



Fanconi Hope Conference

Beth Ann Lee

The Registry and How to Interact with Me And The Late Effects Study

How Did I Become Involved with Fanconi Hope?



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For Patients and Families



Ask Beth – our Fanconi Hope
FA Coordinator

We aim to provide support for families and individuals affected by FA by providing information and also guidance on where additional support and other services may be found. We now have a UK FA Coordinator, Beth Lee, to help you in dealing with all aspects of Fanconi Anaemia. Please email Beth and she will either reply by email or arrange to call you at a mutually convenient time, whichever you prefer.

beth.lee@fanconihope.org

You can read more about Beth Lee, her role and the Study itself [here](#)

Fanconi Hope works in partnership with the US-based **Fanconi Anemia Research Fund (FARF)**, the largest and longest standing FA support organisation and shares many of their resources. Please refer to their [website](#) for further information.

Please note that **Fanconi Hope** is run by a number of FA parents in their own time. There are no employed administrative staff or professional counsellors. We are happy to be contacted by email/phone and to share our experiences. However, we are

not in a position to provide professional counselling, advice, or ongoing support, and as with all FA-affected families, we are also having to deal with the personal challenges that FA presents.

There are many options available for free professional support, counselling and other services in the UK. See [Support Available](#) page for more information.

Key documents you should know about:

- Clinical research and liaison nurse for Fanconi Hope charity
- One day per week
- Families can email me at **beth.lee@fanconihope.org**

- **Dr Stefan Meyer**

Paediatric Oncology

University of Manchester

Royal Manchester Children's and Christie Hospital

Clinician Trustee - Fanconi Hope

- **Dr Marc Tischkowitz**

Department of Medical Genetics, University of Cambridge

- **Dr Kate Chandler**

Genetic Medicine, Manchester

Reasons For The Study ?

- We believe there are over 300 individuals in the UK with Fanconi Anaemia.
- It is evolving from a rare and serious paediatric condition to a condition affecting a considerable number of younger and increasingly older adults.
- Not much is known about what happens in the long term and how problems are managed.

Therefore we want to collect data about the genetic problem and clinical course

- From this data we will get a much better idea of the problems affecting individuals with FA during their lifetime and how best to manage them .

The studies objectives are:

- Over time establish a database of clinical and genetic data of FA patients in the UK ...the UK FA registry
- Evaluate current care pathways and management of FA in children and adults.
- Collect data from Fanconi patients over time in order to further improve clinical care.
- Enable communication for international clinical work (see what medical approach works best).

Data Capture Form

Data capture FA study:

Name DOB Place Lead clinician

Age at Diagnosis:

Mutations:

Consanguinity: yes/no

Family History:

Physical findings

Height

Microcephaly OFC

CNS MRI findings

|

Skin abnormalities

Thumb/radial abnormalities: Details

Other skeletal abnormalities

GI abnormalities

Renal abnormalities:

Who can take part?

- All patients with a diagnosis of FA can be included in the study.
- Patients and or parents will need to sign a consent.
- Patients can withdraw from the study at any time, just need to inform me.
- If you are interested in the study take the opportunity to talk with me or the consultants about it and we will answer any questions you may have.

During the study

- We would be pleased if you would keep in contact from time to time and let me know your progress.
- I would contact you 6 monthly to yearly over several years to document changes and keep the records up to date.
- All the data will be recorded on password protected hardware.
- This will only work if clinicians and patients work together.

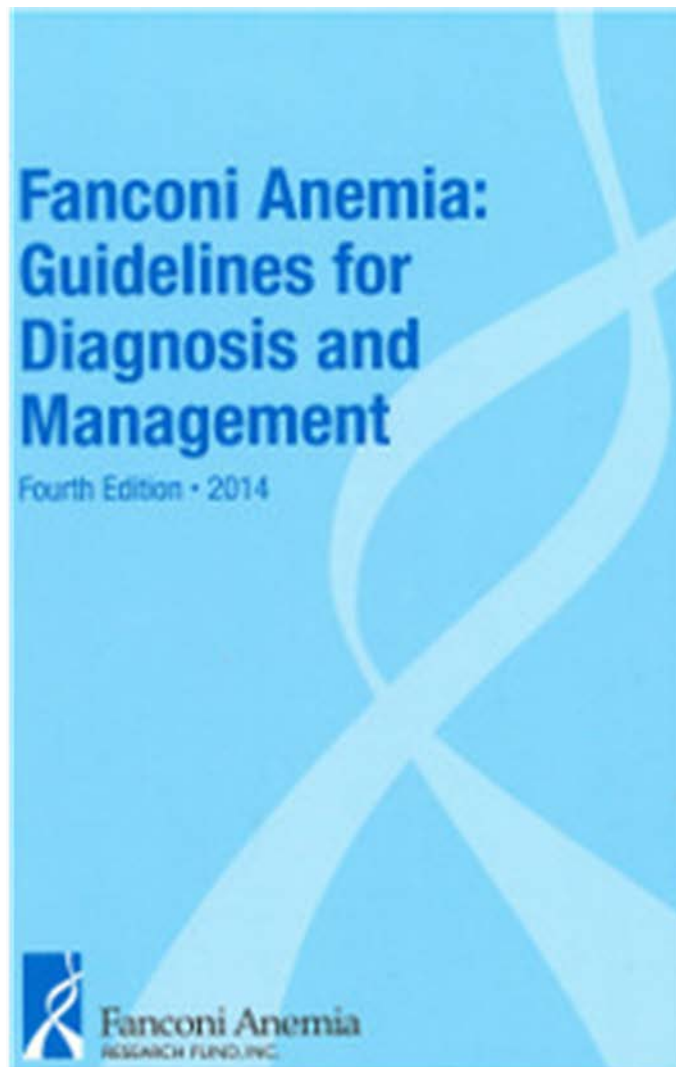


Thank you for listening, Any Questions ??

Beth Ann Lee

What families Say to Me...

- Sometime I feel like our lives revolve around FA.
- It would be nice if there was an end to it.
- Sometimes I don't give enough attention to my other children.
- I feel lonely and isolated, no one understands.
- I hate coming to the hospital.
- I have to try and stop myself from being over-protective.
- I feel guilty and responsible for the disease.



- Record age of BMT.
- Record surveillance e.g. ENT screening , upper GI screening, Gynae screening.
- Record Management of endocrine issues.
- Record Management of adult cancers in FA .
- Record Outcome of therapeutic interventions.

- Families can email with any concerns or questions.
- We can arrange a phone call if desired.
- Give advice and information.
- Support families.
- Collect data for the Study.

- Even though there are Guidelines for the management of FA these mainly address FA in childhood.
- The extent to which these guidelines are implemented is not known and may vary significantly between UK centres– we are finding out this is the case
- We need to learn more about the best management of FA in adults.

- Collaborative study led via the Charity and Patients.
- Enable translational research ("translate" findings in fundamental **research** into medical practice and meaningful health outcomes)
- Enable pooling of data with other FA studies