The Section of Pediatric Cardiology at Texas Children’s Hospital and Baylor College of Medicine has a well-established reputation for excellence in patient care. The John Welsh Cardiovascular Diagnostic Laboratory was established as part of our commitment to providing the highest quality of service and cutting-edge medical and scientific technology to our patients and the Cardiology/Medical community.

**GENETIC TESTING**

The John Welsh Cardiovascular Diagnostic Laboratory offers state-of-the-art genetic testing for DNA mutations associated with cardiac disease. All testing is performed using automated DNA sequencing of the coding region and splice junctions of each gene. A small blood sample is required for each test. Testing can also be performed on tissue samples. Many of our tests are the result of years of research performed in the Phoebe Willingham Muzzy Pediatric Molecular Cardiology Laboratory, which is the research branch of our laboratory. The close relationship between our research and diagnostic laboratories allows us to identify novel disease-associated genes and mutations in the research laboratory and transfer this knowledge directly to the patient through the diagnostic laboratory, allowing “bench-to-bedside” service.

**VIRAL PCR ANALYSIS**

The detection of viral genomic nucleic acid sequences by polymerase chain reaction (PCR) or reverse transcription (RT)-PCR provides a highly sensitive and specific method for the diagnosis of viral infection. We offer rapid molecular testing of multiple tissues, including heart, liver, lung, and brain tissue, and bodily fluids for viruses. Multiple DNA and RNA viruses can be detected in a single tissue sample as small as a needle biopsy or in less than 1cc of fluid. These tests are focused specifically for viruses identified as causing heart disease in children and adults, as well as in the fetus. Viral infection has been shown to cause myocarditis, dilated cardiomyopathy, endocardial fibroelastosis, and transplant rejection. Viral PCR analysis also has a broad range of other clinical applications.

**CONSULTING SERVICES**

The John Welsh Cardiovascular Diagnostic Laboratory also offers free medical consultation services via telephone or e-mail to any physician or genetic counselor. Our Medical Director, Dr. Daniel Penny, is the Chief of Pediatric Cardiology at Texas Children’s Hospital, and our Laboratory Director, Dr. Yuxin Fan is certified by the American Board of Medical Genetics in Clinical Molecular Genetics. Thus, we are able to offer expert insight into the clinical and molecular aspects of the diseases for which we offer testing. It is the goal of the John Welsh Cardiovascular Diagnostic Laboratory to not only offer the highest quality of molecular diagnostic testing possible but also to provide physicians and genetic counselors with the information that they need to provide their patients an optimal standard-of-care.
GENETIC TESTING

GENETIC TESTING

TESTS OFFERED

- Actin alpha 2 (ACTA2) – Thoracic Aortic Aneurysms and Aortic Dissections (AAT6)
- Calsequestrin 2 (CASQ2) – Catecholamine-Induced Polymorphic Ventricular Tachycardia
- Caveolin 3 (CAV3) – Caveolinopathies, Long QT Syndrome, Sudden Infant Death Syndrome
- Desmin (DES) – Cardiomyopathy, Distal Myopathy, Desmin-related Myopathy, DCM
- Emerin (EMD) – Emery-Dreifuss Muscular Dystrophy
- FBN1 – Marfan Syndrome, Ectopia Lentes, Aortic Aneurysm
- G4.5 (TAZ) – Barth Syndrome, DCM, LVNC, EFE
- KCNJ2 – Andersen-Tawil Syndrome, Long QT Syndrome
- Lamin A/C (LMNA) – Emery-Dreifuss Muscular Dystrophy, DCM, Cardiomyopathy with Conduction Defects, Limb-Girdle Muscular Dystrophy, Partial Lipodystrophy, Charcot-Marie-Tooth, Mandibuloacral Dysplasia, Hutchinson-Gilford Progeria Syndrome
- LAMP2 – Danon Disease, Hypertrophic/Dilated Cardiomyopathy with Skeletal Myopathy and Wolff-Parkinson-White
- MYH11 – Thoracic Aortic Aneurysms and Aortic Dissections (AAT4)
- NKX2.5 – Isolated Nonsyndromic Congenital Heart Disease/Defects
- SCO2 – HCM, Encephalopathy, COX Deficiency
- SURF1 – Leigh Syndrome, COX Deficiency
- TBX5 – Holt-Oram Syndrome
- TGFBR1 – Loeys-Dietz Syndrome, Thoracic Aortic Aneurysms and Aortic Dissections (AAT5), Furlong Syndrome, Marfan Syndrome
- TGFBR2 – Loeys-Dietz Syndrome, Thoracic Aortic Aneurysms and Aortic Dissections (AAT3), Marfan Syndrome
- ZASP/CYPHER/LDB3 – DCM, LVNC, Myofibrillar Myopathy

REASONS FOR REFERRAL

- Molecular Confirmation of disease diagnosis
- Family history of disease
- Carrier Testing – If appropriate
- Prenatal Diagnosis – Please call before sending specimens

SPECIMENS ACCEPTED

- Blood – Preferred
- Tissue – Multiple organ sources accepted

METHODOLGY

Genomic DNA is analyzed for mutations by automated fluorescent DNA sequencing of all coding exons, as well as the exon/intron junctions, in both the forward and reverse orientations

GENETIC TESTING

GENETIC TESTING

PANELS OFFERED

- Panel A: Marfan Syndrome and MFS-related Disorders ($4,400)
  - Fibrillin 1 (FBN1) Sequencing ($1650)
  - TGF-beta receptor 1 (TGFBR1) Sequencing ($600)
  - TGF-beta receptor 2 (TGFBR2) Sequencing ($600)
  - Actin alpha 2 (ACTA2) Sequencing ($750)
  - MYH11 Sequencing ($1500)
- Panel B: Congenital Heart Diseases (CHD) - Holt-Oram Syndrome, ASD, TOF, Heterotaxy, RVOT, TGA, DORV ($1,000)
  - TBX5 Sequencing ($750)
  - NKX2.5 Sequencing ($450)
- Panel C: Dilated Cardiomyopathy-DCM ($2,500)
  - Desmin (DES) Sequencing ($750)
  - Lamin A/C (LMNA) Sequencing ($750)
  - Tafazzin (TAZ, G4.5) Sequencing ($600)
  - LAMP2 Sequencing ($750)
  - ZASP/CYPHER/LDB3 Sequencing ($800)
- Panel D: Left Ventricular Noncompaction-LVNC ($1,850)
  - Tafazzin (TAZ, G4.5) Sequencing ($600)
  - ZASP/CYPHER/LDB3 Sequencing ($800)
  - Lamin A/C (LMNA) Sequencing ($750)
- Panel E: Muscular Dystrophies with Cardiomyopathy ($2,500)
  - Lamin A/C (LMNA) Sequencing ($750)
  - Caveolin 3 (CAV3) Sequencing ($450)
  - Emerin (EMD) Sequencing ($450)
  - Desmin (DES) Sequencing ($750)
- Panel F: Hypertrophic Cardiomyopathy-HCM ($2,500)
  - Caveolin 3 (CAV3) Sequencing ($450)
  - SCO2 Sequencing ($300)
  - SURF1 Sequencing ($750)
  - LAMP2 Sequencing ($750)
  - ZASP/CYPHER/LDB3 Sequencing ($800)
- Panel G: Long QT Syndrome-LQTS ($750)
  - KCNJ2 Sequencing ($450)
  - Caveolin 3 (CAV3) Sequencing ($450)
- Panel H: Other Arrhythmia Disorders ($1,600)
  - Calsequestrin 2 (CASQ2) Sequencing ($800)
  - Desmin (DES) Sequencing ($750)
  - KCNJ2 Sequencing ($450)
- Panel I: Sudden Infant Death Syndrome-SIDS ($450)
  - Caveolin 3 (CAV3) Sequencing ($450)
- Panel J: Atrioventricular Block ($2,000)
  - Lamin A/C (LMNA) Sequencing ($750)
  - Desmin (DES) Sequencing ($750)
  - Emerin (EMD) Sequencing ($450)
  - NKX2.5 Sequencing ($450)
TESTING INFORMATION
JOHN WELSH CARDIOVASCULAR
DIAGNOSTIC LABORATORY

VIRAL PCR ANALYSIS

TESTS OFFERED
- Adenovirus
- Enterovirus
- Parvovirus
- Influenza A Virus
- Respiratory Syncytial Virus
- Viral Panel (Adenovirus, CMV, EBV, Enterovirus, Parvovirus)

- Cytomegalovirus
- Epstein-Barr Virus
- Mumps Virus
- Rubella Virus

REASONS FOR REFERRAL
- Molecular confirmation of viral infection
- Routine surveillance of transplant patients

SPECIMENS ACCEPTED
- Tissue – Multiple Organ Sources Accepted
- Bodily Fluids
- Blood – Please Call

METHODOLOGY
- DNA and RNA are simultaneously isolated from the specimen, oligonucleotide primers are used to detect a portion of the genomic DNA or RNA of the virus using PCR or RT-PCR, respectively.

SERVICE FEES
- Single Virus - $370
- Viral Panel - $740
- Discounts offered for larger sample volumes

ESTIMATED TURNAROUND TIMES (TAT)

GENE SEQUENCING ANALYSIS:

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<tr>
<th>GENE</th>
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<tbody>
<tr>
<td>ACTA2</td>
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<tr>
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<td>TAZ (G4.5)</td>
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<tr>
<td>ZASP (LDB3)</td>
<td>4-6 Weeks</td>
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<tr>
<td>FBN1</td>
<td>6-8 Weeks</td>
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PRENATAL GENETIC TESTING: 2-4 Weeks

KNOWN FAMILIAL MUTATION: 1-2 Weeks

VIRAL TESTING: 1-2 Weeks

BILLING INFORMATION

ONE OF TWO FOLLOWING BILLING OPTIONS MUST BE INDICATED BELOW.
- The Self-Pay option must include payment with sample. If the billing information for referring institution is incomplete, the referring physician or laboratory will be automatically charged, or sample processing will not be initiated.

PAYMENT METHODS:
- SELF-PAY (PAYMENT-IN-FULL)
- Check or Money Order
- Credit Card (Please complete the Credit Card Authorization Form)

- REFERRING INSTITUTION/MD
- Please provide existing account number or call Julie at 832-824-4152 to establish one with us

SAMPLE SHIPPING INFORMATION

SEND SAMPLES MONDAY THRU THURSDAY
OVERNIGHT TO:
John Welsh Cardiovascular Diagnostic Laboratory
Baylor College of Medicine, Feigin Center
1102 Bates Avenue, Suite 480.02
Houston, TX 77030
Website: www.bcm.edu/pediatrics/welsh

CONTACT INFORMATION

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