

ADDENDUM (OUTCOMES OF ISHGG, 2016):

A) Publications:

1. **Adeyemo WL** Genetics and Genomics Etiology of Non-Syndromic Orofacial Clefts. Invited Commentary, MGGM. 2017. (Accepted).
2. **Ariani Y**, Soeharso P et al. Genetic and genomic medicine in Indonesia. 2017 (In preparation).
3. **Ariani Y**, Soeharso P et al. Loss of heterozygosity in children with multiple congenital anomaly. 2017. (In preparation).
4. **Ariani Y**, Priambodo R et al. Specific amino acid changes of congenital nephrotic syndrome in Indonesia. 2017 (In preparation).
5. **Ariani Y**, Priambodo R et al. Ancestral diversity; a global challenge to future disease understanding. 2017. (In preparation).
6. **Balbuena HR**. Genetics and Genomic Medicine in Cuba. 2017 (In preparation).
7. **Belhassan K**, Ouldin K, Sefiani AA. Genetics and genomic medicine in Morocco:the present hope can make the future bright. MGGM. 2016; 4:588-598.
8. Gowans LJJ, Busch TD, Mossey PA, Eshete MA, **Adeyemo WL** et al. The Prevalence, Penetrance and Expressivity of Aetiologic IRF6 variants in Orofacial Clefts Patients from sub-Saharan Africa. MGGM. 2017. (Accepted).
9. Kruszka P, Porras AR, Sobering AK, Ikolo FA, La Qua S, Shotelersuk V, **Uwineza A**, et al. Down syndrome in diverse populations. Am J Med Genet Part A. 2017; 173: 42-53.
10. Kruszka P, Porras AR, Sobering AK, Ikolo FA, La Qua S, Shotelersuk V, Chung BHY, Mok GTK, **Uwineza A**, Mutesa L, Moresco A, Obregon MG, Sokunbi OJ, Kalu N, Joseph DA, Ikebudu D, Ugwu CE, Okoromah CAN, Addissie YA, Pardo KL, Brough JJ, Lee N-C, Girisha KM, Patil SJ, Ng ISL, Min BCW, Jamuar SS, **Tibrewal S**, Wallang B, Ganesh S, **Sirisena ND**, Dissanayake VHW, Paththinige CS, Prabodha LBL, Richieri-Costa A, **Muthukumarasamy P**, Thong M-K, Jones KL, Abdul-Rahman OA, **Ekure EN**, et al. Down syndrome in diverse populations. Am J Med Genet Part A. 2017; 173A:42–53.
11. Kruszka P, Addissie YA, McGinn DE, Porras AR, Biggs E, Share M, Crowley TB, Chung BHY, Mok GTK, **Muthukumarasamy P**, ThongM-K, **Sirisena ND**, Dissanayake VHW, Paththinige CS, Prabodha LBL, Mishra R, Shotelersuk V, **Ekure EN**, et al. 22q11.2 Deletion Syndrome in Diverse Populations. Am J Med Genet Part A. 2017. (In press).
12. Kruszka P, **Muthukumarasamy P**. et al. Noonan Syndrome in Diverse Populations. 2017. (In preparation).
13. **Okafor FU**. Prevalence And Projection Of Retinoblastoma In Tertiary Health Institution In Edo State, Nigeria. 2017. (in preparation).
14. Putoux A, Alqahtani A, Pinson L, Paulussen AD, Michel J, Besson A, **Uwineza A**, et al. Refining the phenotypical and mutational spectrum of Taybi-Linder syndrome. Clin Genet. 2016.
15. Temtamy SA, **Ahmed FD**. Genetics and Genomic Medicine in Egypt: steady pace. MGGM 2017. (Accepted).
16. **Sirisena ND** and Bonham VL. Global Efforts Needed to Increase Ancestral Diversity Within Genomics Research: Sri Lanka. Special Issue of Journal of Community Genetics. 2017. (In preparation).

17. **Sirisena ND**, Anchala Kuruppu, Adebawale Adeyemo, et al. Genetic Determinants of Sporadic Breast Cancer in a Cohort of Sri Lankan Postmenopausal Women. 2017. (In preparation).
18. **Tibrewal S, Lallar M, Sirisena ND**. Chapter on Ocular Genetics in a book for Post-graduate students in India.

B) Grants:

1. **Adeoye AM**. Renewal Application for Stroke Investigative Research And Education Network (SIREN). Submitted to H3Africa. National Institutes of Health (NIH) and National Institute of Neurological Disorders and Stroke (NINDS) (Grant 1U54HG007479-01).
2. **Adeyemo WL** (PI) and Butali A (Co-PI). Investigating the Role of Genetics and Genomics of Non-syndromic clefts in Africa. (H3 Africa, U01).
3. **Ariani Y**. DIPI (Submitted to Indonesian Science Fund).
4. **Ariani Y**. PITA (Submitted to University of Indonesia).
5. **Ariani Y**. PUPT (Ministry of Research, Technology and Higher Education, Received). 2017-2018.
6. **Deniz E** (PI). Genome-Wide Screening With CRISPR/Cas9 and Modelling of Resistance Mechanisms Developed Against Cytotoxic Drugs in Cancer Treatment. (Submitted to Scientific and Research Council of Turkey (TUBITAK)).
7. **Ekure EN** (PI), **Uwineza A** (Co-PI), Muenke M et al. Genomic and Environmental Factors Influencing Congenital Heart Disease Risk in Africa. (Submitted to H3Africa, U01).
8. Mutesa L (PI), **Uwineza A** (Co-PI) et al. Transgenerational Epigenomics of Trauma and PTSD in Rwanda. (Submitted to H3Africa, U01).
9. **Muthukumarasamy P** (PI). The diagnostic value of chromosomal microarray in a developing nation. (To be submitted to University Of Malaya Special Research Fund Assistance).
10. **Muthukumarasamy P** (PI). The uptake of amniocentesis for prenatal diagnosis and its implications in Turkey and Malaysia. (To be submitted to University Of Malaya Special Research Fund Assistance).
11. **Tibrewal S** (PI) and Kumar V (Co-PI). Genetics of eye disorders in North India. (Submitted to INDO-US NIH Grant).

C) Collaborations (NIH and Other Institutions):

NIH:

1. **Adeoye AM**, Rotimi CN and Adeyemo AA. Pharmacogenomics of antihypertensive medication response in treatment naïve newly diagnosed hypertensive African Population. (University of Ibadan, University College Hospital Ibadan, Nigeria and NHGRI, NIH).
2. **Adeoye AM**, Rotimi CN and Adeyemo AA. Exploring association between lipids (or a panel of lipids-lowering variants) and stroke risk and types. The baseline analysis of blood sample from SIREN project will be done. (University of Ibadan, University College Hospital Ibadan, Nigeria and NHGRI, NIH).
3. **Adeyemo A**, Griffith A and Friedman TB. Genetics of Non-Syndromic Hearing Loss in Nigeria. (University College Hospital, Ibadan, Nigeria and NIDCD, NIH).

4. **Ahmed FD.** Atlas of Human Malformation syndromes in Diverse populations (Williams Syndrome, <https://research.nhgri.nih.gov/atlas/condition/>). (The National Research Center, Egypt and NHGRI, NIH).
5. **Ekure EN,** Muenke M, Adeyemo A, Kruszka P. Ongoing research on Genetic bases of Congenital heart disease in Africa. (University of Lagos, Lagos University Teaching Hospital Idi-Araba, Lagos, Nigeria and NHGRI, NIH).
6. **Ekure EN,** Muenke M, Adeyemo A, Kruszka P. Ongoing research on Genetic bases of acquired heart disease in Africa (Rheumatic heart disease and Endomyocardial fibrosis). (University of Lagos, Lagos University Teaching Hospital Idi-Araba, Lagos, Nigeria and NHGRI, NIH).
7. **Sirisena ND,** Kruszka P and Muenke M. Atlas of Human Malformation syndromes in Diverse populations (Down Syndrome, 22q11.2 Syndrome, Turner Syndrome, Noonan Syndrome, Williams Syndrome). (University of Sri Lanka and NHGRI, NIH).
8. **Sirisena ND,** Kruszka P and Muenke M. Diagnostic evaluation of patients with various craniosynostosis syndromes, congenital heart diseases and other congenital malformations. (University of Sri Lanka and NHGRI, NIH).
9. **Sirisena ND** and Bonnemann C. Diagnostic evaluation of patients with complex neuromuscular & neurogenetic conditions without a definite diagnosis. (University of Sri Lanka and NINDS, NIH).
10. **Sirisena ND,** Carsten B, Tennekoon G. Satellite Neurogenetics Symposium in Sri Lanka (Sept. 2017); Clinics (trials/therapies) for children with neuromuscular/neurodegenerative diseases. (University of Sri Lanka, NINDS, NIH and University of PA).
11. **Seven M** and Calzone K. A Global Nursing Alliance to Accelerate Integration of Genomics into Everyday Health Professional Practice. (Koç University School Of Nursing, İstanbul, Turkey and NCI, NIH).

Other Institutions:

12. **Adeyemo A** and Werely C. South-South Institutional partnership on Genetic research in Hearing Loss (University College Hospital, Ibadan, Nigeria and Stellenbosch University, South Africa).
13. **Malasa L** and Makani J. initiated dialogue for working on the Sickle Cell Programme, Tanzania. (Muhimbili National Hospital and Muhimbili University of Health and Allied Sciences, Tanzania).
14. Temel S, Celiker A, **Deniz E** et al. Personalized medicine and treatment approaches in hereditary arrhythmogenic and hypertrophic cardiomyopathy patients. (proposed; Near East University, NORTHERN CYPRUS, Medical Doctor, TURKEY, Acibadem University, TURKEY, Baskent University, TURKEY and Pamukkale University, TURKEY).

D) New Research Projects:

1. **Adeoye AM.** Exploring the Phenomics, Genomics and Environmental determinants of left ventricular mass among offspring of Hypertensive African Blacks: a Family screening study
2. **Adeoye AM.** Genetics of hypertension treatment response in drug naïve newly diagnosed hypertensives.
3. **Ahmed DF.** Predictive Cytogenetic Biomarkers for Non-disjunction disorders.

4. **Ariani Y.** FISH for 22q11.2 deletion syndrome.
5. **Ariani Y.** Molecular diagnosis for glycogen storage disease.
6. **Ariani Y.** Amino acid profile of stunted toddler.
7. **Ariani Y.** IgF1 gene polymorphism in stunted toddler.
8. **Ariani Y.** Molecular diagnosis for Mucopolysaccharidosis.
9. **Deniz E.** The use of induced pluripotent stem cell differentiated cardiomyocytes for gene therapy purpose at cardiac insufficiency caused by different origins: the investigation of the roles of new target molecules in this therapy by molecular and electrophysiological approaches.
10. **Deniz E.** Molecular Genetic and Functional Analysis of the Underlying Mechanisms of Primary Immune Deficiencies.
11. **Lallar M.** Functional Assessment of Neural Tube Defects, mRNA sequencing of amniotic fluid of NTD patients; Started a high-risk pregnancy clinic for mothers with genetic diseases; In the process of starting a metabolic clinic; Started support groups for patients with genetic disorders (Downs Syndrome).
12. **Uwineza A.** Developing a birth defect registry for Rwandan Teaching hospital in collaboration with CDC: Contact person: Jennifer William.
13. **Uwineza A.** Description of disorders of sex development(DSD) in children and adolescents in referral hospitals in Rwanda.
14. **Seven M** and Pasalak SI. Determination of the effect of genetic literacy on the utilization of prenatal screening test in pregnant women.

E) Other Initiatives:

1. **Adeoye AM.** Appointed as a Committee member for workshops in Human Genetics and Genomics at the 'Center for Genomic and Precision Medicine', College of Medicine, University of Ibadan, Nigeria. Aim is to interact with local and international scholars in genomics and precision medicine and; to include genomics in the curriculum for the medical and nursing students.
2. **Adeyemo A.** Appointed to coordinate workshop activities at University College Hospital, Ibadan, Nigeria to broaden the knowledge of the academic community on the different components of genetic research in Nigeria.
3. **Adeyemo WL.** Coordinated and lecture at a Bioinformatics workshop at the College of Medicine, University of Lagos, Nigeria.
4. **Ariani Y.** Speaker at International Conference and Exhibition, Indonesian Medical and Education Research Institute. Genetic evaluation of congenital malformation. 2016; Board member of Revitalization of Indonesian Society of Human Genetics; Weekly discussion on birth defect cases; Started a hospital birth defect registry; Developing web-based birth defect database; Developing rare disease integrated clinic in the teaching hospital; Negotiating national insurance coverage for genetic/metabolomics testing.
5. **Belhassan K.** Opportunity to be a guest researcher in Dr. Muenke's lab at NHGRI, NIH.
6. **Deniz E.** Introduction of a course (Spring Semester) "Molecular Genetics" for undergraduates, Dept. of Molecular Biology and Genetics, Acibadem University, Istanbul/Turkey.

7. **Eurke E.** Co-opted into curriculum review committee of the Faculty of Pediatrics, West African College of physicians. Responsible for reviewing genetics for Pediatric residency training; Lectured on 'Prenatal and Newborn Genetic screening', BRAINS Genomic and Bioinformatics workshop for faculty at College of Medicine, University of Lagos, Nigeria; Guest lecturer on 'Prenatal and newborn genetic screening in Nigeria How far?' at the Lagos University Medical Society 2016 Annual Scientific Conference, advocated for newborn screening; Incorporated Genetic counselling in practice.
8. **Lallar M.** Expanded newborn screening, based on US model to include nearby institutions. Plan to present a draft to the health secretaries for funding to include larger populations.
9. **Malassa L.** Educated Senior Faculty (Vice Chancellor, Dean) Pediatrics Department and Department of Biochemistry and Molecular Biology of Hubert Kairuki Memorial University and staff of Kairuki Hospital and Muhimbili National Hospital on importance of Genetics and Genomics in health care, the undiagnosed disease program and information on the electronic Atlas of Human Malformation syndromes in Diverse populations (Downs Syndrome).
10. **Muthukumarasamy P.** Implementation of Newborn screening (Blood spot) and Oxygen saturation monitoring as part of newborn screen; Genetic testing for rare disorders; Increasing awareness among health professionals of the availability of genetic tests and the use of the right test in a resource limited country, importance of pre- and post-test counselling; introduction to resources unlockinglifescode.org, OMIM, National Library of Medicine.
11. **Okafor FU.** Lectured 'Ingraining Genetics and Genomics into Reproductive Health Nursing Education In Nigeria', to undergraduates, post-graduates and faculty, Dept. of Nursing Science, University of Benin.
12. **Seven M.** Lectured on 'Genetic and Nursing' in pathophysiology courses for Master's Degree in nursing at Dept. of Nursing, Koç University School Of Nursing, İstanbul, Turkey.
13. **Uwineza A.** Outpatient consultation for patients with Genetic diseases in CHU (University Teaching Hospital of Kigali, Rwanda) average 30 patients per months; Teaching the course of Medical genetics in undergraduate in General Medicine.