

# Advanced Cardio Genetic Testing

## NEXT GENERATION SEQUENCING (NGS)

Genetic variations have been described in association with cardiomyopathies, arrhythmias, and many cardiac disorders. The genetic cause of these heart disorders can now be determined and can aid in diagnosis and provide valuable information about prognosis and treatment.

### DIAGNOSIS

Observation of a known or expected pathogenic variant by genetic testing can solidify a clinical diagnosis and direct appropriate patient management and surveillance. Additionally, testing allows for screening of at-risk family members.

### PROGNOSIS

Genotype-phenotype relationships have been established in several forms of cardiac disease. Knowledge of the causative genetic variant can provide information around the natural history of the disease, response to pharmacotherapy, and circumstances that may trigger the condition.

### TREATMENT

Therapeutic options exist for many forms of cardiac disease. Treatment may include drugs or surgical interventions for optimal patient management. For example, use of beta blockers for management of patients with Long QT syndrome, or early, pre-emptive intervention with an implantable cardiac defibrillator may be advocated in patients with dilated cardiomyopathy. Combining NGS with pharmacogenetics may also afford significant insight into which drug(s) will metabolize most efficiently in the patient and provide precision care.



## ARRHYTHMIC DISORDERS/CHANNELOPATHIES

### BRUGADA

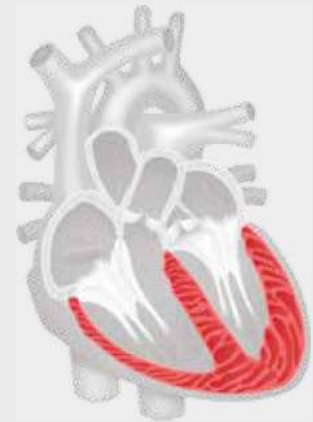
- Can be used in asymptomatic patient with typical ECG changes to identify those needing follow up
- Provides confirmation of diagnosis and possible prognostic information in those with clinical symptoms
- Identifies family members requiring further follow up

### SHORT QT SYNDROME

- Assists in risk stratification
- Assists in decision to place ICD
- Identifies family members requiring further workup

### LONG QT SYNDROME

- Provides confirmation of diagnosis in those with clinical signs/symptoms
- Predicts response/nonresponse to beta blockers
- Assists in decision to place ICD
- Identifies family members requiring further workup



### CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA (CPVT)

- Provides confirmation of diagnosis in patients with compatible clinical signs/symptoms
- Identifies family members requiring further workup
- Identifies infants at higher risk of SIDs who may need prophylactic treatment

## RIGHT VENTRICLE DISORDER

### ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY

- Confirms diagnosis in those with definite clinical syndrome
- May assist in diagnosis in suspected cases with multiple criteria
- Assists in identifying family members who need lifestyle advice and further follow up



## CORONARY VESSEL DISEASE

### FAMILIAL HYPERCHOLESTEROLEMIA

- Confirm diagnosis, pinpoints molecular mechanism of disease in patients with clinical syndrome or appropriate family history
- Directs therapeutic decision for use of novel, expensive, narrow therapeutic index drugs



## DISORDERS OF AORTA & AORTIC VALVES

### AORTIC DISEASES

- Confirms phenotypic diagnosis of Marfan's or other familial aortopathy
- Guides timing of surgical intervention
- Provides prognostic information
- Identifies family members who need imaging and followup

### NOONAN'S SYNDROME

Confirms diagnosis in those with typical phenotypical features

Differentiates Noonan's from other congenital diseases

Allows identification of subclinical cases among family members needing evaluation of heart and kidneys



## CARDIOMYOPATHIES

### HYPERTROPHIC CARDIOMYOPATHY

- Confirms clinical diagnosis
- Identifies those who could benefit from enzyme replacement (Fabry's disease) or early transplant (Danon disease)
- Identifies patients at high risk of arrhythmia despite mild hypertrophy
- Identifies family members needing further workup

### DILATED CARDIOMYOPATHY

May assist in diagnosis of those with dilated cardiomyopathy and significant conduction disorders

Aids in identifying patients at very high risk of sudden cardiac death

Identifies family members requiring further workup



MyGenetx also offers additional genetic testing to help identify risks related to cardiovascular disease.

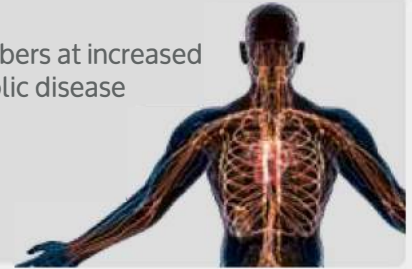
## VASCULAR

### HAPTOGLOBIN GENOTYPE

- Identifies those at increased risk for cardiovascular disease
- In diabetic patients, haptoglobin 2-2 genotype confers 2-10x risk for cardiovascular disease: treating these patients with vitamin E reduces their risk several fold
- **Haptoglobin testing is proprietary to MyGenetx**

### DVT/THROMBOPHILIA

- Identifies those patients with unexplained DVT who would benefit from extended or lifelong anticoagulation
- Identifies family members at increased risk for thromboembolic disease



## PHARMACOGENETICS

### PHARMACOGENETICS (PGt)

PGt genes are involved in the metabolism of many clinically important medications:

- Antiplatelets (clopidogrel)
- Beta Blockers (metoprolol, carvedilol)
- Antiarrhythmics (flecainide, propafenone)

Actionable reports offer timely guidance on patient treatment which can reduce secondary events.

### ANTICOAGULANT PANEL

- Identifies increased risk of hemorrhage for patients who cannot metabolize warfarin, which can account for 15% of severe adverse effects
- Minimizes the risk of bleeding and thrombotic complications

MyGenetx offers a full pharmacogenetic panel that tests over a dozen genes and their drug-to-gene interactions.

**THE RIGHT PATIENT. THE RIGHT DRUG. THE RIGHT DOSE.**

## SPECIMEN COLLECTION

NGS requires either oral saline rinse or traditional venipuncture (one purple-top, EDTA tube). All necessary materials for collecting and shipping are provided with the collection kit. The specimen is then shipped via UPS to our CLIA-certified molecular laboratory for analysis. Results are available within 10-14 days.

## ABOUT MYGENETX

MyGenetx is a CLIA-certified lab focused on molecular and advanced diagnostic testing. MyGenetx is the primary resource for transitioning and implementing precision-guided medicine. Our medical team consists of experts in research and clinical product development for genetics.

MyGenetx understands the importance of educating both the provider and patient. We are dedicated to providing relevant, up-to-date knowledge on the ever-changing world of genetic testing.