as species when using the Identity, Region or Gene file type for the input target file.
    - With the update from dbSNP 147 to dbSNP 155, many variants became multi-allelic as more humans have been genotyped.
    - Check box within user interface allows conversion of multi-allelic SNPs to bi-allelic.
        - For multi-allelic variants where exactly one minor allele is greater than 1% frequency, the minor allele with >1% frequency will be chosen for the bi-allelic design
        - For multi-allelic variants where all minor alleles are <1% frequency, the minor allele with the highest frequency will be chosen for the bi-allelic design.

    - For multi-allelic variants where more than one minor allele is >1% frequency, the design will be returned in the excluded content file. The design can proceed using flanking sequences or HGVS naming convention to specify the intended design. Illumina Concierge services are available for design support.

- Enable design of multi-nucleic variants using HGVS naming convention.
    - HGVS naming convention can be used to specify the multi-allelic variant as bi-allelic targets.
    - The bi-allelic targets can be designed and recombined during analysis using GenomeStudio™ software.
- Enabled design of Infinium I indels for all species.
    - 'Force_Infinium_I' available as column in the Sequence file type for the input target file.
        - Accepts inputs of TRUE or FALSE

RESOLVED ISSUES

Defect repairs (bug fixes) from v2.0.0

- Added validation and messaging in the user interface to prevent non-alphanumeric characters from being used in the design name.
- Allows large region files with greater than 200 entries to be used.
- Allows design files with greater than 200 chromosomes to be used.
- Improved error messaging when:
    - Legacy Illumina IDs are used in the existing design file.
    - Non-alphanumeric characters such as "|", "Ö", "&", "-", etc, are used in the input file

KNOWN ISSUES

- Non-specific failures occur when:
    - Loci names include two dashes ("--") in the input file
    - A M or MT chromosome designation is used for equine as species
    - Submitting sequence files with certain formatting errors. The workflow terminates without allowing access to the error file.
- When using an existing design file, a dropdown exists but the stand type cannot be changed