SCHENTENDET ET CONSUL

South-North and South-South Collaborations in Medical Genomics Research in Africa

Gerrye Mubungu, MD, PhD



- 1. Experience of medical genomics in DRC
- 2. Challenges for establishing genetics and WGS
- 3. Present and future collaborations



1. Experience

Prof Gerrye Mubungu

Pediatrics and Genetics

Center for Human of Genetics/ CRMRND

University of Kinshasa

Center for Rare and Undiagnosed Diseases

Ensure access to diagnosis and multidisciplinary care for patients with rare and undiagnosed diseases

Provide training on the correct management of rare and undiagnosed diseases

Build and maintain a database and disseminate information related to rare and undiagnosed diseases

Organize the prevention of rare and undiagnosed diseases

Perform and supervise translational research targeting rare and undiagnosed diseases

Support organizations of parents and patients with rare and undiagnosed diseases

Perform, in collaboration with the Clinical Pharmacology Unit, clinical trials of orphan drugs and gene therapies

Develop therapeutic protocols for rare and undiagnosed diseases adapted to local realities and meeting international standards



Clinical services



Figure 1. Clinical phenotypes



- Intellectual Disability/Developmental delay
- Inherited Retinal Disease
- Skeletal dysplasia
- Skin & Pigmentary diseases

The University Hospitals and

outside in the specialized schools





Training of the care providers





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Educate the population and potential research participants Create trust and aware







Community engagement





2. Challenges for establishing genetics and WGS



1 patient = 1 geneticist + n Non-geneticists







2. Challenges for establishing genetics and WGS





3. Present and future collaborations



The main source of access to genomic sequencing for rare diseases patients in Africa is research and philanthropy.



High "potential" demand, low offer



NGS provided opportunity to reach diagnosis

Ihope (WGS) : N= 44



- Negative
- Pathogenic/LP variants
- VUS

DDD_DRC (WES) : N=68

DD variants found	n	%
Patients sequenced	68	100
Pathogenic/Likely Pathogenic	25	36.8
VUS	13	19.1
No relevant SNVs	30	44.1

PROSPECT COLLABORATION



- Interactions +++ between African

countries(Sud-Sud)

- Interactions +++ Nord-Sud

- Projects importants for Africa
- Capacity building
- Infrastructures for genomic

research

- Training local researchers
- Well-defined Data management



Acknowledgements





Thank you for your attention

