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

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OUTLINE

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

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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

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

Support organizations of parents and patients with rare and undiagnosed diseases
Clinical services

The University Hospitals and outside in the specialized schools

Figure 1. Clinical phenotypes

- Intellectual Disability/Developmental delay (48%)
- Inherited Retinal Disease (14%)
- Skeletal dysplasia (6%)
- Skin & Pigmentary diseases (9%)
- Other (3%)

20%
Phenotype and Sample QC filters Ref Pop freq Freq in Disease pop In silico Inheritance Sample QC Beacon Or MatchMaker

- Medical information
- Genetic testing
- General Clinical examination
- Dysmorphism evaluation
Training of the care providers
Educate the population and potential research participants
Create trust and aware
2. Challenges for establishing genetics and WGS

- Area: 2,345 millions km²
- Population: 95.894.118 hab.

- Limited clinical and molecular expertise
- Poorly organized health system
- Heavy burden of mystical belief and stigmatization
- No governmental support for research
- No access to genetic diagnostic
- No access to reference data
- Depth of genomic diversity unknown
- 4 geneticists
1 patient = 1 geneticist + n Non-geneticists

Need for capacity non-geneticists
2. Challenges for establishing genetics and WGS

- Insufficient collaboration
- No data sharing
- Limited expertise
- Real Necessity of Genomic
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

DDD_DRC (WES) : N=68

<table>
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<tr>
<th>DD variants found</th>
<th>n</th>
<th>%</th>
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<tbody>
<tr>
<td>Patients sequenced</td>
<td>68</td>
<td>100</td>
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<tr>
<td>Pathogenic/Likely Pathogenic</td>
<td>25</td>
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<tr>
<td>VUS</td>
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<tr>
<td>No relevant SNVs</td>
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<td>44.1</td>
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</table>
- 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

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