International Fanconi Anemia Testing Resources

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Country	Institution	Test
Australia	**Children's Hospital at Westmead** Sydney, Australia **Zhan he Wu**, **MD, PhD, FFSc (RCPA)** 0061-2-98453236 ***zhan.wu@health.nsw.gov.au***	DEB/MMC diagnostic testing for patients and family *(Accredited by the National Association of Testing Authorities, Australia).* Next generation sequencing on trial basis.
Canada	**Hospital for Sick Children** Toronto, Canada **Yigal Dror**, **MD** (416) 813-5630 ***yigal.dror@sickkids.ca***	DEB/MMC diagnostic testing *(Licensed under the Ontario Ministry of Health and Long Term Care).* Canadian Inherited Marrow Failure Registry (Enrollment of patients on this research study includes data collection and sample repository).
Germany	**Heinrich Heine University Medical Center** ENT (HNO) Lab Düsseldorf, Germany **Helmut Hanenberg**, **MD** 49-211-811-0832 or 0830 Fax: 0839 ***Hanenberg@uni-duesseldorf.de***	Complementation Group (Research). Use of retroviruses for *A, B, C, D2, E, F, G, I, J, L, M, N, O, P* in primary T-cells, bone marrow or fibroblast cells or in lymphoblast cell lines (LCL). FANCD2 western blot. Coordinator of the Fanconi Anemia Registry (FAR01) in Germany, Austria, and Switzerland, together with Christian Kratz, MD, Hannover Medical School, Germany.
	University of Würzburg Würzburg, Germany **Detlev Schindler**, **MD**, **PhD** +49-931-888 ext. 4088, 4098, or 4075 ***schindler@biozentrum.uni-wuerzburg.de***	MMC Diagnostic Testing *(Certified under Governmental and Societies).* FANCD2 western blot. Prenatal Diagnosis. Mutation Analysis *(Certified under Governmental and Societies).* Complementation Group. Carrier testing.

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Netherlands	**VU University Medical Center** Amsterdam, Netherlands **Hans Gille**, **PhD** 31-20-444-8346 ***jjp.gille@vumc.nl***	Mutation analysis *FANCA (Clinically Certified).* Mutation analysis *FANCC, -E, -F,* and *-G (Clinically Certified).* Mutation analysis of other *FANC* genes *(Clinically Certified).* Prenatal DNA diagnosis *(Clinically Certified).*
Republic of Serbia	**Laboratory for Medical Genetics Mother and Child Health Care Institute of Serbia** St. Radoja Dakica 6-8 11070 Belgrade, Republic of Serbia **Marina Djurisic**, **PhD** 381-11-3108273 ***genetika@imd.org.rs***	DEB diagnostic testing performed in patients with symptoms of FA and on request of the physician/hematologist. Test siblings of DEB-positive patients, but only if requested.
Spain	**Hematopoiesis and Gene Therapy Division**, **CIEMAT/CIBER of Rare Diseases (CIBERER)** Madrid, Spain ***www.ciemat.es*** **Spanish Fanconi Anemia Network:** ***www.redfanconi.org*** **Juan Bueren**, **PhD**, Head of Hematopoiesis and Gene Therapy 34-91-346-6518 ***juan.bueren@ciemat.es*** **José A. Casado**, Senior Researcher 34-91-496-2525 ***jose.casado@ciemat.es*** **Aurora de la Cal**, Secretary of the Spanish FA Research Network 34-91-496-2518 ***aurora.delacal@ciemat.es***	Coordination of the Spanish FA Registry. FA subtyping by retroviral complementation (*A,C,G,E,F*) in T-cells (Research). Studies of G2/M cell cycle arrest in fibroblasts in FA patients with mosaicims in peripheral blood and subtyping by reversion of G2/M arrest with retroviral vectors (*A,C,G*) (Research).

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Spain	**Universitat Autónoma de Barcelona/CIBER of Rare Diseases (CIBERER)** Barcelona, Spain Spanish Fanconi Anemia Network: ***www.redfanconi.org*** ***http://safcro.uab.cat*** **Jordi Surrallés**, **PhD** 34-935-811-830 *jordi.surralles@uab.cat* **Maria Roser Pujol**, **PhD** Cytogeneticist (UAB-CFS Technical Director) ***MariaRoser.Pujol@uab.cat***	Coordination of the Spanish FA Biorepository (biobank of DNA and cell lines from FA patients). DEB diagnostic testing, diagnosis of mosaicism (certified by Societies). Studies of G2/M cell cycle arrest in fibrobasts, FANCD2 ubiquitination and foci formation, western blot of FA proteins. (Research) Mutation analysis (Research) for Spanish gypsy population. Carrier testing is available for family members when mutations are known in the affected case (sibling, etc.) Genetic characterization (subtyping and mutation analysis) of FA patients is done in the lab for Spanish families under the financial umbrells of research grants. Service is not free of charge to non-Spanish patients.

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 results may only be used for research purposes, meaning results may not be shared with referring physicial/family.