# Standardization of Pharmacogenetic Nomenclature

**Lisa Kalman, PhD**
Coordinator, GeT-RM
CDC
LKalman@cdc.gov

GeT-RM
Genetic Testing Reference Materials
PROGRAM

# The Disclaimer:

The fi- di- gs a- d co- clusio- s i- this report are those of the author a- d do - ot - ecessarily represe- t the official positio- of the Ce- ters for Disease Co- trol a- d Preve- tio- /the Age- cy for Toxic Substa- ces a- d Disease Registry.

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Lisa Kalma- has - o co- flicts of i- terest to report

# Reference Materials: Essential for Quality Testing

## The Problem

Characterized reference materials are not publicly available for most genetic tests; laboratories often use uncharacterized, non-renewable clinical materials for test validation and QC

## Consequences

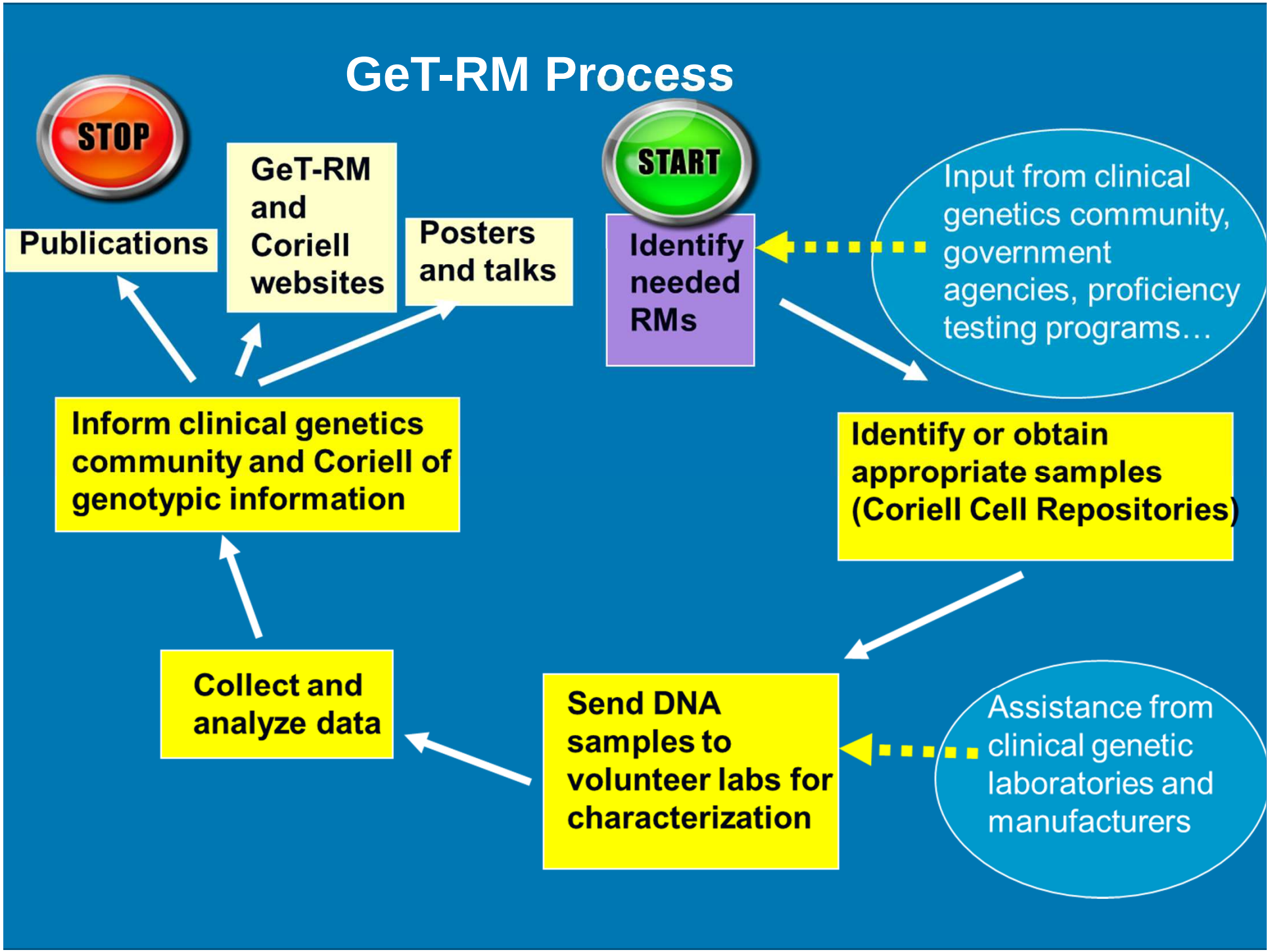
The quality of genetic tests may be compromised

# Ge- etic Testi- g Refere- ce Material Program (GeT-RM)

**A collaborative CDC-based program to improve the availability of refere- ce materials for ge- etic testi- g**

# Roles of GeT-RM

- Information exchange about RMs for genetic testing
- Monitor RM needs
- Facilitate submission, development and characterization of RMs


GeT-RM Process
STOP
START
Publications
GeT-RM and Coriell websites
Posters and talks
Identify needed RMs
Input from clinical genetics community, government agencies, proficiency testing programs…
Inform clinical genetics community and Coriell of genotypic information
Identify or obtain appropriate samples (Coriell Cell Repositories)
Collect and analyze data
Send DNA samples to volunteer labs for characterization
Assistance from clinical genetic laboratories and manufacturers

# GeT-RM Projects

## Completed Projects

- Fragile X
- Hu- ti- gto- Disease
- Cystic Fibrosis
- Ashke- azi Jewish Pa- el
  - 9 disorders i- cludi- g Tay-Sachs a- d Ca- ava- disease
- BRCA1/2
- MTHFR
- Multiple e- docri- e - eoplasia , Type 2A
- Alpha1-a- titrypsi- deficie- cy
- Pharmacoge- etics (20 loci)
- Duche- - e muscular dystrophy
- Myoto- ic dystrophy

## O- goi- g Projects

- Rett Sy- drome
- Next-Ge- Seque- ci- g
- Cytoge- etics
- Molecular o- cology
- HLA (6 loci)

# Pharmacogenetics 1

- Involved 13 laboratories (clinical + commercial)
- Assays: laboratory developed tests + many commercial platforms
- Characterized 107 DNA samples for 20 loci (same samples were used in a HLA study)
  - 5 mai- pharmacoge- etic loci (*CYP2D6, CYP2C19, CYP2C9, VKORC1 and UGT1A1*) characterized by multiple labs
  - 9 other pharmacoge- etic loci characterized by 1 lab (*CYP4F2, EPHX1, ABCB1, HLAB, KIF6, CYP3A4, CYP3A5, TPMT, DPD*)
  - 6 - o- -pharmacoge- etic loci characterized by 1 or 2 labs (*F5, F2, HFE, MTHFR, AAT, PAI1*)

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# Characterization of 107 Genomic DNA Reference Materials for *CYP2D6*, *CYP2C19*, *CYP2C9*, *VKORC1*, and *UGT1A1*

## *A GeT-RM and Association for Molecular Pathology Collaborative Project*

Victoria M. Pratt,* Barbara Zehnbauer,†‡ Jean Amos Wilson,§ Ruth Baak,¶ Nikolina Babic,‖ Maria Bettinotti,* Arlene Buller,** Ken Butz,†† Matthew Campbell,‡‡ Chris Civalier,§§ Abdalla El-Badry,¶ Daniel H. Farkas,¶¶ Elaine Lyon,‡‡ Saptarshi Mandal,‖ Jason McKinney,‖ Kasinathan Muralidharan,* LeAnne Noll,*** Tara Sander,*** Junaid Shabbeer,§ Chingying Smith,¶ Milhan Telatar,††† Lorraine Toji,‡‡‡ Anand Vairavan,¶ Carlos Vance,‡‡ Karen E. Weck,§§ Alan H.B. Wu,§§§ Kiang-Teck J. Yeo,‖ Markus Zeller,¶ and Lisa Kalman†

*From the Quest Diagnostics Nichols Institute,* Chantilly Virginia; the Centers for Disease Control and Prevention,† Atlanta, Georgia; Washington University School of Medicine,‡ St. Louis, Missouri; Berkeley HeartLab, Inc.,§ South San Francisco, California; AutoGenomics Inc.,¶ Vista, California; University of Chicago,‖ Chicago, Illinois; Quest Diagnostics, Nichols Institute,** San Juan Capistrano, California; ParagonDx, LLC,†† Morrisville, North Carolina; ARUP Laboratories,‡‡ Salt Lake City, Utah; University of North Carolina,§§ Chapel Hill, North Carolina; Sequenom Center for Molecular Medicine,¶¶ Grand Rapids, Michigan; Idaho Technology, Inc.,‖‖ Salt Lake City, Utah; Children's Hospital of Wisconsin,*** Milwaukee, Wisconsin; Specialty Laboratories, Inc.,††† Valencia, California; Coriell Cell Repositories,‡‡‡ Camden, New Jersey; and the University of California,§§§ San Francisco, California

**Pharmacogenetic testing is becoming more common; however, very few quality control and other reference materials that cover alleles commonly included in such assays are currently available. To address these needs, the Centers for Disease Control and Prevention's Genetic Testing Reference Material Coordination Program, in collaboration with members of the pharmacogenetic testing community and the Coriell Cell Repositories, have characterized a panel of 107 genomic DNA reference materials for five loci (*CYP2D6*, *CYP2C19*, *CYP2C9*, *VKORC1*, and *UGT1A1*) that are commonly included in pharmacogenetic testing panels and proficiency testing surveys. Genomic DNA from publicly available cell lines was sent to volunteer laboratories for genotyping. Each sample was tested in three to six laboratories using a variety of commercially available or laboratory-developed platforms. The results were consistent among laboratories, with differences in allele assignments largely related to the manufacturer's assay design and variable nomenclature, especially for *CYP2D6*. The alleles included in the assay platforms varied, but most were identified in the set of 107 DNA samples. Nine additional pharmacogenetic loci (*CYP4F2*, *EPHX1*, *ABCB1*, *HLAB*, *KIF6*, *CYP3A4*, *CYP3A5*, *TPMT*, and *DPD*) were also tested. These samples are publicly available from Coriell and will be useful for quality assurance, proficiency testing, test development, and research.** *(J Mol Diagn 2010, 12:835–846; DOI: 10.2353/jmoldx.2010.100090)*

Many laboratories are testing for pharmacogenetic (PGx) markers, common genetic variants that are usually considered only when a patient is likely to be exposed to a

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R.B., A.E.-B., C.S., A.V., and M.Z. are employees of AutoGenomics (manufacturer of several pharmacogenetic assays used in this study); M.B., A.B., and K.M. are employees of Quest Diagnostics Inc.; J.M. is an employee of Idaho Technology (manufacturer of the reagents used to genotype *CYP2C9* and *VKORC1* loci for this project); L.N. and T.S. received donated reagents from Applied Biosystems Inc.; L.T. is employed by the Coriell Institute for Medical Research, Coriell Cell Repositories, which maintains and distributes these biomaterials to investigators for research, education and use as reference materials; K.W. is a consultant to ParagonDx. V.P. is an employee of Quest Diagnostics and owns stock.

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Standard of practice is not being defined by this article, and there may be alternatives.

Address reprint requests to Lisa Kalman, Ph.D., Laboratory Research and Evaluation Branch, Division of Laboratory Science and Standards, Office of Surveillance, Epidemiology, and Laboratory Services, Centers for Disease Control and Prevention, 1600 Clifton Road, Mailstop G23, Atlanta, GA 30333. E-mail: LKalman@cdc.gov.



**Pratt, VM et al.**
**J Mol Diag 2010 12:835-846**

# Pharmacogenetics 2

- Second study to increase number of characterized pharmacogenetic genes/alleles
- Compared a number of available PGx panel tests (231 pharmacogenetic loci, over 2000 SNPs)
- Selected 137 Coriell cell lines that contained variants in as many of the SNPs included in the PGx panels as possible

# Pharmacogenetics 2

Study Design:

DNA from 137 Coriell cell lines are being characterized in 9 labs with a number of PGx platforms:

- Affymetrix DMET (231 genes, 1931 SNPs)
- Sequenom iPLEX ADME (36 genes)
- Sequenom iPLEX expanded panels (*CYP2D6, CYP2C19, VKORC1, CYP2C9, UGT1A1*)
- AutoGenomics (*CYP2D6, CYP3A4, CYP3A5, NAT2*)
- GenMark (*CYP2C19, CYP3A4/3A5, CYP2C9/VKORC1*)
- Luminex (*CYP2D6, CYP2C19, CYP2C9/VKORC1*)
- Custom Life Tech Taqman array- (17 genes)
- Targeted NGS assays – 3 labs
- LifeTech Ion Torrent NGS assay- 2 labs

# Pharmacogenetics 2

Participating Laboratories:

- Robin Everts- Agena Biosciences (Sequenom)
- Uli Broeckel/Praful Aggarwal- University of Wisconsin
- Stuart Scott- Icahn School of Medicine at Mount Sinai
- Ruth Baak- AutoGenomics
- Vicky Pratt- Indiana University
- Elaine Mardis/Bob Fulton- Washington University
- Steven Scherer- Baylor Medical College
- Debbie Nickerson/Adam Gordon- University of Washington
- Audrey Papp/Wolfgang Sadee- Ohio State University

# Nomenclature Issues Identified During the PGx 2 Study

# Nomenclature Issues

Not all assays test the same SNPs for each gene, which can lead to incorrect assignment of haplotypes

Example: CYP2C19

- *4 (1A>G; 99C>T, 80161A>G)
- *4B (-3402C>T; -806C>T; 1A>G; 99C>T; 80161C>T)
- *17 (-3402C>T; -806C>T; 99C>T; 80161C>T)

If platform does not test for *4 (c.1A>G), how does one distinguish *4B (no activity) or *17 ( increased activity)?

# Nomenclature Issues

**Laboratories report genotype of same gene using different nomenclature**

## Example 1: SLC22A2

| Sample | | Lab 1 | Lab 2 |
|---|---|---|---|
| PGxT117 | | *1/*2B,*2A/*3A | WT / WT |
| PGxT118 | | *3D/*3D | S270A / S270A |
| PGxT119 | | *2B/*2B | WT / WT |
| PGxT120 | | *1/*2B,*2A/*3A | WT / WT |
| PGxT121 | | *1/*1 | WT / WT |
| PGxT122 | | *1/*3E,*2A/*3D,*2B/*6;*3A/UNK | WT / S270A |
| PGxT123 | | -;*1/UNK,*7/UNK | R400C / K432Q |
| PGxT124 | | *1/*7 | WT / R400C |

# Nomenclature Issues

**Laboratories report ge- otype of same ge- e usi- g differe- t - ome- clature**

## Example 2: VKORC1

| Sample | Lab 1 | Lab2 | Lab3 | Lab4 | Lab5 | Lab6 | Lab7 |
|---|---|---|---|---|---|---|---|
| PGxT101 | H2/H5 / H2/H5 | H2/H2 | AA | AA | *2/*2 | *2 / *2 | AA |
| PGxT102 | *2B / *3; H2/H5 / H7B | H2/H7;H6/UNK,H1/UNK,H3/UNK,H4/UNK,UNK/UNK | GA | GA | *1/*2 | *2 / *3 | AG |
| PGxT103 | H2/H5 / H2/H5 | H2/H2 | AA | AA | *2/*2 | *2 / *2 | AA |
| PGxT104 | *2/H1 / H7B; *2A / *3 | H1/H7;H6/UNK,H3/UNK,H4/UNK,UNK/UNK | GA | GA | *1/*2 | *2 / *3 | AG |
| PGxT105 | BHT2RE / BHT2RE | H9/H9 | GG | GG | *1/*1 | *4 / *4 | GG |
| PGxT106 | *2/H1 / *2/H1 | H1/H1 | AA | AA | *2/*2 | *2 / *2 | AA |

# Nomenclature Issues

**Laboratories report ge- otype of same ge- e usi- g differe- t - ome- clature**

**AND**
**Differe- t assays look at differe- t SNPs**

## Example 2: VKORC1

| Sample | Lab 1 | Lab2 | Lab3 | Lab4 | Lab5 | Lab6 | Lab7 |
|---|---|---|---|---|---|---|---|
| PGxT101 | H2/H5 / H2/H5 | H2/H2 | AA | AA | *2/*2 | *2 / *2 | AA |
| PGxT102 | *2B / *3; H2/H5 / H7B | H2/H7;H6/UNK,H1/UNK,H3/UNK,H4/UNK,UNK/UNK | GA | GA | *1/*2 | *2 / *3 | AG |
| PGxT103 | H2/H5 / H2/H5 | H2/H2 | AA | AA | *2/*2 | *2 / *2 | AA |
| PGxT104 | *2/H1 / H7B; *2A / *3 | H1/H7;H6/UNK,H3/UNK,H4/UNK,UNK/UNK | GA | GA | *1/*2 | *2 / *3 | AG |
| PGxT105 | BHT2RE / BHT2RE | H9/H9 | GG | GG | *1/*1 | *4 / *4 | GG |
| PGxT106 | *2/H1 / *2/H1 | H1/H1 | AA | AA | *2/*2 | *2 / *2 | AA |

## Problems reporting sequence results: which strand?

- The Affymetrix DMET and other assays report genotypes from the coding strand and do not refer to the Human Reference Genome (e.g. hg19)

- Other sequencing assays report from the Human Reference Genome (not always same as coding strand)

## Problems reporting sequence results: reference genotype

**Example: rs10276036**

- Reference genotype for Affymetrix DMET= AA
- Reference genotype for deep sequencing labs = CC

Where does AA come from? It is based on the most common allele observed in HapMap data.

## Problems reporting sequence results: SNPs with non-variant calls

- Sequencing labs only report genotypes that are different from the reference sequence (this is how the VCF is usually structured)

- In order to determine the haplotype, it is important to know which SNPs were tested, but found to be the same as reference!

# Problems reporting sequence results: SNPs with non-variant calls

| | Ge- otype data Assay # | | | | | Refere- ce ge- otype Assay # | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| rsSNP | 1 | 2 | 3 | 4 | 5 | 1 | 2 | 3 | 4 | 5 |
| RS10276036 | | AG | CT | CT | CT | | AA | CC | CC | CC |
| RS1045642 | CT | CT | AG | AG | AG | CC | CC | AA | AA | AA |
| RS1128501 | | GG | | | | | GG | | | |
| RS1128502 | | AA | | | | | AA | | | |
| RS1128503 | CT | CT | AG | AG | AG | CC | CC | AA | AA | AA |
| RS1202183 | | AA | | | | | AA | | | |
| RS17064 | | AA | | | | | AA | | | |
| RS2032581 | | AA | | | | | AA | | | |
| RS2032582 | GT | GT | AC | AC | AC | GG | GG | AA | AA | AA |

Yellow shading indicates discordant data

# Problems reporting se
# SNPs with non-v

Assays use differe- t refere- ce seque- ce

| | Ge- otype Assay # | | | | | Refere- ce ge- otype Assay # | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| rsSNP | 1 | 2 | 3 | 4 | 5 | 1 | 2 | 3 | 4 | 5 |
| RS10276036 | | AG | CT | CT | CT | | AA | CC | CC | CC |
| RS1045642 | CT | CT | AG | AG | AG | CC | CC | AA | AA | AA |
| RS1128501 | | GG | | | | | GG | | | |
| RS1128502 | | AA | | | | | AA | | | |
| RS1128503 | CT | CT | AG | AG | AG | CC | CC | AA | AA | AA |
| RS1202183 | | AA | | | | | AA | | | |
| RS17064 | | AA | | | | | AA | | | |
| RS2032581 | | AA | | | | | AA | | | |
| RS2032582 | GT | GT | AC | AC | AC | GG | GG | AA | AA | AA |

# Problems reporting sequence results: SNPs with non-variant calls

Some assays do - ot report ge- otypes of SNPs with - o- - varia- t ge- otypes

Refere- ce ge- otype

| rsSNP | Assay # 1 | 2 | 3 | 4 | 5 | 1 | 2 | 3 | 4 | 5 |
|---|---|---|---|---|---|---|---|---|---|---|
| RS10276036 | | AG | CT | CT | CT | | AA | CC | CC | CC |
| RS1045642 | CT | CT | AG | AG | AG | CC | CC | AA | AA | AA |
| RS1128501 | | GG | | | | | GG | | | |
| RS1128502 | | AA | | | | | AA | | | |
| RS1128503 | CT | CT | AG | AG | AG | CC | CC | AA | AA | AA |
| RS1202183 | | AA | | | | | AA | | | |
| RS17064 | | AA | | | | | AA | | | |
| RS2032581 | | AA | | | | | AA | | | |
| RS2032582 | GT | GT | AC | AC | AC | GG | GG | AA | AA | AA |

## Nomenclature?

## Next Steps:

Develop standardized nomenclature for pharmacogenetics!

- Assembled group of stakeholders including:
  - Testing laboratories
  - Commercial test developers
  - Database providers (NIH/NCBI and gene specific)
  - Gene specific nomenclature groups
  - Pharmacogenomics Research Network (PGRN)
  - PharmGKB
  - Clinical Pharmacogenetics Implementation Consortium (CPIC)
  - HUGO, HGVS Nomenclature curators
- Have started discussions to identify issues and possible solutions

# Possible Solutions???

**Problem**

- Not all assays test the same SNPs for each gene. Lack of standardization can cause ambiguous haplotype assignments.

**Solution?**

- Create defined list of SNPs that MUST be assayed for each gene (similar to ACMG list of 23 cystic fibrosis variants)-based on CPIC recommendations?
- Labs must clearly state the genotype (variant or not variant) of each SNP on the “must test” list, or indicate that the SNP was not tested.

# Possible Solutions???

**Problem**

- There are many nomenclatures used to report results of a single gene

**Solution?**

- Use HGVS nomenclature to describe variant SNPs
- Refer to rsSNP #
- Can also state results in * nomenclature to provide continuity

# Possible Solutions???

**Problem**

- Not all platforms define the reference sequence of the same SNPs in the same way (some use the coding strand, some use the positive strand, some use reference genotype from a different population)

**Solution?**

- Use the Human Reference Genome (e.g. hg19) and/or LRG as reference sequence

## Other issues to consider…

- How can non-variant SNPs (i.e. SNPs with the reference sequence) be represented in VCF? (forced calling?)
- How to deal with sequencing data that detects many new variants that do not have described haplotypes?
- How should databases handle changing nomenclature? (esp. related to *alleles)
- How should haplotypes be represented in databases such as ClinVar?

# Standardized haplotype nomenclature

**Developed prototype tables of VKORC1, TPMT and CYP2C19**

- The rsSNPs that compose each haplotype are shown
- Table can be used to translate between different nomenclature systems
- HGVS for each SNP is provided

# Standardized haplotype nomenclature

| VKORC1: Official Haplotypes and Nomenclature Version: Sept. 30, 2014 | | | yellow- SNPs that define * haplotypes | Developed by Dr. Johan den Dunnen and Dr. Lisa Kalman based on tables from PharmGKB. Contact Lisa Kalman (Lkalman@cdc.gov) for questions or to provide feedback | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| **Genome (GRCh38, hg38)** | NC_000016.10 | g.31099660G>A | g.31099180G>T | g.31097388C>G | g.31096368C>T | g.31094233A>C | g.31094032G>A | g.31093557G>A | g.31093188C>G | g.31092475A> |
| **Genome (GRCH37, hg19)** | NC_000016.9 | g.31110981G>A | g.31110501G>T | g.31108709C>G | g.31107689C>T | g.31105554A>C | g.31105353G>A | g.31104878G>A | g.31104509C>G | g.31103796A> |
| **Reference LRG** | ? | | | | | | | | | |
| **Reference Gene (RefSeqGene)** | NG_011564.1 | g.296C>T | g.776C>A | g.2568G>C | g.3588G>A | g.5723T>G | g.5924C>T | g.6399C>T | g.6768G>C | g.7481T>C |
| **Reference Transcript** | NM_206824.1 | c.-4931C>T | c.-4451C>A | c.-2659G>C | c.-1639G>A | c.173+324T>G | c.173+525C>T | c.173+1000C>T | c.173+1369G>C | c.174-1133T>C |
| **Reference Protein** | | | | | | | | | | |
| **Common name?** | | | | | -1639G>A | | 6009C>T | 6484C>T | | 7566C>T |
| **LOCATION in gene** | | 5' exon 1 | 5' exon 1 | 5' exon 1 | 5' exon 1 | 1i | 1i | 1i | 1i | 1i |
| **Haplotype Id** | **VKORC1** | **rs7196161** | **rs17880887** | **rs17881535** | **rs9923231** | **rs2884737** | **rs17708472** | **rs9934438** | **rs8050894** | **rs2359612** |
| PA165816585 | H1 (consistent with *2) | G | G | C | T | A | G | A | G | A |
| PA165816586 | H2 (consistent with *2) | G | G | C | T | C | G | A | G | A |
| PA165816587 | H3 (consistent with *1) | G | G | C | C | A | G | G | G | G |
| PA165816588 | H4 | G | G | C | C | A | G | G | C | A |
| PA165816589 | H5 (consistent with *2) | A | G | C | T | C | G | A | G | A |
| PA165816590 | H6 (consistent with *1) | A | G | C | C | A | G | G | C | G |
| PA165816591 | H7 (consistent with *3) | A | G | C | C | A | G | G | C | G |
| PA165816592 | H8 (consistent with *3) | A | T | C | C | A | G | G | C | G |
| PA165816593 | H9 (consistent with *4) | A | T | G | C | A | A | G | C | G |

| VKORC1: Official Haplotypes and Nomenclature Version: Sept. 30, 2014 | | | yellow- SNPs that define * haplotypes | Developed by Dr. Johan de based on tables from Pha (Lkalman@cdc.gov) for qu | | |
|---|---|---|---|---|---|---|
| **Genome (GRCh38, hg38)** | NC_000016.10 | g.31099660G>A | g.31099180G>T | g.31097388C>G | g.31096368C>T | g.31094233A>C |
| **Genome (GRCH37, hg19)** | NC_000016.9 | g.31110981G>A | g.31110501G>T | g.31108709C>G | g.31107689C>T | g.31105554A>C |
| **Reference LRG** | ? | | | | | |
| **Reference Gene (RefSeqGene)** | NG_011564.1 | g.296C>T | g.776C>A | g.2568G>C | g.3588G>A | g.5723T>G |
| **Reference Transcript** | NM_206824.1 | c.-4931C>T | c.-4451C>A | c.-2659G>C | c.-1639G>A | c.173+324T>G |
| **Reference Protein** | | | | | | |
| **Common name?** | | | | | -1639G>A | |
| **LOCATION in gene** | | 5' exon 1 | 5' exon 1 | 5' exon 1 | 5' exon 1 | 1i |
| **Haplotype Id** | **VKORC1** | **rs7196161** | **rs17880887** | **rs17881535** | **rs9923231** | **rs2884737** |
| PA165816585 | H1 (consistent with *2) | G | G | C | T | A |
| PA165816586 | H2 (consistent with *2) | G | G | C | T | C |
| PA165816587 | H3 (consistent with *1) | G | G | C | C | A |
| PA165816588 | H4 | G | G | C | C | A |
| PA165816589 | H5 (consistent with *2) | A | G | C | T | C |
| PA165816590 | H6 (consistent with *1) | A | G | C | C | A |
| PA165816591 | H7 (consistent with *3) | A | G | C | C | A |
| PA165816592 | H8 (consistent with *3) | A | T | C | C | A |
| PA165816593 | H9 (consistent with *4) | A | T | G | C | A |

# Next Steps….

- The PGx Nomenclature Workgroup will continue to discuss these issues
- Sub-workgroups will develop models to show how our proposed nomenclature could be used to address problems identified in the nomenclature. We plan to make models using 4 genes: TPMT, CYP2C19, VKORC1 and NAT2
- We are preparing a manuscript that describes the issues with PGx nomenclature and our suggestions for addressing these problems

Lisa Kalman: Lkalman@cdc.gov

**For more information please contact Centers for Disease Control and Prevention**

1600 Clifton Road NE, Atlanta, GA 30333
Telephone: 1-800-CDC-INFO (232-4636)/TTY: 1-888-232-6348
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