2017 International Summit in Human Genetics and Genomics Report

The Second International Summit in Human Genetics and Genomics was hosted by NHGRI on the NIH campus, from August 31st –September 29th, 2017. It was the second of the 5- year (2016-2020) trans-NIH initiative to advance genetics and genomics in research and medicine in low to middle income countries (LMIC; defined by the World Bank). Based on last year's success, four additional ICs (NCATS, NCI, NIAID, NIDA) participated this year.

As mortalities from more common diseases and disorders decline, genetic disorders and congenital birth defects consume a disproportionate amount of resources allotted to health and medical care. Many LMICs lack expertise in genetics and genomics which impacts the provision of care related to genetic diseases and limits the training in this field. This takes a toll on the welfare of these nations. The International Summit is helping to alleviate these issues by filling the knowledge gap in human genetics and genomics.

With the generous support from Institutes at NIH (FIC, NCATS, NCI, NEI, NHLBI, NHGRI, NIAID, NIDA, NIDCD, NIDCR, NIMHD, NINR) and donations made to the Foundation for the NIH (FNIH) by Mayo Clinic and the March of Dimes, 26 candidates from 24 countries (refer to world map on reverse side) attended the 2017 Summit. Participants included research scientists (3), physicians (12) physician/scientists (5), dentist/scientist (3), a nurse and genetic counselor, and two PhD candidates, one of whom was a physician.

The curriculum included 12 days of didactics in genomic research and clinical genetics medicine, and 4 days of tailored training in the participants' areas of interest. There were 58 speakers delivering over 60 lectures on a variety of topics. In addition to didactics, the training included a hands-on bioinformatics workshop, a grant writing session, exposure to genetics clinics and patients, and 7 fieldtrips. The field trips included visits and lectures at academic and commercial institutions providing both clinical and laboratory services (Children's National Health System, GeneDx, Johns Hopkins University, NIH Clinical Center, NIH Intramural Sequencing Center, Kennedy Krieger Institute and MedStar Washington Hospital Center). These visits helped inform participants of the spectrum of experts involved in the provision of genetic services and testing. Based upon feedback, the patient panel was a favorite of the Summit. It included several patients that shared their insights, their struggles and continuing needs, because of the genetic diseases affecting them or their family members. One participant described the Summit as "life changing and career enhancing."

To measure outcomes, we assessed knowledge, interest and learning, daily, through pre- and post-surveys. The results indicated that the Summit was a unique learning opportunity for participants and speakers, and they strongly encouraged its continuance. All activities were rated as imparting knowledge and important to include in next year's summit.

Our one year outcomes from the 2016 Summit have been remarkable (ISHGG 2016_Annual Addendum). Participants have established collaborations (27) with investigators at NIH, other US-based academic institutions, and amongst themselves. Many have published articles in genetics/genomics related to their field of expertise (51) and written/received grants to/from NIH or other funding institutions (24). Our assessment of outcomes will continue for 5 years. Based on the survey feedback (2017) and the annual outcomes (2016) on the Summits, the Summit has trained 45 professionals from 37 countries, and is making great strides in achieving its goals. Due to its demand and success, we hope to expand and host up to 30 candidates in 2018. This however, will only be possible through partnership and support from our Institutes and Centers at NIH and the FNIH.

International Summit in Genetics and Genomics Outcomes
October 2016 – September 2017

A) Publications:

1. **Adeoye AM**, Ovbiagele B et al. SIREN Team as part of H3Africa Consortium. Exploring Overlaps Between the Genomic and Environmental Determinants of LVH and Stroke: A Multicenter Study in West Africa. Glob Heart. 2017 Mar 13; pii: S2211-8160 (17) 30001-7.
2. Akinyemi R, Arnett DK, Tiwari HK, Ovbiagele B, Sarfo F, Srinivasasainagendra V, Irvin MR, **Adeoye AM**, et al. SIREN Investigators. Interleukin-6 (IL-6) rs1800796 and cyclin dependent kinase inhibitor (CDKN2A/CDKN2B) rs2383207 are associated with ischemic stroke in indigenous West African Men. J Neurol Sci. 2017 Aug 15; 379:229-235.
3. Akinyemi R, Hemant K. Tiwari, Donna K. Arnett, Bruce Ovbiagele, Marguerite Ryan Irvin, Kolawole Wahab, Fred Sarfo, Vinodh Srinivasasainagendra, **Adeoye AM** et al. APOL1, CDKN2A/CDKN2B and HDAC9 polymorphisms and small vessel ischemic stroke. Acta neurologica Scandinavica. 2017 Sept 11; 1-9.
4. **Adeoye AM,** Ogah OS et al. SIREN Team as part of the H3Africa Consortium. Prevalence and Prognostic Features of ECG Abnormalities in Acute Stroke: Findings from the SIREN Study Among Africans. Glob Heart. 2017 Mar 13; pii: S2211-8160 (17) 30002-9.
5. **Adeoye AM**, Oladimeji Adebayo et al. Assessment of measures of adiposity that best correlate with blood pressure among hypertensive Africans. Ann Ib Postgrad Med. 2017 (In Press).
6. **Adeoye AM**, Yemi R. Raji et al. Circadian Blood Pressure variation among people with Chronic Kidney Diseases: A pilot Study in Ibadan Niger Postgrad. Med J. 2017 (In Press)
7. **Adeyemo WL** Genetics and Genomics Etiology of Non-Syndromic Orofacial Clefts. Invited Commentary, MGGM. 2017; 5: 3-7.
8. **Ariani Y**, Soeharso P et al. Genetic and genomic medicine in Indonesia. MGGM. 2017; 5:103-109.
9. **Ariani Y**, Soeharso P et al. Loss of heterozygosity in children with multiple congenital anomaly. 2017 (Manuscript in preparation).
10. **Ariani Y**, Priambodo R et al. Ancestral diversity; a global challenge to future disease understanding. 2017 (In Review).
11. **Belhassan K**, Ouldim K, Sefiani AA. Genetics and genomic medicine in Morocco: the present hope can make the future bright. MGGM. 2016; 4:588-598.
12. Bouchikhi EI, **Belhassan K**, et al. Novel NKX25 germline mutation in a Moroccan child with transitional atrioventricular septal defect (tAVSD). 2017 (Accepted for publication at the Turkish journal of pediatrics).
13. Bouchikhi EI, Bouguenouch L, Moufid FZ, Houssaini MI, **Belhassan K** et al. NKX2-5 molecular screening and assessment of variant rate and risk factors of secundum atrial septal defect in a Moroccan population. Anatol J Cardiol 2017; 17: 217-23.
14. Bouchikhi EI, **Belhassan K**, et al. GATA4 molecular screening and assessment of environmental risk factors in a Moroccan cohort with tetralogy of Fallot. 2017. (Submitted, Balkan Medical Journal. Manuscript ID is BalkMedJ20170402).
15. Céspedes-Garro C, Naranjo M-E G, Rodrigues-Soares F, LLerena A, Duconge J, Montané-Jaime LK, **Roblejo H**, et al. Pharmacogenetic research activity in Central America and the Caribbean: a systematic review. Pharmacogenomics. 2016 Oct. 17; (15):1707-1724.
16. **Ekure EN**, Fidelia Bode-Thomas et al. Congenital Heart Defects in Nigerian Children: Preliminary data from the National Pediatric Registry. WJPCHS 2017. (In press).
17. Feoktistova YF, Domech CR, Bacallao EC, **Roblejo Balbuena H**, Jiménez ZR, Peralta EM. Análisis de los polimorfismos p. K832R y p. T991T en pacientes cubanos con diagnóstico clínico de la enfermedad de Wilson. Revista Habanera de Ciencias Médicas. 2017; 16 (2).
18. Giri V, Srivastava P, **Lallar M**, Phadke SR. Genetic Clinics. 2017; 10: 9-17.

19. Gowans LJL, 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; 5: 164-171.
20. **Guio H**. Genetics in Peru: Before and after Mendel. 2017. (Manuscript in preparation).
21. **Guio H**, Galarza M, Pellon O, Gomez H, Olivera M, Jaramillo L, Vigil C. Circulating exosomal microRNAs as biomarkers for non-small-cell lung cancer in a high TB burden setting. 2017. (Manuscript in preparation).
22. Harris DN, Ruczinski I, Yanek LR, Becker LC, Becker D, **Guio H**, et al. Evolution of Hominin Polyunsaturated Fatty Acid Metabolism: From Africa to the New World. BMC Biology. 2017. (Accepted).
23. Harris DN, Song W, Shetty AC, Levano K, Cáceres O, Padilla C, Borda V, Santos ET, O'Connor TD, **Guio H**. The Evolutionary Genomic Dynamics of Peruvians Before, During, and After the Inca Empire. 2017. (In peer review).
24. **Hussen DF**, Marwa Shehab, Mona Mekkawy, et al. Diagnostic value of Cytogenetic Biomarkers in Cohesion defective disorders. Middle East Journal of Medical Genetics. (Accepted, August 2017).
25. Ismail S, Essawi M, Sedky N, Hassan H, Fayez A, Helmy N, Shehab M, **Farouk D**, et al. Roberts syndrome: Clinical and cytogenetic studies in 8 Egyptian patients and molecular studies in 4 patients with genotype /phenotype correlation. Genetic Counseling. 2016; 27 (3): 305-323.
26. 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.
27. Kruszka P, Addissie YA, McGinn DE, Porras AR, Biggs E, Share M, **Uwineza A**, Crowley TB, Chung BHY, Mok GTK, **Muthukumarasamy P**, ThongM-K, **Sirisena ND**, Dissanayake VHW, Paththinige CS, Prabodha LBL, Mishra R, Shotelersuk V, **Ekure EN**, Sokunbi OJ, Kalu N, Jones KL, Kaplan JD, Abdul-Rahman OA, Vincent L, Love A, **Belhassan K**, Ouldim K, Bouchikhi IE, Shukla A, Girisha KM, Pati SJ, **Sirisena ND**, et al. 22q11.2 Deletion Syndrome in Diverse Populations. Am J Med Genet Part A. 2017; 173 (4):879-888.
28. Kruszka P, Porras AR, Addissie YA, Moresco A, Medrano S, Mok GTK, Leung GKC, Tekendo-Ngongang C, **Uwineza A**, Thong M-K, **Muthukumarasamy P**, Honey E, **Ekure EN**, Jones KL, Kaplan JD, Abdul-Rahman OA, Vincent L, Love A, **Belhassan K,** Ouldim K, Bouchikhi IE, Shukla A, Girisha KM, Patil SJ, **Sirisena ND** et al. Noonan Syndrome in Diverse Populations. Am J Med Genet A. 2017; 173 (9):2323-2334. Cover image by **Muthukumarasamy P.**
29. **Lallar M**, Srivastava A, Phadke SR. Hyperekplexia: A forgotten diagnosis clinched by next-generation sequencing. Neurol India, 2017; 65 (5): 1065-1067.
30. **Lallar M**, Phadke SR. MECP2 gene related disorders. Genetic Clinics. 2017; 10: 5-9.
31. Maddirevula S, AlZahrani F, Anazi S, Almureikhi M, Ben-Omran T, Abdel-Salam GMH, Hashem M, Ibrahim N, Abdulwahab FM, Meriki N, Bashiri FA, Thong M-K, **Muthukumarasamy P** et al. GWAS signals revisited using human knockouts. Genet. Med. June 2017. DOI: 10.1038/gim.2017.78.
32. **Malasa L**. A case report on Schizencephally. (In preparation).
33. Moufid FZ, Bouguenouch L, Elbouchikhi I, Houssaini MI, **Belhassan K** et al. Comparative effectiveness of universal molecular screening versus revised Bethesda guidelines to identify Lynch syndrome among unselected patients with colo-rectal cancer. 2017. (Submitted to PLOS ONE Journal. 2017. PONED1701394).
34. Ngim CF, Lai NM, Hong JY, Tan SL, Ramadas A, Muthukumarasamy P, Thong MK. Growth hormone therapy for people with thalassaemia (protocol). Cochrane database of systematic reviews (Online) 2016 (7). DOI: 10.1002/14651858.CD012284.
35. **Okafor FU**. Prevalence and Projection Of Retinoblastoma In Tertiary Health Institution In Edo State, Nigeria. NJEHETR. 2017. (Accepted).

36. Paththinige CS, **Sirisena ND**, et al. A child with multiple congenital anomalies due to partial trisomy 7q22.1→qter resulting from a maternally inherited balanced translocation: A case report and review of literature. 2017. BMC Medical Genetics (Submitted).
37. 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.
38. **Roblejo Balbuena H**, Marcheco Teruel B. Genetics and genomic medicine in Cuba. MGGM. 2017; 5 (3):196-201.
39. Rokickia D, Pajdowskab M, Trubickac J, Thong M-K, Ciara E, Piekutowska-Abramczuk D, Pronicki M, Sikora R, Haidar R, Ołtarzewski M, Jabłońska E, **Muthukumarasamy P** et al. 3-Methylglutaconic aciduria, a frequent but underrecognized finding in carbamoyl phosphate synthetase I deficiency. Clin Chim Acta. 2017; 471:95-100. DOI: 10.1016/j.cca.2017.05.023.
40. **Seven M**, Pasalak SI, Guvenc G, Kok G. The Knowledge Level and Educational Needs of Turkish Oncology Nurses Regarding the Genetics of Hereditary Breast and Ovarian Cancers. J Contin Educ Nurs. 2017. (Accepted).
41. **Sirisena ND, Dissanayake V.H.W.** Focusing attention on ancestral diversity within genomics research: a potential means for promoting equity in the provision of genomics based healthcare services in developing countries. J Community Genet. July 2017; DOI 10.1007/s12687-017-0311-y.
42. **Sirisena ND** and Bonham VL. Global Efforts Needed to Increase Ancestral Diversity Within Genomics Research: Sri Lanka. Special Issue of J of Community Genet. 2017. (In preparation).
43. **Sirisena ND**, Anchala Kuruppu, Adebowale Adeyemo, et al. Genetic Determinants of Sporadic Breast Cancer in a Cohort of Sri Lankan Postmenopausal Women. BMC Cancer. 2017. (In peer review).
44. Sokunbi OJ, **Ekure EN**, et al. Pulmonary Hypertension among 5 to 18-Year-Old Children with Sickle Cell Anemia in Nigeria. PLOS ONE. 2017. (In press).
45. Temtamy SA, **Ahmed DF.** Genetics and Genomic Medicine in Egypt: steady pace. MGGM 2017; 5(1):8-14.
46. **Tibrewal S, Lallar M, Sirisena ND.** Chapter on Ocular Genetics in Dutta's Textbook of Ophthalmology, for Post-graduate students in India. (Book in Press).
47. Tora A, **Tadele G**, et al. Health beliefs of school-age rural children in podoconiosis affected families: a qualitative study. Southern Ethiopia Journal of PLOS NTD. 2017; https://doi.org/10.1371/journal.pntd.0005564.
48. Ayode D, Tora A, Farrell D, **Tadele G** et al. Association between causal beliefs and shoe wearing to prevent podoconiosis: a baseline study. Am J Trop Med Hyg. 2016; 94 (5):1123–1128, doi:10.4269/ajtmh.15-0342
49. Tora A, Ayode D, **Tadele G**, et al. Interpretations of education about gene-environment influences on health in rural Ethiopia: The context of a neglected tropical disease. Journal of International Health. 2016; doi:10.1093/inthealth/ihw016.
50. Ayode D, Tora A, Farrell D, **Tadele G**, et al. Dual Perspectives on Stigma: Reports of Experienced and Enacted Stigma by Those Affected and Unaffected by Podoconiosis. Journal of Public Health Research. 2016; 5:689
51. Zühlke L, Karthikeyan G, Engel ME, Rangarajan S, Mackie P, Cupido-Katya Mauff B, Islam S, Daniels R, Francis V, Ogendo S, Gitura B, Mondo C, Okello E, Lwabi P, Al-Kebsi MM, Hugo-Hamman C, Sheta SS, Haileamlak A, Daniel W, Goshu DY, Abdissa SG, Desta AG, Shasho BA, Begna DM, ElSayed A, Ibrahim AS, Musuku J, Bode-Thomas F, Yilgwan CC, Amusa GA, Ige O, Okeahialam B, Sutton C, Misra R, Abul Fadl A, Kennedy N, Damasceno A, Sani MU, Ogah OS, Elhassan TO, Mocumbi AO, **Adeoye AM** et al. Clinical Outcomes in 3343 Children and Adults With Rheumatic Heart Disease From 14 Low- and Middle-Income Countries: Two-Year Follow-Up of the Global Rheumatic Heart Disease Registry (the REMEDY Study). Circulation. 2016 Nov 8;134 (19):1456-1466.

B) Grants:

1. **Adeoye AM** (PI), Owolabi M (Multiple PI). Left ventricular hypertrophy in AfRicans and hypertension GEnomics (LARGE). (NIH R21, PAR-16-052, Submitted).
2. **Adeyemo A** (PI). RCT on the action of Dexamethasone in preventing ototoxicity in cancer patients receiving Cisplatin. Part of the project will explore the genetic mutations predisposing to Cisplatin ototoxicity. Local grant from the University of Ibadan, University College Hospital Ibadan, Nigeria (Received).
3. **Adeyemo A** (PI), **Uwineza A**. Genetic Variation of Heritable Deafness in Newborns (hedef): and African perspective. University of Ibadan. (Submitted to H3Africa).
4. Nwaorgu OGB (PI), Fasunla AJ, Onakoya PA, **Adeyemo A**, et al. Hearing Health Care for Adults: Improving Access and Affordability (NIH R01, FOA number: PA-17-202. Part of the proposal includes genetic studies on deafness. (University of Ibadan, University College Hospital Ibadan, Nigeria, and other centers- Nigeria) (Submitted).
5. **Ariani Y**. Enzyme assay for detecting I2S enzyme activities of MPS 2 patients. (Submitted to the Indonesian Medical Education and Research Center, IMER, Accepted).
6. **Butali A (PI), Adeyemo WL**. Whole Genome Sequencing (WGS) of African and Asian Orofacial Clefts Case-Parent Triads. (NIH funded, Received). It is the first NIH funded WGS for any chronic disease or trait in Africa, including oral clefts. Kids First X01 HL140516-01.
7. **Deniz E** (PI), Keskin ZT et al. Genome-Wide Screening With CRISPR/Cas9 and Modelling of Resistance Mechanims Developed Against Cytotoxic Drugs in Cancer Treatment. (Acibadem University and Sabanci University, Turkey). (Scientific and Research Council of Turkey (TUBITAK), Received).
8. Inan EA, Can A, Keskin ZT, **Deniz E** et al. The potential role and mechanisms of action of brain natriuretic peptide as a diabetes preventive target. (Akbara University, Ankara University, Acibadem University, Kemerburgaz University, Turkey). (Submitted to Scientific and Research Council of Turkey (TUBITAK)).
9. **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).
10. **Guio H** (PI), Levano K. To develop a new kit to extract DNA using magnetic pearls which need minimal facilities to promote molecular biology research in remote areas (INBIOMEDIC). (INNOVATE, Received).
11. **Guio H** (PI), Levano K. To establish a program called "Sowing Science" to promote research at two levels (a) High school: through cartoons and communication (Skype) with Peruvian researchers who live abroad and who have made important contributions to science. This is to help break myths and mental barriers in entrepreneurship. (b) University: By developing videos for writing protocols, research papers, genetics and genomics, molecular biology etc. (INBIOMEDIC). (INNOVATE, Received).
12. Hou, L, Sagay A, Ogunsola FT, Rob M, **Adeyemo WL** (Co-investigator). Epigenomic Biomarkers of HIV-Associated Cancers in Nigeria. (NIH U54, Received).
13. Mutesa L (PI), **Uwineza A** (Co-PI), Uddin M (PI) et al. Transgenerational Epigenomics of Trauma and PTSD in Rwanda. (Carl R. Woese Institute for Genomic Biology (IGB), University of Illinois at Urbana-Champaign). (Preliminary acceptance by H3Africa. Pending final decision).
14. **Muthukumarasamy P** (PI). The diagnostic value of chromosomal microarray in a developing nation. (To be submitted to University Of Malaya Special Research Fund Assistance).
15. **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).
16. Owolabi M (PI), **Adeoye AM** (PI). Genomic Epidemiology to Neutralize hEmorrhagic sTroke In Continental Africans (GENETIC). Wellcome Trust grant (Alliance for Accelerating Excellence in Science in Africa-AESA, Submitted).
17. Owolabi M (PI), **Adeoye AM**. Precision African non-Communicable diseases center of Excellence (PACE) (African Research University Alliance-ARUA, Submitted).
18. Sjarif DR (PI), **Ariani Y** (Co-investigator). Genetic study on IDS gene of MPS 2 Indonesia patients. (Submitted to PITA, University of Indonesia, Accepted).

19. Sjarif DR (PI), **Ariani Y** (Co-investigator). Genetic study on PTS gene of PTPS type PKU. (Submitted to PITA, University of Indonesia, Accepted).
20. Sjarif DR (PI), **Ariani Y** (Co-investigator). Developing diagnostic method for unknown multiple congenital anomalies. (PUPT, Ministry of Research, Technology and Higher Education, Received for 2017).
21. Sjarif DR (PI), **Ariani Y** (Co-investigator). FISH method development for 22q11.2 deletion syndrome. (Submitted to the Indonesian Medical Education and Research Center, IMER, Accepted).
22. Sjarif DR (PI), **Ariani Y** (Co-investigator). Developing GAGs (Heparan sulfate and Dermatan sulfate) detection to do MPS2 screening. (Submitted to PUPT, Ministry of Research, Technology and Higher Education, 2018).
23. **Uwineza A** (PI). Genetic etiology of epilepsy in Rwandan patients with intellectual disability'. Funded by University of Rwanda for Post-doctoral grant and SIDA-SAREC. (Received). Data collection and preliminary analysis underway.
24. **Uwineza A**. The "TWAS Equipment Grant to Support Research Projects in Rwanda" was sponsored by TWAS 2015 Agricultural Sciences Prize winner Prof. Feng-Min Li (Institute of Arid AgroEcology, School of Life Sciences, Lanzhou University, China). (Received).

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. Genetic mutations responsible for deafness in Nigerians. (University College Hospital, Ibadan, Nigeria and NIDCD, NIH).
4. **Adeyemo A** and Prokunina L. Characterization of HPV genotypes in head and neck cancer patients in Nigeria. (University College Hospital, Ibadan, Nigeria and NCI, NIH).
5. **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).
6. **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).
7. **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).
8. **Malasa L** and Makani J. In conversation about collaborating on the Sickle Cell Program in Tanzania.
9. **Roblejo Balbuena**, Lierena A. Working on a PhD in Pharmacogenetics and Schizophrenia. (Univeristy of Havana, Cuba and CICAB Clinical Research Center at Extremaduar University Hospital and Medical School, Spain).
10. **Sirisena ND**, Kruszka P and Muenke M. Atlas of Human Malformation syndromes in Diverse populations (Submitted photos for Down Syndrome, 22q11.2 Syndrome, Noonan Syndrome, Williams Syndrome, Cornelia de Lange syndrome and Turner syndrome). (University of Sri Lanka and NHGRI, NIH).
11. **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).
12. **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).

13. **Sirisena ND** and Sharan S. Functional assays on five variants of uncertain significance in the BRCA2 gene identified in some of the Sri Lankan patients with hereditary breast cancer. (University of Sri Lanka and NCI/NIH).
14. **Sirisena ND** and Adeyemo A. PLINK analysis of genotype data on sporadic breast cancer in a cohort of Sri Lankan postmenopausal women. (University of Sri Lanka and NHGRI/NIH).
15. **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).
16. **Uwineza A.** Kruzska P, Muenke M. et al. Targeted gene sequencing of Rwanda patient with Noonan syndrome. (Center of Human Genetic of Rwanda / School of Medicine and Pharmacy / College of Medicine and Health Sciences/ University of Rwanda and NHGRI/NIH).

Other Institutions:

17. **Adeoye AM,** Bamidele Tayo et al. Cardiovascular And Renal Events In People With Chronic Kidney Disease. (University of Ibadan, University of Arizona College of Medicine, Tucson, AZ, Loyola University Chicago Stritch School of Medicine, Maywood, IL).
18. **Adeoye AM**, Karaye KK, Mark Loeb et al. A Randomized Controlled Trial of Influenza Vaccine to Prevent Adverse Vascular Events (IVVETrial). (University of Ibadan, Bayero University,Kano,and McMaster University, Canada).
19. **Adeoye AM**, Bongani Mayosi and Stuart J. Connolly. INVestIgation of rheumatiC AFTreatment Using vitamin K antagonists, rivaroxaban or aspirin Studies (INVITUS trial). (University of Ibadan, University of Cape Town, and Population Health Research Institute, Canada).
20. **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).
21. **Guio H** et al. Signed an Agreement with EUROESPES to develop pharmacogenomics.
22. **Guio H** et al. Signed an Agreement with GENYCA to develop Nutrigenomics.
23. **Guio H** et al. Signed an Agreement with SOPHIAGENETICS to test BRCA1/2.
24. **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).
25. Mulder N, Wonkam A and **Uwineza A**. Review of the of Sickle cell Disease and ontology. (University of Cape Town, H3Bionet and Center of Human Genetic of Rwanda / School of Medicine and Pharmacy / College of Medicine and Health Sciences/ University of Rwanda).
26. **Okafor FU** and Abad PJ. Study on the Nurses' knowledge, attitude and belief on genetics and genomics in Nigeria and Philippines. (University of Benin, Nigeria and University of Philippines, Manilla).
27. 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. **Adeoye AM.** Maternal Cardiovascular Risk Assessment And Fetal Outcome (MARCAF study): Community Based study
4. **Adeoye AM.** Carotid Intima Media Thickness and Lipid Markers of Atherosclerosis among Hypertensives.
5. **Ahmed DF**. Predictive Cytogenetic Biomarkers for Non-disjunction disorders.
6. **Ahmed DF** (collaborator). Nicotine dependence as an environmental health problem, the efficacy of different approaches for its management. 3-year project.
7. **Ahmed DF** (collaborator). Using of microarray technique in diagnosis of agenesis of corpus callosum.

8. **Ahmed DF**. Preclinical diagnosis of Alzheimer's disease. Project has been accepted by the Egyptian Academy of Scientific Research and Technology (July 2017). Pending evaluation by the International Centre for Genetic Engineering and Biotechnology (ICGEB) in Italy.
9. **Ariani Y**. FISH fpr 22q11.2 deletion syndrome.
10. **Ariani Y**. Molecular diagnosis for glycogen storage disease. Waiting for budget from faculty.
11. **Ariani Y**. Amino acid profile of stunted toddler; IgF1 gene polymorphism in stunted toddler.
12. **Ariani Y**. Molecular diagnosis for Muccopolysaccharidosis type 2. Looking for a collaborator to test the new variant of IDS gene in exon 9.
13. **Ariani Y**. Developing GAGs (heparan and dermatan sulfate) assay from urine for early detection and evaluation of MPS 2; Developing enzyme assay for I2S activity for MPS 2.
14. **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. Research Group- Assistant Researcher.
15. **Deniz E**. Molecular Genetic and Functional Analysis of the Underlying Mechanisms of Primary Immune Deficiencies. Research Group- Advisor.
16. **Deniz E**. Genetic variants in glioblastomas.
17. **Deniz E**. Identifying the roles of long noncoding RNA's (lncRNA) on cancer development.
18. **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).
19. **Uwineza A.** Developed a birth defect registry for Rwandan Teaching hospital. Developed and completed a "core outcome set" for a congenital abnormalities surveillance program using Delphi techniques. Results will be published.
20. **Uwineza A.** Description of disorders of sex development (DSD) in children and adolescents in referral hospitals in Rwanda.
21. **Seven M** and Pasalak SI. Determination of the effect of genetic literacy on the utilization of prenatal screening test in pregnant women. Study has been approved and data is being collected.

E) Other Initiatives/Accomplishments/Presentations:

1. **Adeoye AM**. Established the 'Center for Genomic and Precision Medicine' at the College of Medicine, University of Ibadan, Nigeria with a mandate to regularly organize workshop in Human Genetics and Genomics. The 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; Organized a one-day workshop/symposium on application of precision medicine with the theme: 'REVOLUTIONIZING DIABETES AND STROKE CARE IN AFRICA WITH PRECISION MEDICINE' (July 18th, 2017). Another has been scheduled for later in the year.
2. **Adeyemo A**. Was invited to join the Center for Genomic and Precision Medicine, due to Summit selection; Nominated to head the workshop committee of the Center, to train the academic community in genomics and precision medicine; First successful workshop in "Revolutionizing Diabetes and Stroke Care in Africa with Precision Medicine" was held in July 2017, that included local and international speakers (Professor Louis Philipson, Director, Kovler Diabetes Center, University of Chicago).
3. **Adeyemo WL**. Coordinated and lectured at a Bioinformatics workshop at the College of Medicine, University of Lagos, Nigeria; Obtained a Certificate in Genetics and Genomics from Stanford University (April 2017); Won the Best Prize in Oral Presentations for Genetics of Third Molar Impactions at the Faculty of Dental Sciences, College of Medicine University of Lagos (July 2017); Promoted to a full Professor; Delivered the Inaugural Lecture as a Professor on "Genome Editing, Surgical Editing: A Surgeon-Scientist Narrative of Orofacial Cleft Research and Care", at the University of Lagos (October 2017).

4. **Ahmed DF**. Submitted an abstract for oral presentation at the10th conference of the African Society of Human Genetics, Egypt; Nov. 2017; Gave a lecture at The National Research Center on the ISHGG and benefits of collaboration (Oct. 2016); Oral presentation in The Human Genetics & Genome Research Conference under the theme "Path to the Future of Human Genetics" (Cairo, Egypt, Nov. 2016); Lectured in a course on Basic Genetics under the theme "Human Genome: Actions and Interactions" at The National Research Centre-Egypt (Feb, 2017).
5. **Ariani Y**. Weekly discussion (Skype) on birth defect cases with other centers in Indonesia; Started a hospital birth defect registry; Developed a "Rare disease team of excellence" at Cipto Mangunkusumo Hospital, which is now a national referral hospital; Leading an advocacy efforts to educate the Dean and Directors of the hospital for negotiating national insurance coverage for genetic/metabolomics testing; Speaker at the International Conference and Exhibition, Indonesian Medical and Education Research Institute on 'Genetic evaluation of congenital malformation' (2016); Board Member of Revitalization of Indonesian Society of Human Genetics; Member of Newborn screening working group of Indonesian Pediatric society; Attended the 10th International Society of Neonatal Screening, Asia Pacific Regional Meeting (Mongolia, Aug. 2017) to revitalize the existing work and optimize goals in increasing the coverage of newborn screening in Indonesia, in collaboration with the Indonesian Ministry of Health and private hospitals; Presented a poster 'Genetic abnormality of two children presenting with failure to thrive' at the Indonesian Pediatric Congress 2017 (Indonesia, Aug. 2017); Presented 'A novel variant at exon 9 of IDS gene of Indonesian MPS 2 patient at the 13th International Congress of Inborn Errors of Metabolism, (Brazil, September 2017).
6. **Belhassan K**. Opportunity to be a guest researcher in Dr. Muenke's lab at NHGRI, NIH; Will graduate as a Medical Geneticist (Oct. 2017) from Fes Faculty of Medicine and Pharmacy, University Sidi Mohammed Ben Abdelah, Morocco; Applying for the Laboratory Genetics and Genomics Fellowship in the USA, including at NIH.
7. **Deniz E.** Introduction of a course (Spring Semester) "Molecular Genetics' for undergraduates, Dept. of Molecular Biology and Genetics, Acibadem University, Istanbul/Turkey;
8. **Eurke E**. Co-opted into curriculum review committee of the Faculty of Pediatrics, West African College of physicians. Responsible for reviewing genetics for Pediatirc residency training (Oct. 2016); Lectured on 'Prenatal and Newborn Genetic screening', BRAINS Genomic and Bioinformatics workshop for faculty at College of Medicine, University of Lagos, Nigeria (Dec 6th, 2016; Guest lecturer on 'Prenatal and newborn genetic screening in Nigeria How far?'(Dec. 7th, 2016) at the Lagos University Medical Society 2016 Annual Scientific Conference, advocated for newborn screening; Echocardiographic Screening of 3901 Nigerian School Children for Rheumatic Heart Disease- Preliminary Report. Presented at the Rheumatic Heart Disease from Molecules to the Global Community Meeting organized by Magdi Yacoub Heart Foundation & PASCAR in Cairo, Egypt (Jan. 2017); Congenital Heart Defects in Nigerian Children: Preliminary data from the National Pediatric Registry. Presented at the 7th World Congress of Pediatric Cardiology & Cardiac Surgery in Barcelona, Spain (July, 2017); To deliver a presentation titled "Addressing the Burden of Congenital Heart Diseases in Nigeria" at a plenary in the Nigerian Cardiac Society Annual General and Scientific Conference in Benin, Nigeria (Sept. 2017); Incorporated Genetic counselling in practice. Families with 22q11.2 deletion syndrome, hypertrophic cardiomyopathy, Down syndrome, Turner syndrome, Noonan syndrome, and Alagille syndrome are among those being counselled; Promoted to a Full Professor of Pediatrics at the University of Lagos, Nigeria (backdated to Feb. 2017).
9. **Guio H**. Presenting at the ASHG Conference (Oct. 2017) on the Peruvian Genome Project: A new reference of Andean haplotypes to study genome populations; Exploring opportunities for establishing a Latin American Consortium to improve and increase data related to genetic variations in Latino America Countries; Developing opportunities for healthcare workers in personalized medicine.
10. **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; Demonstrator in the 16th ICMR course of Medical Genetics and Genetics Counselling (July, 2017) attended by clinicians from all over India; Demonstrator in "Medical Genetics- pedigree to Genome" (01 course, Mar. 2017) attended by senior residents from various super specialities of SGPGI; Lectured on 'Genome Test: Ethical & Psychological

Issues' at the " CME on diagnosis of Genetic Disorders in NGS era and Next Generation Sequencing workshop" organised by the department of Medical Genetics, IGBI New Delhi and Centogene, India Pvt Ltd (Aug. 2017).

11. **Malassa L**. Educated Senior Faculty (Vice Chancellor, Dean, Directors, Administartors) Department of Pediatrics and Child Health, and Department of Biochemistry and Molecular Biology of Hubert Kairuki Memorial University on importance of collaborations and opportunities available at NIH, including training programs, grants and fellowships, emphasized the role of Genetics and Genomics in health care, especially in low resource settings, and provided information on the undiagnosed disease program and the weblink to the Genetic and Rare Diseases Information Center. Many healthcare professionals have found this information useful; Provided information to the Department of Pediatrics and to Kairuki Hospital on the procedures to send photos for publication in the electronic Atlas of Human Malformation syndromes in Diverse populations (Downs Syndrome).
12. **Muthukumarasamy P**. Implementation and improved uptake of Newborn screening (Blood spot) at University Malaya Medical Centre, Kuala Lumpur due to continued education of pregnant mothers, both antenatal and postnatal; Inclusion of a clinical specialist nurse in metabolic medicine to better facilitate newborn screening; Continued education of the unit's genetic counselor to expedite and improve counseling services within the unit; Lectures and presentations on upcoming genetic services, online resources (unlockinglifescode.org, OMIM, National Library of Medicine) and newer genetic testing and gene therapy to nurses, undergraduate and postgraduate doctors and professors; Elected as Trust Fellow in Pediatrics (ST4) at Great Ormond Street Hospital, London via the Medical Training Initiative (MTI) scheme (Oct. 2017) and to continue a subspecialty in Genetics.
13. **Okafor FU**. Introduced the concept of genetic pedigree to students of Department of Nursing Science program, following sensitization with the academic staff; Lectured on genetics and genomics during the Nigerian University Nursing Students Association (NUNSA, University of Benin branch) week. It was well attended by students and College of Medical Science staff; As doctoral degree candidate developed the Ph.D. seminar 'Incorporating Genetics and Genomics into Reproductive Health Nursing Education and Practice in Nigeria' for presentation to the staff of College of Medical Sciences, Centre of Excellence of Reproductive Health Innovation [CERHI] and the general public; After which the concept of genetics and genomics will be introduced to the Educational Committee of Nursing and Midwifery Council of Nigeria; Community Vanguard has been established, to track patients having suspected cases of retinoblastoma in the immediate community.
14. **Roblejo Balbuena H**. Given lectures in advances in Medical Genetics across Cuba; Will be teaching a pre-congress, genetics technologies course at the International Congress of Community Genetics (Nov. 14-17, 2017) in Havana, Cuba; Promoted to Auxillary Professor and Auxillary Researcher.
15. **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; Invited to be a supervisor for a Ph.D. dissertation on counselling for prenatal genetic screening and diagnosis at Ankara University, Turkey; Invited to be on the organizing committee (Turkey Representative) for The International Society of Nurses in Genetics Congress, USA (Nov. 2017).
16. **Sirisena ND**. Obtained membership for Sri Lanka in the Undiagnosed Diseases Network International (UDNI, June 2017). Prof. Vajira H.W. Dissanayake will represent Sri Lanka at the 5th UDNI Conference (Stockholm Aug. 2017). Meeting will serve as a forum to initiate research collaborations on rare undiagnosed diseases within the network; Won the Korean Breast Cancer Foundation Scholarship award for outstanding oral presentation at the Global Breast Cancer Conference (South Korea, April 2017) for the paper 'Genetic determinants of sporadic breast cancer in a cohort of Sri Lankan postmenopausal women: Sirisena ND, Kuruppu A, Adeyemo A et al'; Will present 'Genetic variants associated with risk and clinico-pathological profile of sporadic breast cancer in Sri Lankan women'. at The Landscape of Genetic Variants in Asian Founder Populations – from Near to Far East, 2nd International Conference on Founder Populations (India Nov. 2017).

17. **Tibrewal S.** Expanded genetics clinic at Shroff Charity Eye Hospital (SCEH); Appointed a genetics counselor at SCEH; Attended the Asian Eye Genetics Consortium at Aditya Jyot Hospital, Mumbai (Nov. 2016).
18. **Tadele G.** Promoted to a Full professor (Jan. 2017).
19. **Uwineza A.** Outpatient consultation for patients with Genetic diseases in CHU (University Teaching Hospital of Kigali, Rwanda and Rwanda Medical Hospital); Launched the Rwanda Association of Down syndrome (March, 2017); Teaching the course of Medical genetics in undergraduate in General Medicine; Teaching the Module of Biochemistry and Molecular Biology in General Medicine and Pharmacy Year I;; Attended the 2nd Sickle Cell Disease Ontology Workshop in Cape Town, South Africa (29 May – 02 June 2017). The main objective of the workshop was to finalize and release the SCDO.