

# International Fanconi Anemia Treatment Resource Guide

*Editors' Note and Disclaimer: The resources provided in this guide are not meant to be inclusive. The reader should not consider the resources listed here to be endorsements by the Fanconi Anemia Research Fund, Inc.*

## 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, PhD**, Head Haematology Department  
Telephone: 612-9845-0000 Fax 612-9845-3332  
E-mail: [juliec@chw.edu.au](mailto:juliec@chw.edu.au)
- **Professor Peter Shaw**, Head BMT Service  
Telephone: 612-9845-0000 Fax: 612-9845-2171  
E-mail: [peters@chw.edu.au](mailto:peters@chw.edu.au)
- **Ida O'Brien**, BMT Follow up Clinical Nurse Consultant  
Telephone: 612-9845-0000 Fax: 612-9845-2171
- **Dr. Meredith Wilson**, Clinical Geneticist & Head Department of Clinical Genetics  
Telephone: 612-9845-3273 Fax: 612-9845-3204
- **Dr. Michael Bellmore**, Head Orthopaedic Surgery  
Telephone: 612-9845-0000 Fax: 612-9845-2702  
E-mail: [MichaeB3@chw.edu.au](mailto:MichaeB3@chw.edu.au)
- **Professor Chris Cowell**, Head Endocrinology Department  
Telephone: 612-9845-0000 Fax: 612-9845-3170  
E-mail: [chrisc@chw.edu.au](mailto:chrisc@chw.edu.au)
- **Dr. Carsten E. Palme, FRACS**, Surgeon, Otolaryngology Head & Neck Surgery  
Telephone: 612-9845-6025 Fax: 612-9893-7440  
E-mail: [Cartsen\\_Palme@wsahs.nsw.gov.au](mailto:Cartsen_Palme@wsahs.nsw.gov.au)
- **Mark Schifter**, Oral Medicine and Oral Pathology  
Telephone: 612-9845-8716 Fax: 612-9845-5740  
E-mail: [mark\\_schifter@wsahs.nsw.gov.au](mailto:mark_schifter@wsahs.nsw.gov.au)

A 350 bed tertiary care paediatric institution, providing comprehensive care for the majority of children in NSW with all diagnoses. This includes a comprehensive team of medical professionals working in disease-specific multidisciplinary teams to manage complex disorders with multisystem manifestations, such as Fanconi anaemia. We offer comprehensive management of new and previously diagnosed FA patients, including:

- ◆ Diagnostic testing and screenings of siblings;
- ◆ Genetic counselling: coordination of complementation studies or gene testing via overseas facilities to identify causative mutations in affected individual discussion of family planning options for future pregnancies, including prenatal diagnosis or IVF-pre-implantation genetic diagnosis for those with identified mutations;
- ◆ Orthopaedic management, including specialist hand, upper limb and congenital hand clinic;
- ◆ Haematological management (Julie Curtin);
- ◆ Blood and marrow transplantation (Peter Shaw): comprehensive program of related, unrelated and haploidentical marrow, PBSC and cord blood transplant;

*(Continued on the next page)*

## AUSTRALIA, cont'd

### The Children's Hospital at Westmead Sydney, cont'd

- ◆ Co-ordinated long term follow up and ongoing surveillance, with particular reference to BMT follow up (Ida O'Brien) oral (Mark Schifter), Head and neck cancer screening (Carsten Palme) 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

- **Tina Carter, FRACP, PhD**, Clinical Director of Bone Marrow Transplantation Program  
Telephone: 08-9340-8222  
E-mail: [Tina.Carter@health.wa.gov.au](mailto:Tina.Carter@health.wa.gov.au)

### Sydney Children's Hospital

Cord & Marrow Transplant Program

High Street Randwick, 2031

Sydney, NSW Australia

- **Tracey O'Brien, FRACP, MBChB, MHL, BSc**, Program Director, Paediatric and Adolescent Oncologist/Hematologist  
Telephone: 61-2-93821733 Fax: 61-2-93821789  
E-mail: [Tracey.O'Brien@sesiahs.health.nsw.gov.au](mailto:Tracey.O'Brien@sesiahs.health.nsw.gov.au)

#### **Services Offered:**

Sydney Children's Hospital is a large tertiary pediatric hospital. Over 750 pediatric transplants have been performed at the center since establishment of the transplant program in 1975. The center is considered a national leader in unrelated marrow and cord transplants with particular interest in transplant for non-malignant diseases. The center director has specialized training and expertise in treatment of Fanconi anemia offering a well established multi-disciplinary comprehensive pre- and post-transplant service for patients with FA.

## **BRAZIL:**

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

Rua General Carneiro, 181 - 15º andar  
80001-970-Curitiba-PR, Brazil

- **Carmem Bonfim, MD**, Director of Pediatric Stem Cell Transplant Program, Stem Cell Transplant in Genetic Diseases, Coordinator of Fanconi Anemia Outpatient Clinic  
Telephone: 55-41-3360-1007 Fax 55-41-3360-1015  
E-mail: *carmem\_bonfim@terra.com.br*
- **Ricardo Pasquini, MD, PhD**  
Telephone: 55-41-3360-1007 Fax: 55-41-3360-1015  
E-mail: *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.

## **CANADA:**

### **The Hospital for Sick Children, The University of Toronto**

555 University Avenue  
Toronto, Ontario, Canada M5G 1X8

- **Yigal Dror, MD, FRCP(C)**, Associate Professor of Paediatrics, Division of Hematology/Oncology; Director, Marrow Failure and Myelodysplasia Program; Scientist, Program in Cell Biology, Research Institute  
Telephone: (416) 813-5630 Fax: (416) 813-5327  
E-mail: *yigal.dror@sickkids.ca*
- **Adam Gassas, MBChB, MSc, MRCP, DCH, MRCPCH**, Assistant Professor of Pediatrics; Division of Hematology/Oncology/Blood and Marrow Transplant Program  
Telephone: (416) 813-8227 Fax: (416) 813-5327  
E-mail: *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, U of T  
Telephone: (416) 946-2266 Fax: (416) 946-6585  
E-mail: *Jeff.Lipton@uhn.on.ca*

## FRANCE:

### 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  
E-mail: [gerard.socie@sls.aphp.fr](mailto:gerard.socie@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  
E-mail: [thierry.leblanc@sls.aphp.fr](mailto: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  
E-mail: [jean.soulier@sls.aphp.fr](mailto:jean.soulier@sls.aphp.fr)

#### Services Offered:

- ◆ 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  
E-mail: [wolfram.ebell@charite.de](mailto:wolfram.ebell@charite.de)

#### Services Offered:

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

*(Germany continued on the next page)*

## GERMANY, cont'd:

### Heinrich Heine University Medical Center

Children's Hospital

Moorenstrasse 5

40225 Düsseldorf, Germany

- **Helmut Hanenberg, MD**, Department of Pediatric Hematology, Oncology and Clinical Immunology, Department of Pediatrics, Wells Center for Pediatric Research, Indiana University School of Medicine, Indianapolis, IN  
Telephone: 49-211-811-6103 or -6568 Fax: 49-211-811-9436  
E-mail: *Hanenberg@uni-duesseldorf.de*
- **Eunike Velleuer, MD**, Department of Pediatric Hematology, Oncology and Clinical Immunology  
Telephone: 49-211-811-7662 Fax: 49-211-811-7634  
E-mail: *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  
E-mail: *lesch@med.uni-duesseldorf.de*

#### Services Offered:

- ◆ Primary diagnosis, blood work, transfusions and bone marrow harvests of pediatric patients with hematological, malignant or immunological disorders (also for the National Marrow Donor Program - NMDP);
- ◆ Autologous and allogenic stem cell transplantation;
- ◆ FA multidisciplinary out- and in-patient care;
- ◆ FA complementation group analysis on peripheral blood T-cells, bone marrow cells, fibroblasts and B-cell lines for groups A, B, C, D1, D2, E, F, G, I, J, L, M, N;
- ◆ Close collaboration with Detlev Schindler, University of Würzburg, on FA-related clinical and scientific issues;
- ◆ Cooperation with Ruud Brakenhoff, University of Amsterdam, on "Progression of FA-SCC and Development of a Non-Invasive Screening Method for Precursor Lesions";
- ◆ Preclinical testing of alternative treatment and gene therapy approaches in animal models in cooperation with Wade Clapp, Laura Haneline, Karen Pollok and Kenneth Cornetta at the Indiana University School of Medicine, Indianapolis, IN;
- ◆ Close cooperation with the two German FA family support groups: Deutsche FA-Hilfe e.V. (Ralf Dietrich) and Aktionskreis Fanconi Anemia e.V.

## INDIA:

### Apollo Specialty Hospital

320 "Padma Complex"

Anna Salai

Chennai 600 035, India

*refain@vsnl.net*

- **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  
E-mail: *vbaburao@hotmail.com*

#### Services Offered:

- ◆ Chromosomal breakage study using MMC and DEB test;
- ◆ Cytogenetic study from bone marrow and FISH analysis;
- ◆ FANCD2 western blot.

## IRAN:

### Hematology-Oncology and SCT Research Center

Tehran University of Medical Sciences

Shariati Hospital

Kargar Ave.

14114, Tehran, Iran

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

- **Ardehsir Ghavamzadeh, MD**, Professor of Medicine, Adult Hematologist Oncologist, President of Hematology-Oncology and SCT Research Center  
Telephone: 98-21-84902635 Fax: 98-21-88004140  
E-mail: [ghavamza@sina.tums.ac.ir](mailto:ghavamza@sina.tums.ac.ir)
- **Amir Ali Hamidieh, MD**, Assistant Professor of Medicine, Pediatric Hematologist Oncologist, Director of Fanconi Anemia Transplant Program  
Telephone: 98-21-84902645 Fax: 98-21-88004140  
E-mail: [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, haploidentical and cord blood stem cell transplants.

## 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  
E-mail: [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 (NST). Stem cell transplantation can also be considered from half-matched (haploidentically mismatched) family member while avoiding the risk of graft-versus-host disease.

## ITALY:

### G. Gaslini Children's Institute

Largo G.Gaslini 5  
16147 Genova, Italy

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

#### Services Offered:

- ◆ Primary diagnosis with DEB test, Cell Cycle analysis and MMC sensitivity, D2 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 (University of Dusseldorf, Germany) and Dr. Juan Bueren (CIEMAT, Madrid, Spain) for remaining FA complementation group analyses;
- ◆ Collaboration with Prof Anna Savoia, IRCSS Burlo Garofolo, University of Trieste Italy, for molecular diagnosis of FA;
- ◆ Haematopoiesis Laboratory (cell cultures, cytokine hypersensitivity tests);
- ◆ Cytogenetic Laboratory;
- ◆ Close scientific collaboration with Professor Grover Bagby (OHSU, 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;
- ◆ 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.

(Italy continued on the next page)

## ITALY, cont'd:

### 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  
E-mail: *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  
Ishigaura, Kanagawa, 259-1193, Japan

- **Hiromasa Yabe, MD**, Stem Cell Transplantation  
Telephone: 81-463-93-1121 Fax: 81-463-95-9567  
E-mail: *yabeh@is.icc.u-tokai.ac.jp*
- **Miharu Yabe, MD**, Stem Cell Transplantation  
Telephone: 81-463-93-1121 Fax: 81-463-93-8607  
E-mail: *miharu@is.icc.u-tokai.ac.jp*
- **Shunichi Kato, MD**, Stem Cell Transplantation  
Telephone: 81-463-93-1121 Fax: 81-463-91-6235  
E-mail: *skato@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;
- ◆ Umbilical cord blood bank and cell processing center.

## 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  
E-mail: *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.

## SINGAPORE:

### National University Health System, Singapore

Paediatric Haematology/ Oncology/ BMT

University Children's Medical Institute

5 Lower Kent Ridge Road

Singapore 119074

Republic of Singapore

*www.nuh.com.sg/\_kids/index.htm*

#### Physicians:

- **Thuan-Chong Quah, MBBS**  
E-mail: *paqtc@nus.edu.sg*
- **Allen Yeoh Eng-Juh, MBBS**  
E-mail: *paeyej@nus.edu.sg*
- **Poh-Lin Tan, MBBS**  
E-mail: *paev6@nus.edu.sg*
- **LeLe Aung, MD**  
E-mail: *paev2@nus.edu.sg*
- **Pei-Lin Koh, MBBS**  
E-mail: *paev30@nus.edu.sg*

#### BMT Nurse Clinician:

- **Laura Tan**  
E-mail: *Laura\_Tan@nuh.com.sg*

#### BMT Coordinator:

- **Zin Mar Myo**  
E-mail: *Zin\_Mar\_Myo@nuh.com.sg*

#### International Patient Liaison Centre:

Telephone (24hr Helpline): (65) 67792777

E-mail: *iplc@nhg.com.sg*

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## SINGAPORE, cont'd

### National University Health System, Singapore, cont'd

#### Services Offered:

- ◆ Paediatric Haematology/ Oncology/ Blood and Marrow Transplantation;
- ◆ Paediatric Subspecialties;
- ◆ Comprehensive Laboratory Service;
- ◆ Stem Cell Laboratory.

## SOUTH AFRICA:

### Chris-Hani Baragwanath Hospital

Division of Haematology and Oncology

- **Dr. Linda Wainwright**  
Clinic Telephone: 0800333555 Office Telephone: 0119339594  
Email: *rosalind.wainwright@wits.ac.za*

### Johannesburg Hospital

Division of Haematology and Oncology

Dept. of Pediatrics

Private Bag X39

Johannesburg 2000

South Africa

- **Prof J 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, and University of the Witwatersrand

PO Box 1038

Johannesburg 2000

South Africa

- **Dr. Amanda Krause**  
Telephone: 27-11-489-9219/9224 Fax: 27-11-489-9226  
Email: *amanda.krause@nhls.ac.za*

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

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

### Unitas Hospital

Pediatric Oncology/Haematology Unit

Box 12552

Lyttleton 0140, Gauteng

Pretoria, South Africa

- **Charmaine Jacobs, MD**, Pediatric Oncologist  
Telephone: 01-267-780-76 Mobile Phone: 27-82-82-07435  
E-mail: *cjaconko@mweb.co.za*

## SPAIN:

### 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  
E-mail: [jsevilla@salud.madrid.org](mailto:jsevilla@salud.madrid.org)

### Universitat Autònoma de Barcelona

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

Web: [www.uab.es/surralleslab](http://www.uab.es/surralleslab)

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

- **Jordi Surrallés, PhD**, Full Professor of Genetics (UAB-CFS Scientific Director)  
Telephone: 34-935-811-830 Fax: 34-935-812-387  
E-mail: [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  
E-mail: [MaríaRoser.Pujol@uab.cat](mailto:MaríaRoser.Pujol@uab.cat)

#### Services Offered:

- ◆ DEB chromosomal breakage testing;
- ◆ Diagnosis of mosaicism;
- ◆ MMC-induced G2/M-cell phase arrest assay;
- ◆ FANCD2 monoubiquitination assay;
- ◆ Spanish contact for prenatal diagnosis;
- ◆ FA subtyping by retrovirus mediated gene transfer, in collaboration with Dr. J.A. Bueren's laboratory at CIEMAT, Madrid;
- ◆ Genetic counseling;
- ◆ Maintenance of a database integrating genetic and clinical data;
- ◆ Maintenance of a biobank of Spanish FA patients with cryopreserved transformed cell lines and DNA;
- ◆ An FA registry with approximately 170 (Spanish) families.

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.

# THE NETHERLANDS:

## Leiden University Medical Center

PO Box 9600

Leiden, 2300 RC, The Netherlands

- **Elisabeth T. Korthof, MD**, Department of Pediatric Stem Cell Transplantation, Pediatric Hemato-Oncologist  
Telephone: 31-71-526-4131 Fax: 31-71-524-8198  
E-mail: *E.T.Korthof@lumc.nl*

## UMC Utrecht

Kinderarts - Hematoloog

Wilhelmina Kinderziekenhuis

Postbus 85090

3508 AB Utrecht

Huispostnr KC 03.063.0, The 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 The Netherlands.  
Telephone: 31 (0)88 7554344 Fax: 31 (0)88 7555350  
E-mail: *m.bierings@umcutrecht.nl*

## VU University Medical Center Amsterdam

Department of Clinical Genetics

Van der Boechorststraat 7

NL-1081 BT Amsterdam

The Netherlands

### **Diagnosis:**

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

### **Contacts:**

- **Hans Gille, PhD**  
E-mail: *jpp.gille@vumc.nl*
- **Resie Vervenne, PhD**  
E-mail: *r.vervenne@vumc.nl*

### **Research:**

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

### **Contacts:**

- **Josephine C. Dorsman, PhD**  
E-mail: *jc.dorsman@vumc.nl*
- **Hans Joenje, PhD**  
E-mail: *joenje@vumc.nl*
- **Johan P. de Winter, PhD**  
E-mail: *j.dewinter@vumc.nl*

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## THE NETHERLANDS, cont'd

### VU Medical Center Amsterdam, cont'd

#### Services Offered:

#### Diagnosis:

- ◆ Diagnosis is by chromosomal breakage analysis, including mosaicism;
- ◆ Molecular diagnosis by mutation screening of all known FA genes;
- ◆ Prenatal diagnosis;
- ◆ Genetic counseling.

#### 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 role in sporadic cancer.

#### Collaboration in The Netherlands:

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

## TURKEY:

### İstanbul University

Bone Marrow Bank & Department of Medical Biology, Istanbul Medical Faculty  
Çapa 34390 İstanbul, 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  
E-mail: *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  
E-mail: *Phil.Darbyshire@bch.nhs.uk*

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

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: 44-117-3428628  
E-mail: *colin.steward@uhbristol.nhs.uk*

#### **Services Offered:**

- ◆ Comprehensive diagnostic and routine haematological care in specialized clinic;
- ◆ Coordination of complementation group analysis, mutation screening, somatic mosaicism studies and other genetic testing;
- ◆ 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, MRCP, MRCPC, FRCPath**, Consultant Pediatric Hematologist  
Telephone: 44-20-788-66164  
E-mail: *Josu.delaFuente@imperial.nhs.uk*

## UNITED KINGDOM, cont'd:

### Yorkshire Paediatric Haematology Unit

St James's University Hospital  
Fanconi Anaemia - Multidisciplinary Clinic  
Beckett Street  
Leeds LS9 7TF United Kingdom

- **Professor Sally E Kinsey, MD**, Paediatric Haematologist  
Telephone: 0113-206-4986 Fax: 0113-247-0248  
E-mail: *sally.kinsey@leedsth.nhs.uk*
- **Ben Sykes**, Nurse Co-ordinator  
Telephone: 0113-206-4986 Fax: 0113-247-0248  
E-mail: *benjamin.sykes@leedsth.nhs.uk*

#### **Services Offered:**

The Fanconi Anaemia multidisciplinary clinic provides initial general assessment of cases and confirmation of diagnosis. The multidisciplinary clinic provides direct access to specialists in: haematology, nephrology, genetics, nutrition, dentistry, psychology and life-style advice. In addition there is access to other specialists: hand surgery, hepatology, gynaecology, head and neck surgery, endocrinology and urology.

Patients are assessed and reviewed in Leeds with communication to their referring clinician. Follow up in the multidisciplinary 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 co-ordinated via their referring clinician.

Where appropriate work-up for stem cell transplantation can be arranged in Leeds. The Yorkshire Paediatric Haematology department is a JACIE accredited Bone Marrow Transplant Unit.