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IN RARE FORM

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IN THIS ISSUE

Pharmacogenomics: A Word to Remember

by James Doulgeris

Pharmacogenomics uses our genetic makeup to determine the best drug, dosage and frequency to maximize its effectiveness and minimize side effects – and it's within our reach today.

Pharmacogenomics is the study of how your genetic makeup can affect your response to drugs. It involves identifying genetic variations that influences your ability to metabolize, absorb, and respond to a medication. This information is then used to tailor drug therapy to your unique genetic profile, to optimize treatment outcomes and to reduce the risk of side effects. For example, you may have a specific genetic variation that metabolizes a certain medication more quickly or slowly than someone without that variation, which could affect the dosage or frequency of the drug needed giving you enhanced results.

Pharmacogenomics is an emerging field in medicine and is gaining acceptance as a best practice in many areas of drug therapy including pain management, where genetic variations can have a significant impact on treatment response. However, the use of pharmacogenomics in pain management is not yet widely accepted as a best practice. This is primarily due to lack of standardization, lack of a robust evidence base, and the cost of genetic testing and limited accessibility to genetic testing.

Nonetheless, pharmacogenomics testing in pain management is increasingly considered to be an important tool in individualizing patient care and improving drug safety, but more research and implementation is needed to gain full adoption in the medical community and payment by insurers.

As a result, the use of pharmacogenomics in pain management is an area of active research and development. An increasing cohort of studies agree that genetic variations influence a person's response to certain pain medications, such as opioids. For example, certain genetic variations may affect how quickly a person metabolizes opioids, which could affect their risk of side effects or the effectiveness of the medication including indicating paradoxical reactions where side effects may outweigh benefits or risk of dependency or addiction may be substantially reduced.

Individualized pain management for CRPS lags other fields such as cancer

As research in the field continues to develop and more data is available, it is certain that pharmacogenomics will play an increasingly important role in the individualized management of pain, particularly in patients with chronic pain, where the risk of side effects and addiction are perceived to be high among political and regulatory bodies despite a compelling body of evidence to the contrary.

Pharmacogenomics has the potential to play a role in the individualized management of pain, called precision medicine, particularly for patients with chronic pain or those who have not responded well to traditional treatment options. By identifying genetic variations that may affect a person's response to pain medications, healthcare providers can tailor treatment to an individual's unique genetic profile, which could lead to better treatment outcomes and reduced risk of side effects.

For example, research has shown that certain genetic variations in the genes that encode for enzymes involved in the metabolism of opioids such as CYP2D6, CYP2C19, CYP3A4, and UGT2B7 can affect a person's response to these medications. This information can help healthcare providers select the most effective opioid medication and dosage for a patient to optimize pain control while minimizing side effects.

Pharmacogenomic testing can also help to identify which patients are at the greatest risk of adverse reactions to non-opioid pain medication such as non-steroidal anti-inflammatory drugs (NSAIDs) and anti-seizure medications and those who may not receive a pain management benefit equal to the risk.

While pharmacogenomics has the potential to improve pain management, it is not a standalone solution. Using these tests in conjunction with other clinical information such as

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Zoomalong Escape Room for Kids - Feb 26th

RSDSA and The Coalition Against Pediatric Pain (TCAPP) will host a virtual Escape Room on Sunday, February 26, 2023 at 2pm Eastern for kids ages 5-17. The theme is Maze Runner Grievers. This will be the first of four activities presented by RSDSA and TCAPP for 2023.

The fee is $10 and is refundable when your child attends. The limit is 25 children, first come-first served. The deadline to register is February 24, 2023.

Rare Disease Day is February 28th

February is Rare Disease Month with Rare Disease Day taking place annually on February 28th!

Each year, our friends at National Organization for Rare Disorders, Inc. (NORD) encourage everyone to take pictures wearing striped clothing and share them on social media on February 28th by using the hashtag #ShowYourStripes to raise awareness for rare diseases. We'll give you bonus points if your stripes are orange!

As we lead up to Rare Disease Day, we'll share interesting rare disease facts on our Facebook Page. Head there now to see them!

Interested in Writing for the RSDSA Blog?

We're always looking for Warriors who want to tell the story of their CRPS journey story on the RSDSA blog!

If you're interested in sharing your story with us and fellow Warriors as a form of therapy and/or in order to help those who may be in the same part of their journey as you are, send us an email at alexisdavis@rsds.org.

RSDSA's Young Adult Weekend Retreat is June 2nd-5th

RSDSA is excited to announce that the next Young Adult Weekend Retreat will take place June 2nd-5th in Philadelphia! Young adults with CRPS between the ages of 21 and 31 will spend the weekend with people who fully understand what they are going through.

Our Young Adult Weekend Retreats have been a great success as they offer young adults with CRPS the opportunity to network, attend workshops, learn to advocate for themselves, and build a support system all while leaving time to sightsee and have unique experiences.

Date: June 2-5, 2023
Location: Holiday Inn Express Midway | 1305 Walnut Street | Philadelphia, PA 19107
Cost: $250

Please note that all room fees are based on double occupancy. A $50.00 non-refundable deposit is required to reserve your spot. You can also choose to pay in full at the time of registration. Attendees will have the option to pay by check or credit card.

We have limited space so please register as soon as possible.

Final payment of outstanding balance is due May 20, 2023 with all information and emergency forms. With help from sponsors and donations, RSDSA will cover the additional costs for rooms, meals, and speakers.

If you have any questions or concerns, please contact RSDSA's Young Adult Retreat Committee at yaphili.rsdsa@gmail.com.

We look forward to seeing you there!

Learn More About The Dane Foundation

The Dane Foundation addresses the unique needs of individuals with developmental and physical disabilities. They specialize in providing "quality-of-life" items such as clothing, toiletries, household cleaning supplies, some food, paper towels, toilet paper, and other similar items. Reach out via Info@TheDaneFoundation.org.

Facebook Live with Cynthia Toussaint and John Garrett of For Grace

Join RSDSA for our Facebook Live with Cynthia Toussaint and John Garrett of For Grace at facebook.com/rsdsa on **Thursday, March 2nd at 7:00p Eastern** as they discuss the journey of CRPS and cancer.

Cynthia Toussaint founded For Grace in 2002 to raise awareness about CRPS and five years later expanded the organization's mission to include all women in pain. Cynthia has had CRPS for 40 years. She later developed Fibromylagia and other over-lapping, auto-immune conditions. Her most recent diagnosis is Triple Negative Breast Cancer which she is currently battling.

Toussaint was the first CRPS sufferer to be featured in the New York Times, Los Angeles Times and on the Public Broadcasting System and National Public Radio. She is a consultant for The Discovery Channel, ABC News, FOX News, the National Pain Report and PainPathways, the official magazine of the World Institute of Pain. Also, she is a guide and guest contributor for Maria Shriver's Architects of Change website.

She continues to be a leading advocate for health care reform in California. She was instrumental in changing public opinion which sparked sweeping HMO reform legislation that was signed by Governor Gray Davis in 1999. Her focus has now shifted to creating a single-payer, universal health care plan in California that would provide a model for the rest of the country. Her work is now strongly reflecting her passion for the pain-cancer connection and Post-Traumatic Growth.

John Garrett serves as Director at For Grace and was instrumental in launching the organization in April 2002 along with his partner, Cynthia Toussaint. Garrett has been partner and caregiver to Toussaint for 41 years. He has done extensive research about the gender disparity toward women in pain, compiling a comprehensive library on that issue along with specific chronic pain conditions.

Garrett's work focuses on speech presentation, grant writing, research, media outreach and the development of branding strategies. He has also advised California's Department of Managed Health Care and other state agencies regarding pain management practices in the HMO industry. Commenting on her long-term partnership with Garrett, Toussaint said, "My story as a woman in pain is also a love story because John's support has been total and unwavering. Without his loving presence in my life, I wouldn't be here."

Garrett made numerous writing contributions, reflecting on his role as a caregiver, trauma survivor, witness and partner, in Toussaint's memoir, *Battle for Grace: A Memoir of Pain, Redemption and Impossible Love*. Also, Garrett continues to make public speaking and media appearances that share the virtues and challenges of the caregiving experience.

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patient history and physical examination, comorbidities and other health status indications will deliver the best results. The scarcity of specific skill and experience sets in rare diseases like CRPS/RSD combined with the cost of genetic testing, limited accessibility and insurance coverage are still major challenges that need to be addressed before it can be widely adopted in the field of pain management.

Nonetheless, health insurers are increasingly approving payment for pharmacogenomic studies

Whether or not health insurers will pay for pharmacogenomic studies varies not just on the insurer but the specific circumstances of the test. Often the coverage for pharmacogenomic testing may depend on whether the test is considered medically necessary. The first step is to check with your insurance provider to see if they will cover the cost of the test. You can do this by calling the customer service number on the back of your insurance card or by visiting the insurance provider's website.

When you contact your insurance provider, you should have your policy number and any other relevant information available, such as the specific test you are interested in having done. You should also ask if the test is considered medically necessary and if there are any specific criteria that must be met for the test to be covered.

Even if your insurance does not cover pharmacogenomic testing, they stand to benefit the most. Here is the best way to make your case:

1. **Gather information:** Research the specific test you are interested in having done and the benefits it can provide. Your doctor can help with this task and may be the best person to make this call or provide you with a letter of medical necessity. This may include information on how the test can improve treatment outcomes, reduce the risk of adverse reactions, and lower healthcare costs.
2. **Contact your insurance provider:** Explain the specific test you are interested in having done and the benefits it can provide. Provide any relevant information, such as a letter of medical necessity from your doctor if they are not calling directly.
3. **If your request is denied, make an appeal:** If your insurance provider denies coverage for the test, you have the right to appeal the decision. Your insurance provider is required to provide information on the appeals process and any necessary documentation.
4. **Be persistent:** If at first, you don't succeed in getting coverage, don't give up. Keep in touch with your insurance provider and healthcare provider. Keep providing them with new information and studies about the clinical and economic benefits of pharmacogenomic testing. Enlist your doctors to help.

Please keep in mind that even if your health insurer covers the cost of a pharmacogenomic test, there may still be out-of-pocket costs for the patient, such as co-pays or deductibles. In cases where the insurance coverage is not clear, it is always best to check with the insurance provider to get a better understanding of the specific cost of the test.

If you are paying out of pocket, what is the cost for pharmacogenomic testing?

The cost can vary widely depending on the specific test being done and the laboratory that is conducting the test. Some pharmacogenomic tests can cost a few hundred dollars, while others can cost over a thousand dollars. Additionally, the cost of pharmacogenomic testing may depend on whether the test is being done as part of a research study or as a commercial test.

There are some pharmacogenomic testing companies that offer testing services at a lower cost using saliva sample or a cheek swab and provide the results online.

Is there federal legislation to support or compel insurance coverage for pharmacogenomics?

Currently, there is no federal legislation that specifically supports or compels insurance coverage for pharmacogenomic testing. However, there are a few laws and regulations that may impact coverage for pharmacogenomic testing:

1. The Affordable Care Act (ACA) requires that insurance plans cover certain preventive services, including some genetic tests, without any cost-sharing.

2. The Genetic Information Nondiscrimination Act (GINA) prohibits discrimination based on genetic information in employment and health insurance.

3. The 21st Century Cures Act requires the U.S. Food and Drug Administration (FDA) to establish a program to encourage the development of pharmacogenomic tests and to work with the Centers for Medicare & Medicaid Services (CMS) to ensure that these tests are covered by Medicare.

It's important to note that while these laws and regulations may impact coverage for pharmacogenomic testing, they do not specifically require insurance providers to cover pharmacogenomic tests.

Additionally, some states have legislation that affects pharmacogenomic testing, such as laws that require health insurers to cover certain types of genetic testing, but it's also important to check with the state's laws and regulations regarding pharmacogenomic testing.

To test or not to test?

There is an adage in medicine not to let perfection stand in the way of progress. Waiting for absolute proof that pharmacogenomic testing is effective when studies to date, including those for pain management, strongly indicate that it can be beneficial both clinically and economically, is a personal call between you and your doctor.

Even if your insurer does not cover the testing, the monetary savings can be substantial in the form of reduced medication, procedure, hospitalization and physician visit costs if your health status improves as a result. The quality of life benefits in the form of reduced pain and suffering, increased mobility and activity, of course, are priceless.

This is a conversation I urge you to have, and the purpose of this article is to provide sufficient information to have that conversation.

Want to know more about the science of pharmacogenomic testing? Here are some resources:

Practical Pain Management – The science of Pharmacogenomic Testing
Pharmacogenomics and Precision Medicine – Mayo Clinic - YouTube
Pharmacogenomic Testing Labs – Where to Find Them

Since medically retiring after 35 years in CEO roles in hospitals, medical device companies and value-based care providers, James Doulgeris stays active by advising healthcare companies and systems in the AI and analytics spaces. He also works to improve care and treatments for the rare disease community including the first initiative to use clinical and claims analytics to identify, diagnose and treat rare disease beginning with CRPS/RSD. As an award winning novelist for "The Dyodyne Experiment," Doulgeris is hard at work with his writing partner on its sequel, "Sentience." He is also a contributor writing articles and providing background and interviews on healthcare for national publications and news outlets.

rsdsa
SUPPORTING THE CRPS COMMUNITY

Learn More About RareMDx

RareMDx™ is a novel rare disease-focused, phenotype-driven differential diagnosis tool to transform and expedite the diagnostic process conducted by medical professionals. Based on their selection of signs/symptoms, RareMDx activates its probability algorithms and creates a list with suspected diagnoses of up to ten rare diseases in order of probability. The entire process, from selection to prediction is accomplished in a matter of minutes.

Knowing that patients and parents are very knowledgeable about an undiagnosed medical condition they are affected by, they've also created RareLook™, the layman's terms version of RareMDx. Both digital tools will become available to the rare disease community on Rare Disease Day, February 28. Please visit their website at raremd.com to learn more.

Access to RareLook is free. Please feel free to share this information on social media to increase the chance that others affected by an undiagnosed rare disease can benefit from this innovative development.

RSDSA's 4th Virtual CRPS Awareness Walk

The virtual RSDSA CRPS Awareness Walk is back for it's fourth straight year! From 2020 to now, we have increased our participation and beat our goal all three years. It's your participation and energy that keeps this walk growing. We are so grateful for the CRPS Warriors, caregivers, family, friends, and supporters who make it all possible!

We will open registration on **Wednesday, March 1st.** Be the first in your state to register and we will send you an Amazon gift card for $20. With your help, we will go bigger and better!

We've livestreamed the walk each year and we will do it again, so we look forward to "seeing" you on June 3rd. Although we wish we could see all CRPS Warriors in person, the benefit of our virtual walk is that you can have team members from across the country or even around the globe! This gives supporters the freedom to walk on their own schedule and still feel involved in such a wonderful experience.

Save The Date: 5th Long Island CRPS Awareness Walk & Expo

The 5th Long Island CRPS Awareness Walk & Expo is Saturday, September 9th from 8am – 2pm at Eisenhower Park in East Meadow, NY. For more information or Sponsorship Opportunities please email RSDSALongIsland@gmail.com

Tell Us What You Want to See on Facebook Live

What Facebook Live sessions are you interested in watching this year? Let us know by messaging us on Facebook or by emailing us at alexisdavis@rsds.org.

Create Content for RSDSA's TikTok Account

RSDSA has officially joined TikTok @RSDSAofficial in order to further educate others about CRPS in addition to our events and activities!

We want to ensure more people learn about CRPS, especially Gen Z (people born between 1997-2012) and Millennials (people born between 1981-1996) as they have a large footprint on the app in addition to being future + current medical leaders.

If you or your children, grandchildren, nieces/nephews, godchildren, etc. are interested in creating content for TikTok, please reach out to us at alexisdavis@rsds.org. And don't forget to tag us in any CRPS-related content you create!

We want your feedback!

Please send any suggestions or upcoming events of interest to our community to info@rsds.org and please consider donating at rsds.org/donate.

Thank you to our Title Sponsors

Our title sponsors make RSDSA events and awareness activities possible. Please join us in thanking and supporting them!

Abbott | The Baker Family Charitable Fund | Diana and Peter Smith in memory of Stephanie Theresa Smith | Dr. & Mrs. Lawrence and Judy Zager, in loving memory of Hunter Lia Zager Lynn & Michael Coatney | The Cochran Firm, National CRPS/RSD Lawyers