ADDENDUM

ISHGG_2017 ANNUAL OUTCOME (October 2017- September 2018)

A. Publications: (Total- 147; 2017- 76; 2016- 71)

2017 Batch

1. **Abad PJ**, Lee JMH, Kejriwal S, Chu YWY, et al. Is a Genetic Counselor a Value-Added Healthcare Team Member? Survey Results in Six Asian Countries. J. Genet Couns. 2018. (In review).
2. **Abad PJB**, Laurino MY, Daack-Hirsch S, Abad L, et al. Parent Child Communication about Congenital Adrenal Hyperplasia: Filipino Mothers' Experience. Acta Medica Philippina. 2017; 51(3):175-180.
3. **Abad PJB**, Laurino MY. Preconception Genetic Counseling in a Filipino Couple with a Previous Child with Trisomy 18. Acta Medica Philippina. 2017; 51(3):248-250.
4. Abdel-Hamid MS, Issa MY, **Otaify GA**, et al. PGAP3-related hyperphosphatasia with mental retardation syndrome: Report of 10 new patients and a homozygous founder mutation. Clin. Genet. 2018; 93: 84-91.
5. Abdel-Salam GMH, Abdel-Hamid MS, **Mehrez MI** et al. Further delineation of the oculoauricular syndrome phenotype: A new family with a novel truncating HMX1 mutation. Ophthalmic Genetics. 2018; 39 (2): 215-220.
6. Adhikari S, **Thakur N,** Shrestha U, Shrestha MK, et al. Genetic Analysis of Children with congenital anomalies in three ecological regions of Nepal: A phase II Nepal Pediatric Ocular Diseases Study. Ann Hum Genet. 2018. (Submitted, AHG-MS-18-0177).
7. **Avogbe PH**, Delhomme TM, Gabriel A, et al. Assessment of the diagnostic value of circulating *RB1* and *TP53* mutations for early detection of small cell lung cancers: discovery and validation in two independents cohorts (In preparation).
8. **Avogbe PH,** Manel A, Vian E et al. Urinary and blood circulating *TERT* promoter mutations as non-invasive biomarkers for the detection of primary and recurrent urothelial cancer. 2018. (In preparation).
9. Benevide N, **Casado PL**, et al. Active tactile sensibility of Brånemark protocol prostheses: a double-blind randomized clinical study. Int J Oral Maxillofac Implants. 2018. (Submitted).
10. **Benítez Cordero Y**, Fuentes Smith LE, Marcheco Teruel B. Genetic risk factors in cancer of larynx, breast, colon and prostate. Classic Twin Study in Cuba. 2018. (In preparation).
11. **Benítez Cordero Y**, Suáres Besil B, Labrada Moreno LM. Gastroschisis and Onfalocele: Clinical epidemiological aspects in Cuba in the 2007-2016 periods. 2018. **(**In preparation).
12. Bonato LL, Quinelato V, Borojevic R, Vieira AR, Modesto A, Granjeiro JM, Tesch R, **Casado PL**. Haplotypes of the RANK and OPG genes are associated with chronic arthralgia in individuals with and without temporomandibular disorders. Int J Oral Maxillofac Surg. 2017; 29:31407.
13. Bonato LL, Quinelato V, Felipe Cordeiro PC, Sousa EB, Tesch R, **Casado PL**. Association between temporomandibular disorders and pain in other regions of the body. J Oral Rehab. 2017; 44:9-15.
14. **Casado PL**. Tobacco Smoking as a risk indicator for the development of peri-implant diseases. Implant Dent. 2018. (Submitted).
15. **Casado PL**, Quinelato V, et al. Dental genetic in Brazil: Where we are. Mol Genetic Genomic Med. 2018; 00:1-13.

16. Cordeiro PCF, **Casado PL**, et al. *MMP2* polymorphisms are associated with muscular temporomandibular disorder, submitted to the "Int J Oral Maxillofac Surg. 2018. (Submitted).
17. Costa LC, Fonseca MA, Pinheiro AR, Aguiar TRS, Machado NA. Quinelato V, Bonato LL, Aguiar DP, Vieira T, Almeida FLD, Lobo JC, Jordão M, Lomardo PG, Granjeiro JM, **Casado PL**. Chronic Periodontitis and RANKL/OPG Ratio in Peri-Implant Mucosae Inflammation. Braz Dent J. 2018; 29:14-22.
18. **Dhoro M**, Katzenstein D, Makadzange TA. Impact of HLA/KIR genotypes on HIV disease progression in Africans- A focus on perinatally infected children. Central African Journal of Medicine. 2018. (In review).
19. **Dhoro M**, Kennias Rashirai, Alan Mcgregor, Trust Zaranyika. A retrospective data review of Diabetic ketoacidosis at Parirenyatwa group of Hospitals. Central African Journal of Medicine. 2018. (In review).
20. **Díaz C**., Pardo R., Bustos P. Neurological manifestations associated with spina bifida in adults. Family medicine, SEMERGEN, 2018; 44(4), 276-280.
21. Doyard M, Bacrot S, Huber C, Di Rocco M, Goldenberg A, Aglan MS, Brunelle P, Temtamy S, Michot C, **Otaify GA** et al. *FAM46A* mutations are responsible for autosomal recessive osteogenesis imperfecta. J Med Genet. 2018; 55(4):278-284.
22. El-Ruby M, Alaa El-Din F, El-Dessouky SH, Aglan MS, Mazen I, Ismail N, Afifi HH, Eid MM, Mostafa MI, **Mehrez MI** et al. Identification of a novel homozygous ALX4 mutation in two unrelated patients with frontonasal dysplasia type-2. Am J Med Genet A. 2018; 176:1190-1194.
23. **Fatima SS**. Book title 'Emerging trends in Metabolic Syndrome' Publisher: Paramount Books. 2018.
24. **Fatima SS**, Khalid E, Ladak AA and Ali SA. Colostrum and Mature breast milk analysis of Serum Irisin and Sterol regulatory element-binding proteins-1c in gestational diabetes mellitus. J Matern Fetal Neonatal Med. 2018; 2:1-7.
25. Fonseca MA, Costa LC, Pinheiro AR, Aguiar T, Quinelato V, Bonato LL, Almeida FLD, Granjeiro JM, **Casado PL**. Peri-implant Mucosae Inflammation during Osseointegration Is Correlated with Low Levels of Epidermal Growth Factor/Epidermal Growth Factor Receptor in the Peri-implant Mucosae. Intl J Growth Factors Stem Cells Dent. 2018; 1:17-22.
26. Gokhroo RK, **Nandal R**, Garhwal K, Phogat AK, Tasleem TM, Kaul A. Optical Coherence Tomography (OCT) guided percutaneous coronary intervention (PCI) of Right Coronary Artery (RCA) with high thrombus burden after intracoronary thrombolysis with Urokinase. Indian J Cardiol. 2018; 20:50-53.
27. González-Morón D, **Vishnopolska SA**, et al. Germline and somatic mutations in cortical malformations: Molecular defects in Argentinean patients with neuronal migration disorders. PloS One. 2017; 12(9), e0185103.
28. Hiasat JG, Saleh A, Al-Hussaini M, Al Nawaiseh I, Mehyar M, Qandeel M, Mohammad M, Deebajah R, Sultan I, Jaradat I, Mansour A, **Yousef YA**. The predictive value of magnetic resonance imaging of retinoblastoma for the likelihood of high-risk pathologic features. Eur J Ophthalmol. 2018 Jun 1:1120672118781200. doi: 10.1177/1120672118781200. [Epub ahead of print]
29. Jaradat I, Zewar A, AlNawaiseh I, AlRawashdeh K, Khurma S, Mehyar M, Abdeen G, **Yousef YA**. Characteristics, management, and outcome of patients with uveal melanoma

treated by Iodine-125 radioactive plaque therapy in a single tertiary cancer center in Jordan. Saudi J Ophthalmol. 2018; 32(2):130-133.
30. Joshi G, **Thakur N**, Thapa S, Kunwar AJ, et.al. Distribution of TEM, SHV and CTX-M Genes among extended spectrum ß-lactamase producing clinical isolate of *E. coli* and *K. pneumonia* in tertiary care hospital. 2018. (In preparation).
31. Kirijas M, Genadieva Stavrik S, Senev A, Efinska Mladenovska O, **Petlichkovski A**. HLA-A, -B, -C and -DRB1 allele and haplotype frequencies in the Macedonian population based on a family study. Hum Immunol. 2018; 79(3):145-153.
32. Kirijas M, Stavrik SG, Trajkov D, Mitkovska SH, Senev A, Mladenovska OE, Sibinovska O, **Petlichkovski A**. HLA profile of the donors in the Macedonian Bone Marrow Donor Registry. Int J Immunogenet. 2018. (Accepted).
33. Kyobe S, Musinguzi H, Lwanga N, Kezimbira D, Kigozi E, Katabazi A. F, **Wayengera M**, et al. Selecting a Laboratory Information Management System for Biorepositories in Low- and Middle-Income Countries: The H3Africa Experience and Lessons Learned. Biopreserv Biobank. 2017; 15(2): 111–115.
34. Lal KK, Jarwar R, Farhat S, **Fatima SS**. Association of Vaspin levels and its SNP rs2236242 with Gestational Diabetes at a tertiary care setting. J Pak Med Assoc. 2018. (Accepted).
35. Laurino MY, Leppig KA, **Abad PJ**, Cham B, et al. A Report on Ten Asia Pacific Countries on Current Status and Future Directions of the Genetic Counseling Profession: The Establishment of the Professional Society of Genetic Counselors in Asia. J. Genet Couns. 2018; 21(1):21-32.
36. Maddirevula S, Alsahli S, Alhabeeb L, Patel N, Alzahrani F, Shamseldin HE, Anazi S, Ewida N, Alsaif HS, Mohamed JY, Alazami AM, Ibrahim N, Abdulwahab F, Hashem M, Abouelhoda M, Monies D, Al Tassan N, Alshammari M, Alsagheir A, Seidahmed MZ, Sogati S, Aglan MS, Hamad MH, Salih MA, Hamed AA, Alhashmi N, Nabil A, Alfadli F, Abdel-Salam GMH, Alkuraya H, Peitee WO, Keng WT, Qasem A, Mushiba A, Zaki MS, Fassad MR, Alfadhel M, Alexander S, Sabr Y, Temtamy S, Ekbote AV, Ismail S, Hosny GA, **Otaify GA**, Amr K et al. Expanding the phenome and variome of skeletal dysplasia. Genet. Med. 2018; Apr 5. doi: 10.1038/gim.2018.50.
37. **Mahfoudh W**, Bettaieb I, Ghedira R, et al. Contribution of BRCA1 5382insC mutation in Triple Negative Breast Cancer in Tunisia. 2018. (In preparation).
38. Mboowa G, Mwesigwa S, Katagirya E, Retshabile G, Mlotshwa BC, Williams L, Kekitiinwa A, Kateete D, Wampande E, **Wayengera M**, et al. The Collaborative African Genomics Network (CAfGEN): Applying Genomic technologies to probe host factors important to the progression of HIV and HIV-tuberculosis infection in sub-Saharan Africa. AAS Open Res. 2018; 1:3.
39. **Mgasa A**, Lyimo M, Mugusi, et al. Unprecedented magnitude of anemia among people living with HIV in Dar es salaam, Tanzania—is care and treatment adequate? TJHR. 2018. Manuscript ID is ISSNB:1821-9241. (Submitted).
40. Mlotshwa BC, Mwesigwa S, Mboowa G, Williams L, Retshabile G, Kekitiinwa A, **Wayengera M**,et al. The collaborative African genomics network training program: a trainee perspective on training the next generation of African scientists. Genet Med. 2017; doi: 10.1038/gim.2016.177.
41. Muhorakeye A, **Uwineza A**, **Hitayezu J**, and Cartledge P. Developing a Core Outcome Set for a congenital abnormalities surveillance program in Rwanda - a Delphi consensus study. BMC Pediatrics (BPED-D-18-00534) (In review).

42. **Nandal R**, Garhwal K, Phogat A, Tasleem TM, Kaul A, Jain P, et al. Diabetic ketoacidosis masquerading as anterior wall myocardial infarction: case report and discussion of cardiac electrophysiology in hyperkalemia and management. Indian J Cardiol. 2018; 20:41-47.
43. Okee M, Bayiyana A, Musubika C, Joloba ML, Ashaba-Katabazi F, Bagaya B, **Wayengera M**. In Vitro Transduction and Target-Mutagenesis Efficiency of HIV-1 pol Gene Targeting ZFN and CRISPR/Cas9 Delivered by Various Plasmids and/or Vectors: Toward an HIV Cure. AIDS Res Hum Retroviruses. 2018; 34 (1):88-102.
44. Ortega-Olvera C, Ulloa-Aguirre A, Ángeles-Llerenas A, Mainero-Ratchelous FE, González-Acevedo CE, Hernández-Blanco M de L, Ziv E, Avilés-Santa L, Pérez-Rodríguez E and **Torres-Mejía G***. Thyroid hormones and breast cancer association according to menopausal status and body mass index. Breast Cancer Res. 2018; 20(1):94.
45. **Otaify GA**, Abdel-Hamid MS, **Mehrez MI**, et al. Genetic study of eight Egyptian patients with pycnodysostosis: identification of novel CTSK mutations and founder effect. Osteoporos Int. 2018; 29: 1833–1841.
46. **Otaify GA**, Whyte MP, Gottesman GS, et al. Gnathodiaphyseal dysplasia: Severe atypical presentation with novel heterozygous mutation of the anoctamin gene (ANO5). Bone, 2018; 107:161-171.
47. **Owusu M**, Marfo KS, Acheampong G et al., Gonococcal sepsis in a 32-year-old female: a case report. BMC Res Notes. 2018; 11(1):253.
48. **Owusu M**, Owusu-Dabo E, Acheampong G, et al. Pseudomonas oryzihabitans sepsis in a 1-year-old child with multiple skin rashes: a case report. J Med Case Rep. 2017 11(1):77.
49. Pavkovic M, **Petlichkovski A**, Karanfilski O, Cevreska L, Stojanovic A. FC gamma receptor polymorphisms in patients with immune thrombocytopenia. Hematol J. 2018; 23(3):163-168.
50. Pérez Millán MI, **Vishnopolska SA**, et al. Next generation sequencing panel based on single molecule molecular inversion probes for detecting genetic variants in children with hypopituitarism. Mol Genet Genomic Med. 2018; 6:514-525.
51. Prazeres JC, **Casado PL**, et al. Peri-implant health after supportive mucositis therapy is associated with increased levels of FGF-2. J Periodontol. 2018. (Submitted).
52. Quezada Urban R , Díaz Velasquez CE, Gitler R, Rojo Castillo MP, Sirota Toporek M, Figueroa Morales A, Moreno García O, García Esquivel L, **Torres Mejía G**, Dean M, et al. Comprehensive analysis of germline variants in Mexican patients with hereditary breast and ovarian cancer susceptibility. Cancers. 2018. (Submitted).
53. Quinelato V, Bonato LL, Vieira AR, Granjeiro JM, Tesch R, **Casado PL**. Association Between Polymorphisms in the Genes of Estrogen Receptors and the Presence of Temporomandibular Disorders and Chronic Arthralgia. J Oral Maxillofac Surg. 2017; 76:314.
54. Rajeh A, Barakat F, Khurma S, AlRawashdeh K, Ababneh OH, AlNawaiseh I, Mehyar M, Abdeen G, Jaradat I, Mohammad M, **Yousef YA**. Characteristics, management, and outcome of squamous carcinoma of the conjunctiva in a single tertiary cancer center in Jordan. Int J Ophthalmol. 2018;11(7):1132-1138.
55. Retshabile G, Mlotshwa BC, Williams L, Mwesigwa S, Mboowa G, Huang Z, Rustagi N, Swaminathan S, Katagirya E, Kyobe S, **Wayengera M**, et al. Collaborative African Genomics Network (CAfGEN) of the H3Africa Consortium. Whole-Exome Sequencing Reveals Uncaptured Variation and Distinct Ancestry in the Southern African Population of

Botswana. Am J Hum Genet. 2018; Apr 25. pii: S0002-9297(18)30099-5. doi: 10.1016/j.ajhg.2018.03.010. PMID: 29706352.
56. **Roy S**. A case report on Von Hipple Lindu disease. Horizon (Institutional Journal of Chittagong Eye Infirmary and Training Complex. (Submitted).
57. **Roy S**. Nuruddin. M. Clinical features of retinoblastoma patients—In a tertiary eye care center of Bangladesh. (In preparation).
58. Steinmetz I, Gabriel E. Wagner 1,Kanyala E, Sawadogo M, Soumeya H, Teferi M, Andargi E, Yeshitela B, Atsé-Achi AY, Sanogo M, Bonfoh B, Rakotozandrindrainy R, Shongo CP, Pongombo MS, Ilunga EK, Lichtenegger S, Assig K, May J , Bertherat E, **Owusu M**, et al. Melioidosis in Africa: Time to Uncover the True Disease Load Trop. Med. Infect. Dis. 2018, 3(2),62.
59. Suárez C, Aranibar L, Nakousi N, **Díaz C**, **Pizarro P**. Pigmentary mosaicism: cutaneous and extracutaneous involvement. Cytogenetic characterization. 2018. (In preparation).
60. Sukarova-Angelovska E, **Petlichkovski A**. Genetics in Macedonia-Following the international trends. Mol Genet Genomic Med. 2018; 6(1):9-14.
61. Tariq A, Asghar A, Alam F, **Fatima SS**. AHSG rs4918 Polymorphism poses a weak predisposition to insulin resistance during pregnancy. J Pak Med Assoc. 2018; 68: 698-701.
62. Tesch R, Vieira AR, **Casado PL**, et al. Evaluation of genetic risk related to comt and adbrb2 activity in different diagnostic sub-groups of temporomandibular disorder (TMD) in Brazilian patients. J Oral Rehab. 2018. (Submitted).
63. **Thakur N**, Thapa S, Joshi G, Kunwar AJ et.al. Diagnosis of *Entamoeba hystolytica, E. dispar and E. moshkovskii* in stool samples of rural community of Nepal. 2018. Open Medicine Journal. (Submitted).
64. Thakur S, **Thakur N** et. Al. Microalbuminurea and its risk factors in Type 2 Diabetic Patients. 2018. Open Medicine Journal. (Submitted).
65. Thapa S, **Thakur N**, Joshi G, Kunwar AJ, et.al. Identification of *Salmonella typhi* in suspected blood specimen by conventional culture, serology and advanced PCR targeting hila and their antibiogram. 2018. (In preparation).
66. **Vishnopolska SA**, Turjanski AG, Piñero H et al. Genetics and genomic medicine in Argentina. Mol Genet Genomic Med. 2018; 6:481-491.
67. **Wayengera M,** Kateete DPK, Asiimwe B. Mycobacterium tuberculosis thymidylate kinase Antigen Assays for designating incipient, high-risk latent M.tb Infection. BMC Infect Dis. 2018; 18:133.
68. **Wayengera M**, Mwebaza I, Welishe J, et al. Immuno-diagnosis of Mycobacterium tuberculosis in sputum, and reduction of timelines for its positive cultures to within 3 h by pathogen-specific thymidylate kinase expression assays. BMC Res Notes. 2017; 10(1):368.
69. **Wayengera M**, Mwebaza I, Welishe J, et *al*. Sero-diagnosis of Active *Mycobacterium tuberculosis* Disease among HIV Co-infected Persons using Thymidylate Kinase based Antigen and Antibody Capture Enzyme Immuno-Assays. Mycobact Dis. 2017; 7(2). pii: 241.
70. Yasmeen S, Khan UI, Khan GM, **Fatima SS**. Association of Tissue Inhibitor of Metalloproteinase 2 with Non-Alcoholic Fatty Liver Disease in Metabolic Syndrome. Arch Physiol Biochem. 2018; 18:1-6.
71. **Yaylacioglu FT**, Dincer PR. Genome Editing Technologies: From bench side to bedside. Review. Acta Medica. 2018. (In review).

72. **Yaylacioglu FT**, Dincer PR. Trials to mimick autosomal dominant TGFBI-related corneal dystrophies in zebrafish by using CRISPR/Cas9: Challenges and Possibilities. (In preparation).
73. **Yaylacioglu Tuncay F**, Tefon AB**,** Özdek Ş. Turkish Family with Malattie Levantinese carrying EFEMP1 mutation. Case Report. (In preparation).
74. **Yaylacioglu FT**, Tefon AB, Üçgül AY, Özmen C, Aydın B. Gelatinous drop-like corneal dystrophy (GDLD) and Epithelial Debridement- Diamond Burr Polishing (ED-DBP) treatment. (In preparation).
75. **Yousef YA**, Al-Hussaini M, Nazzal R, Abdeen G, et al. Rosai-Dorfman disease masquerading as Uveal Melanoma: Case report and review of literature. Hematol Oncol Stem Cell Ther. 2018 Jun 15. pii: S1658-3876(18)30061-X. doi: 10.1016/j.hemonc.2018.05.005.
76. **Yousef YA**, Tbakhi A, Al-Hussaini M, AlNawaiseh I, et al. Mutational analysis of the RB1 gene and the inheritance patterns of retinoblastoma in Jordan. Fam Cancer. 2018;17(2):261-268.

2016 Batch

77. Abdullahi ZA, Morenikeji OA, **Adeyemo AA**, Ogunleye, V. O. Helminthiasis and chronic suppurative otitis media in Ijoun Community in Ogun State, Nigeria. J Public Health Epidemiol. 2018; 10(2): 28-33.
78. **Adeoye AM**, Adeabayo Oladimeji, Tayo Bamidele , Adebiyi Adewole, Owolabi Mayowa; Prevalence of ECG abnormalities among adults with metabolic syndrome in a Nigerian Teaching Hospital. Afr Health Sci. 2018. (In review).
79. **Adeoye AM**, Ovbiagele B, Ogah O, et.al: Echocardiographic abnormalities and predictors of 1-month outcome of stroke among West Africans in the SIREN Study. Stroke. 2018. (In review).
80. **Adeoye AM**, Tayo BO, Owolabi MO, et al. Ambulatory blood pressure threshold for black Africans: more questions than answers. J Clin Hypertens. 2018; 20(5):847-849.
81. **Adeyemo AA**, Amodu OK, **Ekure EE**, Omotade OO. Medical genetics and genomic medicine in Nigeria. Mol Genet Genomic Med. 2018;1–8. DOI: 10.1002/mgg3.419
82. Adeniyi OF, **Ekure E**, Olatona FA, Ajayi EO, Nwaoma Nworgu, Nutritional Assessment and Maternal Perception of Toddler Body Size using Toddler Silhouette Scale in Nigeria a Developing Country. IJMA. 2018; 7(1): 9-16.
83. Adetayo AM, **James O**, **Adeyemo WL**, Ogunlewe MO, Butali A. Unilateral cleft lip repair: A comparison of treatment outcome with two surgical techniques using quantitative (anthropometry) assessment. J Korean Assoc Oral Maxillofac Surg. 2018; 44:3-11.
84. Akinyemi RO, Sarfo FS, Akinyemi J, Singh A, Onoja Akpa M, Akpalu A, Owolabi L, **Adeoye AM**, Obiako R, Wahab K, Sanya E. Knowledge, attitudes and practices of West Africans on genetic studies of stroke: Evidence from the SIREN Study. Int J Stroke. 2018; Jul 24:1747493018790059.
85. Akpalu A, Ovbiagele B, Gebregziabher M, Sarfo F, Iheonye H, Akinyemi R, Akpa O, Tiwari H, Arnett D, Wahab K, Lackland D, **Adeoye AM**, Ogbole Goodwin. Differential Impact of Risk Factors on Stroke Occurrence among Men vs. Women in West Africa: the SIREN Study. Stroke. 2018. (In review).

86. **Ariani Y**, Priambodo R, Dewi LM, Pangestika Y, et al. Variations on Exon 3 of 6-Pyruvoyl-Tetrahydropterin Synthase Gene ln lndonesian Population. JPCS. The 2nd Physics and Technologies in Medicine and Dentistry Symposium 2018. (Submitted).
87. **Ariani Y**, Priambodo R, Purwanto MF, Pangestika Y, et al. Novel variations on exon 4 of Iduronate 2-sulfatase gene in six Indonesian patients with Mucopolysaccharidosis type II. JPCS. The 2nd Physics and Technologies in Medicine and Dentistry Symposium 2018. (Submitted).
88. **Ariani Y**, Priambodo R, Purwanto MF, Widyaningrum AR, et al. ldentification of Novel Variation in lntron 3 And 4 of lduronate-2-Sulfatase 6ene on Mucopolysaccharidosis Type ll Patients in Indonesia. JPCS. The 2nd Physics and Technologies in Medicine and Dentistry Symposium 2018. (Submitted).
89. **Ariani Y**, Priambodo R, Putri ANK, Pangestika Y et al. ldentification of a Novel Mutation on Exon 6 of lduronate-2-Sulfatase Gene in an Indonesian Patient With Mucopolysaccharidosis Type ll. JPCS. The 2nd Physics and Technologies in Medicine and Dentistry Symposium. 2018. (Submitted).
90. **Ariani Y**, Priambodo R, Putri DC, Yuliarti K, et al. Mutation Analysis for Exon 4 of 6-Pyruvoyl-Tetrahydropterin Synthase Gene in Indonesian Population. JPCS. The 2nd Physics and Technologies in Medicine and Dentistry Symposium 2018. (Submitted).
91. Bonadonna LV, Saunders MJ, **Guio H**, Zegarra R, Evans CA. Socioeconomic and Behavioral Factors Associated with Tuberculosis Diagnostic. Delay in Lima, Peru. Am J Trop Med Hyg. 2018; 98(6):1614-1623.
92. Bonadonna LV, Saunders MJ, Zegarra R, Evans C, Alegria-Flores K, **Guio H** Why wait? The social determinants underlying tuberculosis diagnostic delay. PLoS One. 2017; 12(9):e0185018.
93. Butali A, Pagán-Rivera K, Dawson DV, Munger R, Eshete MA, **Adeyemo WL,** Mossey PA. Descriptive Epidemiology of Orofacial Clefts in Africa Using data from 46,502 Smile Train Surgeries. J Public Health Epidemiol 2017; 9:114-121.
94. Clark Feoktistova Y, Ruenes Domech C, García Bacallao EF, **Roblejo Balbuena H**, Collazo Mesa T, Clark Feoktistova I, Morales Peralta E, et al. Molecular genetic analysis of exon 3 of the ATP7B gene in Cuban patients with Wilson's disease. Rev Haban Cienc Méd [Internet]. 2018;17(3). http://www.revhabanera.sld.cu/index.php/rhab/article/view/2112.
95. **Ekure EN**. Molecular Diagnosis and Digital Facial Analysis Technology Applications in Congenital Cardiovascular Disorders. Lecture, 5th Faculty of Clinical Sciences Gathering from Univ. of Lagos, Lagos. July, 2018; (published in Book format).
96. **Ekure EN**, Bode-Thomas F, Sadoh WE, Orogade AA et al. and the Nigerian Pediatric Cardiology study group. Congenital Heart Defects in Nigerian Children: Preliminary data from the National Pediatric Registry. World J Pediatr Congenit Heart Surg. 2017; 8(6):699-706.
97. **Ekure EN**, Kalu N, Sokunbi OJ, Kruszka P, et al. Clinical Epidemiology of Congenital Heart Disease in Nigerian Children 2012-2017. Birth Defects Research. 2018. (In press)
98. **Ekure EN**, Kruszka P, Sokunbi OJ, Kalu N et al. Case report on neurofibromatosis with congenital heart defect. (In preparation)
99. **Ekure EN**, Kruszka P, Sokunbi OJ, Kalu N et al. Case report on Tuberous Sclerosis. (In preparation).
100. **Ekure EN**, Sokunbi OJ, Kalu N et al. Echocardiographic Screening of 4107 Nigerian School Children for Rheumatic Heart Disease. (In preparation).

101. Emeka CI, **Adeyemo WL,** Ladeinde AL, Butali A. A comparative study of quality of life of families with children born with cleft lip/ palate before and after surgical treatment. J Korean Assoc Oral Maxillofac Surg 2017; 43:247-255.
102. Eshete MA, Liu H, Li M, **Adeyemo WL,** Gowans LJJ, Mossey PA, Busch T, Deressa W, Donkor P, Olaitan PB, Aregbesola BS, Braimah RO, Oseni GO, Oginni FAudu , R, Onwuamah C. **James O,** et al. Loss-of-Function GRHL3 Variants Detected in African Patients with Isolated Cleft Palate. J Dent Res 2018; 97: 41-48.
103. Flores-Villanueva PO, Ganachari M, **Guio H**, Mejia JA, Granados J An Isolated TCR αβ Restricted by HLA-A*02:01/CT37 Peptide Redirecting CD8+ T Cells To Kill and Secrete IFN-γ in Response to Lung Adenocarcinoma Cell Lines. J Immunol. 2018; 200(8):2965-2977.
104. Galarza M, **Guio H**, Reques J, Omar Piscoya, Mitzi Rodríguez Molecular diagnosis of multidrug resistant tuberculosis in sputum samples by analysis of melting curves in the callao district. Rev Peru Med Exp Salud Publica. 2018. (In press).
105. Ganesh S, Arora P, **Tibrewal S**. Ocular Manifestations of Albers-Schoenberg Disease (Osteopetrosis). J Pediatr Ophthalmol Strabismus. 2018; 55(4):276.
106. Gashegu J, **Uwineza A**, Kubwimana O. Proximal Femoral Focal Deficiency associated with Fibula hemimelia: An uncommon Experience, Case report and review of literature. Rwanda Medical Journal. 2018. (In Review).
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109. **Guio H**, Poterico J, Levano L, Jaramillo L. Genetics and Genomics in Peru. 2018. (In preparation).
110. 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. Genome Biol Evol. 2018. (Submitted).
111. Harris DN, Song W, Shetty AC, Omar 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. Proc Natl Acad Sci U S A. 2018;115(28).
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113. Ifeoluwa A, Adewole A, **Adeoye AM**, Akinyemi A: Right Ventricular Systolic Function in Subjects with Heart Failure Secondary to Hypertensive Heart Disease; African Health Sciences (In review).
114. Jacobs M, Hulst VM, **Adeoye AM**, Tieleman RG, et al; Atrial fibrillation in Africa - an underreported and unrecognized risk factor for stroke: a systematic review. The Lancet Glob Health. 2018. (In review).
115. Jaiyeola MT, **Adeyemo AA**. Quality of life of deaf and hard of hearing students in Ibadan metropolis, Nigeria. PLoS ONE. 2018;13(1): e0190130.
116. Jaramillo L, Levano K, Galaraza M, Caceres O, **Guio H**. Rapid identification technique for drug-resistant Mycobacterium tuberculosis isolates using mismatch-specific cleavage enzyme. Bioinformation. 2018; 14(7):404-407.

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118. Kayode O, **Adeyemo AA**. The Yoruba version of LittlEARS Auditory Questionnaire: evaluation of auditory development in children with normal hearing. Journal of Otology. 2018. (Accepted).
119. Khurana R, **Tibrewal S**, Ganesh S, Tarkar R, Nguyen PT, Siddiqui Z, Dasgupta S. Accuracy of noncycloplegic refraction performed at school screening camps. Indian J Ophthalmol 2018; 66:806-11.
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122. **Lallar M**, Srivastava P, Phadke SR. A large interstitial 11q deletion with isolated mild intellectual disability: Review of literature for genotype phenotype correlation. Clinical Dysmorphology. 2018; May 29. doi: 10.1097/MCD.0000000000000228.
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124. Levano K, Jaramillo L, Capristano S, Tarazona D, **Guio H**. SNPs genomic editing in active tuberculosis progression for new diagnostic method. 2018. (Manuscript in preparation).
125. Levano K, Jaramillo L, Valdivia J, **Guio H**. Macrophages modification by CXCR4 genomic editing and effect on metastatic breast cancer. 2018. (In preparation).
126. Mishra R, Paththinige CS, **Sirisena ND**, Nanayakkara S, et al. Partial trisomy 16q21➔qter due to an unbalanced segregation of a maternally inherited balanced translocation 46,XX,t(15;16)(p13;q21): a case report and review of literature. BMC Pediatr. 2018; 18:4.
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129. **Okafor FU**. Prevalence and prediction of retinoblastoma among patients attending a tertiary health institution in Benin City, Edo State, Nigeria. Nigeria Journal of Education, Health, and Technology Research (NJEHETR). 2018; 10 (1):141-149. http://njehetr.aprehet.org/create-articlesubmissioncnt-13 OR www.aprehet.org.

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134. Paththinige CS, **Sirisena ND**, Kariyawasam UGIU, Ediriweera RC, Kruszka P, Muenke M, Dissanayake VHW. 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. BMC Med Genomics. 2018; 11:44.
135. Prabodha LBL, **Sirisena ND,** Dissanayake VHW. Susceptible and Prognostic Genetic Factors Associated with Diabetic Peripheral Neuropathy: A Comprehensive Literature Review. Int J Endocrinol. 2018; Article ID 8641942, 9 pages.
136. Prendergast EA, Perkins S, Engel ME, Cupido B, Francis V, Joachim A, Al Kebsi M, Bode-Thomas F, Damasceno A, Abul Fadl A, El Sayed A, Gitura B, Kennedy N, Ibrahim A, Mucumbitsi J, **Adeoye AM**, et al. REMEDY investigators. Participation in research improves overall patient management: insights from the Global Rheumatic Heart Disease registry (REMEDY). Cardiovasc J Afr. 2018; 29 (2):98-105.
137. Priambodo R, **Ariani Y**, Pangestika Y, Hafifah CN, et al. A Novel Silent Mutation at Exon 9 of lduronate-2-Sulfatase Gene in an lndonesian Patient with Mucopolysaccharidosis Type ll. JPCS. The 2nd Physics and Technologies in Medicine and Dentistry Symposium 2018. (Submitted).
138. Ranawaka R, **Sirisena ND**, Dayasiri KC, et al. The first Sri Lankan family with Dent disease‑1 due to a pathogenic variant in the *CLCN5* gene: a case report. BMC Res Notes. 2017; 10:539.
139. Ratnasamy V, Suganthan S, **Sirisena ND,** Grüning N, Brandau O, et al. Dyskeratosis congenita with a novel genetic variant in the DKC1 gene: a case report. BMC Med Genet. 2018; 19:85.
140. **Roblejo Balbuena H**. CYP2D6 polymorphism, ethnic background and extrapyramidal side-effects of antipsychotics in Cuban inpatients with schizophrenia. (In preparation).
141. **Seven M,** Paşalak Ş.İ, Guvenc G, Kok G. Knowledge Level and Educational Needs of Turkish Oncology Nurses Regarding the Genetics of Hereditary Breast and Ovarian Cancer. J Contin. Educ. Nurs. 2017; 48(12):570-576.
142. **Seven M,** Paşalak ŞI, Şahin E. Akyüz A. Pregnant Women's Genetics Literacy and Their Use of Prenatal Screening and Diagnostic (In Review).

143. Singh B, Mandal K, **Lallar M**, Narayanan DL, et al. Next Generation Sequencing in Diagnosis of MLPA Negative Cases Presenting as Duchenne/ Becker Muscular Dystrophies. Indian J Pediatr. 2018; 85(4):309-310. DOI 10.1007/s12098-017-2455-5.
144. **Sirisena ND** and Dissanayake VHW. Taking Genomics from the Bench to the Bedside in Developing Countries. In: Translational and Applied Genomics Series - Genomic Medicine in Emerging Economies – Genomics for Every Nation". Lopez-Correa C and Patrinos GP (Ed.) Academic Press (Elsevier). July 2018.
145. **Sirisena ND**, **Adeyemo AA**, Kuruppu AI, Samaranayake N, Dissanayake VHW. Genetic Variants Associated with Clinicopathological Profiles in Sporadic Breast Cancer in Sri Lankan Women. J Breast Cancer. 2018; 21(2): 165-172.
146. **Sirisena ND**, **Adeyemo AA**, Kuruppu AI, Neththikumara N, Samaranayake N, Dissanayake VHW. Genetic determinants of sporadic breast cancer in Sri Lankan women. BMC Cancer. 2018; 18:180.
147. **Tibrewal S**, Kekunnaya R. Risk of Anterior Segment Ischemia Following Simultaneous Three Rectus Muscle Surgery: Results from a Single Tertiary Care Centre, Strabismus. 2018; 26:2, 77-83.

B. Grants: (Total- 49; 2017- 30; 2016- 19)

2017 Batch

1. **Avogbe PH** (PI), Sehonou J, Le Calvez-Kelm F, McKay JD, Sanni A (Co-PI). Circulating tumor DNA biomarkers and early detection of hepatocellular carcinoma. (IARC/WHO funded, Accepted).
2. **Casado PL** (PI), Alves T. Grants to Initiate Technology Training for student in molecular biology (miRNA isolation). FAPERJ (Fundação de Amparo à Pesquisa do Estado do Rio de Janeiro, FAPERJ, Dec. 2017. IT235321, Received).
3. **Casado PL** (PI), Regina C. Grants for Training students for Starting Research Experience. from (Conselho Nacional de Desenvolvimento Científico e Tecnológico, CNPq) / UFF (Fluminense Federal Univ., Aug. 2018, IC180261, Received).
4. **Dhoro M.** Triggering factors and underlying mechanisms of diabetic ketoacidodis in newly diagnosed type 2 diabetic patients in Zimbabwe (NIH K43 award, to be submitted).
5. **Dhoro M.** Retrospective review of congenital disorders at Parirenyatwa Hospital in Harare, Zimbabwe (Univ. of Zimbabwe Research Fund, to be submitted).
6. Dinçer P (PI), Kürekçi GK, Kural E, **Yaylacıoğlu Tuncay F**, Önal G, Ünsal S. Preparation and maintenance of CRISPR/Cas9 platforms in zebrafish specific to rare diseases. (Local grant from the Univ. of Hacettepe, Ankara, Turkey, TAY-201712735, Received).
7. **Fatima SS** (PI), Rehman R, Khan U, Mohammed N. Epigenetic signature of high contribution susceptibility gene variant for diabetes and obesity. (Univ. Research Council, Aga Khan Univ., Accepted).
8. **James O** (PI), **Adeyemo WL**. A Comparison of 2-Octyl Cyanoacrylate (Dermabond®) tissue adhesive and sutures in the repair of Cleft Lip at the Lagos Univ. Teaching Hospital - A Randomized Controlled Clinical trial. (CRC Research Grant (CRC/2018/07), Univ. of Lagos Research Grant, Received).

9. Jha AK (PI), **Thakur** N (Co-PI). Population based screening of sickle cell disorder of Tharu community of Bardiya district. (Nepal Health Research Council under Ministry of Health and Population, Nepal, Received).
10. Kadir MM (PI-PAK), Ali MK (PI-USA), Narayan KM, Hu FB, Ahmad S, **Fatima SS** (Co-PI). Molecular Signatures Associated with Obesity and Risk of Diabetes in Pakistan (Agency: HEC-USAID-NIH under PAK-US Science & Technology Cooperation Program Phase-7, Received).
11. Kateete D (PI), Jjingo, **Wayengera M** (Co-I). Nurturing Genomics and Bioinformatics Research Training in Africa (BreCA). (NIH/NIAIDS/Forgarty, 1U2RTW010672-01 Received).
12. Kunwar A. (PI), **Thakur N** (Co PI). Detection of *Helicobacter pylori* from water sources and biofilm of Kathmandu valley using Real Time PCR. (The World Academy of Sciences, TWAS; No. 17-478 RG/BIO/AS_I – FR3240297773, Received).
13. Matshaba M (PI), CAfGEN Investigators, **Wayengera M** (Co-I)**.** Collaborative African Genomics Network (CAFGEN). (NIH/NIAID, Received).
14. **Mgasa A**. Afya Bora Consortium Fellowship in Global Health Leadership- 2018/2019. (Afya Bora Consortium, Received).
15. **Otaify GA** (PI), Abdelhamid MS (Co-PI). Targeted next-generation sequencing in the diagnosis and prevention of skeletal dysplasias with joint deformities. (Young researcher STDF grant funded by Science and Technology Development in Egypt, Submitted)
16. **Owusu M** (PI), Sylverken J, Nguah SB, Peprah AB, Adu-Sarkodie Y (all Co-PIs). Microbiome of lung infections among children in Ghana (European and Developing Countries Clinical Trial Partnership, EDCTP CDF, Received).
17. Sukarova E (PI), **Petlichkovski A** (PI). The role of genetic polymorphisms of MTHFR in children with developmental disorders associated with clefts of body structures. (Medical Faculty in Skopje, Univ. Sv. Kiril i Metodij, Received).
18. **Thakur N** (PI), Thapa S (PI). Distribution and comparison of Human Papilloma Virus Genotype in Cervical Cancer in Three Different Ecological Regions of Nepal. (Nepal Health Research Council, Received).
19. Tofoski G (PI), **Petlichkovski A** (PI). Association of genetic polymorphisms in killer cells immunoglobulin genes with pregnancy complications in Republic of Macedonia. (Medical Faculty in Skopje, Univ. Sv. Kiril i Metodij, Received).
20. **Torres-Mejía G** (PI). Strategic project for the prevention and control of breast cancer. (Consejo Nacional de Ciencia y Tecnología (CONACyT), Project number: 549, 2018, Approved).
21. **Wayengera M** (PI). Development and Evaluation of 3 vectors administered via iliac-bone marrow infusion(s) for purposes of delivering anti-HIV-1 pol gene targeted ZFN and CRISPER-Cas9 to CD4+ve T cell progenitors and reservoirs. (NIH/NIAID, AN # 4199590, RFA-AI-18-016, Submitted).
22. **Wayengera M** (PI). Development and Validation of the next generation of xenogenetic therapeutics based on AcsI and ApoI nucleases for sustained HIV-1 remission (NIH/NIAID, AN # 4199591, RFA-AI-18-017, Submitted).
23. **Wayengera M** (PI). Experimental testing of a Gene –Therapeutic Vaccine for Malaria using purified Reticulocytes from donor Bone Marrow. (Grand Challenges Africa, 2018 Drug Discovery Call, application #6207, Submitted).
24. **Wayengera M** (PI). Instrumental Access Application. (Seeding Labs, 2018, Submitted).

25. **Wayengera M** (PI). Receiver Operator Characterization of Novel EBOV/MARV-GP Epitopes using 2014-2015 Sierra Leonean Ebola Patient-Samples at the NICD BSL-IV. (EDCTP, TMA2016CDF-1545, Received).
26. **Wayengera M** (PI), Joloba M, Jerome K. Validation of reversal of cervical neoplasia by target-mutagenesis of the high-risk HPV oncogenes E6 and E7 in HeLa cell-lines. (World Cancer Research Fund International Regular Grant Programme, 2018-2019, Application # 7954, Submitted).
27. **Wayengera M** (PI), Joloba M, Roscigno G. Development and Validation of Mycobacterium Tuberculosis Thymidylate Kinase based Lateral Flow and Microfluidics platforms as Superior Alternative Replacement for Smear-microscopy and traditional Culture. (EDCTP, RIA - 2018 - Diagnostics – LoI, application #s RIA2018D-LoI-2063, Submitted).
28. **Wayengera M** (PI), Nyakarahuka L, Nelson RE, Owino M. Establishment and positively impacting the KAPs around rapid diagnostic tests (RDTs) in an on-going outbreak setting of the Democratic Republic of Congo, DRC. (IDRC/SSHRC/CIHR Rapid Research Fund for EVD Outbreaks, 2018, Submitted).
29. **Yousef YA**, Tbakhi A, Al-Hussaini M, Mehyar M. Mutational analysis of the RB1 gene and the inheritance patterns of retinoblastoma in Jordan. (King Hussein Cancer Center Intramural grant, 70K USD, Received).
30. Abildinova G (PI), Tolegen N (Co-PI). Molecular-genetic analysis of the subtelomeric and centromere region of the chromosome with mental retardation (State of Kazakhstan, Submitted).

2016 Batch

31. **Adeoye AM** (PI Nigeria), Karmacharya B (PI Nepal), Di Cesare M, Hakim F, et al. The GOALPoST Study: improving Global access to Oral AnticoaguLants to Prevent Stroke in aTrial fibrillation. (World Heart Federation Emerging Leaders Grant Award, Received).
32. **Adeoye AM** (PI), Abiola B, Adebayo O. Association between selected molecular biomarkers and ambulatory blood pressure pattern in African Chronic Kidney disease and hypertensive patients compared with normotensive controls (SYMBOLIC STUDY). (International Society of Hypertension Research Scholar Grant Award, Received).
33. 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). Awarded.
34. Deniz G (PI), **Deniz E** et al. Identification and integration of clinical use of immunogenetic substructures in the diagnosis and treatment of primary immunodeficiencies; mainly antibody deficiencies, combined immunodeficiencies and phagocyte system disorders. (Acibadem Univ. and Istanbul Univ., Turkey). (Scientific and Research Council of Turkey (TUBITAK), Submitted).
35. **Guio H (PI)**. To develop a new kit to extract DNA using magnetic pearls which need minimal facilities to promote molecular biology research in remote areas. (INNOVATE PERU, Received).
36. **Guio H (PI)**. Circulating microRNAs as biomarkers for tuberculosis infection in a high TB burden setting. (Peru science and technology fund, Received).
37. **Guio H (PI)**. Susceptibility to Guillain-Barré syndrome is associated with Gene polymorphisms IL17, iCAM-1 and CD1(INBIOMEDIC, Received).

38. **Guio H**, Shady R **(PI)**. Susceptibility and resistance genes identification of the human coprolites microbiome in Caral 2000 B.C. (Peru science and technology fund, Applied).
39. **Guio H (PI),** Valdivia J (Co-PI). Development of a microfluidic system (Lab-on a-chip) for the detection of circulating tumor cells in regions of low and medium resources. (Medical College of Peru, Received).
40. Hou, L, Sagay A, Ogunsola FT, Murphy M, A**deyemo WL** (Co-investigator). Epigenomic Biomarkers of HIV-Associated Cancers in Nigeria. (NIH U54, Received).
41. Hou, L, Sagay A, Ogunsola FT, Murphy M, **Adeyemo WL** (Co-investigator) Northwestern/Nigeria Research Training Grant for HIV and Malignancies. (NIH/NCI, Applied).
42. Mohamed AM. (PI), **Ahmed DF** (collaborator). Copy Number Variance and Gene Expression in Diagnosis of genetic causes of Intellectual Disability (Center of Excellence for Human genetics – The National Research Center- Egypt) (Received). Skilled to join this project after attending a detailed demonstration on microarrays with Dr. Settara Chandra, at the NIH Intramural Sequencing Center (NISC) as a part of the advanced training of the summit.
43. Mutesa L, Uddin M, Wildman D, Jansen S. (Multiple PIs), **Uwineza A** (co-investigator). Transgenerational Epigenomics of Trauma and PTSD in Rwanda. Human Heredity and Health in Africa (H3Africa): Research Projects (U01, NIH, Accepted).
44. 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. Proposal includes genetic studies on deafness. (Univ. of Ibadan, Univ. College Hospital Ibadan, Nigeria, and other centers- Nigeria, Revised).
45. **Uwineza A** (PI). Genetic etiology of epilepsy in Rwandan patients with intellectual disability'. Funded by Univ. of Rwanda for Post-doctoral grant and SIDA-SAREC. (Received).
46. **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 Univ., China, Received).
47. Sjarif DR (PI), **Ariani Y** (co-PI). Identification of Iduronate 2-sulfatase gene defect using mRNA from Indonesian MPS type II patients. (PITTA 2018, Univ. of Indonesia, Accepted).
48. Sjarif DR (PI), **Ariani Y (co-PI).** The establishment of method to detect Iduronate 2-sulfatase enzyme activity. (PUPT Ministry of Research, Technology and High Education Republic of Indonesia, Received).
49. Sjarif DR (PI), **Ariani Y (co-PI).** Variants analysis of glucosylceramidase beta (GBA) gene in type I Gaucher disease from Indonesia. (PITAA 2018, Univ. of Indonesia, Accepted).

C. Collaborations (NIH and Other Institutions):

NIH: (Total- 12; 2017- 3; 2016- 9)

2017 Batch

1. **Lotz-Esquivel S.** Atlas of Human Malformation Syndromes in Diverse Populations (Williams syndrome, https://research.nhgri.nih.gov/atlas/condition/williams-syndrome/). (Hospital Nacional de Niños, Costa Rica; NHGRI/NIH).
2. **Mgasa A**, Muenke M, **Ekanem E**, Pranoot T, Patil SJ et al. The International Genetic Basis of Congenital Heart Disease Study Collaborative Project**. (**Submitted for approval at Muhimbili Univ. of Health and Allied Science, Tanzania; 2018; NHGRI/NIH).

3. **Owusu M**, Kruzka P, Muenke M, et al., Genetics of congenital heart disease in Ghana. (Kwame Nkrumah Univ. of Science and Technology; NHGRI/NIH).

2016 Batch

4. **Adeyemo A**, Griffith A and Friedman TB. Genetic mutations responsible for deafness in Nigerians. (Univ. College Hospital, Ibadan, Nigeria; NIDCD/NIH).
5. **Adeyemo A** and Prokunina L. Characterization of HPV genotypes in head and neck cancer patients in Nigeria. (Univ. College Hospital, Ibadan, Nigeria; NCI/NIH).
6. **Ahmed FD**, Kruszka P, Muenke M**.** Atlas of Human Malformation syndromes in Diverse populations (Williams Syndrome, Down syndrome & Turner syndrome (https://research.nhgri.nih.gov/atlas/condition/). (The National Research Center, Egypt; NHGRI/NIH).
7. **Belhassan K**, Muenke M, Ouldim. Genetic diseases in diverse populations. (Genetics Department of Hassan II Univ. Hospital, Fes, Morocco; NHGRI/NIH).
8. **Ekure EN**, Kruszka P and Muenke M. Atlas of Human Malformation Syndromes in Diverse Populations: (College of Medicine, Univ. of Lagos, Nigeria/Lagos Univ. Teaching Hospital, Nigeria; NHGRI/ NIH).
9. **Sirisena ND** and Bonnemann C. Diagnostic evaluation of patients with complex neuromuscular & neurogenetic conditions without a definite diagnosis. (Human Genetics Unit, Faculty of Medicine, Univ. of Colombo, Sri Lanka; NINDS/NIH).
10. **Okafor F, Abad PJ,** Saligan L**.** Study on Nurses' knowledge, attitude and belief on genetics and genomics in Nigeria and Philippines. Proposal stage. (Univ. of Benin, Nigeria; Univ. of Philippines Manila; NINR/NIH).
11. **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. (Human Genetics Unit, Faculty of Medicine, Univ. of Colombo, Sri Lanka; NCI/NIH).
12. **Sirisena ND**, Kruszka P and Muenke M. Diagnostic evaluation of patients with various craniosynostosis syndromes, congenital heart diseases and other congenital malformations. (Human Genetics Unit, Faculty of Medicine, Univ. of Colombo, Sri Lanka; NHGRI/NIH).

Other Institutions: (Total- 39; 2017- 23; 2016- 16)

2017 Batch

1. **Abad, PJ**, MY Laurino, MR Tumulak, et al. Family Functioning and Supportive Care Needs of a Child Diagnosed with a Rare Disease. In discussion (Univ. of the Philippines Manila; Seattle Cancer Care Alliance, Seattle, Washington; Univ. of South Australia).
2. **Avogbe PH** (PI), Le Calvez-Kelm F, and McKay JD. Circulating tumor DNA biomarkers and early detection of hepatocellular carcinoma (Univ. of Abomey-Calavi, Benin; International Agency for Research on Cancer/WHO).
3. **Casado PL**. FDI Consensus in Peri-implant Disease Project.Peri-implant Disease Project. Consensus from FDI (Fluminense Federal Univ., Rio de Janeiro, Brazil; World Dental Federation, 2018).
4. **Casado PL** and Alexandre Rezende Vieira. Researches related to genetic basis of temporomandibular disorder/ peri-implant disease/ periodontal disease in partnership with Oral Biology Laboratory (Fluminense Federal Univ., Rio de Janeiro, Brazil; Univ. of Pittsburgh, USA).

5. **Casado PL** and Georgios E. Romanos. Researches publications about growth factors in peri-implant diseases. (Fluminense Federal Univ., Rio de Janeiro, Brazil; School of Dental Medicine, Stony Brook Univ., USA).
6. **Dhoro M**, Mochly-Rosen D, Grimes KV. In discussions about collaboration and assistance on the diabetes project. (Univ. of Zimbabwe; Stanford Univ.).
7. **James O, Avogbe PH** et al. Genetic Polymorphism Predisposing to Nonsyndromic Orofacial Clefts Risk: An Evaluation of Major Findings of the Genome-Wide Association Studies in Unstudied African Populations (Faculty of Dental Sciences, College of Medicine, Univ. of Lagos, Nigeria; Univ. of Abomey-Calavi, Department of Biochemistry and Cellular Biology, Benin).
8. Kaliki. S, Fabihan.D. **Roy S**. Global Clinical Presentations of Retinoblastoma. (London School of Hygiene, UK; L.V.Prasad Eye Institute, Hyderabad, India; Chittagong Eye Infirmary, Bangladesh and 98 more centers around the world)
9. Kaliki. S, Fabihan.D, **Roy. S.** Lag time of Retinoblastoma presentation. (London School of Hygiene, UK; L.V. Prasad Eye Institute, Hyderabad, India; Chittagong Eye infirmary, Bangladesh; 14 more institutes around the world).
10. **Lotz-Esquivel S,** Saborío-Rocafort M, Badilla-Porras R, et al. Genetic diagnoses for monogenic diseases (MSUD, GM1 and Crigler-Najjar type 1) and familial genetic diseases of unknown genetic etiology. In discussions (deCode, Iceland, Hospital Nacional de Niños Costa Rica; Univ. of Pennsylvania, USA).
11. **Lotz-Esquivel S,** Saborío-Rocafort M, Badilla-Porras R, et al. Natural History Study for MSUD. In discussions (Hospital Nacional de Niños, Costa Rica; Univ. of Pennsylvania, USA).
12. **Mahfoudh W,** Rebbeck T and Klingner TM**.** BRIDGE **(***BRCA1/2*International Diversity by Geography and Ethnicity project. In discussions (Univ. of Monastir, Tunisia; Univ. of Cambridge).
13. **Mgasa A** and Shukuru. Establish registry to record and track outcome of children born with congenital birth defects. In discussion (Temeke Regional Refferal Hospital, Tanzania; National Blood Transfusion Service, Tanzania; Ministry of Health Community Development, Gender, Elderly and Children,Tanzania).
14. **Mgasa A**. Utilization of genetic laboratory for DNA extraction before transfer of genetic materials. In discussion (Biochemistry and Genetics Departments at Muhimbili Univ. of Health and Allied Science; National Blood Transfusion Service; Ministry of Health Community Development, Gender, Elderly and Children, Tanzania).
15. **Mgasa A.** Genetic counselling services to patients with congenital heart diseases and their parents. In discussion (Jakaya Kikwete Cardiac Institute; National Blood Transfusion Service; Ministry of Health Community Development, Gender, Elderly and Children, Tanzania.
16. **Tolegen N,** Shevcov A. Research human gene and genomic on neuromuscular diseases (National Science Biotechnology Center, Department of Applied Genetics; National Scientific Center for Mothers and Children at Department of Clinical-genetic diagnosis).
17. **Vishnopolska, SA** and Sally Camper. Genetics of Hypopituitarism. (Univ. of Buenos Aires, Buenos Aires, Argentina; Univ. of Michigan).
18. **Vishnopolska, SA** and Jacob Kitzman. Development of Molecular Inversion Probes (MIPs) for detecting SNV in mendelian diseases. Development of MAVEs for massive parallel functional studies of SNV (Univ. of Buenos Aires, Buenos Aires, Argentina; Univ. of Michigan).

19. **Wayengera M,** Jerome K. Validation of reversal of cervical neoplasia by target-mutagenesis of the high-risk HPV oncogenes E6 and E7 in HeLa cell-lines. (Univ. of Washington/Fred Hutchinson Cancer Research Centre, USA; College of Health Sciences, Makerere Univ.).
20. **Wayengera M** and Prof. Paweska J. "Receiver Operator Characterization of Novel EBOV/MARV-GP Epitopes using 2014-2015 Sierra Leonean Ebola Patient-Samples at the NICD BSL-IV". (NICD P4 Lab, Center for Emerging Zoonotic Diseases, NHLS, South Africa; College of Health Sciences, Makerere Univ.).
21. **Wayengera M**. Pitching of Tuberculosis Rapid Diagnostic Test (TBrdt) for Venture Capital Investiment. (Through Advancing Health Innovation in Africa (AHIA) program**,** Emory Univ., USA; College of Health Sciences, Makerere Univ.).
22. **Wayengera M**. and Roscignio G. Development and Validation of Mycobacterium tuberculosis Thymidylate Kinase based Lateral Flow and Microfluidics platforms as Superior Alternative Replacement for Smear-microscopy and traditional Culture. (Foundation for Innovation of New Diagnostics; College of Health Sciences, Makerere Univ.).
23. **Yousef YA**, Tbakhi A, Al-Hussaini M, AlNawaiseh I, Gallie B, Jewett F, et al. Mutational analysis of the RB1 gene in Jordanian Population. (King Hussein Cancer Center, Amman, Jordan; Impact Genetics Lab, Canada).

2016 Batch

24. **Adeoye AM**, Bongani M, Connolly SJ. INVestIgation of rheumatiC AFTreatment Using vitamin K antagonists, rivaroxaban or aspirin Studies (INVITUS trial). (Univ. of Ibadan; Univ. of Cape Town; Population Health Research Institute, Canada).
25. **Adeoye AM**, Karaye KK, Loeb M, Yusuf S. A Randomized Controlled Trial of Influenza Vaccine to Prevent Adverse Vascular Events (IVVETrial). (Univ. of Ibadan, Bayero Univ., Kano; McMaster Univ., Canada).
26. **Adeoye AM**, Tayo B, Ojo A, Cooper R. Cardiovascular And Renal Events In People With Chronic Kidney Disease (Univ. of Ibadan; Univ. of Arizona College of Medicine, Tucson, AZ; Loyola Univ. Chicago Stritch School of Medicine, Maywood, IL).
27. **Adeyemo AA**, **Adeoye AM**, et.al. Correlations between Hypertension and Hearing Loss In Patients Attending Cardiology Clinic At The Univ. College Hospital, Ibadan. (Department of Ear Nose and Throat (ENT) and Department of Medicine Univ. College Hospital, Nigeria).
28. **Adeyemo WL.** Extending the Phenotype of Non-Syndromic Orofacial Clefting (Univ. of Lagos; Univ. of Iowa, USA; Univ. of Pittsburgh, USA)
29. **Adeyemo WL.** Epigenomic Biomarkers of HIV-Associated Cancers in Nigeria (Univ. of Lagos, Univ. of Jos, Nigeria; Northwestern Univ., USA)
30. Bukini D (PI), Makani J, Manji K, Mbekenga C, **Malasa L**, Tutuba H. Newborn Screening for Sickle Cell Disease: Exploring ethical, social and social based implications. (Muhimbili Univ. of Health; Allied Sciences and MUHAS Sickle Cell Centre).
31. **Ekure EN** (PI), Bode-Thomas F, Sadoh WE, Orogade AA et al**.** Nigerian Pediatric cardiac disease registry (Univ. of Lagos, Lagos; Univ. of Jos; Univ. of Benin; Ahmadu Bello Univ.; Univ. of Port Harcourt; Univ. of Nigeria; Nsukka Usmanu Danfodiyo Univ.; Sokoto, Bayero Univ.; other Univs. in Nigeria).
32. Husien AA, **Hussen DF,** Hammad SA, Refaat KM. Studying cases of Alzheimer disease and correlating cytogenetic biomarkers with severity of cognitive status. (The National Research Centre, Egypt; El Aini Hospital, Cairo University, Egypt).

33. Mulder N, Wonkam A, **Uwineza A.** Review of the of Sickle Cell Disease and ontology. (Univ. of Cape Town, H3Bionet; College of Medicine and Health Sciences/ Univ. of Rwanda).
34. Mutesa L, Jansen S, **Uwineza A**, Uddin M, Wildman D. Transgenerational Epigenomics of Trauma and PTSD in Rwanda. (Woese Institute for Genomic Biology, Univ. of Illinois, Urbana- Champaign; CMHS, Univ. of Rwanda).
35. **Roblejo HB**, Llerena Ruiz A. Working on a PhD in Pharmacogenetics and Schizophrenia. (Univ. of Havana, Cuba; Clinical Research Center in the Univ. Hospital Infanta Cristina en Badajoz Spain).
36. Sjarif DR, **Ariani Y,** Priambodo R. Variants analysis of IDS, PTPS and GBA gene. (Faculty of Medicine Univ. of Indonesia; Cipto Mangunkusumo Hospital Jakarta; Faculty of Mathematic and Natural Science Univ. of Indonesia; Dr. Mohammad Hoesin Hospital South Sumatra; Dr Soetomo Hospital East Java; Sanglah Hospital Bali; Dr. Sardjito Hospital Yogyakarta).
37. Sjarif DR, **Ariani Y,** Priambodo R. mRNA detection method to optimize IDS gene defect identification. (Faculty of Medicine Univ. of Indonesia; Cipto Mangunkusumo Hospital Jakarta; Faculty of Mathematic and Natural Science Univ. of Indonesia; Dr. Mohammad Hoesin Hospital South Sumatra; Dr Soetomo Hospital East Java; Sanglah Hospital Bali; Dr. Sardjito Hospital Yogyakarta).
38. **Tibrewal S**, Vanita V. Phenotypic-genotypic correlation of pediatric cataract cases in north India. (Shroff Charity Eye Hospital; Guru Nanak Dev Univ., Punjab).
39. Urio F (PI), Nkya S, **Malasa L**, Mgaya J, Makani J, Menzel S. Genetic determinants of F cells in Sickle Cell Disease (Muhimbili Univ. of Health and Allied Sciences; MUHAS Sickle Cell Centre).

D. New Research Projects: (Total- 43; 2017- 29; 2016- 14)

2017 Batch

1. **Abad, PJ**. Content Analysis of Messages in a CAH Support Group Social Media Page: Implications in Genetic Counseling. Proposed.
2. **Avogbe PH**. Characterization of human papillomavirus (HPV) genotypes from cervical specimen of women living in Cotonou, Benin.
3. **Benítez Cordero Y**, Suárez Besil B, et al. Characterization of the major congenital defects in Cuba in the 2007-2020 periods. Integrated System for its Management.
4. **Casado PL**. Association between peri-implant supportive therapy and growth factors levels.
5. **Casado PL**. Association of polymorphisms for metalloproteinasis and muscular temporomandibular disorder.
6. **Casado PL**. Characterization of S. pneumoniae levels in gingival crevicular fluid.
7. **Casado PL.** Analysis of miRNA in subjects under TMD and bruxism.
8. **Cordero YB**, Suáres Besil B et al. Characterization of the major congenital defects in Cuba in the 2007-2020 periods. Integrated System for its Management.
9. **Dhoro M.** Determine the status of congenital disorders at Parirenyatwa Hospital, Harare, Zimbabwe.
10. **Dhoro M.** Establishment of a biorepository inventory for the Univ. of Zimbabwe, College of Health Sciences, to support genomics research.
11. **Fatima SS**. Genomics and Environmental determinants of obesity in South Asians.

12. **Fatima SS**, Shahid S. Markers of feto-maternal cardiovascular risk assessment in preeclampsia.
13. **James O**. Syndromes associated with Orofacial clefts: the Lagos Univ. Teaching Hospital experience. (Research topic developed after the summit to collate and possibly find the genetic and environmental predisposing factors to these anomalies).
14. **Lotz-Esquivel S**, Alvarado-Aguilar MV and Acuña-Feoli JA. Acute intermittent porphyria: a case series from 2007 – 2017.
15. **Nandal R**, Gokhroo RK. Clinical Characteristics, risk factors, Angiographic Profile and in Hospital Mortality in Acute Coronary Syndrome Patients in young north Indian Population.
16. **Nandal R,** Gokhroo RK. OCT characteristics of young patients with re-canalized vessels post thrombolysis (< 50 % stenosis on angiography), and their follow up for 6 months with repeat coronary angiography and OCT.
17. **Otaify GA**. Targeted next-generation sequencing in the diagnosis and prevention of skeletal dysplasias with joint deformities. young researcher STDF grant funded by Science and Technology Development (Submitted)
18. Penón-Portmann M, **Lotz-Esquivel S**, et al. Wilson's disease: Mutation analysis of the ATP7B gene and characterization of a Costa Rican pediatric cohort.
19. **Roy S**. Outcome of periocular capillary haemangioma with oral propranolol in an outpatient basis.
20. **Roy S**. Demographic profile and clinical presentation of thyroid eye disease in Bangladesh.
21. Sukarova E, **Petlichokovski P**. The role of genetic polymorphisms of MTHFR in children with developmental disorders associated with clefts of body structures.
22. Tofoski G, **Petlichokovski P**. Association of genetic polymorphisms in killer cells immunoglobulin genes with pregnancy complications in Republic of Macedonia.
23. Vaca-Paniagua Felipe (PI), **Torres-Mejía G** (Collaborator), Díaz-Velásquez Clara Estela (collaborator), Martínez-Razo Gabriel (collaborator), Moreno-Macías Hortensia, Romieu Isabelle. Identification of pathogenic variants of susceptibility to early-onset breast cancer in young women (CONACyT, 000000000285879).
24. **Vishnopolska SA**. Functional Studies of SNVs in Hypopituitarism and Diseritropoyetic Anaemia.
25. **Wayengera M** (Co- investigator). Integrated Biorepository of H3Africa (IBRH3AU).
26. **Wayengera M** (Co- investigator). Nurturing Genomics and Bioinformatics Research Training in Africa (BreCA).
27. **Wayengera M** (Co- investigator). Collaborative African Genomics Network (CAFGEN). The goal of this training grant is to build capacity for genetics and genomic training study among collaborating African Institutions through research on the genetic determinants of pediatric HIV and TB disease-progression.
28. **Yaylacioglu FT**, Ünlü M. Genetic analysis of a family with anterior segment dysgenesis.
29. **Yousef YA**. The Impact of RB1 Gene Mutation Type in Retinoblastoma Patients on Clinical Presentation and Management Outcome.

2016 Batch

30. **Adeoye AM,** Owolabi MO. Exploring the Phenomics, Genomics and Environmental determinants of left ventricular mass among offspring of Hypertensive African Blacks: AFamily screening study.

31. **Adeoye AM**, Ayede AI. CARdiovascular rIsk Factors and diseases among pregnant women and their Foetal outcome in Ibadan (CARdIFF Study)
32. **Adeoye AM**, Kuti M. Carotid Intima Media Thickness and Lipid Markers of Atherosclerosis among Hypertensives.
33. **Adeoye AM**, Raji RY. The role of Renin Angiotensin Aldosterone pathway genetic polymorphisms in circadian blood pressure variations in Nigerian patients with chronic KIDney disease (RAASKID Study).
34. **Deniz E**. Identification and Functional Investigation of New Long Noncoding RNAs (lncRNA) Role in DNA Damage Responses.
35. **Ekure EN**, Kruszka P, Sokunbi OJ, Kalu N et al. Case reports on genetic diseases associated with congenital heart defects (Tuberous sclerosis, Neurofibromatosis etc.)
36. **Lallar M,** Srivastava P, Phadke SR. Clinical exome experience of clinicians – questionnaire based approach.
37. **Malasa L**, Luzzatto L and Makani J. Estimation of proportional of intravascular and exravascular hemolysis in Sickle Cell Disease.
38. Mutesa L, Uddin M, Wildman D, Jansen S, **Uwineza A**. Transgenerational Epigenomics of Trauma and PTSD in Rwanda. NIH. (H3Africa funded Research Project).
39. Nkya S, Mtei L, **Malasa L**, et al. Newborn Screening for Sickle Cell Disease in Dar es Salaam, Tanzania.
40. Olatosi O, **Adeyemo WL**, Butali A. Investigating the genetic causes of early childhood caries.
41. Sjarif DR, **Ariani Y,** Priambodo R. Estabishment Iduronate 2-sulfatse enzyme activity measurement in type II MPS patients.
42. Sjarif DR, **Ariani Y,** Priambodo R. mRNA identification method to detect IDS gene defect in Indonesia type II MPS patients.
43. Sjarif DR, **Ariani Y,** Priambodo R. Variants analysis of GBA gene in Gaucher disease Indonesian patients.

E. Ongoing Research Projects: (Total-35; 2017- 21; 2016- 14)

2017 Batch

1. Abimana D (Investigator), **Uwineza A**. (Supervisor), **Hitayezu J**. (Co-supervisor): Description of disorders of sex development (DSD) in children and adolescents in referral hospitals in Rwanda. Study completed. (Obtained the idea of initiating a registry for recording and tracking of congenital birth defects, including DSD, from the Summit, and knowledge gained helped organize writing of scientific paper).
2. **Casado PL**. Analysis of 7 different techniques to isolate microRNA.(Knowledge from the bioinformatics class at the Summit provided clarity on the relationship of miRNA with DNA and messenger RNA which helped improve the execution of this project).
3. **Casado PL**. Genetic profile of patients that underwent peri-implant maintenance therapy: relationship with peri-implant disease incidence. (Knowledge from the Summit helped improve sample collection, medical history analysis, selection of the main related genes in multifactorial peri-implant diseases).
4. **James O**. Cardiovascular Anomalies In Patients With Oro-facial Clefts: A case-Controlled Prospective Study. (Summit gave more insight into the findings and interpretation of results).

5. Le Calvez-Kelm F (PI/IARC/WHO), **Avogbe PH** (Co-investigator), et al. Urinary circulating *TERT* promoter mutations as non-invasive biomarkers for the detection of primary and recurrent urothelial cancer. (Summit helped meet researchers with similar interests for advice).
6. Le Calvez-Kelm F, **Avogbe PH** (Co-investigator), et al. Whole transcriptional profiling and gene expression analysis in patients with urothelial carcinoma. (Summit helped meet researchers with similar interests for advice).
7. **Lotz-Esquivel S**, Matarrita-Quesada B, Monge-Bonilla C, Kuhn-Delgadillo K. Cardiac surgery in Costa Rica: patient characterization at Hospital San Juan de Dios from 2010 to 2015. (Knowledge from Summit helped write scientific paper on congenital heart diseases in adults).
8. Marcheco Teruel B, **Benítez Cordero Y**, Suárez Besil B, et al. Evaluation of the impact of the National Program for Diagnosis, Handling and Prevention of Genetic diseases and congenital related defects. (Summit helped reaffirm usefulness of data from different specialties for-development of personalized medicine; clinical validity of gene-disease associations; registries for genetic epidemiological studies).
9. Medina Reyes Y, Barrios Martínez A, **Benítez Cordero Y**, et al. Contribution of supernumerary chromosomal markers to the appearance of clinical phenotypes. (Summit lectures introduced tools for molecular genetics used to identify chromosomal markers).
10. Mutesa L (PI), **Hitayezu J.** (Co-investigator), **Uwineza A**. (Co-investigator). Newborn Screening in Rwanda: A pilot survey to screen for congenital hypothyroidism and congenital adrenal hyperplasia. Data collection completed. (Learnt of newborn screening and its benefits in low-income countries at the Summit; knowledge gained helped organize writing of scientific paper).
11. **Otaify GA**. Co-PI in project entitled "Clinical and molecular study of patients with inherited metabolic bone disorders" in National Research Centre Grant. (Knowledge gained from Summit helped improve reporting of results).
12. **Otaify GA**. (Collaborator) Predictive Cytogenetic Biomarkers for Non-disjunction disorders. National Research Centre Grant. (Knowledge gained from Summit helped improve reporting of results).
13. **Otaify GA**. (Collaborator) Immunogenetic approach in diagnosis and prognosis of some genetic disorders. National Research Centre Grant. (Knowledge gained from Summit helped improve reporting of results).
14. **Otaify GA**. (Collaborator) "Recent Advances in the Diagnosis, Management and Research of Genetic Diseases". STDF Grants for Centers of Scientific Excellence (STDF-CSE). (Knowledge gained from Summit helped improve reporting of results).
15. **Owusu M**. The microbiome of lung infections among HIV and non-HIV children in Ghana: A case-control study (MOLIC Study). (Knowledge from the Summit helped develop protocols and reshape the study design to obtain sufficient data from this work).
16. Sabina Rinaldi (PI), **Torres-Mejía G** (Co-PI). Molecular Subtypes of Premenopausal Breast Cancer in Latin American Women (PRECAMA): a multicenter population-based case-control study (IARC 2012-). (Summit informed of state-of-the-art in genetics and genomics; inclusion of gene-environmental interaction in breast cancer).
17. **Wayengera M.** Development of lateral flow tests to serve as Rapid Diagnostic Tests to replace smear microscopy for TB. (Summit improved career standing and helped draw more funding).
18. **Wayengera M.** Development of Microfluidics MEMS for ultra-rapid TB culture detection. (Summit improved career standing and helped draw more funding).

19. **Wayengera M**. Animal testing for a gene-therapeutic cure for HIV-1. (Summit improved career standing and helped draw more funding).
20. **Wayengera M**. Validation and ROC characterization of duo EBOV and MARV rapid diagnostic biomarkers. (Summit improved career standing and helped draw more funding).
21. **Yaylacioglu Tuncay F,** Dinçer P. Evaluation of phenotypic effects of genome editing mediated TGFBI variation on zebrafish cornea. (Through the Summit met researchers who helped resolve issues with the project).

2016 Batch

22. **Ekure E** (PI, Nigeria), Kruszka P, Adeyemo A, Muenke M et al**.** Genetic basis of congenital heart disease in Africa. (Summit gave insight into genetics of congenital heart defects and relationship with other birth defects. Learnt practical application of bioinformatics to the data set from this study).
23. **Ekure E.** Nigerian Pediatric cardiac disease registry. (Summit emphasized driving this registry as it is a perfect tool to track congenital heart defects not just locally but nationally making a national genetic study on congenital heart defects much easier.
24. Hammad SA (PI), **Ahmed DF.** (Co-investigator) Predictive Cytogenetic Biomarkers for Non-disjunction disorders. (Center of Excellence for Human genetics – The National Research Center- Egypt). (Continued from 2016-17).
25. Helmy N (PI), **Ahmed DF** (collaborator). Using of microarray technique in diagnosis of agenesis of corpus callosum. (Center of Excellence for Human genetics – The National Research Center- Egypt). (Continued from 2016-17).
26. Mahmoud A (PI) **Ahmed DF** (collaborator). Nicotine dependence as an environmental health problem, the efficacy of different approaches for its management. 3-year project. (The National Research Centre, Egypt). (Continued from 2016-17).
27. **Malasa L**. A case report on Schizencephally. (With knowledge gained from the Summit on genetic diseases, was able to provisionally diagnose the condition by CT Scan and now is writing a case report).
28. Ogunlewe MO, James O, **Ekure E.** Cardiovascular anomalies in patients with Congenital Oro-facial clefts. (Summit strengthened partnership with some of the collaborators on this project).
29. **Roblejo HB.** CYP2D6 genetic polymorphism and extrapyramidal side effects of antipsychotic drugs in patients with schizophrenia. (With knowledge gained from the Summit, added the analysis of Ancestry).
30. Sjarif DR, **Ariani Y,** Priambodo R, Pratanu L. CNVs detection in multiple congenital anomaly patients using Infinium cyto-SNP 850K bead chip. (Continued from 2017).
31. Sjarif DR, **Ariani Y**, Yuliarti K, Hafifah CN. Genotype-phenotype analysis of type II MPS patients from Indonesia. (Continued from 2017).
32. Sjarif DR, **Ariani Y**, Priambodo R. Urine heparan and dermatan sulfate measurement as a screening method in neonatal period. (Continued from 2017).
33. Sjarif DR, **Ariani Y**, Priambodo R**.** Variant analysis of IDS gene in 30 Mucopolysaccharidos type II patients type II patients. (Continued from 2017).
34. Sjarif DR, **Ariani Y**, Priambodo R**.** Variant analysis of PTS gene in PKU. (Continued from 2017).
35. **Uwineza A** (PI), **Hitayezu J.** (Co-investigator). Genetic etiology of epilepsy in Rwandan patients with intellectual disability'. Data collection completed, data analysis ongoing.

(Knowledge gained from the Summit helped include microdeletion and microduplication of genes in addition to organizing writing of the scientific paper).

F. **Other Initiatives/Accomplishments/Presentations:**

2017 Batch

1. **Abad PJ**. Chosen as Director Philippines (2018-2020), Transnational Alliance of Genetic Counseling (TAGC, incorporated 2006 by Univ. of South Carolina), to foster genetic counseling education, communication, collaboration transnationally, among academic institutions; Co-organized the 2nd Preconference in Genetic Counseling (Nov. 2017), in conjunction with the 12th Asia Pacific Conference in Human Genetics; Membership Director of Professional Society of Genetic Counselors in Asia; Presented at the preconference on 'The results of the survey on the roles and added value of genetic counselors in the Asia Pacific region'; Invited to hold a 1-day workshop on genetic counseling (Ho Chi Minh City, Viet Nam, Apr. 2018), by Center for Reproductive Health, Viet Nam National Univ., and by the KT Biotechnology Inc., attended by physicians and laboratory technicians; Invited by Philippine Oncology Nurses Association (Jan. 2018), as a resource person on the scope of nursing practice in cancer genetics and genomics; Invited by the School of Nursing of the Centro Escolar Univ., Manila, Philippines, (Jun. 2018, Faculty Development Seminar), as a resource person on the role of nurses in genetics.
2. **Avogbe PH.** Invited as a Visiting Scientist to join the Genetic Cancer Susceptibility Group/ International Agency for Research on Cancer/WHO (Lyon, France); Co-opted into Curriculum Review Committee (Jun. 2018), Department of Biochemistry and Molecular Biology, Faculty of Science and Techniques; Lectured Genetics and Genomics to graduate level students at Univ. of Abomey-Calavi, Benin, expanded curricula to include three additional courses (from 2019); Leading advocacy efforts for integration of Genomics into medical curricula.
3. **Casado PL**. Reviewer for Archives of Oral Biology and International Journal of Growth Factors and Stem Cells in Dentistry (2017-2018); Invited to join the Peri-implant Disease Project organized by FDI World Dental Federation and International Congress of Implantologists, to address lack of awareness on peri-implant diseases and to update recommendations/guidelines for oral health professionals (Zurich, Switzerland, May 2018); Invited to present on " Peri-implant diseases: how to proceed in clinical practice? at the Dental Treatment Congress (EuroScieCon 2018) – 25th International Conference of Dental Treatment (Zurich, Switzerland, Sep. 2018); Mentored students in molecular biology and research through grant funding from FAPERJ and CNPq (Univ. of Fluminense Federal).
4. **Cordero YB**. Updated medical genetics professionals on advances in Genetics and Genomics, Human Heredity and Variation, NCBI Genetic databases; Lectured pre-conference course on epidemiological surveillance of congenital defects, at the International Congress of Community Genetics (Havana, Cuba, Nov. 2017).
5. **Dhoro M.** Included human genetics and genomic research in the curriculum; Used resources obtained from the Summit to compile teaching material; Lectured on a) genetic basis of disease and gene therapy, b) diagnosis of genetic disorders, c) genetic counselling, d) genomic medicine and pharmacogenetics to Master's level students (medical doctors, postgraduates) in Clinical Pharmacology; Based on evaluation of courses, students are amazed at the technology and research capacity in Zimbabwe; Increased interest and awareness for genetic counselling

training, currently role is fulfilled by doctors; Received complimentary membership and travel award from the International Society for Biological and Environmental Repositories (ISBER) to attend the annual meet (Dallas, Texas, 2018), crucial for the bio-banking project; Invited to Stanford Univ. to meet with experts in endocrinology and diabetes, for mentorship and collaborations.

6. **Díaz C**. Completed internships in Cytogenetics, Clinical Genetics and endocrinology, Clinical Genetics, Infant Neurology, Clinical Genetic and Neonatology (Chile); Summit strengthened medical curriculum; Presented clinical case at 57 Chilean Congress of Pediatrics (Suractivo, Concepción, Chile); Attended graduate level class of 'Mucopolisacaridosis II' organized by SHIRE (Rio de Janeiro Brazil); Attended 'Epidemiology of congenital anomalies in the genomic era' of the Latin American Center for Interdisciplinary Training (CELFI), Ministry of Science, Technology and Productive Innovation, Univ. of Buenos Aires (Buenos Aires, Argentina); Coordinator and speaker for a course 'Experiences in Neurology and Psychiatry' (Nov. 2018, Chile) to bridge genomics and clinics.
7. **Fatima SS**. Educated Faculty and Staff at Department of Biological and Biomedical Sciences on opportunities available at NIH (training programs, grants and fellowships); Conducted research workshops for undergraduate medical, nursing and biomedical students, emphasized the role of good protocol writing and the need for including Genetics and Genomics in research and health care, especially in low resource settings; Trained research technician in field of bioinformatics and next generation genomic raw data analysis, based on the advance training received at NIH; Plans underway to offer a short course on Genomics and precision medicine in Metabolic Syndrome to nurses, undergraduate and postgraduate professionals; Presented a poster on Molecular Basis of NAFLD at the 78th Scientific Session American Diabetes Association (Florida, USA, Jun. 2018); Presented a paper on Molecular Dynamics study of Adipokine at the 1st PROBE conference, Univ. of Karachi (Karachi, Pakistan, Jan. 2018); Submitted an abstract for oral presentation at the South Asian Association of Physiology (SAAP) VI Conference (Lahore, Pakistan, Dec. 2018); Became a member of The International Society for Developmental Origins of Health and Disease, which promotes research in the fetal and developmental origins of disease, involves scientists from various fields; Member of Translational Research on Functional Foods/Complementary Therapies in Cardio-metabolic Disorders in Pakistani population.
8. **Hitayezu J**. Established the Rwandan Down Syndrome Organization, included children and families with Down Syndrome; Member, Rwanda Society of Human Genetics; Presented to inform colleagues, local health professionals, hospital pediatric staff (Dec. 2017) about the Summit; Active Co-supervisor in developing a birth defect registry in Rwanda; Helped with the 11th Conference of the African Society of Human Genetics (AfSHG) (Kigali, Rwanda, Sept. 2018); Received Travel Award from AAP on International Child Health and Section on International Medical Graduate to present on 'A cross-sectional study to evaluate adherence to the Ten Steps to Successful Breastfeeding at a referral hospital in Rwanda' at the NCE meeting (Nov. 2018); No specific registration of cases, 1 case/ week seen in the weekly general pediatric consultation, suspect cases sent to a genetic clinic, children with Down syndrome encouraged to join the Rwanda Down syndrome organization;
9. **Lotz-Esquivel S**. Invited to join the Multidisciplinary Clinic for Rare and Orphan Diseases at Hospital San Juan de Dios, San José, Costa Rica, a National Reference Center for adult population with genetic and metabolic conditions, to start a database on metabolic disorders and congenital syndromes (102 patients currently with diverse pathologies); Consolidation of

information resulted in efficient multidisciplinary consult (one visit), better communication and understanding, resulted in improved quality of life for patients and families; Submitted application as a volunteer intern to World Health Organization to aid in the inclusion of Genetics and Genomics in Universal health coverage; Invited by Dr. Stephen G. Kaler (NICHD/NIH) to attend the 11th International Copper Meeting: Bridging Clinical and Fundamental Research in Copper Biology (Sorrento, Italy, Sept. 2018); Invited to attend Aarhus Wilson's Symposium (Aarhus Univ., Denmark, May 2019).

10. **Mahfoudh W**. Selected for the African Cancer Leaders Institute (2017), to help develop the next generation of leaders in cancer research, advocacy, education, policy, and clinical practice in Africa; Presented a poster 'Screening for common BRCA1 mutations in Tunisian women with triple negative breast cancer' at the 11th International Conference of the African Organization for Research and Training in Cancer (Rwanda, Nov. 2017); Awarded the C.V. Raman Fellowship (Jun. 2018) for African Researchers to undertake research in Next Generation Sequencing (NGS), statistical analysis of NGS data and application of comparative genomics methods to identify novel drug targets, at the International Centre for Genetic Engineering and Biotechnology, New Delhi, India.
11. **Mehrez MI**. Provided guidance to colleagues in developing lectures for oro-dental scientific day, based on learnings from the Summit; Attended as Faculty the basic course in Human Genetics, (National Research Centre, Cairo, Egypt, Mar. 2018); Organized the oro-dental genetics department scientific day (May 2018); Applied for the USaid graduate scholarships to attend training as part of a higher education initiative; Speaker at the 10th Sofia dental meeting (Bulgaria, Oct. 2017); Speaker at the 10th conference of the African Society of Human Genetics (Cairo, Nov. 2017); Submitted abstract for 11th Annual African Society of Human Genetics Conference (Kigali, Rwanda); Identified 92 genetic disorder cases; Referred 11 cases for management of dental condition; 10 cases experienced better of quality of life due to oral rehabilitation resulting in better appearance, speech and eating; Currently recruiting patients in Blood & Geno-dermatosis and Oro-dental Genetics clinics, National Research Centre, Cairo, Egypt where "PhenoDP" was explained.
12. **Mgasa A**. Initiated project to create awareness among clinicians and policy makers on the role of genetics in disease causation; Provided Summit feedback to the Director of Clinical Services, presented report to the Ministry of Health Community Development Gender, Elderly and Children Tanzania (Oct. 2017); Discussed future collaboration with NIH with Biochemistry Departmental Head and at the Government Chief Medical Officer and Director's Meeting (Feb. 2018, approval granted); Discussed procedures to conduct genetic studies and genetic material transfer with Chief Government Chemist (Feb. 2018); Presented on the role of genetic counselling to patients and families of children with congenital heart disease at Jakaya Kikwete Cardiac Institute (Apr. 2018).
13. **Nandal R**. Lectured an Introductory Clinical genetics to undergraduate and post graduate students (Nov. 2017) and Genetics in Cardiology to DM colleagues and professors (Dec. 2018); Focused on congenital heart diseases in neonates and Familial Hyperlipidemia with follow up and management of identified patients and families; Involved in genetic studies of such families; Maintained data on children with cardiac defects, in the pediatric cardiology clinic (OPD, wards); Identified ~50 congenital heart diseases (simple and complex), Marfan syndrome (2), Fabry disease (1), Familial Hyperlipidemia (3); Patients were treated, counseled and/or referred; Referred patients with Fabry disease, Marfan syndrome, suspected of Noonan syndrome for genetic analysis and extended family screening; Families were informed and

relieved as questions were answered after a prolonged period of time due to rarity of the condition.

14. **Otaify GA**. Helped organize a Human Genetics workshop in conjunction with Clinical Genomics and NGS course at the European School of Genetic Medicine, Bertinoro, Italy, web casted lecture to National Research Centre (Cairo, Egypt, Apr. 2018); Lectured a newly introduced course on Basics of Human Genetics to undergraduate students in Faculty of Physiotherapy, Cairo Univ. for 14 weeks, summit helped structure it; Lectured on "Genetics of Limb Development in Basic Course of Human Genetics held at National Research Centre (Mar. 2018); Presented at the10th conference of the African Society of Human Genetics (Nov. 2017, Egypt); Presented at the 14th Middle East Metabolic Group meeting (Greece, Feb. 2018) Poster presentation at ASHG (2018, California, USA); Outpatient consultation, management and counseling improved for patients with genetic diseases including hereditary bone disorders as a result of the Summit (15-20 new cases/week, 20 follow-ups for treatment or continued investigation till diagnosis); Participated in medical Caravans in rural Fayoum governorate, Egypt to raise awareness on genetic disorders and risks due to consanguinity, measures to prevent recurrence and decrease the burden on families and societies.
15. **Owusu M.** Selected as a fellow for the Africa Research Excellence Fund for training young African researchers in research leadership; On the committee for organizing a conference on Genetics at the Child Health Department of the Komfo Anokye Teaching Hospital (Ghana, Dec. 2018).
16. **Petlichkovski A**. Implemented NGS platform; Improved program for specialization in laboratory clinical genetics with inclusion of the latest recommendations for diagnosis and treatment of genetic conditions; Included Summit lectures and data in the curriculum at the Medical Faculty.
17. **Pizarro P**. Presented clinical case "Sexual differentiation disorder, possible case of Campomelic Acromegal Dysplasia" at the 57 Pediatrics Congress, South Active Convention Center (Concepción, Chile, Nov. 2017); Participated in weekly meetings at the Department of Genetics, Biomedical Sciences Institute, Univ. of Chile, to educate colleagues on the Summit, its contribution to knowledge and in creating bonds between people from different parts of the world; Presented Dr. William A. Gahl's (Clinical Director/NHGRI; Head, Section on Human Biochemical Genetics Human Genetics Branch/ NHGRI) lecture "Diagnosis and Treatment of Patients with Inborn Errors of Metabolism or Other Genetic Disorders" to educate physicians and graduate school students interested in genetics about the management of such patients at NIH and for similar implementation in Chile; Participated in the national registry of congenital anomalies (RENACH) along with other doctors, nurses and midwives; Collaborated with countries in South America, to form Latin American collaborative study of congenital malformations (ECLAMC), which holds annual meetings to discuss international contingency issues. Informed them of the importance of completing the implementation and improvement of registries; During internships at hospitals and clinics educated physicians (pediatricians, neonatologists, gynecologists, pediatric neurologists) in genetics and increased their interest in the field; Integrated learnings from the Summit to attain similar standards of practice in genetics to improve patient care, management of their condition and quality of life.
18. **Roy S.** Speaker at 15th Annual conference of Bangladesh Ophthalmological Society (Dhaka, May 2018); Speaker at 14th SARRC Ophthalmological Association(SAO) Conference (Kathmandu, Jun. 2018); Developed a new registration form for the hospital to identify, track and monitor congenital malformations and genetic diseases; Identified 84 genetic disorder

cases, treated 64 patients and referred 19 patients for medical management ; 32 cases experienced better quality of life due to better cosmetic appearance of eye and visual rehabilitation; Based on learnings from the Summit delivered regular awareness/education sessions to colleagues on ocular genetic diseases and systemic associations.

19. **Thakur N**. Improved patient care from diagnosis to counseling as a result of the Summit; Diagnosed 186 patients at the national referral center, as a result of learnings from the Summit; Collaborations established with India for Non-Invasive Prenatal Testing (NIPT), sequencing etc. of samples (not done before in Nepal), resulted in affordable diagnosis as opposed to travelling to India; Guest speaker presented on Genetic Testing in Oncology (based on oncogenetics lectures at the Summit), at 4th SFO-N International Cancer Conference (Nov. 2017); Organized 2nd symposium on Medical Genetics (Mar. 2018); Guest Speaker presented on 'Genetics of breast cancer' (based on oncogenetics lectures at the Summit), at 3rd NCHCON-2018 Updates in Breast Cancer, National Cancer Conference (Mar. 2018); Organized International Brain Research Organization- Asia Pacific Regional Committee (IBRO-APRC) Associate School and 3rd Annual Meeting of Neuroscience Society of Nepal (May 2018), in collaboration with Neuroscience Society of Nepal; Strengthened curriculum for postgraduate residents (MD) at Department of Pathology, National Academy for Medical Sciences (NAMS), Bir Hospital through compulsory one month rotation in Genetics and Genomics, initiated by the ability to design short courses due to attendance of the Summit.
20. **Tolegen N**. Presented 'Frequency and Structure of Congenital and Hereditary Pathology" at Actual Issues Health Protection of Mother and Child Conference (Astana, Kazakhstan, Oct. 2017); To present a Report: 'Chromosomal Aberrations in Reproductive Pathology' at Innovative Approaches to the Modernization of Medical Education, Science and Practice (Semey, Kazakhstan, Nov. 2018). **Vishnopolska, SA**. Lectured two postgraduate courses: "Human Genetics" and "School of Clinical Genomics"; Participative member in a grant for Precision Medice from Agencia Nacional de Promoción Cientifica y Tecnologica (ANPCyT).
21. **Torres-Mejía G.** Invited speaker on 'Epidemiology, Genetics and genomics in the XXIX', at the National Meeting in Medicine Research (Nuevo León, Mexico, Oct. 2017).
22. **Vishnopolska S**. Taught two post graduate courses Huma Genetics and School of Clinical Genomics; Member in a grant for Precision Medicine from Agencia Nacional de Promoción Cientifica y Tecnologica (ANPCyT).
23. **Wayengera M.** Lectured Medical genetics in undergraduate in General Medicine at Makerere Univ. College of Health Sciences, Kampala, Uganda; Participated as MakCHS-Select Advisory Representative, Biotechnology and Biosafety (a.k.a GMO) Bill; Organizing Committee Member for School of Biomedical Sciences Annual Health Conference; Member of Research, Grants and Contracts Committee-MakCHS; MakCHS Representative of the 50 Scientists' Advisory Committee for the African Network for New Diagnostic Innovation (ANDI) Pre-Symposium to the African Union Heads of State meeting on Ebola and other disease Technologies; Adjunct member of the Ebola National Task-Force of the Emergency Operations Centre-EOC; Speaker for the Café Scientifique Project, Uganda; Uganda and sub-Saharan Africa Representative on the Young and Early Career Investigators (YECI) working group for the Global HIV Vaccine Enterprise; Advised and directed the lay and scientific community about the scientific, ethical, legal and social implications of an HIV Vaccine; Served as the only Clinical Geneticist for Uganda; In-charge and Snr Consultant Geneticist for Unit of Genetics & Genomics (UGG), a super specialty reference centre for rare Mendellian disorders under the New Mulago National Referral Hospital; Proposed to the AESA/Bill and

Melinda Gates Grand Challenges Africa (2017), for establishment of a cohort of markers for R&D of a new-born screening DNA chip for the most frequent rare Mendellian diseases in Uganda; Diagnosed and or treated 156 patients with congenital abnormalities in the past year; Provided medical diagnosis and options for treatment to affected persons and/ or families; Reduced financial burdens by educating patients against witch-doctors; Counselled to empower patients to live harmoniously with their diagnosis; Developing local capacity and infrastructure for gene therapy for sickle cell anemia based on HIV-1 gene therapy model; Acquired updated advanced training materials for the curriculum from the Summit; Enrolled in an MSc Genetics & Genomics Program approved by the Univ.; Proposed establishment of a clinical and molecular genetics laboratory for patient care and, for research and training; Working with the Uganda Cancer Institute to develop a National Cancer Policy as mandated by WHO, and integrating aspects of genetics and genomic testing and counselling for individualized, stratified (on basis of ethnicity and tribe) population-wide cancer prevention, screening, diagnosis, and prognostic-follow-up. This could have a multiplier effect on local approach to other common complex diseases like diabetes mellitus, Hypertension etc.

24. **Yaylacıoğlu FT**. Received Fulbright scholarship for a Visiting researcher in the Department of Genetics in Yale Univ. between (Oct. 2017-Jun. 2018) under Dr. Kaya Bilguvar; Observership at Yale Medical School, Retina Clinic (Mar.- Jun. 2018) under Prof. Dr. Ron Adelman;
25. **Yousef YA.** Identified cases of Retinoblastoma (25); Referred patients (50) with familial and non-familial Retinoblastoma (genetic test positive), for counselling and management of condition; Informed patients negative for germline disease the extremely low risk of transmitting the disease to their off-springs.

2016 Batch

26. **Adeoye AM**. Elected as Fellows of American College of Cardiology and European Society of Cardiology (2018); Selected Emerging Leader in the field of cardiovascular medicine by World Heart Federation (2018); Participated actively in a Think Thank Seminar for emerging leaders at Duke Kunshan Univ. Kunshan, China (Jun. 2018); Awarded travel grant to present abstract titled "Apolipoproteins and Carotid Intima-Media Thickness among a Nigerian Hypertensive population-A community based study" at the International Society of Hypertension Conference (Beijing, China, Sept. 2018); Present an abstract "Prevalence And Clinical Correlates Of Blunted Heart Rate Dip In Chronic Kidney Disease: Findings From Ibadan Cardiovascular And Renal Event In People With Chronic Kidney Disease (Creckid) Study" at the World Cardiology Congress (Dubai, Dec. 2018).
27. **Adeyemo A**. On the steering team for commencement of Universal Newborn Hearing Screening, Univ. College Hospital, Ibadan.
28. **Adeyemo WL.** Delivered Inaugural Lecture as Professor on 'Genome Editing, Surgical Editing: A Surgeon-Scientist Narrative of Orofacial Cleft Research and Care', at the Univ. of Lagos (Oct. 2017); Coordinated a Genomics workshop, delivered 4 lectures (Introduction to Genomics, Genomics and Other Omics, Techniques and Tools in Genomics), to junior faculty at the College of Medicine Univ. of Lagos (Jul. 2018); Coordinated Bioinformatics workshop, delivered 2 lectures (Introduction to Genome Databases and SNPedia), for junior faculty at the College of Medicine Univ. of Lagos (Nov. 2017); Mentored 4 junior faculty, awarded Seed Grant for research projects in genomics, at the College of Medicine Univ. of Lagos.

29. **Ahmed DF**. Member in the Community Awareness Committee of Human Genetics and Genome Research Division at the National Research Center (estab. Jan. 2018), to raise awareness on genetic diseases in rural areas of Egyptian governorates according to a scheduled program to improve the quality of life; Selected as a Faculty in the Egyptian Committee of Pathology Training (ECPT) and lectured on the Basic Genetics held at The National Research Centre in collaboration with ECPT, Egypt (Mar. 2018); Presented at the10th conference of the African Society of Human Genetics under the theme "Human Genetics and Genomics in Africa", Egypt, (Nov. 2017); Presented poster at " Atypical Dementias; Diagnosis and Emerging therapies" Conference (Italy, Nov. 2017); Obtained a scholarship to attend the 13th Goldrain course on Clinical Cytogenetics (Italy, Aug. 2018).
30. **Ariani Y**. With the help of senior scientists and physicians proposed to start a registry for MPS type II and type IV; Helped establish the Indonesian MPS and Rare Disease foundation; Participated in crowdfunding campaign with Indonesia MPS and Rare Disease Foundation to help rare disease patients; Collaborated with Rainbow Foundation, Indonesia to evaluate the psychological impact of such patients on parents and family members; Helped provide biomolecular training for students from Biomolecular Division, Faculty of Mathematic and Natural Science.
31. **Belhassan K**. Completed training as guest researcher in Dr. Muenke's lab/NIH (Jun. 2018), planning to pursue collaboration and research with his lab team; Applied and admitted into an American Board of Medical Genetics and Genomics (ABMGG, USA) program, in Laboratory Genetics and Genomics (Jul. 2018 – Jun. 2020).
32. **Deniz E.** Established a new program 'Molecular and Translational Biomedicine' (Fall, 2018), for grad students at Dept. of Molecular Biology and Genetics, Acibadem Univ., Istanbul, Turkey; As a result of the Summit, supervise 3 grad students in genomics and genome editing projects.
33. **Ekure E**. Co-opted into Curriculum Review Committee for Faculty of Pediatrics, West African College of physicians. Responsible for reviewing genetics for Pediatric residency training (New curriculum published in 2017), introduced Genomics into the with Thompson and Thompson Genetics in Medicine as a recommended read; Selected as the Annual Gathering Speaker, for the Faculty of Clinical Sciences, College of Medicine, Univ. of Lagos- received Distinguished Speaker Award for the Faculty Lecture on Precision Medicine: Molecular Diagnosis and Digital Facial Analysis Technology Applications in Congenital Cardiovascular Disorders (Jul. 2018); Established, national coordinator for A National Pediatric Cardiac Disease Register for Congenital Heart Defects seen in 17 Tertiary centers across Nigeria; Established an electronic Pediatric echocardiography register; Saw 262 cases of congenital heart defects in children in the past year (Lagos Univ. Teaching Hospital, Nigeria), 43 were syndromic, 21 had identified genetic syndromes, Down syndrome (14)- commonest identified genetic syndrome; All cases were counselled; Obtained medical management for the required cases; Only 47 out of 231 had surgery due to funding constraints; Genetic counselling enabled provision of accurate prognostic information to families, facilitated early identification of important organ system involvement and appropriate referrals, and contributed to improved quality of life for patients and families.
34. **Lallar M**. Lectured on significance of genetics in modern medicine at the departmental and institutional level (post graduates, other specialists); Systemic data maintained (excel file) in OPD, for follow up, tracking and recalling of patients; Referred 400 patients in 6 months for genetic diagnosis and counseling; Routinely provided prenatal diagnosis for chromosomal

anomalies, Thalassemia, other single gene disorders; Hypertransfuion therapy, factor replacement, ERT etc., provided at SGPGIMS, Lucknow, India; Improvement in patients as measured and monitored by clinical outlook and blood tests; Routinely referring 3-4 patients/week with familial cancers for surveillance protocols; Prenatal diagnosis and extended family screening provided to all families.

35. **Malasa L**. Initiated project to create awareness among clinicians and policy makers on the role of genetics in disease causation (2017); Appointed as a Tutorial Assistant in the Department of Haematology and Blood Transfusion of Muhimbili Univ. of Health and Allied Sciences (May 2018); Attended the 3rd Sickle Cell Disease Ontology workshop, which aimed at finalizing and releasing the first version of Sickle Cell Disease Ontology (Cape Town, South Africa, Jun. 2018); Presented on Newborn Screening for Sickle Cell Disease in Tanzania at the Sixth MUHAS scientific conference held at Kisenga LAPF International Conference Centre (Dar es Salaam, Tanzania, Jun. 2018).
36. **Okafor F.** Participated in Nursing & Midwifery Council of Nigeria meeting with HODs of Nursing Departments in Nigerian Universities (Abuja, Jul. 2018); Participated in Workshop/Conference on Introduction to Monitoring, Evaluation and Learning, Assoc. of African Universities (Accra, Ghana, Aug. 2018); Proposed Ph.D. dissertation- Knowledge, belief and practice of reproductive health genetics and genomics of nursing institution lecturers in Edo State, Nigeria; Speaker at the Nigeria Ophthalmic Nurses Association Annual National Conference 'Prevention of Blindness in Nigeria, the way forward' at Lagos Univ. Teaching Hospital, Lagos, Nigeria (Oct. 2018).
37. **Roblejo Balbuena H**. Promoted to Auxillary Professor and Auxillary Researcher; Based on learning from the Summit, updated curriculum in clinical genetics for residents, genetic counselors and professional associated with medical genetics; Lectured on advances in Medical Genetics across Cuba; Lectured on genetic technologies prior to International Congress of Community Genetics (Havana, Cuba, Nov. 2017); Lectured on Genetic Cardiopathies and Heart Failure at the XXX Central American and Caribbean Congress of Cardiology and IX Cuban Congress of Cardiology (Havana, Cuba, Jun. 2018); Trained in pharmacogenetics in Clinical Research Center, Univ. Hospital Infanta Cristina en Badajoz, Spain (Jul. 2018); Evaluated ~113 genetic cases of monogenic, chromosomal or multifactorial etiology; Registered 56 new cases in the Genetic Diseases' Register of the Pediatric Hospital Center Havana in this period; Improved genetic counselling skills.
38. **Seven M.** Developed a course 'Introduction to Genetics for nurses' for undergraduate nursing program; Invited Speaker, Genetic Counselling In Breast Cancer at BREASTANBUL Conference (İstanbul, Turkey, Oct. 2018); Invited Speaker, Risk factors in gynecologic cancer and genetics, National Gynecologic Oncology Cngress (Antalya, Turkey, Nov. 2018).
39. **Sirisena ND.** Received a training scholarship from the Overseas Special Training Programme of the National Science Foundation, Sri Lanka to attend the Genetics, Molecular and Cell Biology Short-term Training Fellowship (Laboratory of Molecular Biology, Univ. of Verona, Italy, May 2018), and obtained hands-on laboratory training on gene editing using Crispr/Cas9 experiments; Received the Daphne Attygalle Award for Best Paper in Cancer, for the poster presentation titled "Implementation of Multi-gene Panel Testing for Hereditary Cancer Predisposition in Sri Lanka: Initial Experiences", at the Sri Lanka Medical Association 130th Anniversary International Medical Congress (2017); Presented a paper on "Genetic Determinants of Sporadic Breast Cancer in Sri Lankan Women" at the Seminar on Epigenetics organized by the International Council for Science, Regional Office for Asia and Pacific in

collaboration with the Human Genetics Unit, Faculty of Medicine, Univ. of Colombo, Sri Lanka (Nov. 2017); Poster presentation on "Pattern of germline genetic variants in cancer predisposing genes in a Sri Lankan cohort with inherited cancer syndromes", at 3rd Variant Effect Prediction Training Course organized by the Human Variome Project (New Castle Univ. Medicine Malaysia, Aug. 2018); Strengthened the healthcare system in clinical genetic services, at peripheral centers, through trained MSc graduates (6); 600 patients with various genetic diseases and congenital birth defects were offered genetic counseling and diagnostic services at the Human Genetics Unit, referred for appropriate supportive therapies (physiotherapy, speech therapy, occupational therapy and specialized medical management) for improved quality of life;

40. **Tibrewal S**. In-charge of Genetic Clinic at Dr. Shroff Charity Eye Hospital, Delhi; Provided genetic counselling, referred patients with ocular hereditary disorders for genetic tests, maintained records in databases; Hold pedigree data on patients- 32 pediatric cataract, 59 retinitis pigmentosa, 30 high myopia, 33 glaucoma, 5 high hyperopia families, 36 other retinal dystrophy, 40 anterior segment disorders (total 230); Collected blood samples for genetic analysis of cases presently underway at Guru Nanak Dev Univ., India.
41. **Uwineza A.** Lectured Medical Genetics and Genomics in General Medicine and graduate school (MMED, pathology/pediatrics and Gynecology and obstetrics) curriculum; Established the Rwandan Down Syndrome Organization, included children and families with Down Syndrome; Creation of Rwanda Society of Human Genetics, Vice Chairperson; Member of the organizing committee of the 11th Conference of African Society of Human Genetics and 12th H3Africa Consortium (Kigali-Rwanda Sept. 2018); Supervisor, birth defect registry in Rwanda, developed an initial "Core Outcome Set" for testing in clinical practice; Started a pilot program in newborn screening in Rwanda (congenital hypothyroidism and congenital adrenal hyperplasia) to identify major challenges; Out-patient consultations done weekly in Kigali Referral Teaching Hospital, ~560 patients referred for consultation to Human Genetics Center, abnormal karyotypes found in 135 patients, identified genetic conditions - Down Syndrome (135), Patau syndrome (8), Edwards syndrome (12), Turner syndrome (5), Klinefelter syndrome (1), Unbalanced translocation (9), Unidentified marker (1); Improved quality of life for patients with Sickle cell Disease and Osteogenesis imperfecta through appropriate management of condition.