The Kaduna State Newborn Screening Program: Communicating Results to Families

I.P Ijei-Enesi *MBBS, FMCPath*

Haematologist/ARISE Fellow

Barau Dikko Teaching Hospital/Kaduna State University

Nigeria

This project has received funding from the European Union's Horizon 2020 research and innovation programme under the Marie Skłodowska-Curie grant agreement No. 824021

Aims

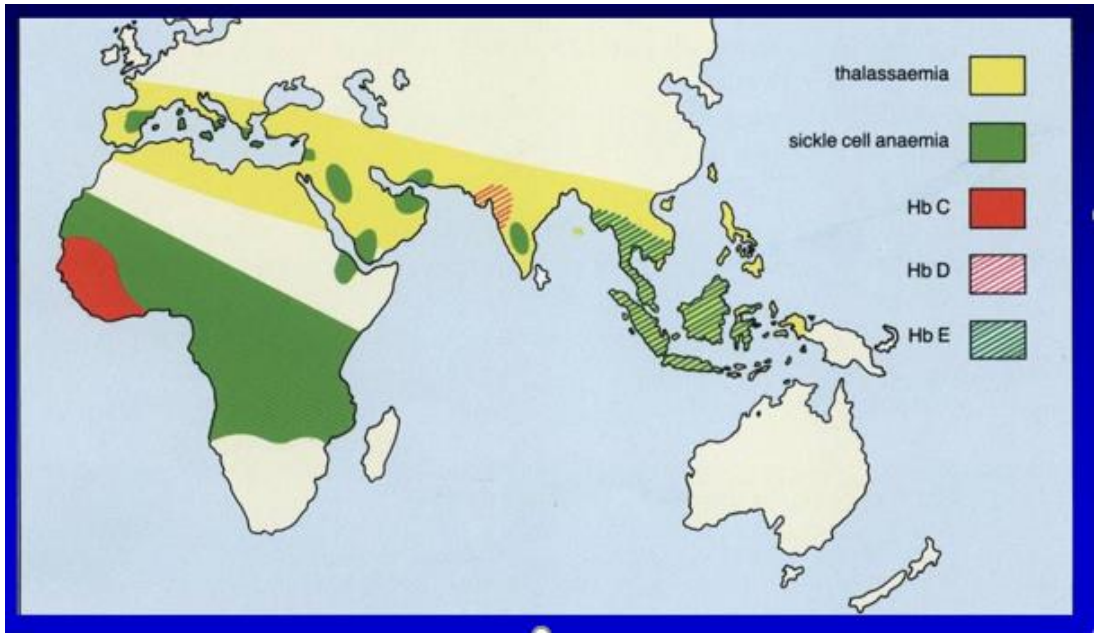
- To provide participants with an overview of the Kaduna State newborn screening (NBS) program pathway
- To outline the types of results generated via the NBS
- To introduce participants to a simple approach to breaking “difficult’ news
- To delineate potential areas of concern to parents/caregivers of affected children

Outline

- Burden of SCD
- Inheritance of SCD
- Pathophysiology and clinical manifestation
- NBS & SCD
  - The journey so far
  - Goals of the NBS program
  - The screening pathway- Kaduna State
  - Reporting results
  - Staff involved in communicating results
  - Breaking 'difficult' news- The *'SPIKES'* approach
  - FAQs
- Summary & Resources

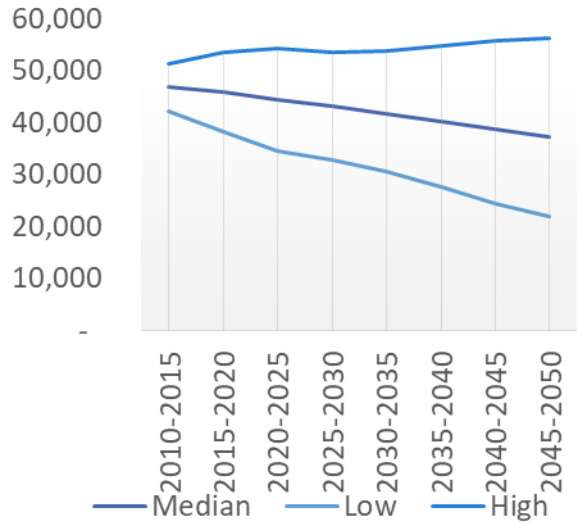
Burden of SCD

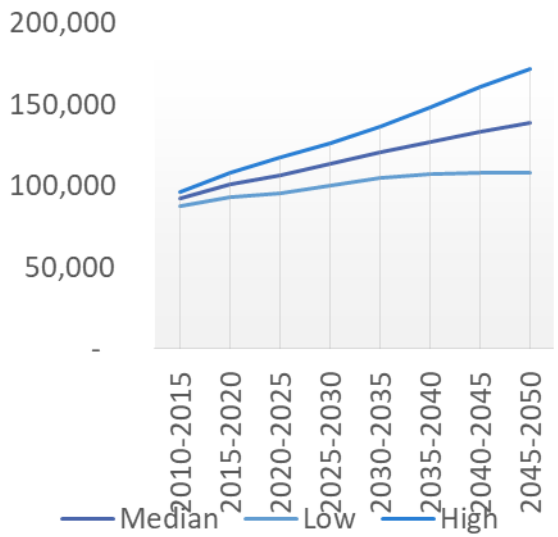
- Sickle Cell Disorder (SCD) is the most common monogenetic disorder worldwide with highest burden in Sub-Saharan Africa.
- The most clinically severe SCD is SCA (Hb SS). Others include; Hb SC disease, Hb Sβ-thalassaemia etc.
- Nigeria has the highest prevalence of SCA in the world and accounts for > 150,000 annual births with the disorder.
- About 5% of the world's population carry the trait for haemoglobin disorders.
- In Nigeria, 2-3% of the population are estimated to be living with SCA while up to 25-30% carrier state prevalence is reported
- In Kaduna state, approx. 1.7% of >4,000 babies/annum have SCA Inusa *et al* (2015)

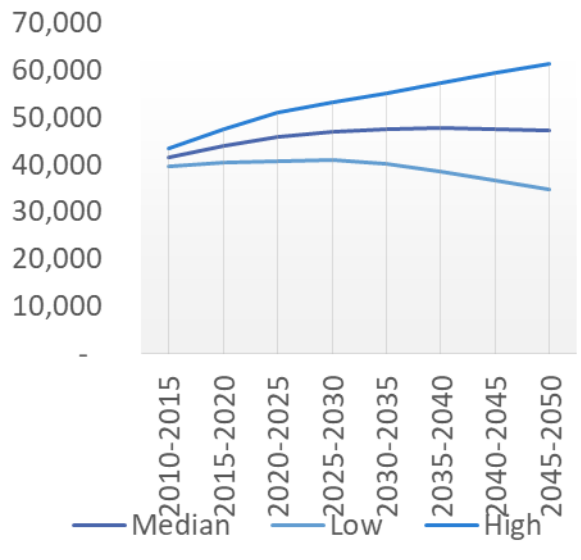


Sickle-cell anaemia projections(births)

	India		Nigeria		DRC	
	2010	2050	2010	2050	2010	2050
All. Freq.	0.0017		0.0138		0.0140	
Pop. (x1,000)	1,205,625	1,620,051	159,708	440,355	62,191	155,291
CBR*	20.7	13.1	41.5	29.0	42.9	27.5
SCA births	**46,790**	**37,157**	**92,330**	**138,680**	**41,561**	**47,301**



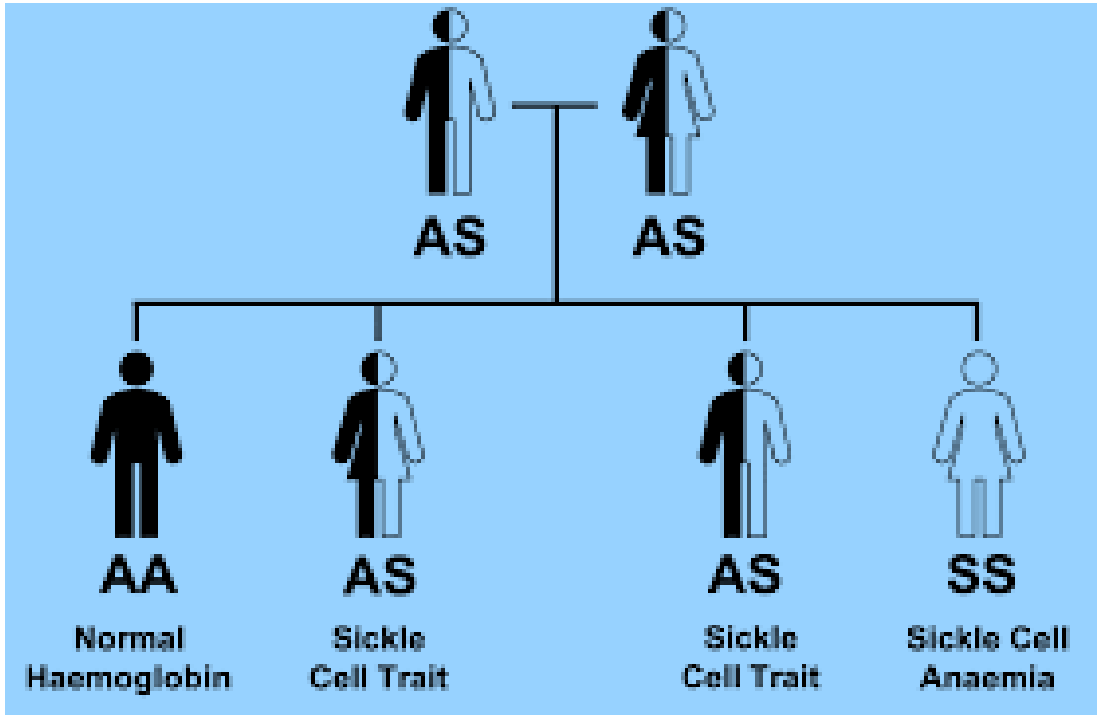




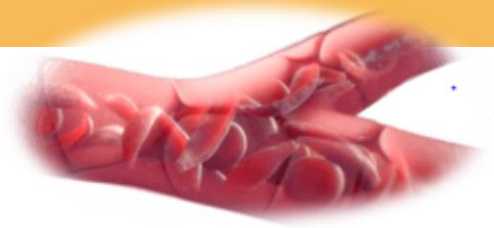
Piel *et al* (2013) *PLOS Med (Courtesy, Baba Inusa 2020)*

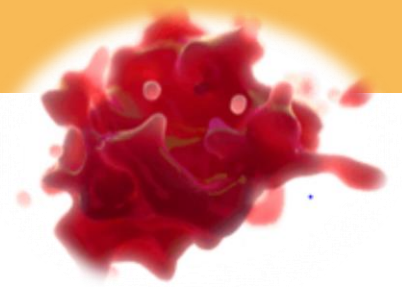
Inheritance of SCD

- Autosomal recessive inheritance pattern
- Possible outcomes in ANY pregnancy
  - Hb AA 25% or 1 in 4 chance
  - Hb AS 50% or 2 in 4 chance
  - Hb SS 25% or 1 in 4 chance
- Not influenced by gender



Pathophysiology of SCA





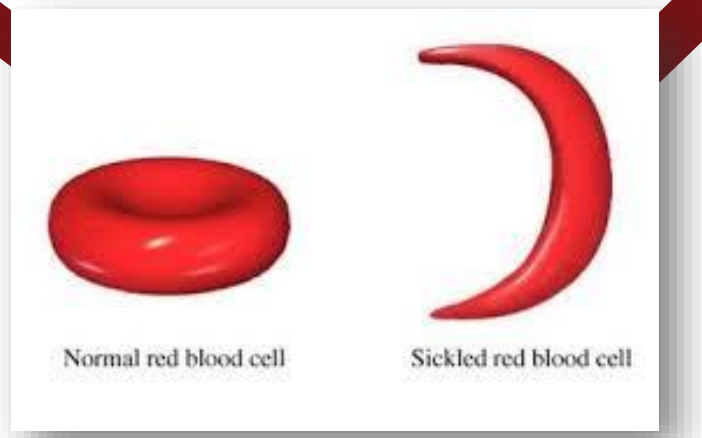
Single nucleotide change
GAG → GTG
Glutamic acid → valine
At position 6 of the β-globin chain
$\alpha^{A}_{2}\ \beta^{S}_{2}\ (\alpha^{A}_{2}\beta^{6}_{2}\ ^{Glu\text{-}Val})$
Alters physicochemical properties of haemoglobin molecule

Trapping of sticky, rigid sickle shaped cells and impaired blood flow "Vaso-occlusion"

Bone pain, AVN, OM

Retinopathy, Nephropathy, Priapism etc.

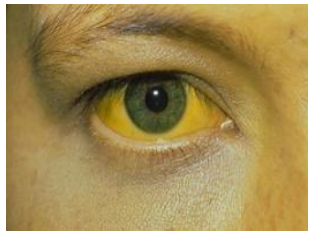
ACS, stroke



Reduced red cell lifespan "Haemolysis"

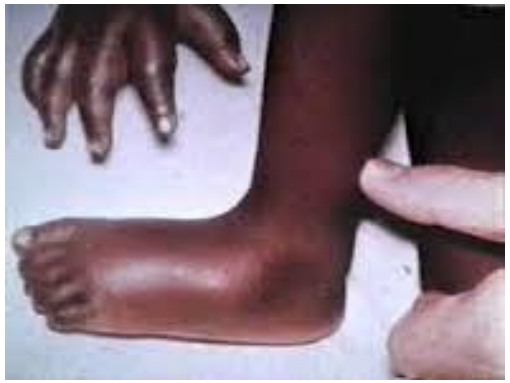
Anaemia, jaundice, cardiomegaly

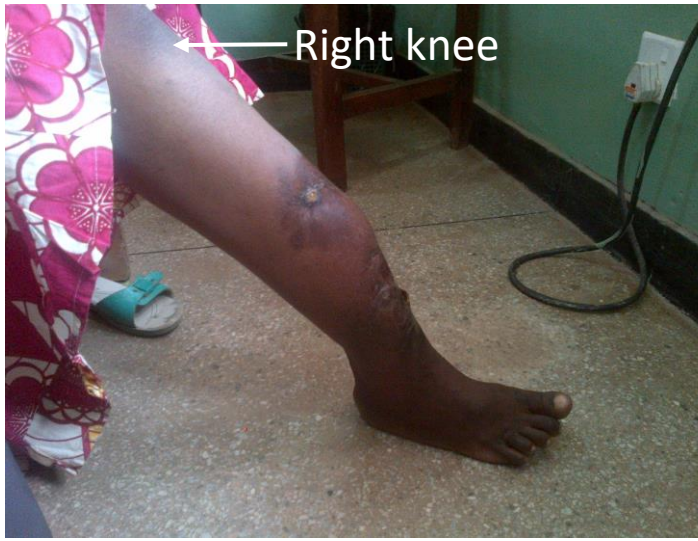
Clinical Manifestations of SCD



- It is a lifelong disorder punctuated by acute and chronic events

ACUTE	CHRONIC
• Painful episodes- Hand-and-foot syndrome, bone pain crises	• SC nephropathy • Osteonecrosis • Osteomyelitis • Leg ulcers
• Anaemic crises- Hyperhaemolytic, sequestration, aplastic	• Delayed puberty • Pulmonary hypertension etc.





*Risk factors: Dehydration, extremes of temperature, infections etc.

Newborn screening and SCD

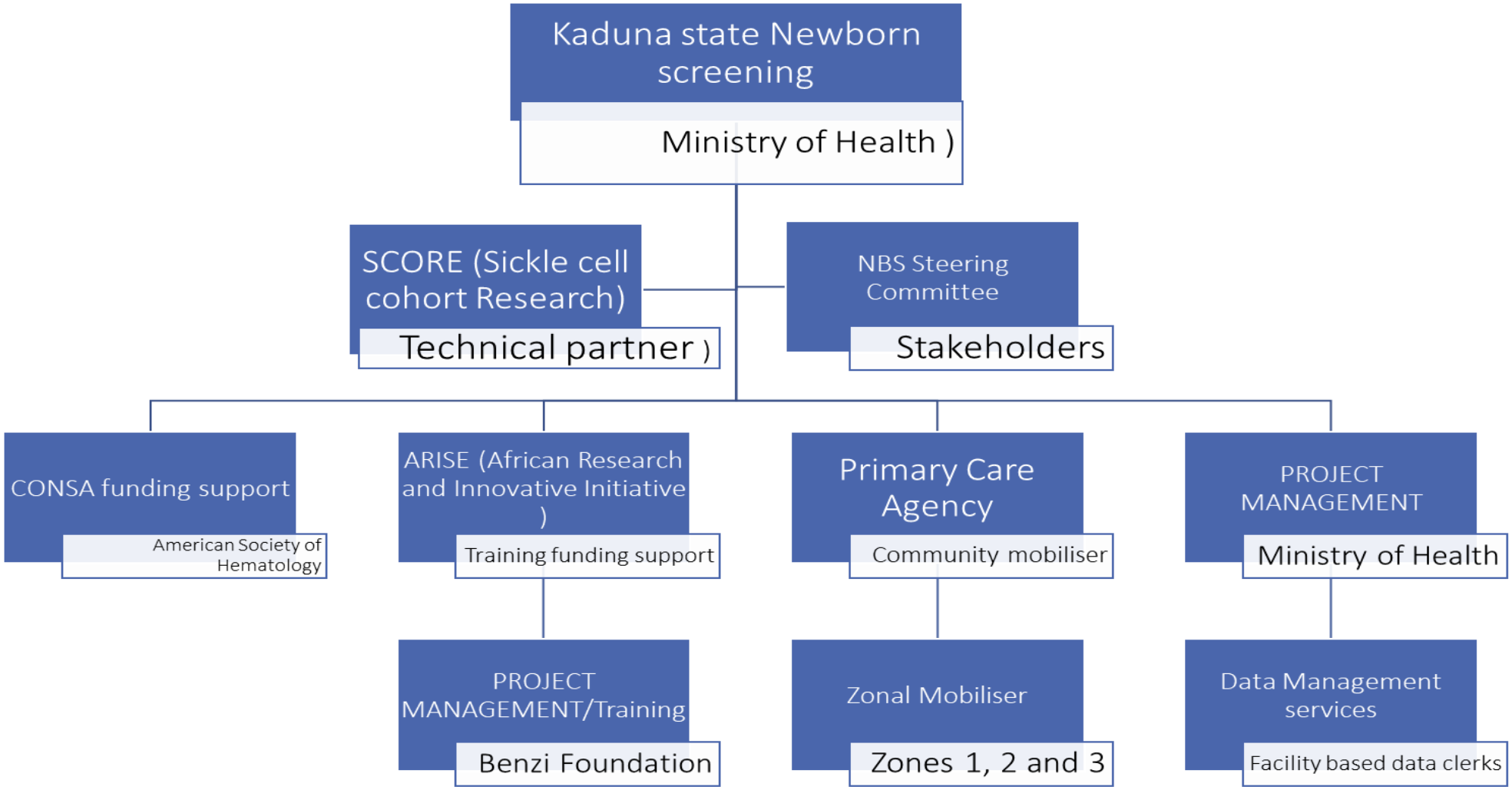
Journey of NBS-SCD in Kaduna- a collaborative effort

KD NBS-SCD diagnosis & Management

Focus Group Discussion with stakeholders

Courtesy; Baba Inusa (2022)

KADSCAP- Roles & Responsibilities



Little 'Giant' Steps

Workshop on SCD Research

***SCORE**/Health systems strengthening workshops*

SCORE-KDS Health systems strengthening workshops/AfroSicklenet/ ASCAT

Revised MoU with Kaduna state Govt./SCORE

2007 | 2009-2011 | 2012 | 2013-2014 | 2016-2018 | 2019 | 2020 | 2022

- Implementation
- Roll out

EU-UNDP SCD N-253 JMDI project (Michigan state University, GSTT, Zankli and Fantsuam Foundation)
APHL IEF Platform (APHL)

Seed NBS reagents exhausted, Line of communications opened with KDSG

ARISE
CONSA
RCPath LabSkill
Africa/IEF
CESTRA
Installation training

Implementation of the Screening Programme

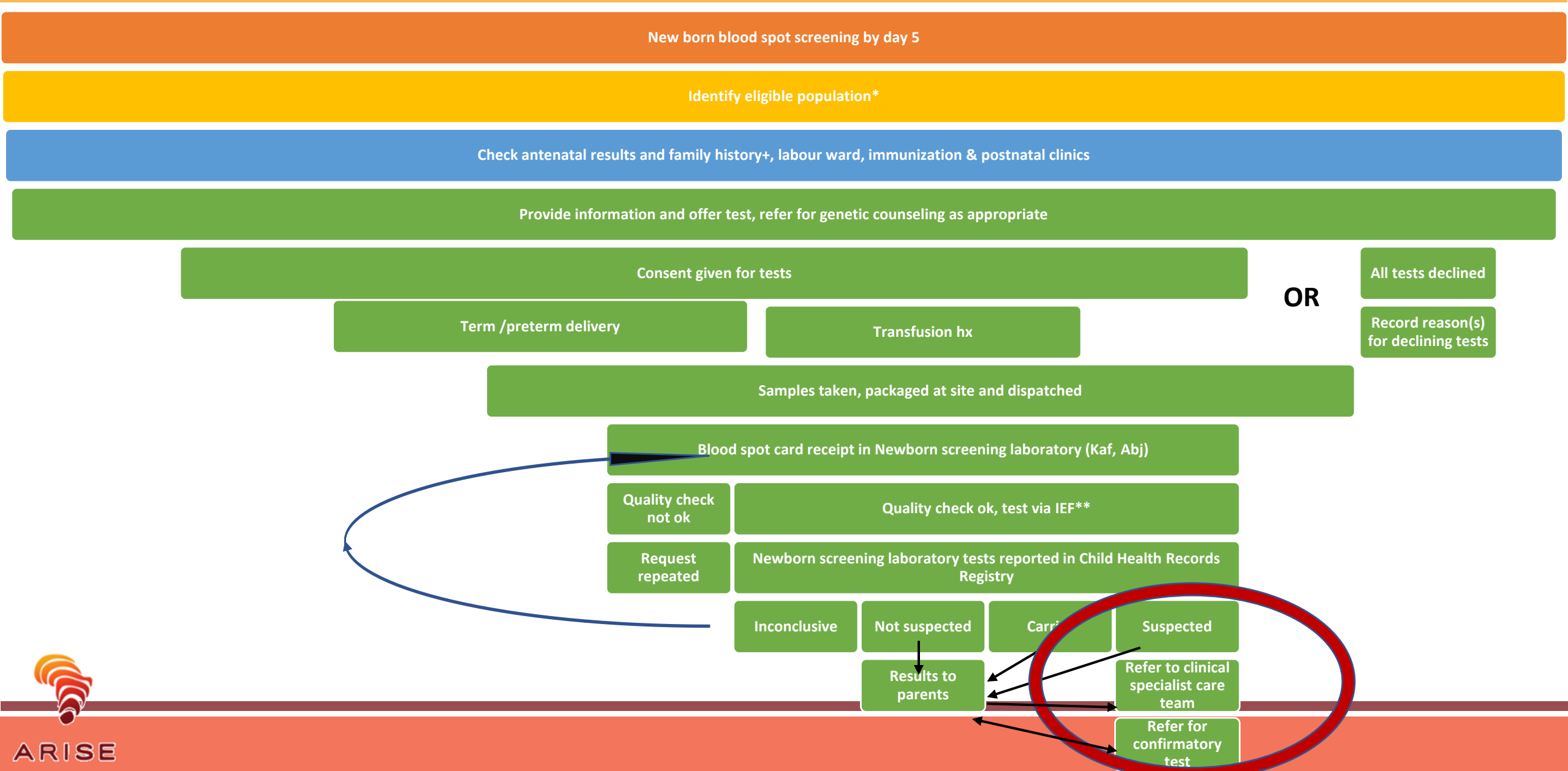
DBS Sampling & Treatment Hubs- Kad, Zx, Kaf
IEF Testing Sites- Kaf, Abj

Goals of NBS Program

- Early accurate diagnosis of SCD
- Prompt initiation into a pathway of care by PHC/specialist HCW
  - Penicillin prophylaxis
  - Malaria prophylaxis
  - Access to comprehensive care etc.
- Continuous support via family education, health advice, advocacy etc.

DBS Sampling

THE NBS SCREENING PATHWAY- Kaduna State



ARISE

NBS Result Reports

- ***Inconclusive;*** Not released from the lab, retested according to protocol

- ***Not suspected;*** Result FA, no haemoglobin disorder suspected

- ***Carrier;*** Result FAS, has trait for a haemoglobin disorder, asymptomatic, genetic counseling required

- ***Suspected;*** Presumed haemoglobin disorder

Suspected cases

- IEF results;

Bands identified on IEF	Inference	Action to take
FS	HB SS (Sickle cell anaemia)	For confirmation by repeat IEF ± HPLC after 3 months; Refer to clinical pathway including specialist in haemoglobin disorders
FSC	Hb SC disorder	"
FSA	Hb Sβ-thalassaemia	"
FSV	Hb S/Variant	"

TREATMENT HUBS:

Ahmadu Bello University Teaching Hospital, Zaria

Barau Dikko Teaching Hospital, Kaduna

Institute of Child Health, Banzazzau, Zaria

Sir Patrick Ibrahim Yakowa Memorial Hospital, Kafanchan

Challenging cases

- There is a risk of missed diagnosis with screening methods
- Cases of Hb Sβ-thalassaemia (FSA) may be reported as sickle cell trait (FAS)
- Preterm infants (especially <33/52) may give false negative results
- Transfusion history is important

Staff who should be involved in communicating results for affected babies

- Should have a clinical background
- Should have basic knowledge about SCD, Hb variants and inheritance patterns
- Should be able to differentiate between carrier states and clinically significant disease
- Minimal training required for ‘not suspected’ and ‘carrier’ results

Laying the foundation

- It begins at first contact!!!!
- Tell parents the truth! Be empathetic
- Do not set unreasonable expectations
  "This is the best test for diagnosing SCD"
- Refrain from personal misleading interjections and colorations
  "By God's grace/in sha Allah, it will be negative"

Principles of Counselling- *I C N S*

Informative

- Accurate, unbiased
- Address myths, misconceptions
- Consider language, level of education
- May require ≥1 session

Confidential

- Maintain confidentiality to encourage full disclosure

Non-directive

- Client(s) decision is key
- Non-imposition of counsellor's opinion(s)

Supportive

- Support client(s) decision
- No judgement
- Respect for the right of others
- Allow client to feel secure to discuss potential consequences of choice(s)

Breaking ‘Difficult’ News

- This is a somber task with implications to the infant and family- physical, socioeconomic, psychological etc.
- Should be done in person as much as is possible
- Parents are initially notified by phone call to come in for their results
- *Privacy* should be maintained for the discussion
- Avoid using the term ‘abnormal haemoglobin’ or normal haemoglobin
- Some advocate for the use of words like ‘unusual’, ‘variant’
- Parents may exhibit anxiety, disappointment or sadness. Sometimes anger or denial
- Reporting staff should be calm, provide reassurance and support

The Calgary-Cambridge Framework for Breaking 'Difficult' News

- **S** · *Setting up*
- **P** · *Perception*
- **I** · *Invitation*
- **K** · *Knowledge*
- **E** · *Emotion/Empathy*
- **S** · *Summary*

The 'SPIKES' Acronym

A Critical Look at the 'SPIKES' Acronym

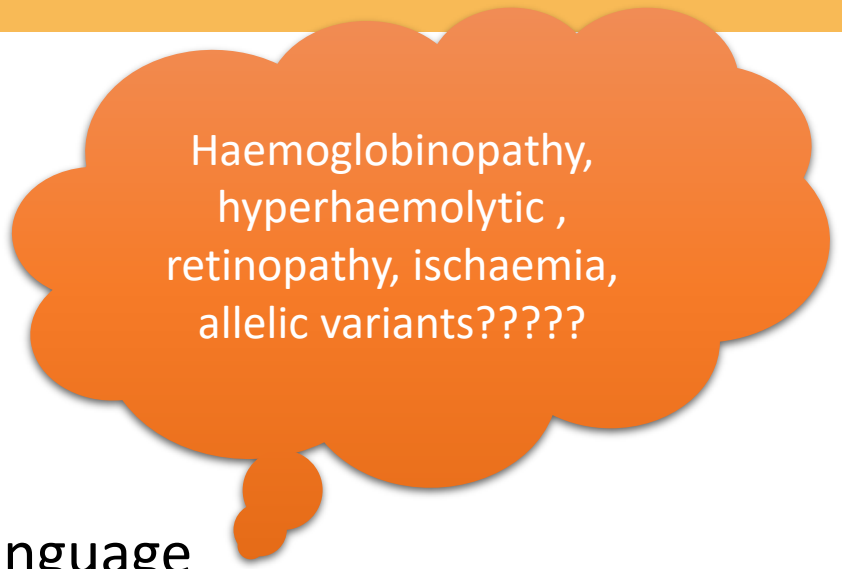
- ***Setting up;***
  - A comfortable, quiet and private location is required
  - Prepare for the discussion in advance
  - Have an overview of SCD and FAQs handy
  - Seat family comfortably and offer refreshments if available
  - Review any previous information disclosed by the parents or caregivers
  - Staff posture is important
    - No phone or other interruptions
    - Professional but empathetic
    - Maintaining eye contact
    - Listening and responding as required

A Critical Look at the 'SPIKES' Acronym

- ***Perception;***
  - Aids determination of how the news will be conveyed
  - Establish emotional states, expectations, knowledge contexts
  - Use of open-ended questions
    - How does the news make you feel? Vs Do you feel sad about the news?
  - Assess if client is able and willing to receive the news one is about to deliver
  - Allow time to analyze the message

A Critical Look at the 'SPIKES' Acronym

- ***Invitation;***
  - Obtain permission to share current news
  - Share as much as client can handle
  - Use simple, clear terms; avoid technical, complex language
  - Observe their response(s) to what you are saying, be sensitive & pace accordingly



A Critical Look at the 'SPIKES' Acronym

- ***Knowledge;***
  - Provide information on enrollment, follow up
  - Address questions (see FAQs)
  - Give reassurance of ongoing support

A Critical Look at the 'SPIKES' Acronym

- ***Emotion/Empathy;***
  - Encourage parents/caregivers to express themselves
  - Respect their wishes
  - Anticipate a potential wide range of reactions (Kubler-Ross stages of grief)
    - Denial
    - Anger
    - Bargaining
    - Depression
    - Acceptance
  - No two responses are the same; Validate their response, give them time to express themselves

A Critical Look at the 'SPIKES' Acronym

- ***SUMMARY/STRATEGY;*** This helps to establish a clear plan for the care of the affected individual. It eases stress
  - Ensure client has a clear understanding of the discussion by eliciting feedback
  - Client should be provided with materials for further reading
  - The client could take down notes from the discussion for later reference
  - Discuss treatment plan if clients are ready
  - They can prepare questions for a next meeting

Community Based FAQs

- What is SCD? What is sickle cell trait?
- How common is it?
- What causes SCD? If I have a child with SCD, will my other children have it?
- How is SCD diagnosed?
- Are the tests accurate? How many times does one need to be tested?
- What are common complications of SCD?
- How long does a person with SCD live?
- What is/are treatment(s) and/or services for SCD? Is there a cure?
- What is the experience of your facility in managing SCD?
- What kind of research is being conducted at NBS centers?
- Are there people i/we can contact for assistance/information?

Conclusion

- Delivering ‘difficult’ news is challenging to the recipient and the staff involved
- Building a good rapport helps
- Lay the foundation early
- Adopt principles of counselling; Informative, confidential, non-directive, supportive
- The ‘SPIKES’ approach is a useful tool
- Give your clients time- ‘It’s at their pace, not on your clock’
- No ambiguity! No judgement!

Acknowledgement & Appreciation

- *Thank you for your time*
- *Questions?*

Resources

- Piel FB, Hay SI, Gupta S, Weatherall DJ, Williams TN (2013) *Global Burden of Sickle Cell Anaemia in Children under Five, 2010–2050: Modelling Based on Demographics, Excess Mortality, and Interventions.* PLOS Medicine 10(7): e1001484. https://doi.org/10.1371/journal.pmed.1001484

- Inusa BP, Daniel Y, Lawson JO, Dada J, Matthews CE, et al. (2015) *Sickle Cell Disease Screening in Northern Nigeria: The Co-Existence of B- Thalassemia Inheritance*. Pediat Therapeut 5: 262. doi:10.4172/2161-0665.1000262. Pediat Therapeut ISSN:2161-0665 Pediatrics, an open access journal Volume 5 • Issue 3 • 1000262

- Breaking Bad News: Guidance for Staff. ARISE guidelines

- Protocol of SCD Screening Pathway. ARISE guidelines

- Community Based Frequently Asked Questions on Sickle Cell Disease. ARISE. 2021

- Protocol for Reporting newborn screening results for sickle cell disease to parents. NHS. April 2012

- Hemoglobinopathies: Current Practices for Screening, Confirmation and Follow-up. CDC, APHL. Dec 2015

- Frommel C. *Newborn Screening for Sickle Cell Disease and Other Hemoglobinopathies: A Short Review on Classical Laboratory Methods—Isoelectric Focusing, HPLC, and Capillary Electrophoresis.* Int J Neonatal Screen. 2018 Dec; 4(4): 39. Published online 2018 Dec 5. doi: 10.3390/ijns4040039 PMCID: PMC7548892

ARISE

African Research And Innovative Initiative For Sickle Cell Education

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.

This project has received funding from the European Union's Horizon 2020 research and innovation programme under the Marie Skłodowska-Curie grant agreement No. 824021