BIOGRAPHICAL SKETCH

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NAME: Wonkam, Ambroise

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

POSITION TITLE: Professor and Director of GeneMAP (Genetic Medicine of African Populations) Research Center

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 Yaounde, Cameroon	M.D.	1995	Medicine
School of Genetic Medicine, Italy	Certificate	2002	Human Genetics
Bar Harbor Courses, USA	Certificate	2003	Human Genetics
University of Geneva, Switzerland	D.MedSc.	2003	Cell Biology
University of Geneva, Switzerland	Specialist Diploma	2005	Medical Genetics
University of Cape Town, South Africa	Ph.D.	2015	Human Genetics

A. Personal Statement

Dr. Ambroise Wonkam is Professor and Director, of the McKusick-Nathans Institute, and Department of Genetic Medicine, Johns Hopkins University, School of Medicine Baltimore, USA. He also hods a position of professor of medical genetics, and Director of GeneMAP (Genetic Medicine of African Populations), and immediate past Deputy Dean Research at the Faculty of Health Sciences, University of Cape Town, South Africa.

After a MD training from the Faculty of Medicine and Biomedical Sciences, University of Yaoundé I (Cameroon), he completed a thesis in Cell Biology in the department of Morphology, and a specialist traing as medical geneticist at University of Geneva (Switzerland) and a PhD in Human Genetics (University of Cape Town, South Africa). His research interests and international recognition by the academic community are reflected in > 230 peer-reviewed publications.

Dr. Wonkam has been investigating numerous monogenic conditions of High burden in Africa, e.g. Sickle cell disease and inheritable hearing Impairment in Africa with focus on the use of genetics in public health intervention. He has a traceable record of studying genomic factors that affect the SCD phenotype, specifically studying HbF-promoting loci and co-inheritance of SCD and alpha-thalassemia in Cameroon and genomic variants affecting Kidney Dysfunctions. Moreover, his research has emphasized the huge genetic and locus heterogeneity in rare diseases in Africa including congenital hearing loss, and particularly genes variants, and novel genes, as well as innovative finding in the genetic architecture that will inform the pathobiology globally. In this application, he will build of what is presumed to be the World's largest collection of Whole Exome Sequencing Data of individual living with Hearing impairment, with African ancestry. He has successfully advocated for more focus on Intellectual (syndromic and non-syndromic) by contributing to establish a section on Case report in Diverse Population in the American Journal of Medical Genetics (PMC7255818).Dr Wonkam was the principal investigator (PI) of a NIH funded H3Africa grant aiming to examine ethical issues relating to sickle cell genomics research in Cameroon, Tanzania and Ghana. Since 2017, He established the **Sickle Africa Data Coordinating Center (SADaCC)** to support the activities of the Sickle Pan African Research Consortium (SPARCo) site in Tanzania, Nigeria, Mali, Uganda, Zimbabwe, Zambia and Ghana (https://www.sickleinafrica.org/). SADaC and SickleInAfrica have built significant infrastructure to contribute in many ways to the current proposal.

a. **Wonkam A**, Chimusa ER, Mnika K, Pule GD, Ngo Bitoungui VJ, Mulder N, Shriner D, Rotimi CN, Adeyemo A. Genetic modifiers of long-term survival in sickle cell anemia. *Clin Transl Med*. 2020 Aug;10(4):e152. PMCID: PMC7423184
b. Makani J, Sangeda RZ, Nnodu O, Nembaware V, Osei-Akoto A, Paintsil V, Balandya E, Kent J, Luzzatto L, Ofori-Acquah S, Olopade OI, Pallangyo K, Minja IK, Jonas M, Mazandu GK, Mulder N, Ohene-Frempong K, **Wonkam** A. SickleInAfrica. *Lancet Haematol.* 2020 Feb;7(2):e98-e99. PMCID: PMC7255817
c. Geard A, Pule GD, Chetcha Chemegni B, Ngo Bitoungui VJ, Kengne AP, Chimusa ER., **Wonkam A.** Clinical and genetic predictors of renal dysfunctions in sickle cell anemia in Cameroon*, Br J Haematol.* 2017 Aug;178(4):629-639. PMCID: PMC5660286
d. Makani J, Ofori-Acquah SF, Tluway F, Mulder N, **Wonkam** A. Sickle cell disease: Tipping the balance of genomic research to catalyse discoveries in Africa. *Lancet.* 2017 Jun 17;389(10087):2355-2358. PMCID: PMC5612389

Ongoing and recently completed projects that I would like to highlight include:

U24 HL135600
Wonkam (PI)
04/01/2017 – 04/30/2026
Sickle Africa Data Coordinating Center (SADaCC)

U01 HG007459
Wonkam (PI)
09/30/2013 – 07/31/2017
Exploring perspectives in sickle genomic and public health interventions

U01 HG009716
Wonkam (PI)
09/15/2017 – 06/30/2023
Hearing Impairment Genetics Studies in Africa (HI-GENES Africa)

AESA/Welcome Trust (H3AFull/17/001)
Wonkam (PI)
09/01/2018 – 08/31/2021
Hearing Impairment Genetics Studies in Africa (HI-GENES Africa)

R01DC016593
Leal (PI)
09/01/2019 – 08/31/2023
Identification and Functional Evaluation of Autosomal Recessive Non-Syndromic Hearing Impairment Genes in sub-Saharan Africans

U01 MH127692
Wonkam (PI)
09/20/2021 – 07/31//2026
Understanding of Big data in Genomics Medicine in Africa (PUBGEM-Africa)

U54 HG009790
Wonkam (PI)
9/20/2017 – 06/30/2023
Individual Findings in Genetic Research in Africa (IFGeneRA)

Welcome Trust (107755Z/15/Z)
Aswandare (PI)
09/01/2021 – 09/31/2025
WACCBIP II-DELTAS

B. Positions, Scientific Appointments, and Honors

Positions and Employment

2022- Director, of McKusick-Nathans Institute, and Department of Genetic Medicine, Johns Hopkins University School of Medicine Baltimore, MD 21205, USA
2018 - 2021 Deputy Dean Research, Faculty of Health Sciences, University of Cape Town, South Africa
2017 - Director GENEMAP (Genetic Medicine of African Populations) Research Center
2016 - Professor/senior consultant, Division of Human Genetics, Department of Medicine, Faculty of Health Sciences, University of Cape Town, South Africa
2012 - 2015 Associate Professor/Senior Consultant, Division of Human Genetics, Department of Medicine, Faculty of Health Sciences, University of Cape Town, South Africa
2009 - 2012 Senior lecturer / senior specialist, Division of Human Genetics, Department of Medicine, Faculty of Health Sciences, University of Cape Town, South Africa
2007 - 2009 Senior lecturer /consultant/ Head, Genetics and Developmental Biology & Service of Medical genetics Gyn&Obst, Faculty of Medicine and Biomedical Sciences- University of Yaoundé I – Cameroon & Pediatric Hospital, Yaoundé – Cameroon
2005 - 2006 Medical geneticist, consultant/lecturer, Service of Medical Genetics, Geneva University Hospital (HUG); Faculty of Medicine, University of Geneva, Switzerland
2001 - 2005 *Medical intern (registrar) in Medical genetics,* Service of Medical Genetics, Geneva University Hospital (HUG), Faculty of Medicine, University of Geneva, Switzerland
1998 - 2001 *Research fellow in Cell biology,* Department of Morphology, Faculty of medicine, University of Geneva (Switzerland)

Other Experience and Professional Memberships

2021- H3Africa consortium, Chair
2019 - Associated Editor of the *American Journal of Human Genetics*
2018 - Associate Editor the *Journal of Community Genetics*
2018 - Academic Editor of *PLOS One*
2017 - President of the *African Society of Human Genetics*
2017 - Associate Editor of the *American Journal of Medical Genetics*

Honors

2024 Fellow, The world Academy of Sciences (UNESCO)
2023 Human Genome Organization Faculty Scholar
2020 **Alan Pifer Award,** the prestigious award is the UCT vice-chancellor's annual prize in recognition of outstanding welfare-related research.
2014 Human Genome Organization, future of Genetic Award
2014 Clinical Genetics Society International Award, British Society of Genetic Medicine, UK
2009 3rd March of Dime International Conference on Birth Defect and Disability (ICBD) Award, New Delhi, India
2003 Denber-Pinard Prize for the best thesis, Faculty of Medicine, University of Geneva, Switzerland

Current Memberships

- African Society of Human Genetics, President
- American Society of Human Genetics, Associate Editor, *American Journal of Human Genetics*
- European Society of Human Genetics, Member
- International Federation of Human Genetics Societies, Board member

B. Contributions to Science

1. Sickle Cell Disease: Genetic and Public Health Intervention

Dr. Wonkam has used genetics to address public health interventions. He has tractable record of studying psychosocial burden of SCD and genomic factors that affect the SCD phenotype, specifically HbF-promoting

loci and co-inheritance of SCD and alpha-thalassemia in Cameroon. He is PI of an NIH funded H3Africa grant aiming to examine ethical issues relating to sickle cell genomics research in Cameroon, Tanzania and Ghana (http://www.h3africa.org/consortium/projects).

a. **Wonkam A**, Chimusa ER, Mnika K, Pule GD, Ngo Bitoungui VJ, Mulder N, Shriner D, Rotimi CN, Adeyemo A. Genetic modifiers of long-term survival in sickle cell anemia. *Clin Transl Med*. 2020 Aug;10(4):e152. PMCID: PMC7423184
b. Makani J, Sangeda RZ, Nnodu O, Nembaware V, Osei-Akoto A, Paintsil V, Balandya E, Kent J, Luzzatto L, Ofori-Acquah S, Olopade OI, Pallangyo K, Minja IK, Jonas M, Mazandu GK, Mulder N, Ohene-Frempong K, **Wonkam** A. SickleInAfrica. *Lancet Haematol.* 2020 Feb;7(2):e98-e99. PMCID: PMC7255817
c. Geard A, Pule GD, Chetcha Chemegni B, Ngo Bitoungui VJ, Kengne AP, Chimusa ER., **Wonkam A.** Clinical and genetic predictors of renal dysfunctions in sickle cell anemia in Cameroon*, Br J Haematol.* 2017 Aug;178(4):629-639. PMCID: PMC5660286
d. Makani J, Ofori-Acquah SF, Tluway F, Mulder N, **Wonkam** A. Sickle cell disease: Tipping the balance of genomic research to catalyse discoveries in Africa. *Lancet.* 2017 Jun 17;389(10087):2355-2358. PMCID: PMC5612389

2. Intellectual disability in Diverse populations

Dr Wonkam has performed numerous researches on syndromic and non-syndromic condition associated with ID in Africa, notably his co-edit a section on Case report in Diverse Population in the American Journal of Medical Genetics.

a. Girisha KM, **Wonkam A,** Muenke M. Introducing in AJMG Part A: Case reports in diverse populations. *Am J Med Genet A.* 2018 Jul;176(7):1547-1548. PMC7255818
b. Kengne Kamga K, Nguefack S, Minka K, Wonkam Tingang E, Esterhuizen A, Nchangwi Munung S, De Vries J, **Wonkam A.** Cascade Testing for Fragile X Syndrome in a Rural Setting in Cameroon (Sub-Saharan Africa). *Genes (Basel).* 2020 Jan 28;11(2):136. PMCID: PMC7074341
c. Dowsett L, Porras AR, Kruszka P, Davis B, Hu T, Honey E, Badoe E, Thong MK, Leon E, Girisha KM, Shukla A, Nayak SS, Shotelersuk V, Megarbane A, Phadke S, Sirisena ND, Dissanayake VHW, Ferreira CR, Kisling MS, Tanpaiboon P, Uwineza A, Mutesa L, Tekendo-Ngongang C, **Wonkam A**, Fieggen K, Batista LC, Moretti-Ferreira D, Stevenson RE, Prijoles EJ, Everman D, Clarkson K, Worthington J, Kimonis V, Hisama F, Crowe C, Wong P, Johnson K, Clark RD, Bird L, Masser-Frye D, McDonald M, Willems P, Roeder E, Saitta S, Anyane-Yeoba K, Demmer L, Hamajima N, Stark Z, Gillies G, Hudgins L, Dave U, Shalev S, Siu V, Ades A, Dubbs H, Raible S, Kaur M, Salzano E, Jackson L, Deardorff M, Kline A, Summar M, Muenke M, Linguraru MG, Krantz ID. Cornelia de Lange syndrome in diverse populations. *Am J Med Genet A.* 2019 Feb;179(2):150-158. PMCID: PMC6367950
d. Kruszka P, Porras AR, de Souza DH, Moresco A, Huckstadt V, Gill AD, Boyle AP, Hu T, Addissie YA, Mok GTK, Tekendo-Ngongang C, Fieggen K, Prijoles EJ, Tanpaiboon P, Honey E, Luk HM, Lo IFM, Thong MK, Muthukumarasamy P, Jones KL, Belhassan K, Ouldim K, El Bouchikhi I, Bouguenouch L, Shukla A, Girisha KM, Sirisena ND, Dissanayake VHW, Paththinige CS, Mishra R, Kisling MS, Ferreira CR, de Herreros MB, Lee NC, Jamuar SS, Lai A, Tan ES, Ying Lim J, Wen-Min CB, Gupta N, Lotz-Esquivel S, Badilla-Porras R, Hussen DF, El Ruby MO, Ashaat EA, Patil SJ, Dowsett L, Eaton A, Innes AM, Shotelersuk V, Badoe Ë, **Wonkam A**, Obregon MG, Chung BHY, Trubnykova M, La Serna J, Gallardo Jugo BE, Chávez Pastor M, Abarca Barriga HH, Megarbane A, Kozel BA, van Haelst MM, Stevenson RE, Summar M, Adeyemo AA, Morris CA, Moretti-Ferreira D, Linguraru MG, Muenke M. Williams-Beuren syndrome in diverse populations. *Am J Med Genet A*. 2018 May;176(5):1128-1136. PMCID: PMC6007881

3. Genetic of Hearing Loss in Africa

Dr. Wonkam has a proven record on hearing loss research in Africa. Specifically, his research group has convincingly shown that Mutations in *GJB2*, *GJB6* and *GJA1* are not a major cause of non-syndromic deafness in Africans. Subsequently, he has investigated numerous African families using a massively parallel targeted sequencing platform, and Whole Exome sequencing to illustrate, and identified novel variants in known genes and numerous novel candidate genes.

a. **Wonkam A**, Manyisa N, Bope CD, Dandara C, Chimusa ER. Whole Exome Sequencing Reveals Pathogenic Variants in *MYO3A, MYO15A* and *COL9A3,* and Differential Frequencies in Ancestral Alleles in Hearing Impairment Genes Among Individuals from Cameroon. *Hum Mol Genet.* 2020 Oct 20:ddaa225. PMCID: PMC7861016
b. Oluwole OG, Esoh KK, Wonkam-Tingang E, Manyisa N, Noubiap JJ, Chimusa ER, **Wonkam A**. Whole exome sequencing identifies rare coding variants in novel human-mouse ortholog genes in African individuals diagnosed with non-syndromic hearing impairment. *Exp Biol Med (Maywood).* 2020 Sep 30:1535370220960388. PMCID: PMC7871117
c. Adadey SM, Tingang Wonkam E, Twumasi Aboagye E, Quansah D, Asante-Poku A, Quaye O, Amedofu GK, Awandare GA, **Wonkam A**. Enhancing Genetic Medicine: Rapid and Cost-Effective Molecular Diagnosis for a GJB2 Founder Mutation for Hearing Impairment in Ghana. *Genes (Basel).* 2020 Feb;11(2):132. PMCID: PMC7074138
d. Lebeko K, Sloan-Heggen CM, Noubiap JJ, Dandara C, Kolbe DL, Ephraim SS, Booth KT, Azaiez H, Santos-Cortez RL, Leal SM, Smith RJ, **Wonkam A**. Targeted genomic enrichment and massively parallel sequencing identifies novel nonsyndromic hearing impairment pathogenic variants in Cameroonian families. *Clin Genet.* 2016 Sept;90(3):288-290. PMCID: PMC5324826

4. **Capacity Building and Genetic Education**

Seminal work of Dr. Wonkam has provided evidence of the major need to increase genetic knowledge at all levels of medical education in Africa, through capacity building and the development of effective genetic services. He has invested in medical genetic research and reported on an effective framework to increase capacity in human and medical genetics in Africa.

a. McGuire AL, Gabriel S, Tishkoff SA, **Wonkam A**, Chakravarti A, Furlong EEM, Treutlein B, Meissner A, Chang HY, López-Bigas N, Segal E, Kim JS. The road ahead in genetics and genomics. *Nat Rev Genet.* 2020 Oct;21(10):581-596. PMCID: PMC7444682
b. **Wonkam A,** Njamnshi AK, Angwafo FF 3rd. Knowledge and attitudes concerning medical genetics amongst physicians and medical students in Cameroon (sub-Saharan Africa). *Genet Med.* 2006 Jun;8(6):331-338. PMID: 16778594
c. **Wonkam A,** Nature. 2021 Feb;590(7845):209-211. PMID: 33568829
d. **Wonkam A**, as member of the H3Africa Consortium. Research capacity. Enabling the genomic revolution in Africa. *Science.* 2014 Jun 20;344(6190):1346-1348. PMCID: PMC4138491

5. **Medical Research Ethics in Genetics**

Dr Wonkam has important work in research ethics in Africa, notably on equity, communities' engagement and, return of individual/incidental genetic results.

a. **Wonkam** A, de Vries J. Returning incidental findings in African genomics research. *Nat Genet.* 2020 Jan;52(1):17-20. PMCID: PMC7255819
b. **Wonkam** A, Hurst SA. Acceptance of abortion by doctors and medical students in Cameroon. *Lancet.* 2007 Jun 16;369(9578):1999. PMID: 17574089
c. **Wonkam** A, de Vries J, Royal CD, Ramesar R, Angwafo FF 3rd. Would you terminate a pregnancy affected by sickle cell disease? Analysis of views of patients in Cameroon. *J Med Ethics.* 2014 Sep;40(9):615-620. PMID: 23918815
d. **Wonkam** A, Kenfack MA, Muna WF, Ouwe-Missi-Oukem-Boyer O. Ethics of human genetic studies in sub-Saharan Africa: the case of Cameroon through a bibliometric analysis. *Dev World Bioeth.* 2011 Dec;11(3):120-127. PMID: 21781234

Complete List of Published Work in MyBibliography:
https://www.ncbi.nlm.nih.gov/myncbi/1bundm-HQAU5X/bibliography/public/ (*n* = 238)