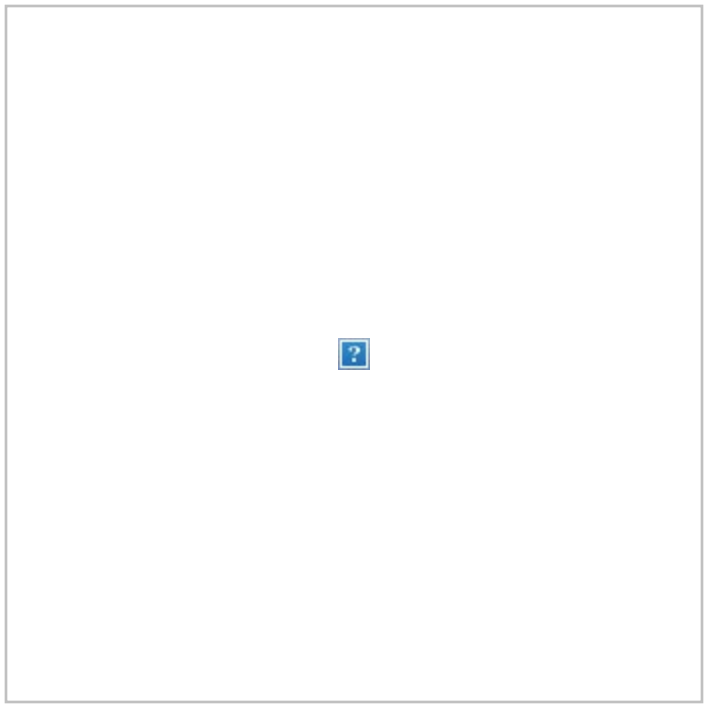
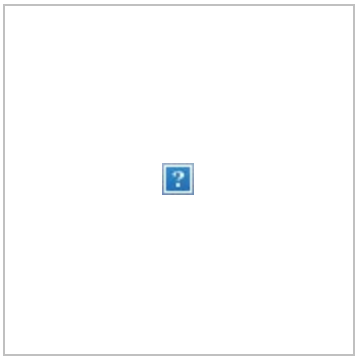
From: CHAUHAN, Dharmisha (THE ROYAL MARSDEN NHS FOUNDATION TRUST)
Subject: Issue 2: North Thames GMS Alliance Pharmacy Genomic Champions Newsletter



Pharmacy Genomic Champions Network



Issue 2
21st September 2022

North Thames GMS Alliance
PHARMACY AND GENOMIC MEDICINE

Dear Genomic Champions,

I started this role just over a year from today, and the importance of pharmacy in genomic medicine is becoming more and more evident. The role of pharmacogenetics to ensure an individual is treated with the right drug at the right dose on the NHS has approached, and to date there are two commissioned pharmacogenetic tests available on the NHS via the **NHS National Test Directory**.

- The first is **DPYD**, a pharmacogenetic test for fluoropyrimidine (5-FU) anti-cancer treatment, to identify individuals who would be at a greater risk of severe 5-FU chemotherapy toxicity.

- The second is **m.1555A>G**, a mitochondrial gene to help detect individuals who have a greater risk of ototoxicity with aminoglycoside treatment.

Over the next few years, we will see more pharmacogenetic tests within our clinical areas, it may continue to occur in specialised settings first, but the ultimate vision involves broader pharmacogenetic panel testing for a range of commonly prescribed drugs.

However, as this is developing, we must not forget the roles we already play. Within cancer care, pharmacists are involved in helping to ensure the right patient receives the right drug based on their **tumour genetic profile**, and these **drug-gene or drug-biomarker matches** are pivotal in the role of precision medicine to improve patient outcomes. Outside of cancer care, pharmacists working within cystic fibrosis have seen the positive impact of immunomodulators for individuals who harbour the **cystic fibrosis transmembrane conductance regulator (CFTR) gene**. The approvals of drug-gene or drug-biomarker clinically actionable variants will only increase over time, **across all disease indications** and therefore the role of pharmacists in genomic medicine will continue to evolve.

Since the last newsletter, the North Thames GMS Alliance team have continued to work hard on several projects to ensure equitable access to genetic testing and supporting services to improve clinical pathways. A few of our pharmacy highlights are provided below.

In addition, we are committed to **improve the genomic literacy** amongst **ALL** pharmacy professionals and this includes pharmacy technicians. We have lunchtime webinars, podcasts and champion engagement sessions, please see our future events section at the bottom of this newsletter for more information on how to get involved.

Please share this newsletter with your networks/teams and any questions please do not hesitate to contact me.

Kind regards,

Dharmisha Chauhan
Lead Genomic Medicine Pharmacist

Contents

National: Aminoglycosides and m.1555A>G testing

Together with Veronica Chorro-Mari, our senior pharmacist within the NT GMS Alliance team, we carried out two educational and training sessions on the importance of testing for a mitochondrial gene variant m.1555A>G linked to risk of ototoxicity after a single dose of an aminoglycoside, such as gentamicin.

Currently, this test which has an **approximate turnaround time of 10 days** and is currently conducted within our North Thames paediatric population. However, to ensure equitable access to all, we are working at a national level with the other Genomic Medicine Alliance Pharmacists to implement equitable testing within cystic fibrosis patients first. Other clinical areas will follow after identifying the priority patient cohorts on long-term aminoglycoside across England. This work also includes close collaborative working with our North Thames

Genomic Laboratory Hub.

The current test directory states that testing should be considered for individuals i**n whom significant exposure to aminoglycosides posing risk of ototoxicity is likely to occur.**

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.

A recording of our aminoglycoside and m.1555A>G educational and training package will be available soon on the North Thames website.

If you have any questions in the interim, please do contact us. Contact details provided below.

Pharmacogenetics and Mental Health

A Regional Transformational Project

Over the last four months, I have worked to place together a mental health specialist interest group. Within the group we have 6 specialist mental health pharmacists, psychiatrists, a clinical fellow and patient and public member representatives. Together we will help to interpret pharmacogenetic reports from a clinical trial, which is recruiting 2000 patients, in which 400 patients will have pharmacogenetic testing undertaken for CYP2D6 and CYP2C19, amongst other CYP enzymes to help guide treatment.

Pharmacogenetics is one element of medicines optimisation, and the aim will be to understand how the reports can be interpreted alongside clinical factors we routinely consider when reviewing medications e.g., renal and hepatic function, therapeutic drug monitoring and consideration of co-morbidities, amongst other patient factors. Further updates

from this project will be provided within the next pharmacy champions engagement session.

As experience builds, MDT workshops will be developed to share learning and best practice through case studies. Additionally, by the end of the project a pharmacogenetic manual for selective serotonin reuptake inhibitors and/or tricyclic antidepressants will be produced.

A call for interest: I am looking for a community pharmacist or PCN pharmacist with expertise in mental health. If this is you and you would like to be involved within the mental health specialist interest group, please contact me. Will be great to have a community representative on board!

Familial Hypercholesterolemia and the role of the pharmacist

By Dominic Studart

Clinical Nurse Specialist

Familial Hypercholesterolaemia (FH) is a genetic condition that impairs the liver's ability to remove excess cholesterol from the blood. Lifelong exposure to elevated LDL cholesterol greatly increases the incidence of premature cardiovascular disease (CVD) in FH individuals. If untreated, half of men with FH will have a heart attack or stroke before age 50 and a third of women before age 60. FH is very treatable and involves a combination of medical therapy such as statins, alongside healthy diet, and lifestyle. It is estimated that 1 in 250 people have FH, but currently only 8% have been diagnosed in England.

Genetic testing for FH testing is now available in the NHS and cascade genetic testing of 1st degree relatives is a cost-effective means of identifying affected family members. However, identifying index test cases is the greatest issue in reducing the incidence of CVD in these high-risk patients. The NHS long term plan aims to increase FH detection to 25% by 2024. To support this goal North Thames GMSA is leading on

a pilot to showcase the power of the primary care workforce to screen their patients for FH genetic testing.

The pilot utilises the UCLPartners (UCLP) FH searches and proactive care framework for FH screening in primary care. The UCLP FH searches identify patients in the primary care electronic health records that may have FH according to NICE guidelines. The FH proactive care framework is led by primary care Pharmacists/Nurses, who work together with Healthcare Assistants to screen these patients for FH. Other causes of hypercholesterolaemia e.g., diabetes or hypothyroid must be excluded, and family history data of premature CVD is collected. This information can then be used to identify patients who would benefit from FH genetic testing. First degree relatives of those found to have FH will also be offered FH testing. This work is supported by collaboration with the FH nurses and clinicians at St Bartholomew's Hospital.

For more information and to get involved in screening for FH in primary care with the UCLP searches and proactive care frameworks:

https://www.nice.org.uk/guidance/cg71/resources/familial-hypercholesterolaemia-identification-and-management-pdf-975623384005

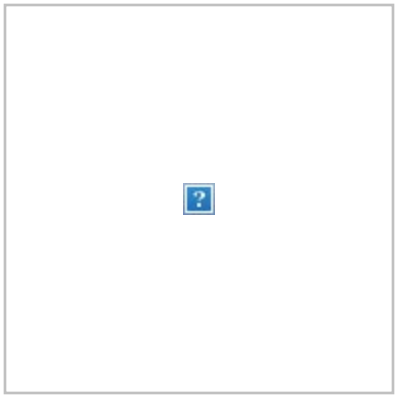
https://www.genomicseducation.hee.nhs.uk/about-us/supporting-the-national-transformation-projects/transformation-project-familial-hypercholesterolaemia/#toggle-id-6

https://s31836.pcdn.co/wp-content/uploads/Implementing-UCLPartners-Familial-Hypercholesterolaemia-Searches-and-Screening_Final.pptx

https://www.youtube.com/watch?v=8u0uTOQrxsE

Upcoming Educational Events:

Pharmacy Champion Engagement Session



Our next pharmacy champion engagement session will be held in November. Details will be released soon.

In the meantime, you can access our **Genomics Now** podcast series available on Amazon music, Google podcasts, Spotify and iTunes.

Series 1: NHS Genomic Medicine Service
Learn how the GMS evolved, how NHS infrastructure has changed to allow genomic testing to become part of routine care and the healthcare areas the GMS is committed to deliver.

Series 2: Primary Care Genomics Medicine
Includes advice and explanations for general practitioners to guide the use of genomics in their practice and discusses the vital role that primary care plays in supporting the equity of access to genomic tests. **Includes episodes on direct-to-consumer testing and pharmacogenomics.**

Series 3: Genomics in Disease
Explores the role of genomics in disease, and the ways in which genomics can be used to improve patient care in various clinical scenarios including cancer, rare disease, and prenatal medicine.

Contact

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North Thames GMS Alliance
Hosted by UCLPartners