Laboratory Support and FAQs

Genetic Services collaborates with laboratories throughout the implementation of molecular genetic testing by providing:

- Interpretive services through TeleGene™
- Access to experienced Geneticists and Genetic Counselors
- Genetics education
- Laboratory forms review and development
- Patient education materials
- Support for all CF testing platforms

Laboratory FAQs

- Pregnant women
- Reproductive partner of CF carrier
- Preconception couple
- Sperm or egg donors
- Prenatal diagnosis for CF carrier couple
- Male with congenital absence of the vas deferens
- Individual with a family history of CF
- Symptomatic individual
- Newborn screen positive individual
- Individuals with fetal echogenic bowel on ultrasound

The Access Genetics Molecular Information System, TeleGene™, is a secure web portal through which Genetic Services provides expert real-time interpretation of complex test results and customized patient-specific reports.

Interpretive services offered with molecular testing for:

- Cystic Fibrosis
- Inherited Thrombophilia (Factor V Leiden, Prothrombin and MTHFR)
- Other Molecular Tests

Interpretive services provide:

- Concise yet comprehensive, personalized reports
- Interpretive review by Board Certified Genetic Counselors
- Final consultative review and sign-out by a Board Certified Molecular Geneticist

Personalized Reports Include:

- Individualized risk assessments
- Clear and concise genotype results
- Clinical relevance of the genotype
- Recommendations for further assessment when appropriate
- Implications for family members

To arrange Genetic Services for your practice or laboratory

CONTACT:
Toll Free: 888-250-4407
Email: salesandmarketing@access-genetics.com
Access Genetics Genetic Services

Access Genetics provides web-based software, laboratory materials, and networked expertise working seamlessly to improve molecular diagnostics and patient care. Genetic Services adds confidence in molecular genetic test results and provides decision support by:

- Supporting healthcare providers
- Offering patient support through Genetic Counseling
- Collaborating with partner clinicians & laboratories
- Providing personalized interpretive services for:
  - Cystic Fibrosis
  - Inherited Thrombophilia
  - Other Molecular Tests
- Genetic Services are provided by a network of:
  - Molecular Geneticists Board Certified by the American Board of Medical Genetics
  - Genetic Counselors Board Certified by the American Board of Genetic Counseling

Provider Support and FAQs

Genetic Services is a resource for healthcare providers to:

- Refer patients for Genetic Counseling
- Discuss a test result or testing requirements
- Seek case consultation regarding a patient history
- Provide community resources for patients
- Stay informed of developments in genetics

Provider FAQs

- **Will you speak to my patient about this test result?**
  - With your authorization, Genetic Services works directly with your patients. Call 1-888-250-4407 to arrange Genetic Counseling

- **Who should be offered Cystic Fibrosis (CF) carrier screening?**
  - In April 2011, The American College of Obstetricians and Gynecologists (ACOG) issued updated recommendations (Number 486) that CF carrier screening be offered to all couples seeking prenatal or preconception care with appropriate genetic counseling, education, and follow up. These recommendations correspond with the recommendations of the American College of Medical Genetics (ACMG).

- **What are the most recent treatment guidelines for Inherited Thrombophilia?**
  - Treatment guidelines are published biannually in the Journal Chest.
  - References available through Genetic Services

- **Can you provide carrier frequencies for Cystic Fibrosis?**
  - Carrier frequencies vary by ethnicity (see below).

- **What are the detection rates for the ACOG 23 mutation panel?**
  - Detection rates vary by ethnicity (see below).

<table>
<thead>
<tr>
<th>Ethnic Group</th>
<th>Detection Rate</th>
<th>Carrier Risk Before Testing</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ashkenazi Jewish</td>
<td>94%</td>
<td>1/24</td>
</tr>
<tr>
<td>Non-Hispanic Caucasian</td>
<td>88%</td>
<td>1/25</td>
</tr>
<tr>
<td>Hispanic American</td>
<td>72%</td>
<td>1/58</td>
</tr>
<tr>
<td>African American</td>
<td>64%</td>
<td>1/61</td>
</tr>
<tr>
<td>Asian American</td>
<td>49%</td>
<td>1/94</td>
</tr>
</tbody>
</table>
Access Genetics provides web-based software, laboratory materials, and networked expertise working seamlessly to improve molecular diagnostics and patient care. Genetic Services adds confidence in molecular genetic test results and provides decision support by:

- Cystic Fibrosis
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- Other Molecular Tests
- Molecular Geneticists Board Certified by the American Board of Medical Genetics
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**Patient Support and FAQs**

Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease (National Society of Genetic Counselors, 2005). Genetic Services offers Genetic Counseling to physician referred patients.

- **Genetic Counseling Services Provided by:**
  - Board Certified Genetic Counselors
- **Genetic Counseling Includes:**
  - Analysis of Family History
  - Review of Health History
  - Patient Education
  - Individualized Risk Assessment
  - Discussion of Informed Consent
  - Community Resource Referrals
- **Available through**
  - Phone Counseling
  - Access TeleGene™ Video Counseling (Coming soon)

**Patient FAQs**

- **My partner and I are Cystic Fibrosis (CF) carriers. What is our chance of having a child with CF?**
  - CF is autosomal recessive. When both parents are CF carriers, there is a 25% chance with each pregnancy to have an affected child.

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**Ethnic Group**

- **Ashkenazi Jewish**
  - Detection Rate: 1/24
  - Carrier Risk Before Testing: 1/25
- **Non-Hispanic Caucasian**
  - Detection Rate: 1/58
  - Carrier Risk Before Testing: 1/61
- **Hispanic American**
  - Detection Rate: 1/94
  - Carrier Risk Before Testing: 1/94
- **African American**
  - Detection Rate: 1/25
  - Carrier Risk Before Testing: 1/25
- **Asian American**
  - Detection Rate: 1/24
  - Carrier Risk Before Testing: 1/24

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**Providers, please CONTACT a Genetic Counselor or arrange Genetic Counseling, by contacting Genetic Services at:**

Toll Free: 888-250-4407
Direct: 952-400-7763
Email: geneticservices@access-genetics.com
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- **What proportions of Inherited Thrombophilia are accounted for by Factor V Leiden and Prothrombin mutations?**
  - These two tests diagnose up to 97% of patients with Inherited Thrombophilia (see below)

<table>
<thead>
<tr>
<th>Name</th>
<th>Percent Explained</th>
</tr>
</thead>
<tbody>
<tr>
<td>Factor V Leiden (FVL)</td>
<td>65 - 75%</td>
</tr>
<tr>
<td>Prothrombin G20210A</td>
<td>20 - 22%</td>
</tr>
</tbody>
</table>

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Genetic Services Offers...

- Genetic Counseling directly to patients
- Genetic Counseling provided by ABGC Board Certified Genetic Counselors
- Analysis of family history and individualized risk assessments
- Consultations for complicated patient histories
- Personalized interpretive services for molecular genetic testing
- Support to healthcare providers through the process of molecular genetic testing
- Collaboration with clinical laboratories on molecular genetic testing
- Education to help you stay informed of developments in genetics
- Direct access to Board Certified Molecular Geneticists and Genetic Counselors
- Expert, real-time interpretations of genetic test results
- Customized patient specific reports
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