International Fanconi Anemia
Treatment Resource Guide

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

- Dr. Julie Curtin, MD, PhD, Head Haematology Department
  Telephone: 612-9845-0000  Fax 612-9845-3332
  Email: juliec@chw.edu.au

- Professor Peter Shaw, Head BMT Service
  Telephone: 612-9845-0000  Fax: 612-9845-2171
  Email: peter.shaw@health.nsw.gov.au

- Ida O’Brien, Clinical Nurse Consultant BMT Follow Up
  Telephone: 612-9845-0000  Fax: 612-9845-2171
  Email: IdaO1@chw.edu.au

- A/Prof. Meredith Wilson, Clinical Geneticist
  Telephone: 612-9845-3273  Fax: 612-9845-3204
  Email: merediw@chw.edu.au

- A/Prof. David Little, Director Orthopaedic Research
  Telephone: 612-9845-3352  Fax: 612-9845-3180
  Email: DavidL3@chw.edu.au

- Professor Chris Cowell, Head Endocrinology Department
  Telephone: 612-9845-0000  Fax: 612-9845-3170
  Email: chrisc@chw.edu.au

- Dr. Carsten E. Palme, FRACS, Surgeon, Otolaryngology Head & Neck Surgery
  Telephone: 612-9845-3170  Fax: 612-9893-7440
  Email: Carsten_Palme@wsahs.nsw.gov.au

- Mark Schifter, Oral Medicine and Oral Pathology
  Telephone: 612-9845-8716  Fax: 612-9845-5740
  Email: mark_schifter@wsahs.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

Sydney Children’s Hospital
Blood & Marrow Transplant Program
Kids Cancer Centre
Sydney Children’s Hospital
High St, Randwick
Sydney, NSW, 2031, Australia
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 Anaemia Registry (Dr. Arleen Auerbach).

AUSTRIA

St. Anna Children’s Hospital
Department of Hematology and Oncology

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: tobrien@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

A/Prof. Richard Cohn, Head, Oncology, Kids Cancer Centre
Telephone: 02-93821731 Fax: 02-93821789
Email: 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

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

Services Offered:
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 Anaemia Registry (Dr. Arleen Auerbach).

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

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

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;
- Mutation analysis (FANCA, C and G) and FANCD2 by western blot.

CANADA

Hospital for Sick Children, University of Toronto
555 University Ave.
Toronto, Ontario, Canada M5G 1X8

Yigal Dror, MD, FRCP(C), Associate Professor of Paediatrics; Director, Marrow Failure and Myelodysplasia Program; Division of Hematology/Oncology; Scientist, Program in Cell Biology, Research Institute
Telephone: 416-813-5630 Fax: 416-813-5327
Email: 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

**Princess Margaret Hospital**
Ontario Cancer Institute
University of Toronto
610 University Ave
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

**University of Manitoba**
ON2021a-675 McDermot Ave.
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 or jdoyle2@hsc.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
Email: xsusan28@gmail.com

**COLOMBIA**

**Clínica Las Americas**
Diagonal 75 B #2 A80 Ofic. 520
Medellín
Antioquia, Colombia

• Dr. Amado Karduss, Adult Hematology/Stem Cell Transplantation
  Telephone: 57 4 3409314
  Email: amaka@une.net.co

• Dr. Yadira Coll, Pediatric Hematology
  Telephone: 57 4 3409314
  Email: yacoll@une.net.co

**Services Offered:**
- Evaluation;
- Medical treatment;
- FA stem cell transplantation.

**Dr. Adriana Linares**
Telephone: 571-343-6600 extension 1364, 1362, or 1280
Email: alinaresb@gmail.com

- **Clínica Colsubsidio**
  Carrera 24 # 63 - 50
  Bogotá, Colombia

- **National University of Colombia**
  Carrera 45 No 26-85
  Building Uriel Gutierrez
  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 Sébrurier
75935 Paris cedex 19, France

• Thierry Leblanc, MD, Department of Pediatric Hematology
  Telephone direct: 33-1-4403-4185
  Secretary: 33-1-7128-2535 Fax: 33-1-4403-4740
  Email: thierry.leblanc@rdb.aphp.fr

• Jean-Hugues Dalle, MD, PhD, Department of Pediatric Hematology
  Telephone direct: 33-1-4403-1241
  Secretary: 33-1-4403-5388 Fax: 33-1-4403-4740
  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);

Hôpital Saint-Louis
1, Ave. 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.socié@sls.aphp.fr

• Thierry Leblanc, MD, Department of Pediatric Hematology
  Telephone direct: 33-1-4249-9725
  Secretary: 33-1-4249-9721 Fax: 33-1-4249-9823
  Email: thierry.leblanc@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
  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
• Wolfram H. Ebell, MD
  Telephone: 49-30-450-566-014
  Fax: 49-30-450-566-919
  Email: wolfram.ebell@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

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

- 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

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 Dusseldorf, 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 Dusseldorf, 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

Services Offered:
- Evaluation of new patients;
- Hematopoietic stem cell transplantation from HLA-matched siblings, matched unrelated donors and haploididentical family donors;
- Co-transplantation of bone marrow with mesenchymal stem cells to enhance engraftment and to prevent graft-versus-host-disease (GvHD);
- Extracorporal 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: 91-44-2433-1741, ext. 2509

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 (in process of development);
- 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

**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.

**Sahyadri Hospital**
Pune’s Sahyadri Hospital (Deccan Gymkhana branch)
30C, Erandawane, Karve Road
Pune - 411 004, Maharashtra
India

- **Dr. Shashikant Apte**
  Telephone: 67213000 or 25403040
  Email: shashikant.apte@gmail.com

**IRAN**

**Tehran University of Medical Sciences**
Shariati Hospital
Hematology-Oncology and SCT Research Center
Kargar Ave.
14114, Tehran, Iran
[http://horcsct.tums.ac.ir](http://horcsct.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

- **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

**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

**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
Services Offered:

- Mutation detection for *FANCA*, *FANCC*, and *FANCG*, 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

- **Johanna Svahn**, MD, Hematology Unit  
  Telephone: 39-010-5636-694  Fax: 39-010-5636-714  
  Email: 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

**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 Tunour 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

- **Silverio Perrotta**, MD  
  Telephone and Fax: 39-0815-665421  
  Email: silverio.perrotta@unina2.it

- **Sofia Maria Rosaria Matarese**, MD  
  Telephone and Fax: 39-0815-665444  
  Email: Sofia.MariaRosaria.MATARESE@unina2.it

- **Francesca Rossi**, MD  
  Telephone and Fax: 39-0815-665342  
  Email: 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

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: yabe@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

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.

KENYA

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

- Dr. Grace W. Kitonyi MBChB, FRCPA
  Haematologist/Haematopathologist

Telephone: 254 20 2721815
Mobile phone: 254722385336
Email: gwkitonyi@yahoo.com
Cc: gkitonyi@uonbi.ac.ke

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: yabe@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

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 117
Ashrafieh, Beirut, Lebanon

- Dr. Roula Farah
  Associate Professor of Pediatrics and Pediatric Hematology/Oncology
  President, CHANCE Association
  Cell phone: 961-3-681-309
  Email: roula.fs@dm.net.lb

Services Offered:
- Complete clinical evaluation, treatment and follow-up of new and previously diagnosed FA patients;
- 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: 52 55 5281 0073 Fax: 52 55 5280 2942 Mobile: 52 55 2690 1450
Email: clarag@unam.mx or cgorodea@fundacioncompartevida.org.mx
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
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Marisol González, MD
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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

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

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: jjp.gille@vumc.nl

- Resie Vervenne, PhD
  Email: r.vervenne@vumc.nl

- Aggie Nieuwint, MSc
  Contact for chromosomal breakage testing.
  Email: awm.nieuwint@vumc.nl
Research:
Telephone: 31-20-444-8270 Fax: 31-20-444-8285

• Josephine C. Dorsman, PhD
  Email: jc.dorsman@vumc.nl

• Hans Joenje, PhD
  Email: joenje@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

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

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

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.

• Dr. Nyree Cole
  Telephone: 64 9 307 4919
  Email: 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

• Myungshin Kim, MD, PhD, Professor, Department of Laboratory Medicine
  Telephone: 82-2-2258-1645 Fax: 82-2-2258-1719
  Email: 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.
- 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

**International Patient Liaison Centre:**  
Telephone (24hr Helpline): (65) 67792777  
Email: 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, otorhinolaryngology, 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

**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

**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

**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: 2772495346 Fax: 27-12-6647848  
  Email: 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
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: cbelendez.hgugm@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

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
- 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 Diaz Foundation
Madrid, Spain

www.ciemat.es
Spanish Fanconi Anemia Network: www.redfanconi.org
- Juan Bueren, PhD, Head of Division of Hematopoietic Innovative Therapies (HIT)/CIBER of Rare Diseases (CIBERER)/IIS Jiménez Diaz Foundation
  Telephone: 34-91-346-6518
  Email: juan.bueren@ciemat.es
- José A. Casado, Senior Researcher of Division of Hematopoietic Innovative Therapies (HIT)/CIBER of Rare Diseases (CIBERER)/IIS Jiménez Diaz Foundation
  Telephone: 34-91-496-2525
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- Paula Río, Researcher of Division of Hematopoietic Innovative Therapies (HIT)/CIBER of Rare Diseases (CIBERER)/IIS Jiménez Diaz Foundation
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  Email: paula.rio@ciemat.es
- Susana Navarro, Senior Researcher/CIBER of Rare Diseases (CIBERER)/IIS Jiménez Diaz Foundation
  Telephone: 34-91-346-0891
  Email: s.navarro@ciemat.es
- Mª Luz Lozano, Technical Assistance/CIBER of Rare Diseases (CIBERER)/IIS Jiménez Diaz Foundation
  Telephone: 34-91-346-6255
  Email: luz.lozano@ciemat.es
- Aurora de la Cal, Secretary, Spanish FA Research Network
  Telephone: 34-91-496-2518
  Email: 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
- 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
- Maria Roser Pujol, PhD, Cytogeneticist (UAB-CFS Technical Director). Accredited by Spanish Society of Human Genetics
  Fax: 34-935-812-387
  Email: MariaRoser.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

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

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

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

- 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

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

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 MRCPCH FRCPath**
  Consultant Pediatric 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

**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.

- **Ben Sykes**, Nurse Co-ordinator
  Telephone: 0113-392-7379  Fax: 0113-392-6629
  Email: benjamin.sykes@leedsth.nhs.uk

Telephone: 0113-392-8191  Fax: 0113-392-8488
Email: sally.kinsey@leedsth.nhs.uk