TESTING FOR FRAGILE X SYNDROME: 
NATIONAL FRAGILE X FOUNDATION GUIDELINES

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National Fragile X Foundation

THE FRAGILE X DNA TEST

Until 1991, the only laboratory test for fragile X syndrome was a chromosome test. In 1991 a DNA test, the FMR-1 gene test, was introduced. This test is the most accurate test (99+%)) for detecting fragile X syndrome. The chromosome test is still available through most labs and is used for a variety of diagnostic purposes. In some cases, your physician may want to have both the DNA test and the chromosome test performed. However, if you are testing specifically for fragile X syndrome, please be certain that your physician is requesting the FMR-1 gene test. Below are the differences between the two tests:

<table>
<thead>
<tr>
<th>DNA Test</th>
<th>Chromosome Test</th>
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</thead>
<tbody>
<tr>
<td>Name: FMR-1 gene test</td>
<td>Name: chromosome analysis</td>
</tr>
<tr>
<td>Sample in: EDTA lavender top tube</td>
<td>Sample in: Sodium-Heparin green top tube</td>
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<tr>
<td>Cost: $250-325</td>
<td>Cost: $500-700</td>
</tr>
<tr>
<td>Turnaround time: about 2 weeks</td>
<td>Turnaround time: about 1 month</td>
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</table>

For prenatal testing for fragile X syndrome, the labs that we suggest are indicated below with an asterisk. Please contact the labs directly for special handling instructions before collecting a sample to be used for prenatal testing.

PAYING FOR THE TEST

When testing for fragile X syndrome, you have two options for payment:

1. You pay for the test: Most laboratories will charge between $250-325 for fragile X testing. If you opt to pay for the test yourself (out-of-pocket), we suggest that you use one of the labs listed below. All labs require a referral from a physician in order to do testing. Please have your physician’s office contact the lab at the number listed to obtain further information regarding referral forms, exact cost of the test, and payment.

2. Your insurance pays for the test: In order to have your insurance cover the cost of your test for fragile X syndrome, you must have the test performed at a laboratory that is approved by your insurance company. Contact your insurance company directly and ask for the list of labs that they are contracted with for fragile X testing. The labs that you have access to may or may not be listed below. If any are listed below, request that your physician send your sample to that laboratory. Please either contact the lab directly or have your physician’s office contact the lab at the number listed to obtain further information regarding referral forms.

If none of your labs are listed below, you have two options. You may decide to pay for the cost of the test yourself and use one of the labs listed (see section above entitled You pay for the test). Your other option is to have your insurance cover the cost of the test, meaning that you use one of the labs from their list.
FRAGILE X SYNDROME TESTING LABORATORIES

The National Fragile X Foundation has compiled the following list of laboratories which we suggest using when testing for fragile X syndrome using the FMR-1 gene test. This list does not include every laboratory that offers FMR-1 gene testing, however these are the facilities with which we are most acquainted. **Using a lab that is not listed below does not mean that your results will not be accurate.** It simply means that we may not be familiar with that particular laboratory or their process of testing. Please call the National Fragile X Foundation at (800) 688-8765 with any questions.

Kimball Genetics
101 University Boulevard #350, Denver, CO 80206
Contact: Mary Linden or Juli Murphy (303) 320-1807
**prenatal testing (samples can include: amniotic fluid, chorionic villus, PUBS)**

Institute of Basic Research
Department of Genetics, 1050 Forest Hill Road, Staten Island, NY 11050
Contact: Sally Nolin (718) 494-5293
**prenatal testing (samples can include: amniotic fluid, chorionic villus)**

Genetics & IVF
3020 Javier Road, Fairfax, VA 22031
Contact: Ann Maddalena (800) 552-4363
**prenatal testing (samples can include: amniotic fluid, chorionic villus, PUBS)**

Mayo Medical Laboratories
200 1st Street SW, Rochester, MN 55905
Contact: (800) 533-1710
**prenatal testing (samples can include: amniotic fluid, chorionic villus, PUBS)**

Baylor DNA Diagnostic Laboratory
One Baylor Plaza, Room T536, Houston, TX 77030
Contact: (800) 226-3624
**prenatal testing (samples can include: amniotic fluid)**

Genzyme Genetics
1 Mountain Road, Framingham, MA 01710
Contact: Genetic Coordinator (800) 255-7357
**prenatal testing (samples can include: amniotic fluid)**

Center for Human and Molecular Genetics—UMDNJ
185 South Orange St #MSB-621, Newark, NJ 07103-2499
Contact: James Dermody (973) 972-3170

DIANON Systems
200 Watson Boulevard, Stratford, CT 06615
Contact: Amy Wirtheim (800) 328-2666 ext. 4547
**prenatal testing (samples can include: amniotic fluid, chorionic villus)**

Athena Diagnostics
Four Biotech Park, 377 Plantation Street, Worcester, MA 01605
Contact: (800) 394-4493

Nichols Institute/Quest Diagnostics
33608 Ortega Highway, San Juan Capistrano, CA 92575
Contact: (800) 642-4657