A Multi-dimensional Approach towards Comprehensive Care and Prevention of Sickle Cell Disease in India: Experience from CSIR-Sickle Cell Anaemia Mission

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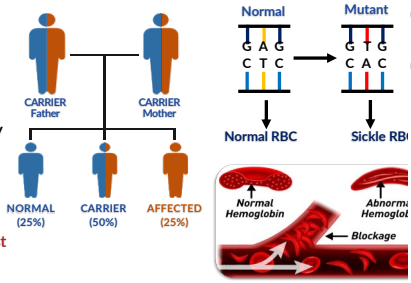
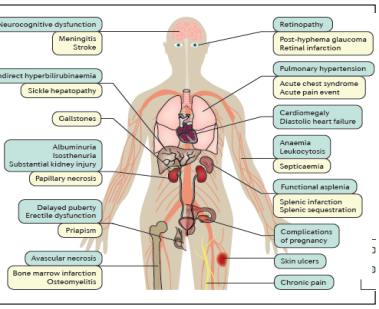
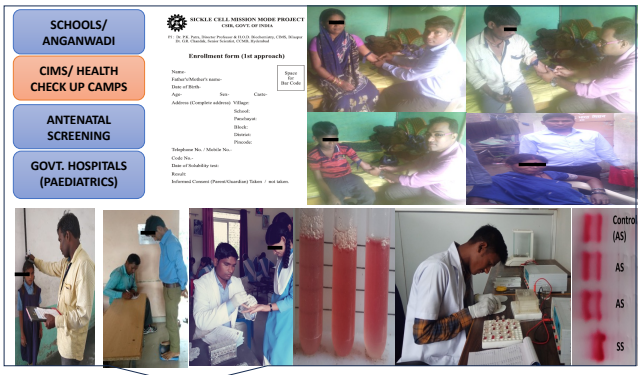
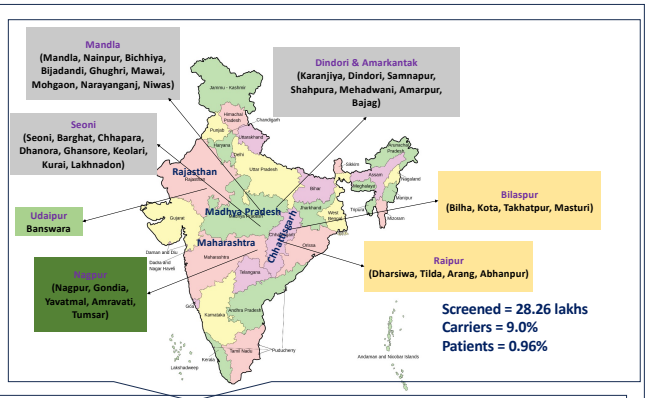
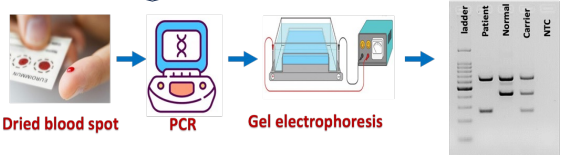
- A genetic disease due to a point mutation in beta globin gene
- Leads to sickling of RBCs in low oxygen state and early destruction
- Affects multiple organs in the body; No permanent cure
- India has high disease burden (8-10%), both general and tribal people
- Phenotypic heterogeneity & variable response to Hydroxyurea therapy
- Early diagnosis with proper supervision improves quality of life
- Cost effective high-throughput screening system is key to early diagnosis, leading to preventive and management strategies
- Genetic testing, prenatal diagnosis and genetic counselling is the surest way to eradicate the disease





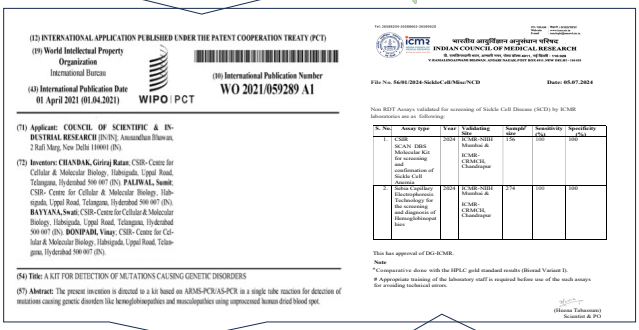






		WB/DBS-PCR			
		AA	AT	TT	Total
HPLC	AA	209	2	0	211
	AS	4	5882	26	5912
	SS	0	26	850	876
	Total	213	5910	876	6999

Sensitivity	99%
Specificity	98%
Positive Predictive Value	99%
Negative Predictive Value	98%
Accuracy	99%



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(54) Title: A KIT FOR DETECTION OF MUTATIONS CAUSING GENETIC DISORDERS

(57) Abstract: The present invention is directed to a kit based on ARMS-PCR/AS-PCR in a single tube reaction for detection of mutations causing genetic disorders like hemoglobinopathies and musculopathies using unprocessed human dried blood spot.

भारतीय आयुर्विज्ञान अनुसंधान परिषद
INDIAN COUNCIL OF MEDICAL RESEARCH

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Non RDT Assays validated for screening of Sickle Cell Disease (SCD) by ICMR laboratories are as following:

S. No.	Assay type	Year	Validating Site	Sample size	Sensitivity (%)	Specificity (%)
1.	CSIR-IGIB's DBS Molecular Kit for screening and confirmation of Sickle Cell Anemia	2024	ICMR-NIHI Mumbai & ICMR-CRMCHL Chandrapur	156	100	100
2.	Sebia's Capillary Electrophoresis Technology for the screening and confirmation of Hemoglobinopathies.	2024	ICMR-NIHI Mumbai & ICMR-CRMCHL Chandrapur	274	100	100

This has approval of DG-ICMR.

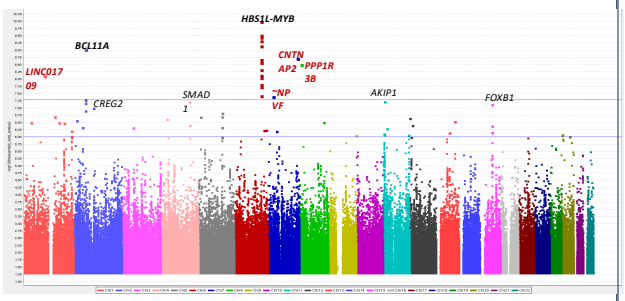
Note

* Comparative done with the HPLC gold standard results (Biorad Variant I).

Appropriate training of the laboratory staff is required before use of the such assays for avoiding technical errors.

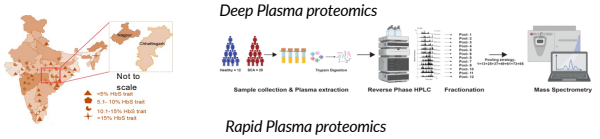
(Heena Tabassum)
Scientist & PO

Genome-Wide Association Study of Hb F levels

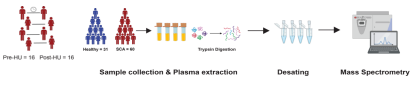


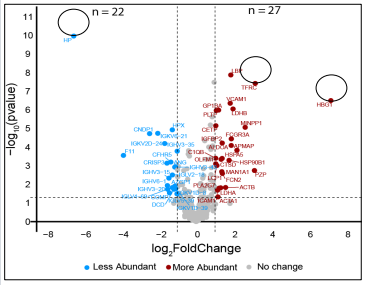
Plasma proteomics for identification of Biomarkers

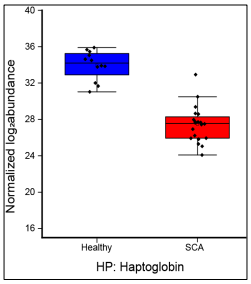
Deep Plasma proteomics



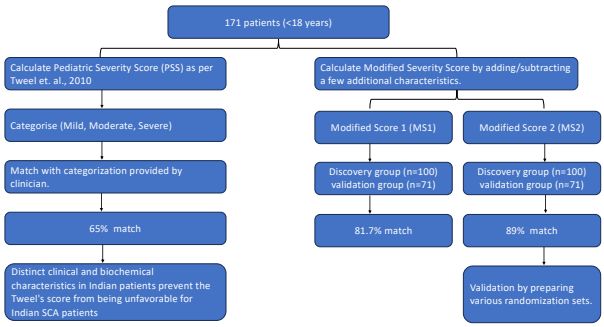
Rapid Plasma proteomics







Generation of Disease Severity Score for Indian SCA patients



Strategy for SCA eradication