

U.S. Fanconi Anemia Testing Resources

U.S. Fanconi Anemia Testing Resources

Institution	Test
<p>Cincinnati Children’s Hospital Medical Center Fanconi Anemia Comprehensive Care Center Bone Marrow Failure Clinic Cincinnati, OH</p> <p>Erica L. Goodridge, RN, BSN, CPN, Care Manager, Fanconi Anemia Comprehensive Care Center 513-636-3218 Toll-free: 1-800-344-2462, ext. 3218 <i>erica.goodridge@cchmc.org</i></p>	<p>DEB/MMC diagnostic testing using blood or bone marrow samples.</p> <p>Complementation analysis for <i>A, B, C, E, F, G, L, I</i> (<i>I</i> analysis via skin testing only) and <i>FANCD2</i> (via western blot). Testing done with blood, skin biopsy or amniocytes. Note: This testing must be requested by clinicians or researchers.</p> <p>Molecular testing to find specific mutations for patients with <i>FANCA, FANCC,</i> or <i>FANCG</i>.</p> <p>Prenatal diagnostic molecular testing for <i>FANCA, FANCC,</i> or <i>FANCG</i>.</p> <p>Carrier detection testing for patients with a family history of FA, from complementation groups <i>FANCA, FANCC,</i> or <i>FANCG</i>.</p>
<p>Dana-Farber Cancer Institute Boston, MA</p> <p>Alan D’Andrea, MD 617-632-2080 <i>alan_dandrea@dfci.harvard.edu</i></p> <p>Lisa Moreau, MS 617-632-6302 <i>lisa_moreau@dfci.harvard.edu</i></p>	<p>DEB/MMC diagnostic testing using blood, bone marrow, or skin fibroblasts.</p> <p><i>FANCD2</i> western blot (Research).</p> <p>FA carrier testing.</p>
<p>Indiana University School of Medicine Wells Center, Riley Hospital for Children Indianapolis, IN</p> <p>Helmut Hanenberg, MD 317-278-9290 or 274-2980 Fax 317-274-2852 <i>hhanenbe@iupui.edu</i></p>	<p>Complementation Group (Research): Use of retroviruses for <i>A, B, C, DI, D2, E, F, G, I, J, L, M, N, O, P, Q</i> in primary T-cells, bone marrow cells, or fibroblast cells or in lymphoblast cell lines (LCL) (Research).</p> <p><i>FANCD2</i> western blotting (research).</p>
<p>Indiana University School of Medicine Cytogenetic Lab, Dept. of Medical & Molecular Genetics Indianapolis, IN</p> <p>Gail H. Vance, MD 317-274-2243/317-278-0172 Fax 317-278-1616 <i>ghvance@iupui.edu</i></p>	<p>Diagnostic testing: Cytogenetic breakage studies with DEB on peripheral blood and fibroblast specimens.</p> <p>Mutation testing for <i>FANCA, FANCC,</i> and <i>FANCG</i>. Collaborative mutation testing and research with Gail Vance, MD, and Shaochun Bai, PhD, Medical & Molecular Genetics, Indianapolis, IN.</p>

Institution	Test
<p>Oregon Health & Science University Portland, OR</p> <p>Susan Olson, PhD, FACMG (cytogenetics) 503-494-5964 <i>olonsu@ohsu.edu</i></p> <p>C. Sue Richards, PhD, FACMG (molecular) 503-494-5400 <i>richarsu@ohsu.edu</i></p>	<p>DEB/MMC diagnostic testing - blood and skin fibroblasts.</p> <p>Next-Generation sequencing comprehensive Fanconi panel is now available for all <i>FANC</i> genes. Exon-centric microarray is also available for detection of deletions and duplications in all <i>FANC</i> genes.</p> <p>Complementation and full gene mutation analysis by Sanger sequencing for <i>A, C, G, E, F</i> as well as targeted sequencing of any exon is available. Deletion/duplication analysis for <i>FANCA</i> is also available by MLPA. Prenatal testing available for known mutations. Custom sequencing for known mutations in Fanconi genes other than <i>A, C, G, E, F</i> is also available. Please see GeneTests for further information.</p> <p>FA carrier testing.</p>
<p>Prevention Genetics 3700 Downwind Drive Marshfield, WI 54449</p> <p>Michael Chicka, PhD 715-387-0484 <i>michael.chicka@preventiongenetics.com</i></p>	<p>FA gene sequencing and deletion/duplication analysis for molecular diagnosis. Genes analyzed (at press): <i>FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCI/BRIP1, FANCL, FANCM, FANCN/PALB2, FANCO/RAD51C, FANCP/SLX4, FANCO/ERCC4</i></p> <p>Tests offered include:</p> <ul style="list-style-type: none"> • Next-Generation and Sanger Sequencing FA Panels for identifying the particular FA gene and variant(s) of interest; • Sanger sequencing of individual FA genes when gene of interest is known; • Sanger confirmation of all pathogenic variants and variants of unknown significance found by Next-Generation Sequencing; • Detection of large deletions/duplications using custom designed, high-density, gene-centric array CGH technologies; • Targeted variant analysis for familial variants and for confirming research findings; • Prenatal testing.
<p>Quest Diagnostics Chantilly, VA</p> <p>Dr. Steven Schonberg 800-336-3718 x7285</p> <p>Quest Diagnostics San Juan Capistrano, CA</p> <p>Joy Redman, MS, MBA, CGC 800-642-4657, ext. 4279 <i>Joy.B.Redman@questdiagnostics.com</i></p>	<p>DEB diagnostic testing performed on blood, bone marrow, and skin fibroblasts (in addition to prenatal testing): the doctor needs to specify DEB test, otherwise it is possible that a DNA carrier test for <i>FANCC</i> may be done.</p> <p>Mutation analysis for <i>FANCC</i> (IVS4+4A>T and 322delG).</p> <p>Prenatal diagnosis.</p>

U.S. Fanconi Anemia Testing Resources

Institution	Test
<p>University of Minnesota Physicians Outreach Laboratories Minneapolis MN</p> <p>Bharat Thyagarajan, MD, PhD, Director Matt Bower, MS, CGC, Contact 612-624-8948 <i>mbower1@fairview.org</i></p>	<p>Next Generation sequencing. These methods will detect point mutations and small indel mutations (<18bp).</p> <p>Copy number analysis for large deletions/duplications is under development and will be available in 2015.</p> <p>Targeted mutation analysis available in families with known mutations. Lab requires a positive control to develop primers.</p> <p>Complementation and analyze genes: <i>FANCA, FANCB, FANCC, FANCD1, FANCD2, FANCE, FANCF, FANCG, FANCI, FANJ (BRIP1), FANCL, FANCM, FANCN (PALB2), FANCO(RAD51C), FANCP(SLX4), FANQ (ERCC4)</i>.</p>

Please note:

- The laboratories listed are CLIA (Clinical Laboratories Improvement Amendments)-certified.
- Estimated costs for diagnostic tests range from approximately \$1,000 - \$1,200. Check with your insurance company for coverage. The average turnaround time for testing results is about 10 working days. For more information, please contact the labs directly.
- Additional testing information for Fanconi anemia is available at: <http://geneclinics.org>.
 - Select “Gene Reviews”.
 - Select “Search by Disease”.
 - Enter “Fanconi anemia” into the text box.
- “Research” indicates that the results will be used for research purposes only and may not be shared with referring physician/family.