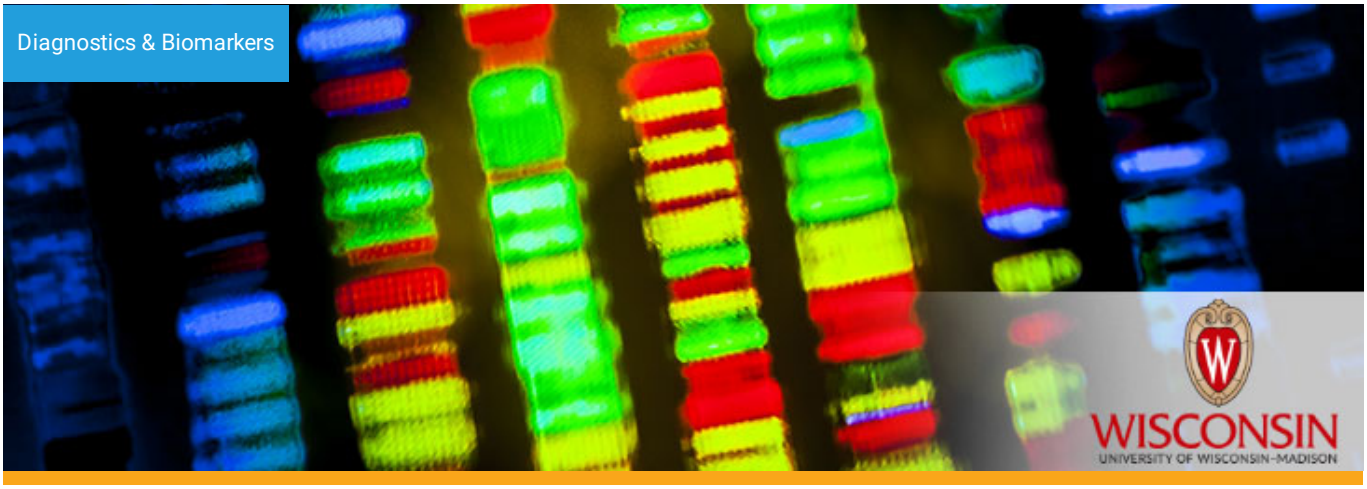


# IDENTIFYING DISEASE-CAUSING HUMAN DDX41 GENETIC VARIANTS

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## The Invention

UW-Madison researchers have developed a genetic rescue assay that discriminates the activities of wild type DDX41 from those of human myeloid malignancy-associated germline and somatic DDX41 mutants. The researchers used CRISPR/CaS9 to engineer heterozygous DDX41 progenitor cells and demonstrated that the cells express ~50% lower DDX41 protein. This system was then used to devise a genetic rescue assay in which wild type or disease-linked variants of DDX41 are replaced at near physiological levels. From here, the researchers leveraged RNA-seq to define a restricted cohort of transcripts that are DDX41-regulated, with the transcription levels providing a quantitative metric of DDX41 function. This was combined with the differentiation state of the cells to provide another quantitative metric of DDX41 function.

Ultimately, these findings could be extended to structure/function assessments for clinical genetic curation and leveraged to elucidate mechanisms and interventions that promote and/or oppose DDX41 function.

## Additional Information

**For More Information About the Inventors**

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**Tech Fields**

- Diagnostics & Biomarkers : Diagnostics

For current licensing status, please contact Andy DeTienne at adetienne@warf.org or 608-960-9857