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Variant Effect Mapping Protocol Collection

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We use this collection and it's working

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Keywords: DMS, variant effect mapping, VE mapping, deep mutational scanning, human cell-based assay, yeast-based assay, functional assay, MAVE, Multiplex Assay of Variant Effect, MAVE, POPcode, double-tag POPcode, Precision Oligo-Pool Based Code Alteration, TileSeq, multiplexed assays of variant effect, supporting clinical variant interpretation, measurement of variant effect, clinical variant interpretation, comprehensive atlas of variant effect, measured variant function, functional impact of all possible missense variant, disease phenotype, variant effect mapping protocol collection advance, variant effect, based functional assay, variant effect map, functional assay, associated gene, gene, enabled multiplexed assay, including variant, directed mutagenesi, possible missense variant, variant, generating mutagenic library, based assay, functional score, mutagenic library

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Abstract

Advances in cellular engineering and sequencing have enabled **Multiplexed Assays of Variant Effect (MAVEs)**, a technological approach to proactively measure the functional impact of all possible missense variants in a disease-associated gene, including variants not yet observed in the clinic. The resulting "lookup tables" of measured variant function are commonly referred to as **variant effect maps**. There is a growing international effort to generate a comprehensive atlas of variant effects, with value not only for defining sequence–structure–function relationships and supporting clinical variant interpretation, but also for demonstrating quantitative correlations between functional scores and disease phenotypes.

The protocols in this collection are organized sequentially and guide the user through preparing the gene of interest with a stop codon, performing site-directed mutagenesis, validating a scalable human cell- and yeast-based functional assay, generating mutagenic libraries, and executing a MAVE in a human cell line or in yeast. These workflows enable the measurement of variant effects *en masse* using complementary readouts — including growth-based assays and fluorophore reporters — to assess outputs such as total enzymatic activity and protein stability.

Files

SEARCH

Protocol

NAME

Gene of Interest with STOP codon

VERSION 1

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Protocol

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Site-Directed Mutagenesis

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Protocol

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Validation of a Yeast-Based Functional Assay

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Protocol

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Validation of a Mammalian Cell-Based Fluorescent Reporter Assay

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Validation of a Mammalian Cell-Based Growth Assay

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Generation of Mutagenic Libraries using a POPCode Method

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Multiplex Assay of Variant Effect in Yeast

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Large-Scale Fluorescence-Based Mammalian Cell Reporter Assay

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Protocol

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Large-Scale Growth-Based Mammalian Cell Assay

VERSION 1

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