Implementing Pharmacogenetics into Primary Care: An Informatics Approach

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DOI

- HEE Genomics Education Programme: Primary Care Lead
- RCGP Joint Clinical Champion for Genomics in Primary Care (with Imran Rafi)
- PI for HAPPY Project: Funded by InnovateUK in collaboration with Congenica
- GP, Affinity Care PCN

Agenda

- Introduction to pharmacogenomics (PGx)
- PGx in the NHS
- The role of primary care

- Research into implementation strategies
- Opportunities and challenges
- The informatics approach

Medicines are an important part of NHS care and help many people get well

We spend £20.9 billion a year on medicine (£1 in every £7 that the NHS spends) and they are a major part of UK economy (COVID19 and Brexit economy impacts)

However, quality, safety and increasing cost continue to be an issue…

- Around **5-8% of hospital admissions** are medicines related, many preventable/avoidable (>13% in over 65s)
- Up to **50% of patients** do not take their medicines as intended
- Use of **multiple medicines is increasing** – over 1 million people now take 8 or more medicines a day, many of whom are older people (over 65s) – **'Problematic Polypharmacy'**
- **The challenges of Deprescribing**

Slide courtesy of Ravi Sharma, RPS Director for England

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CYP2C19 genotype testing to guide clopidogrel use ischaemic stroke or transient ischaemic attack

Diagnostics guidance [DG59] Published: 31 July 2024 Register as a stakeholder

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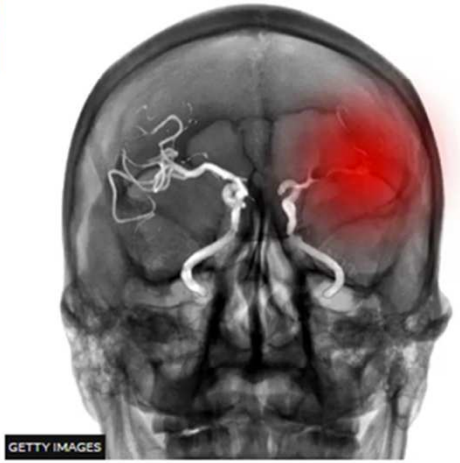
Health

Offer gene test to stroke patients, NHS told

19 May



GETTY IMAGES

By Michelle Roberts

Digital health editor

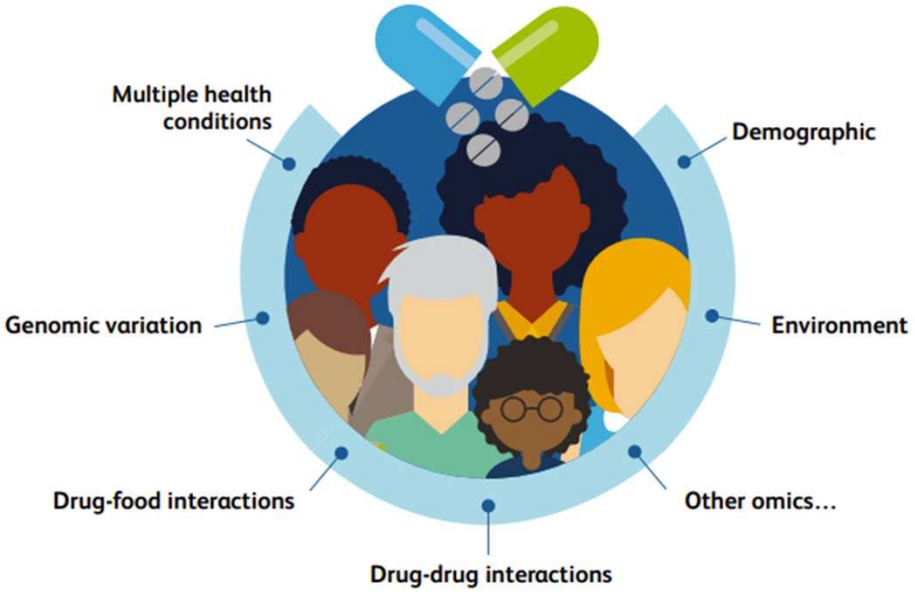
People who have had a stroke caused by a blood clot should have a DNA test to see whether they can be given a drug to help prevent more brain attacks, new draft guidelines for the NHS say.

Pharmacogenomics: a prescribing tool

Genomics vs Genetics

- The study of an organism's complete set of genetic information.
- The genome includes both genes (coding) and non-coding DNA.
- 'Genome': the complete genetic information of an organism.

- The study of heredity
- The study of the function and composition of single genes.
- 'Gene': specific sequence of DNA that codes for a functional molecule.

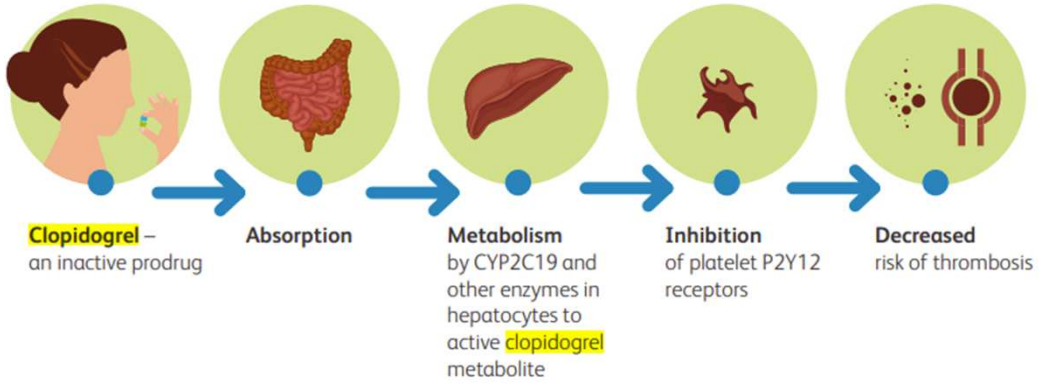


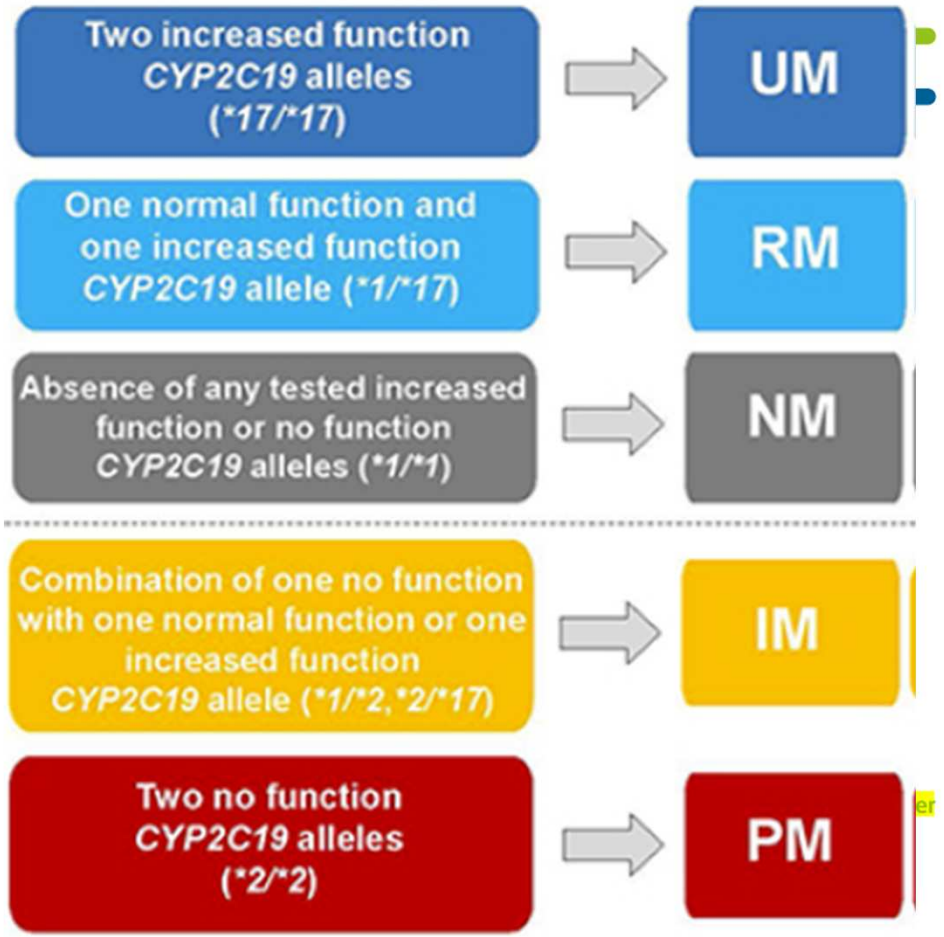
Pharmacogenomics: a prescribing tool

Right drug, right dose, right time

We can use a person's genomic information to **predict** their responses to certain drugs to improve treatment and avoid a potentially **serious** adverse reaction

Clopidogrel CYP2C19 effects



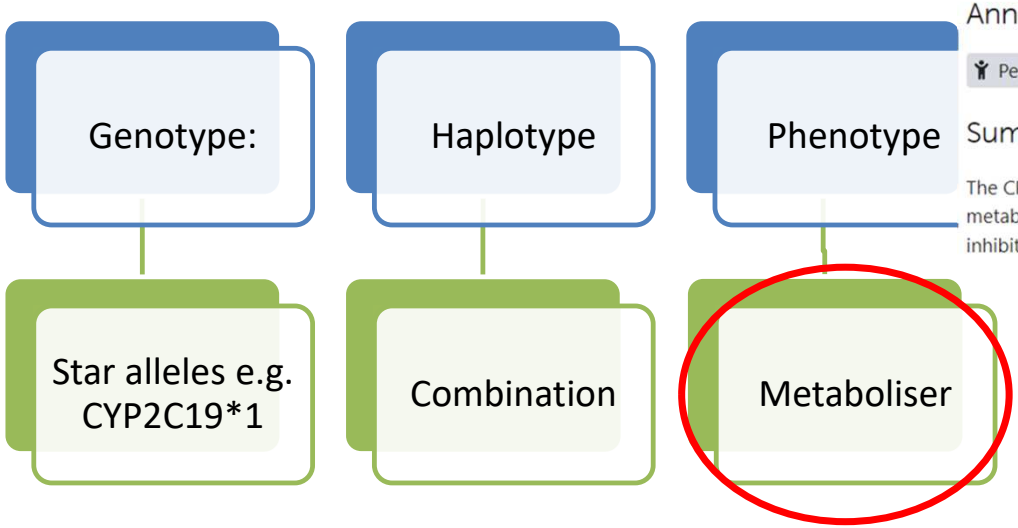


Prescription of Clopidogrel to CYP2C19 intermediate or poor metabolisers results in poorer clinical outcomes…..

- Increased rates of MI and CVA post-stenting
- Increased rates of CVA after initial event
- Alternatives more effective but expensive, bleeding risk (Tigacrelor / Prasugrel)

Personalised prescribing_main report_2_0 (2).pdf: Personalised prescribing: Using Pharmacogenomics to improve patient outcomes (RCP/BPS 2022)

PGx Data Pipeline



Annotation of CPIC Guideline for clopidogrel and CYP2C19

Pediatric

Summary

The CPIC Dosing Guideline for clopidogrel recommends an alternative antiplatelet therapy for CYP2C19 poor or intermediate metabolizers (cardiovascular indications: prasugrel or ticagrelor if no contraindication; neurovascular indications: alternative P2Y12 inhibitor if clinically indicated and no contraindication.)

PGx in the NHS: 4 tests currently available

Abacavir – *HLA-B*57:01*

A 33-year-old man with bilateral pneumonia is found to be HIV positive and agrees to commence antiretroviral therapy (ART). He undergoes genetic testing, which shows that he does not carry the HLA allele *HLA-B*57:01*, and is commenced on abacavir/lamivudine/dolutegravir, which he tolerates.

- Abacavir is a nucleoside reverse transcriptase inhibitor (NRTI) that inhibits HIV replication.

Fluoropyrimidines – *DPYD*

A 68-year-old man goes to his GP with altered bowel habits and is diagnosed with a left-sided colon adenocarcinoma. The oncologist undertakes *DPYD* genetic screening which reveals the patient carries a reduced-function *DPYD* genetic variant. He commences chemotherapy, which includes capecitabine at a 50% reduced starting dose. He tolerates this reduced dose and it is cautiously incremented to 75% of the standard dose over subsequent cycles.

- Fluoropyrimidines are antimetabolite chemotherapy drugs indicated in gastrointestinal, breast and head and neck cancer treatment. Fluoropyrimidines include parenterally administered 5-fluorouracil (5-FU) and its oral inactive forms (prodrugs) capecitabine and tegafur.
- Approximately 10–40% of patients who receive fluoropyrimidine therapy develop serious adverse

- NHS England and the relevant NHS organisations in Scotland, Wales and Northern Ireland *DPYD* genomic testing in October/November 2020, making routine testing for the four vari

R65 Aminoglycoside exposure posing risk to hearing

Testing Criteria

Significant exposure to aminoglycosides posing risk of ototoxicity

This indication would be relevant to:

1. individuals with a predisposition to gram negative infections for example due to known respiratory disease (e.g. bronchiectasis, cystic fibrosis) or due to structural or voiding genitourinary tract disorders, OR
2. individuals with hearing loss who have been exposed to aminoglycosides

NICE › BNF › Drugs › Azathioprine

Azathioprine

Pre-treatment screening

Thiopurine methyltransferase

The enzyme thiopurine methyltransferase (TPMT) metabolises thiopurine drugs (azathioprine, mercaptopurine, tioguanine); the risk of myelosuppression is increased in patients with reduced activity of the enzyme, particularly for the few individuals in whom TPMT activity is undetectable. Manufacturer advises consider measuring TPMT activity before starting azathioprine, mercaptopurine, or tioguanine therapy. Seek specialist advice for those with reduced or absent TPMT activity.

Primary care

genes

MDPI

Review

Pharmacogenomics for Primary Care: An Overview

Victoria Rollinson *,†, Richard Turner † and Munir Pirmohamed

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Table 1. Commonly used drugs in primary care with available pharmacogenomics guidelines.

Class	Drug	Gene	Actionable Result	Guideline Availability		Therapeutic Recommendations [1]
				DPWG	CPIC	
Lipid lowering agents	Atorvastatin	*SLCO1B1*	rs4145096 (521T > C) carriers	√	-	AD
	Simvastatin	*SLCO1B1*		√	√	LD, AD, M
Antidepressants	Citalopram	*CYP2C19*	PM, UM	√	√	LD (PM), AD (PM, UM)
	Sertraline	*CYP2C19*	PM, UM	√	√	LD (PM), AD (PM, UM)
	Amitriptyline	*CYP2C19* *CYP2D6*	PM, RM, UM IM, PM, UM	- √	√ √	LD (PM), AD (PM, RM, UM) LD (IM, PM), AD (PM, UM)
Analgesics	Codeine	*CYP2D6*	IM, PM, UM	√	√	AD (UM, PM), M (IM)
	Tramadol	*CYP2D6*	IM, PM, UM	√	-	AD (IM, PM, UM), ID (IM, PM), LD (UM)
Anti-platelet	Clopidogrel	*CYP2C19*	IM, PM	√	√	AD
Anticoagulant	Warfarin	*VKORC1* *CYP2C9* *CYP2C* region	*VKORC1 c.-1639G > A* **2, *3, *5, *6, *8, *11* rs12777823	√ √	√ √ √	LD [2] LD [2] LD
Anticonvulsant	Carbamazepine	*HLA-B* *HLA-A*	*HLA-B*15:02* detected *HLA-A*31:01* detected	- -	√ √	AD, M AD, M
Antibiotic	Flucloxacillin	*HLA-B*	*HLA-B*57:01* detected	√	-	AD, M
Contraception	Oestrogen-containing contraceptives	*F5*	rs6025 (p.R534Q) carriers	√	-	AD
Xanthine oxidase inhibitor	Allopurinol	*HLA-B*	*HLA-B*58:01* detected	-	√	AD

[1] = Where a drug-gene pair has guidance available from both CPIC and DPWG, the CPIC recommendations are detailed here. [2] = VKORC1 -1639G > A and CYP2C9 alleles are often combined together into an algorithm, alongside clinical variables, to guide initial warfarin dosing. Please note that CYP2C9 *1/*2 does not lead to recommended warfarin dose changes unless the VKORC1 -1639A allele is also present, and VKORC1 -1639GA does not lead to a dose change unless a CYP2C9 reduction-of-function allele is also present [9]. AD = alternative drug; CPIC = Clinical Pharmacogenetics Implementation Consortium; DPWG = Dutch Pharmacogenetics Working Group; LD = lower dose; ID = increase dose; M = consider additional monitoring (e.g., routine CK surveillance for simvastatin); PM = poor metaboliser; UM = ultra-rapid metaboliser; √ = guideline available; - = no guideline available.

99.4% of people carry a Pharmacogenomic variant:

Drug metabolism (Cytochrome, CYP)

Hypersensitivity (HLA)

Longitudinal exposure of English primary care patients to pharmacogenomic drugs: An analysis to inform design of pre-emptive pharmacogenomic testing

James E. Kimpton, Iain M. Carey, Christopher J.D. Threapleton, Alexandra Robinson, Tess Harris, Derek G. Cook, Stephen DeWilde, Emma H. Baker

First published: 27 August 2019 | https://doi.org/10.1111/bcp.14100 | Citations: 3

- 58% patients prescribed at least 1 PGx drug (GDP)
- 89% patients 70+ y.o.a. prescribed at least 1 PGx drug
- 47% patients prescribe 2 or more PGx drugs
- 7% patients prescribed 5 or more PGx drugs

- PGx most frequent prescribed:
  - Analgesia
  - Gastroprotection
  - Psychiatric conditions
  - Cardiovascular conditions

Primary Care context:

CYP2C19 metaboliser phenotype is relevant for prescribing decisions

SSRIs: Poor metabolisers:

- Citalopram: alternative or dose reduction (prolonged QTc)
- Sertraline: dose reduction

PPIs: Lansoprazole, Omeprazole, Pantoprazole

- Ultra-rapid: Increased starting dose
- Normal / intermediate: Increased dose in oesophagitis / H Pylori

Case study (HAPPY research study):

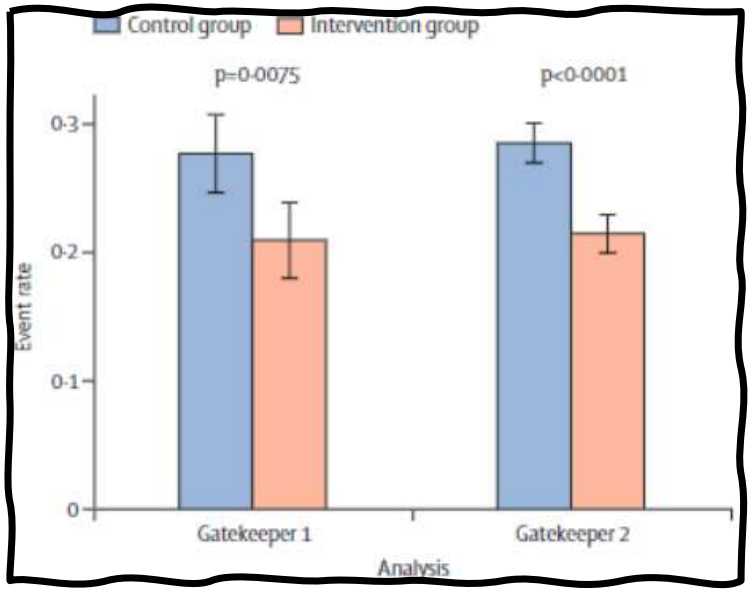
- 71-year-old lady admitted acutely via A+E with significant anaemia (Hb 67), Oesophagitis on Endoscopy. (UM)
- Colleague enquiry: 68-year-old with reduced SLCO1B1 function, Terbinafine prescription
- 68-year-old lady with treatment resistant depression, Citalopram 40mg. CYP2C19 PM. Psychiatrist advice: increased frequency ECG and rv if QTc abnormality.

The Value of Panel Based Pharmacogenetic Testing Has Been Demonstrated – The Question is How Can we Implement

Articles

A 12-gene pharmacogenetic panel to prevent adverse drug reactions: an open-label, multicentre, controlled, cluster-randomised crossover implementation study

Jesse J Swen, Cathelijne H van der Wouden, Lisanne EN Manson*, Heshu Abdullah-Koolmees, Kathrin Blagec, Tanja Blagus, Stefan Böhringer, Anne Cambon-Thomsen, Erika Cecchin, Ka-Chun Cheung, Vera HM Deneer, Mathilde Dupui, Magnus Ingelman-Sundberg, Siv Jonsson, Candace Joefield-Roka, Katja S Just, Mats O Karlsson, Lidija Konta, Rudolf Koopmann, Marjolein Kriek, Thorsten Lehr, Christina Mitropoulou, Emmanuelle Rial-Sebbag, Victoria Rollinson, Rossana Roncato, Matthias Samwald, Elke Schaeffeler, Maria Skokou, Matthias Schwab, Daniela Steinberger, Julia C Stingl, Roman Tremmel, Richard M Turner, Mandy H van Rhenen, Cristina L Dávila Fajardo, Vita Dolžan, George P Patrinos, Munir Pirmohamed, Gere Sunder-Plassmann, Giuseppe Toffoli, Henk-Jan Guchelaar, on behalf of the Ubiquitous Pharmacogenomics Consortium†*



The PREPARE Trial Demonstrated a 30% Reduction in ADRs

Review > Pharmacogenomics. 2021 Aug;22(12):761-776. doi: 10.2217/pgs-2021-0032.
Epub 2021 Sep 1.

Pharmacogenomic testing to support prescribing in primary care: a structured review of implementation models

Judith Hayward [1 2], John McDermott [3 4], Nadeem Qureshi [5], William Newman [3 4]

- Pre-test counselling and family history
- Role of the pharmacist
- Point of care CDSS
- Integrated with other patient factors affecting prescribing and guidance

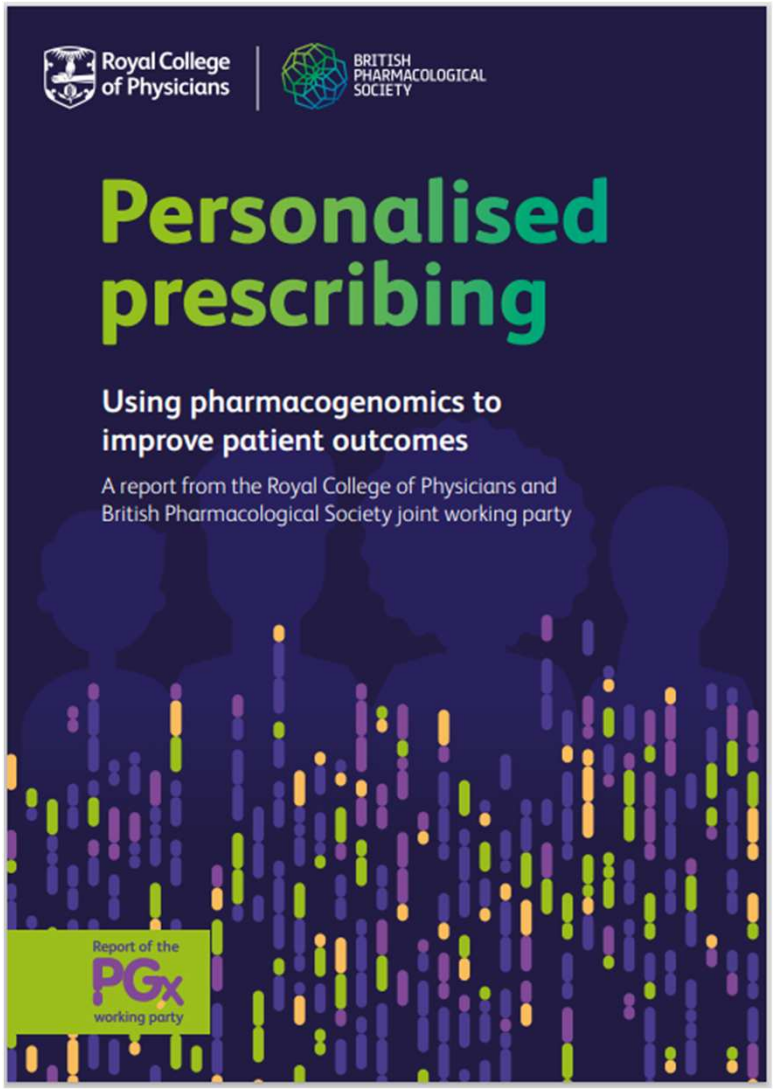
Implementation into Primary Care is feasible

Existing prescribing infrastructure and MDT working is key

- National infrastructure
- CDSS
- SMR
- Medicines Optimisation

- Evidence for clinical utility
- Development of the PCP and Pharmacist with expertise
- Definition of roles and responsibilities with MDT working
- Defined clinical pathways
- Integrated education
- Reimbursement / commissioning

Considerations for implementation:



Stakeholder organisations

This report is endorsed by the following organisations:

Association of Cancer Physicians
British Society for Allergy and Clinical Immunology
British Society for Genetic Medicine
British Society for Haematology
British Thoracic Society
Clinical Genetics Society
Genomics England
Royal College of Anaesthetists
Royal College of General Practitioners
Royal College of Psychiatrists
Royal College of Radiologists, Faculty of Clinical Oncology
Royal Pharmaceutical Society
UK Kidney Association

Acknowledgements

Thank you to all the clinicians and patient representatives who gave their time, insight and experience to support the development of this report.

We would also like to acknowledge staff from the RCP and BPS who supported the working party and report production, including Anna Zecharia, Sophie Joseph, Simon Land, Sophia McCully, Charles Whalley, Dina Koulama, Karen Porter, Jordan Marshall, Janet Leggett-Jones, Dr Norma O'Flynn, Karen Reid and Victoria Wilson.

This report is dedicated to the memory of Professor Donal O'Donoghue, who championed the creation of the working party and co-chaired it at its inception.

Ethnicity and PGx variation

Carbamazepine – *HLA*

REUTERS

World Business Markets Breakingviews Video More

HEALTHCARE & PHARMA FEBRUARY 15, 2021 / 11:26 PM / UPDATED A YEAR AGO

Bristol-Myers, Sanofi ordered to pay Hawaii $834 million over Plavix warning label

Family history in primary care: understanding GPs' resistance to clinical genetics — qualitative study

Jonathan Mathers, Sheila Greenfield, Alison Metcalfe, Trevor Cole, Sarah Flanagan and Sue Wilson

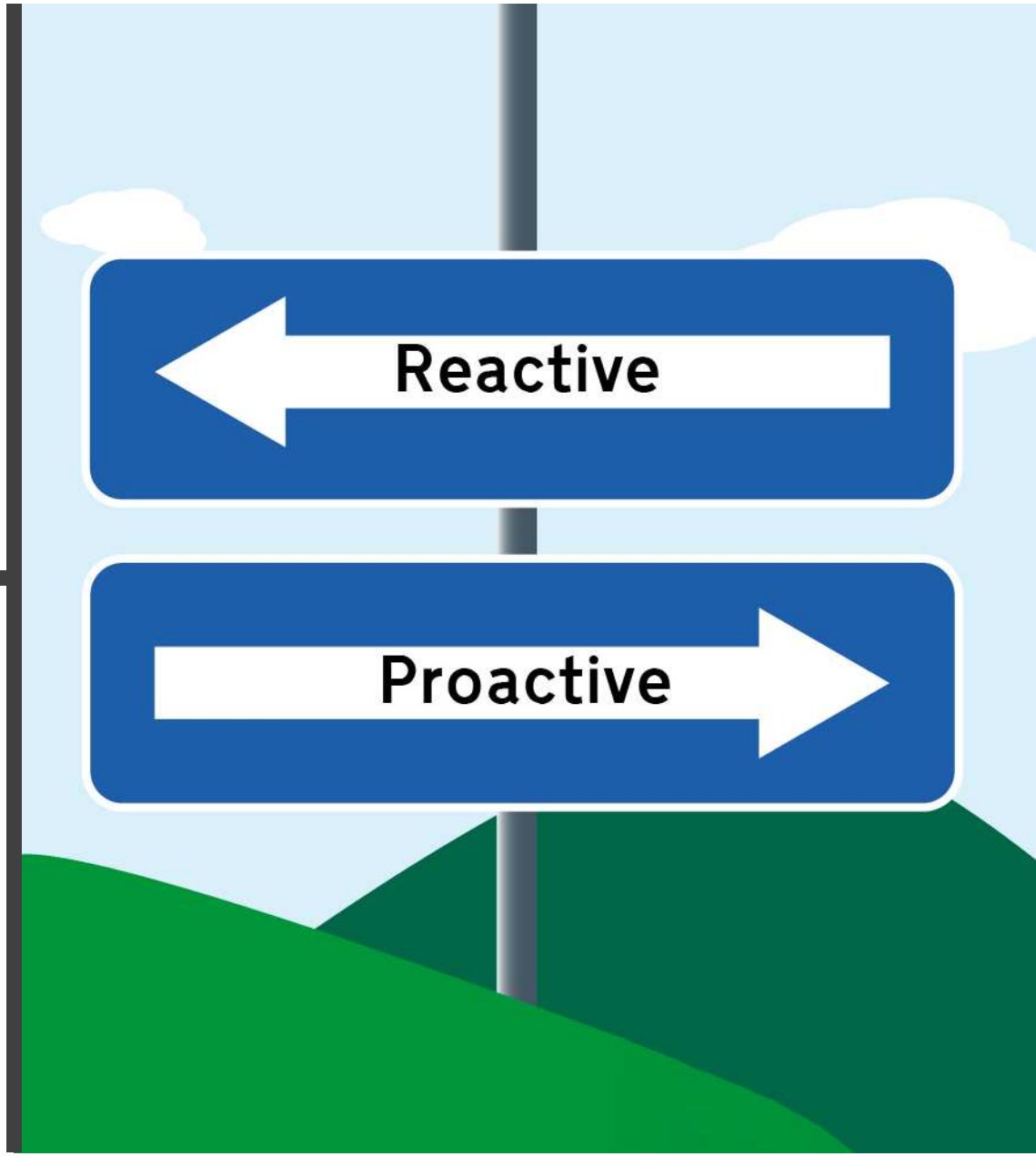
GPs seek information about genetics when they perceive it to be relevant e.g. family history

Primary-care providers' perceived barriers to integration of genetics services: a systematic review of the literature

Natalie A. Mikat-Stevens MPH ✉, Ingrid A. Larson RN, CPNP & Beth A. Tarini MD, MS

Genetics in Medicine **17**, 169–176(2015) | Cite this article

Lack of point-of-care tools identified as barrier



Genomics education in primary care: Evidence

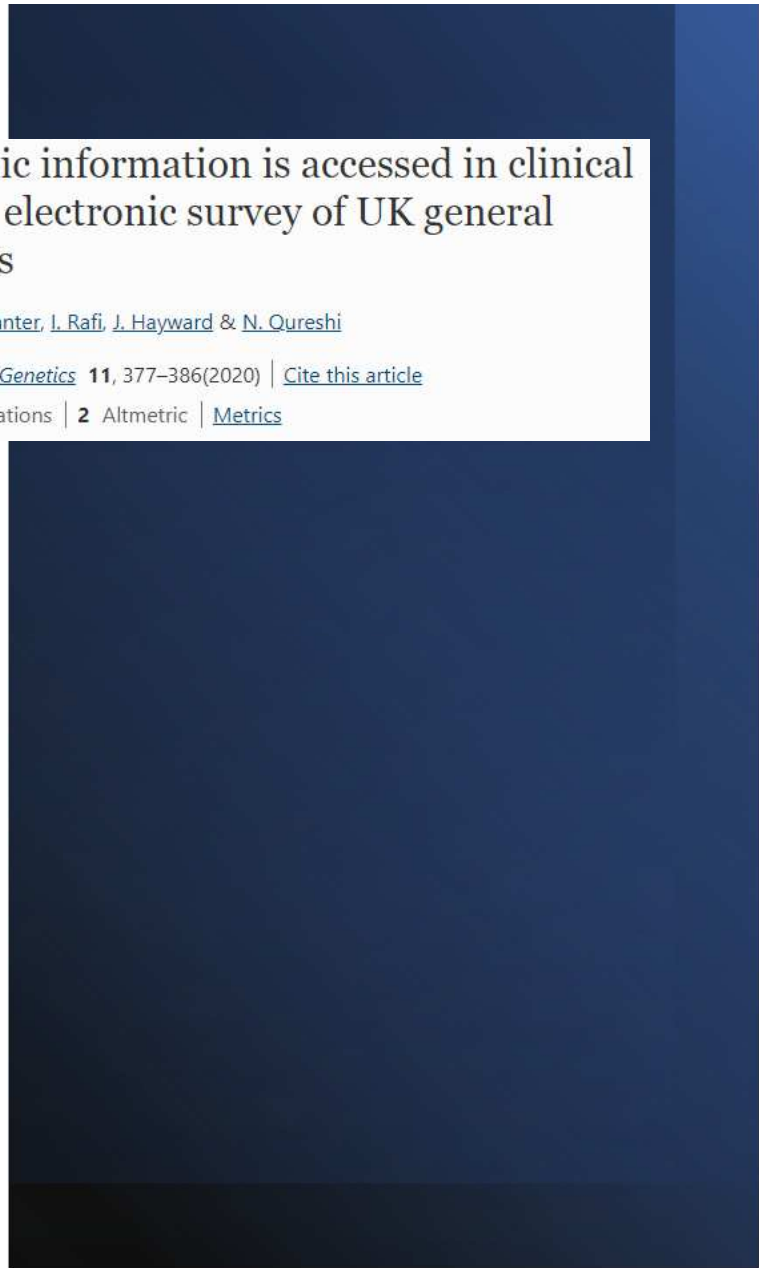
via concise, relevant and primary-care focussed resources

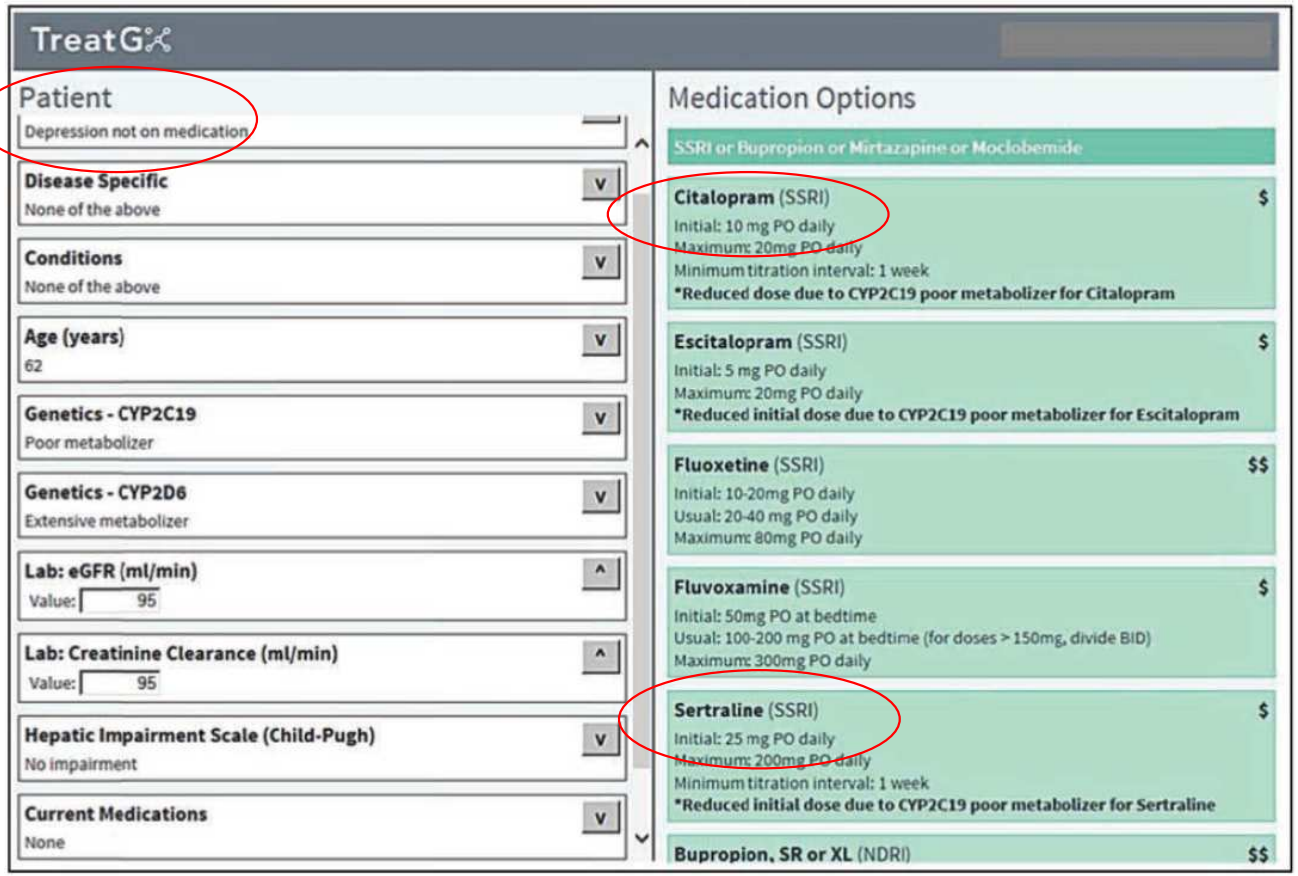
through familiar sources rather than genomic-specific websites;

Intranet repositories, clinical guidelines and pathways, local expertise, disease-specific websites

Pathways should utilize clinical ‘touch-points’ to give information, e.g. design of test reports

Clear preference towards online modules and resources (duration 30-60 mins).



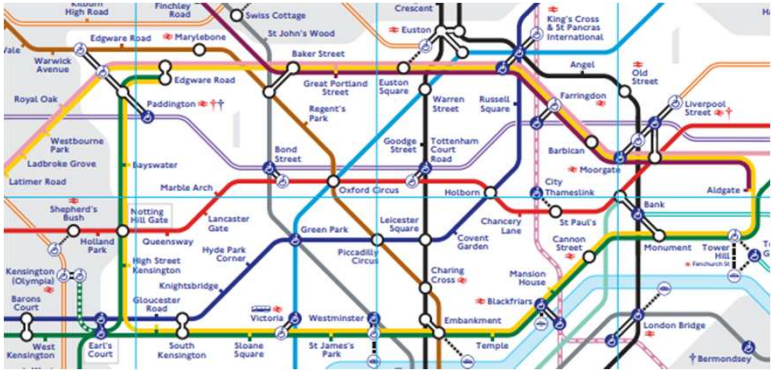


Medication Decision Support System: 236 times over 3 months

Dawes et al, 2016

NHSE Pharmacogenomics Digital Group: Data standards and test case

Unified Genomic Record (UGR)



Guidance (PRSB) and evidence all supports implementation with point-of-care prescribing support, integrating other factors affecting prescribing and up-to-date guidance

All implementation challenges lead to informatics solutions

Point-of-care educational tool: supported by guidance / GeNotes

Pharmacogenomics in primary care

BCS Primary Health Care Specialist Group
Annual conference, 15/11/2024

fava

The background

- We all respond differently to medicines
- Changes in our genetic code influences our response
    - This is known as '**pharmacogenomics**' or '**PGx**'
- Testing for these gene changes can personalise prescribing
- This can improve the safety and effectiveness of medicines

The problem

– Medicines are not personalised today!

– ‘Trial-and-error’ prescribing

– Costs related to medicines are high

– Implementation is limited

The opportunity

- This is about common medicines!
  - Statins, antidepressants, PPIs, clopidogrel
- These gene changes are common!
  - Most of us carry at least one gene change linked to our response to medicines

The opportunity

What if doctors and patients knew what medicines were safe and effective from the start?

Our research

Characterizing pharmacogenetic programs using the consolidated framework for implementation research: A structured scoping review

John H. McDermott[1,2]*, Stuart Wright[3], Videha Sharma[4], William G. Newman[1,2], Katherine Payne[3] and Paul Wilson[5]

Our research

ORIGINAL ARTICLE | Open Access

Public preferences for pharmacogenetic testing in the NHS: Embedding a discrete choice experiment within service design to better meet user needs

John H. McDermott, Videha Sharma, William G. Newman, Paul Wilson, Katherine Payne, Stuart Wright

First published: 14 April 2024 | https://doi.org/10.1111/bcp.16058

Our research

Article | Open access | Published: 09 August 2024

Understanding general practitioner and pharmacist preferences for pharmacogenetic testing in primary care: a discrete choice experiment

John H. McDermott ✉, Videha Sharma, Glenda M. Beaman, Jessica Keen, William G. Newman, Paul Wilson, Katherine Payne & Stuart Wright

The Pharmacogenomics Journal **24**, Article number: 25 (2024) | Cite this article

Our research

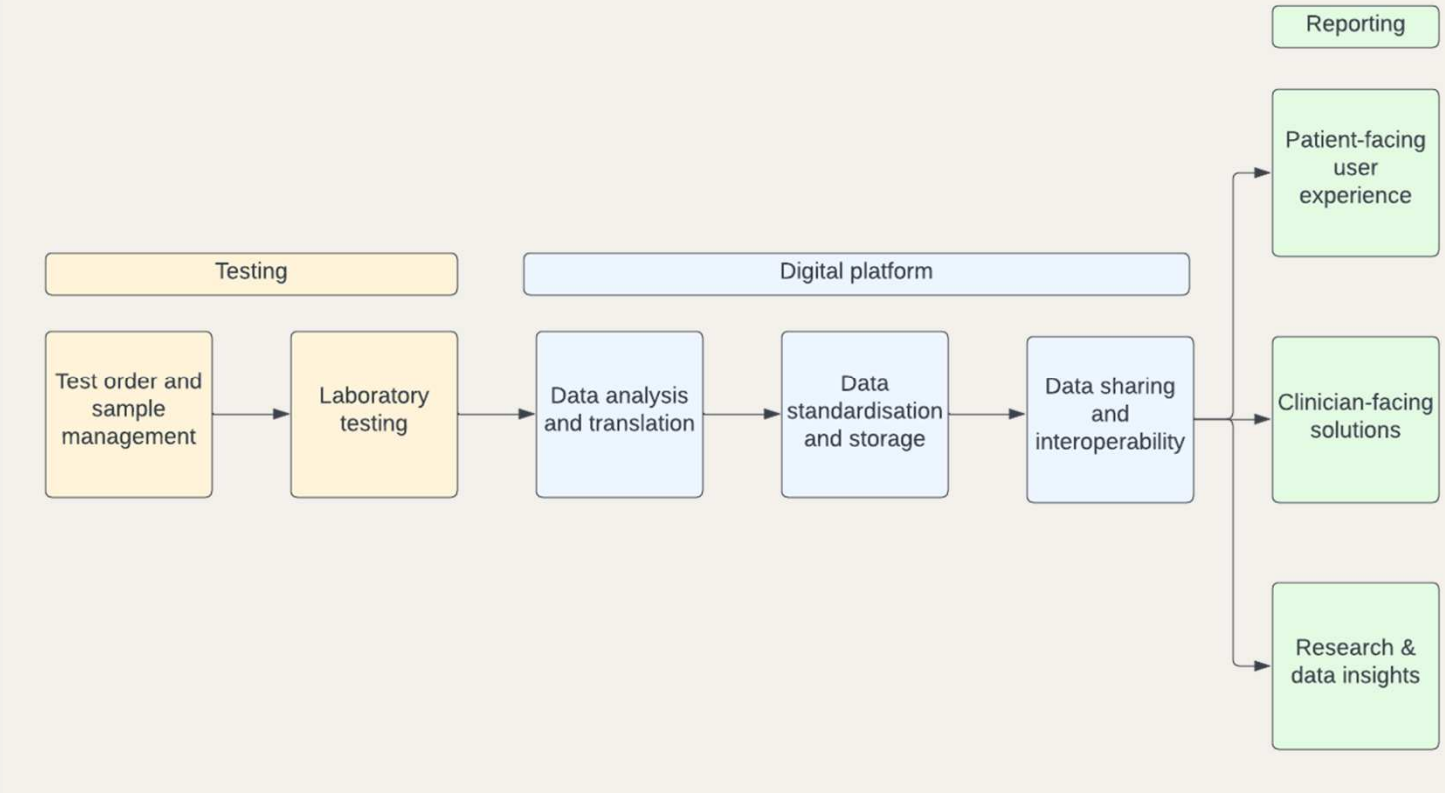
Pharmacogenetics Clinical Decision Support Systems for Primary Care in England: Co-Design Study

Videha Sharma[1, 2]; John McDermott[3, 4]; Jessica Keen[3]; Simon Foster[1]; Pauline Whelan[1]; William Newman[3]

Personalised medicine

- The potential benefits for PGx are significant
- Benefits are only realised if PGx is *integrated* in everyday care
  - Real-time clinical decision support to personalise prescribing decisions

The service


Testing
Digital platform
Test order and sample management
Laboratory testing
Data analysis and translation
Data standardisation and storage
Data sharing and interoperability
Reporting
Patient-facing user experience
Clinician-facing solutions
Research & data insights

The service

- **Patient selection:** How we decide who to test and how to phase roll out
- **Test ordering:** The operational process required to complete the test order and who is responsible for completing details including consent
- **Samples:** How and when samples are taken from patients and how samples are packaged and posted to the central lab
- **Receiving results:** The process around receiving results and logging them on local systems alongside the ideal workflow, if system integration was possible
- **Actioning results:** Who receives the results and is responsible for ensuring these are actioned, recorded and communicated
- **Prescribing:** Altering prescriptions, if required and communicating results to patients

The challenge

- **Patient selection:** How we decide who to test and how to phase roll out
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- **Prescribing:** Altering prescriptions, if required and communicating results to patients

The challenge

Myriad Genetics Stock Tanks After UnitedHealthcare Pulls Coverage of PGx Panels

Nov 04, 2024 | staff reporter

This story has been updated from a previous version to include a statement from Myriad and an updated stock quote.

NEW YORK – Shares of Myriad Genetics' stock were down around 4 percent Monday morning after tumbling about 18 percent on Friday following an announcement by UnitedHealthcare that it will no longer pay for pharmacogenetic multigene panel tests beginning Jan. 1, 2025.

In a note to investors, JP Morgan analyst Rachel Vatnsdal wrote that the payor was the largest and first one to reimburse for Myriad's GeneSight test, beginning in 2019. According to Vatnsdal, GeneSight represented approximately 18 percent of the company's total revenues in 2023.

In its new medical policy document, UnitedHealthcare now says that "the use of pharmacogenetic multigene panels (five or more genes) for the evaluation of drug-metabolizer status is unproven and not medically necessary for any indication due to insufficient evidence of efficacy."

The challenge

- It isn't that the use of PGx multi-gene panels for the evaluation of drug metabolizer status is unproven but the ability to impact *prescribing is challenging*

- Supporting prescribing decisions is dependent on digital infrastructure and integration with existing systems (this is really hard!)

- Innovation + infrastructure development → transition into a 'service'

GeneSight® Psychotropic

Pharmacogenomic Test

genesight

Patient, Sample
Date of Birth: MM/DD/YYYY
Clinician: Sample Clinician

Order Number: 0000000
Report Date: MM/DD/YYYY
Reference: 000000

Questions about report interpretation?
Contact our Medical Information team:
855.891.9415 | medinfo@genesight.com

Antidepressants

Non-Smokers | Smoking is defined as the daily inhalation of burning plant material (cigarettes, marijuana), and **excludes** vaping and e-cigarettes. This is used to determine medication results.

Use as Directed		Moderate Gene-drug Interaction		Significant Gene-drug Interaction	
desipramine (Norpramin®)		**venlafaxine** (Effexor®)	1	**bupropion** (Wellbutrin®)	2
desvenlafaxine (Pristiq®)		**selegiline** (Emsam®)	3	**amitriptyline** (Elavil®)	3
levomilnacipran (Fetzima®)		**fluoxetine** (Prozac®)	1,4	**paroxetine** (Paxil®)	4,6
nortriptyline (Pamelor®)		**clomipramine** (Anafranil®)	1,7	**escitalopram** (Lexapro®)	1,4,6
trazodone (Desyrel®)		**fluvoxamine** (Luvox®)	4,7	**sertraline** (Zoloft®)	1,4,6
vilazodone (Viibryd®)				**imipramine** (Tofranil®)	1,6,7
vortioxetine (Trintellix®)				**citalopram** (Celexa®)	1,4,6,8
duloxetine (Cymbalta®)	7			**doxepin** (Sinequan®)	1,6,7,8
mirtazapine (Remeron®)	7				

Clinical Considerations

1: Serum level may be too high, lower doses may be required.
2: Serum level may be too low, higher doses may be required.
3: Difficult to predict dose adjustments due to conflicting variations in metabolism.
4: Genotype may impact drug mechanism of action and result in moderately reduced efficacy.
6: Use of this drug may increase risk of side effects.
7: Smoking status changes the results of this medication. **See next section labeled Smokers for smoking results.**
8: FDA label identifies a potential gene-drug interaction for this medication.

All psychotropic medications require clinical monitoring. Medications should not be changed based solely on the test results. The results of this test are intended to supplement other clinical information considered by a healthcare provider within the context of a comprehensive medical evaluation.
This report is not intended to imply that the drugs listed are approved for the same indications or that they are comparable in safety or efficacy. The brand name is shown for illustrative purposes only; other brand names may be available. The prescribing physician should review the prescribing information for the drug(s) being considered and make treatment decisions based on the patient's individual needs, the characteristics of the drug prescribed, and the risk and safety information provided in the drug's labeling. Propranolol and oxcarbazepine prescribed for neuropsychiatric disorders might be considered off-label. Please consult their respective FDA drug labels for specific guidelines regarding their use.

The GeneSight Psychotropic test interpretations are based on a thorough review of published peer-reviewed literature, internal research, and FDA label information when applicable. The clinical validity and utility of the GeneSight Psychotropic test have been evaluated for patients with major depressive disorder who failed at least one psychotropic medication in multiple clinical studies.

Clinical requirements

- Prescribing happens in many clinical settings
  - Majority in primary care
- Mix of prescribers (end-users, who are unfamiliar with genomics!)
  - Doctors, specialist nurses, pharmacists, etc
- This is a ‘new’ clinical concept – it isn’t exactly an evaluation, diagnosis, or an allergy/intolerance
- Clinicians only need (want) to know the clinically relevant information
  - This is derived from raw genetic data – but is distinctly different
  - This needs to be presented within existing systems and workflows

The data requirements

- Genomic data is for life
- Genomic sequencing machines produce a range of outputs
- Health IT landscapes are varied, as are clinical decision support system

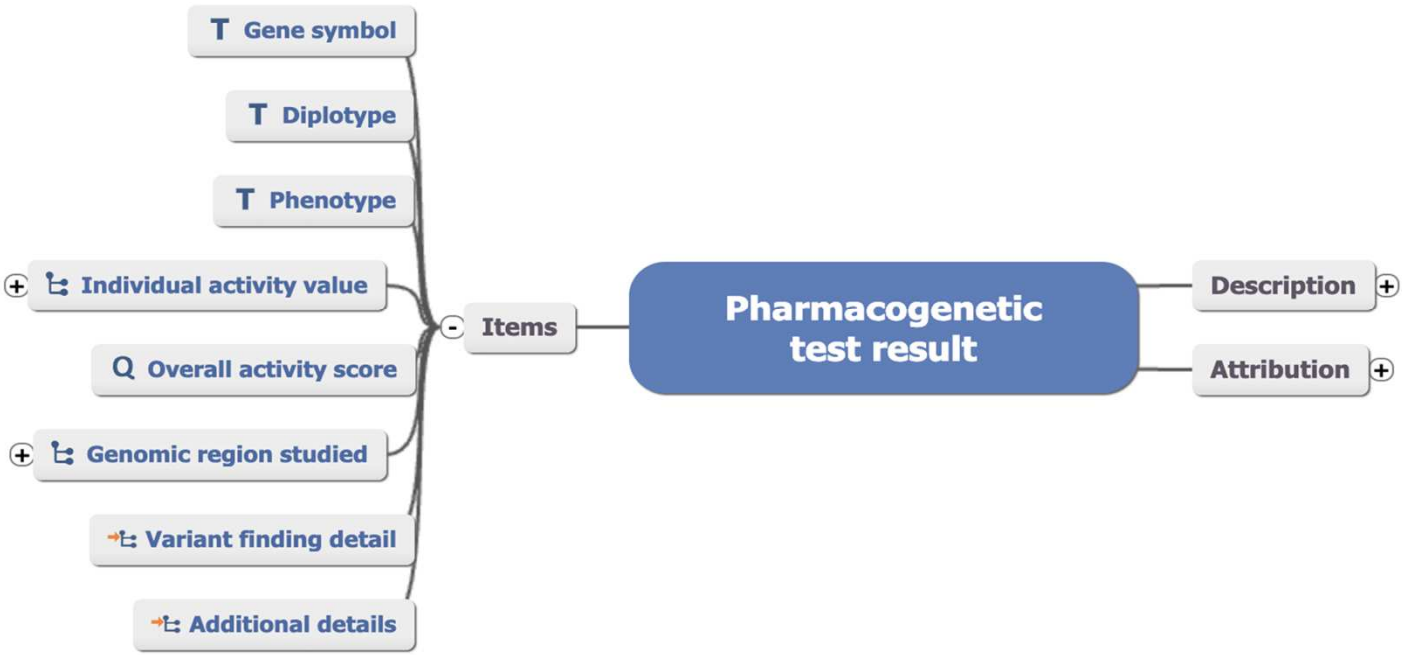
- This requires a vendor-neutral or agnostic approach based on open standards
- Strategy: *interoperable by design*

What is part of the clinical record?

What are my DNA variants?	Variants: NC_000010.10(CYP2C19):g.96612 4	No
What significant genes do I have?	Genotype: CYP2C9*1/*1: *1/*2	As background evidence only
For those genes that are biologically significant what is the impact?	Phenotype: CYP2C19 - Poor metaboliser	Key record fragment
What are the therapeutic implications?	Therapeutic implication: Clopidogrel - suggest alternative	Dynamic

Thank you to Ian McNicoll!

Open data model



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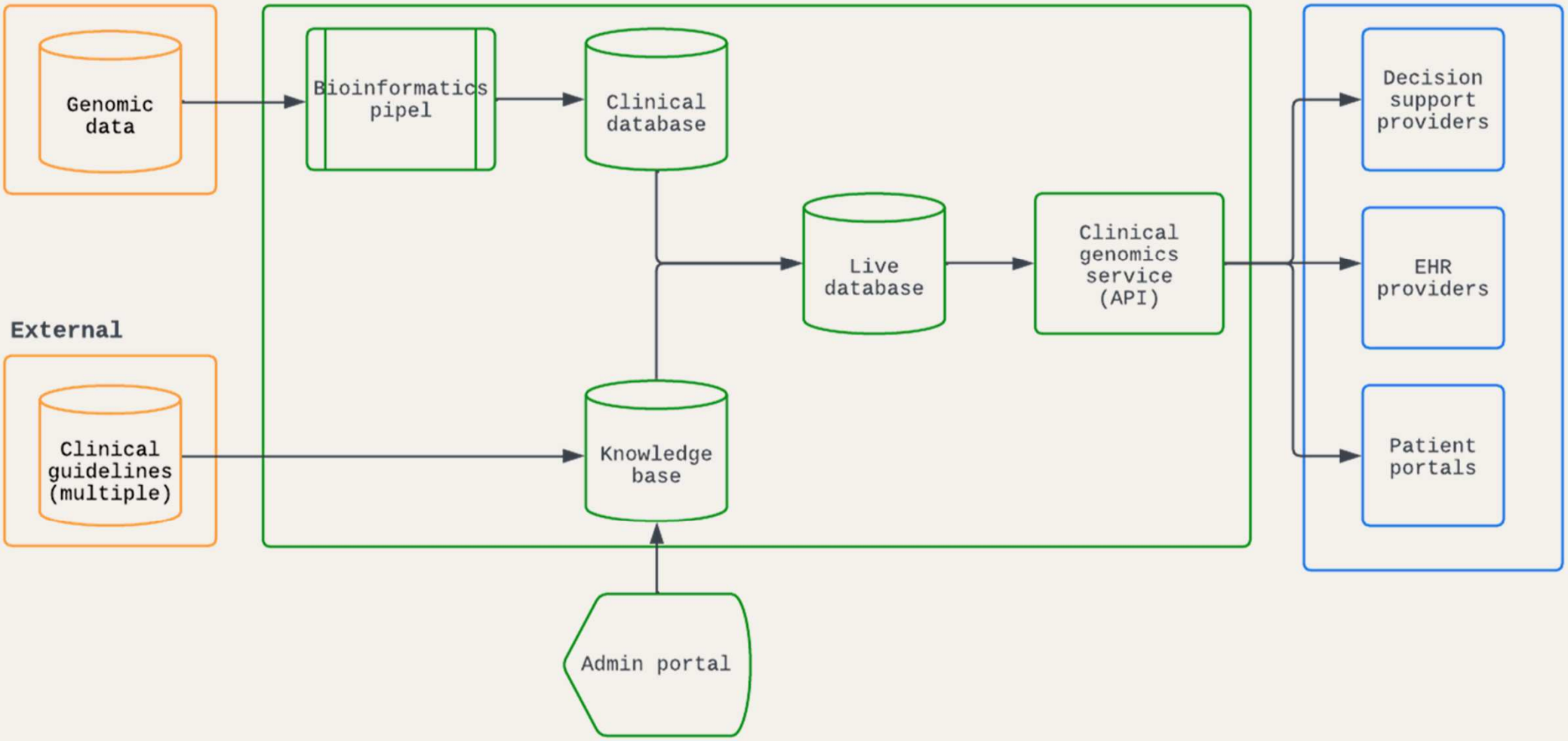
Genomic data
Bioinformatics pipel
Clinical database
Live database
Clinical genomics service (API)
Decision support providers
EHR providers
Patient portals
External
Clinical guidelines (multiple)
Knowledge base
Admin portal

Food for thought

- Is this ready for implementation?
- Are the digital and data challenges key to implementation?
- Is our strategy viable?

Feedback and questions