SUPPORTING INFORMATION FOR THE ARTICLE:

Long-read single-molecule real-time (SMRT) full gene sequencing of cytochrome P450-2D6 (*CYP2D6*)

Wanqiong Qiao^{1*}, Yao Yang^{1*}, Robert Sebra^{1,2}, Geetu Mendiratta¹, Andrea Gaedigk^{3,4}, Robert J.

Desnick¹, and Stuart A. Scott¹

* These authors contributed equally to this manuscript and should be regarded as joint first authors.

¹ Department of Genetics and Genomic Sciences, Icahn School of Medicine at Mount Sinai, New York, NY 10029, USA.

² Icahn Institute for Genomics and Multiscale Biology, Icahn School of Medicine at Mount Sinai, New York, NY 10029, USA.

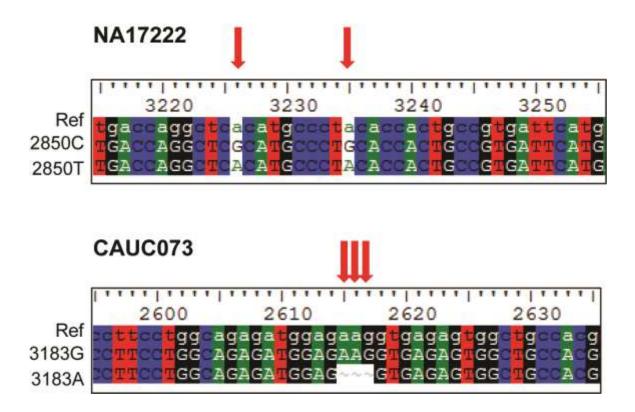
³ Division of Clinical Pharmacology, Toxicology & Therapeutic Innovation, Children's Mercy Kansas City, Kansas City, MO 64108, USA.

⁴ School of Medicine, University of Missouri-Kansas City, Kansas City, MO 64108, USA.

CORRESPONDENCE TO:

Stuart A. Scott, PhD

Assistant Professor Department of Genetics and Genomic Sciences Icahn School of Medicine at Mount Sinai One Gustave L. Levy Place, Box 1497 New York, NY, 10029 Tel. 212-241-3780 Fax. 212-241-1464 E-mail: <u>stuart.scott@mssm.edu</u>



Supp. Figure S1. Allele-specific PCR and Sanger sequencing results of NA17222 and CAUC073 based on 2850C>T and 3183G>A, respectively. Red arrows denote the locations of interrogated variants by Sanger sequencing. The 3226A>G (rs61736517; p.H352R) and 3235A>G (rs202102799; p.Y355C) variants detected in NA17222 by *CYP2D6* SMRT sequencing were found in *cis* with 2850C (classified as **108*), and the 3183G>A (rs59421388; p.V338M) variant detected in CAUC073 was on the same haplotype as the 2615_2617delAAG (rs5030656; p.K281del) allele (classified as **109*).

Supp. Tables S1-S5 are available in a separate Excel file under the Supporting Information for this article.

Supp. Table S1. *CYP2D6* variant coordinates according to common M33388.1 GenBank reference and HGVS nomenclature

Supp. Table S2. *CYP2D6* variants and diplotypes identified by SMRT sequencing in the positive control samples

Supp. Table S3. *CYP2D6* Allele-specific PCR (AS-PCR) results and phased diplotype determination for NA17222 and CAUC073

Supp. Table S4. *CYP2D6* variants and diplotypes identified by SMRT sequencing in the duplication control samples

Supp. Table S5. *CYP2D6* variants and diplotypes identified by SMRT sequencing in the discordant control samples