

US Fanconi Anemia Testing Resources

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

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Oregon Health & Science University Portland, OR • Susan Olson, PhD, FACMG (cytogenics) 503-494-5964 <i>olsonsu@ohsu.edu</i> • Markus Grompe, MD (complementation) 503-494-6888 <i>grompem@ohsu.edu</i> • C. Sue Richards, PhD, FACMG (molecular) 503-494-5400 <i>richarsu@ohsu.edu</i>	DEB/MMC diagnostic testing
	Complementation group (Research)
	Mutation Analysis for A, C, G, E, F (Please see GeneTests for further information at geneclinics.org)
Quest Diagnostics Chantilly, VA • Aimee Martin, MGC, CGC 800-336-3718, ext. 7148 <i>aimee.d.martin@questdiagnostics.com</i> • Dr. Steven Schonberg 800-336-3718 x7285	DEB diagnostic testing: the doctor needs to specify DEB test, otherwise it is possible that a DNA carrier test for FANCC may be done.
	Mutation analysis for FANCC (IVS4+4A>T)
	Prenatal diagnosis
Quest Diagnostics, San Juan Capistrano, CA • Joy Redman, MS, MBA, CGC 800-642-4657, ext. 4279 <i>Joy.B.Redman@questdiagnostics.com</i>	Mutation analysis for FANCC (IVS4+4A>T and 322delG)

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>
 - 1) Select "Gene Reviews"
 - 2) Select "Search by Disease"
 - 3) Enter "Fanconi anemia" into the text box

