



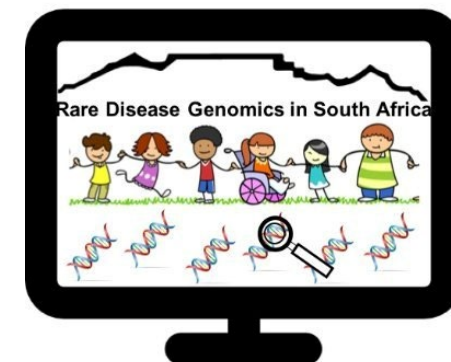
# MEDICAL GENOMICS RESEARCH IN AFRICA

## SOUTH-NORTH AND SOUTH-SOUTH COLLABORATIONS



**Shahida Moosa**

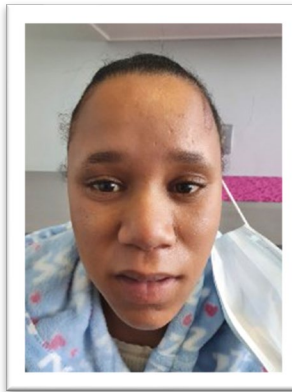
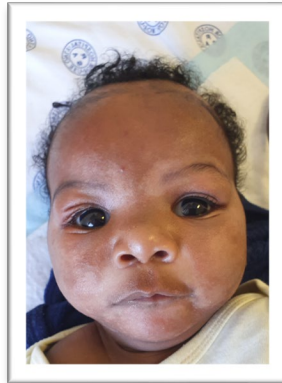
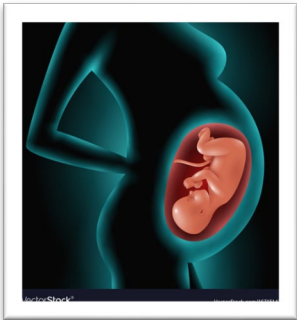
**Head: Medical Genetics / Associate Professor**  
**Tygerberg Hospital / Stellenbosch University**  
**[shahidamoosa@sun.ac.za](mailto:shahidamoosa@sun.ac.za)**



# Medical Genetics: Tygerberg Hospital



2<sup>nd</sup> largest hospital in South Africa  
Small team: MGs, GCs  
Full spectrum of patients  
Prenatal – Paeds – Adult - Cancer  
Limited access to genetic testing



## UNDIAGNOSED

# Genomics Research: Stellenbosch



## Biomedical Research Institute

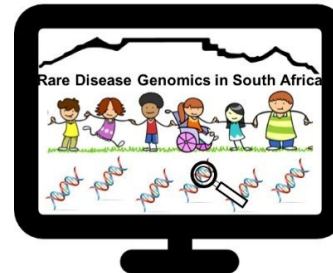
- Largest genomics facility in Africa
- Rare Disease Genomics group

Sub-Saharan Africa's first

## *Undiagnosed Disease Programme*

- 450 patients enrolled over 2.5 years
- >50% diagnostic yield on exome seq

- As of 2023: In-house WGS



Until every African with a  
**RARE DISEASE**  
is diagnosed!

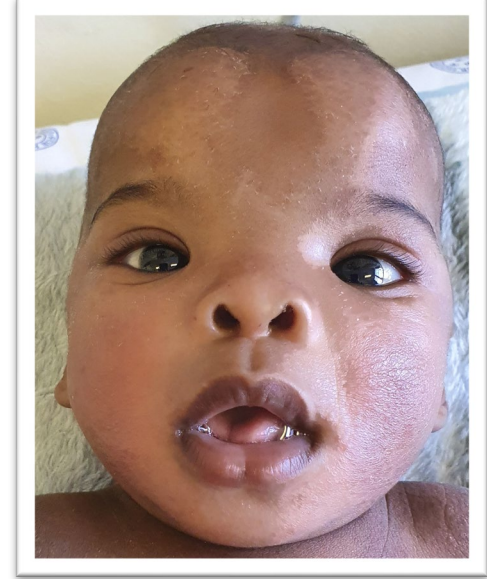
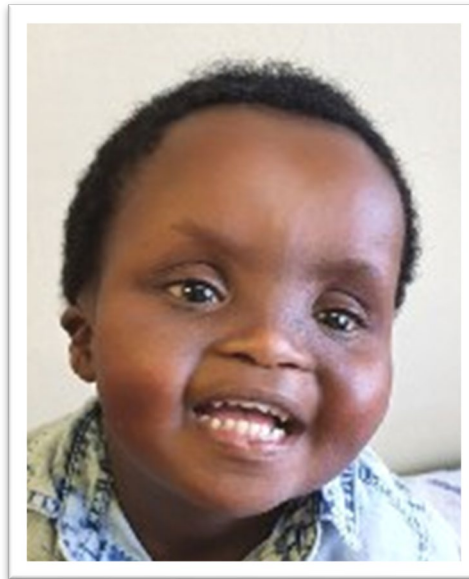
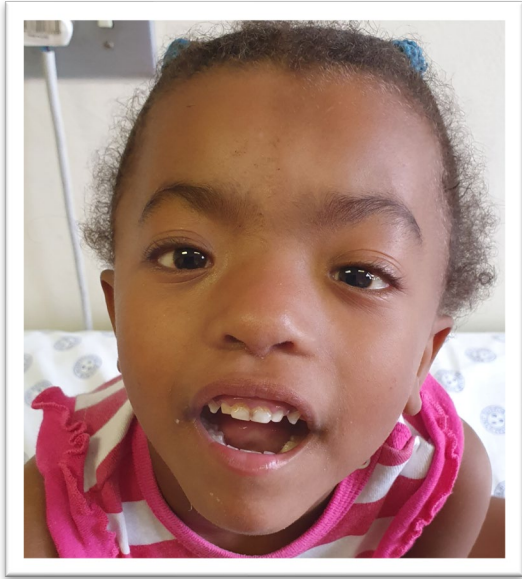
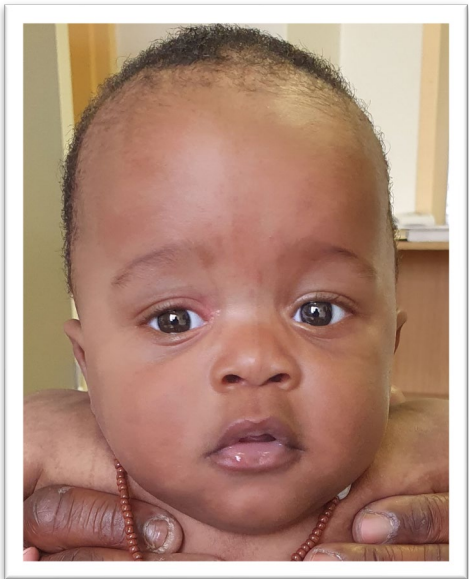
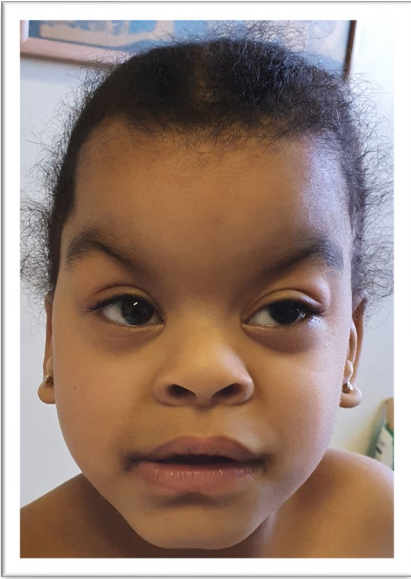


# Genomics in South Africa: UDP

>50% diagnostic  
yield

Many first diagnoses  
in Africa

Understanding of  
conditions in our  
populations



# Medical Genetics in South Africa: challenges



3 Medical Genetics departments  
2 Paediatricians with interest in Genetics

10 Medical Geneticists in Public Service

5 Medical Geneticists in Private Practice /  
other

Population ~60 million

**6/9 Provinces without access to  
Medical Genetics**

★ Medical Genetics clinical service

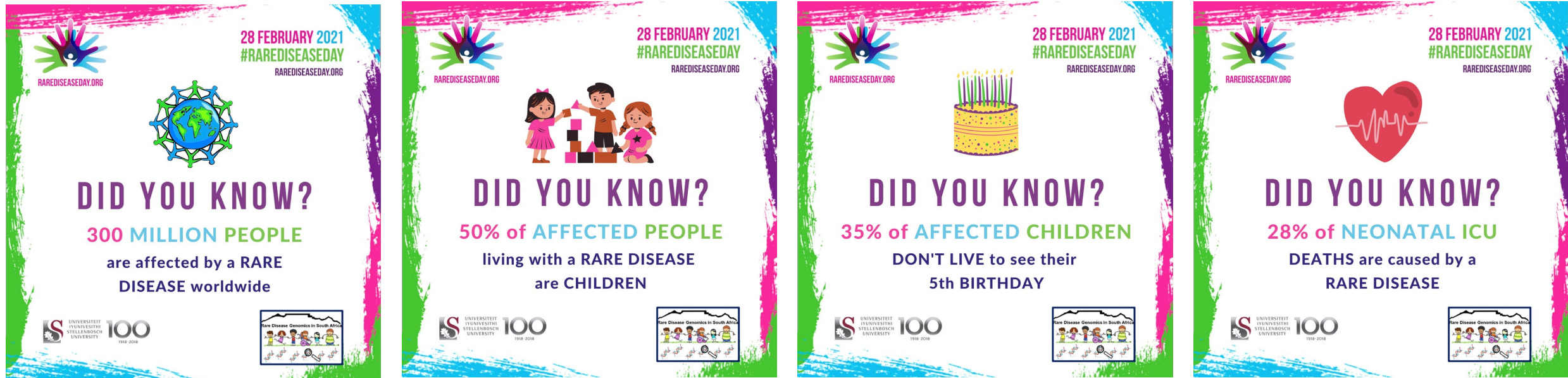
★ Medical Genetics clinical service with research



## Little government support



# Challenges – Lack of prioritisation



**“Genomics is not a priority for Africa!”**

# Challenges – Lack of prioritisation

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## “Medical genomics is not a priority!”



Infant mortality?

Under 5 mortality?

UN SDGs?

Funding for  
research

Funding for  
training

Investment in  
infrastructure

Policy



# Challenges: Stereotypes

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## Before I started the UDP:

“too difficult to do in Africa”

“requires specialized infrastructure”

“understudied populations too complex for this to be worthwhile”

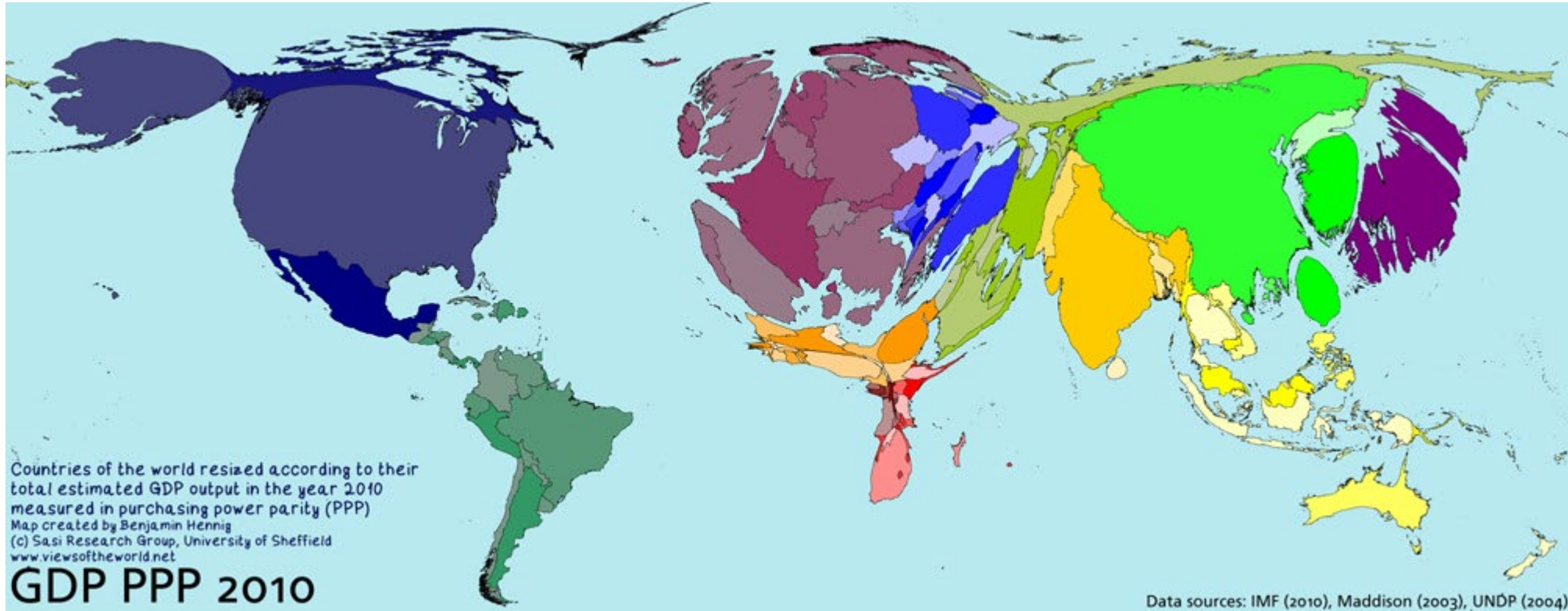
“necessary skills lacking”

“Africans would not be able to understand”

“Africans would not be interested”

“too early for Africa”

# Challenges – Underrepresentation



**Our population is underrepresented**

- **Global databases**
- **Research <10%**
- **Diagnostics?**

# Challenges – Underrepresentation

## “Representation matters”

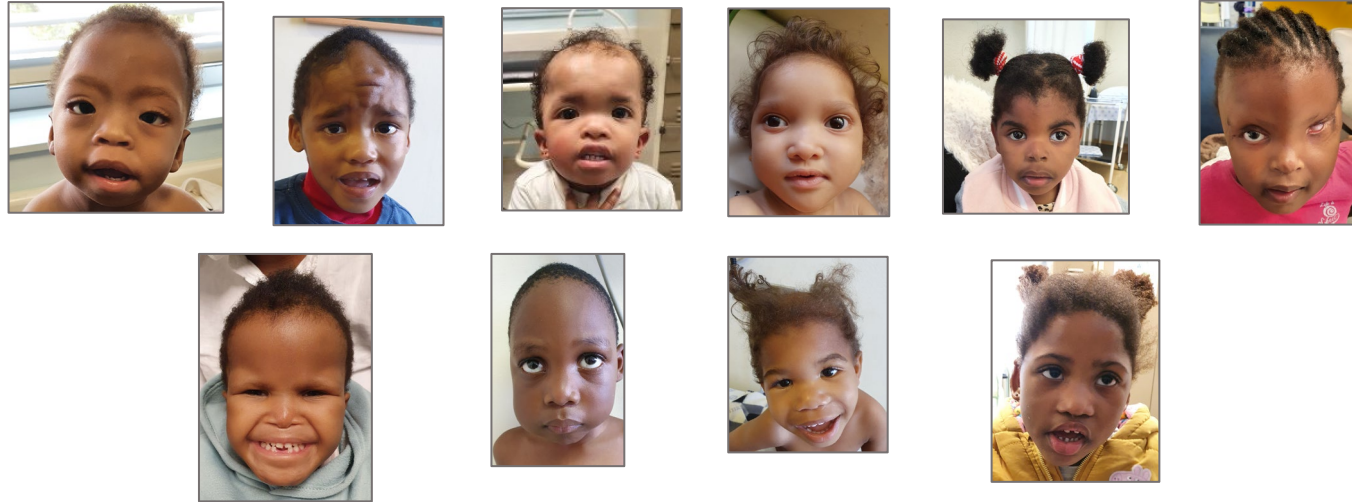
Pathogenic?

Benign?

Likely  
Pathogenic?

Likely Benign?

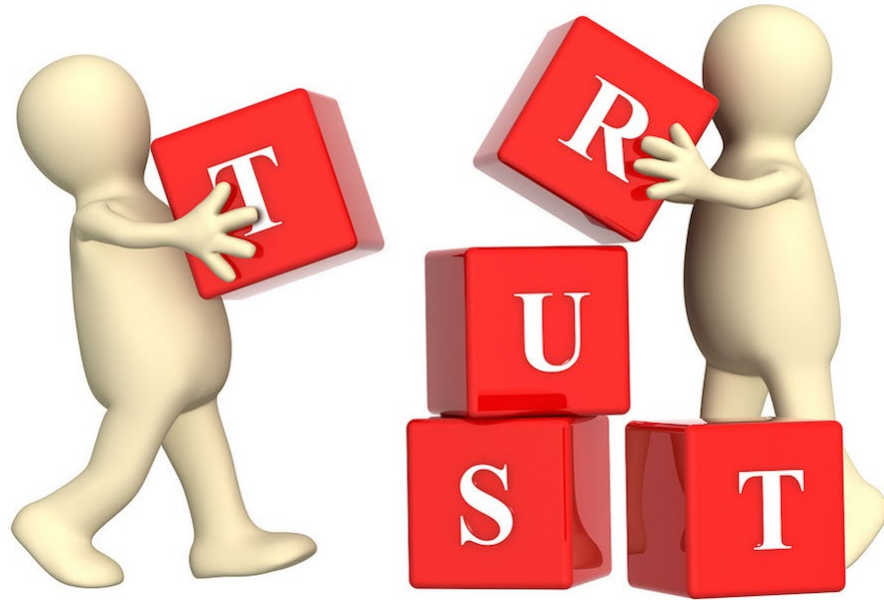
Variant of  
uncertain  
significance?



**In-house database >200 samples: 3 common SNPs identified**  
**All classified as “likely pathogenic” in ClinVar – repercussions?**

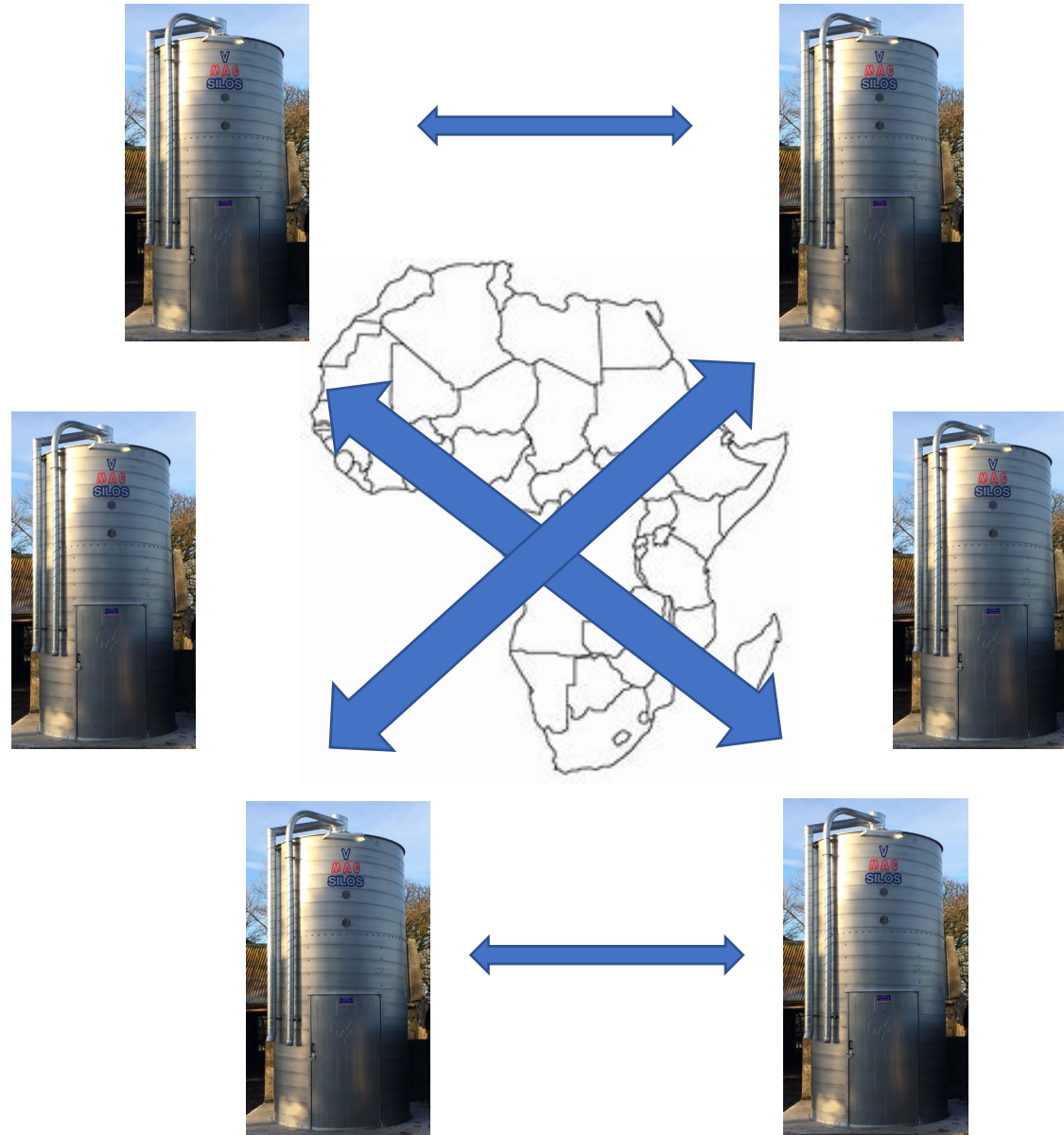
# Challenges – Rebuilding Trust

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# Challenges – Data sharing



# Collaboration is key!

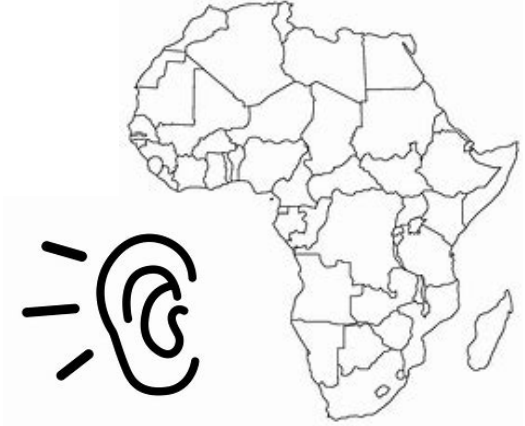
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**“Ubuntu-inspired” research  
and excellence**



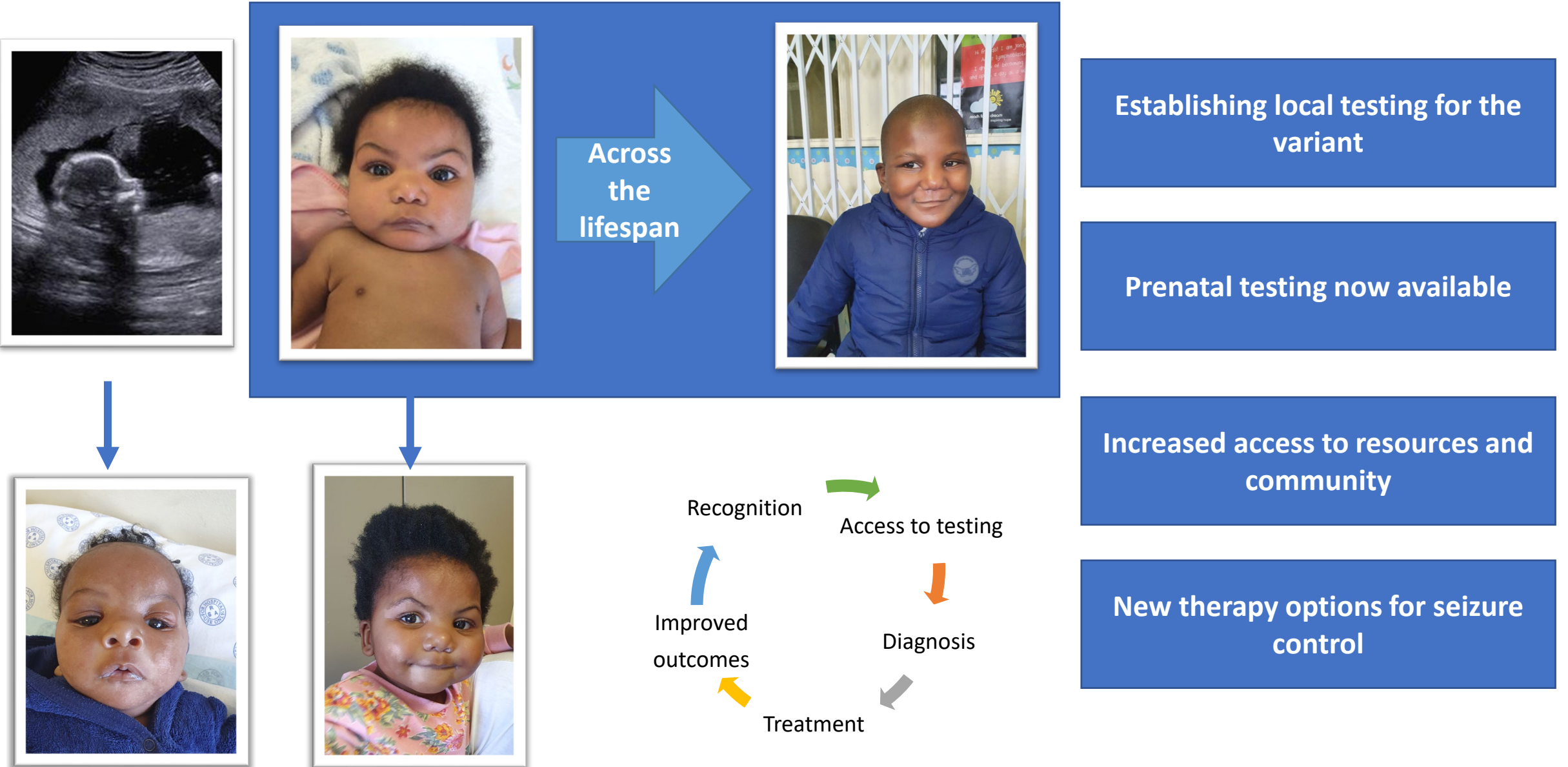
**Co-creation of knowledge  
and solutions**



**Listen to the Africans!**



# Collaboration is key: South-South on Mabry syndrome



# Collaboration is key: South-South

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**Expansion of UDP to neighboring countries**

**Pan-African initiatives**

**Training of African students, scientists, healthcare workers**

**Others:**

**SA – Brazil**

**SA - India**



A world map showing the distribution of major world religions. The map is color-coded: Christianity is shown in shades of blue and green, Islam in orange and red, Hinduism in yellow, Buddhism in light green, and other religions in various colors. Major countries are labeled, including the United States, Canada, Mexico, Brazil, Russia, China, India, and Australia. The map also shows the distribution of major world religions across the globe.

**“Genomics for Health in Africa”**  
**ARUA/The Guild**  
**Bern-Tuebingen-Stellenbosch Unis**

# Africa is ready!



Variant testing  
Single gene testing  
Gene panels  
Microarray  
Exome

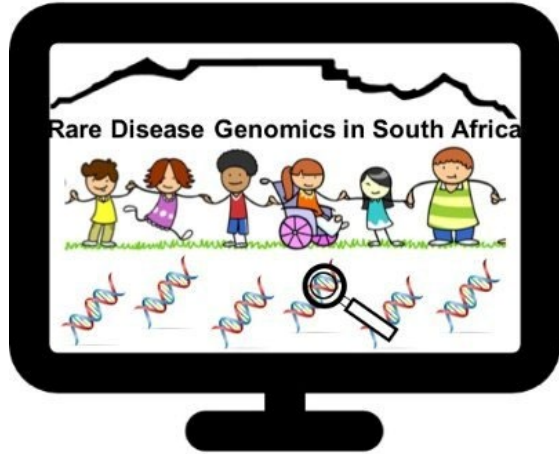


WGS



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# Thank you



 [RareGenomicsRSA](#)



**“Until every African with a  
#RareDisease is diagnosed!”**

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