Exclusive Opportunity

Diagnostic for Fragile-X Syndrome
Emory Tech ID: 05027

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Licensing Associate
Fragile X Syndrome

- Most common inherited cause of mental impairment and autism
- Prevalence: 1 in 4000 males and 1 in 6000 females, indiscriminately.
- Onset: 20 months to life.
- Diagnosis: Voluntary PCR test
- Treatment: Symptoms only.
## Fragile X: Symptoms

### Cognitive Impairment
- learning disabilities
- mental retardation

### Behavioral Anomalies
- ADD
- Hyperactivity
- anxiety and unstable mood
- autistic behaviors

### Physical Effects
- elongated face
- prominent jaw
- large ears
- flat feet
- hyper extensible joints
- Seizures (epilepsy)
- Premature Ovarian failure
Fragile X: FMR1 Gene

Mutation in the Fragile X mental retardation 1 (FMR1) gene

<table>
<thead>
<tr>
<th>Men</th>
<th>Women</th>
</tr>
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<tbody>
<tr>
<td>1/800</td>
<td>1/260</td>
</tr>
<tr>
<td>1/4000</td>
<td>1/6000</td>
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</tbody>
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Technology

Diagnostic and Population Screen

1. Tests for Hypermethylation FMR1

2. CGG convert to UGG
   • Methylated C is protected

3. Measure Uracil.

Blood Sample: 88 males, 2 Known Fragile X Positive
Fragile-X Diagnostic

Pooled Sample: One Test

Methylated FMR1 DNA

1 mosaic FX male + 95 normal male pool

96 normal male pool
## Advantages: Cost and Time

<table>
<thead>
<tr>
<th>Unmet Need</th>
<th>Fragile X</th>
<th>Diagnostic</th>
<th>Next Steps</th>
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<tbody>
<tr>
<td>Current Tests</td>
<td>Emory Diagnostic</td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Cost</strong></td>
<td>$300-400</td>
<td>$5</td>
<td></td>
</tr>
<tr>
<td><strong>Time</strong></td>
<td>7-14 Days</td>
<td>Hours</td>
<td></td>
</tr>
<tr>
<td><strong>Patients per Test</strong></td>
<td>1</td>
<td>96 or 384</td>
<td></td>
</tr>
<tr>
<td><strong>Other Diseases?</strong></td>
<td>No</td>
<td>Autism, Prader-Willi, Klinefelter, Aneuploidies</td>
<td></td>
</tr>
<tr>
<td><strong>Patients per Test</strong></td>
<td>1</td>
<td>96, 384, etc</td>
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# Further Development

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## Current Activities

1. Scale up proof of principle: 100,000 Samples from Georgia State Labs
2. Positioning for mandatory newborn screens: 4,000,000 screens per year

## Next Steps

1. Assess compatibility with therapeutic strategy
   - **Rationale:** Reduced costs of patient recruitment.
   - **Current Costs:** $300 X 4000 = $1,200,000/patient
2. Develop Genetic Counseling and Therapy Protocols

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12/8/2006