ARISE - African Research and Innovative Initiative for Sickle cell Education: Improving Research Capacity for Service Improvement

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Domains of the ARISE action and main aims



An EU funded project launched in 2019 with the aims:

- to see a universal newborn screening and early infant diagnosis programme successfully implemented and effectively embedded within regional and national policies in SSA
- to build African personnel capacity to achieve excellence in science and a sustainable research capacity using SCD projects as learning laboratories

ARISE project

Specific objectives	Work Programme	Output		
✓ To evaluate the	Scientific coordination and project managed	160 staff members		
 prevalence of SCD in target countries ✓ To establish laboratory diagnosis and quality assurance systems ✓ To test the feasibility for establishing newborn and early infant screening for SCD ✓ To develop best practices in clinical management for acute and chronic complications in SCD 	eHealth technologies to support a Newborn screening programme [FGB]	Disse	 With improved ability: ✓ implementation science 	
	Improving laboratory diagnostics and quality assurance systems for population screening [RCPath/INSERM]	 ✓ epidemiology and statistics ✓ clinical management 		
	Newborn SCD screening, screening for neurocognitive complications, clinical care and antibiotic prophylaxis [GSTT]	equiremer Ind Comm	 ✓ clinical management of SCD ✓ genetic counselling ✓ TCD 	
	Training in molecular diagnostic and genetic counselling and conducting epidemiological and genetic research [CING]	nts [FGB] unication [GS	 ✓ research protocols development ✓ communication 	
	Training and support for clinical research	TT]		

[LSHTM]

The partnership



Staff exchange program



Training

On-site

- 24 secondments focused on different themes related to SCD diagnosis and management
- Training topics: clinical management protocols, genetic counselling, data analysis to estimate the burden of SCD in Nigeria

Virtual training

- Virtual Training Learning Programme launched in April 2020 in response to the COVID-19 lockdown travel restrictions
- Open webinars with international experts on SCD
- Temporary replacement of f2f learning
- Very high attendance and appreciation rates
- Resources freely available on the ARISE website





On-site training



Virtual events enlarged the ARISE training chances



https://www.ariseinitiative.org/education-training/arise-virtual-teaching-programme/

Joint research



A combined gap analysis and baseline needs assessment to implement a laboratory improvement plan (WP3)



Analysis on country-specific ethical and legal requirements for the processing of personal and health data and the handling of human samples in Nigeria and Kenya (WP2)



"Pilot study to map and assess selected clinical centres managing SCD patients" in Nigeria (WP2)



"Pilot study to assess the needs of SCD patients and healthcare workers involved in SCD management" in Nigeria (WP2)



Epidemiological surveys and data collection on haemoglobinopathies, including a questionnaire for the collection of data in epidemiological studies (WP5)



Assessment of genetic and haematological modifiers of disease severity among patients with Sickle Cell Disease in Kaduna state, Northern Nigeria (WP5)

Research projects on "Molecular diagnostics for haemoglobinopathies", "Genotypephenotype correlations and genetic modifiers of haemoglobinopathies" (WP5) Ethical-regulatory support/oversight for all activities



Laboratory Gap Analysis and baseline assessment



ARISE

A survey to define laboratory capacity and **areas for improvements** has been prepared and delivered twice (in 2019 and 2021) to laboratories in SSA Countries

- There is the need for each laboratory to progress towards an application for accreditation.
- Early standardisation and rebranding of documents previously labelled as SOPs.
- An understanding that SOPs need to be updated via a document management system.
- QA leadership for each laboratory identified prior to interim assessment.



Analysis of the organisation of healthcare centres managing Sickle Cell Disease in Nigeria (pilot)

Background

In the framework of the European research project "African Research and Innovative initiative for Sickle cell Education: Improving Research Capacity for Service Improvement" – ARISE project, a <u>cross-sectional</u>, web-based pilot survey was designed to map and assess the organisation of Nigerian centres managing <u>Sickle Cell Disease (SCD) patients</u>. To achieve effective NBS, it is essential to describe the state of SCD centres that will provide ongoing care for infants diagnosed with SCD

Methods

The questionnaire for the online survey was developed in collaboration with the ARISE Steering Committee in line with published criteria for comprehensive SCD management. It investigated facilities' characteristics including SCD management, screening services, counselling and education. Questionnaire was administered to selected facilities (16, including paediatrics and adults' services) using an online system (Limesurvey). SPSS software was used for the analysis.

National Health Research Ethics Committee of Nigeria approval number for this study: NHREC/01/01/2007-27/07/2020

Results



10 answers were received, from 2 secondary and 8 tertiary care facilities. They include University of Abuja Teaching Hospital (UATH), University of Ilorin Teaching Hospital (UITH).and both adults and paediatrics facilities from Ahmadu Bello Teaching Hospital (ABUTH), Barau Dikko Teaching Hospital (BDTH) General Hospital Kafanchan (GHK), Federal Medical Centre, Katsina (FMC Katsina).

Results show that:

- there is <u>no online SCD patient registry</u> at the investigated facilities
- all the facilities perform basic laboratory tests on SCD patients
- most of facilities (9/10) adopt the National Guidelines for the control and management of SCD patients
- The percentage of coverage of individuals screened receiving follow up education/counseling/treatment ranges from 45% to 56%, mostly adults.

Screening services are paid out of pocket by the patients in 8 facilities or sponsored by other non-government organisations in 2.

Concerning test type for SCD screening, all facilities perform Alkaline Gel Electrophoresis, more than one half blood film. Full details of test type is available in fig.2

Screening test Facilities	Sickling test	Haemogl obin Solubility Test/ Sickledex	Alkaline Gel EF	Acid gel EF	Blood film	HPLC	IEF
UATH			+		+	•	
UITH	-	•	+	•		•	•
ABUTH (adult)			+		+	•	
ABUTH (paed)	-		+		+	+	
BDTH (adult)			+		+	•	
BDTH (paed)			+			•	
GHK (adult)	-		+			•	
GHK (paed)		•	+	•		•	•
FMC (adult)	+	+	+	-	+		-
FMC (paed)			+		+		

Fig. 2 type of tests performed in the investigated facilities

Only 1 of these 10 centres of excellence has SCD screening services delivered by a community staff. 5 centres delivered SCD screening services as part of special campaigns and 6 as part of maternal and child health care services (fig. 1)



Conclusion and next steps

There is potential for an excellent management of SCD patients in Nigeria.

<u>Improvements are possible</u>, especially in terms of online registries to assess data for planning and research, health insurance and good policy to encourage patients to attend the clinics.. After this initial pilot testing, the survey will be reviewed and extended to cover around 70% of Nigerian centres managing SCD patients Results presented at ASCAT 2020 and published in the Hemasphere Journal

To be extended during 2022-23



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https://journals.lww.com/hemasphere/Documents/Abstracts%20of%20ASCAT_EHA_BSH%202020.pdf



Pilot study to assess the needs of patients and health workers involved in SCD management

satisfied
 dissatisfied



Results presented at ASCAT 2020 and published in the Hemasphere Journal

To be extended during 2022-23

https://journals.lww.com/hemasphere/Documents/Abstracts%20of%20ASCAT_EHA_BSH%202020.pdf

Molecular biology-based research projects

Molecular diagnostics for haemoglobinopathies

- Diagnosis
- Prenatal diagnosis by CVS
- Preimplantation genetic testing
- Diagnostic laboratory organisation

Genome-wide association study (GWAS) of haemoglobinopathy phenotypes

Multi-ethnic genotyping array using a customdesigned component Epidemiological surveys and data collection on haemoglobinopathies

- Healthcare infrastructure and policies
- Screening and services
- Disease prevalence and incidence
- Frequency of pathogenic variants

Protocols finalised and shared with the ethics team





Implementation Science

IS is aimed to find how do we get "what works" to the people who need it, with greater speed, fidelity, efficiency, quality, and relevant coverage



IS tools are used to translate research health benefits to patient communities and to enhance the engagement of community, public and policy stakeholders

Advocacy visit in Nigeria in 2020



https://www.ariseinitiative.org/search/?_sf_s=implementation%20science

Built-in return mechanism



TRAIN THE TRAINER WORKSHOP-September 2019,

ABUJA

- Venue: The laboratory practical session took place at the International Foundation Against Infectious Disease (IFAIN) Laboratory.
- Attendees: Fifty delegates were nominated by their institutional leads to attend the workshop
- Faculty: 27 experts, renowned for their expertise and contribution to improving lives of patients living with Sickle Cell disease delivered lectures.

Full event report is available here: <u>https://www.ariseinitiative.org/wp-</u> <u>content/uploads/2020/11/Final-train-the-trainer-</u> <u>brochure.pdf</u>

NBS Programme

2 NBS programmes implemented in Zambia and Kaduna State – Nigeria, supported by the Consortium on Newborn Screening in Africa for SCD (**CONSA**)

Objectives:

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- To establish yearly screening of 10,000 newborns for SCD
- To establish a shared early intervention and follow-up clinical protocol
- To establish a SCD registry

Kaduna

- More than 3,000 samples collected
- 3 facilities involved in samples collection, 2 laboratories for data analysis
- About 2% of positive subjects
- Periodic training and group meetings

Zambia

- NBS integrated in the Expanded Programme on Immunization and the HIV Early Infant Diagnosis Programme
- 3 facilities involved in the screening (ADCH, UTHs-CH and UTHs-WNBH)
- Samples collected (DBS) are tested at TDCR lab by using the Isoelectric focusing (IEF) machine

Networking

ARISE

HemaSphere

ARISE and INHERENT

Networking

- The primary aim of INHERENT is to study the role of genetic modifiers in hemoglobinopathies through a large-scale, multiethnic genome-wide association study (GWAS)
- ARISE is one of the 9 founding consortia acknowledging the need of addressing challenges of previous studies related to small sample sizes and low statistical power → the target sample size is at least 30,000 individuals with hemoglobinopathies
- INHERENT membership currently includes >110 organizations, spanning 44 countries worldwide
- INHERENT is led by the Cyprus Institute of Neurology and Genetics, that is also leader of ARISE Work Package 5
- Participation in INHERENT is open for any group that can submit a minimum of 30 DNA samples with their core phenotypic description (contact <u>petrosk@cing.ac.cy</u>)

https://www.inherentnetwork.org/

Kountouris P, Stephanou C, Archer N, Bonifazi F, et al. The International Hemoglobinopathy Research Network (INHERENT): An international initiative to study the role of genetic modifiers in hemoglobinopathies. Am J Hematol. 2021 Nov 1;96(11):E416-E420. doi: 10.1002/ajh.26323

What's next?

What's next?

Keeping the focus on SCD management and NBS, we have planned:

- \circ to move the leadership from Europe to Africa,
- to change our approach from the individual capacity building to African generated evidence base for policy and practice,
- to expand the educational programme establishing PhD and Masters courses,

In conclusion, we aim at exploiting the potential of clinicians and researchers trained for improved SCD management and NBS

Thanks a lot!

This presentation reflects only the author(s)'s view and the EU Research Executive Agency (REA) is not responsible for any use that may be made of the information it contains.

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