U.S. Fanconi Anemia Testing Resources
## U.S. Fanconi Anemia Testing Resources

<table>
<thead>
<tr>
<th>Institution</th>
<th>Test</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Cincinnati Children’s Hospital Medical Center</strong>&lt;br&gt;Fanconi Anemia Comprehensive Care Center Bone Marrow Failure Clinic&lt;br&gt;Cincinnati, OH</td>
<td>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.</td>
</tr>
<tr>
<td><strong>Dana-Farber Cancer Institute</strong>&lt;br&gt;Boston, MA</td>
<td>DEB/MMC diagnostic testing using blood, bone marrow, or skin fibroblasts. FANCD2 western blot (Research). FA carrier testing.</td>
</tr>
<tr>
<td><strong>Indiana University School of Medicine</strong>&lt;br&gt;Wells Center, Riley Hospital for Children&lt;br&gt;Indianapolis, IN</td>
<td>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 cells or in lymphoblast cell lines (LCL) (Research). FANCD2 western blotting (research).</td>
</tr>
<tr>
<td><strong>Indiana University School of Medicine</strong>&lt;br&gt;Cytogenetic Lab, Dept. of Medical &amp; Molecular Genetics&lt;br&gt;Indianapolis, IN</td>
<td>Diagnostic testing: Cytogenetic breakage studies with DEB on peripheral blood and fibroblast specimens. Mutation testing for FANCA, FANCC, and FANCG. Collaborative mutation testing and research with Gail Vance, MD, and Shaochun Bai, PhD, Medical &amp; Molecular Genetics, Indianapolis, IN.</td>
</tr>
<tr>
<td>Institution</td>
<td>Test</td>
</tr>
<tr>
<td>-------------------------------------</td>
<td>-------------------------------------------------------------------------------------------------------------------------------------</td>
</tr>
</tbody>
</table>
| **Oregon Health & Science University** Portland, OR  
Susan Olson, PhD, FACMG (cytogenics)  
503-494-5964  
olsonsu@ohsu.edu  
C. Sue Richards, PhD, FACMG (molecular)  
503-494-5400  
richarsu@ohsu.edu | DEB/MMC diagnostic testing - blood and skin fibroblasts.  
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.  
FA carrier testing. |
| **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, FANCI/J/BRIP1, FANCL, FANCM, FANCN/PALB2, FANCO/RAD51C, FANCP/SLX4, FANCQ/ERCC4  
Tests offered include:  
• 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. |
| **Quest Diagnostics**  
Chantilly, VA  
Dr. Steven Schonberg  
800-336-3718 x7285 | 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. |
| **Quest Diagnostics**  
San Juan Capistrano, CA  
Joy Redman, MS, MBA, CGC  
800-642-4657, ext. 4279  
Joy.B.Redman@questdiagnostics.com |
<table>
<thead>
<tr>
<th>Institution</th>
<th>Test</th>
</tr>
</thead>
<tbody>
<tr>
<td>University of Minnesota Physicians Outreach Laboratories Minneapolis MN</td>
<td>Next Generation sequencing. These methods will detect point mutations and small indel mutations (&lt;18bp). Copy number analysis for large deletions/duplications is under development and will be available in 2015. 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), FANQ (ERCC4).</td>
</tr>
<tr>
<td>Bharat Thyagarajan, MD, PhD, Director Matt Bower, MS, CGC, Contact 612-624-8948 <a href="mailto:mbower1@fairview.org">mbower1@fairview.org</a></td>
<td></td>
</tr>
</tbody>
</table>

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](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.