
بسم الله الرحمن الرحيم

Establishment of Hemoglobinopathies Registry

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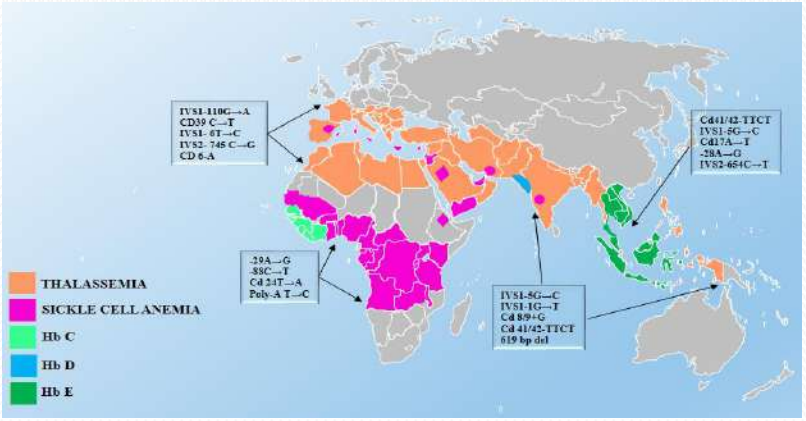
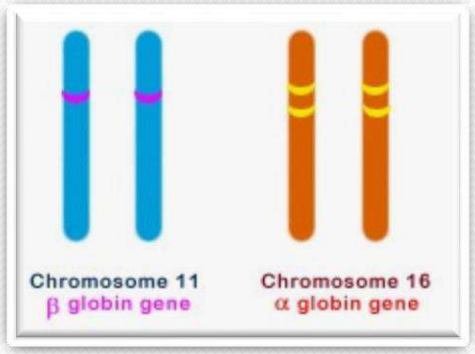
Hemoglobinopathies

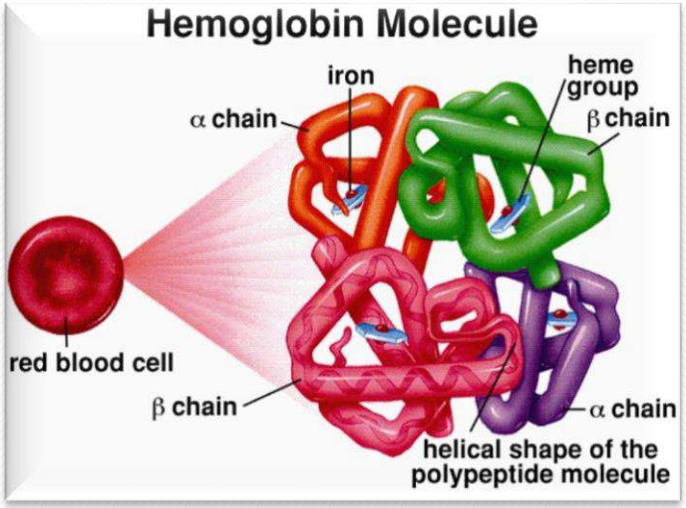
- Are a group of blood disorders affecting Red Blood Cells
- Include Thalassemias and Sickle Cell Disease
- Over 5% of world population is healthy carriers of Hemoglobinopathies
- 300,000 babies are born with hemoglobinopathies all over the world
- 60,000-70,000 thalassemia major Disease



Thalassemia

- Heterogeneous group of hereditary, hemoglobin disorders
- Genetic deficiency of one or more globin chains
- α thalassemia (due to deficiency of α genes)
- β thalassemia (due to mutation of β genes)





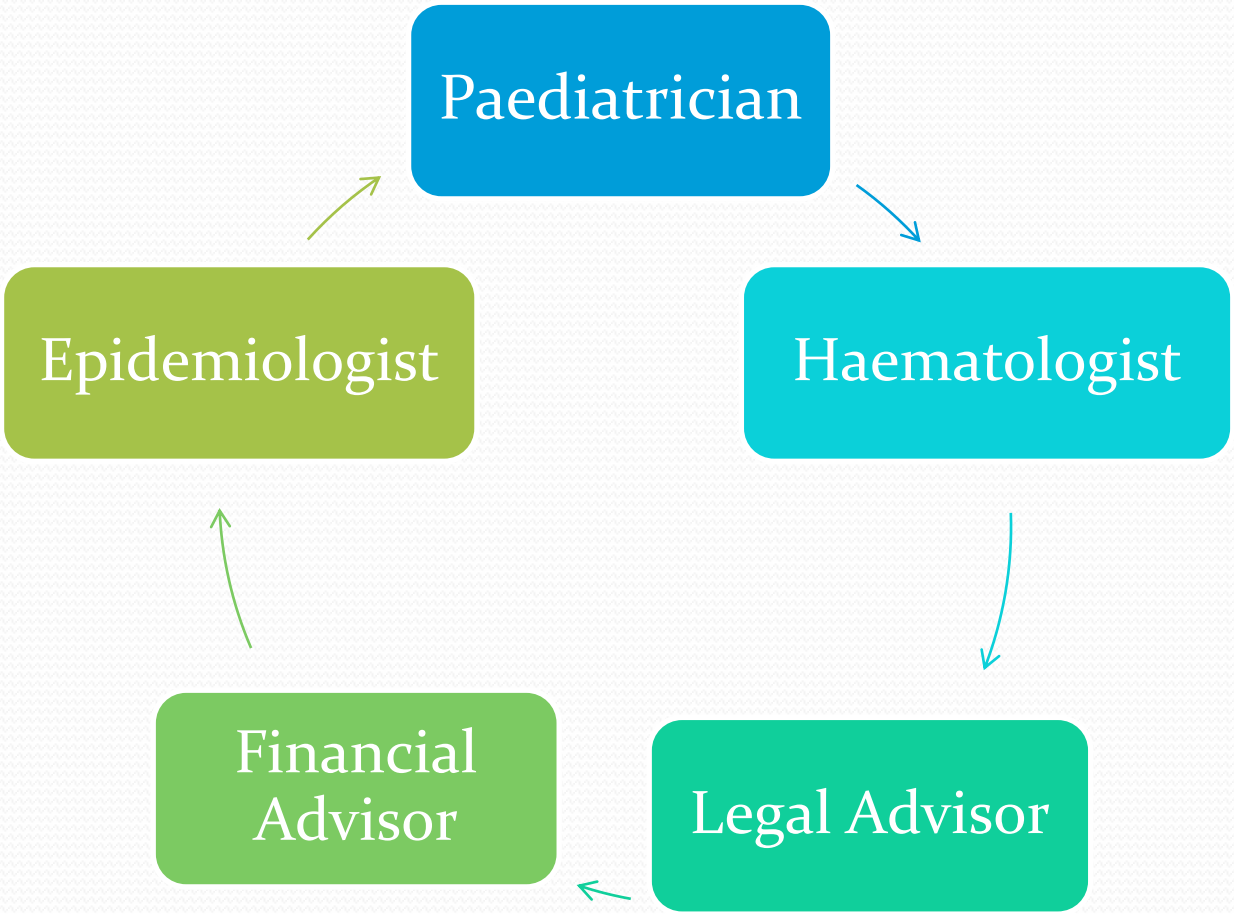
Thalassemia in Pakistan

- 6000 new cases born per year
- 500 new cases/month, means 17 new cases /day
- 60,000 or more registered patients
- No definite record of registered patients is available throughout Pakistan

Objectives

- To recognize thalassemia & other hemoglobinopathies as a common health problem & to find the exact disease burden
- To find the exact number of Thalassemics so that optimum treatment facilities could be provided to them at their home town
- To find out complications of the disease with facility of diagnosis and treatment of these complications
- It will serve as electronic medical file for thalassemics and will be available at every thalassemia treatment center

Governing Body



Geographical Definition

- At this phase, the registry is for Punjab
- All districts of Punjab will be included
- Names of hospitals & thalassemia centers will be available
- It will be covering other

provinces of Pakistan in steps

Material & Methods

- All thalassemics & persons with other hemoglobinopathies will be registered
- Inclusion Criteria
  1. Diagnosed cases of Thalassemia Major
  2. Diagnosed cases of Thalassemia Intermedia
  3. Diagnosed cases of other hemoglobinopathies
  4. Domicile of Punjab
- Exclusion Criteria
  1. Un diagnosed cases (Investigations offered for diagnosis)
  2. Any blood disorder other than hemoglobinopathies
  3. Domicile of other provinces of Pakistan

Preparation of Registry

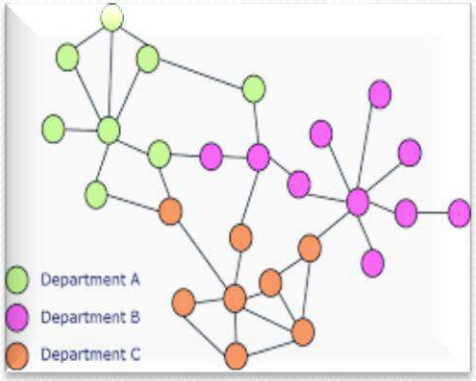
- With the help of Punjab Information Technology Board (PITB)
- Approval from Ethical Committee of FJMU
- Case report form for each thalassemic will be filled with consent form
- The system will be able to detect repeated registries for any thalassemic receiving health care in more than one center

Confidentiality

- Different levels of access to data will be defined
- Thalassemics can access to their own data
- The data cannot be edited by them
- A dedicated operator will have access to the data in each organization. He will be authorize to edit the data of his organization
- Central Administration has access to all data
- Central Administration will be responsible to create Statistical reports

Links

- Software has links to
  1. Thalassemia Federation of Pakistan
  2. All related organizations
  3. Punjab Health Department
  4. Punjab Blood Bank Authority
  5. Punjab Thalassemia Prevention Project
  6. Punjab Information Technology Board



Data Collection of Centers

- All the centers invited to fill an electronic form for the collection of following information

  1. Center size (classification depending upon No. of registered Thalassemics)

- Centers size

  Small (Less then 50)

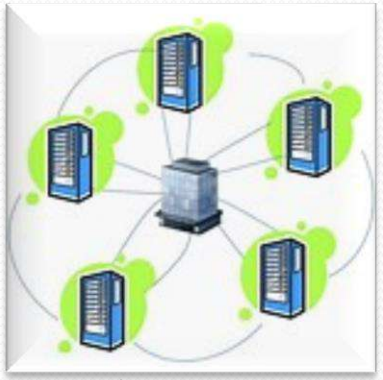
  Medium (51-100)

  Large (101-500)

  Very Large (more than 500)

Data Collection of Centers

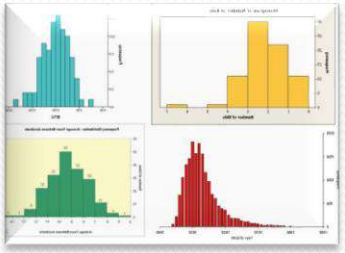
- Availability of clinical, diagnostic & monitoring services including
    a. Transfusion Services
    b. Standard Laboratory tests
    c. Ferratin Assessment
    d. Standard medication availability



- Specialized departments & Laboratories (Hepatology, Cardiology, Endocrinology, Orthopaedics, Gastroenterology, etc)
- Iron overload monitoring facilities (cardiac & liver MRI, liver biopsy)
- Paediatric Clinical Units
- Adult Clinical Units

Data Collection of Thalassemics

- Data collection forms in Urdu
- Demographics of Thalassemics
- Disease characteristics of Thalassemics
- Management of disease by regular and adequate Red Cell Transfusions



- Close monitoring of Iron Overload
- Appropriate iron Chelation Therapy
- Assess the efficacy of Haemoglobinopathy Control Program

Quality Control of Filled Forms

- Admin is able to randomly select 5% of registered Thalassemics to data control
- Directions for filling the forms will be available in a dedicated part

- There will be short directions in each part upon pause of the cursor
- Data base is accessible only for statistical reports by Headquarter
- Language of program is English
- Technical problems & security issues will be covered by PITB

Demographic Information

Date of Entry	
File Number	
National ID card # (Father)	
National ID card # (Mother)	
Name of Thalassemic	
National ID card # (Personal)	
Family Number	
Gender	Male Female
Date of Birth	
Caste/Ethinicity	
District	
Tehsil	
Address	

Extended Screening

- Date of Genetic Counselling of Parents
- Extended Carrier Screening Offered
- Date of Genetic Counselling & Screening of the Family
- Number of Family Members Screened
- Number of Carriers Detected (Male/Female)
- Number of Non Carriers (Male/Female)
- CVS facility availed by the at risk couples

Results

- District wise distribution of thalassemics

District	Tehsil	Centers (n)	Patients (n)

Results

- Diagnosis of registered Thalassemics

Diagnosis	Number	%
β- Thalassemia Major		
β- Thalassemia Intermedia		
β/S- Thalassemia		
β/E- Thalassemia		
HbH Disease		
S/S Disease		

Results

- The most common mutations of β- Thalassemia

β gene Mutation	Homozygous	Compound Heterozygous	Total β Allele	%

Physical Examination

Date of 1st Physical Examination	
Weight	
Height	
Hepatomegaly	
Splenomegaly	
Facial disfigurement	
Spleenectomy	
Growth Failure	

Important Thalassemia Complications

Date of Lab Results	
Bile Stones	
Diabetes Mellitus	
Cardiomyoathy	
Cardiac Arrythemias	
Hypothroidism	
Hypoparathyroidism	
Osteoporosis	
Pubertal Disorders	
Fertility Disorders	

Laboratory Data

Date		
Mean Pre transfusion Hb		gr/dl
Fasting Blood Sugar		mg/ml
S. Ferratin		ng/ml
S. Calcium		mg/dl
AST		u/l
ALT		u/l
Phosphorus		mg/dl
Blood Urea		mg/dl
S. Uric Acid		mg/dl

Annual Laboratory Data

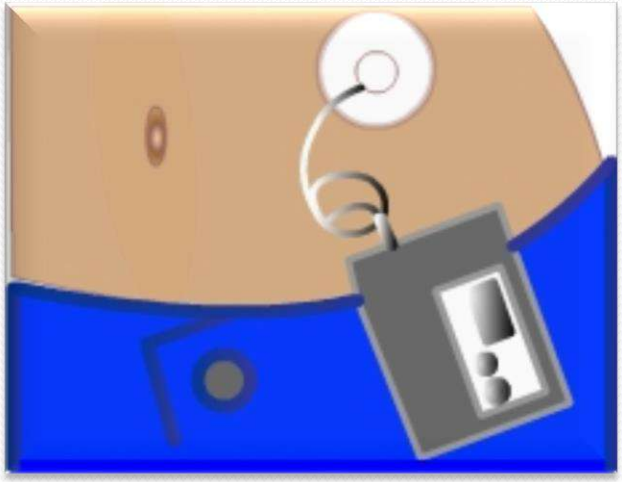
Date of Lab Result		
TSH		
FBS		
2hrs after breakfast		
OGTT		
HCV Antibody		
HIV Antibody		
HBS Antigen		
Zinc		
Vit D		
PTH		
MRI T2*		

List of Medicines

- ☐ Calcium
- ☐ Calcitriol
- ☐ Digoxin
- ☐ Estrogens
- ☐ Folic Acid
- ☐ Insulin
- ☐ Levothyroxine
- ☐ Oral Hypoglycaemic
- ☐ Penicillin
- ☐ Progesterone
- ☐ Testosterone
- ☐ Vit D

Iron Chelators

- DFO
- DFX
- DEF



Conclusion

- This registry will help to calculate the exact disease burden
- Will help to find the birth rate of new thalassemic's
- Will be helpful to optimize thalassemics management
- Help in implementing the prevention program
- To achieve further improvement in survival & quality of life of thalassemics
- Will be a source for health system decision makers


Questions?

THANK YOU

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