6

32

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CARDIOLOGY

14

2

Phosphorus
Diagnostics

Genomics, illuminated.

We focus on your peace of mind.

Our expert-guided assay designs, powerful data analytics, and rigorous variant interpretation ensure accurate results for your patients.

Each test is flexible, because each case is unique.

Our comprehensive and customizable panels are designed to handle the complexity of each patient's clinical history.

Simple, well-vetted patient reports are our forte.

We create reports you and your patients can understand, giving you the depth and simplicity you need to confidently use and communicate results.

Consider us another set of hands.

Our knowledgeable support team is here to help. From genetic counseling, to lab support, to facilitating family member testing, we want your workflow to be seamless.

We take care of billing so you don't have to.

Transparent prices and billing policies are our priority, so you can spend time with your patients – not insurance companies.

We're driven by a broader mission.

We're scientists at the core, and we want to work with you to discover the genetic basis of complex inherited disease.

Cardiomyopathies & Arrhythmias

A genetic test for a variety of inherited arrhythmias and cardiomyopathies that can lead to sudden cardiac death. The test includes customizable panels for:

- Hypertrophic cardiomyopathy (HCM)
- Dilated cardiomyopathy (DCM)
- Arrhythmogenic cardiomyopathy
- Left ventricular non-compaction cardiomyopathy (LVNC)
- Restrictive cardiomyopathy (RCM)
- Catecholaminergic polymorphic ventricular tachycardia (CPVT)
- Long QT Syndrome (LQTS)
- Short QT Syndrome (SQTS)
- Brugada Syndrome (BrS)

1 in 200

people have inherited cardiomyopathy or arrhythmia

133 GENES

Semsarian, C. et al. (2015). Journal of the American College of Cardiology, 65(12), 1249-1254.

Why Cardiogenetic Testing?

CONFIRMATION OF DIAGNOSIS

Heart conditions are often asymptomatic and can be missed using clinical criteria alone. Genetic testing can be used to reduce false positives and negatives and further refine etiology.

EARLY ID OF AFFECTED PATIENT & FAMILIES

Evidence shows that when disease is identified early and treated aggressively, morbidity and mortality can be reduced significantly. Cascade testing of family members helps identify those affected so they can take necessary proactive steps.

TREATMENT PLANNING

Identifying a mutation may guide treatment plans in some cases, which could include more frequent screening, an implanatable defibrillator (ICD) or pacemaker, medications specific to a patient's phenotype, and physical activity restrictions.

Familial Hypercholesterolemia

A genetic test for familial hypercholesterolemia (FH), a disorder associated with high cholesterol levels in the blood, which can lead to early-onset cardiovascular disease. Due to vast underdiagnosis in the population, less than 1% of affected individuals, many of whom are children, are aware of their condition.

Our test looks directly at the genes that control cholesterol levels to determine if they are functioning correctly or if FH-causing mutations are present. It covers four genes that account for the majority of FH diagnosis: *APOB*, *LDLR*, *LDLRAP1*, and *PCSK9*.

1 in 250

people is estimated to have familial hypercholesterolemia.

4 GENES

Mendelson, M. et al. (2015). Circulation, Vol. 123, Issue Supplement 3

Why Our Tests?

COMPREHENSIVE VARIANT DETECTION

Powered by full next-generation sequencing and deletion/duplication analysis for all genes. All variants are rigorously assessed by expert Ph.D. scientists, and all positive variants are confirmed.

CUSTOMIZABLE PANEL OPTIONS

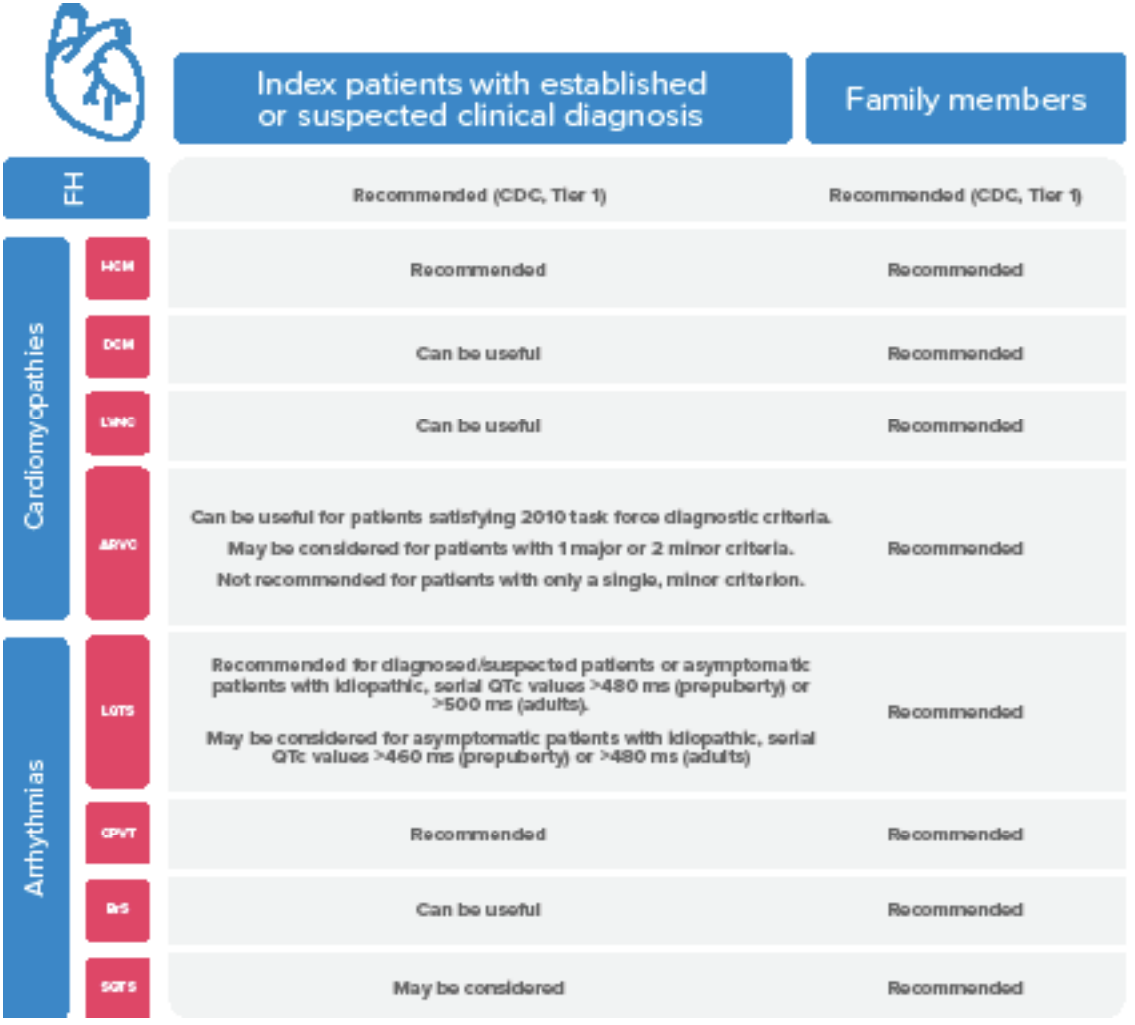
Select from expert curated gene panels or choose individual genes based on the unique clinical phenotype of your patient. Additional genes may be ordered up to 90 days after testing at no cost to the patient if an expanded disease panel is deemed clinically necessary.

RAPID TURNAROUND & SUPPORT

3-4 week turnaround from sample receipt in the lab to report delivery. Our knowledgeable client services and genetic counseling team is available via phone or email to answer questions from clinical staff or patients and their family members.

Supported by national medical guidelines

Prominent national societies have emphasized the importance of genetic testing for cardiomyopathies, arrhythmias, and familial hypercholesterolemia, including the Heart Rhythm Society (HRS), the European Heart Rhythm Association (EHRA), and the Centers for Disease Control (CDC) .



Ackerman, M. J., Priori, S. G., Willems, S., Berul, C., Brugada, R., Calkins, H., ... & Hershberger, R. E. (2011). HRS/EHRA expert consensus statement on the state of genetic testing for the channelopathies and cardiomyopathies. Europace, 13(8), 1077-1109.

Rigorous variant curation approach

MULTI-STEP VARIANT REVIEW

Phosphorus follows rigorous guidelines for variant curation. Interpretations are vetted through a multi-step quality control process that involves internal validations by Ph.D. scientists and external consultations with noted experts in cardiovascular genetics.

CONSCIENTIOUS VUS RESOLUTIONS

In the event a variant of unknown significance (VUS) is found in an index patient, Phosphorus may provide family member testing to better classify and resolve the mutation. If a VUS is reclassified, Phosphorus contacts relevant individuals to apprise them of the change in variant status.

ROBUST VARIANT DATABASE

The Phosphorus Variant Database is built on examination and interpretation of major online databases (OMIM, Clinvar, dbSNP, and other publicly available databases) and extensive literature review, as well as expert review and computational analysis of novel variants observed in patients.

Simple testing workflow

1

SAMPLE COLLECTION

Sample kits are available for both blood and saliva collection and are shipped directly to providers' offices. Patient's requisition form with selected test/disease panels and consent forms must be sent with the kit inside the Prepaid FedEx Clinical Pak (included in the kit).

2

DNA ANALYSIS

Patient DNA is extracted and processed using next-generation sequencing technology in our CLIA-certified lab. Data is analyzed using Phosphorus Elements proprietary bioinformatics software.

3

REPORTING

All variant interpretations are vetted through a rigorous quality control process that follows the latest ACMG guidelines and involves internal validations by Ph.D. scientists. Patient reports are sent back to providers via secure email or our online clinic portal.

4

GENETIC COUNSELING & FOLLOW-UP

Our board-certified genetic counselors assist in explanation of variants, genes, and conditions. By examining patients' medical histories in addition to test results, they help to facilitate the most informed actions for patients and at-risk family members.

Dedicated to advancing genetic research

We're scientists at the core. By leveraging the power of cutting-edge genomic technologies, collaborative data sharing, and exceptional relationships with partnering clinics and laboratories across the globe, we seek to improve understanding of Mendelian and complex diseases. Phosphorus strengthens this objective through additional IRB-approved studies, integrating genomic data with clinical outcomes to address complex biological questions and translating these results to personalized clinical care informed by each patient's DNA.

CARDIOGENETIC RESEARCH STUDIES

AGTC
TCAG

- Novel variant identification in cardiomyopathies, arrhythmias, and FH to establish novel genotype-phenotype associations and new clinical risk factors
- Detection of genetic factors associated with complex disorders such as atherosclerosis, coronary artery disease, atrial fibrillation, and other lipidemias

- Whole genome sequencing for sudden cardiac death
- Refinement of incidence rates by condition and within differing ethnic populations

Contact us to learn more about our research studies

Committed, responsive billing support

- Phosphorus offers comprehensive billing support modeled on responsible medical guidelines.
- We aim for price transparency so that patients, physicians, and clinics know ahead of time what they will pay.
- Multiple billing options are available, including insurance billing, and patient pay.
- Phosphorus accepts many commercial insurance plans and submits claims on your behalf as an out-of-network laboratory.

At Phosphorus, affordable testing is a priority. For specific cost estimates and billing assistance, please contact us at billing@phosphorus.com or call 855-746-7423. We are happy to help.

Ready to run this testing in your in-house lab? We can help!

CONTACT US TO LEARN MORE
support@phosphorus.com
1-855-746-7423

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Diagnostics

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