



Detecting CYP2D6 Hybrid Alleles and Preventing Sample Mischaracterization with the MassARRAY® System

Dr. Houda Hachad is an entrepreneurial scientist with deep experience in pharmacology and pharmacogenetics. At the University of Washington, she co-developed a drug interaction database and a pharmacogenetics database. Both leverage revenue-based funding and are widely used by pharmaceutical companies, regulatory agencies, and academic institutions worldwide. She is Vice President of Clinical Operations at AccessDx.

“Bottom line, if you are detecting CNV by looking only at a specific region, you are likely missing hybrid alleles.”

– Houda Hachad

Dr. Hachad offers insights and recommendations for overcoming common lab challenges, achieving a comprehensive CYP2D6 profile and more accurate PGx results.

Q Why is CYP2D6 copy number detection so important to PGx testing?

A CYP2D6 influences the metabolism of around 25% of all commonly used medications, so accurately characterizing it is critical. While characterization of the alleles based on SNPs and INDELS is important, it is incomplete without an understanding of the gene copy number. Without proper characterization of copy number, you cannot properly determine the various phenotypes.

Q What are CYP2D6 hybrid alleles and how do they affect copy number detection?

A CYP2D6 hybrid alleles are structural variations that change the functionality of the enzyme. These alleles are fairly common. It is essential to distinguish between hybrids and other CYP2D6 alleles to accurately characterize a sample's metabolizer status. Unfortunately, depending on the technology being used, that can be difficult to do.



Houda Hachad

Pharm.D, M.Res

Vice President of Clinical Operations, AccessDx



Q Why is *CYP2D6* copy number detection so important to PGx testing?

A CYP2D6 influences the metabolism of around 25% of all commonly used medications, so accurately characterizing it is critical. While characterization of the alleles based on SNPs and INDELS is important, it is incomplete without an understanding of the gene copy number. Without proper characterization of copy number, you cannot properly determine the various phenotypes.

Q What are *CYP2D6* hybrid alleles and how do they affect copy number detection?

A CYP2D6 hybrid alleles are structural variations that change the functionality of the enzyme. These alleles are fairly common. It is essential to distinguish between hybrids and other CYP2D6 alleles to accurately characterize a sample's metabolizer status. Unfortunately, depending on the technology being used, that can be difficult to do.

Q Why are *CYP2D6* hybrid alleles difficult to detect?

A Most PGx copy number detection methods cannot detect hybrid alleles. They only look at one, two, or maybe three regions on the gene. That is not enough. You need a technology that looks broadly across the gene to catch these structural changes. The VeriDose® CYP2D6 CNV Panel from Agena Bioscience® is a great example of a technology that is well-suited for hybrid allele detection. The single-well panel interrogates 22 points along the gene to identify hybrid alleles and provide accurate copy number calling.

Explore the MassARRAY® for PGx testing – visit agenabio.com/pgx to learn more.

The MassARRAY® System is For Research Use Only. Not for use in diagnostic procedures.

Agena Bioscience, Inc.
4755 Eastgate Mall
San Diego, CA 92121
Phone: +1.858.882.2800
Web: agenabio.com

Order Desk: +1.858.202.9301
Order Desk Fax:
+1.858.202.9220
orderdesk@agenabio.com
www.agenabio.com

MassARRAY, VeriDose and Agena Bioscience are registered trademarks
of Agena Bioscience, Inc. © 2019-2022 Agena Bioscience, Inc.
GEN0025 02

