

Fanconi Anemia: Guidelines for Diagnosis and Management

Fourth Edition • 2014



Fanconi Anemia
RESEARCH FUND, INC.

Fanconi Anemia: Guidelines for Diagnosis and Management, Fourth Edition, a publication of the Fanconi Anemia Research Fund, is the result of a Consensus Conference held in Herndon, Va., April 5-6, 2013. It replaces earlier editions published in 1999, 2003, and 2008. These guidelines are published for physicians who provide care for FA patients, and for patients and families who wish to secure optimal treatment by improving their understanding of all facets of Fanconi anemia, medical consultation, and appropriate referral.

These guidelines begin with detailed information on diagnosis and evaluation of FA. Subsequent chapters examine more specific health issues faced by persons with FA, followed by two chapters on psychosocial considerations that bear upon the well-being of the person with FA and his or her extended family. The guidelines conclude with a comprehensive checklist and diagnostic criteria for physicians and medical specialists.

These guidelines are commended in the profound hope that they will better serve the lives of patients who have this serious and life-threatening disorder.

“Dave and Lynn Frohnmayer [through the Fanconi Anemia Research Fund] have really transformed this world that they entered.”

—Francis Collins, MD, PhD, Director, National Institutes of Health
Winner, National Medal of Science, 2009

“The Fanconi Anemia Research Fund is the single best orphan disease research support group in the world. It is a model for demonstrating how a small group of dedicated families can speed the pace of scientific progress and bring realistic hope to patients with an otherwise fatal disorder.”

—David Nathan, MD, Distinguished Professor, Harvard Medical School
former Director, Dana-Farber Cancer Institute; Winner, National Medal of Science, 1990

“The tireless efforts of the Fanconi Anemia Research Fund have demonstrated that a rare genetic disorder can provide a vital key to understanding the basic biological process of cancer itself. The importance of these efforts to the advancement of medical science cannot be overstated.”

—Lee Hartwell, PhD, President Emeritus, Fred Hutchinson Cancer Research Center
Winner, Nobel Prize 2001

Lynn and Dave Frohnmayer started the Fanconi Anemia Research Fund, Inc. in 1989 to find effective treatments and a cure for Fanconi anemia and to provide education and support services to affected families worldwide.

The Fanconi Anemia Research Fund:

- Awards research grants. FARF has funded more than 105 investigators at 56 institutions totaling more than \$17 million.
- Publishes clinical, scientific, and family support information. These publications reach thousands of researchers, physicians, and families worldwide.
- Convenes an annual international scientific symposium at which researchers from around the world present research results.
- Provides invaluable support to families worldwide, including online support groups, annual family meetings, and meetings for adults with FA.

Fanconi Anemia Research Fund
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