BIOGRAPHICAL SKETCH

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NAME: LUMAKA ZOLA, Aimé

eRA COMMONS USER NAME (credential, e.g., agency login): AIMELZ

POSITION TITLE: Associate Professor of Genetics and Pediatrics

EDUCATION/TRAINING *(Begin with baccalaureate or other initial professional education, such as nursing, include postdoctoral training and residency training if applicable. Add/delete rows as necessary.)*

INSTITUTION AND LOCATION	DEGREE *(if applicable)*	Completion Date MM/YYYY	FIELD OF STUDY
University of Kinshasa (UNIKIN), Faculty of Medicine	Master	06/2005	Medicine
University of Kinshasa (UNIKIN), Faculty of Medicine	Specialization	06/2012	Pediatrics
University of Leuven (KULeuven) Belgium	Certificate	06/2012	Human Genetics
University of Leuven (KU Leuven) Belgium	PhD	08/2015	Human Genetics

Please refer to the Biographical Sketch sample in order to complete sections A, B, C, and D of the Biographical Sketch.

A. Personal Statement

Strong research experience:
I obtained my PhD degree at the KU Leuven (Belgium). During my PhD research, I personally conducted the consent process and the recruitment of patients from specialized institutions across Kinshasa, in DR Congo, in strict accordance to the ELSI. The strong influence of religious and mystical believes in Sub-Saharan Africa, makes challenging the implementation of research studies that require blood sampling. Luckily, I was able to develop a unique approach that facilitated a trust-relationship with families. I built a large network of institutions and initiated patients' organisations. This ability to connect with patients and institutions is a key feature for researchers in developing countries.
Moreover, during our thesis, I was strongly involved in the implementation of the first DNA extraction facility in DR Congo. This was another challenge since water supply and furniture in electricity are irregular and unstable in sub-Saharan Africa. We had to identify local resources and apply them in order to circumvent energy problems and ensure the proper functioning of the new facility. That ability to solve serious problems using local resources is one of the contributions I will bring to the Consortium.
I have acquired a strong experience in the identification of known and new mutations in known genes as well as in the identification of novel candidate genes. I personally applied a wide range of genetic testing going from the G-banding karyotype to the Whole Genome Sequencing. In addition, I also gained experience in the clinical characterisation of genetic disorders including: investigation on family and patient's history, drawing and interpreting pedigree, identification of possible transmission model and identification, description of morphological traits in patients using standard terminology, and establish or exclude concordances with other patients in the family, in the research cohort and/or in the literature.
I have the ability to work flexible hours. I'm familiar with working in a group, having intra and inter group's exchanges, contributing to discussions, producing data, preparing and reporting data.

Experience in developing and handling large database:
During my PhD I acted as both data-entry clerk and database manager. My research dataset contained a total of 298 individuals, including relatives and parents. I collected and recorded their contacts, demographic and clinical data, information about samples, tests performed, future plans and other important observations.

Moreover, due to my background of medical doctor, I had previous experience with data management, ethical and legal issues. That previous aptitude was helpful during my thesis.

B. Positions and Honors

List in chronological order previous positions, concluding with the present position. List any honors. Include present membership on any Federal Government public advisory committee.

Positions and Employment

2006-2008	Consultant, SS Assistance/Noir et Blanc, DR Congo
2008-2012	Fellow, Department of Pediatrics, University of Kinshasa, DR Congo
2011-2012	Predoctoral Fellow, Department of Human Genetics, KU Leuven, Belgium
2011-2015	Lecturer, Department of Pediatrics, University of Kinshasa, DR Congo
2013-	Adjunct-Head of Department, Department of Genetics, Institut National de Recherche Biomédicales, DR Congo
2015-	Adjunct Director of the Center for Human Genetics, University of Kinshasa, DR Congo
2015-	Assistant Professor, Department of Pediatrics, University of Kinshasa, DR Congo

Other Experience and Professional Memberships

2011-	Member, Belgian Society for Human Genetics (BeSHG)
2012-	Member, Sociètè des Pédiatres du Congo Démocratique (SOPECOD)
2013-	Founding Member, Congolese Society for Human Genetics (CoSHG)

Honors

2014	Best poster at the 15th congress of the BeSHG 2015, Belgium

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C. Contribution to Science

1. **Description of the phenotype associated with known genetic conditions**.

Only few patients with African ethnicities are reported in the literature and in clinical databases. Moreover, it is well known that some facial characteristics such as thick lips, broad nose are frequent in people with African ethnicities. Therefore, the clinical presentation of patient with African ethnicities carrying a known condition is still unknown for many syndromes. My publications have shown that the phenotype in African patients with a known genetic condition may be different from what is reported for Caucasian. Through some case reports, we were able to show that this difference is a major cause for delayed/missed diagnostic, delayed care and paucity of African patients in databases.

a. **Lumaka A**, Bone D, Lukoo R, Mujinga N, Senga I, Tady B, Matthijs G, Lukusa TP. Werdnig-Hoffmanndisease: Report of the first case clinically identified and genetically confirmed in central Africa (Kinshasa-Congo)(2009). Genetic Counseling. 20(4):349-358.
b. Mbuyi-Musanzayi S, **Lumaka A**, Yogolelo Asani B, Lubala Kasole T, Lukusa Tshilobo P, Kalenga Muenze P, Tshilombo Katombe F, Devriendt K.(2014). Preaxial polydactyly of the foot: variable expression of trisomy 13 in a case from central Africa. Case Rep Genet.; 2014:365031.
c. Gerrye Mubungu, **Aimé Lumaka**, Rosette Matondo, Gloire Mbayabo, Deborah Tuka, Claudarche Kayembe, Didier Mulowhe, Antoine Molua, Bruno-Paul Tady, Emmanuel Nkidiaka, Paulo Bunga, Prosper Lukusa Tshilobo, Koenraad Devriendt (2014). Skinfold over toenail is pathognomonic for the popliteal pterygium syndrome in a Congolese family with large intrafamilial variability. Clinical Case Reports doi: 10.1002/ccr3.101
d. Mbuyi-Musanzayi S, Lubala Kasole T, **Lumaka A**, Kayembe Kitenge T, Kabamba Ngombe L, Kalenga Muenze P, Lukusa Tshilobo P, Tshilombo Katombe F, Banza Lubaba Nkulu C, Devriendt K. Meningocele in a congolese female with beckwith-wiedemann phenotype. Case Rep Genet. 2014;2014:989425.

e. **Lumaka, A**., Lukoo, R., Mubungu, G., Lumbala, P., Mbayabo, G., Mupuala, A., Lukusa Tshilobo P. and Devriendt, K. (2016), Williams–Beuren syndrome: pitfalls for diagnosis in limited resources setting. Clinical Case Reports. doi: 10.1002/ccr3.476

2. **Clinical consequences of genome variability of people of African Ethnicities.**

As only few patients from Africa have been described in existing databases, one can question whether the known genetic defects are the same in African as in Caucasians. We have released few cases reports showing that testing African patients may reveal novel genetic defects and suggest novel mechanisms for known diseases.

a. **Lumaka A**, Mubungu G, Nsibu C, Tady BP, Lukusa T, Devriendt K (2012). X-linked adrenal hypoplasia congenita: a novel DAX1 missense mutation and challenges for clinical diagnosis in Africa. Eur J Pediatr.; 171(2):267-70
b. **Lumaka Aimé**, Mubungu Gerrye, Mukaba Papino, Mutantu Pierre, Luyeye Gertrude, Corveleyn Anniek, Tady Bruno-Paul, Lukusa Tshilobo Prosper, Devriendt Koenraad. (2014). A novel heterozygous mutation of three consecutive nucleotides causing Apert Syndrome in a Congolese family. Eur J Med Genet.; 57(4):169-173.
c. Toni Kasole LUBALA, **Aimé LUMAKA**, Sébastien MBUYI-MUSANZAYI, Olivier MUKUKU, Didier MALAMBA-LEZ, Oscar Numbi LUBOYA, Prosper LUKUSA-TSHILOBO. (2015). Evidence of fragile X syndrome with mosaic size mutation in Bantu patient from Central Africa (submitted)

3. **Sickle Cell Anemia**

SCA is the first genetic disease and severely affect population of DR Congo, I have involved myself in the exciting area of SCD research. The first project targeted genetic factors influencing the fetal hemoglobin values and the clinical presentation of SCD in Congolese patients. We have reported that hematological parameters and genetic factors behind the elevated residual values of fetal hemoglobin are associated with less severe forms of the disease.

a. Michel Ntetani Aloni; Tite Minga Mikobi; **Aimé Zola Lumaka**; Didine Kinkodi Kaba; Jean Marie Mbuyi-Muamba; Prosper Lukusa Tshilobo; Koenraad Devriendt; Gert Matthijs; Valérie Race. (2015). Protective BCL11A and HBS1L-MYB polymorphisms in Congolese Sickle Cell Anemia Patients. (Submitted to Plos One)
b. Tite Minga Mikobi, **Aimé Lumaka**, Michel Ntetani Aloni, Didine Kinkodi Kaba, Jean-Marie Mbuyi Muamba, Prosper Lukusa Tshilobo, Koenraad Devriendt, Gert Matthijs, Valérie Race : Genetic Cofactors Modulators in Congolese Sickle Cell Anemia patients. Ann. Afr. Med., 2014, 7(4): 1752 – 1761.

D. Research Support

I acknowledge having never applied or received funding from NIH agencies before. However, I have benefited from supports from funding agencies in Europe as listed below.

Ongoing Research Support

VLIR-UOS TEAM- project (Belgium) Aimé (Co-Investigator) 10/04/15-31/12/18
Title: Sickle Cell Disease in DR Congo and determinant of Hydroxy urea treatment
The goal of this study is to prepare the implementation of hydroxyl-urea treatment at large scale in DR Congo. This study will determine whether there are specific criteria for selection of patients to receive HU and evaluate genetic and non-genetic factors that can influence that treatment in young and adult Congolese SCD patients.
Role: Co-Investigator

Fond Mérieux (France), Aimé (Co-PI) 01/02/15-31/12/15
Title: Genetic determinant of Ebola infection outcome
During Ebola virus infection majority of patients die while only very few survive. The goal of this project is to identify genetic determinants of the outcome during Ebola virus infection using PBMC collected during the recent Ebola outbreak in DR Congo.
Role: Co-PI

Own Funding Aimé (Co-Supervisor) 01/03/15-31/12/16
Title: Association between genetic polymorphisms and pulmonary functions in SCD.
This project aims at evaluating respiratory functions in SDC patients in DR Congo and investigates further association between pulmonary functions and carriage of certain polymorphisms influencing fetal hemoglobin values.
Role: Co-supervisor

Completed Research Support

KUL-IRO (Belgium) Aimé (young Investigator) 01/10/12-30/09/15
Title: Genetics Etiological study of Intellectual Disability in Kinshasa
The goal of this study was to determine causal genetic defects in patients affected with ID and followed in specialized institutions for ID across Kinshasa.
Role: Investigator