

# **TESTING FOR FRAGILE X SYNDROME: NATIONAL FRAGILE X FOUNDATION GUIDELINES**

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## **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:

### **DNA Test**

Name: FMR-1 gene test  
Sample in: EDTA lavender top tube  
Cost: \$250-325  
Turnaround time: about 2 weeks

### **Chromosome Test**

Name: chromosome analysis  
Sample in: Sodium-Heparin green top tube  
Cost: \$500-700  
Turnaround time: about 1 month

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 1<sup>st</sup> 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