U.S. Fanconi Anemia Testing Resources

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Institution	Test
Cincinnati Children's Hospital Medical Center **Fanconi Anemia Comprehensive Care Center Bone Marrow Failure Clinic** Cincinnati, OH **Erica L. Goodridge**, **RN**, **BSN**, **CPN**, Care Manager, Fanconi Anemia Comprehensive Care Center 513-636-3218 Toll-free: 1-800-344-2462, ext. 3218 ***erica.goodridge@cchmc.org***	DEB/MMC diagnostic testing using blood or bone marrow samples. Complementation analysis for *A, B, C, E, F, G, L, I* (*I* analysis via skin testing only) and FANCD2 (via western blot). Testing done with blood, skin biopsy or amniocytes. Note: This testing must be requested by clinicians or researchers. Molecular testing to find specific mutations for patients with *FANCA, FANCC,* or *FANCG*. Prenatal diagnostic molecular testing for *FANCA, FANCC,* or *FANCG*. Carrier detection testing for patients with a family history of FA, from complementation groups *FANCA, FANCC,* or *FANCG*.
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. FANCD2 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 *A, B, C, D1, D2, E, F, G, I, J, L, M, N, O, P,* Q in primary T-cells, bone marrow cells, or fibroblast cell lines or in lymphoblast cell lines (LCL) (Research). FANCD2 western blotting (research).
Indiana University School of Medicine Cytogenetic Lab, **Dept. of Medical & Molecular Genetics** Indianapolis, IN **Gail H. Vance**, **MD** 317-274-2243/317-278-0172 Fax 317-278-1616 ***ghvance@iupui.edu***	Diagnostic testing: Cytogenetic breakage studies with DEB on peripheral blood. Mutation testing for *FANCA, FANCC,* and *FANCG*. Collaborative mutation testing and research with Gail Vance, MD, and Shaochun Bai, PhD, Medical & Molecular Genetics, Indianapolis, IN.

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Oregon Health & Science University Portland, OR **Susan Olson**, **PhD**, **FACMG** (cytogenics) 503-494-5964 ***olsonsu@ohsu.edu*** **Markus Grompe**, **MD** (complementation) 503-494-6888 ***grompem@ohsu.edu*** **C. Sue Richards**, **PhD**, **FACMG** (molecular) 503-494-5400 ***richarsu@ohsu.edu***	DEB/MMC diagnostic testing Next-Generation sequencing comprehensive Fanconi panel is now available for all *FANC* genes. Exon-centric microarray is also available for detection of deletions and duplications in all FANC genes. Complementation and full gene mutation analysis by Sanger sequencing for *A*, *C*, *G*, *E*, *F* as well as targeted sequencing of any exon is available. Deletion/ duplication analysis for *FANCA* is also available by MLPA. Prenatal testing available for known mutations. Custom sequencing for known mutations in Fanconi genes other than *A*, *C*, *G*, *E*, *F* is also available. (Please see GeneTests for further information.)
Prevention Genetics 3700 Downwind Drive Marshfield, WI 54449 **Michael Chicka**, **PhD** 715-387-0484 ***michael.chicka@preventiongenetics.com***	FA gene sequencing and deletion/duplication analysis for molecular diagnosis. Genes analyzed (at press): *FANCA*, *FANCB*, *FANCC*, *FANCD2*, *FANCE*, *FANCF*, *FANCG*, *FANCI*, *FANCJ*/*BRIP1*, *FANCL*, *FANCM*, *FANCN*/*PALB2*, *FANCO*/*RAD51C*, *FANCP*/*SLX4*, *FANCQ*/*ERCC4* Tests offered include: • Sanger sequencing of individual FA genes • Next-Generation and Sanger Sequencing FA Panels for detecting point mutations and varying sizes of deletions/duplications with 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
Quest Diagnostics Chantilly, VA **Dr. Steven Schonberg** 800-336-3718 x7285 **Quest Diagnostics** San Juan Capistrano, CA **Joy Redman**, **MS**, **MBA**, **CGC** 800-642-4657, ext. 4279 ***Joy.B.Redman@questdiagnostics.com***	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 *FANCC* may be done**. Mutation analysis for *FANCC* (IVS4+4A>T and 322delG) Prenatal diagnosis

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University of Minnesota Physicians Outreach Laboratories Minneapolis MN **Bharat Thyagarajan**, **MD**, **PhD**, Director **Matt Bower**, **MS**, **CGC**, Contact 612-624-8948 ***mbower1@fairview.org***	Next Generation sequencing. These methods will detect point mutations and small indel mutations (<18bp). Copy number analysis for large deletions/duplications is under development and will be available in 2014. Targeted mutation analysis available in families with known mutations. Lab requires a positive control to develop primers. Complementation and analyze genes: *FANCA*, *FANCB*, *FANCC*, *FANCD1*, *FANCD2*, *FANCE*, *FANCF*, *FANCG*, *FANCI*, *FANJ* (*BRIP1*), *FANCL*, *FANCM*, *FANCN* (*PALB2*), *FANCO*(*RAD51C*), *FANCP*(*SLX4*). Analysis of *FANQ* (*ERCC4*) will be available by summer 2014.

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.