BIOGRAPHICAL SKETCH

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NAME: LUKUSA TSHILOBO, Prosper

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

POSITION TITLE: Professor of Paediatrics and Clinical Cytogenetics

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
UNAZA (Université Nationale du Zaïre), Kinshasa, DR Congo	MD	07/1974	Medicine
UNAZA (Université Nationale du Zaïre), Kinshasa, DRC Congo	Paediatrician	06/1979	Pediatrics
W.H.O.– I.N.S.E.R.M. (Paris)	DIploma	06/1980	Epidemiology
KUL (Katholieke Universiteit Leuven), Belgium	Ph.D	06/1991	Medical Genetics

A. Personal Statement

I was trained as pediatrician in the University of Kinshasa in DR Congo. As pediatrician, I conducted a research study that identified the etiological factors underlying intra-uterine growth retardation in DR Congo. I also described the growth parameters of small for age newborns and questioned the accuracy of Last Menstrual Period for the calculation of fetal age. Later I joined the KU Leuven for my PhD training in Human Genetics. After my PhD, I worked in the Center for Human Genetics of KU Leuven for about 30 years and during this period I accumulated strong expertise in research that I knew would be useful for Africa. I also conducted researches on Y chromosome and sexual morbidity published in international journals.

Later, I joined my home country in order to promote research in genetics of human development and contribute to the improvement of genetic services to patients. We have, for the first time, implemented a DNA extraction facility in Kinshasa and established the first molecular diagnostic facility for Sickle Cell Anemia (SCA) in the center of Africa.

I'm co-founder and current president of the Congolese Society for Human Genetics.

I initiated a research project on genetic etiology of developmental disorders and intellectual disability in Kinshasa. This study is completed. Other project are investigating the facial dysmorphism in Congolese patients (see publications), congenital malformations in DR Congo and specific clinical phenotype in well-known genetic conditions.

Beside the genetic of ID/DD, I launched a small project aiming to study genetic and environmental modifiers of the response to the hydroxyurea treatment among Congolese patients. Of interest, I have initiated a first research project to elucidate genetic and biochemical determinants of the clinical variability in SCA among Congolese patients (see publications). We expect new guidelines for treatment, with treatment to be proposed based on the results of this research. In addition, we are launching another research project aiming to explore association between respiratory functions and some genetic markers.

All those projects are carried out in collaboration with other researchers and institutions involved in similar researches.

Altogether, my research expertise and contribution to the science are in the field of genetics, mainly the genetics of development and cognition, and secondarily in SCA.

B. Positions and Honors

Positions and Employment

1974 - 1979: Assistant-Lecturer in Paediatrics, UNAZA, Kinshasa.
1979 - 1981: Lecturer in Neonatology, Department of Paediatrics, UNAZA, Kinshasa.
1980 - 1983: Supervisor of Interns and Residents in Paediatrics, UNAZA, Kinshasa.
1981 - 1084: Associate Professor in Paediatrics, University of Kinshasa (UNIKIN), DRC.
1984 - 1991: Research Fellow in Medical Genetics, Faculty of Medicine, K.U.L, Belgium.
1991 - 1999: Post Doc Researcher in Medical Genetics, K.U.L., Leuven, Belgium.
1999 - 2011: Research Associate, Centre for Human Genetics, K.U.L.
Since 2008: Appointed as IOM (International Organization for Migration) Expert for Human Genetics in central Africa
Since 2011: Resident Professor, Human Genetics and Pediatrics, UNIKIN – UNILU.
Since 2013: Head Medical Genetics Department, INRB (Institut National de Recherche Biomédicale).
Since 2015: Director Laboratory and Centre for Human Genetics, University of Kinshasa

Scientific Societies and Professional Memberships

- 2006: Member Belgian Society of Human Genetics
- 2006: Member European Society for Human Genetics
- 2008: Member Belgian Society of Pediatrics
- 2012: Member «Société de Pédiatrie du Congo Démocratique»
- 2013: Founding member and President Congolese Society for Human Genetics

Honors

2014: Honorary Member Belgian Society of Human Genetics

C. Contribution to Science

1. Contribution related to genetics of development and cognition

Developmental genetic condition may be characterized by clinical manifestations making it recognizable by clinicians. However, better delineation of clinical entities requires a large number of patients (cohorts or families). Genetic defect must be undoubtedly determined so that phenotype-genotype correlation may be established. We described clinical presentation and genetic particularities of patients carrying certain well-known syndromes. Our research tackled chromosomal syndromes, including duplications (1q, 2q,7q, 9p, 12q) and deletions (2q, 7q, 10q, 11q) as well as monogenic disorders.

- Lukusa T, Van Buggenhout G, Devriendt K, Meireleire J, Van Goethem G, Roelen L, Fryns JP. Zygodactyly as the most striking physical anomaly in an adult male patient with pure partial trisomy 1q. Ann Genet. 1998; 1(4):199-204.
- Lukusa T, Fryns JP. Syndrome of facial, oral, and digital anomalies due to 7q21.2-->q22.1 duplication. Am J Med Genet. 1998; 80(5): 454-458.
- Lukusa T, Devriendt K, Holvoet M, Fryns JP. Severe mental retardation-distal arthrogryposis in the upper limbs and complex chromosomal rearrangements resulting from a 10q25-->qter deletion. Clin Genet. 1998; 54(3): 224-230.
- Lukusa T, Devriendt K, Jaeken J, Fryns JP. Mild dysmorphic signs in two male sibs with partial trisomy 2q32.1-->q35 due to maternal ins(14;2) translocation. Clin Dysmorphol. 1999; 8(1): 47-51.
- Lukusa T, Devriendt K, Holvoet M, Fryns JP. Dicentric chromosome 9 due to tandem duplication of the 9p11-q13 region: unusual chromosome 9 variant. Am J Med Genet. 2000; 91(3): 192-197
- Lukusa T, Holvoet M, Vermeesch JR, Devriendt K, Fryns JP. Partial monosomy 11q and trisomy 12q: variable expression in two siblings. Genet Couns. 2003; 14(2): 155-164.
- Lukusa T, Vermeesch JR, Holvoet M, Fryns JP, Devriendt K. Deletion 2q37.3 and autism: molecular cytogenetic mapping of the candidate region for autistic disorder. Genet Couns. 2004;

15(3): 293-301.

- Lukusa T, Vermeesch JR, Fryns JP. De novo deletion 7q36 resulting from a distal 7q/8q translocation: phenotypic expression and comparison to the literature. Genet Couns. 2005; 16 (1): 1-15.

2. Contribution to description of dysmorphic features in African patients with known genetic defects:

- Lumaka, A., Bone, D., Lukoo, R., Mujinga, N., Senga, J., Tady, B., Matthijs, G., **Lukusa-Tshilobo, P**. (2009). Werdnig-Hoffmann disease: Report of the first case clinically identified and genetically confirmed in central Africa (Kinshasa-Congo). *Genetic Counseling, 20* (4), 349-358.
- Lumaka Zola, A., Mubungu Lumbono, G., Nsibu Ndosimao, C., Tady Muyala, B., **Lukusa-Tshilobo, P**., Devriendt, K. (2012). X-linked adrenal hypoplasia congenita: a novel DAX1 missense mutation and challenges for clinical diagnosis in Africa. *European Journal of Pediatrics, 171* (2), 267-270.
- Lumaka A, Mubungu G, Mukaba P, Mutantu P, Luyeye G, Corveleyn A, Tady B-P, **Lukusa Tshilobo P**, Devriendt K. (2014). A novel heterozygous mutation of three consecutive nucleotides causing Apert syndrome in a Congolese family. *European Journal of Medical Genetics,* 57(4):169-173; doi: 10.1016/j.ejmg.2014.01.004. Epub 2014 Jan 28
- Sébastien Mbuyi-Musanzayi,[1,2] Aimé Lumaka,[3,4] Bienvenu Yogolelo Asani,[2,5] Toni Lubala Kasole,[2,6] **Prosper Lukusa Tshilobo**,[3,4] Prosper Kalenga Muenze,[2,7]François Tshilombo Katombe,[1] and Koenraad Devriendt[3] (2014). Preaxial Polydactyly of the Foot: Variable Expression of Trisomy 13 in a Case from Central Africa. Case Reports in Genetics, Article ID 365031
- Mubungu G, Lumaka A, Matondo R, Mbayabo G, Tuka D, Kayembe C, Mulowhe D, Molua A, Tady BP, Nkidiaka E, Bunga P, **Lukusa Tshilobo P**, Devriendt K. (2014) Skinfold over toenail is pathognomonic for the popliteal pterygium syndrome in a Congolese family with large intrafamilial variability. Clin Case Rep., 2(6): 250-253 ; doi: 10.1002/ccr3.101. Epub 2014 Sep 15.
- 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. (2014). Meningocele in a congolese female with Beckwith-Wiedemann phenotype. Case Rep Genet, Article ID 989425; doi: 10.1155/2014/989425. Epub 2014 Dec 28 (4 pages)
- Lubala TK, Mbuyi-Musanzayi S, Lubala N, Luboya ON, Kalenga PM, Devriendt K, **Lukusa PT**. (2015). Mirror-image gastroschisis in monochorionic female twins. Eur J Med Genet. 2015 Mar 14. pii: S1769-7212(15)00057-9. doi: 10.1016/j.ejmg.2015.03.001.
- **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

3. Contribution related to sickle cell disease (SSD)

DR C is ranking third among the most affected countries for SCD. About 1 in 3 inhabitants are heterozygous and 2% are homozygotes. The last group experiences the most severe manifestations of the diseases including episodes of severe anemia, bone and joint pains or chest pain. Even without these acute manifestations, SCD patients permanently suffer hemolytic anemia that manifests by jaundice of bulbar conjunctives that is associated to delayed growth. Studies have shown that severity of manifestations is inversely correlated with residual fetal hemoglobin values. Hydroxy-urea has proven to be an efficient medication that increases values of fetal hemoglobin and improves the quality of live in treated SCD patients. Our researches looked for hematological, biochemical and genetic modifiers of the disease severity. We also interrogated the influence of parvovirus B19 on the course of the disease.

Related publications:

- Mikobi TM, **Lukusa Tshilobo P**, Aloni MN, Mvumbi Lelo G, Akilimali PZ, Muyembe-Tamfum JJ, Race V, Matthijs G, Mbuyi Mwamba JM (2015). Correlation between the Lactate Dehydrogenase Levels with Laboratory Variables in the Clinical Severity of Sickle Cell Anemia in Congolese

Patients. PLOS ONE 10(5): e0123568.doi:10.1371/journal.pone.0123568

- 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.
- Tite Minga Mikobi, Jean-Marie Mbuyi-Muamba, Michel Ntetani Aloni, Georges Lelo Mvumbi, Pierre Zalagie Akilimali, Jean-Jacques Tamfum Muyembe, **Prosper Tshilobo Lukusa** : Iron status and hematologie parameters modifications in congolese suffering from Sickle Cell Anemia. Ann. Afr. Med., 2014, 7(4): 1762 – 1770
- Pierre Nsele Mutantu, Tite Minga Mikobi, Angèle Dilu-Keti, Mwepu Ngandu, Angèle Palaba, Florence Matsieba, Tony Wawina, **Prosper Lukusa**, Jean-Jacques Muyembe, Steve Ahuka-Mundeke: Infection à parvovirus B19 chez les drépanocytaires suivis au Centre de Médecine Mixte et Anémie SS. Ann. Afr. Med., 2015, 7(2): 1641 – 1642.

D. RESEARCH SUPPORT

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

Ongoing Research Support

VLIR-UOS TEAM- project (Belgium) Prosper (Co-PI) 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-PI

Fond Mérieux (France), Prosper (PI) 01/02/15-31/12/15
Title: Genetic determinant of Ebola infection outcome
During Ebola virus infection majority of patients dye 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: PI

Own Funding Prosper (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: Supervisor

Completed Research Support

KUL-IRO (Belgium) Prosper (Co-PI) 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: Co-PI