

# International Fanconi Anemia Treatment Resources

Editors' note and disclaimer: The resource list provided in this guide is not comprehensive. Inclusion on the list does not imply endorsement by the Fanconi Anemia Research Fund, Inc. Families are encouraged to ask clinicians about their experience treating patients with Fanconi anemia.

## AUSTRALIA

### The Children's Hospital at Westmead

Sydney, NSW 2145, Australia

[www.chw.edu.au/prof/services/fanconi\\_anaemia](http://www.chw.edu.au/prof/services/fanconi_anaemia)

- **Dr. Julie Curtin, MD, PhD**, Head Haematology Department  
Telephone: 612-9845-0000 Fax 612-9845-3332  
Email: [julie.curtin@health.nsw.gov.au](mailto:julie.curtin@health.nsw.gov.au)
- **Professor Peter Shaw**, Head BMT Service  
Telephone: 612-9845-0000 Fax: 612-9845-2171  
Email: [peter.shaw@health.nsw.gov.au](mailto:peter.shaw@health.nsw.gov.au)
- **Ida O'Brien**, Clinical Nurse Consultant BMT Follow Up  
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- **A/Prof. Meredith Wilson**, Clinical Geneticist  
Telephone: 612-9845-3273 Fax: 612-9845-3204  
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- **A/Prof. David Little**, Director Orthopaedic Research  
Telephone: 612-9845-3352 Fax: 612-9845-3180  
Email: [david.little@health.nsw.gov.au](mailto:david.little@health.nsw.gov.au)
- **Professor Chris Cowell**, Head Endocrinology Department  
Telephone: 612-9845-0000 Fax: 612-9845-3170  
Email: [chris.cowell@health.nsw.gov.au](mailto:chris.cowell@health.nsw.gov.au)
- **Dr. Carsten E. Palme, FRACS**, Surgeon, Otolaryngology Head & Neck Surgery  
Telephone: 612-9845-6025 Fax: 612-9893-7440  
Email: [Carsten\\_Palme@specialistservices.com.au](mailto:Carsten_Palme@specialistservices.com.au)
- **Mark Schifter**, Oral Medicine and Oral Pathology  
Telephone: 612-9845-8716 Fax: 612-9845-5740  
Email: [mark.schifter@swahs.health.nsw.gov.au](mailto:mark.schifter@swahs.health.nsw.gov.au)

#### Services Offered:

This 350-bed tertiary care paediatric institution provides comprehensive care for the majority of children in NSW with all diagnoses. This includes a comprehensive team of medical professionals working in disease-specific multi-

disciplinary teams to manage complex disorders with multi-system manifestations, such as Fanconi anaemia. We offer comprehensive management of new and previously diagnosed FA patients, including:

- Diagnostic testing and screening of siblings;
- Genetic counseling; coordination of gene testing via overseas facilities to identify causative mutations in affected individuals; discussion of family planning options for future pregnancies, including prenatal diagnosis or pre-implantation genetic diagnosis for families with identified mutations;
- Orthopaedic management, including specialist hand, upper limb and congenital hand clinic;
- Haematological management;
- Blood and marrow transplantation: comprehensive program of related, unrelated and haploidentical marrow, PBSC and cord blood transplant;
- Co-ordinated long-term follow-up and ongoing surveillance, with particular reference to BMT follow-up, oral, head and neck cancer screening and endocrine follow-up (Chris Cowell);
- Translational research on non-radiotherapy-based BMT conditioning including a well-established drug pharmacokinetic laboratory for real time tailoring of drug doses in individual patients.

### Princess Margaret Hospital for Children

Roberts Road Subiaco

Perth, Western Australia 6008, Australia

- **Catherine Cole, MBBS, FRACP, FRCPA**, Clinical Director of Bone Marrow Transplantation Program
- **Tina Carter, FRACP, PhD**, Bone Marrow Transplant Physician  
Telephone: 08-9340-8222  
Email: [Tina.Carter@health.wa.gov.au](mailto:Tina.Carter@health.wa.gov.au)
- **Shanti Ramachandran, MBBS, FRACP, MPaeds**, Paediatric Oncologist-Clinical Haematologist  
Telephone: 08-9340-8222  
Email: [Shanti.Ramachandran@health.wa.gov.au](mailto:Shanti.Ramachandran@health.wa.gov.au)

## **Sydney Children's Hospital**

Blood & Marrow Transplant Program  
Kids Cancer Centre  
Sydney Children's Hospital  
High St, Randwick  
Sydney, NSW, 2031, Australia

- **A/Prof Tracey O'Brien**, Director, Blood and Marrow Transplant Program, Deputy Director and Head of Clinical Service, Kids Cancer Centre  
Telephone: 02-93821733 Fax: 02-93821789  
Email: [t.obrien@unsw.edu.au](mailto:t.obrien@unsw.edu.au)
- **Dr. Susan Russell**, Head Haematology, Kids Cancer Centre  
Telephone: 02-93821733 Fax: 02-93821789  
Email: [Susan.Russell@sesiahs.health.nsw.gov.au](mailto:Susan.Russell@sesiahs.health.nsw.gov.au)
- **A/Prof. Richard Cohn**, Head, Oncology, Kids Cancer Centre  
Telephone: 02-93821731 Fax: 02-93821789  
Email: [Richard.Cohn@sesiahs.health.nsw.gov.au](mailto:Richard.Cohn@sesiahs.health.nsw.gov.au)
- **Anne Senner**, BMT Coordinator, Blood and Marrow Transplant Program, Kids Cancer Centre  
Telephone: 02-93821733 Fax: 02-93821789  
Email: [Anne.Senner@sesiahs.health.nsw.gov.au](mailto:Anne.Senner@sesiahs.health.nsw.gov.au)

**Lucy Maurice**, BMT Clinical Nurse Consultant, Blood and Marrow Transplant Program, Centre for Children's Cancer and Blood Disorders  
Telephone: 02-93821655  
Email: [Lucy.Maurice@sesiahs.health.nsw.gov.au](mailto:Lucy.Maurice@sesiahs.health.nsw.gov.au)

### **Services Offered:**

Sydney Children's Hospital is a large tertiary paediatric hospital with comprehensive multi-disciplinary services. We offer evaluation of new and previously diagnosed patients with specialists in haematology, bone marrow transplantation, genetics, endocrinology, nephrology, surgery and adolescent medicine. We offer on-site DEB testing, facilitation of DNA mutation testing, family genetic screening and referral for pre-implantation genetic diagnosis. In addition, comprehensive social work, psychology, family support networks are available. The transplant program is one of the oldest and largest national programs with over 900 transplants performed since establishment of the in 1975. The centre is a national leader in unrelated marrow and cord transplants with particular interest in transplant for non-malignant diseases. The program offers a well established multi-disciplinary comprehensive pre- and post-transplant service for patients with FA. The centre director has specialized knowledge in FA having trained in a leading U.S. FA transplant centre, and the centre collaborates with

the International Fanconi Anemia Registry (Dr. Arleen Auerbach).

## **AUSTRIA**

### **St. Anna Children's Hospital**

Department of Hematology and Oncology  
Kinderspitalgasse 6  
Vienna A - 1090, Austria

- **Prof. Wolfgang Holter**, Medical Director  
Email: [aerztliche.direktion@stanna.at](mailto:aerztliche.direktion@stanna.at)

### **Services Offered:**

- Stem cell transplants for FA patients.

## **BRAZIL**

### **Hospital de Clinicas, University of Parana**

Serviço de Transplante de Medula Óssea - 15º andar  
Rua General Carneiro, 181 - 15º andar  
80060-900-Curitiba-PR, Brazil

- **Carmem Bonfim, MD**, Director of Pediatric Stem Cell Transplant Program, Stem Cell Transplant in Genetic Diseases, Coordinator, Fanconi Anemia Outpatient Clinic  
Telephone: 55-41-3360-1000 Fax 55-41-3360-1015  
Email: [carmembonfim@gmail.com](mailto:carmembonfim@gmail.com)
- **Ricardo Pasquini, MD, PhD**, Senior Professor  
Federal University of Parana  
Telephone: 55-41-3360-1000 Fax: 55-41-3360-1015  
Email: [pasquini@hc.ufpr.br](mailto:pasquini@hc.ufpr.br)

### **Services Offered:**

- Comprehensive clinic for FA patients;
- Out-patient unit for diagnostic procedures and treatment;
- Cytogenetics laboratory performing DEB chromosomal breakage testing and conventional cytogenetics of bone marrow;
- Transplant unit performing related and unrelated bone marrow and umbilical cord transplants;
- Haploidentical transplants;
- Mutation analysis (*FANCA*, *C* and *G*) and *FANCD2* by western blot.

## CANADA

### Hospital for Sick Children

#### University of Toronto

555 University Avenue  
Toronto, Ontario, Canada M5G 1X8

- **Yigal Dror, MD, FRCP(C)**, Professor of Paediatrics; Director, Marrow Failure and Myelodysplasia Program; Head, Hematology Section; Division of Hematology/Oncology; Scientist, Program in Genetics and Genome Biology, Research Institute  
Telephone: 416-813-5668 Fax: 416-813-5327  
Email: [yigal.dror@sickkids.ca](mailto:yigal.dror@sickkids.ca)
- **Adam Gassas, MBChB, MSc, MRCP, DCH, MRCPCH**, Associate Professor of Pediatrics; Section of Blood & Marrow Transplant, Division of Hematology/Oncology  
Telephone: 416-813-8227 Fax: 416-813-5327  
Email: [adam.gassas@sickkids.ca](mailto:adam.gassas@sickkids.ca)

### Princess Margaret Hospital

Ontario Cancer Institute  
University of Toronto  
610 University Avenue  
Toronto, Ontario, M5G 2M9, Canada

- **Jeffrey H. Lipton, FRCPC, MD, PhD**, Leukemia and Allogeneic Stem Cell Transplant Programs, PMH Professor of Medicine, University of Toronto  
Telephone: 416-946-2266 Fax: 416-946-6585  
Email: [Jeff.Lipton@uhn.on.ca](mailto:Jeff.Lipton@uhn.on.ca)

### University of Manitoba

ON2021a-675 McDermot Avenue  
Winnipeg, Manitoba, Canada R3E 0V9

- **John J. Doyle, MD, FRCPC, FAAP**, Associate Professor, Dept. of Pediatrics and Child Health, University of Manitoba; Head, Pediatric Hematology/Oncology/BMT  
Telephone: 204-787-4135 Fax: 204-786-0195  
Email: [john.doyle@cancercare.mb.ca](mailto:john.doyle@cancercare.mb.ca)

## CHINA

### Tianjin First Central Hospital

No. 24, Fukang Road, Nankai  
District, Tianjin, China

**Mingfeng Zhao, MD, PhD**

### Capital Institute of Pediatrics

No. 2 Yabao Road  
Chaoyang District  
100020 Beijing, China

**Xiaodong Shi, MD, PhD**

Telephone: 13911601076

## COLOMBIA

### Clínica Las Americas

Diagonal 75 B #2 A80 Ofic. 520  
Medellin  
Antioquia, Colombia

- **Dr. Amado Karduss**, Adult Hematology/Stem Cell Transplantation  
Telephone: 57 4 3409314  
Email: [amaka@une.net.co](mailto:amaka@une.net.co)
- **Dr. Yadira Coll**, Pediatric Hematology  
Telephone: 57 4 3409314  
Email: [yacoll@une.net.co](mailto:yacoll@une.net.co)

#### **Services Offered:**

- Evaluation;
- Medical treatment;
- FA stem cell transplantation.

### Dr. Adriana Linares

Hospital de La Misericordia  
Av Caracas 1-67  
Pediatric Hematology/Oncology Unit  
Telephone: 571-381-1970 extension 1309  
Email: [alinaresb@gmail.com](mailto:alinaresb@gmail.com)

- **Universidad Nacional de Colombia**  
Departamento de Pediatría  
Unidad de Oncohematologie PEDIATRICA  
Av Caracas 1-67 4 piso  
Bogotá, Colombia

**Services Offered:**

- Pediatric hematology oncology, clinical evaluation, medical treatment, and therapy.

**Centro Médico Imbanaco de Cali**

Bone Marrow Transplant Unit  
Carrera 38 A No. 5A 100  
Cali, Valle, Colombia

- **Oscar W. Ramirez, MD, MPhil**  
Telephone: 572-682-1000 or 572-518-6000  
Email: *oscar.ramirez@imbanaco.com.co* or *oramirez1167@gmail.com*

**Services Offered:**

- Pediatric hematology oncology;
- Hematopathology;
- Clinical evaluation;
- Medical therapy;
- Stem cell transplantation related and unrelated.

**CZECH REPUBLIC****University Hospital Motol**

Department of Paediatric Haematology and Oncology V  
Uvalu, 84  
150 06 Prague, Czech Republic

- **Prof. Petr Sedlacek**  
Telephone: 420 2 2443 6552 Fax: 420 2 2443 6519  
Email: *petr.sedlacek@lfmotol.cuni.cz*

**Services Offered:**

- Stem cell transplants for FA patients.

**DENMARK****University Hospital of Aarhus at Skejby**

Brendstrupgaardsvej 100  
DK-8200 Aarhus N, Denmark

- **Niels Clausen, MD**, Department of Pediatrics  
Telephone: 45-7845-1673  
Email: *niecla@rm.dk*

**Services Offered:**

- Diagnosis, treatment and follow-up for children with Fanconi anemia.

**FRANCE****Hôpital Robert Debré**

48, boulevard Sérurier  
75935 Paris cedex 19, France

- **Thierry Leblanc, MD**, Department of Pediatric Hematology  
Secretary: 33-1-7128-2535 Fax: 33-1-4003-4740  
Email: *thierry.leblanc@rdb.aphp.fr*
- **Jean-Hugues Dalle, MD, PhD**, Department of Pediatric Hematology  
Telephone direct: 33-1-4003-1241  
Secretary: 33-1-4003-5388 Fax: 33-1-4003-4740  
Email: *jean-hugues.dalle@rdb.aphp.fr*

**Services Offered (primarily for children):**

- Bone marrow, peripheral stem cell and cord blood transplants;
- FA diagnosis: breakage tests, cell cycle, FANCD2 analysis, mosaicism on skin biopsy, retroviral group complementation, bone marrow karyotype (Dr. Jean Soulier);
- Gene sequencing on grouped patients, in collaboration with Institut Curie (Pr. Dominique Stoppa-Lyonnet);
- Head and neck cancer evaluation and treatment (Dr. Francis Pascal and Dr. Arnaud Rigolet);
- Cell therapy laboratory (Dr. Jerome Larghero);
- Gene therapy laboratory (Dr. Philippe Leboulch).

**Hôpital Saint-Louis**

1, Avenue Claude Vellefaux  
75010 Paris, France

- **Gérard Socié, MD, PhD**, Department of Hematology Transplantation  
Telephone direct: 33-1-4249-9824  
Secretary: 33-1-4249-9639 Fax: 33-1-4249-9636  
Email: *gerard.socie@sls.aphp.fr*
- **Jean Soulier, MD, PhD**, Genome Rearrangements and Cancer Group, Hematology Laboratory  
Telephone: 33-1-4249-9891 Fax: 33-1-4249-4027  
Email: *jean.soulier@sls.aphp.fr*
- **Arnaud Rigolet, MD**, Department of Oral and Maxillofacial Surgery  
Telephone: 33-1-42-49-41-92 Fax: 33-1-1-49-49-91-92  
Email: *arnaud.rigolet@sls.aphp.fr*

**Services Offered (primarily for teens and adults):**

- Bone marrow, peripheral stem cell and cord blood transplants;
- FA diagnosis: breakage tests, cell cycle, FANCD2

analysis, mosaicism on skin biopsy, retroviral group complementation, bone marrow karyotype (Dr. Jean Soulier);

- Gene sequencing on grouped patients, in collaboration with Institut Curie (Pr. Dominique Stoppa-Lyonnet);
- Head and neck cancer evaluation and treatment (Dr. Francis Pascal and Dr. Arnaud Rigolet);
- Cell therapy laboratory (Dr. Jerome Larghero);
- Gene therapy laboratory (Dr. Philippe Leboulch).

## GERMANY

### Charité, Campus Virchow Klinikum

#### University Medicine, Berlin

Children's Hospital  
Augustenburger Platz 1  
13353, Berlin, Germany

- **Jörn-Sven Köhl, MD**

Telephone: 49-30-450 566432

Fax: 049-030-450-566919

Email: [jskuehl@charite.de](mailto:jskuehl@charite.de)

#### **Services Offered:**

- Pediatric bone marrow transplant service;
- Stem cell laboratory;
- Stem cell bank;
- Cytogenetic laboratory;
- German Fanconi anemia registry.

### Hannover Medical School

Department of Pediatric Hematology and Oncology  
Carl-Neuberg-Str. 1  
30625 Hannover, Germany

- **Christian Peter Kratz, MD, Director**

Telephone: 49 (0)511 532 6711 Fax: 49 (0)511 532 9120

Email: [kratz.christian@mh-hannover.de](mailto:kratz.christian@mh-hannover.de)

#### **Services Offered:**

- Natural history study for patients with Fanconi anemia;
- Guidance in clinical decision making.

### Heinrich Heine University Medical Center

Children's Hospital  
Moorenstrasse 5  
40225 Düsseldorf, Germany

- **Eunike Velleuer, MD, Department of Pediatric**

Hematology, Oncology, and Clinical Immunology

Telephone: 49-211-811-7687 Fax: 49-211-811-7634

Email: [velleuer@uni-duesseldorf.de](mailto:velleuer@uni-duesseldorf.de)

- **Arndt Borkhardt, MD, Director, Department of Pediatric Hematology, Oncology, and Clinical Immunology**

Telephone: 49-211-811-7680 Fax: 49-211-811-6206

Email: [lesch@med.uni-duesseldorf.de](mailto:lesch@med.uni-duesseldorf.de)

#### **Services Offered:**

- Clinical care of FA patients;
- Cooperation with Ralf Dietrich, German FA family support group (Deutsche FA-Hilfe e.V.) on "Reducing the burden of squamous cell carcinoma in Fanconi anemia;"
- Cooperation with Birgit Henrich, University of Duesseldorf, Microbiology; Microbiom of the oral cavity in FA;
- Cooperation with Ruud Brakenhoff, University of Amsterdam, on "Progression of FA-SCC and Development of a Non-Invasive Screening Method for Precursor Lesions;"
- Cooperation with Prof. Stefan Biesterfeld, University of Duesseldorf, Cytopathology; non-invasive testing of oral lesions (Cytology, DNA-Cytometry);
- Cooperation with Prof. Torsten Remmerbach, University of Leipzig, development of alternative non-invasive methods for the detection of oral cancers.

### University of Tuebingen

Children's University Hospital  
Department of Hematology/Oncology and General Pediatrics  
Hoppe-Seyler-Strasse 1  
72076 Tuebingen, Germany

- **Prof. Rupert Handgretinger, MD, Chairman**

Telephone : 49-7071-298-4744 Fax: 49-7071-294713

Email: [Rupert.Handgretinger@med.uni-tuebingen.de](mailto:Rupert.Handgretinger@med.uni-tuebingen.de)

#### **Services Offered:**

- Evaluation of new patients;
- Hematopoietic stem cell transplantation from HLA-matched siblings, matched unrelated donors and haploidentical family donors;
- Co-transplantation of bone marrow with mesenchymal stem cells to enhance engraftment and to prevent graft-versus-host-disease (GvHD);
- Extracorporeal photopheresis in small children for the prevention and treatment of GvHD;
- Adoptive transfer of virus-specific T-cells to prevent viral infections post-transplant;
- Long-term follow-up studies (growth, endocrinology, quality of life).

## INDIA

### Apollo Specialty Hospital

320 Padma Complex  
Anna Salai  
Chennai 600 035, India

- **Sheila Mohan, MBBS, DCH, PhD**  
Telephone: 91-44-2491-2943  
Mobile: 91-92-8211-4661
- **Revathi Raj, MBBS, DCH, MRCP (Path)**  
Telephone: 00- 91-44-33151294

#### Services Offered:

- Complete evaluation, treatment and follow-up of new and previously diagnosed FA patients;
- Provide information and assistance for physicians in the care of their FA patients;
- Cancer screening and specialist consultations;
- Diagnostic testing (MMC test) and screenings of siblings;
- Complementation grouping ;
- Genetic counseling;
- Stem cell transplantation;
- Maintenance of national database: Registry for Fanconi Anemia in India (REFAIN).

### National Institute of Immunohaematology (ICMR)

13th floor, New Multistoried Building  
K.E.M. Hospital Campus, Parel  
Mumbai 400012, India

- **Dr. V. Babu Rao**  
Telephone: 91-22-24138518 or 24138519  
Fax: 91-22-24138521  
Email: [vbaburao@hotmail.com](mailto:vbaburao@hotmail.com)

#### Services Offered:

- Chromosomal breakage study using MMC and DEB test;
- Cytogenetic study from bone marrow and FISH analysis;
- FANCD2 western blot;
- Molecular testing of complementation groups *FANCA*, *FANCC*, *FANCG*;
- Prenatal diagnosis of FA after mutation detection in proband.

## IRAN

### Tehran University of Medical Sciences

Shariati Hospital  
Hematology-Oncology and SCT Research Center  
Kargar Avenue  
14114, Tehran, Iran

<http://horecst.tums.ac.ir>

- **Ardeshir Ghavamzadeh, MD**, Professor of Medicine  
Adult Hematologist Oncologist  
President of Hematology-Oncology and SCT Research Center  
Telephone: 98-21-84902635 Fax: 98-21-88004140  
Email: [ghavamza@sina.tums.ac.ir](mailto:ghavamza@sina.tums.ac.ir)
- **Amir Ali Hamidieh, MD**, Associate Professor of Medicine  
Pediatric Hematologist Oncologist  
Director of Fanconi Anemia Transplant Program  
Telephone: 98-21-84902645 Fax: 98-21-88004140  
Email: [aahamidieh@sina.tums.ac.ir](mailto:aahamidieh@sina.tums.ac.ir)

#### Services Offered:

- Pediatric and adult FA patients can be seen by specialists in pediatric and adult medicine;
- Pediatrics stem cell transplantation, 11-bed ward;
- Adults stem cell transplantation, 25-bed ward;
- Stem cell laboratory;
- Cord blood bank;
- Cytogenetic laboratory;
- Complete evaluation, treatment and follow-up of new and previously diagnosed FA patients;
- Diagnostic testing (DEB and MMC test) and screenings of siblings;
- Genetic counseling;
- Matching sibling, unrelated, haploidentical and cord blood stem cell transplants;
- HLA registry (Iranian Stem Cell Donor Program, ISCDP).

## ISRAEL

### International Center for Cell Therapy & Cancer Immunotherapy (CTCI)

Weizman Center, Top Ichilov  
14 Weizman Street  
Tel Aviv 64239, Israel

- **Shimon Slavin, MD**, Professor of Medicine, Scientific & Medical Director of CTCI  
Telephone: 972-77-777-9255 Fax: 972-77-777-9247  
Email: [slavin@CTCIcenter.com](mailto:slavin@CTCIcenter.com)

### Services Offered:

Allogeneic stem cell transplantation using well-tolerated reduced intensity conditioning towards minimizing the risk of the transplant procedure by using nonmyeloablative stem cell transplantation. Stem cell transplantation can also be considered from half-matched (haploidentically mismatched) family member while avoiding the risk of graft-versus-host disease.

### Schneider Children's Medical Center of Israel

14 Kaplan Street  
PO Box 559  
Petach Tikvah 49202  
Israel

- **Hannah Tamary, MD**, Director of Pediatric Hematology, Professor of Pediatrics  
Telephone: 972-3-9253669 Fax: 972-3-9253042  
Email: [htamary@post.tau.ac.il](mailto:htamary@post.tau.ac.il)
- **Joanne Yakobovich, MD**, Senior Pediatric Hematologist
- **Jerry Stein, MD**, Director of Pediatric Bone Marrow Transplantation

### Services Offered:

- Mutation detection for all Fanconi anemia genes, genetic counseling, and prenatal diagnosis;
- Comprehensive clinical care.

## ITALY

### G. Gaslini Children's Institute

Largo G.Gaslini 5  
16147 Genova, Italy

- **Carlo Dufour, MD**, Head, Hematology Unit  
Telephone: 39-010-5636-694 Fax: 39-010-5636-714  
Email: [carlodufour@ospedale-gaslini.ge.it](mailto:carlodufour@ospedale-gaslini.ge.it)
- **Johanna Svahn, MD**, Hematology Unit  
Telephone: 39-010-5636-694 Fax: 39-010-5636-714  
Email: [johanna.svahn@ospedale-gaslini.ge.it](mailto:johanna.svahn@ospedale-gaslini.ge.it)
- **Enrico Cappelli, PhD**, Hematology Laboratory  
Telephone: 39-010-5636-693 Fax: 39-010-5636-714  
Email: [enricocappelli@ospedale-gaslini.ge.it](mailto:enricocappelli@ospedale-gaslini.ge.it)

### Services Offered:

- Primary diagnosis with DEB test, cell cycle analysis and MMC sensitivity, FANCD2 Ubiquitination (WB) on peripheral blood cells and fibroblasts;
- FA complementation group analysis by retroviral vectors on peripheral blood T-cells, bone marrow cells, fibroblasts and B-cell lines for groups A, C, G, D2;

- Collaboration with Dr. Helmut Hannenberg, Riley Hospital for Children, Indianapolis and Dr. Juan Bueren, CIEMAT, Madrid, Spain for remaining FA complementation group analyses;
- Collaboration with Prof. Juan Carlos Belmonte Ispizua, University LaJolla, San Diego, CA for IPS;
- Collaboration with Dr. Anna Savoia, IRCSS Burlo Garofolo, University of Trieste, Italy, for molecular diagnosis of FA;
- Collaboration with Dr Paolo Degan, Mutagenesis Lab, Scientific Tumour Institute, Genova, Italy, for redox activity;
- Haematopoiesis laboratory (cell cultures, cytokine hypersensitivity tests);
- Cytogenetic laboratory;
- Collaboration with European Centres of the EBMT (European Society for Blood and Marrow Transplantation) for data collection and retrospective studies;
- Close scientific collaboration with Dr. Grover Bagby (OHSU, Portland, OR, USA) on pathogenetic mechanism of marrow failure in FA;
- Comprehensive care and long-term follow-up of new and previously diagnosed patients, including hematopoietic stem cell transplantation with related, unrelated, matched or mismatched donors (Dr. Edoardo Lanino, HSC Unit, G. Gaslini Children's Hospital), head and neck cancer evaluation, endocrinology, orthopedic and nephrology clinic, psychological support;
- Ante-natal diagnosis available for patients in whom mutations are known;
- Experimental protocol for treatment of FA patients with bone marrow failure and no HSC donor available, using Etanercept (anti TNF-alpha); Eudract Code (Italian Ministry of Health Registration Code) 2005-003096-20. This study has now been published.

### Second University of Naples

Clinica Pediatrica 1, Department of Paediatrics  
Via De Grecchio 4  
80138 Napoli, Italy

- **Bruno Nobili, MD**  
Telephone and Fax: 39-0815-665432  
Email: [bruno.nobili@unina2.it](mailto:bruno.nobili@unina2.it)
- **Silverio Perrotta, MD**  
Telephone and Fax: 39-0815-665421  
Email: [silverio.perrotta@unina2.it](mailto:silverio.perrotta@unina2.it)
- **Sofia Maria Rosaria Matarese, MD**  
Telephone and Fax: 39-0815-665444  
Email: [SofiaMariaRosaria.MATARESE@unina2.it](mailto:SofiaMariaRosaria.MATARESE@unina2.it)

- **Francesca Rossi, MD**  
Telephone and Fax: 39-0815-665342  
Email: [francesca.rossi@unina2.it](mailto:francesca.rossi@unina2.it)

**Services Offered:**

- Complete clinical evaluation, treatment and follow-up of new and previously diagnosed FA patients;
- Diagnostic testing (chromosomal breakage with Mitomycin C, DEB test, cell cycle analysis, karyotype, bone marrow examination, cytogenetic analysis, FANCD2 ubiquitination (WB) on peripheral blood cells and fibroblasts);
- Transfusion of irradiated blood and platelet products if needed;
- Information, education and assistance to families of patients with FA and referring physicians;
- Coordination of consultations with other subspecialties for management of FA-related disorders.

**University of Pavia**

Pediatric Haematology and Oncology Unit  
Fondazione IRCCS, Policlinico San Matteo  
Piazzale Golgi, 2  
27100, Pavia, Italy

- **Prof. Franco Locatelli, MD**  
Telephone: 39-0382-502607 Fax: 39-0382-501251  
[f.locatelli@smatteo.pv.it](mailto:f.locatelli@smatteo.pv.it)

**Services Offered:**

- Hematopoietic stem cell transplantation from HLA-identical or disparate family donors and from unrelated volunteers;
- Stem cell laboratory;
- Cell therapy laboratory;
- Stem cell bank;
- Cytogenetic laboratory;
- Molecular diagnosis of Fanconi anemia.

**JAPAN**

**Tokai University**

Department of Cell Transplantation and Regenerative  
Medicine  
Shimokasuya 143  
Isheara, Kanagawa, 259-1193, Japan

- **Hiromasa Yabe, MD, Stem Cell Transplantation**  
Telephone: 81-463-93-1121 Fax: 81-463-95-9567  
Email: [yabeh@is.icc.u-tokai.ac.jp](mailto:yabeh@is.icc.u-tokai.ac.jp)

- **Miharu Yabe, MD, Stem Cell Transplantation**  
Telephone: 81-463-93-1121 Fax: 81-463-95-9567  
Email: [miharu@is.icc.u-tokai.ac.jp](mailto:miharu@is.icc.u-tokai.ac.jp)

**Services Offered:**

- Evaluation of new and previously diagnosed patients;
- Expertise in pediatric and adult stem cell transplantation from matched sibling donor or alternative donor;
- Special interest in new conditioning regimens for patients with FA to improve long-term quality of life.
- Direct collaboration with Prof. Minoru Takata, the Kyoto University, for mutation analysis.

**KENYA**

**Kenvatta National Hospital**

Haematology & Blood Transfusion Unit  
Department of Human Pathology  
University of Nairobi, School of Medicine  
Nairobi, Kenya

- **Dr. Grace W. Kitonyi MBChB, FRCPath**  
Haematologist/Haematopathologist  
Telephone: 254 20 2721815  
Mobile phone: 254722385336  
Email: [gwkitonyi@yahoo.com](mailto:gwkitonyi@yahoo.com)  
Cc: [gkitonyi@uonbi.ac.ke](mailto:gkitonyi@uonbi.ac.ke)
- **Dr. Jessie N. Githanga MBChB, MMED Path**  
Haematologist/Haematopathologist  
Telephone: 254 20 2721815  
Mobile phone: 254721245721  
Email: [jgithanga3@gmail.com](mailto:jgithanga3@gmail.com)
- **Dr. J. A. Rajab, MBChB, MMED, Path, MPH**  
Haematopathologist/Public Health Specialist  
Telephone: 254 20 2721815  
Mobile phone: 254722707421  
Email: [amjamilla@yahoo.com](mailto:amjamilla@yahoo.com)

**Services Offered:**

- Diagnosis and management of all patients with FA including counseling of families;
- Coordination of consultations with other sub-specialities for management of FA related disorders.



## LEBANON

### St. George Hospital University Medical Center

Dept of Pediatrics and Pediatric Hematology/Oncology  
Office 121  
Ashrafieh, Beirut, Lebanon

- **Dr. Roula Farah, MD, FAAP**

Associate Professor of Pediatrics and Pediatric Hematology/Oncology  
President, CHANCE Association  
Cell phone: 961-3-681-309  
Email: [roula.fs@dm.net.lb](mailto:roula.fs@dm.net.lb)

#### **Services Offered:**

- Complete clinical evaluation, treatment and follow-up of new and previously diagnosed FA patients;
- Prenatal diagnosis;
- Diagnostic testing (chromosomal breakage with mitomycin C, karyotype, bone marrow examination, etc.);
- Genetic testing for *FANCA* gene mutations;
- Transfusion of irradiated blood and platelet products if needed;
- Support with financial coverage for diagnostic studies if patient cannot afford it;
- Information, education and assistance to families of patients with FA and referring physicians;
- Genetic counseling;
- Maintenance of the National Fanconi Anemia Registry.

## MEXICO

### Centro Medico Nacional “La Raza”

Instituto Mexicano Del Seguro Social  
Division de Trasplantes  
Seris y Zaachila  
Col. La Raza, 02990  
Mexico DF, Mexico

- **Dr. Inès Montero Ponce**
- **Dr. Ana Lilia Hernández Moreno**

### Prof. Dr. Clara Gorodezky

Head of Department of Immunology & Immunogenetics, InDRE  
Secretary of Health  
President of the Board of Fundacion Comparte Vida, A.C.  
Francisco P. Miranda #77 - Laboratory Building, 2nd Floor, Mexico DF, 01480, Mexico  
Phone: +5255 6383 0041 & +5255 5062 1600 ext. 59314 & 59397

Email: [clarag@unam.mx](mailto:clarag@unam.mx) or  
[cgorodea@fundacioncompartevida.org.mx](mailto:cgorodea@fundacioncompartevida.org.mx)  
[www.fundacioncompartevida.org.mx](http://www.fundacioncompartevida.org.mx)

#### **Services Offered:**

- HLA Lab, ASHI accredited laboratory;
- Access to DONORMO (Mexican Unrelated Bone Marrow Donors Registry) as well as the BACECU (Mexican Unrelated Cord Blood Bank);
- HLA typing and all related studies for solid organ transplantation;
- Search DONORMO, BACECU, and BMDW, etc. and perform all the workup;
- Referrals to physicians for Fanconi anemia patients;
- We work with all the hospitals in Mexico and with many transplant centers abroad;
- We belong to BMDW and are a Coop Registry of NMDP.

### Instituto Nacional de Pediatría

Instituto de Investigaciones Biomédicas  
UNAM  
Insurgentes Sur 3700-C  
Col. Insurgentes Cuicuilco  
4530 Mexico City, DJ, Mexico

#### **Sara Frias, PhD**

Telephone: 52-55-1084-5533  
Email: [sarafrias@biomedicas.unam.mx](mailto:sarafrias@biomedicas.unam.mx)

#### **Marisol González, MD**

Telephone: 55-320-51010  
Email: [mary\\_solg@hotmail.com](mailto:mary_solg@hotmail.com)

#### **Services Offered:**

- Clinical and laboratory diagnosis of Fanconi anemia;
- Hematology in bone marrow and blood;
- Analysis of spontaneous chromosomal aberrations and chromosomal breakage test induced with DEB and MMC in lymphocytes, fibroblasts and amniocytes;
- Analysis of G2/M cell cycle halt by flow cytometry in lymphocytes and fibroblasts.

### Unidad de Citogenética

Servicio de Hemato-Oncología Pediátrica, Unidad de Citogenética, Nuevo Hospital Civil  
Dr. Juan I Menchaca  
Laboratorio de Citogenética Genotoxicidad y Biomonitorio  
CUCS, Universidad de Guadalajara

#### **Juan I. Menchaca, MD**

Nuevo Hospital Civic  
División de Pediatría,  
Servicio de Hematología Pediátrica  
Unidad de Citogenética,  
Salvador de Quevedo y Aubieta #750, CP-44340

Guadalajara, Jalisco, México  
Telephone: 52-33-3883440 ext. 52866

**Enrique Corona Rivera, MD**

Universidad de Guadalajara  
Centro Universitario de Ciencias de la Salud  
Instituto de Genética Humana  
Laboratorio de Citogenética genotoxicidad y Biomonitorio  
Edificio P 2do Nivel  
Sierra Mojara #950, S.L., CP-44340  
Guadalajara, Jalisco, México  
Telephone: 52-33-10585200 ext. 33647

**Lucina Bobadilla-Morales, MD, PhD**

Main contact: Diagnosis, research, and patient management  
Email: [lucinabo@cucs.udg.mx](mailto:lucinabo@cucs.udg.mx)

**Alfredo Corona-Rivera, PhD**

**Research and diagnostic**  
Email: [alcoronar@gmail.com](mailto:alcoronar@gmail.com)

**Fernando Sánchez-Zubieta, MD**

**Pediatric Hemato-oncology services**  
Email: [dfernados59@hotmail.com](mailto:dfernados59@hotmail.com)

**Services Offered:**

- Clinical and laboratory diagnosis of Fanconi anemia;
- Hematology in bone marrow and blood;
- Analysis of spontaneous chromosomal aberrations and chromosomal breakage test induced with MMC in lymphocytes;
- MLPA molecular biology studies in Fanconi anemia patient samples;
- Genetic counseling for patients, parents, and relatives;
- Diagnostic evaluation for Fanconi anemia and other bone marrow failure syndromes;
- Consultation and education for new and previously diagnosed patients and their families;
- Comprehensive clinical management of pediatric Fanconi anemia patients;
- Hematopoietic stem cell progenitors transplantation;
- Performance of HLA- A\*, B\*, C\*, DQ\*, DR\*, high resolution typing;
- Cytogenetics, FISH, molecular biology, cryopreservation and flow cytometry facilities to alternative studies.

## **NETHERLANDS**

### **Leiden University Medical Center**

Department of Pediatrics/BMT unit  
PO Box 9600  
Leiden, 2300 RC, Netherlands

- **Arjan C Lankester, MD, PhD**, Program Director Stem Cell Transplantation, Department of Pediatrics/BMT unit  
Telephone: 31-71-5264131 Fax: 31-71-5248198  
Email: [A.Lankester@lumc.nl](mailto:A.Lankester@lumc.nl)

### **UMC Utrecht**

Kinderarts - Hematoloog  
Wilhelmina Kinderziekenhuis  
Postbus 85090  
3508 AB Utrecht  
Huispostnr KC 03.063.0, Netherlands

- **Marc Bierings, MD, PhD**, Represents the Dutch Fanconi Anemia Guideline on behalf of the Dutch Childhood Oncology Group (DCOG), and Chairs the Guideline Committee for FA in Netherlands.  
Telephone: 31 (0)88 7554344 Fax: 31 (0)88 7555350  
Email: [m.bierings@umcutrecht.nl](mailto:m.bierings@umcutrecht.nl)

### **VU University Medical Center Amsterdam**

Department of Clinical Genetics  
Van der Boechorststraat 7  
NL-1081 BT Amsterdam, Netherlands

**Diagnosis:**

Telephone: 31-20-444-8346 Fax: 31-20-444-8293

- **Hans Gille, PhD**  
Email: [jgp.gille@vumc.nl](mailto:jgp.gille@vumc.nl)
- **Resie Vervenne, PhD**  
Email: [r.vervenne@vumc.nl](mailto:r.vervenne@vumc.nl)
- **Pino Poddighe, PhD**  
Contact for chromosomal breakage testing.  
Email: [p.poddighe@vumc.nl](mailto:p.poddighe@vumc.nl)

**Research:**

Telephone: 31-20-444-8270 Fax: 31-20-444-8285

- **Josephine C. Dorsman, PhD**  
Email: [jc.dorsman@vumc.nl](mailto:jc.dorsman@vumc.nl)

**Services Offered:**

**Diagnosis:**

- Diagnosis by chromosomal breakage analysis, including mosaicism;
- Molecular diagnosis by mutation screening of all known FA genes;
- Prenatal DNA testing (if mutations are known).

**Research:**

- Identification of the genes responsible for FA and “overlapping” conditions;

- Understanding the FA pathway and how a defect in the pathway causes FA-specific clinical symptoms;
- Discovery of FA pathway defects and their role in sporadic cancer.

#### **Collaboration in Netherlands:**

- **M. Bierings, MD, PhD**, Stem Cell Transplantation, Clinical Coordinator for FA families in The Netherlands, Utrecht University Medical Center
- **E.T. Korthoff, MD, PhD**, Stem Cell Transplantation, Leiden University Medical Center
- **R.H. Brakenhoff, PhD**, Dept. Otolaryngology/Head-Neck Surgery, VU University Medical Center Amsterdam.

## **NEW ZEALAND**

### **Christchurch Hospital**

#### **Children's Haematology Oncology Centre**

Private Bag 4710  
Christchurch 8140, New Zealand

- **Dr. Siobhan Cross**, Paediatric Haematologist  
Telephone: 64 3 364 0640, ext. 87500  
Email: [Siobhan.Cross@cdhb.govt.nz](mailto:Siobhan.Cross@cdhb.govt.nz)

#### **Services Offered:**

- FA diagnosis (samples are sent to Australia), follow up/surveillance for FA patients under age 18.

### **Starship Children's Hospital**

Grafton  
Auckland 1023, New Zealand

- **Dr. Nyree Cole**  
Telephone: 64 9 307 4919  
Email: [Nyreec@adhb.govt.nz](mailto:Nyreec@adhb.govt.nz)

## **REPUBLIC OF KOREA**

### **Seoul St. Mary's Hospital**

#### **The Catholic University of Korea**

Blood & Marrow Transplantation Center  
#222 Banpo-daero, Seocho-gu  
Seoul, 137-701, Republic of Korea  
Telephone: 82-10-9407-2220 Fax: 82-2-537-4544

- **Nack-Gyun Chung, MD**, Professor, Department of Pediatrics, Hematology/Oncology/BMT  
Telephone: 82-2-2258-6188 Fax: 82-2-537-4544  
Email: [cngped@catholic.ac.kr](mailto:cngped@catholic.ac.kr)

- **Myungshin Kim, MD, PhD**, Professor, Department of Laboratory Medicine  
Telephone: 82-2-2258-1645 Fax: 82-2-2258-1719  
Email: [microkim@catholic.ac.kr](mailto:microkim@catholic.ac.kr)

#### **Services Offered:**

- Evaluation of new and previously diagnosed patients;
- Breakage test, sequence analysis;
- Diagnosis and management of all patients with FA including counseling of families;
- Expertise in stem cell transplantation from matched sibling donor or alternative donor.

## **SAUDI ARABIA**

### **King Faisal Specialist Hospital & Research Center**

Riyadh, Saudi Arabia

- **M. Ayas, MD**, Head, Section of Pediatric Stem Cell Transplantation Department of Pediatric Hematology Oncology  
Telephone: 966-1-442-3934 Fax: 966-1-442-4662  
Email: [mouhab@kfshrc.edu.sa](mailto:mouhab@kfshrc.edu.sa)

#### **Services Offered:**

- Diagnosis: cytogenetics testing including DEB chromosomal breakage testing and conventional cytogenetics of bone marrow and peripheral blood;
- Comprehensive care for FA patients, including blood and platelet transfusion;
- Allogeneic stem cell transplant: matched related bone marrow and matched/partially matched unrelated cord blood transplants;
- Haploidentical stem cell transplantation for FA patients with no matched related donors.

## **SINGAPORE**

### **National University Hospital, Singapore**

Department of Paediatrics  
1E Kent Ridge Road  
NUHS Tower Block Level 12  
Singapore 119228  
Republic of Singapore  
<http://www.nuh.com.sg/ucmi/>

- **Poh-Lin Tan, MBBS, MMED** (Paediatrics)  
Email: [poh\\_lin\\_tan@nuhs.edu.sg](mailto:poh_lin_tan@nuhs.edu.sg)

### **International Patient Liaison Centre:**

Telephone (24hr Helpline): (65) 67792777

Email: [iplc@nhg.com.sg](mailto:iplc@nhg.com.sg)

### **Services Offered:**

- FACT-accredited paediatric blood and marrow transplant;
- Paediatric sub-specialties: adolescent medicine, allergy, cardiology, developmental paediatrics, endocrinology, gastroenterology, genetics, haematology, infectious diseases, intensive care, nephrology, neurology, oncology, otolaryngology, psychiatry, pulmonology, rheumatology, surgery.

## **SOUTH AFRICA**

### **Chris-Hani Baragwanath Hospital**

Division of Haematology and Oncology

Johannesburg, South Africa

- **Dr. Linda Wainwright**

Clinic Telephone: 0800333555

Office Telephone: 0119339594

Cell: 0828219985

Email: [rosalind.wainwright@wits.ac.za](mailto:rosalind.wainwright@wits.ac.za)

### **Johannesburg Hospital**

Division of Haematology and Oncology

Dept. of Pediatrics

Private Bag X39

Johannesburg 2000, South Africa

- **Prof. Janet Poole**

Telephone: 27(0)114883294/0 Fax: 27(0)114884152

Cell: 27(0)824695762

Email: [Janet.Poole@wits.ac.za](mailto:Janet.Poole@wits.ac.za)

### **NHLS, Division of Human Genetics**

### **University of the Witwatersrand**

PO Box 1038

Johannesburg 2000, South Africa

- **Dr. Amanda Krause**

Telephone: 27-11-489-9219/9224

Mobile: 27-82-809-5797

Fax: 27-11-489-9226

Email: [amanda.krause@nhls.ac.za](mailto:amanda.krause@nhls.ac.za)

### **Services Offered (Chris-Hani Baragwanath, Johannesburg Hospital, and NHLS):**

- Genetic counseling service for families with FA;
- Diagnostic, carrier, preclinical and prenatal testing.

### **Unitas Hospital**

PO Box 15225

Lyttleton, 0140, Pretoria

Gauteng, South Africa

- **Dr. Charmaine Jacobs, MD**, Pediatric

Oncology/Haematology Unit

Telephone: 27-12-6778076

Mobile Phone: 27724195346 Fax: 27-12-6647848

Email: [cjaconko@mweb.co.za](mailto:cjaconko@mweb.co.za)

### **Services Offered:**

- Diagnosis and management of FA patients and their families of all age groups;
- Coaching and counseling families towards a healthy and optimum lifestyle with FA;
- Coordination of consultations with other sub-specialties for management of FA-related disorders;
- Motivational speaking presentation on living a life with Fanconi with meaning and purpose;
- Workshops on: Lifestyle management for Fanconi families - empowering the patients to live their best life.

### **University of the Free State**

PO Box 339(G69)

Bloemfontein 9300, South Africa

- **Prof. D.K. Stones**, Paediatric Oncology and Haematology, Department of Paediatrics and Child Health

Telephone: 27-51-4052820

Mobile: 083-444-7233

Fax: 27-51-4443230

Email: [StonesDK@UFS.ac.za](mailto:StonesDK@UFS.ac.za)

### **Services Offered (Universitas Hospital, Bloemfontein and NHLS):**

- Genetic counseling service for families with FA;
- Diagnostic, carrier, preclinical and prenatal testing in conjunction with genetics department and NHLS.

## **SPAIN**

### **Hospital General Universitario Gregorio Marañón**

Pediatric Hematology

C/Doctor Esquerdo, 46

28007 Madrid, Spain

- **Dr. Cristina Belendez**

Telephone: 91-529-00-37

Email: [crisrina.belendez@salud.madrid.org](mailto:crisrina.belendez@salud.madrid.org)

## **Hospital Universitario de la Princesa**

C/Doctor Esquerdo, 45  
28028 Madrid, Spain

- **Dr. Ángela Figuera**  
Telephone: 91-520-24-31  
Email: [afiguera.hlpr@salud.madrid.org](mailto:afiguera.hlpr@salud.madrid.org)

## **Hospital Vall d'Hebron**

Pediatric Oncology  
P. de la Vall d'Hebron 119-129  
Barcelona 08035, Spain

- **Dr. Cristina Díaz de Heredia**  
Telephone: 93-488-30-93

## **University Hospital Niño Jesús**

Avenida Menéndez Pelayo, 65  
28009 Madrid, Spain

- **Julian Sevilla, MD**, Department of Hematology and Oncology  
Telephone: 91-503-5938 Fax: 91-503-5960  
Email: [jsevilla.hnjs@salud.madrid.org](mailto:jsevilla.hnjs@salud.madrid.org)
- **Dr. Marta Prudencio**, Department of Hematology and Oncology  
Telephone: 91-503-5938 Fax: 91-503-5960

## **CIEMAT/CIBERER**

Division of Hematopoietic Innovative Therapies (HIT)  
CIEMAT/ CIBER of Rare Diseases (CIBERER)  
IIS Jiménez Díaz Foundation  
Madrid, Spain  
[www.ciemat.es](http://www.ciemat.es)

Spanish Fanconi Anemia Network: [www.redfanconi.org](http://www.redfanconi.org)

- **Juan Bueren, PhD**, Head of Division of Hematopoietic Innovative Therapies (HIT)/CIBER of Rare Diseases (CIBERER)/IIS Jiménez Díaz Foundation  
Telephone: 34-91-346-6518  
Email: [juan.bueren@ciemat.es](mailto:juan.bueren@ciemat.es)
- **José A. Casado**, Senior Researcher of Division of Hematopoietic Innovative Therapies (HIT)/CIBER of Rare Diseases (CIBERER)/IIS Jiménez Díaz Foundation  
Telephone: 34-91-496-2525  
Email: [jose.casado@ciemat.es](mailto:jose.casado@ciemat.es)
- **Paula Río**, Researcher of Division of Hematopoietic Innovative Therapies (HIT)/CIBER of Rare Diseases (CIBERER)/IIS Jiménez Díaz Foundation  
Telephone: 34-91-346-0890  
Email: [paula.rio@ciemat.es](mailto:paula.rio@ciemat.es)

- **Susana Navarro**, Senior Researcher/CIBER of Rare Diseases (CIBERER)/IIS Jiménez Díaz Foundation  
Telephone: 34-91-346-0891  
Email: [s.navarro@ciemat.es](mailto:s.navarro@ciemat.es)
- **M<sup>a</sup> Luz Lozano**, Technical Assistance/CIBER of Rare Diseases (CIBERER)/IIS Jiménez Díaz Foundation  
Telephone: 34-91-346-6255  
Email: [luz.lozano@ciemat.es](mailto:luz.lozano@ciemat.es)
- **Aurora de la Cal**, Secretary, Spanish FA Research Network  
Telephone: 34-91-496-2518  
Email: [aurora.delacal@ciemat.es](mailto:aurora.delacal@ciemat.es)

### **Services Offered:**

- Coordination of the FA Spanish registry;
- Coordination of the FA gene therapy clinical trial;
- FA subtyping by retrovirus mediated gene transfer;
- Analysis of the hematopoietic reserve in bone marrow samples from FA.

## **Universitat Autònoma de Barcelona**

Chromosome Fragility Service (UAB-CFS)  
Department of Genetics and Microbiology  
08193 Bellaterra (Cerdanyola del Valles)  
Barcelona, Spain

<http://safcro.uab.es/>

Spanish Fanconi Anemia Network: [www.redfanconi.org](http://www.redfanconi.org)

- **Jordi Surrallés, PhD**, Professor of Genetics (UAB-CFS Scientific Director)  
Telephone: 34-935-811-830 Fax: 34-935-812-387  
Email: [jordi.surralles@uab.es](mailto:jordi.surralles@uab.es)
- **Maria Roser Pujol, PhD**, Cytogeneticist (UAB-CFS Technical Director). Accredited by Spanish Society of Human Genetics  
Fax: 34-935-812-387  
Email: [MaríaRoser.Pujol@uab.cat](mailto:MaríaRoser.Pujol@uab.cat)

### **Services Offered:**

- DEB chromosomal breakage testing in blood and skin fibroblasts;
- Diagnosis of mosaicism;
- MMC-induced G2/M-cell phase arrest assay;
- FANCD2 monoubiquitination and foci formation assays;
- Spanish contact for prenatal diagnosis;
- Genetic counseling;
- Maintenance of a biobank of Spanish FA patients with cryopreserved transformed cell lines and DNA.

Services for genetic analysis are offered free of charge to Spanish FA patients through the Spanish Fanconi Anemia Network, which is funded by the Spanish Ministry of Health. Chromosome fragility testing in blood lymphocytes or skin

fibroblast is free of charge to Spanish patients when the results are positive. The cost of the DEB test in blood lymphocytes to non-Spanish patients or to Spanish patients with a negative result is 290 euros. The cost of the DEB test in fibroblasts to non Spanish patients and to Spanish patients with a negative result is 450 euros.

## **TURKEY**

### **Hacettepe University**

Inherited Bone Marrow Failure Center  
Ankara, Turkey 06100

- **Sule Unal, MD**, Director, Associate Professor  
Telephone: +90 312 305 1170  
Email: [suleunal@hacettepe.edu.tr](mailto:suleunal@hacettepe.edu.tr)

#### **Services Offered:**

- NGS screening is available for genetic diagnosis of FA.
- FA patients are followed by experienced physicians in their fields. Each patient is seen by the same hematologist, ENT, O&G, plastic surgeon on visits. Both adult and pediatric patients are followed and the facility is approved by the Turkish Ministry of Health.
- Hematopoietic stem cell transplantation unit has good results with FA patients.

### **Istanbul University**

Bone Marrow Bank & Department of Medical Biology  
Istanbul Medical Faculty  
Çapa 34390 Istanbul, Turkey

- **Nevin Yalman, MD**, President, Fanconi Anemia Association, Professor, Pediatric Hematologist  
Telephone: 9-212-414-20-00, x 31208  
Fax: 9-212-635-11-68  
Email: [nevinyalman@gmail.com](mailto:nevinyalman@gmail.com)

## **UNITED KINGDOM**

### **Birmingham Children's Hospital**

Steelhouse Lane  
Birmingham, West Midlands B4 6NH, United Kingdom

- **Phil Darbyshire, MD**, Consultant Pediatric Hematologist  
Telephone: 44-121-333-9999  
Email: [Phil.Darbyshire@bch.nhs.uk](mailto:Phil.Darbyshire@bch.nhs.uk)

### **Bone Marrow Transplant & Late Effects Clinic**

Department of Haematology  
Royal Hallamshire Hospital  
Glossop Road  
Sheffield S10 2JF, United Kingdom

- **Professor John Snowden, BSc (Hons), MBChB, MD, FRCP, FRCPath**  
Consultant Haematologist  
Telephone: 0114-271-3357 Fax: 0114-271-3997  
Email: [John.Snowden@sth.nhs.uk](mailto:John.Snowden@sth.nhs.uk)
- **Dr Diana Greenfield, BSc (Hons), RN, PhD**  
Consultant Nurse  
Telephone: 0114-226-5592 Fax: c/o 0114-271-3754  
Email: [Diana.Greenfield@sth.nhs.uk](mailto:Diana.Greenfield@sth.nhs.uk)

#### **Services Offered:**

- The joint Bone Marrow Transplant and Late Effects Clinic provides adult Fanconi anaemia patients access to a multi-disciplinary team providing initial general assessment, confirmation of diagnosis and subsequent coordination of screening and support. The multi-disciplinary clinic provides access to other specialists at our hospital including: endocrinology, respiratory, gynaecology, urology, gastroenterology, maxillo-facial, radiology, genetics, nutrition, psychiatry, psychology and lifestyle advice. There is close transitional working with the paediatric services run by Prof. Ajay Vora at Sheffield Children's Hospital NHS Foundation Trust.
- Patients are assessed and reviewed in Sheffield with communication to their referring clinician. Nurse-led follow-up and support is recommended on an annual basis for all those patients who are medically stable.
- Where appropriate, haematopoietic stem cell transplantation can be arranged in Sheffield. Both the Sheffield Teaching Hospitals NHS Foundation Trust and Sheffield Childrens' Hospital are JACIE-accredited centres.

### **Bristol Royal Hospital for Children**

Oncology Day Beds  
Level 6  
Upper Maudlin Street  
Bristol, BS2 8BJ, United Kingdom

- **Colin Steward, MA, BM, BCh, PhD, FRCPCH, FRCP**, Department of Paediatric Haematology and Bone Marrow Transplantation  
Telephone: 44-117-3428044 Fax: 44-117-3420170  
Email: [colin.steward@uhbristol.nhs.uk](mailto:colin.steward@uhbristol.nhs.uk)

#### **Services Offered:**

- Comprehensive diagnostic and routine haematological care in specialized clinic;

- Coordination of complementation group analysis, mutation analysis and somatic mosaicism studies;
- Expertise in androgen and transplantation therapy;
- Specialists in genetics, haematology, bone marrow transplantation (from sibling, matched and mismatched unrelated, haploidentical and cord donors), endocrinology, gastroenterology and radiotherapy;
- 16-bed inpatient area and outpatient bone marrow transplantation clinic.

### **St. Mary's Hospital**

Praed Street, Paddington  
London W2 1NY, United Kingdom

- **Josu de la Fuente, PhD FRCP MRCPath FRCPa**  
Consultant Paediatric Hematologist  
Telephone: 44-20-788-66164  
Email: *Josu.delaFuente@imperial.nhs.uk*

### **Yorkshire Paediatric Haematology Unit**

Leeds General Infirmary  
Fanconi Anaemia – Multi-disciplinary Clinic  
Great George Street  
Leeds LS1 3EX, United Kingdom

- **Professor Sally E. Kinsey, MD**, Paediatric Haematologist

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#### **Services Offered:**

The Fanconi Anaemia multi-disciplinary clinic provides initial general assessment of cases and confirmation of diagnosis. The multi-disciplinary clinic provides direct access to various specialists.

Patients are assessed and reviewed in Leeds with communication to their referring clinician. Follow-up in the multi-disciplinary clinic is recommended on a yearly basis. Geographically local individuals can have routine follow-up (including blood counts, biochemistry, ultrasound examinations, bone marrow review etc) in Leeds. Individuals living more distant can have care coordinated via their referring clinician.

Where appropriate, work-up for stem cell transplantation can be arranged in Leeds.