



v.2.2.0

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Page 2 of 3

Introduction

DesignStudio Microarray Assay Designer is a web-based software tool that allows customers to design custom and semi-custom InfiniumTM 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.2.2.0. As a web application, the new release affects all customers. For information on how to use the system, see the <u>DesignStudio Microarray Assay Designer Online Help.</u> 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:
 - o EX-24
 - o EX-48
 - o HTS
 - o XT
- 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:
 - o Minimum and maximum number of markers per chip
 - o Minimum number of chips that need to be ordered
 - o Manufacturing conversion rate
- Provides standard inputs for custom designs
- Generates designs using:
 - o rsIDs from dbSNP (human only)
 - o Genomic coordinates (human only)
 - o Gene names
 - o Existing Illumina IDs (ILMN IDS)
 - o Flanking sequences and alleles
 - o Previous score file
- Alerts to potential issues with submitted design



Page 3 of 3

- Outputs list of input targets that were unsuccessful
- Output file containing the sequence of each design and predicted success

RELEASE V.2.2.0 HIGHLIGHTS

• Ability to design EX-24 and EX-48 iSelect BeadChips

NEW FEATURES IN DETAIL

- Options within Select Assays to design EX-24 iSelect and EX-48 iSelect BeadChips
 - o Updates to Compare Assays table to show new BeadChip types and specifications
- Out-of-date rsIDs submitted are automatically updated to the revised merged rsID per dbSNP
 - o Identity error file lists submitted rsID alongside revised merged rsID
 - o Score file automatically includes revised merged rsID
- rsIDs submitted with capital letters are automatically lowercased for consistent formatting
- Designs that use the automated strategy to convert from multi-allelic to bi-allelic are more clearly indicated in Review and Order Design page and output files
- Score file name updated from 'Failure_Codes' to 'Warning_Codes' to more accurately reflect the purpose

KNOWN ISSUES

- Non-specific failures occur when:
 - O Loci names include two dashes ("--") in the input file
 - o 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
- Rare, specific Infinium II indels designing as Infinium I with a 3' ambiguity error message