

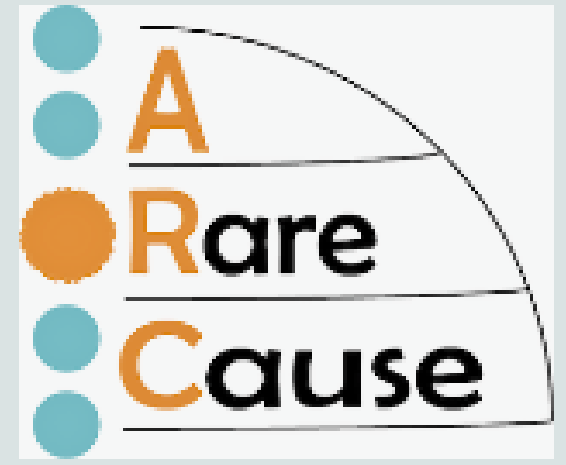


# WHEN INFRASTRUCTURE FAILS: WHY DRIED BLOOD SPOTS ARE AFRICA'S LIFELINE FOR BIOLOGICAL SAMPLING

INNOVATIVE SAMPLING METHODS  
OVERCOMING HEALTHCARE  
CHALLENGES



# DISCLOSURES



CMO for Decentra Health ( For benefit company)

CCIO for A Rare Cause ( Nonprofit of England and Wales)

Consultant FYMCA Medical Ltd ( For profit UK company)

All unreferenced opinions are those of the presenter from his own experience

Presenter is a non prescribing physician

This presentation was prepared with the assistance of artificial intelligence (AI) tools, which were used to support language refinement and to generate or source illustrative images. All content was reviewed and curated by the author



# THE CORE PROBLEM: FRAGILE DIAGNOSTIC INFRASTRUCTURE

## Infrastructure Fragility

Frequent power outages and fragmented transport networks disrupt reliable biological sampling in many African contexts.

## Cold-Chain Maintenance Challenges

Cold-chain systems are expensive and difficult to maintain, risking degradation of biological samples before analysis.

## Mismatch of Technology and Environment

Conventional venous blood sampling relies on ideal infrastructure, which often fails in unstable environments causing invalid results.

## Need for Robust Sampling Solutions

Diagnostic approaches must function reliably despite infrastructure weaknesses, highlighting the importance of alternative sampling methods.



# WHAT ARE DRIED BLOOD SPOTS (DBS)?

## DBS Sampling Method

DBS involves collecting small blood volumes on filter paper from finger or heel pricks, then air-drying them.

## Storage and Transport Benefits

Dried blood spots can be stored and transported at room temperature without immediate processing or centrifugation.

## Analytical Compatibility

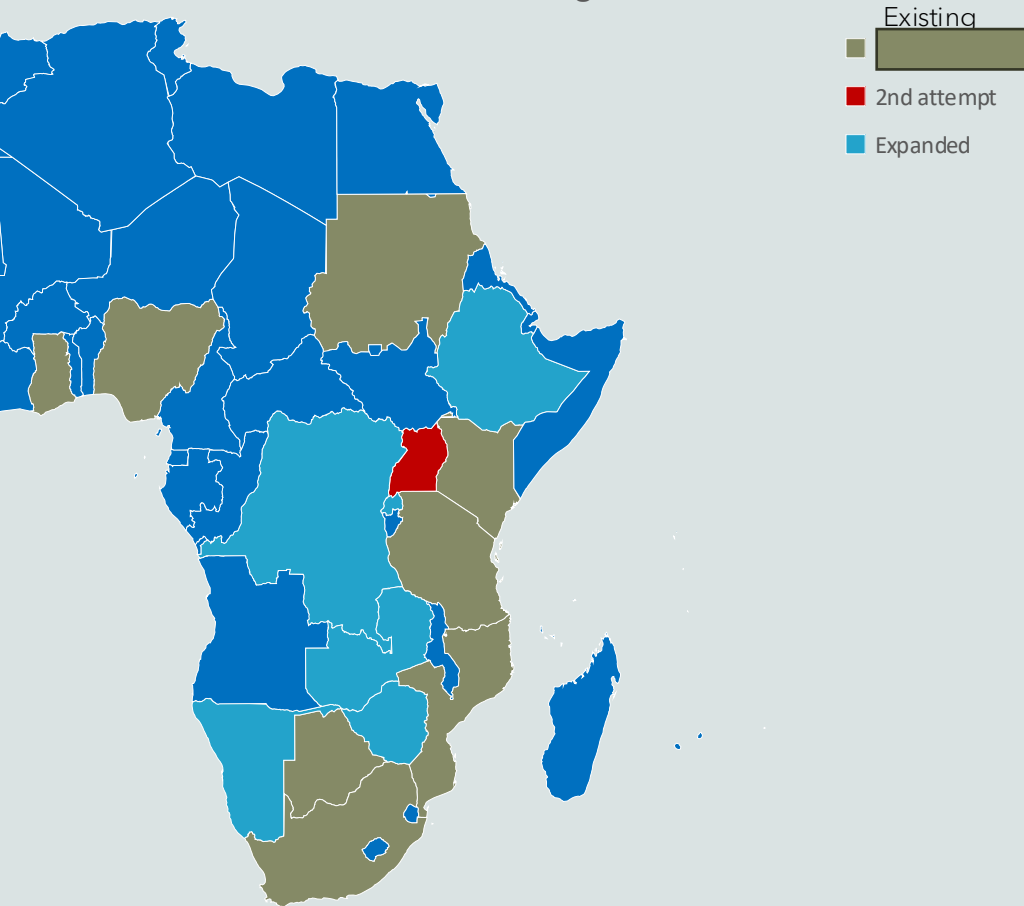
DBS samples work well with modern analytical techniques like PCR, ELISA, and mass spectrometry.

## Ideal for Decentralized Healthcare

Simplicity and durability make DBS suitable for healthcare settings with limited infrastructure.



LSD-IMPACT network coverage



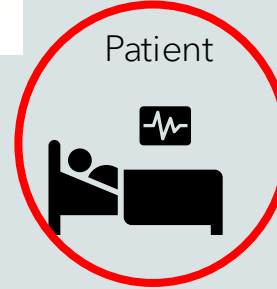
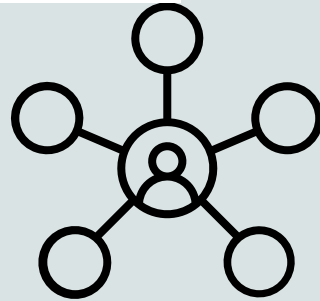
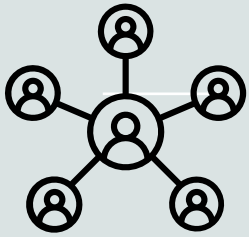
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# LSD-IMPACT AFRICAN NETWORK

- 15 African countries onboarded
- DBS-based 6-plex LSD testing
- Training mandatory before referrals

# DIAGNOSTICS- AFRICAN ROADMAP

FYMCA trained clinicians



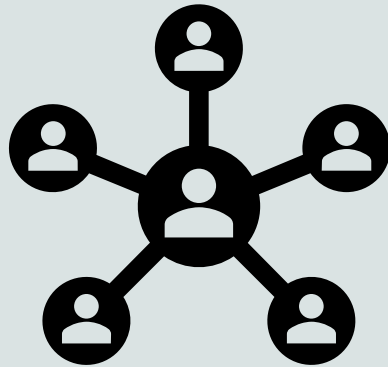
Patient

CHM central laboratory

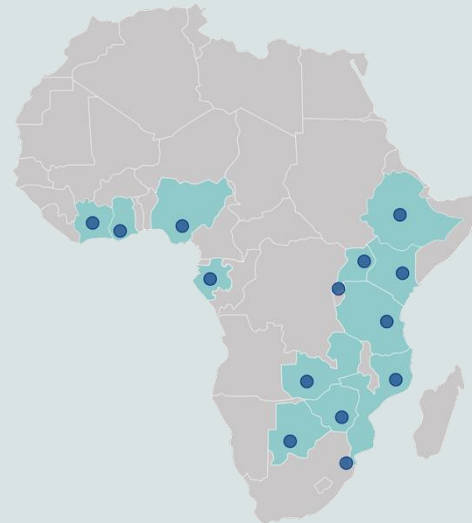


FYMCA/PLIEM trained laboratory staff

Plex 6 samples



Lancet Africa network



Medical support network



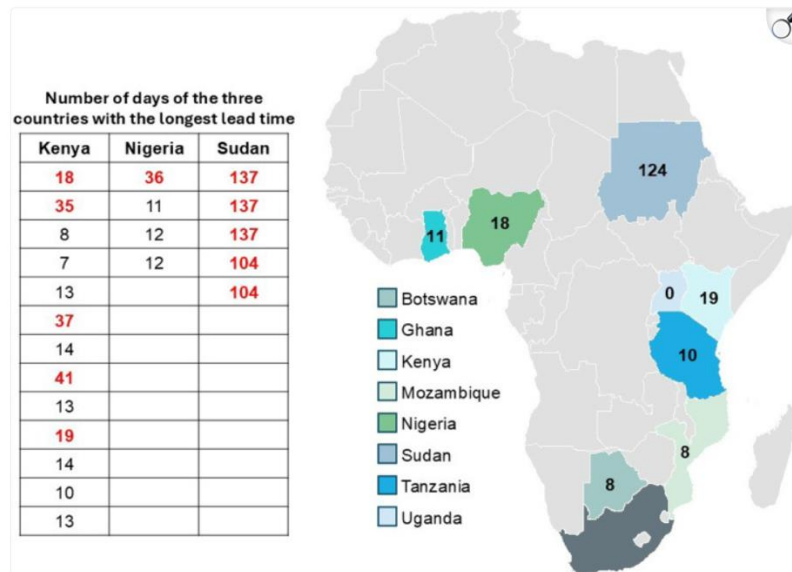
Negotiations ongoing to expand free testing  
Currently developing genetic testing services.



# GEOGRAPHY AND EQUITY

- Pathology networks reduce delay
- Sudan > 100 days due to courier reliance
- Time-to-lab did not affect diagnosis

Fig. 5.



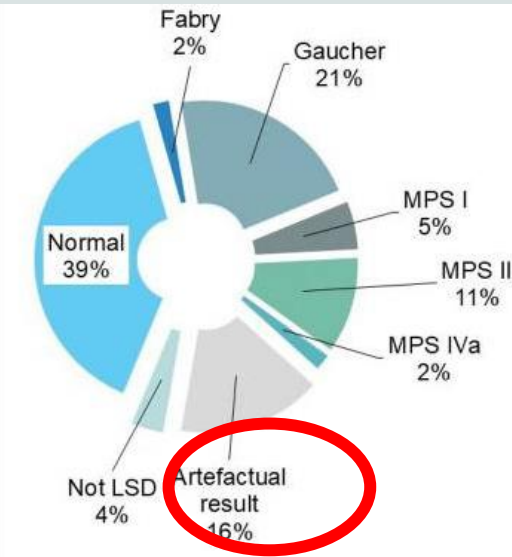
Average number of days until registered per country

Swanepoel AC, Hendriksz CJ, Mukhwana R, Oduwole A, Abdalla AT, Ameyaw E, Muze KC, Auruku A, Pinto F, Joel D, Aleksovska V, Collin-Histed T, Odero R, Conradie EH. Improving access to rare disease diagnostics in Africa: insights from a multinational pilot study. *Orphanet J Rare Dis.* 2026 Feb 5;21(1):44.

doi: 10.1186/s13023-026-04202-y. PMID: 41545220; PMCID: PMC12879359.

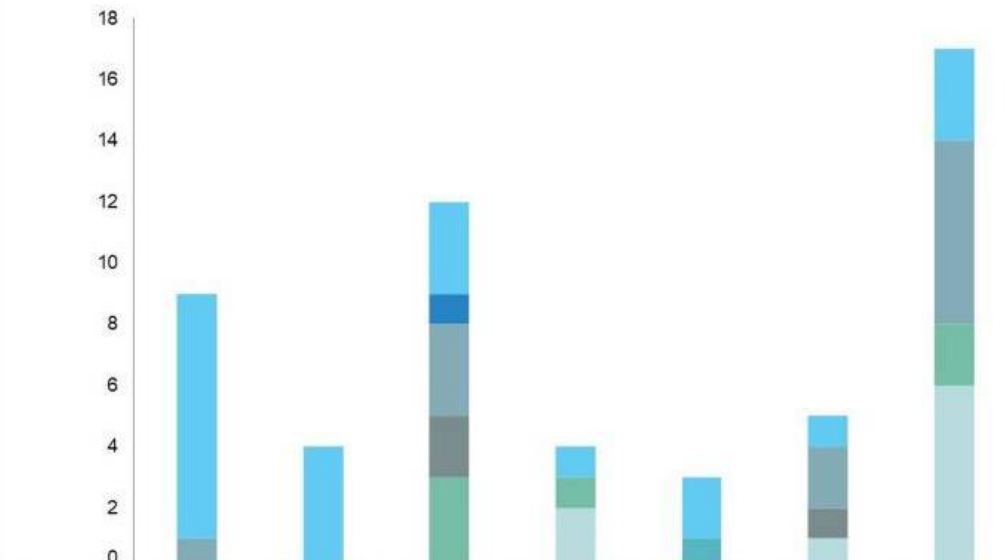
# WHAT THE DATA SHOW

- 98 referrals, 43 diagnoses
- ~45% diagnostic hit rate compared to usually 2-4%
- Gaucher most common diagnosis



Gender distribution for diagnosed LSD

Diagnosis	Total n	Males n (%)	Females n (%)
Fabry	1	1 (100)	0
Gaucher	12	3 (25)	9 (75)
MPS (All)	10	8 (80)	2 (20)
⇒ MPS I	3	1 (33.3)	2 (66.7)
⇒ MPS II	6	6 (100)	0
⇒ MPS IVa	1	1 (100)	0



LSD Diagnosed	Botswana	Ghana	Kenya	Mozambique	Nigeria	Sudan	Tanzania
Normal	8	4	3	1	2	1	3
Fabry			1				
Gaucher	1		3			2	6
MPS I			2			1	
MPS II			3	1			2
MPS IVa					1		

Swanepoel AC, Hendriksz CJ, Mukhwana R, Oduwole A, Abdalla AT, Ameyaw E, Muze KC, Auruku A, Pinto F, Joel D, Aleksovska V, Collin-Histed T, Odero R, Conradie EH. Improving access to rare disease diagnostics in Africa: insights from a multinational pilot study. Orphanet J Rare Dis. 2026 Feb 5;21(1):44.

doi: 10.1186/s13023-026-04202-y PMID: 41645220 PMC ID: PMC12879359

# SUCCESS STORY: CROSS-BORDER AND REGIONAL SHIPPING

## Regulatory Challenges in Shipping

Cross-border transport of biological samples faces strict regulatory hurdles and limited certified shipping services.

## DBS as Non-Dangerous Goods

Dried blood spots are shipped as 'Not Restricted' cargo, bypassing dangerous goods classification and easing transport.

## Cost and Time Efficiency

Avoiding UN3373 classification reduces delays and costs, enabling broader collaboration and sample sharing.

## Importance of Regulatory Alignment

Success depends on regulatory fit alongside biological stability, allowing samples to move freely within existing systems.



# OPERATIONAL CHECKLIST: CUSTOMS DECLARATION

## Importance of Customs Declarations

Effective customs declarations ensure shipments move smoothly and avoid delays in program timelines.

## Use of Minimal Nominal Value

Declaring a minimal nominal value instead of zero reduces customs issues and facilitates faster clearance.

## Impact on Program Continuity

Proper customs documentation prevents delays and maintains sample integrity for uninterrupted program operations.



# AFRICAN CLIMATE CONTEXT

- Humidity affects enzyme stability
- Tropical regions show artefacts
- Simple mitigations identified- air drying of samples

Use of dedicated envelope and desiccant



# CONCLUSION: DBS AS A MODEL FOR RESILIENT DIAGNOSTICS

## Scientific Reliability of DBS

DBS provide scientifically reliable diagnostic results even in challenging infrastructure conditions.

## Logistical and Regulatory Simplicity

DBS are logistically resilient and simple to regulate, enhancing access and equity in healthcare.

## Systems-Level Diagnostic Solution

DBS act as a systems-level solution, continuing to generate actionable data despite infrastructure failures.

## Future Diagnostic Model

DBS offer a powerful model for future diagnostics, especially under climate change and logistic/infrastructure pressures.



# SESSION SNAPSHOT

## Community Name:

W3 Exchange

## Session Title:

African Task Force Virtual Advisory Board

## Session Objectives:

There are multiple challenges which are unique to Africa that limits the care that patients with rare diseases receive. Although lack of finances is given as the most common cause for poor services, it is believed that a lack of diagnostic abilities and data to support clinicians and patient groups are the primary limitations. As such, this virtual session is being used to allow physicians and patient groups to collaborate with one another in hopes that the information collected can be used to develop models that will support the development of services for the rare disease community across Africa.

## Duration:

Online session spanned 30 days: August 15 – September 13, 2019

Within3

The screenshot shows the W3 Exchange website interface. At the top, there is a navigation bar with links for Help, Guidelines, and Contact Us. Below this, the current room is identified as FYMCA\_2019\_08. The main content area is titled 'African Taskforce Virtual Advisory Board' and includes a language selector (English and español), a 'HIDE TOPIC DETAILS' button, and a date indicator 'Aug 15, 2019 - Sep 13, 2019' with a 'Closes in 29 days' notification. The text below the title provides an introduction to the session, explaining the challenges in Africa and the purpose of the virtual advisory board. It also includes sections for 'Virtual Engagement', 'Instructions', 'Resource Library', and 'Timing'.

**W3 Exchange** Help | Guidelines | Contact Us  
Current Room FYMCA\_2019\_08

Current Topic All Topics Webcasts Moderator Tools Admin

English español

### African Taskforce Virtual Advisory Board

Aug 15, 2019 - Sep 13, 2019  
Closes in 29 days

⌘ HIDE TOPIC DETAILS

Here is just a short introduction why we as FYMCA Medical Ltd and Care Beyond Diagnosis embarked on trying to find a solution and we will share more about our work and ideas on the virtual platform we have invited you to join.

There are multiple problems which are unique to Africa which limits the care that rare diseases patients receive across the continent. There are already some excellent organizations developed in some African countries and collaboratives to work together and this taskforce will in no way want to compete or impinge on their current work but hope to support their goals and integrate were possible. A lot of the excellent work that has been done has been led by the patient organizations but looking from the outside there seem to be a lack of coordination between medical professionals, patient groups, diagnostic laboratories and decision makers of both politics and health care funders.

This Taskforce is an effort to provide a platform to firstly identify communal problems and then try and find solutions to them. We believe by working collectively we can help and offer our experiences from working with many clinicians and patient groups from resource poor countries and from our own experiences in health care systems which are already developed.

Although lack of finances is given as the commonest cause for poor services, we believe it is the lack of data to support clinicians and patient groups that is the ultimate limitation, and this will not change until we can improve the diagnostic abilities and provide better data to those making decisions. We believe there are also opportunities to develop some models to support development of services but only if we can manage to all work together and allow everyone an opportunity to develop. Hopefully we can develop some documents which can be used by individual groups to help move things forward for the greater good of the rare disease community across Africa.

This is only one perspective and we have put the following questions and statements out as a starting point for discussions. Please feel free to add additional resources, references ideas and obstacles to the lists.

**Virtual Engagement:**  
One of the benefits of using this platform is that not only are you able to answer the session's questions, but you are able to engage directly with your fellow advisors. Please be sure to take advantage of all the key features of this platform that will allow you to engage in insightful dialogue with your colleagues and moderators. These including commenting, replying, tagging users, and attaching documents to your comments.

**Instructions:**  
We ask that you access the platform regularly throughout the session to review and respond to all posted questions, as well as comments posted by the session moderators and your peers. Your active and ongoing participation is important, as this will provide us with valuable and timely insights and give you the opportunity to share and learn from your colleagues.

**Resource Library:**  
If you have any questions regarding the platform, please review the orientation video or Advisor Orientation slide deck which are posted in the Resources Library on the session's home page. Additionally, you can click "contact us" or send an email to support@within3.com with any technical questions you may have.

**Timing:**  
The session is open today, Thursday, August 15th through Thursday, September 13th at 5:00pm EST.

We appreciate your participation and look forward to your feedback.

This is a screenshot from the live program and is for reference only

# African Taskforce – Countries Involved

## African Countries Represented:

South Africa • Ghana •  
Botswana • Kenya •  
Nigeria • Sudan •  
Tanzania • Rwanda  
Mauritania • Niger • Ivory  
Coast • Congo Brazzaville  
• Tunisia • Cameroon •  
Zimbabwe • Senegal

## International Contributors:

United  
Kingdom  
United  
States of  
America  
Canada

*Pan-African participation*

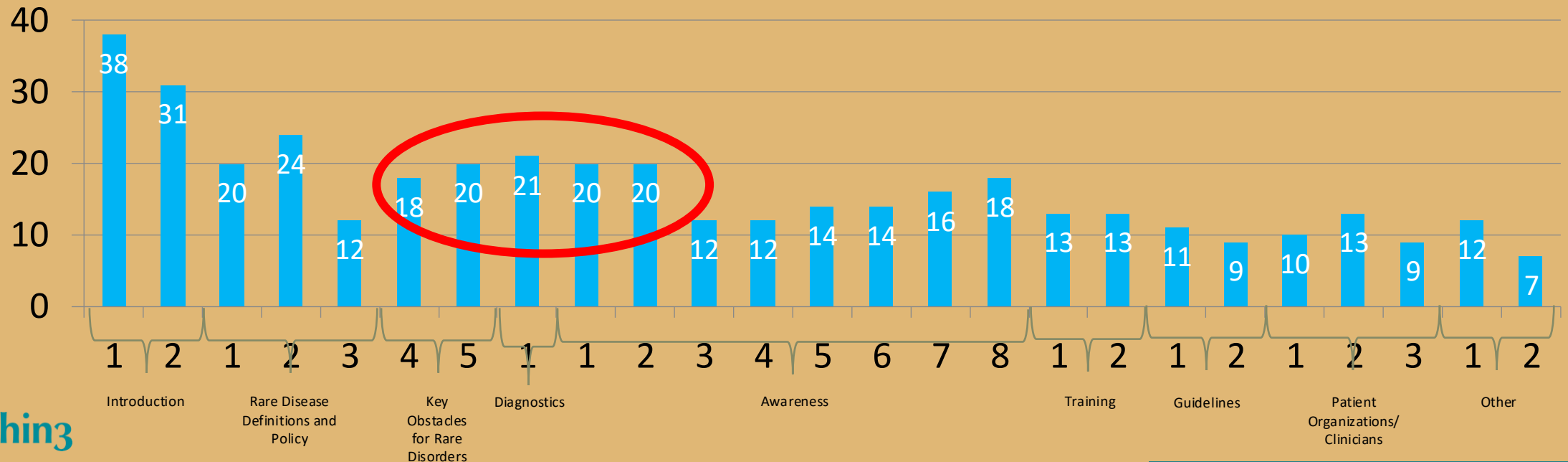
# ADVISOR RESPONSES BY QUESTION

**428**  
total advisor responses

## Top Questions

by total advisor responses

- S1-Q1** Please complete the online Gapminder test to allow yourself some perspective on a fact-based worldview. After you have completed the test you will be able to compare your country with others.
- S1-Q2** Please introduce yourself or your organization and let others understand what your involvement in rare disorders in Africa is.
- S2-Q2** Do you agree with this definition or do you feel it needs to be specific for your country, continent or should depend on Gross Domestic Product?



## SESSION PARTICIPATION METRICS

Metric	African Taskforce Virtual Advisory Board
Session Duration (in days)	30
Number of Advisors	22
Number of Questions	27
Number of Advisor Words Per Question	592
Number of Advisor Responses per Question	16
Number of Moderator Responses	100
Number of Moderator Responses per Question	4

# ADVISOR POST-SESSION SURVEY RESULTS (CONT.)

## *What Advisors Specifically Found to be the Most Valuable Part of This Experience*

*(N=4)*

“I had the opportunity to learn more about the challenges in the management of people with rare diseases in other African countries. Contributions from other advisors about how to improve the rare disease situation in Africa were valuable.”

“Being able to understand everyone else’s challenges that they experience and to reflect that South Africa is not too bad to be working in.”

“I valued the broad spectrum of persons on the platform – clinicians, patients, doctors, advocates and their extensive geographical coverage. Why? It made me appreciate the common challenges and strides we have in Africa as a whole.”

“The ability to partake at a time suited to your personal schedule was refreshing and allowed for continuous and consistent input.”

## *Specific Topics From the Advisory Board that Advisors Would Like to Spend More Time On*

*(N=4)*

- Getting a definition for rare diseases for Africa
- Development of specific guidelines
- Rare disease definition, genetics & diagnostics
- Registries / surveillance definitions