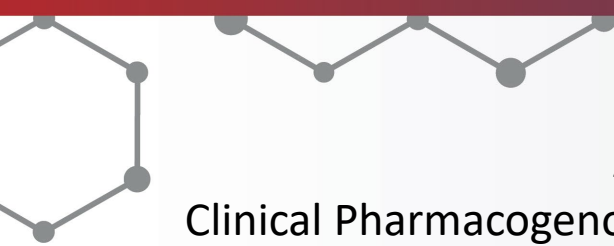




IMPLEMENTATION OF CLINICAL PHARMACOGENOMICS ACROSS A MULTI-STATE HEALTH SYSTEM



Natasha Petry, PharmD, MPH, BCACP

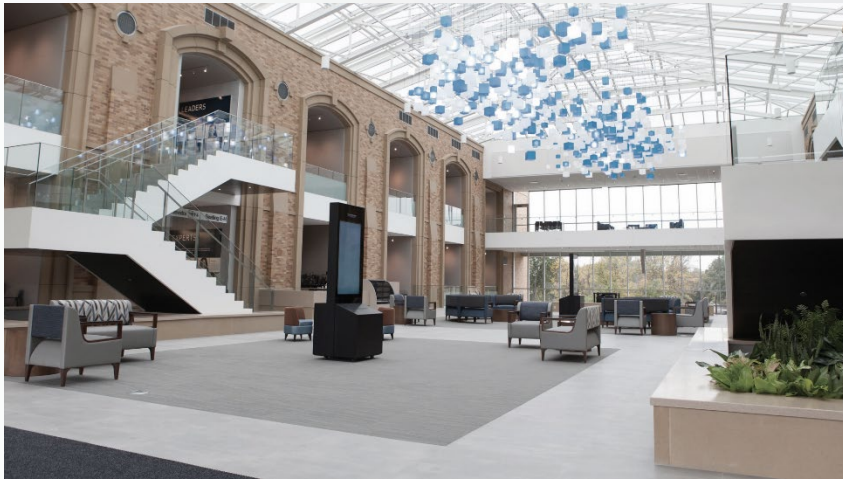
Associate Professor of Practice, North Dakota State University

Clinical Pharmacogenomics Pharmacist and Clinical PGx PGY2 Residency Program Director, Sanford Health



OBJECTIVE

Describe methods used to implement pharmacogenomics into a large rural healthcare system.



Sanford Health

Sanford Health is one of the largest rural health systems in the United States and is dedicated to:

- integrated delivery of health care
- **genomic medicine**
- senior care and services
- global clinics
- research
- affordable insurance

The logo for Sanford Health, featuring the word "SANFORD" in a dark blue serif font with a stylized "S" and "D" that incorporate a circular element, and the word "HEALTH" in a smaller, light blue sans-serif font below it.

SANFORD[®]
HEALTH

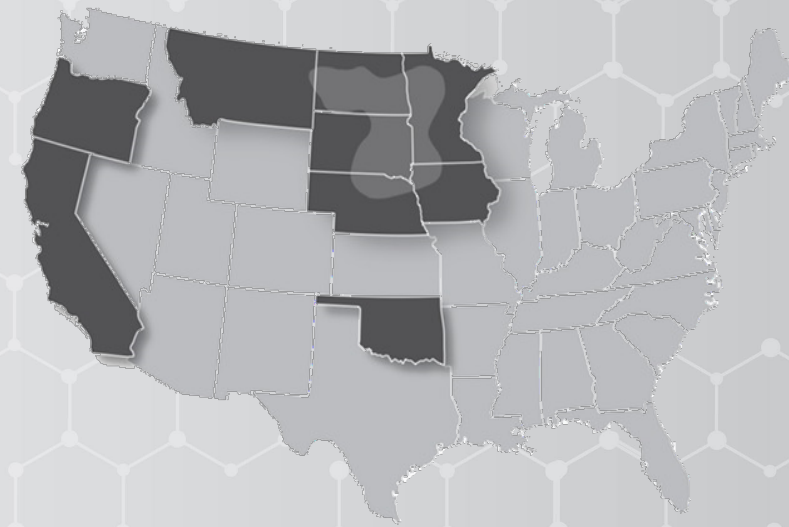
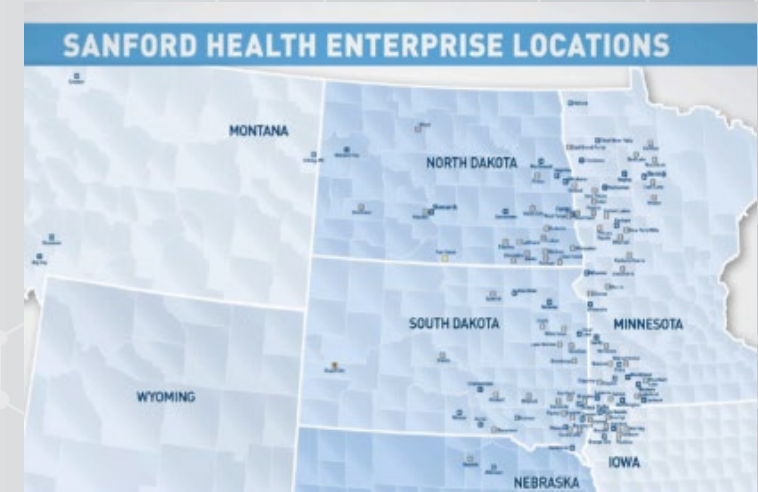
Sanford Health

Predominantly the upper Midwest

- Spans across 9 states

Geographical footprint of over 250,000 square miles

- >2 million patients
- 44 medical centers and 482 clinics
- >48,000 employees and 1,350 physicians



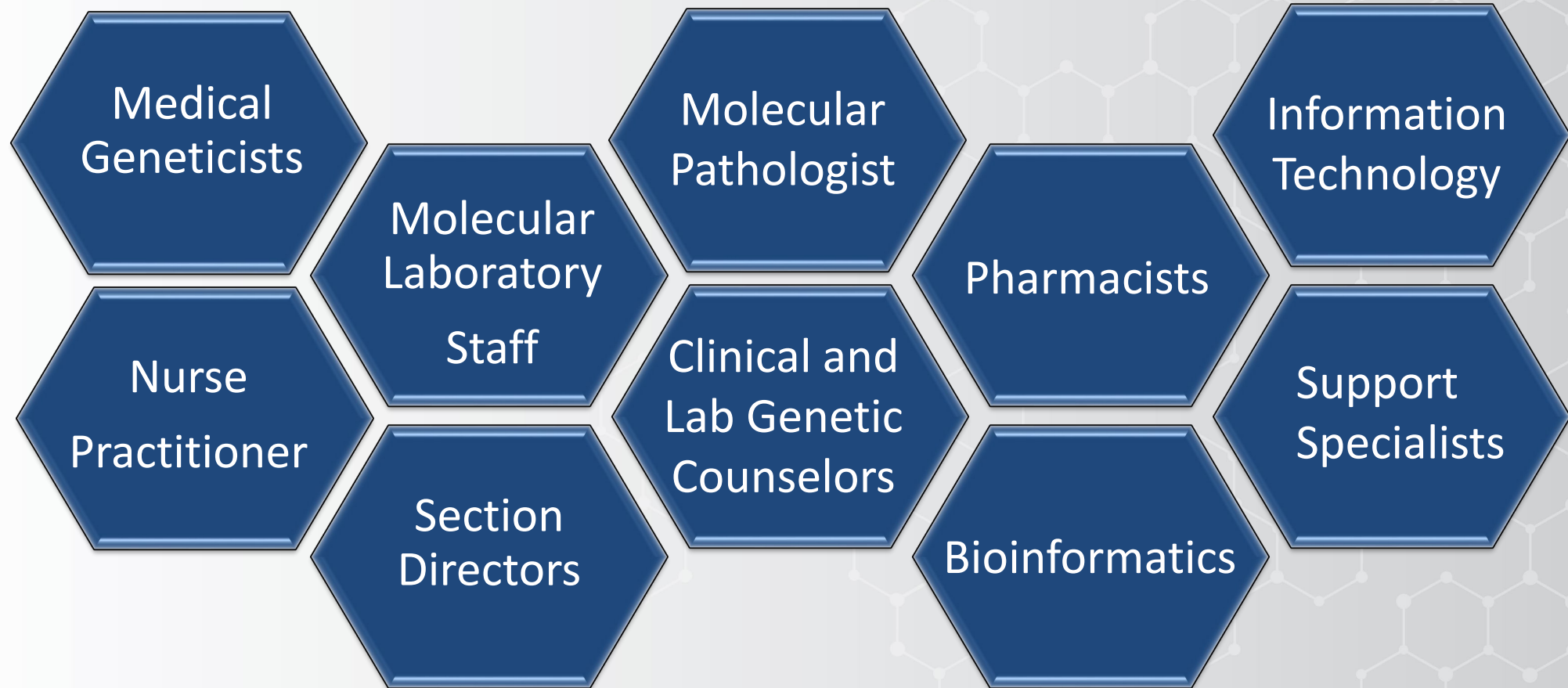
History of Imagenetics

Established in 2014 after a \$125 million gift from philanthropist Denny Sanford

Internal Medicine and Genetics = Imagenetics



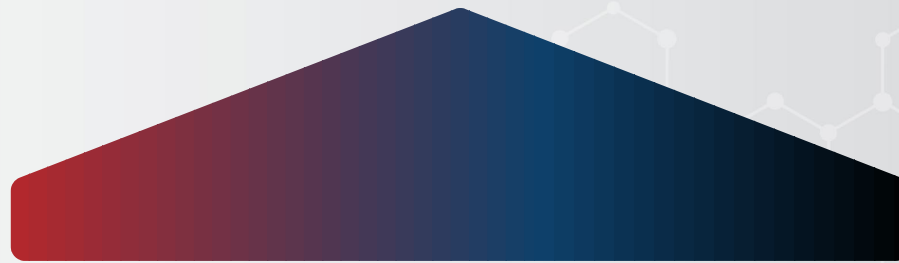
Multi-disciplinary Team



PGX PHARMACIST TIMELINE

- 2014
 - Natasha Petry is still employed by Internal Medicine department– starts working with Dr. Russ Wilke
- 2017/2018
 - **April Schultz** hired as manager (later promoted to Director of Operations and PGx services) – 1.0 FTE
 - **Jordan Baye** hired as 0.5 FTE South Dakota State University Faculty and 0.5 FTE Imagenetics
 - **Natasha Petry** hired as 0.5 FTE North Dakota State University Faculty and 0.5 FTE Imagenetics
- 2019
 - **Amanda Massmann** and **Joel Van Heukelom** hired each at 1.0 FTE (Amanda later promoted to clinical lead and Joel to supervisor)
- 2020
 - **Kristen Jacobsen** hired as Clinical PGx pharmacist (1.0 FTE)

Pillars of Sanford Imagenetics





Clinical Care

PGx Initiatives



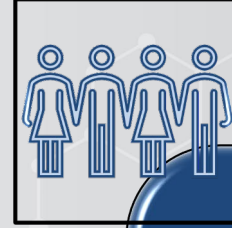
Single Gene Testing

- *CYP2C19* – cath lab



PGx Panel Initiatives

- Psychiatry
- Sanford Employee Health Plan
- Transplant
- Veterans
- Medicare Align



Population Screening

- Sanford Chip
- Specific requirements
- PGx panel
- Medically Actionable Predisposition

In House Medical Genetics Lab

CAP Accredited

Digital Droplet PCR
(ddPCR) for *CYP2D6* copy
number assessment

Ease of
ordering
PGx testing

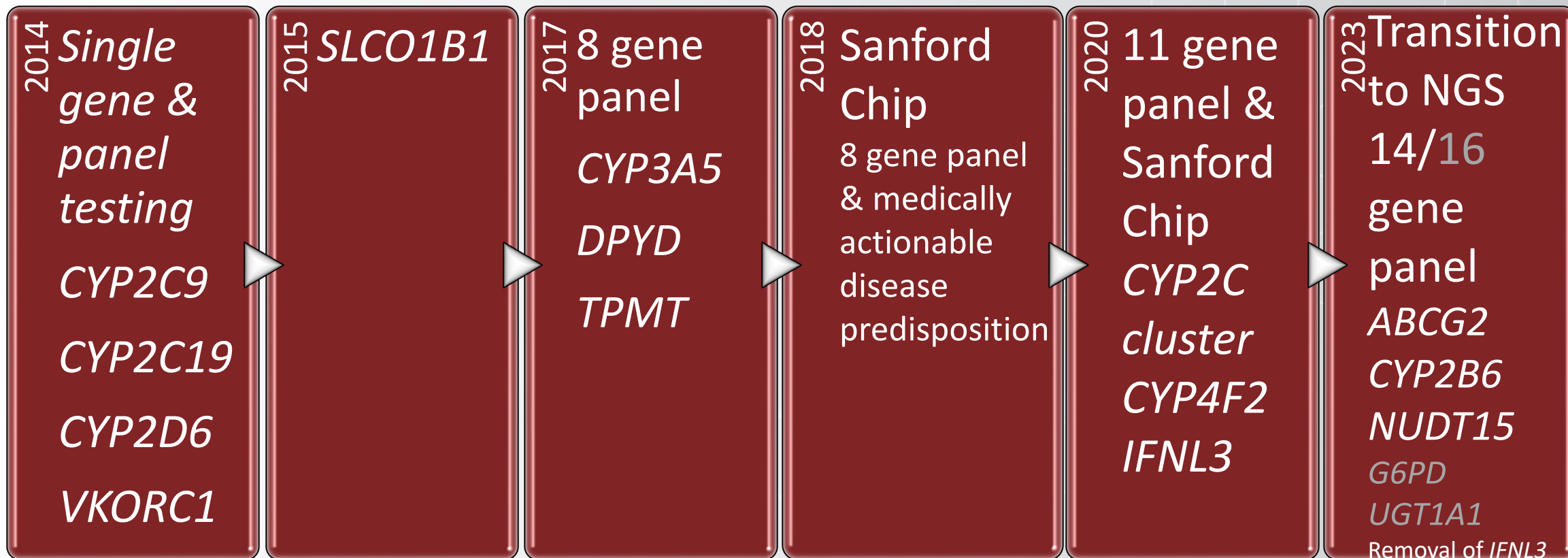
Translating
Research to
Clinical
Screening and
Diagnostics

Expanding
Diagnostic
Testing
Capabilities

Support
research
activities in a
CLIA/ CAP
environment



Evolution of PGx testing



Patient name: 3_2, Rows
DOB: 04/01/1990



MRN: Row3_2
Sex: female
Specimen ID: Row3_2
Specimen Type: blood
Test Name: COMPREHENSIVE PHARMACOGENETICS PANEL
Ordering Provider: Shubhi
Date Collected: 11/01/2023
Date Resulted: 11/21/2023

Patients should not stop their medication(s) or make any changes to their medication(s) without consulting with their provider first.

DRUG-GENE ASSOCIATION

MEDICATION CATEGORY	BASED ON PHARMACOGENETICS STANDARD DOSING PER YOUR PROVIDER	BASED ON PHARMACOGENETICS PROVIDER MAY DISCUSS ALTERNATIVES
Anti-infective	Abacavir (Ziagen) Atazanavir (Reyataz) Efavirenz (Sustiva; Atripla) Voriconazole (Vfend)	Dapsone (Aczone) Nitrofurantoin (Macrobid; Macroductin) Primaquine (Primaquine) Tafenoquine (Aarakoda; Krintafel)
Anticoagulant	Warfarin (Coumadin; Jantoven)	
Antiplatelet		Clopidogrel (Plavix)
Cholesterol		Atorvastatin (Lipitor) Fluvastatin (Lescol XL) Lovastatin (Mevacor) Pitavastatin (Livalo) Pravastatin (Pravachol) Rosuvastatin (Crestor) Simvastatin (Zocor)
Gastrointestinal	Metoclopramide (Reglan) Ondansetron (Zofran)	Dexlansoprazole (Dexilant) Lansoprazole (Prevacid) Omeprazole (Prilosec) Pantoprazole (Protonix)

Electronically signed by: Massmann, Amanda
Medical Laboratory Director: Rachel Starks, MD, PhD
Sanford Medical Genetics Laboratory 1321 W 22nd Street, Sioux Falls, SD, 57105 (605) 312-GENE (4363)
Date signed: 11/21/2023
CLIA number: 43D0658889

Patient name: 3_2, Rows
DOB: 04/01/1990



PHARMACOGENETIC RESULTS

GENE	RESULTS	PHENOTYPE
CYP2B6	*1/*22	Rapid Metabolizer
CYP2C19	*1/*9	Likely Intermediate Metabolizer
CYP2C9	*1/*8 Activity Score: 1.5	Intermediate Metabolizer
CYP2D6	*10/*10 Activity Score: 0.5	Intermediate Metabolizer
CYP3A5	*1/*1	Normal Metabolizer
G6PD	B/Canton	Decreased or Normal Function
HLA-B *57:01 screen	See Comment	Unknown Phenotype
NUDT15	*3/*6	Poor Metabolizer
SLCO1B1	*1/*5	Decreased Function
TPMT	*1/*3A	Intermediate Metabolizer
UGT1A1	See Comment	Unknown Phenotype

GENE	VARIANTS	ZYGOSITY	PHENOTYPE
ABCG2 (NM_004827.3)	c.421C>A (p.Q141K)	Homozygous	Poor Function
CYP2C cluster (NC_000010.10)	No Variants Detected	Homozygous	Low Sensitivity
CYP4F2 (NC_000019.9)	No Variants Detected	Homozygous	Normal Function
DPYD (NM_000110.4)	No Variants Detected Activity Score: 2	Homozygous	Normal Metabolizer
VKORC1 (NM_024006.5)	No Variants Detected	Homozygous	Low Warfarin Sensitivity

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Regions excluded due to inadequate sequencing coverage rs2395029

The panel includes the following targets:

ABC2	CYP2D6	G6PD
rs2231142 G>T	rs1135840 G>C rs72549346 CAC>CACAC rs72549347 G>A	rs72554664 C>T rs72554665 C>G,A rs5030069 C>T
CYP2B6	rs59421388 C>T rs28371725 C>T rs79292917 C>T rs5030867 T>G rs16947 A>G	rs76723693 A>G rs137852330 G>A rs5030868 G>A rs267606836 G>A rs1050829 T>C rs267606835 G>C rs1050828 C>T
CYP2C Cluster	rs5030656 CTCT>CT rs72549352 G(7)>G(8) rs35742686 T>C rs3892097 C>T rs5030865 C>T, A rs5030655 A>-- rs61736512 C>T rs1135822 A>T rs28371706 G>A, T rs774671100 A>AA rs5030862 C>T rs1065852 G>A	rs2395029 T>G
CYP2C19	rs4244285 G>A rs4986893 G>A rs28399504 A>G rs12248560 C>T rs56337013 C>T rs72552267 G>A rs72558186 T>C rs41291556 T>C rs17884712 G>A rs6413438 C>T rs12769205 A>G rs375781227 G>A rs118203759 C>G rs118203757 G>A rs140278421 G>C rs192154563 C>T	rs746071566 GAGTCG(3)>GAGTCG(2),GAGTCG(4) rs116855232 C>T rs147390019 G>A rs1186364061 G>A rs766023281 G>C [GRCh37/hg19] chr13:48611985 A>G
CYP3A5	rs41303343 A>AA rs10264272 C>T rs776746 C>T	rs2306283 A>G rs11045819 C>A rs4149056 T>C
CYP4F2	rs2108622 C>T	rs1142345 T>C,G rs1800584 C>T rs74423290 G>C rs9533570 C>T rs1800460 C>T rs72552730 C>T rs1800462 C>G rs267607275 A>G rs9333569 T>C
CYP2C9	rs1799853 C>T rs1057910 A>C rs28371686 C>G rs9332131 AA>A rs7900194 G>A rs28371685 C>T rs9332239 C>T rs72558187 T>C rs72558190 C>A rs56165452 T>C rs72558192 A>G	rs1801268 C>A rs67376798 T>A rs3910290 C>T rs72549305 GG>G rs55886062 A>C rs56038477 C>T rs78060119 C>A rs75017182 G>C rs1801266 G>A rs115232898 T>C rs72549309 ATGA[2]>ATGA
UGT1A1	rs3064744 TA[7]>TA(6),TA(8),TA(9) rs4148323 G>A rs35350960 C>A	rs9923231 C>T
VKORC1		

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PGx Clinical Service

PGx Clinic (pre and post visits)

Clinical Decision Support

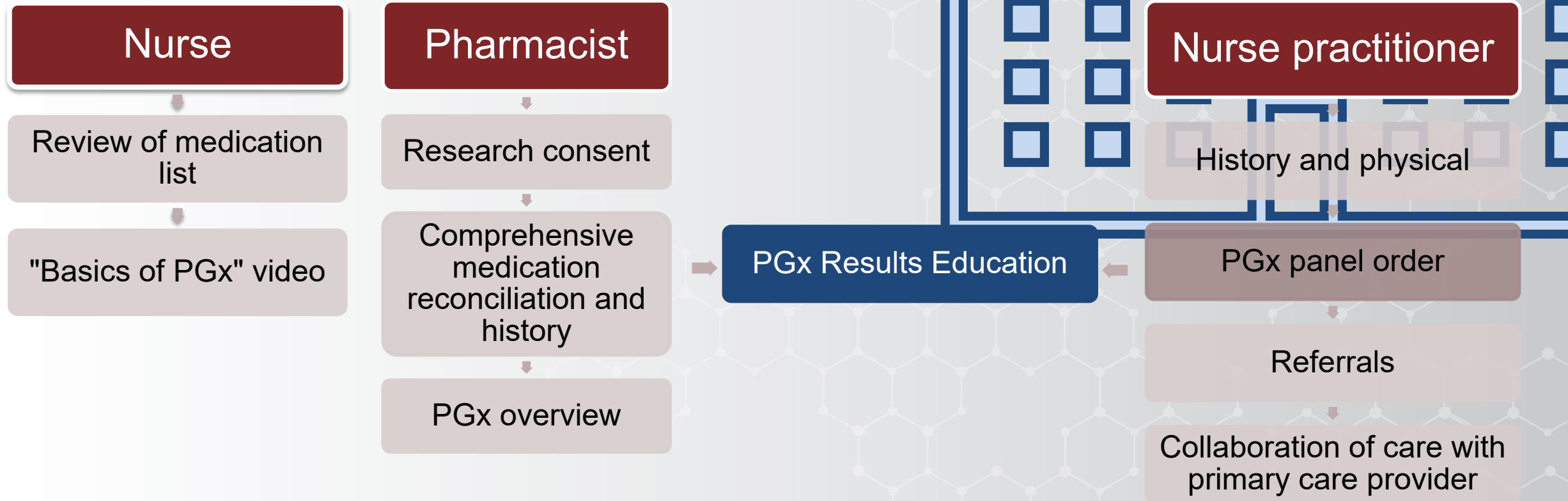
Patient Portal Result Messages

Clinical Note within EMR

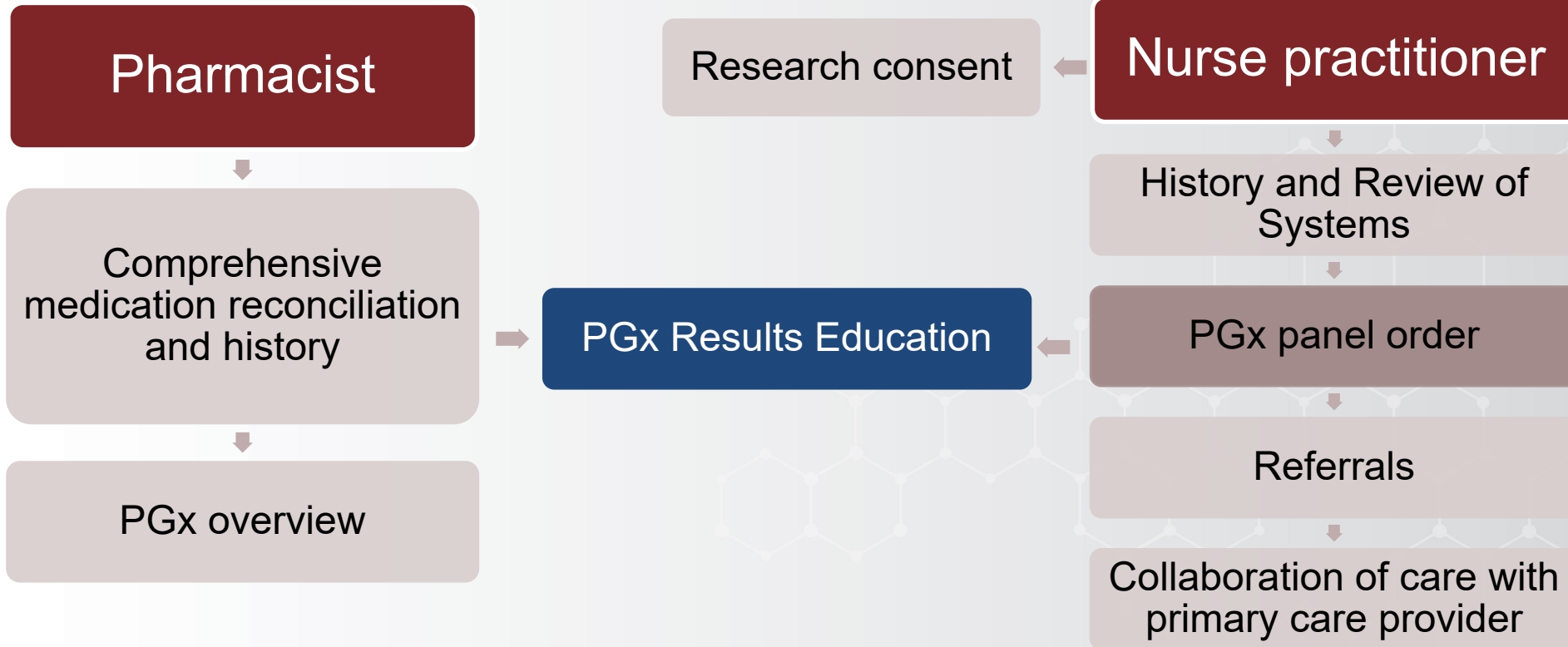
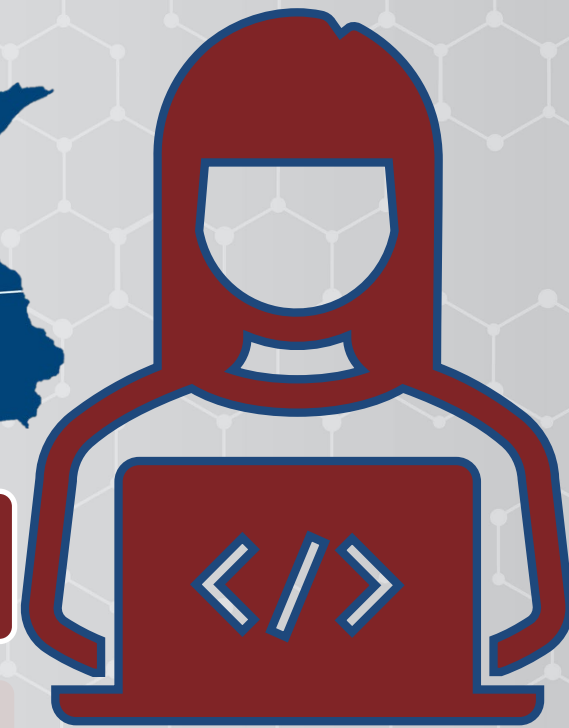
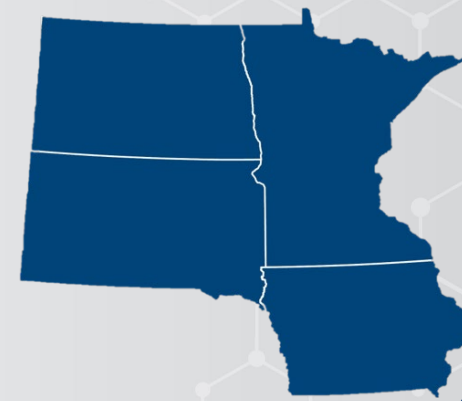
Actionable Notes Routed to Providers

All PGx Results Reviewed

PGx Clinic Model



PGx Virtual Clinic Model



Discrete Data

Screenshot
before transition
to updated panel

- Data is stored in a particular manner that can be easily retrieved and turned into relevant information
- Laboratory components may include:
 - Genotype/diplotype
 - Phenotype
 - Activity score

GENETICS	
MISCELLANEOUS GENETIC ...	
PHARMGX PANEL (11 GENE)	
CYP2C Cluster Phenotype	High Sensit...
CYP2C Cluster Genotype	g.96405502...
CYP2C19 Phenotype	Rapid Meta...
CYP2C19 Genotype	*1/*17
CYP2D6 Phenotype	Intermediat...
CYP2D6 Genotype	*2/*3
CYP2D6 Activity Score	Activity Sco...
CYP2C9 Phenotype	Normal Met...
CYP2C9 Genotype	*1/*1
CYP2C9 Activity Score	Activity Sco...
VKORC1 Phenotype	High Warfa...
VKORC1 Genotype	-1639G>A ...
CYP3A5 Phenotype	Poor Metab...
CYP3A5 Genotype	*3/*3
CYP4F2 Phenotype	Normal Acti...
CYP4F2 Genotype	c.1297G>A ...
DPYD Phenotype	Normal Met...
DPYD Activity Score	Activity Sco...
IFNL3 Phenotype	Favorable ...
IFNL3 Genotype	rs12979860...
SLCO1B1 Phenotype	Normal Fun...
SLCO1B1 Genotype	*1/*1
TPMT Phenotype	Normal Met...
TPMT Genotype	*1/*1

PGx Result Notification

The screenshot displays the Epic BestPractice interface. At the top, the browser title is "TST - Classic Hyperspace - Sanford Health TST - 29 BestPractice". The navigation bar includes "Epic", "Dragon Login", "Receiving", "Outstanding List", "Result Entry and Verification", "Specimen Inquiry (By Patient)", "Specimen Update", "Addendum", "Print", and "Log Out". The user is identified as "SANFORD HEALTH TST AMANDA M. Beaker".

The "In Basket" section shows a list of message categories on the left: "My Messages", "My Open Encounters 4/4", "My Open Charts 5/7", "BestPractice 29/35" (highlighted), "My Unsigned Orders 2/2", "Patient Notifications 1/1", "Attached & Covering... 0/0", "Follow-up", "Search", and "Sent Messages".

The main content area shows a table of messages under the "BestPractice" category, with 29 new and 35 total messages. The table has columns for "Msg Date", "Status", "Subject", "Patient", "Comment", "Expiration", "Visit...", and "Room".

Msg Date	Status	Subject	Patient	Comment	Expiration	Visit...	Room
03/03/2023 08:40 AM	New	MAP Positive Varian...	Imagenetics, Ch...		07/01/2023 0...	02/10...	LABSCMOLGEN
Department: SANFORD CLINIC MOLECULAR GENETICS RECEIVING LABORATORY							
03/03/2023 12:55 PM	New	CYP2C19 Result Re...	Cupid, Gene		07/01/2023 0...	03/02...	LABSCMOLGEN
Department: SANFORD CLINIC MOLECULAR GENETICS LABORATORY							
03/03/2023 12:56 PM	New	Actionable Result	Cupid, Gene		07/01/2023 0...	03/02...	LABSCMOLGEN
Department: SANFORD CLINIC MOLECULAR GENETICS LABORATORY							
03/03/2023 12:58 PM	New	PharmGx (11 GENE)...	Imagenetics, Army		07/01/2023 0...	11/14...	LABSCIMAGEN
Department: SANFORD CLINIC IMAGENETICS LABORATORY							

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Genomic Indicators

Genomic Indicators

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Add a new indicator [+ Add](#)

On

Disease



Monoallelic mutation of KCNQ1 gene [Edit](#)

Shared:

Mutations, or disease-causing variants, in KCNQ1 may increase the risk for arrhythmia. Variants in KCNQ1 have been associated with Long QT Syndrome (L...
[Long QT Syndrome](#)

Drug



Amitriptyline-Altered Enzyme Function [Edit](#)

[View Results](#) Shared:

Based on genetic results, amitriptyline may not be recommended due to potential for toxicity and lack of efficacy. The patient is predicted to be a CYP2D6 inter...
[Hicks, et al. Guideline \(CPIC\) for CYP2D6 and CYP2C19 Genotypes and Dosing of Tricyclic Antidepressants. July 2017.](#)



Atorvastatin-Poor Enzyme Function [Edit](#)

[View Results](#) Shared:

Based on genetic results, atorvastatin doses greater than 20 mg are not recommended due to increased risk of myopathy and rhabdomyolysis. May consider c...
[Cooper-DeHoff, et al. The Clinical Pharmacogenetics Implementation Consortium Guideline for SLCO1B1, ABCG2, and CYP2C9 and statin-associated musculoskeletal symptoms. 2022](#)



Azathioprine-Decreased Enzyme Function [Edit](#)

[View Results](#) Shared:

Based on genetic results, patient is at increased risk of myelosuppression with azathioprine therapy. This phenotype displays a 1:100,000 chance for severe T...
[Relling, et al. Guideline for Thiopurine Dosing Based on TPMT and NUDT15 Genotypes. May 2019.](#)



Capecitabine-Decreased Enzyme Function [Edit](#)

[View Results](#) Shared:

Based on genetic results, capecitabine is not recommended due to serious adverse events including death. Patient is VERY high risk of myelotoxicity, neuroto...
[Amstutz, et al. Clinical Pharmacogenetics Implementation Consortium \(CPIC\) Guideline for Dihydropyrimidine Dehydrogenase Genotype and Fluoropyrimidine Dosing. 2017 Update.](#)



Citalopram-Decreased Enzyme Function [Edit](#)

[View Results](#) Shared:

Based on genetic results, citalopram is not recommended due to increased risk of toxicity. Consider a 50% reduction in starting dose for escitalopram, citalopra...
[Hicks, et al. Guideline for CYP2D6 and CYP2C19 Genotypes and Dosing of Selective Serotonin Reuptake Inhibitors](#)

Patient Portal

Patient Message

To: Chamanda Imagenetics

Regarding: Chamanda Imagenetics

Pharmacogenomics Results Review

Dear Chamanda Imagenetics,

A pharmacist has reviewed your pharmacogenomics (PGx) test results. If the PGx results impact any of your medications, your doctor will be notified. Your genes are one of many factors affecting how medications work, and your provider may or may not decide to change your medications at this time.

Your PGx results will remain part of your medical record. They may be used to help guide your doctor in choosing the best medication and dose for you in the future.

Follow the steps below to access your Genetic Profile:

1. Log in to your My Sanford Chart account
2. Select the Menu icon
3. Find the My Record section
4. Click Genetic Profile

Save as QuickAction | © 2023 Epic Systems Corporation

Conversation List

Pharmacogenomics Results Review

Participants: Amanda

A pharmacist has reviewed your pharmacogenomics (PGx) test results. If the PGx results impact any of your medications, your doctor will be notified. Your genes are one of many factors affecting how medications work, and your provider may or may not decide to change your medications at this time.

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Follow the steps below to access your Genetic Profile:

1. Log in to your My Sanford Chart account
2. Select the Menu icon
3. Find the My Record section
4. Click Genetic Profile

Your healthcare team must know which medications you are taking. This includes medications your doctor prescribes or those you can buy without a prescription, such as herbal supplements, pain relievers, or antacids.

If you have any updates to your medication lists call (605)-312-GENE (4363)
If you would like to schedule an appointment with the PGx Clinic call (605)-404-4000

Reply

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Medication

Do **not** stop taking your medication(s) or make any changes to your medication(s) without talking with your doctor first.

This section has a list of medications that may be affected by your genes. These genetic results will be reviewed by a member of your healthcare team. If you are taking any of these medications now or in the future, talk with your doctor. If you have any questions, please call (888) 424-2332 or (605) 312-GENE.



Amitriptyline

Also known as Elavil

Discussion with your doctor may be warranted based on genetic results if this medication is prescribed.

[More information...](#)

SANFORD HEALTH | MyChart by Epic | Log out

Menu | Messages | Visits | Test Results | Billing Summary

Chamanda

My Genetic Profile

Disease

This section has information about your genes and how they affect your health. These genetic results will be reviewed by a member of your healthcare team. Speaking with a genetic counselor or geneticist may be helpful. If you have any questions, please call (888) 424-2332 or (605) 312-GENE.

Long QT Syndrome | KCNQ1 Gene Variant | [More information...](#)

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Resources

- [What is genetic testing?](#)
- [What can I learn from testing?](#)
- [What are the different types of genetic tests?](#)
- [What are the benefits and drawbacks of genetic testing?](#)
- [How do I decide whether to be tested?](#)
- [Where can I find more information about genetic testing?](#)

All screenshots on this slide © 2024 Epic Systems Corporation.

Interruptive Alerts


BestPractice Advisory







Based on this patient's genetics:
Simvastatin doses greater than 20 mg are not recommended due to increased risk of myopathy and rhabdomyolysis.

Reference: Cooper-DeHoff, et al. The Clinical Pharmacogenetics Implementation Consortium Guideline for SLCO1B1, ABCG2, and CYP2C9 and statin-associated musculoskeletal symptoms. 2022

Remove the following orders? _____

<input checked="" type="checkbox"/> Remove	<input type="checkbox"/> Keep	 simvastatin (ZOCOR) 40 mg tablet Take 1 tablet (40 mg) by mouth every night at bedtime, Disp-90 tablet, R-4, e-Prescribing
--	-------------------------------	--

Apply the following? _____

<input type="checkbox"/> Order	<input checked="" type="checkbox"/> Do Not Order	 rosuvastatin (CRESTOR) 5 mg daily
<input type="checkbox"/> Order	<input checked="" type="checkbox"/> Do Not Order	 atorvaSTATin (LIPITOR) 10 mg tablet
<input type="checkbox"/> Order	<input checked="" type="checkbox"/> Do Not Order	 pravastatin (PRAVACHOL) 40 mg tablet
<input type="checkbox"/> Order	<input checked="" type="checkbox"/> Do Not Order	 CK

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Accept **Dismiss**

Non-interruptive Alerts

metoclopramide (REGLAN) 10 mg tablet ✓ Accept ✗ Cancel

⚠ Pharmacogenomic Warning ⬆

Maximum recommended total daily dose is 30 mg for treatment of GERD based on genetic results. Maximum recommended total daily dose is 20 mg for treatment of diabetic gastroparesis based on genetic results.

Product: **METOCLOPRAMIDE HCL 10 MG PO TABS** View Available Strengths

Sig Method: **Specify Dose, Route, Frequency** Use Free Text Taper/Ramp Combination Dosage

Dose: mg **5 mg** mg

Prescribed Dose: 5 mg
Prescribed Amount: 0.5 tablet

Route: **Oral**

Frequency: **3 times a day before meals** Before meals and at bedtime
4 times a day prn 4 times a day

Duration: Doses **Days** 30 days 90 days 1 year

Starting: Ending: First Fill:

Dispense: Days/Fill: Full (0 Days) 30 Days 90 Days

Quantity: tablet Refill:

⚠ Next Required ✓ Accept ✗ Cancel

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Education

Educational Tools & Approaches

Internal



In-services & Champions



Internal Website



Modules



Pocket Cards

External



Community Lectures



External Website



Patient Instructions through
the EMR



Medication Specific Resources

Residency Program



PGY2 – CLINICAL PHARMACOGENOMICS PHARMACY RESIDENCY

Preparing pharmacy residents to be leaders in pharmacogenomics while providing exceptional patient care opportunities in the clinical setting.

FAST FACTS

- 1 resident position
- ASHP-accredited
- Start date July 1, 2024 (flexible)
- Longitudinal rotations with opportunities to explore electives in areas of interest!

PROGRAM STRUCTURE

Required Longitudinal Experiences:

- Orientation
- Administration
- Clinical Decision Support
- Clinical Pharmacogenomics I
- Clinical Pharmacogenomics II
- Education
- Research

Following the orientation learning experience, residency learning experiences are generally longitudinal and built upon the three pillars of Imagenetics: Clinical Care, Education, and Research. Experiences related to the three pillars include:

Clinical Care

- Provide evidence-based interpretation of PGx results to optimize pharmacotherapy

- Staff multidisciplinary PGx clinic
- Assist with clinical decision support maintenance and updates
- Address PGx-related drug info questions

Education

- Contribute to educational materials for patients and healthcare providers
- Teach and precept students, PGY1 residents, and other learners
- Obtain teaching certificate

Research

- Evaluate PGx drug literature and statistics
- Conduct a year-long research project

QUALIFICATIONS

All residency candidates must meet the following prerequisites:

- Earned a PharmD degree from an accredited college of pharmacy
- Hold an active pharmacy license, or be eligible for licensure in South Dakota
- Be participating in or have completed an American Society of Health-System Pharmacists (ASHP) accredited PGY1 residency program or one in the ASHP accreditation process.

APPLICATION

Application materials and deadline will be managed through PhORCAS:

- Letter of interest
 - Address clinical areas of interest and reasons for pursuing PGY2 in PGx
- Curriculum Vitae
- Academic transcripts
- Three reference submissions

Applicants considered for a position will be invited for an interview with pharmacy administration, preceptors, and current residents. They will be required to review and present a brief case study.



PROGRAM CONTACT INFORMATION

Natasha Petry, PharmD, MPH, BCACP
Residency Program Director
PGY2 Clinical Pharmacogenomics

natasha.petry@sanfordhealth.org
(701) 234-6016

Sanford Imagenetics
1321 W. 22nd Street
Sioux Falls, SD 57105



STIPEND AND BENEFITS

The stipend is competitive and updated annually. Residents are provided paid health and vision insurance and 20 days of Allowed Time Away (ATA), which are separate from paid holidays off. All required travel expenses are also covered.

SANFORD HEALTH

Sanford Health, one of the largest health systems in the United States, is dedicated to the integrated delivery of health care, genomic medicine, senior care and services, global clinics, research and affordable insurance.

Sanford USD Medical Center is the largest hospital in South Dakota (545 patient beds) and academic teaching institution for the University of South Dakota Sanford School of Medicine. Sanford USDMC provides an exciting learning environment to disciplines including medicine, pharmacy and nursing.

ABOUT SIOUX FALLS, SOUTH DAKOTA

With over 200,000 people, Sioux Falls is a great place to live. This vibrant city offers everything from parks and entertainment to shopping and music. With easy access to outdoor activities like biking, boating, hunting and fishing, there is something for everyone. Sioux Falls strives to maintain one of the healthiest environments in which to live, work and raise a family.

FOR MORE INFORMATION

Websites:

- Imagenetics: imagenetics.sanfordhealth.org
- PGY2 Residency: www.sanfordhealth.org/residency-programs/pharmacy-residency

Scan the QR codes to access more information or ask a question:



Other Training Opportunities

- APPE
- IPPE



NDSU NORTH DAKOTA
STATE UNIVERSITY

Other schools upon request

- PGY1 Resident Electives
 - Sioux Falls Sanford
 - Fargo Sanford
- High school student shadowing
- NIH Laboratory Genetics and Genomics Fellowship elective



Research

Selected Publications

> [Pharmacogenomics](#). 2019 Aug;20(12):903-913. doi: 10.2217/pgs-2019-0043.

Implementation of wide-scale pharmacogenetic testing in primary care

Natasha Petry^{1 2}, Jordan Baye^{1 2 3}, Aissa Aifaoui¹, Russell A Wilke^{4 5}, Roxana A Lupu^{4 5}, John Savageau⁶, Britni Gapp⁶, Amanda Massmann¹, Deidre Hahn², Catherine Hajek^{1 5}, April Schultz^{1 5}

> [Front Genet](#). 2021 Mar 12;12:626845. doi: 10.3389/fgene.2021.626845. eCollection 2021.

Precision Population Medicine in Primary Care: The Sanford Chip Experience

Kurt D Christensen^{1 2 3}, Megan Bell⁴, Carrie L B Zawatsky^{5 6}, Lauren N Galbraith¹, Robert C Green^{3 5 6 7}, Allison M Hutchinson⁴, Leila Jamal^{8 9}, Jessica L LeBlanc¹, Jennifer R Leonhard¹⁰, Michelle Moore⁴, Lisa Mullineaux¹¹, Natasha Petry^{12 13}, Dylan M Platt⁴, Sherin Shaaban^{14 15}, April Schultz^{4 16}, Bethany D Tucker⁴, Joel Van Heukelom^{4 16}, Elizabeth Wheeler⁴, Emilie S Zoltick¹, Catherine Hajek^{4 16}; Imagenetics Metrics Team

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[Free PMC article](#)

Abstract

Genetic testing has the potential to revolutionize primary care, but few health systems have developed the infrastructure to support precision population medicine applications or attempted to evaluate its impact on patient and provider outcomes. In 2018, Sanford Health, the nation's largest rural nonprofit health care system, began offering genetic testing to its primary care patients. To date, more than 11,000 patients have participated in the Sanford Chip Program, over 90% of whom have been identified with at least one informative pharmacogenomic variant, and about 1.5% of whom have been identified with a medically actionable predisposition for disease. This manuscript describes the rationale for offering the Sanford Chip, the programs and infrastructure implemented to support it, and evolving plans for research to evaluate its real-world impact.

Keywords: clinical decision support; precision medicine; genetic testing; pharmacogenomics

> [Genet Med](#). 2022 Jan;24(1):214-224. doi: 10.1016/j.gim.2021.08.008. Epub 2021 Nov 30.

Improved provider preparedness through an 8-part genetics and genomic education program

Catherine Hajek¹, Allison M Hutchinson², Lauren N Galbraith³, Robert C Green⁴, Michael F Murray⁵, Natasha Petry⁶, Charlene L Preys⁷, Carrie L B Zawatsky⁸, Emilie S Zoltick³, Kurt D Christensen⁹; Imagenetics METRICS Team

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PMID: 34906462 PMID: PMC9121992 DOI: 10.1016/j.gim.2021.08.008

> [Pharmacogenomics](#). 2020 Nov;21(17):1207-1215. doi: 10.2217/pgs-2020-0088. Epub 2020 Oct 29.

Malignant hyperthermia susceptibility: utilization of genetic results in an electronic medical record to increase safety

Jordan F Baye^{1 2 3}, Natasha J Petry^{1 4}, Shauna L Jacobson⁵, Michelle M Moore¹, Bethany Tucker¹, Sherin Shaaban⁶, Amanda K Massmann^{1 3}, Nicole M Clark¹, April J Schultz^{1 3}

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PMID: 33118445 DOI: 10.2217/pgs-2020-0088

Abstract

Aim: This manuscript describes implementation of clinical decision support for providers concerned with perioperative complications of malignant hyperthermia susceptibility. **Materials & methods:** Clinical decision support for malignant hyperthermia susceptibility was implemented in 2018 based around our pre-emptive genotyping platform. We completed a brief descriptive review of patients who underwent pre-emptive testing, focused particularly on *RYR1* and *CACNA1S* genes. **Results:** To date, we have completed pre-emptive genetic testing on more than 10,000 patients; 13 patients having been identified as a carrier of a pathogenic or likely pathogenic variant of *RYR1* or *CACNA1S*. **Conclusion:** An alert system for malignant hyperthermia susceptibility - as an extension of our pre-emptive genomics platform - was implemented successfully. Implementation strategies and lessons learned are discussed herein.

Keywords: clinical decision support; inhalation anesthetics; malignant hyperthermia; malignant hyperthermia susceptibility; personalized medicine; pharmacogenomics.



Lessons Learned

Barriers

Technology
Integration /
Updates

Lack of
Insurance
Coverage

Education
Initiatives

Communication
Across Multiple
Disciplines

Physical
Distance

Maintenance

Strengths



**Emphasis on
physician-patient
relationship**



**Foundation of
education**



**Primarily clinical
focus
(not research)**



**Clinical evaluation is
both prospective
and retrospective**



**In-house lab
imputes discrete
genomics data in
EMR**



**Implementation
efforts focused
on evidence-
based sources**



**Effective use of
technology
within a rural
footprint**



**Support from
administration**

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Thank you for the invitation to speak today!

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