PharmCAT: A Tool for Pharmacogenomics Implementation

Marylyn D. Ritchie, PhD

Director, Biomedical and Translational Informatics Institute

Geisinger Health System

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Geisinger PharmGKB Stanford

in a gene known as BRCA1. A BRCA1 mutation gives her a much higher lifetime risk of developing breast cancer and ovarian cancer. A related mutation in a gene known as BRCA2, which Ms. Barnes does not have, also increases breast and ovarian cancer risk. Her results mean that close biological relatives also could have a BRCA1 mutation. About 5 to 10 percent of all breast cancers are linked to one of these two mutations.

Ms. Barnes, who is 57, and her husband are bringing up three grandchildren, ages 3, 5 and 14. That responsibility kept her calm when she heard her result. "I just said, 'Okay, so what do we do next?'" she recalls. "I have 15 more years to go until they're raised."

A mammogram showed no breast cancer. She and her daughter met with a Geisinger genetic counselor. Her daughter was tested for the BRCA1 mutation. Ms. Barnes talked about prevention with breast surgeon and an oncologist, or cancer doctor. She will have more frequent mammograms and MRIs to closely monitor her breast health. She also decided to have her ovaries and fallopian tubes surgically removed as a preventive measure, lowering her risk of ovarian cancer to almost none.

During the surgery this past summer, her doctor, Ashlee Linn Smith, DO, a gynecologic oncologist, found cancer in one of Ms. Barnes' fallopian tubes. Such cancer is sometimes found during the preventive surgery. The cancer was removed and Ms. Barnes is having chemotherapy. "If I hadn't been in MyCode, I wouldn't have known," she says.

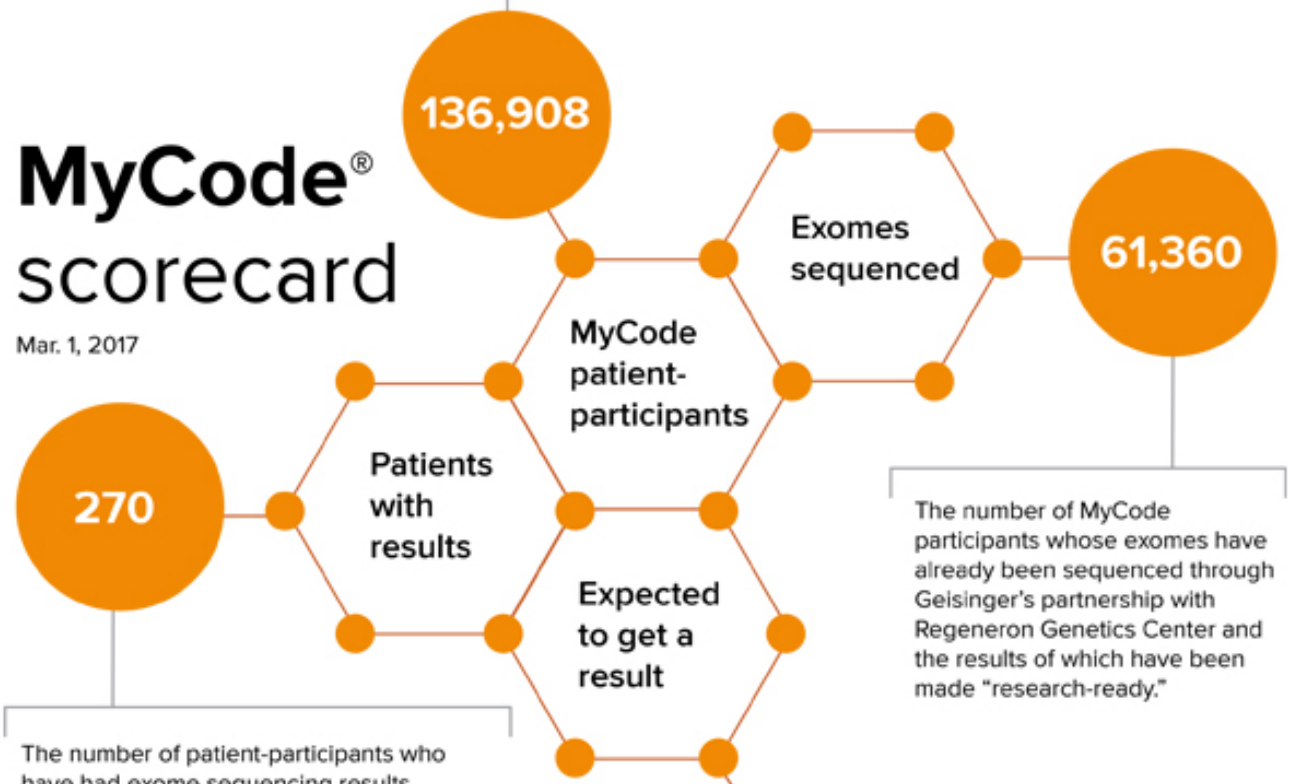
- Barbara Barnes, 57
- Pathogenic *BRCA1* mutation
- Mammogram → no evidence of cancer
- Preventive surgery to remove ovaries and fallopian tubes

- During surgery, cancer found in one of her fallopian tubes

Geisinger Magazine, Fall 2016

MyCode® scorecard

Mar. 1, 2017



The number of Geisinger patient-participants who have consented to take part in the MyCode Community Health Initiative. The number is growing monthly.

The number of MyCode participants whose exomes have already been sequenced through Geisinger's partnership with Regeneron Genetics Center and the results of which have been made "research-ready."

The number of patient-participants who have had exome sequencing results returned to them so far – meaning disease-causing gene variants were found in the MyCode "DiscovEHR" Study; these were verified by an independent, certified lab and that information was returned to the patient through established protocols.

The estimated percentage of MyCode participants who are expected to receive a clinical result indicating that they have a disease-causing variant or variants in one or more of their genes.

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ACMG STATEMENT | Genetics inMedicine

Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2016 update (ACMG SF v2.0): a policy statement of the American College of Medical Genetics and Genomics

Sarah S. Kalia, ScM[1], Kathy Adelman[2], Sherri J. Bale, PhD[3], Wendy K. Chung, MD, PhD[4,5], Christine Eng, MD[6], James P. Evans, MD, PhD[7], Gail E. Herman, MD, PhD[8], Sophia B. Hufnagel, MD[9], Teri E. Klein, PhD[10], Bruce R. Korf, MD, PhD[11], Kent D. McKelvey, MD[12,13], Kelly E. Ormond, MS[10], C. Sue Richards, PhD[14], Christopher N. Vlangos, PhD[15], Michael Watson, PhD[16], Christa L. Martin, PhD[17], David T. Miller, MD, PhD[18]; on behalf of the ACMG Secondary Findings Maintenance Working Group

Disclaimer: These recommendations are designed primarily as an educational resource for medical geneticists and other healthcare providers to help them provide quality medical services. Adherence to these recommendations is completely voluntary and does not necessarily assure a successful medical outcome. These recommendations should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed toward obtaining the same results. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this statement. Clinicians also are advised to take notice of the date this statement was adopted and to consider other medical and scientific information that becomes available after that date. It also would be prudent to consider whether intellectual property interests may restrict the performance of certain tests and other procedures.

To promote standardized reporting of actionable information from clinical genomic sequencing, in 2013, the American College of Medical Genetics and Genomics (ACMG) published a minimum list of genes to be reported as incidental or secondary findings. The goal was to identify and manage risks for selected highly penetrant genetic disorders through established interventions aimed at preventing or significantly reducing morbidity and mortality. The ACMG subsequently established the Secondary Findings Maintenance Working Group to develop a process for curating and updating the list over time. We describe here the new process for accepting and evaluating nominations for updates to the secondary findings list. We also report outcomes from six nominations received in the initial 15 months after the process was implemented. Applying the new process while upholding the core principles of the original policy statement resulted in the addition of four genes and removal of one gene; one gene did not meet criteria for inclusion. The updated secondary findings minimum list includes 59 medically actionable genes recommended for return in clinical genomic sequencing. We discuss future areas of focus, encourage continued input from the medical community, and call for research on the impact of returning genomic secondary findings.

Genet Med advance online publication 17 November 2016

Key Words: exome sequencing; genetic testing; genome sequencing; incidental findings; secondary findings

REPORTER
Are You a Patient?
Click here to visit www.VanderbiltHealth.com
VANDERBILT UNIVERSITY MEDICAL CENTER'S WEEKLY NEWSPAPER

PREDICT helps match patient with proper drug

BY: KATHY WHITNEY

Scyble Van Cleve, right, is the first patient at Vanderbilt to benefit from a new program that puts genetic information in the patient's medical records to help physicians like John McPherson, M.D., choose the drug and dose that will benefit them the most. (photo by Susan Urmy)

10/28/2010 - Had Scyble Van Cleve, a spry 83-year-old from Brentwood, had her heart procedure done a month ago instead of one week ago, she would have been prescribed the standard dose of clopidogrel, a blood thinner used to prevent blood clots from forming around her coronary stents.

Her doctors may not have known that, based on her genes, she needed a different blood-thinning regimen to safeguard her from possible fatal complications.

Thanks to a novel program implemented at Vanderbilt called PREDICT, Van Cleve

PRINT
E-MAIL

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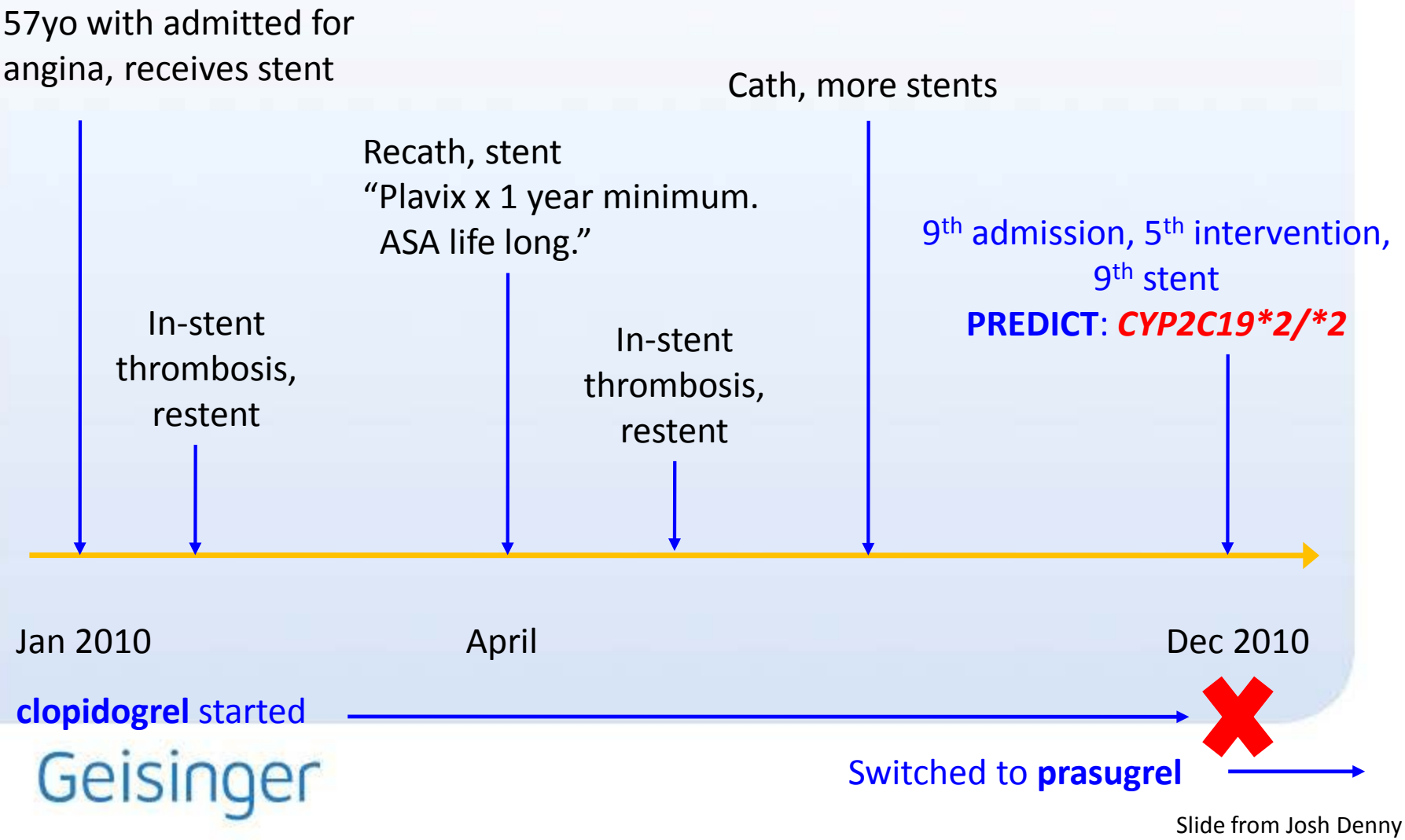
Vanderbilt case: Implementing *CYP2C19* for Clopidogrel



Slide from Josh Denny

What else can we implement?
How do we decide what to implement?

28 CPIC guideline publications* (including updates)

- Genetic information should be used to change prescribing of affected drug
- Preponderance of evidence is high or moderate in favor of changing prescribing
- At least one moderate or strong action (change in prescribing) recommended

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*https://cpicpgx.org/publications/

I've built an automated bioinformatics pipeline for ACMG genes or a few PGx genes, so I can implement all CPIC guidelines now, right?

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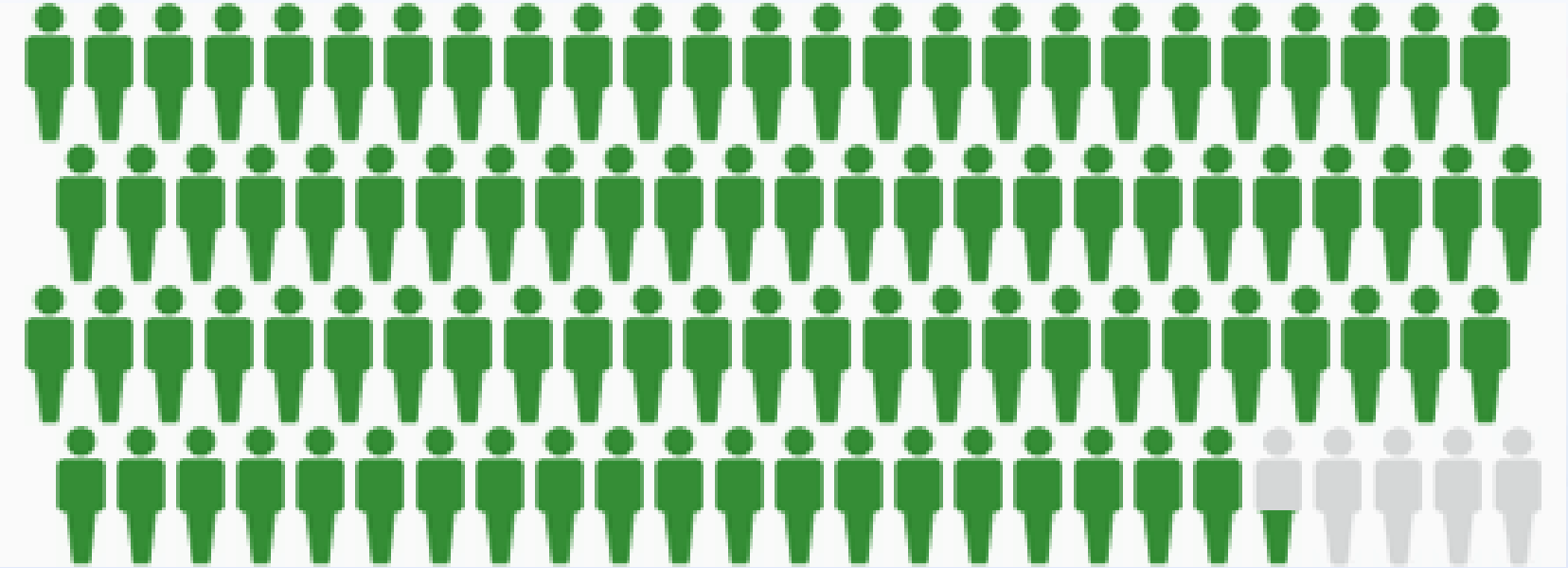
Motivation for PharmCAT - #1



95-96% of individuals have one or more genetic variants in important PGx genes

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Genetic Variation Among 82 Pharmacogenes: The PGRNseq Data From the eMERGE Network

WS Bush[1], DR Crosslin[2], A Owusu-Obeng[3], J Wallace[4], B Almoguera[5], MA Basford[6], SJ Bielinski[7], DS Carrell[8], JJ Connolly[5], D Crawford[1], KF Doheny[9], CJ Gallego[2], AS Gordon[2], B Keating[5], J Kirby[6], T Kitchner[10], S Manzi[11], AR Mejia[3], V Pan[12], CL Perry[11], JF Peterson[6], CA Prows[13], J Ralston[8], SA Scott[3], A Scrol[8], M Smith[12], SC Stallings[6], T Veldhuizen[7], W Wolf[11], S Volpi[14], K Wiley[14], R Li[14], T Manolio[14], E Bottinger[3], MH Brilliant[10], D Carey[15], RL Chisholm[12], CG Chute[9], JL Haines[1], H Hakonarson[5], JB Harley[16], IA Holm[17], IJ Kullo[7], GP Jarvik[2], EB Larson[8], CA McCarty[10], MS Williams[15], JC Denny[6], LJ Rasmussen-Torvik[12], DM Roden[6] and MD Ritchie[15]

The first two authors contributed equally to this work.

[1]Case Western Reserve University, Cleveland, Ohio, USA; [2]University of Washington, Seattle, Washington, USA; [3]Charles Bronfman Institute for Personalized Medicine, Icahn School of Medicine at Mount Sinai, New York, New York, USA; [4]Pennsylvania State University, University Park, Pennsylvania, USA; [5]Children's Hospital of Philadelphia, Philadelphia, Pennsylvania, USA; [6]Vanderbilt University School of Medicine, Nashville, Tennessee, USA; [7]Mayo Clinic, Rochester, Minnesota, USA; [8]Group Health Research Institute, Seattle, Washington, USA; [9]Johns Hopkins University School of Medicine, Baltimore, Maryland, USA; [10]Marshfield Clinic, Marshfield, Wisconsin, USA; [11]Boston Children's Hospital, Boston, Massachusetts, USA; [12]Northwestern University Feinberg School of Medicine, Evanston, Illinois, USA; [13]Children's Hospital Medical Center, Cincinnati, Ohio, USA; [14]National Human Genome Research Institute, Bethesda, Maryland, USA; [15]Geisinger Health System, Danville, Pennsylvania, USA; [16]University of Cincinnati, US Department of Veterans Affairs Medical Center, Cincinnati, Ohio, USA; [17]Department of Pediatrics, Division of Genetics and Genomics and The Manton Center for Orphan Disease Research, Boston, Massachusetts, USA. Correspondence: MD Ritchie (marylyn.ritchie@psu.edu)

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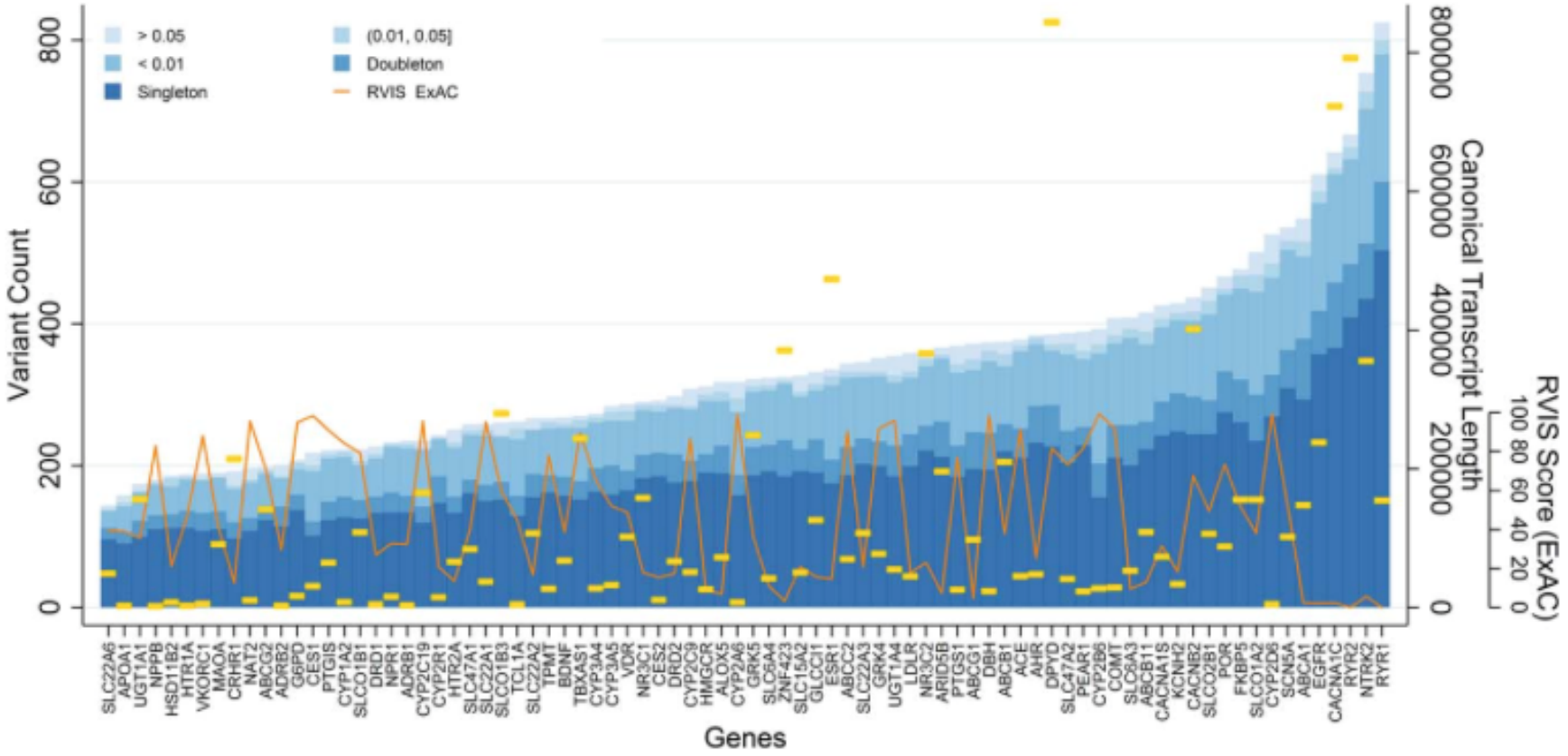

Figure 1 Allelic spectrum of eMERGE-PGx variants. Counts of genomic variants mapping to the canonical transcript of PGRNseq captured genes are plotted by frequency class (over all samples) by gene (x-axis) in ascending order. Gold horizontal lines indicate the size of the canonical transcript in base-pairs. The inset line plot is a percentile rank of genic intolerance (RVIS) scores computed using the ExAC dataset.

~96% of the 5000 subjects has one or more variants in CPIC level-A genes

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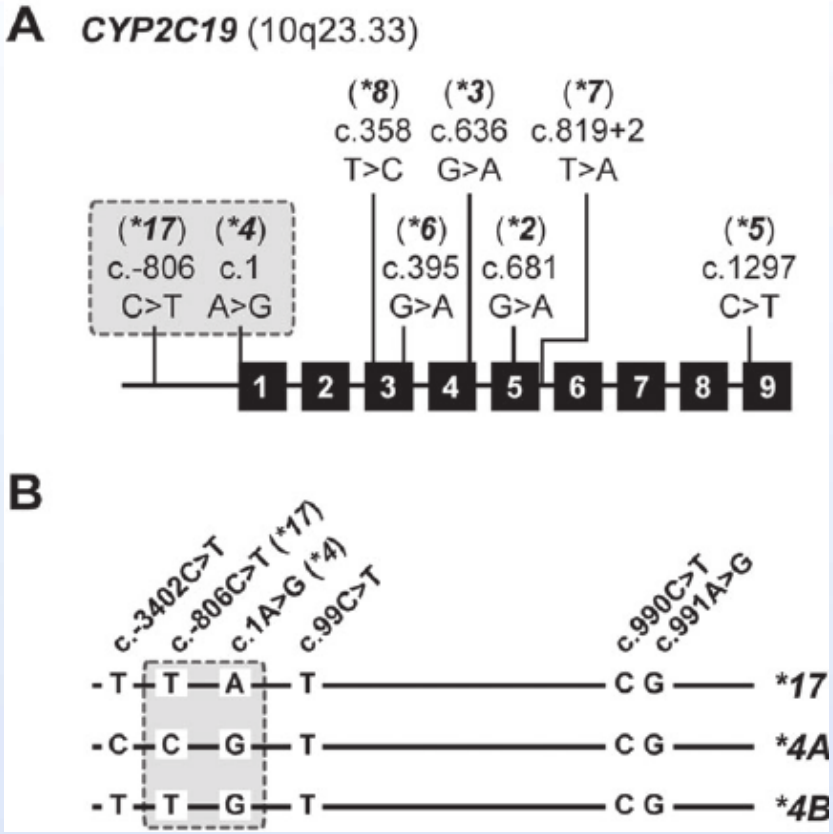
Motivation for PharmCAT - #2



Extracting genomic variants and assigning haplotypes (including star-alleles) from genetic data is challenging

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Motivation for PharmCAT

To automate the annotation of .vcf files with the appropriate haplotypes or diplotypes from the CPIC guideline genes, and generate a report with the corresponding CPIC guideline prescribing recommendations

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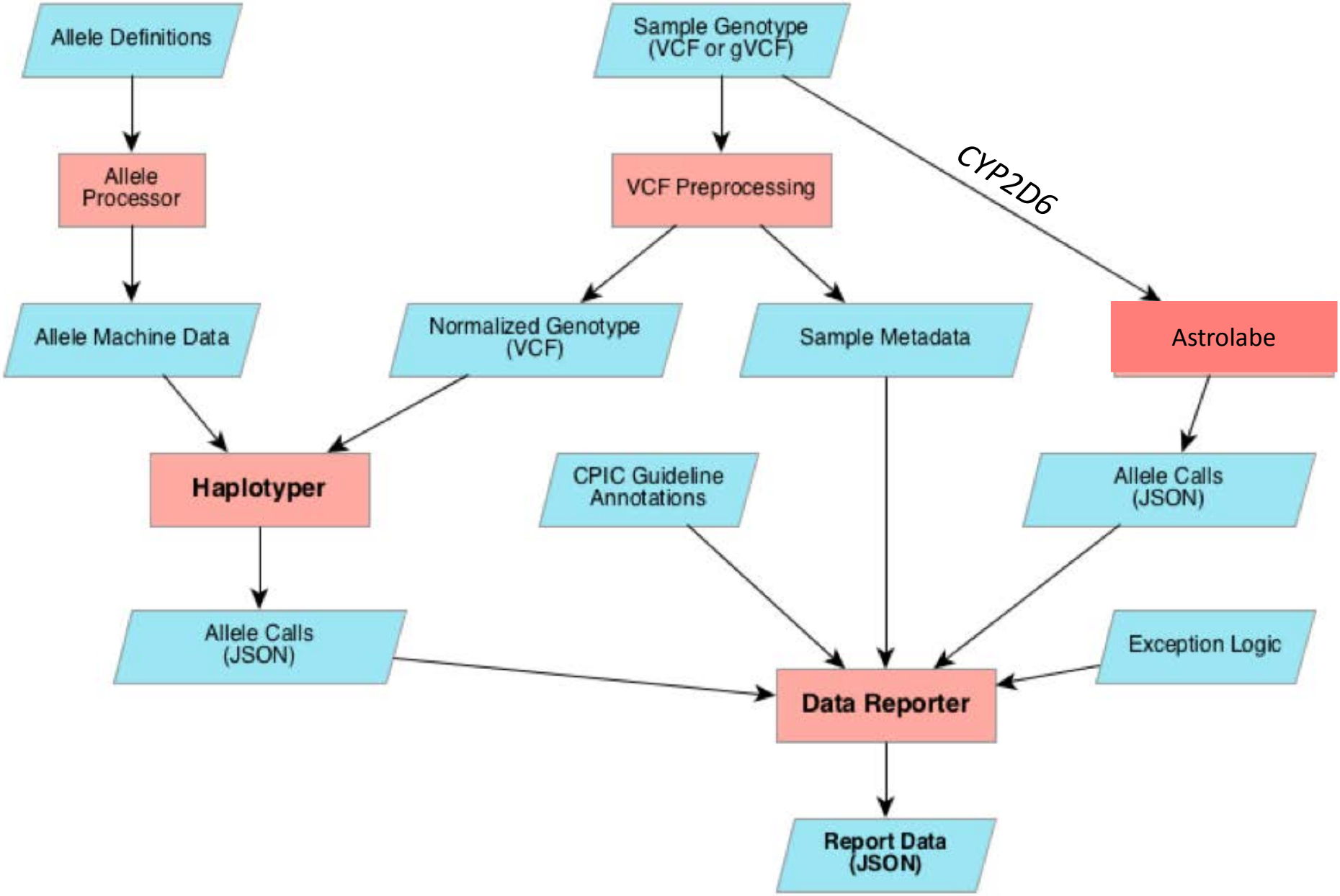
PharmCAT – a Community Effort

- Rules of engagement
  - MPL 2.0 license
  - Code posted in github – updates need to be contributed
- Several collaborative meetings
  - Meeting 1 – March, 2016: think tank
  - Meeting 2 – April, 2016: Hackathon – for programmers
  - Meeting 3 – May, 2016: dissemination, future planning
  - Meeting 4 – January, 2017: evaluation, coding, planning

PharmCAT Workflow



CPIC Guidelines

PharmCAT version 1

- These genes are in process for release in PharmCAT version 1.0
  - *CFTR, CYP2C19, CYP2C9, CYP2D6, CYP3A5, CYP4F2, DPYD, IFNL3, SLCO1B1, TPMT, UGT1A1, VKORC1*
- These genes are more difficult and require more work: *G6PD, HLA-B, CYP2D6**
- *CYP2D6* haplotype calls are coming from Astrolabe
  - Integrated into PharmCAT
  - Will require user license to Astrolabe
  - JSON calls join Data Reporter for PharmCAT report

CYP3A5

- Why is this complicated?

Haplotype Set Id	PA166128212	7/22/15	R28C			Q200R		A337T	T346fs	T398N
Haplotype Set Name	Haplotype Set	*Nucleotide	3699C>T	6986A>G	12952T>C	14665A>G	14690G>A	19386G>A	27131_27132insT	27289C>A
Haplotype Id	CYP3A5	TAG(S)	rs55817950	rs776746	NG_000004.3:g.253133T>C	rs56411402	rs10264272	rs28383479	rs41303343	rs28365083
PA166128213	*1		G	T	A	T	C	C	del	G
PA166128218	*2									T
PA166128219	*3			C						
PA166128231	*4					C				
PA166128232	*5				G					
PA166128233	*6						T			
PA166128234	*7								insA	
PA166128235	*8		A							
PA166128236	*9			Y				T		

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CYP3A5

- Why is this complicated?

Haplotype Set Id	PA166128212	7/22/15	R28C			Q200R		A337T	T346fs	T398N
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PA166128213	*1		G	T	A	T	C	C	del	G
PA166128218	*2									T
PA166128219	*3			C						
PA166128231	*4					C				
PA166128232	*5				G					
PA166128233	*6						T			
PA166128234	*7								insA	
PA166128235	*8		A							
PA166128236	*9			Y				T		

Poor metabolizer (CYP3A5 non-expresser)	An individual carrying two non-functional alleles	*3/*3, *6/*6, *7/*7, *3/*6, *3/*7, *6/*7	Higher ("normal") dose-adjusted trough concentrations of tacrolimus and increased chance of achieving target tacrolimus concentrations	Initiate therapy with standard recommended dose. Use therapeutic drug monitoring to guide dose adjustments	Strong

[a] Typically with other CYP enzymes, an extensive metabolizer would be classified as a "normal" metabolizer, and therefore, the drug dose would not change based on the patient's genotype. However, in the case of CYP3A5 and tacrolimus, a CYP3A5 expresser (i.e. CYP3A5 extensive metabolizer or intermediate metabolizer) would require a higher recommended starting dose and the CYP3A5 non-expresser (i.e. poor metabolizer) would require the standard recommended starting dose.

[b] Additional rare variants such as *CYP3A5*2, *8*, and **9* may be found which are of unknown functional significance. However, if a copy of **1* is present, expected phenotype would be intermediate metabolizer.

CYP3A5

Likely phenotype [a]	Genotypes	Examples of diplotypes [b]	Implications for tacrolimus pharmacologic measures	Therapeutic Recommendations [c]	Classification of recommendations [e]
Extensive metabolizer (CYP3A5 expresser)	An individual carrying two functional alleles	*1/*1	Lower dose-adjusted trough concentrations of tacrolimus and decreased chance of achieving target tacrolimus concentrations	Increase starting dose 1.5 to 2 times recommended starting dose [d]. Total starting dose should not exceed 0.3mg/kg/day. Use therapeutic drug monitoring to guide dose adjustments	Strong
Intermediate metabolizer (CYP3A5 expresser)	An individual carrying one functional allele and one non-functional allele	*1/*3, *1/*6, *1/*7	Lower dose-adjusted trough concentrations of tacrolimus and decreased chance of achieving target tacrolimus concentrations	Increase starting dose 1.5 to 2 times recommended starting dose [d]. Total starting dose should not exceed 0.3mg/kg/day. Use therapeutic drug monitoring to guide dose adjustments	Strong
Poor metabolizer (CYP3A5 non-expresser)	An individual carrying two non-functional alleles	*3/*3, *6/*6, *7/*7, *3/*6, *3/*7, *6/*7	Higher ("normal") dose-adjusted trough concentrations of tacrolimus and increased chance of achieving target tacrolimus concentrations	Initiate therapy with standard recommended dose. Use therapeutic drug monitoring to guide dose adjustments	Strong

[a] Typically with other CYP enzymes, an extensive metabolizer would be classified as a "normal" metabolizer, and therefore, the drug dose would not change based on the patient's genotype. However, in the case of CYP3A5 and tacrolimus, a CYP3A5 expresser (i.e. CYP3A5 extensive metabolizer or intermediate metabolizer) would require a higher recommended starting dose and the CYP3A5 non-expresser (i.e. poor metabolizer) would require the standard recommended starting dose.

[b] Additional rare variants such as *CYP3A5*2, *8*, and **9* may be found which are of unknown functional significance. However, if a copy of **1* is present, expected phenotype would be intermediate metabolizer.

CPIC Haplotype Table – *CYP2C19* example

GENE: CYP2C19	5/27/16															
	Nucleotide change to gene from http://www.cypalleles.ki.se/cyp2c19.htm	-2030C>T	-2020C>A	-1439T>C	-1041G>A	-806C>T	-13G>A	1A>G	7C>T	10T>C	50T>C	55A>C	83A>T	151A>G	12401C>T	12416C>T
	Effect on protein (NP_000760.1)	5' region	5' region	5' region	5' region	5' region	5' region	M1V	P3S	F4L	L17P	I19L	K28I	S51G	R73C	H78Y
	Position at NC_000010.11 (Homo sapiens chromosome 10, GRCh38.p2)	g.94760676C>T	g.94760686C>A	g.94761267T>C	g.94761665G>A	g.94761900C>T	g.94762693G>A	g.94762706A>G	g.94762712C>T	g.94762715T>C	g.94762755T>C	g.94762760A>C	g.94762788A>T	g.94762856A>G	g.94775106C>T	g.9477512 1C T
	Position at NG_008384.2 (CYP2C19 RefSeqGene; forward relative to chromosome)	g.2971C>T	g.2981C>A	g.3562T>C	g.3960G>A	g.4195C>T	g.4988G>A	g.5001A>G	g.5007C>T	g.5010T>C	g.5050T>C	g.5055A>C	g.5083A>T	g.5151A>G	g.17401C>T	g.17416C>T
	rsID	rs113164681	rs111490789	rs17878739	rs7902257	rs12248560	rs367543001	rs28399504	rs367543002	rs367543003	rs55752064	rs17882687			rs145328984	
CYP2C19 Allele	Allele Functional Status															
*1	Normal function	C	C	T	G	C	G	A	C	T	T	A	A	A	C	C
*2	No function															
*3	No function															
*4A	No function							G								
*4B	No function					T		G								
*5	No function															
*6	No function															
*7	No function															
*8	No function															
*9	Decreased function															
*10	Decreased function															
*11	Normal function															
*12	Unknown function															
*13	Normal function															
*14	Unknown function										C					
*15	Normal function											C				
*16	Decreased function															
*17	Increased function					T										
*18	Normal function															
*19	Decreased function													G		
*22	No function															
*23	Unknown function															

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CPIC Guideline – *CYP2C19* example

CPIC Guideline for sertraline and CYP2C19

The CPIC Dosing Guideline for the selective serotonin reuptake inhibitor sertraline recommends to consider a 50% reduction of recommended starting dose and titrate to response or select alternative drug not predominantly metabolized by CYP2C19 for CYP2C19 poor metabolizers.

Annotations for CYP2C19:*1/*2

Type	Annotation
Implications	Reduced metabolism when compared to extensive metabolizers.
Metabolizer Status	Intermediate metabolizer (~18-45% of patients)
Phenotype (Genotype)	An individual carrying one normal function allele or one increased function allele and one no function allele. *The predicted metabolizer phenotype for the*2/*17 genotypes is a provisional classification. The currently available evidence indicates that the CYP2C19*17 increased function allele is unable to completely compensate for the no function CYP2C19*2.*
Recommendations	Initiate therapy with recommended starting dose.
Classification of Recommendation	Strong

For full guideline see https://cpicpgx.org

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Diplotype Caller – *CYP2C19* example

CYP2C19

- *1/*2 (40)

Definition Position	94760676	94760686	94761267	94761665	94761900	94762693	94762706	94762712	94762715	94762755	94762760	94762788	94762856	94775106	94775121	94775160	94775185	94775367	94775416	94775453	94775489	94775
	rs113164681	rs111490789	rs17878739	rs7902257	rs12248560	rs367543001	rs28399504	rs367543002	rs367543003	rs55752064	rs17882687			rs145328984		rs118203756		rs12769205	rs41291556	rs72552267	rs17884712	rs5897
VCF Position	94760676	94760686	94761267	94761665	94761900	94762693	94762706	94762712	94762715	94762755	94762760	94762788	94762856	94775106	94775121	94775160	94775185	94775367	94775416	94775453	94775489	94775
VCF REF,ALTs	C	C	T	G	C	G	A	C	T	T	A	A	A	C	C	G	A	A	T	G	G	G
VCF Call	C|C	C|C	T|T	G|G	C|C	G|G	A|A	C|C	T|T	T|T	A|A	A|A	A|A	C|C	C|C	G|G	A|A	A|A	T|T	G|G	G|G	G|
*1	C	C	T	G	C	G	A	C	T	T	A	A	A	C	C	G	A	A	T	G	G	G
	C	C	T	G	C	G	A	C	T	T	A	A	A	C	C	G	A	A	T	G	G	G
*2	C	C	T	G	C	G	A	C	T	T	A	A	A	C	C	G	A	[AG]	T	G	G	G
	C	C	T	G	C	G	A	C	T	T	A	A	A	C	C	G	A	A	T	G	G	G

- PharmCAT takes the .vcf and the CPIC tables into the Diplotype Caller
- Combines with the CPIC guidelines to generate reports
  - Intermediate
  - Final

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Intermediate report – *CYP2C19* example

Gene: CYP2C19

Matching Allele Call

All variant positions present so all haplotypes considered in analysis.

Diplotype call: CYP2C19:*1/*2

Warnings (none)

Calls at Positions

Position	RSID	Call
94760676	rs113164681	C|C
94760686	rs111490789	C|C
94761267	rs17878739	T|T
94761665	rs7902257	G|G
94761900	rs12248560	C|C
94762693	rs367543001	G|G
94762706	rs28399504	A|A
94762712	rs367543002	C|C
94762715	rs367543003	T|T
94762755	rs55752064	T|T
94762760	rs17882687	A|A
94762788	None	A|A
94762856	None	A|A
94775106	rs145328984	C|C
94775121	None	C|C
94775160	rs118203756	G|G
94775185	None	A|A
94775367	rs12769205	A|A
94775416	rs41291556	T|T
94775453	rs72552267	G|G
94775489	rs17884712	G|G
94775507	rs58973490	G|G
94780574	rs140278421	G|G
94780579	rs370803989	G|G
94780653	rs4986893	G|G
94781858	rs6413438	C|C

- Generates genotype calls at every relevant position
- Includes missing data calls/no calls

PharmCAT report example

PharmCAT Report [test.cftr.reg_inc]

Sections

Diplotype / Genotype Summary

Genotypes called: 12 / 12

Drugs [a]	Gene	Diplotype or Genotype	Allele Functionality [b]	Phenotype [b]	Uncallable Alleles [c]
ivacaftor	CFTR	F508del(TCT)/G542X	N/A	N/A	yes
amitriptyline **escitalopram** **citalopram** **clomipramine** **clopidogrel** **doxepin** **imipramine** **sertraline** **trimipramine** **voriconazole**	CYP2C19	*2/*2	Two no function alleles	Poor Metabolizer	no
phenytoin **warfarin**	CYP2C9	*2/*3	Two decreased function alleles	Poor Metabolizer	no
amitriptyline **clomipramine** **codeine** **desipramine** **doxepin** **fluvoxamine** **imipramine** **nortriptyline** ondansetron **paroxetine** **trimipramine** tropisetron	CYP2D6†	*3/*4	Two no function alleles	Poor Metabolizer	no

PharmCAT Report [test.cftr.reg_inc]

Sections

Diplotype / Genotype Summary

Genotypes called: 12 / 12

Drugs [a]	Gene	Diplotype or Genotype	Allele Functionality [b]	Phenotype [b]	Uncallable Alleles [c]
tacrolimus	CYP3A5†	*1/*7	One normal function allele and one no function allele	Intermediate Metabolizer	no
warfarin	CYP4F2	*1/*1	Two normal function alleles	N/A	no
capecitabine fluorouracil tegafur	DPYD	*1/*1	Two normal function alleles	Normal Metabolizer	yes
peginterferon alfa-2a peginterferon alfa-2b ribavirin	IFNL3†	rs12979860C/rs12979860C	N/A	N/A	no
simvastatin	SLCO1B1†	rs4149056CC	Two decreased function alleles	Poor Function	no
azathioprine mercaptopurine thioguanine	TPMT†	*1/*1	Two normal function alleles	Normal Metabolizer	no
atazanavir	UGT1A1†	*1/*1	Two normal function alleles	Normal Metabolizer	no
warfarin	VKORC1†	-1639A/-1639A	N/A	N/A	no

[a] *The drugs highlighted in red indicate a CPIC recommendation prescribing change based on the person's listed diplotype/genotype (highlighting is not based on CPIC strength of recommendation). See CPIC recommendation section for the classification of the recommendation and further details. Please note, warfarin and peginterferon alpha/ribavirin are highlighted in blue, see CPIC recommendation section for specific prescribing information. The drug is highlighted when multiple diplotypes are presented if any is assocated with a prescribing change. Please see recommendation section for detailed information.*

[b] *Allele functionality and phenotype terms are based on the CPIC term standardization project, PMID:27441996. Guidelines published prior use the term 'extensive' instead of 'normal' metabolizer. CYP2C19*1/*17 is now classified as rapid metabolizer. Guidelines published prior grouped CYP2C19*1/*17 together with *17/*17 as ultrarapid metabolizer.*

[c] *Indicates alleles not considered for the diplotype calls due to missing variant information, please see Allele calls section. Alleles that could not be considered due to missing input might change the metabolizer phenotype and possible CPIC recommendation.*

† *Check the allele call details for this gene for more details about this call.*

For a full list of disclaimers and limitations see the Disclaimer section.

THE PharmGKB BLOG

FRIDAY, APRIL 15, 2016

Pharmacogenomics Clinical Annotation Tool (PharmCAT)

An active area of genomic medicine implementation at many health care organizations and academic medical centers includes development of decision support and return of results around pharmacogenomics. The Clinical Pharmacogenetics Implementation Consortium (CPIC) has established guidelines surrounding gene-drug pairs that can and should lead to treatment modifications based on genetic variants. One of the challenges in implementing pharmacogenomics is the representation of the information in the CPIC guidelines (including star-alleles) and extracting these variants and haplotypes from genetic datasets. In a collaboration between the PGRN Statistical Analysis Resource (P-STAR), the Pharmacogenomics Knowledgebase (PharmGKB), the Clinical Genome Resource (ClinGen), and CPIC, we are developing a software tool to extract all CPIC level-A variants from a genetic dataset (represented as a vcf), interpret the variant alleles, and generate a report. The CPIC pipeline report can then be used to make future treatment decisions.

Summary

- PharmCAT is a Pharmacogenomics Clinical Annotation Tool
- Developed to automate the .vcf → haplotype → CPIC guideline process
- PharmCAT version 1.0 is in testing
  - Goal to release very soon for community feedback
- PharmCAT reports can then be adapted for local implementation into EHR or patient/provider reports

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PharmGKB / PharmCAT

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Code Issues 6 Pull requests 0 Wiki Pulse Graphs

The Pharmacogenomic Clinical Annotation Tool - take genome data, make haplotype calls, get CPIC annotations.

341 commits **1** branch **1** release **6** contributors

Branch: **master** New pull request Find file Clone or download

markwoon Add corner case for not initialized check. Latest commit `4e81c15` 4 days ago

.idea	Update for MarkdownNavigator.	25 days ago
doc	Template TSV update (key-value headers, gene orientation, anchored in…	2 months ago
gradle/wrapper	Use gradle with source.	a month ago
lib	removed jars and moved to gradle	2 months ago

README.md

PharmCAT

Take genome data, make haplotype calls, get CPIC annotations.

Status

Please consider this alpha code.

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- CPIC participants
- P-STAR participants

PharmCAT team: T.E. Klein, M. Whirl-Carrillo, R.M. Whaley, M. Woon, K. Sangkuhl, L.G. Carter, H.M. Dunnenberger, P.E. Empey, A.T. Frase, R.R. Freimuth, A. Gaedigk, A. Gordon, C. Haidar, J.K. Hicks, J.M. Hoffman, M.T. Lee, N. Miller, S.D. Mooney, T.N. Person, J.F. Peterson, M.V. Relling, S.A. Scott, G. Twist, A. Verma, M.S. Williams, C. Wu, W. Yang, M.D. Ritchie

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