REFAIN Registry for Fanconi Anaemia in India

What is Fanconi Anaemia?

Fanconi Anaemia (FA) is a bone marrow failure disorder with an increased risk of malignancy. It is autosomal recessive in the majority and X-linked in a small percentage of cases. Children with FA may have major or minor malformations. Though the original description was based on radial ray deformity, we now know that FA can be associated with malformation in any system. Over 90% of the children develop aplastic anaemia between the ages of 5 to 10 years. Others may present with acute myeloid leukemia (AML) or other malignancies. Diagnosis of FA is confirmed by induced chromosomal breakage test (diepoxybutane / mitomycin C test)

Purpose of early diagnosis

Though FA is a rare disorder, it is the most commonly inherited aplastic anaemia. Consanguinity in certain parts of South India and Tamil Nadu, Andhra Pradesh and Karnataka is high and hence a number of children have been identified with FA. In the last 15 years, over 143 children with FA have been identified and followed up. The only cure for FA at present is bone marrow transplantation (BMT). The next option would be the use of androgens or immunosuppressive agents, but the response is either poor or short lived in most cases.

When should FA be suspected?

- Children or young adults with aplastic anaemia or myelodysplasia
- Children with birth defects
- Patients in whom karyotype analysis reveals increased number of spontaneous breaks
- Patients who are unusually sensitive to Chemotherapy or Radiotherapy

Aim of the Registry

- To disseminate information regarding FA
- To study the spectrum of the disease in Indian children
- To provide access to information on current treatment and follow-up guidelines
- To serve as a foundation for collaborative studies
- To provide family support
- To provide antenatal diagnosis

Diagnostic clinical features of FA in Indian children

Data from REFAIN

Clinical Features	%
Mean age	7.6 yrs
Consanguinity	64%
Aplastic Anaemia	97
Hyper- pigmentation	96
"Fanconi facies"	65
Café au lait spots	78
Thenar hypoplasia	74
Small for date	63
Radial ray anomaly	56
Renal anomaly	29
Ear Anomaly/Impaired hearing	21
Hypo-pigmentation	15
Cardiac anomaly	13
Short stature (-2SD)	11
Microcephaly (-2SD)	13

Would you like to become a member of REFAIN?

Please fill the following details, tear along the dotted lines and send this form to the address given behind

Name:	
Qualification:	
Speciality:	
Area of interest:	
Organisation / Private practice:	
Address:	
Phone:	
e-mail:	

REFAIN Registry for Fanconi Anaemia in India

About the Registry

The registry for Fanconi Anaemia is based at the Apollo Speciality Hospitals, Chennai. We aim to liaise with other existing registries of FA and help raise awareness among medical fraternity in India about FA and its management and link with centres abroad.

Patron

Dr Prathap Reddy
Chairman, Apollo Hospitals Enterprise Ltd.

Committee members of the Registry

Dr Sheila Mohan
Apollo Speciality Hospital, Chennai

Dr Revathi Raj
Apollo Speciality Hospital, Chennai

Dr V Pushpa
Southern Railway Hospital, Chennai

Dr M Venkatadesikalu
Voluntary Health Services Medical Centre, Chennai

Dr V Thilagavathi
ICH & HC, Chennai

Mrs & Mr. Adeshir
Will serve as the lead family for FA in India.
F & I Bone Marrow Foundation
24, Ratanbai Tata Building
38th Road, Bandra
Mumbai - 4000 050
E-mail: fandibmf@vsnl.net

What should a physician do when FA is suspected?

Kindly fill the **PROFORMA** and send it to the registry
Do confirmatory test for FA, if possible
Management of the patient will be in the hands of the treating physician

How to contact us?

Dr. Revathi Raj
Paediatric Haematologist
Apollo Speciality Hospital
320, 'Padma Complex', Anna Salai
Chennai- 600 035. India

Phone: 044 24331741. **Extn:** 2509
E-mail: refain@vsnl.net
Timings: Monday to Saturday, 8:00 am to 4:00 pm

Dr. Sheila Mohan
Paediatrician & Clinical Geneticist
Durgabai Deshmukh General Hospital and Research Centre
Durgabai Deshmukh Road,
Chennai 600 028. India

Phone: 044 24938311
Mobile: (0) 9282114661
E-mail: refain@vsnl.net
Timings: Monday to Friday, 9:00 am to 1:00pm

URL: www.refain.org

Would you like to become a member of REFAIN?
After filling the details given behind, tear along the dotted lines and send this form to the following address

Dr. Revathi Raj
Paediatric Haematologist
Apollo Speciality Hospital
320, 'Padma Complex'
Anna Salai
Chennai- 600 035
India

Physician's Proforma

Kindly fill up the Proforma as accurately as possible and send it to the registry address given behind by post.
Please perform the confirmatory test for FA if available at your centre.

Name in full	
Date	/ / (yyyy/mm/dd)
Age	
Sex	
Male	
Female	
Date of birth	/ / (yyyy/mm/dd)
Address (include parents name)	
Phone number	E-mail

Consanguinity (Pedigree if available)	
History of FA in family (Details, if available)	
History of malignancy in family (Details, if available)	

Anthropometry at birth	Weight	Kgs
	Length	Cms
	Head Circumference	Cms

Complaints

History of bleeds	☐	Skin	☐	Not Observed
	☐	Mucous membrane	☐	Not Observed

History of infections	
Durations of symptoms	

Examination	Weight		Kgs			
	Length		Cms			
	Circumference		Cms			
Anaemia	☐	Present	☐	Absent	☐	Not Observed
Signs of infection	☐	Present	☐	Absent	☐	Not Observed

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Physician's Proforma

Signs of bleed	☐ Skin	☐ Not Observed	
	☐ Mucous membrane	☐ Not Observed	
Pigmentation	☐ Skin	☐ Not Observed	
	☐ Tongue	☐ Not Observed	
	☐ Café au lait spots	☐ Not Observed	
	☐ Hypopigmentation	☐ Not Observed	
Facial dysmorphism (if present, please describe)			
Microphthalmia	☐ Present	☐ Absent	☐ Not Observed
Abnormal Ears	☐ Present	☐ Absent	☐ Not Observed
Hearing Disability	☐ Present	☐ Absent	☐ Not Observed
Hand anomalies	☐ Present	☐ Absent	☐ Not Observed
Thumbs	☐ Absent	☐ Hypoplastic	☐ Not Observed
		☐ Polydactyly	☐ Not Observed
Thenar hypoplasia	☐ Right	☐ Left	☐ Not Observed
Absence of radial pulses	☐ Right	☐ Left	☐ Not Observed
Investigations (X-rays may be avoided)			
Complete Blood Count	Hb	TC	DC	Plateletes
Foetal haemoglobin			
Bone marrow study			
Ultrasonography for renal anomaly			
ECHO/Doppler for cardiac anomaly			
Chromosome breakage study (spontaneous & / or induced)			
Any other relevant investigations			
Transfusions, if any, and last date of transfusion			
Name of the Physician			
Speciality			
Address			
Phone			
Fax		E-mail	

Thank you for filling the form. Please post it to the following address: Dr. Revathi Raj, Paediatric Haematologist
Apollo Speciality Hospital, 320 'Padma Complex',Anna Salai, Chennai - 600035

For further follow-up details you can E-mail to refain@vsnl.net (or) write to the above given postal address.