ADDENDUM

ISHGG THIRD ANNUAL OUTCOME (October 2018- September 2019)

(To avoid a lengthy report, we have tried to mention a project under one category only eg. a publication can also be a grant, a collaboration and an on-going project, however, may not appear in all three categories).

A. Publications: (Total- 177; 2018- 59; 2017- 62; 2016- 56)

2018 Batch

1. Adetiloye A, Erhabor GE, Awopeju O, **Adewole OO**, Onini E, Adewuya O. Challenges of diagnosing and managing bronchiectasis in resource - limited settings: a case study. Pan Afr Med J. 2019; 32:82.
2. Adetiloye A, Erhabor GE, Obaseki DO, Awopeju OF, **Adewole OO**. Impact of Sleep Quality on the Health-Related Quality of Life of Patients with Chronic Obstructive Pulmonary Disease.West Afr. 2018; 35(3):173-179.
3. **Adeyemo TA**, Diaku-Akinwunmi IN, Ojewunmi OO, et al. Barriers t**o** the Use of Hydroxyurea in the Management of Sickle Cell Disease in Nigeria. Hemoglobin. 2019. (Accepted).
4. **Adeyemo TA**, Ojewunmi OO, Oyetunji A, et al. Co-inheritance of alpha thalassemia and BCL11A in Nigerian patients with Sickle Cell Anaemia. 2019. (In preparation).
5. **Adeyemo TA**, Olude E, Bello F, et al. Frequency of Intron 1 and 22 inversion and the clinical characterization of Haemophilia A in Nigeria. 2019. (In preparation).
6. Alva-Diaz C, Alarcon-Ruiz C, Pacheco-Barrios K, Mori N, Traynor BJ, Rivera-Valdivia A, **Lertwilaiwittaya P**, Bird TB, **Cornejo-Olivas M**. Frequency of C9orf72 hexanucleotide repeat expansion in patients with Huntington-like disorders, a systematic review and meta-analysis. Neurology. 2019. (Submitted).
7. Ben Haj Ali A, Amouri A, Sayeb M, Makni S, Hammami W, Naouali C, Dallali H, Romdhane L, Bashamboo A, McElreavey K, Abdelhak S, **Messaoud O**. Cytogenetic and molecular diagnosis of Fanconi anemia revealed two hidden phenotypes: Disorder of sex development and cerebro-oculo-facio-skeletal syndrome. Mol Genet Genomic Med. 2019. 7:e00694.
8. Chikhaoui A, Elouej S, Nabouli I, Jones M, Lagarde A, Ben Rekaya M, **Messaoud O**, Hamdi Y, Zghal M, Delague V, et al. Identification of a ERCC5 c.2333T>C (L778P) Variant in Two Tunisian Siblings With Mild Xeroderma Pigmentosum Phenotype. Front Genet. 2019; 10: 111.
9. Choudhury J, **Dutta AK**. A family of Charcot Marie Tooth Disease with spastic quadriplegia, epilepsy, global developmental delay. Ann Indian Acad of Neurol. 2019. (In review).
10. **Cornejo-Olivas M**, Inca-Martinez M, Machado Castilhos R, Furtado G et al. Hereditary ataxias in Peru: genetic analysis of familial and sporadic cases. Cerebellum. 2019. (Submitted).
11. Cristian S, Wiafe-Addai B, Anorlu RI, **Abdulkareem FB**, Asombang A, Browne D, Makau-Barasa LK, et al. Women in global Oncology forum: Expanding the Global oncology workforce. Chapter NCCP, AORTIC. 2019. (Submitted).
12. **Das S**, Danda S. A case series of pycnodysostosis in Indian patients. 2019. (In preparation).

13. **Das S**, Thomas M, Yoganathan S, Srivastava VM, et al. A descriptive study with genetic analysis in patients of ataxia telangiectasia (AT) from the Indian subcontinent. 2019. (In preparation).
14. **Dueñas-Roque M**, Kruszka P, Muenke M. Turner syndrome in diverse populations. 2019. (In preparation).
15. **Dutta AK**. Schuurs-Hoeijmakers syndrome in a patient from India. Am J Med Genet Part A. 2019; 179A:522–524.
16. **Dutta AK**. Photo-Quiz 45. Genetic Clinics. 2019; 12(3). http://iamg.in/genetic_clinics/photoquiz_link/ph_45/quiz.html.
17. **Dwivedi A**, Gupta S, Saxena D. Association of KDR gene Polymorphisms and Idiopathic Recurrent Pregnancy Loss in Indian Women. J Genet. 2019. (In review).
18. **Dwivedi A**, Mandal K, Moirangthem A, Pandey H, et al. Yield of molecular testing in VHL and isolated VHL associated tumours in an Indian cohort. South Asian J Cancer. 2019. (Accepted).
19. **Dwivedi A**, Srivastava P. Xq28 duplication involving FAM58A gene; facial features resembling STAR syndrome – a case report. 2019. (In preparation).
20. **Eshete MA**, Butali A, Abate F, et al. The Role of Environmental Factors in the Etiology of Nonsyndromic Orofacial Clefts in the Ethiopian Population. J Craniofac Surg. 2019. (Accepted).
21. Figueroa-Ildefonso E, Bademci G, Rajabli F, **Cornejo-Olivas M**, Villanueva RDC, Badillo-Carrillo R, Inca-Martinez M, et al. Identification of Main Genetic Causes Responsible for Non-Syndromic Hearing Loss in a Peruvian Population. Genes. 2019; 10:581.
22. Gelanew M, Fikre A, Abiye H, Yohannes D, **Eshete MA**. The Spectrum of Reconstructive Surgery at Yekatit 12 Hospital Medical College Plastic and Reconstructive Surgery Unit (Y12HMCPRSU). East and Central African Journal of Surgery. 2019. (In review).
23. Gonzales-Saenz C, Cruz-Rodriguez C, Espinoza-Huertas K, Véliz-Otani D, Marca V, Ortega O, Milla-Neyra K, Alvarez-Tejada J, Mazzetti P, **Cornejo-Olivas M**. Distribution of the CAG triplet repeat in ATXN1, ATXN3 and CACNA1A loci in Peruvian population. Cerebellum. 2019. (Submitted).
24. Hamdi Y, Ben Rekaya M, Jingxuan S, Nagara M, **Messaoud O**, Benammar EA, Mrad R, et al. A genome wide SNP genotyping study in the Tunisian population: specific reporting on a subset of common breast cancer risk loci. BMC Cancer. 2018; 18:1295.
25. John DO, Tella BA, Olawale OA, John JN, **Adeyemo TA**, et al. Effects of a 6-week aerobic exercise programme on the cardiovascular parameters, body composition, and quality of life of people living with human immune virus. J Exerc Rehabil. 2018 Oct 31; 14(5):891-898.
26. Kosiyo P, Otieno W, **Gitaka J**, Nyamuni J, Collins Ouma. Correlation between haematological parameters and sickle cell genotypes in children with P. falciparum malaria. 2019. (In preparation).
27. Lawal AO, Phillips AA, Orah NO, Daramola AO, **Abdulkareem FB**. Invasive cribriform carcinoma of the male breast. Ann Trop Pathol 2019; 10:83-85.
28. Mathew M, Tinuola FR, Goel SR, **Taiwo O**, Jalo P. Prevalence of hepatitis B among school adolescents in Jos, Plateau State Nigeria. Texila International Journal of Public Health. 2018; 6 (4).
29. **Mburu S**. Precision medicine strategy in lymphoid tissue neoplasms; The future is here and it's working. 2019. (In preparation).

30. Mellado C, Pardo RA, López-Camelo J, **Nakousi-Capurro N**, Salazar L, Vilca M. Prevalence of red blood cell folate insufficiency among non-pregnant Chilean women of child bearing age. 2019. (In preparation).
31. **Muttamba W**, Kirenga B, Ssengoba W, Katamba A, et al. Prevalence of tuberculosis risk factors among bacteriologically negative and bacteriologically confirmed tuberculosis patients from five regional referral hospitals in Uganda. Am J Trop Med Hyg. 2019; 100 (2): 386-391.
32. **Muttamba W**, Ssengooba W, Kirenga B, Sekibira R, et al. Health seeking behavior among individuals presenting with chronic cough at referral hospitals in Uganda; Missed opportunity for early tuberculosis diagnosis. PLoS ONE. 2019; 14(6):e0217900.
33. Naifar M, Kallel F, HadjKacem F, Boudabous H, Kallel R, Boudawara T, **Messaoud O**, Tbib N, Charfi N, Abid M, et al. Homozygous pArg610del Mutation Unusually Associated With Severe Delay of Growth in 2 Acid Sphingomyelinase Deficiency-affected Sibs. J Pediatr Hematol Oncol. 2019. (In press).
34. Najjingo I, **Muttamba W**, Kirenga BJ, Nalunjogi J, Bakesiima R, et al. Comparison of GeneXpert cycle threshold values with smear microscopy and culture as a measure of mycobacterial burden in five regional referral hospitals of Uganda- A cross-sectional study. PLoS ONE. 2019 May 15. https://doi.org/10.1371/journal.pone.0216901.
35. **Nakousi-Capurro N**, Alegría A, Cares C, Gainza-Lein M, et al. Congenital anomalies and Comorbidities in Down Syndrome Neonates born at Clínica Dávila in Santiago, Chile: 2008-2018. 2019. (In preparation).
36. Ntiamoah P, Monu N, **Abdulkareem FB**, Adeniji KA, Obafunwa JO, Alatise OI, et al. Pathology services in Nigeria: cross-sectional survey results from three Cancer consortia. J Glob Oncol. 2019. (Submitted).
37. Obiajulu FJ, Badmos KB, Awolola NA, Dawodu OO, Adebayo LA, Elesha SO, **Abdulkareem FB**. Autopsy Study of the Liver in Adult Nigerians at Lagos University Teaching Hospital.West Afr J Med. 2019 Jan-Apr; 36(1):18-24.
38. Offor E, **Abdulkareem FB**. Screening endoscopy in Portharcourt, Nigeria. Gastroenterology Insights 2019; 10:7987.
39. Oguntunde OA, Awolola NA, Obiajulu FJN, Daramola ES, **Abdulkareem FB**, Banjo AAF, Baljeet K. The role of p57 in the diagnosis of hydatidiform mole in a resource limited setting Niger Postgrad Med J. 2019. (Submitted).
40. Ojewunmi OO, **Adeyemo TA,** Ayinde OC, Iwalokun B, et al. Current perspectives of sickle cell disease in Nigeria: Changing the narratives. Expert Rev Hematol. 2019; 12 (8): 609-620.
41. **Olojede OC**, Adamson OO, Gbotolorun OM, Emmanuel MM, et al. Surgical management of maxillary sinus tumours in a secondary healthcare facility in Nigeria. J Clin Sci. 2018; 15 (3):140-144.
42. Oseni GO, Jain D, Mossey PA, Busch TD, Gowans LJJ, **Eshete MA**, **Adeyemo WL**, et al. Identification of paternal uniparental disomy on chromosome 22 and a de novo deletion on chromosome 18 in individuals with orofacial clefts. Mol Genet Genomic Med. 2018; 6:924-932.
43. **Otarru S**, **Mgasa A**, **Malasa L**. Genetic and Genomic Medicine in Tanzania: Challenges and Opportunities for Future. Mol Genet Genomic Med. 2019. (Submitted).
44. Rico EMG, Kikuchi A, Saito T, Kumondai M, Hishinuma E, Kaneko A, Chan CW, **Gitaka J**, Oda A, Saito S, Hirasaw N. CYP2D6 genotyping analysis and functional characterization of novel allelic variants in a Ni-Vanuatu and Kenyan population by assessing

dextromethorphan O-demethylation activity. Drug Metab Pharmacokinet. 2019 July 20. https://doi.org/10.1016/j.dmpk.2019.07.003.
45. Romdhane L, **Messaoud O**, Kefi R, Tiar A, Amouri A, et al. The Genetic Epidemiology of Orphan Diseases in North Africa. In: Muntaser Ibrahim and Charles Rotimi ed. The Genetics of African Populations in Health and Disease. ISBN: 9781107072022. 2019 (In press).
46. **Savina O**, Rykov S. Clinical and epidemiological peculiarities of congenital and inherited eye pathologies in children. Archive of the Ukrainian ophthalmology. 2019; 1(7).
47. **Savina O**, Rykov S. Peculiarities of visual organs' conditions in students with genetically determined eye pathology. Theory and practice of tiflopedagogics. Information bulletin of the All-Ukrainian NGO Association of tiflopedagogues of Ukraine. 2019; 18-19:31-33.
48. Sayeb M, Riahi Z, Laroussi N, Bonnet C, Romdhane L, Mkaouar R, Zaouak A, Marrakchi J, Abdessalem G, **Messaoud O**, Bouchniba O, Ghilane N, Mokni M, et al. A Tunisian family with a novel mutation in the gene CYP4F22 for lamellar ichthyosis and co-occurrence of hearing loss in a child due to mutation in the SLC26A4 gene. Int J Dermatol. 2019. (In press).
49. Sheth J, Bhavsar R, **Mistri M**, Pancholi D, Bavdekar A, Dalal A, et al. Gaucher disease: single gene molecular characterization of one-hundred Indian patients reveals novel variants and the most prevalent mutation. BMC Medical Genetics. 2019; 20(1):31.
50. Sheth J, **Mistri M**, Bhavsar R, Pancholi D, Kamate M, et al. Batten disease: Biochemical and Molecular characterization revealing novel PPT1 and TPP1 gene mutations in Indian patients. BMC Neurology. 2018; 18(1):203.
51. Sheth J, Pancholi D, **Mistri M**, Nath P, Ankleshwaria A, Bhavsar R, et al. Biochemical and molecular characterization of adult patients with type I Gaucher disease and carrier frequency analysis of Leu444Pro - a common Gaucher disease mutation in India. BMC Medical Genetics. 2018; 19:178.
52. Suárez C, Aranibar L, **Nakousi-Capurro N**, **Díaz C**, **Pizarro P**. Pigmentary mosaicism in a pediatric Chilean cohort: Cytogenetic characterization, cutaneous and extracutaneous involvement. Actas Dermosifilográficas. 2019. (Submitted).
53. **Utumatwishima JN**, Umukunzi L, Tayebwa E. Compliance to maternal care guidelines is high in patients with complications compared to patients without complications. The tip of an iceberg? A retrospective case-control study. Rwanda Med J. 2019. (Accepted).
54. Véliz-Otani D, Inca-Martinez M, Bampi GB, Ortega O, Jardim LB, Saraiva-Pereira ML, Mazzetti P, **Cornejo-Olivas M**. ATXN10 Microsatellite Distribution in a Peruvian Amerindian Population. Cerebellum. 2019 Jul 24. doi: 10.1007/s12311-019-01057-x.
55. Velez-Pardo C, Lorenzo-Betancor O, Jimenez-Del-Rio M, Moreno S, Lopera F, **Cornejo-Olivas M,** Torres L, Inca-Martinez M, Mazzetti P, et al. The distribution and risk effect of GBA variants in a large cohort of PD patients from Colombia and Peru. Parkinsonism Relat Disord. 2019; 63:204-208.
56. Vishnevetsky A, **Cornejo-Olivas M**. Juvenile Huntington Disease in Peru: Review of 32 Patients and Challenges of Classification. Mov. Disord. 2019. (Submitted).
57. Vishnevetsky A, Zapata Del Mar C, Luis Cam J, **Cornejo-Olivas M**, Creutzfeldt CJ. Palliative Care: Perceptions, Experiences, and Attitudes in a Peruvian Neurologic Hospital. J Palliat Med. 2019; 22(3):250-257.
58. **Wangi KYW**, Sakinah I, Ningsih D.A, Vanawati N, Adiningsih D, Simatupang E.J. Nursing Genetics and Genomics Education in Indonesia: from Philosophy to Policy. Journal of Ethics and Education. 2019. (In review).

59. Zaouak A, Abdessalem G, Mkaouar R, **Messaoud O**, Abdelhak S, Hammami H, Fenniche S. Congenital lamellar ichthyosis in Tunisia associated with vitamin D rickets caused by a founder nonsense mutation in the TGM1 gene. Int J Dermatol. 2019. (In press).

2017 Batch

60. **Abad PJ**, Lee JMH, Kejriwal S, Chu YWY, et al. Who is a Genetic Counselor? Perspective of Asian Genetic Counselors on their Roles and Added Value. J Community Genet. 2019. (In review).
61. **Abad PJ**, Lee JMH, Faradz S, Thuy Doung HH, et al. Genetic Counseling in South East Asia: Opportunities, Challenges, and Future Directions. 2019. (In preparation).
62. **Abad PJ**, Sibulo MSK, Sur ALD. The Newborn Screening Program as an Opportunity to Integrate Genetics in Nursing Practice. Am J Nurs. 2019. (In review).
63. Abdel-Hamid MS, Ismail S, Zaki MS, Abdel-Salam GM, **Otaify GA**, et al. GAPO syndrome in seven new patients: Identification of five novel ANTXR1 mutations including the first large intragenic deletion. Am J Med Genet. Part A. 2019; 179:237-242.
64. Afifi H, Abdel-Hamid M, **Mehrez M**, El-Kamah G, Abdel-Salam G. Lenz-Majewski Syndrome in a Patient from Egypt. Am J Med Genet. 2019. (Submitted).
65. Ahmad S, **Fatima SS**, Rukh G, Smith GE. Gene Lifestyle interactions with relation to Obesity, Cardiometabolic and Cardiovascular Traits among South Asians. Front. Endocrinol. 2019; 10:221.
66. Ali SH, Ali M, Farhat S, **Fatima SS.** Suppressor of cytokine signaling-3 in pregnant females with or without hypertension. Pak J Med Assoc. 2019. (Accepted).
67. Alves G, Reis MGC, **Casado PL,** Brochado AB, Granjeiro JM, Barboza ESP. Evaluation of the receptor activator of nuclear factor-K ligand/osteoprotegerin ratio in the gingival crevicular fluid of patients under periodontal maintenance. Int J Growth Factors Stem Cells Dent. 2019; 2 (1):3-7.
68. Amr K, El-Bassyouni H, Abdel-Hady S, Mostafa M I, **Mehrez MI**, El-Kamah G. Pycnodysostosis: Three Novel Mutations and an Unusual Presentation. 2019. (In preparation).
69. **Avogbe PH**, Manel A, Vian E, Durand G, et al. Urinary TERT promoter mutations as non-invasive biomarkers for the comprehensive detection of urothelial cancer. EBioMedicine. 2019; 6 (44):431-438.
70. Babirye P, Musubika C, Kirimunda S, Downing R, Lutwama JJ, Mbidde EK, Weyer J, Paweska JT, Joloba ML, **Wayengera M.** Identity and validity of conserved B cell epitopes of filovirus glycoprotein: towards rapid diagnostic testing for Ebola and possibly Marburg virus disease. BMC Infect Dis. 2018;18(1):498.
71. Besio R, Garibaldi N, Leoni L, Cipolla L, Sabbioneda S, Biggiogera M, Mottes M, Aglan M, **Otaify GA**, Temtamy SA, Rossi A. Cellular stress due to impairment of collagen prolyl hydroxylation complex is rescued by the chaperone 4-phenylbutyrate. Dis Model Mech. 2019 Jun 1; 12(6):dmm038521.
72. Calandra CR, Mocarbel Y, **Vishnopolska SA**, Toneguzzo V, Oliveri J, Cazado EC, et al. Gordon Holmes Syndrome caused by RNF216 novel mutation in 2 Argentinean siblings. Movement Disorders Clinical Practice. 2019; 6(3):259-262.
73. **Casado PL**, Aguiar T, Pinheiro MPF, Machado A, et al. Smoking as a Risk Factor for the Development of Periimplant Diseases. Implant Dent. 2019; 28:120-124.

74. **Casado PL**, Tesch R, Quinelato V, Cordeiro PCF, et al. Polymorphisms in COMT, ADRB2 and HTR1A genes are associated with temporomandibular disorders in individuals with other arthralgias. Cranio. 2019; 2:1-11.
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76. Cordeiro PCF, Bonato LL, Quinelato V, **Casado PL**. Bruxismo: A Genetic Approac**h**. Faculdade de odontologia de Lins. 2018; 28:35-61.
77. Delhomme TM, **Avogbe PH**, Gabriel A, Alcala N, Leblay N, et al. Needlestack: an ultra-sensitive variant caller for multi-sample next generation sequencing data. Nucleic Acids Res. 2019. (Submitted).
78. **Dhoro M**. Impact of HLA/KIR genotypes on HIV disease progression in Africans- A focus on perinatally infected children. Cent Afr J Med. 2019. (Submitted).
79. **Dhoro M**, Zaranyika T. A retrospective data review of Diabetic ketoacidosis at Parirenyatwa group of Hospitals. Cent Afr J Med. 2018; 64. (Letter to the editor).
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83. Fatima SK, Akhtar A, Khan AR, **Fatima SS**. Distribution and Determinants of Sedentary Lifestyle among Health Care Professionals PJMD. 2019; 8 (02):80-86.
84. Galaviz KI, Narayan KMV, Manders OC, **Torres-Mejía G**, Goenka S, McFarland DA, Reddy KS, et al. The Public Health Leadership and Implementation Academy for Noncommunicable Diseases. Prev Chronic Dis. 2019 Apr 18;1 6:E49.
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87. Hasanreisoğlu Murat, Özdemir HB, **Tuncay FY**, Ertop M, Aktaş Z. Bilateral Anterior Uveitis Revealing Relapsing Polychondritis (Case Report). Turk Oftalmoloji Gazetesi 2019; 49(2):99-101.
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89. **Hitayezu J**, **Uwineza A**, Mukamugema B, et al. Neonatal Screening in Rwanda: A pilot assay for testing congenital hypothyroidism and congenital adrenal hyperplasia. 2019. (In preparation).
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97. **Lotz-Esquivel S**, Acuña-Feoli J, Alvarado-Aguilar M, Penon-Portman M, et al. Delivery of Genetic Services in Costa Rica: Challenges and Opportunities. 2019. (In preparation).
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102. Nunes CHR, Campello M, Pinheiro AR, Cordeiro Filho C, Rangel BC, Villas-Boas R, Mourao CFAB, **Casado PL**. Prosthetic Rehabilitation in a Child with Hereditary Ectodermal Dysplasia. Int Arch BioMed Clin Res. 2019; 5:57-60.

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115. **Thakur N**, Abeysekera G, Wanigasinghe JV, Dissanayake HW. The spectrum of deletions and duplications in the Dystrophin (DMD) gene in a cohort of patients with Duchenne Muscular Dystrophy (DMD) in Sri Lanka. Neurol India. 2019; 67 (3): 714-715.
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117. **Tuncay FY**, Dincer PR. Could autosomal dominant TGFBI-related corneal dystrophies be modelled in zebrafish by using CRISPR/Cas9: Challenges and Possibilities. W J Opthalmol & Vision Res. 2019; 1(3). doi: 10.33552/WJOVR.2019.01.000513.
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119. Ventura-Alfaro CE, Ávila-Burgos L, **Torres-Mejía G**. Adherence of Mexican physicians to clinical guidelines in the management of breast cancer: Effect of the National Catastrophic Health Expenditure Fund. PLoS One. 2019 Mar 20; 14(3):e0212841.
120. Yildiz BK, Aktas Z, Yuksel N, Ozdemir HB, Cingirt M, Gulbahar O, **Tuncay FY**, Hasanreisoglu M. Evaluation of renin and angiotensin II levels in pseudoexfoliation syndrome. Arq Bras Oftalmol. 2018; 81(6):494-499.
121. **Yousef A**, AlNawaiseh T, AlJabari R, Muhsen S, Al-Nawaiseh I. Retinoblastoma awareness among first contact physicians in Jordan. Ophthalmic Genet. 2019; May 28:1-5.

2016 Batch

122. **Adeoye AM**, Adebayo O, Abiola O, Iwalokun B, et al. Association between Selected Molecular BiOmarkers and AmbuLatory Blood Pressure Pattern in African Chronic Kidney Disease and Hypertensive Patients compared with normotensIve Controls: The SyMBOLIC Study Protocol. JIMR Res Protoc. 2019. (In review).
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125. Ajayi SO, **Adeoye AM**, Raji YR, Tayo B, Salako BL, et al. Self-reported Sleep Disorder and Ambulatory Blood Pressure Phenotypes in Patients with or without Chronic Kidney Disease: Findings from Ibadan CRECKID Study. West Afr J Med. 2019; 36(1):61-68.
126. Akpalu A, Gebregziabher M, Ovbiagele B, Sarfo F, Iheonye H, Akinyemi R, Akpa O, Tiwari HK, Arnett D, Wahab K, Lackland D, **Adeoye AM**, et al. Differential Impact of Risk Factors on Stroke Occurrence among Men vs. Women in West Africa: the SIREN Study. Stroke. 2019; 50(4):820-827.
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129. **Ariani Y**, Priambodo R, Sjarif DR. Mutation Analysis of Exon 8 of the Iduronate-2-Sulfatase Gene in Mucopolysaccharidosis Type II Patients in Indonesia. J Nat Sci Biol Med. 2019. (Accepted).
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Onwuamah C, Oseni GO, Owais A, **James O**, et al. Genomic analyses in African populations identify loci for cleft palate. Human Mol Genet. 2019; 28:1038-1051.
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142. **Hussen DF**, Hussein AA, Monzer MA, Hammad SA. Combined markers for preclinical identification of Alzheimer's disease. Psychogeriatric. 2019. (In review).
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dosage in GATA3: A case report and review of literature. BMC Endocr Disord. 2019. (In review).

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149. **Lallar M**, Arora V, Kulshreshtra S, Saxena R, et al. Novel PRUNE gene mutations in three Indian families. 2019. (In preparation).
150. **Lallar M**, Arora V, Saxena R, Puri R, et al. Complete labyrinthine aplasia: a unique sign for targeted genetic testing in hearing loss. Int J Pediatr Otorhinolaryngol. 2019. (In review).
151. **Lallar M**, Phadke S. Exome sequencing: Knowledge, attitude and perspectives of non-genetic clinicians of a developing country. J. Genet Couns. 2019. (In review).
152. **Lallar M**, Srivastava P, Rai A, Saxena D, et al. Cytogenetic microarray in structurally normal and abnormal fetuses: a five-year experience elucidating increasing acceptance and clinical utility. J. Genet. 2019; 98:6.
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154. Levano K, Jaramillo-Valverde L, Delgado E, **Guio H**. Gelatin-coated magnetic nanoparticles-based DNA isolation method: A comparison with commercial DNA isolation kits from whole blood. Journal of Bioscience and Applied Research, 2019; 5 (1):136 -140.
155. Levano K, Jaramillo L, Valdivia J, **Guio H**. Macrophages modification by CXCR4 genomic editing and effect on metastatic breast cancer. 2019. (In preparation).
156. Mohamed AM, **Hussen DF**, Goma HM. The Micronuclei scoring as a biomarker for early detection of genotoxic effect of cigarette smoking. 2019. (In preparation).
157. 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. F1000 Research. 2019. (In review).
158. Oladega AA, **James O**, **Adeyemo WL**. Cyanoacrylate tissue adhesive or silk suture for closure of surgical wound following removal of an impacted mandibular third molar: A randomized controlled study. J Craniomaxillofac Surg. 2019; 47:93-98.
159. Paththinige CS, **Sirisena ND**, Escande F, Manouvrier S, Petit F, et al. Split hand/foot malformation with long bone deficiency associated with BHLHA9 gene duplication: a case report and review of literature. BMC Med Genet. 2019; 20:108.
160. Paththinige CS, **Sirisena ND**, Kariyawasam UGIU, Dissanayake VHW. The Frequency and Spectrum of Chromosomal Translocations in a Cohort of Sri Lankans. BioMed Res Int. 2019 Apr 2: 9797014.
161. Ratna Ria, **Tibrewal S**. The science and art of Pedigree charting. Indian J Ophthalmol. 2019. (Submitted).
162. **Roblejo BH**, Marcheco Teruel B, González Pal S, Bordado T, et al. CYP2D6 genetic polymorphism and genetic ancestry on extrapyramidal side-effects of long-term treatment with classical antipsychotics in Cuban patients with schizophrenia. Clin. Invest. 2018; 9 (1): 1-3.
163. Sahin E. Pasalak SI, **Seven M**. Consanguineous marriage and its effect on reproductive behaviour and usage of prenatal screening. J Genet Couns. 2019. (In review).
164. **Seven M**, Paşalak ŞI, Şahin E. Akyüz A. Genetic Literacy of pregnant women and their use of prenatal screening and diagnostic tests in Turkey. J. Genet Couns. 2019; 28 (3):578-586.

165. **Sirisena ND**, Biswas K, Stauffer S, Cleveland L, et al. Mouse embryonic stem cell-based functional analysis of five unclassified BRCA2 variants identifies one deleterious and four neutral variants. Hum Genet. 2019. (Submitted).
166. **Sirisena ND**, Dissanayake VHW. Challenges of Genomic Medicine Education in Low- and Middle-Income Countries. Front Genet. 2019. (In review).
167. **Sirisena ND**, Dissanayake VHW. Genetics and genomic medicine in Sri Lanka. Mol Genet Genomic Med. 2019 Jun; 7(6):e744.
168. **Sirisena ND**, Samaranayake UMJE, Abath Neto OL, Foley AR, et al. A novel variant in the COL6A1 gene causing Ullrich congenital muscular dystrophy in a consanguineous family. Neuromuscul Disord. 2019. (Submitted).
169. **Sirisena ND**, Samaranayake N, Dissanayake VHW. Electrophoretic mobility shift assays implicate *XRCC2*:rs3218550C>T as a potential low-penetrant susceptibility allele for sporadic breast cancer. BMC Res Notes. 2019. (In review).
170. **Sirisena ND**, Samaranayake N, Dissanayake VHW. Genotype data for single nucleotide polymorphism markers in sporadic breast cancer related genes in a Sri Lankan case–control cohort of postmenopausal women. BMC Res Notes. 2019. (Accepted).
171. **Tibrewal S**, Khurana R, Venkatesh R, Ganesh S. Subconjunctival silicone oil complicating strabismus surgery. GMS Ophthalmol Cases. 2019; 9: Doc12.
172. **Tibrewal S**, Nguyen PTT, Ganesh S, Molinari A. Bilateral Symmetric and Asymmetric Superior Rectus Recession for Patients with Dissociated Vertical Deviation. Asia Pac J Ophthalmol (Phila). 2019; 8(3):218-223.
173. **Tibrewal S**, Subedar K, Sen P, Mohan A, et al. Clinical features of MAC group of disorders in North India – A multicentric study. 2019. (In preparation).
174. Udenze IC, Taiwo IA, Minari JB, **Adeyemo WL**. Insulin resistance and cardiovascular risk in older adult Nigerians with type 2 diabetes. Int J Noncommun Dis. 2019; 4: 21-26.
175. **Uwineza A**, Caberg JH, **Hitayezu J**, Wenric S, Mutesa L, Vial Y, et al. VPS51 biallelic variants cause microcephaly with brain malformations: A confirmatory report. Eur. J. Med. Genet. 2019; 6:103704.
176. Valdivia-Silva J, Perez L, Flores L, Malaga M, **Guio H**. Development of an accessible and low-cost microfluidic (Lab-on-a-chip) system for the detection of circulating tumor cells in rural regions. Acta Medica Peruana. 2019. (In review).
177. Yareta J, Galarza M, Capristano S, Pellón O, Sánchez C, Ballon C, **Guio H**. Differential expression analysis of circulating microRNAs in patients with active tuberculosis and infected with latent tuberculosis. Rev Peru Med Exp Salud Publica. 2019. (Accepted).

B. Grants: (Total- 53; 2018- 21; 2017- 17; 2016- 15)

2018 Batch

1. **Abdulkareem FB**, Badmos KB. CDKN3 mRNA and Immunohistochemistry Expression of Preneoplastic and Invasive Cervical Cancer in LUTH. 2019. (NIH-U54 Seed Grant; Submitted).
2. **Adewole OO** (PI), Adewole TO (Co-PI). Innovative Approaches to improving TB case detection Grant Challenges Africa Phase II. 2019. (African Academy of Science; Submitted).
3. **Adewole OO** (PI), Adewole TO (Co-PI), Ayodele O (Co-PI). Wearable sensor to monitor response and adherence to anti-TB drugs. 2019. (Grand Challenges Exploration; Submitted).

4. Adisa RA, **Abdulkareem FB** (Co-I). Quantification of free circulating mitochondrial DNA content and detection of alterations in the plasma in HIV-associated HCC or Cervical cancer.2 019. (NIH-U54 Seed Grant; Received).
5. Badmos KB, **Abdulkareem FB** (Co-I). Immunohistochemistry expressions of CTLA-4 and PD-L1 in colorectal carcinoma using tissue microarray. 2019. (NIH-BRAINS Mentored research grant; Received).
6. Bankole OB (PI), Sampson JB (PI), Abbey E, Frank S, Ariyo P, **Adeyemo T** (Co-I). Increasing blood availability through intraoperative conservation of blood products in Africa. 2019. (NIH/NHLBI; Applied).
7. **Cornejo-Olivas, M** (PI), Mazzetti P, Veliz-Otani D, Schmidt T. ISIA 2019, International School for Genetics of Inherited ataxias: from genetics to clinics. 2019. (Peruvian grant FONDECYT-CONCYTEC; Received).
8. **Cornejo-Olivas, M** (PI), Veliz-Otani D, Mazzetti P, Fischbeck K, et al. Natural history study of SCA10 in Native American Populations. 2019. (NIH K43 award; In preparation).
9. **Gitaka J** (PI), Obimbo M, Shannon A, Smith R. Evaluating the role of oxidative stress in stillbirths in Kenya. 2019. (Medical Research Council, UK; Submitted).
10. Haddad S (PI), **Messaoud O** (Co-PI), Charfi-Kaddour S, Gargouri M, Drissi LB (PI), et al. Advanced Materials: A Strategic Choice for Energy, Environment and Health. 2019. (Research and Development projects: Tunisian-Moroccan Bilateral Fund; Submitted).
11. Jaja C, **Edem-Hotah J** et al. Analytic and Clinical Efficacy of Microtechnology Point-of-Care Diagnostics for Sickle Cell Disease in Sierra Leone. 2019. (eHealth Africa; In preparation).
12. Kefi R (Tunisian PI), **Messaoud O** (Project lead), Jammoussi H, Bahlous A, Saile R, et al. Multidisciplinary investigation of monogenic forms of diabetes in Tunisia and Morocco. 2019. (Research and Development projects: Tunisian-Moroccan Bilateral Fund; Submitted).
13. **Mburu S** (PI), Anzala O, Waithaka P, Heselmeyer K, et al. Climate change and increased prevalence of Non-Communicable Diseases (NCDs). 2019. (NIH K43 award; In preparation).
14. **Mburu S** (PI), **Gitaka J** (Co-PI). Harnessing the power of Big Data and Bioinformatics to characterize cancer phenotypes and development of new diagnostic and prognostic markers. 2019. (EMBL; Submitted).
15. Menzel S (PI), **Adeyemo TA.** (Co-I) Identification of novel mechanisms of fetal-haemoglobin induction by common genetic variation in patients with sickle cell disease. 2019. (MRC/UK; Applied).
16. **Messaoud O** (PI), Adm M (Co-PI). Simulation of the Arab and German Genomes. 2019. (AGYA/German Federal Ministry of Education and Research Fund; Accepted).
17. **Messaoud O** (PI), Jones M, Abdelhak S. Clinical and genetic characterization of atypical forms of photo-genodermatoses. 2019. (Young Researchers Incentive Project funded by the Tunisian Ministry of Higher Education and Scientific Research; Submitted).
18. **Messaoud O** (PI), Mahmoud AH (Co-PI), Rami C, Drissi B. Functional Materials for Health. 2019. (AGYA/German Federal Ministry of Education and Research Fund; Accepted).
19. Naifar E (PI), **Messaoud O**, Boubaker S, Abdelhak S, Benkahla A, et al. Identification of molecular biomarkers of cancer progression in HPV infections responsible for cervical cancer and cutaneous squamous cell carcinoma. 2019. (Federated Research Project funded by the Tunisian Ministry of Higher Education and Scientific Research; Submitted).

20. **Nakousi-Capurro N** (PI), Bustamante L, Miranda M, Pardo RA, et al. Functional Study of genetic variants associated with isolated congenital anosmia in a Chilean family: An alternative to the study of Central Nervous System development. 2019. (Saval Laboratories, 2018; Received).
21. Romdhane L (PI), **Messaoud O,** Abdelhak S, Ghedira K, Ben Hamda C, et al. Development and establishment of an information portal on genetic diseases in Tunisia. 2019. (Young Researchers Incentive Project funded by the Tunisian Ministry of Higher Education and Scientific Research; Submitted).

2017 Batch

22. **Abad PJ**, Reyes ME, Sur ALD, Ngaya-an FV, et al. Determinants of Parental Adherence to Short-term and Long-term Follow-up in Newborn Screening. 2019. (Newborn Screening Reference Center Grant; Shortlisted).
23. **Avogbe PH** (PI), Sehonou J, Le Calvez-Kelm F, McKay JD, et al. Circulating tumor DNA biomarkers and early detection of hepatocellular carcinoma. 2019. (IARC/WHO, Received).
24. Dinçer P (PI), Kürekçi GK, Kural E, **Tuncay FY**, Önal G, Ünsal S. Preparation and maintenance of CRISPR/Cas9 platforms in zebrafish specific to rare diseases. 2019. (University of Hacettepe, Ankara, Turkey, TAY-201712735; Continued).
25. **Fatima SS** (PI), Rehman R, Khan U, Mohammed N. Epigenetic signature of high contribution susceptibility gene variant for diabetes and obesity. Oct. 2018- Aug. 2019. (University Research Council, Aga Khan University; Received).
26. Jha AK (PI), **Thakur** N (Co-PI). Population based screening of sickle cell disorder of Tharu community of Bardiya district. 2019. (Nepal Health Research Council under Ministry of Health and Population, Nepal; Continued).
27. **Mehrez M I** (PI), Abdel Kader M (Co- PI), El-Kamah G, Mostafa I M. Oral Health related Quality of Life (OHQoL) Assessment Prior and After Oro-dental Management in Epidermolysis Bullosa Patients. 2019. (National Research Centre, Cairo, Egypt; Submitted).
28. **Mgasa A**. Afya Bora Consortium Fellowship in Global Health Leadership- 2018/2019. (Afya Bora Consortium; Received).
29. **Otaify GA** (PI), Abdelhamid MS (Co-PI). Targeted next-generation sequencing in the diagnosis and prevention of skeletal dysplasias with joint deformities. 2019. (Young researcher STDF grant funded by Science and Technology Development in Egypt, Accepted).
30. Pazos CRR, **Casado PL** (PI). Grant to Initiate Technology Training for student in molecular biology. (Fluminense Federal University in partnership with National Council for Scientific and Technological Development (CNPq). Dec. 2018- 2019; Received).
31. **Roy S** (PI), Nuruddin M, Osmani M, Gregor K. Screening of common blinding and ocular tumor among under six-year children in Chittagong district of Bangladesh. 2019. (International Rotary Club, Global Grant; Accepted).
32. Susan Neuhaussen (PI), **Torres-Mejía G** (Consultant), Elad Ziv, Jeffrey Weitzel, et al. Genomics and Breast Cancer in Hispanic Women. (Consejo Nacional de Ciencia y Tecnología (CONACyT), Project number: 349; Continued).
33. **Wayengera M** (PI). Development the Next Generation of Ultra-sensitive, Rapid, Point of Care TB diagnostics for Resource limited settings (NextGENTBDx). 2019. (USAID TB LON; Awaiting co-development).

34. **Wayengera M** (PI). Enabling Local Manufacture and Supply of Pan-filovirus Rapid Diagnostics Tests to Vulnerable Populations within Areas affected by the Ebola Virus Disease outbreak in Eastern Democratic Republic of Congo. 2019. (USAID Creating Hope in Conflict; Applied).
35. **Wayengera M** (PI). Scaling and field testing of 3 Prototypes (Ag, IgM and IgG) of Pan-Filovirus Rapid Diagnostic Tests within the Context of the on-going EVD outbreak in Eastern Democratic Republic of Congo. 2019. (Grand Challenges Africa Round 7 Phase II Closed Call; Applied).
36. **Wayengera M** (PI), Quinn F: Testing prototypes of 3 point of care technology platforms that capture 2 novel TB antigens towards diagnosing active TB disease in HIV-1 infected and exposed children using non-sputum clinical samples. 2019. **(**NIH/NIAID, RFA-AI-19-036; Applied).
37. **Yousef YA** (PI), Tbakhi A, Mehyar M, Mosallam M, et al. Mutational analysis of the RB1 gene and the inheritance patterns of retinoblastoma in Jordan. 2019. (King Hussein Cancer Center Intramural grant; Received).
38. Zlatanovic N (PI), **Petlichkovski A**, Kirijas M, Dobrevski B. Planned genetic testing using NGS technology for patients with cardiological electrophysiological disturbances. 2019. (Innovations Fund of the Republic North Macedonia; Submitted).

2016 Batch

39. Baloglu E (PI), **Deniz E** (Consultant). Identification of protein interacting partners of Na+/K+-ATPase pump in ischemic heart disease model. 2019. (TUBITAK, Turkey; Submitted).
40. Emrence Z (PI), **Deniz E** (Researcher). The Role of NOL7 Gene in the Resistance to ATRA. 2019. (TUBITAK, Turkey; Submitted).
41. **Guio H** (PI). Antimicrobial Resistance in urban and hospital's wastewater of Huánuco determined by metagenomic analysis. 2019. (University of Huanuco; Received).
42. **Guio H** (PI). Interaction of the Genetic-environmental determinants in the development of the immune response of the mestizo and native of the Huánuco region. 2019. (Peru Science and Technology Fund; Submitted).
43. **Guio H** (PI). Susceptibility and resistance genes identification of the human coprolite's microbiome in Caral 2000 B.C. 2019. (Peru science and technology fund; Received).
44. **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. 2019. (INNOVATE PERU; Continued).
45. Habanabakize T. (PI), Rugwizangiga B (PI), **Uwineza A** (Co-PI). Validation of the histological profile of hydatidiform moles using $p57^{KIP2}$ immunophenotyping and conventional karyotyping at the University Teaching Hospital of Kigali (CHUK). 2019. (Kigali Referreal Teaching Hospital. CHUK; Received).
46. Hou, L (PI), Sagay A, Ogunsola FT, Murphy M, **Adeyemo WL** (Co-I), **Abdulkareem FB** (Co-PI). Epigenomic Biomarkers of HIV-Associated Cancers in Nigeria. 2019. (NIH U54; Continued).
47. Mekkawy MK. (PI), **Hussen DF** (Collaborator). Molecular cytogenetic characterization and breakpoint mapping of chromosomal rearrangements towards better management of patients with genetic disorders and carriers. 2019. (The Science and Technology Development Fund (STDF), Egypt; Accepted).

48. Murphy M (PI), Hou, L, Sagay A, Ogunsola FT, **Adeyemo WL** (Co-I), **Abdulkareem FB** (Co-I. Northwestern/Nigeria Research Training Grant for HIV and Malignancies. 2019. (NIH/NCI; Awarded).
49. Mutesa L (PI) **Uwineza A** (Co-PI). Genetic Associations with Pre-term Birth in Rwanda. 2019. (USCF in East Africa Preterm Initiative; Received).
50. Sjarif DR (PI), **Ariani Y** (Co-PI). Identification of Iduronate 2-sulfatase gene defect using mRNA from Indonesian MPS type II patients. 2018 (PITTA, University of Indonesia; Received).
51. Sjarif DR (PI), **Ariani Y** (Co-PI). The development of I2S enzyme actvity from MPS type II patients using leukocytes. 2019. (Indonesian Medical Education and Research Institute; Accepted).
52. Sjarif DR (PI), **Ariani Y** (Co-PI). Variants analysis of glucosylceramidase beta (GBA) gene in type I Gaucher disease from Indonesia. 2018. (PITAA, Univ. of Indonesia; Received).
53. Ustun ST (PI), **Deniz E** (Consultant). Personalized medicine and treatment approaches in rare hereditary arrhythmogenic and hypertrophic cardiomyopathy patients. 2019. (TUBITAK, Turkey; Submitted).

C. Collaborations (NIH and Other Institutions):

NIH: (Total- 20; 2018- 8; 2017- 5; 2016- 7)

2018 Batch

1. **Adewole OO**, Landhi M. Genetic epidemiology of lung cancers in non-smokers. (OAUTHC, Nigeria; NCI/NIH).
2. **Adewole OO**, Levine S. Defining the Role of Apolipoprotein Pathways in African Asthmatics. (OAUTHC, Nigeria; NHLBI /NIH).
3. Bonham V, Jaja C, **Edem-Hotah J**. Research Collaboration on Sickle Cell Disease (SCD) in Sierra Leone- establish a research site in Sierra Leone. (University of Sierra Leone College of Medicine and Allied Health Sciences (Nurisng), Sierra Leone; University South Carolina; NHGRI/NIH; In discussion).
4. **Cornejo-Olivas M**, **Lertwilaiwittaya P**, Traynor B et al. Frequency of C9orf72 hexanucleotide repeat expansion in patients with Huntington-like disorders, a systematic review and meta-analysis. (Instituto Nacional de Ciencias Neurologicas, Peru; Mahidol University, Thailand; NIA/NIH).
5. **Dueñas-Roque M**, Kruszka P, Muenke M. Atlas of Human Malformation Syndromes in Diverse Populations -Prader Willi syndrome, Down Syndrome, Turner Syndrome (Hospital Nacional Edgardo Rebagliati Martins, Perú; NHGRI/NIH).
6. **Messaoud O**, Zghal M, Denguezli M, Khelif A, Kenneth H.K, DiGiovanna JJ. Investigation of Leukemia developed in Xeroderma pigmentosum patients (Institut Pasteur de Tunis; Charles Nicolle Hospital; Farhat Hached Hospital, Tunisia; NCI/NIH).
7. Pasechnikova N, Zborovska O, **Savina O**. Molecular and clinical research in Ukrainian families with the inherited eye diseases. Contract signed. (V. Filatov Institute of Eye Diseases and Tissue Therapy, Ukraine; NEI/NIH).
8. Riazuddin S, **Naeem MA**, Husnain T, Qazi ZA, Butt NH, Ali MH, Assir MZ, Sieving PA, Fielding JH, Riazuddin SA. Hereditary Vision Impairment in Pakistan. (University of

Punjab; Layton Rehmatulla Benevolent Trust (LRBT) Eye Hospital; Allama Iqbal Medical College, Pakistan; Johns Hopkins University; NEI/NIH).

2017 Batch

9. Joloba ML, **Wayengera M**. Integrated Biorepository of H3Africa Uganda - IBRH3AU. (Makrere University, Uganda; NHGRI/NIH).
10. Kateete DP, Jjingo D, Jordan IK, Mardon G, **Wayengera M**. Nurturing Genomics and Bioinformatics Research Training in Africa (BreCA). (Makerere University, Uganda; NIAID/Forgarty/NIH).
11. **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).
12. Matshaba MS, Joloba ML, Kekitiinwa AR, Mardon M, CAfGEN Investigators, **Wayengera M**. Collaborative African Genomics Network (CAFGEN). (Makrere University, Uganda; NIAID/NIH).
13. **Mgasa A**, Muenke M, **Ekanem E**, Pranoot T, Patil SJ et al. The International Genetic Basis of Congenital Heart Disease Study Collaborative Project. **(**Muhimbili University of Health and Allied Science, Tanzania; NHGRI/NIH; Submitted for approval in Tanzania).

2016 Batch

14. **Ekure E**, Kruszka P, Muenke M. Atlas of Human Malformation Syndromes in Diverse Populations. (University of Lagos/Lagos University Teaching Hospital, Nigeria; NHGRI/NIH).
15. **Ekure E**, Kruszka P, Muenke M. Genetics of congenital heart diseases in Africa. (University of Lagos/Lagos University Teaching Hospital, Nigeria; NHGRI/NIH).
16. **Hussen DF**, 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).
17. Mutesa L, Uddin M, Wildman D, Jansen S, **Uwineza A**. Transgenerational Epigenomics of Trauma and PTSD in Rwanda. Phase 1. (University of Rwanda; H3Africa/ NIH).
18. **Sirisena ND**, Bonnemann C. Diagnostic evaluation of patients with complex neuromuscular & neurogenetic conditions without a definite diagnosis. (University of Colombo, Sri Lanka; NINDS/NIH).
19. **Sirisena ND**, Kruszka P, Muenke M. Diagnostic evaluation of patients with various craniosynostosis syndromes, congenital heart diseases and other congenital malformations. (University of Colombo, Sri Lanka; NHGRI/NIH).
20. **Sirisena ND**, Sharan S. Mouse embryonic stem cell-based functional analysis of five unclassified BRCA2 gene variants identified in some of the Sri Lankan patients with hereditary breast cancer. (University of Colombo, Sri Lanka; NCI/NIH).

Other Institutions: (Total- 44; 2018- 20; 2017- 13; 2016- 11)

2018 Batch

1. **Adewole, OO**, **Kemevor-Asima D**, **Ottaru S**, **Edem-Hotah J**. Challenges and Prospects of Genetics and Genomic Medicine Researchers In Developing Countries: Insights from

African Researchers (OAUTHC, Nigeria; Korle Bu Teaching Hospital, Ghana; Catholic University of Health and Allied Sciences (CUHAS), Tanzania; University of Sierra Leone, Sierra Leone).

2. Acharya M, **Dutta AK**, Mukherjee S. Towards understanding genetic architecture underlying hereditary non- syndromic hearing loss (NSHL). (AIIMS, New Delhi; PGIMER; Osmania University; JNCSR; Christian Medical College, India).
3. Awandare GA, Makani J, Ofori-Acquah, SF, Sani MU, **Adeyemo TA**. Sickle Cell Disease Genomics Network of Africa. (University of Ghana, Ghana; Muhimbili University of Health and Allied Sciences, Tanzania; University of Abuja, University of Lagos, & Aminu Kano University Teaching Hospital, Nigeria; University of Pittsburgh).
4. Butali A, **Abebe MK**, Gowans LJJ, **Adeyemo WL**, **Das S**. Investigating the Genetics and Environmental Cause of Orofacial clefts. (Addis Ababa University, Ethiopia; University of Lagos, Nigeria; Kwame Nkrumah University of Science and Technology Cleft Foundation, Ghana; CMC, India; Iowa University; In discussion).
5. **Dutta AK** (PI) and Dr Hillary Martin (PI): Exome-sequencing project in India on rare disorders. (The National Institute of Biomedical Genomics; India; Wellcome Sanger Institute, UK; In discussion).
6. Kingham PT (PI), Efsevia V, Mithat G, Richard F Adebola A, Alatise IS, **Abdulkareem FB** (site PI) et al. Point of Care, Real-Time Urine Metabolomics test to Diagnose Colorectal cancers and Polyps in Low-and Medium-Income Countries. (Memorial Sloan Kettering Cancer Center -MSKCC; multiple centers in Nigeria).
7. Kingham PT, Jesse JS, Martin W, Avinash S, Efsevia V, Robilotti E, Alatise IS, **Abdulkareem FB**, et al. Comparison of Presentation and Outcomes in patients with Colorectal cancer in MSKCC and Nigeria. (MSKCC; multiple centers in Nigeria).
8. **Mburu S**, Anzala, O, Waithaka P, Webale M, Mwaura P. Low-dose Microbial endotoxins exposure and response of cancer characteristics. (Kirinyaga University, KAVI-ICR-University of Nairobi; Murang'a University of Technology, Kenya).
9. **Messaoud O**, Haddad S, Chtourou R, et al. Use of Graphene for molecular biology applications (Institut Pasteur de Tunis; Faculty of Sciences of Tunis; Sidi Thabet Biotechnology Center, Tunisia).
10. **Nakousi-Capurro N**, **Mistri M**, Castillo S, Mutations in PPA2 gene as a cause of sudden unexpected cardiac arrest in a Chilean family. (Universidad De Chile, Chile; Neuberg Centre for Medical Genomics, India).
11. Odukoya LA, Badmos KB, Khramtsova Galina, **Abdulkareem FB**, Olopade OI. Immunohistochemical study and clinicopathologic correlation of COX-2 and HER-2 expression in colorectal carcinoma: a 5-year retrospective study. (University of Lagos, Nigeria; University of Chicago).
12. Olopade OI, Atara Ntekim, Popoola AO, Arowolo OA, Sowunmi Anthonia, Olopade CS, Ibraheem Abiola, **Abdulkareem FB,** et al. Assessing REsponse to neoadjuvant Taxotere and TrAstuzumab in Nigerian women with HER2-positive breast cancer (ARETTA), a multicenter study. (Universities of Lagos, University of Ibadan; Obafemi Awolowo University; Lagos State University, Nigeria; sponsored by-University of Chicago Center for Global Health).
13. Ortega J, Galarreta-Aima C, **Dueñas-Roque M**, **Cornejo-Olivas M**. Ihope, an Ilumina project for genomic testing for rare diseases in Peru. (Ilumina; Hospital Nacional Edgardo Rebagliati Martins; Instituto Nacional de Ciencias Neurologicas, Peru).

14. Pasechnikova N, Zborovska O, **Savina O**, Kremenska U. Genetic diagnostics, medical and genetic consultation for families with retinoblastoma considering IVF. In discussion. (V. Filatov Institute of Eye Diseases and Tissue Therapy; ISIDA-IVF Clinic, Ukraine).
15. **Savina O**, Rykov O, Kremenska U. Genetic testing, medical and genetic consultation for families with aniridia and WAGR syndrome, considering IVF. (Shupyk's National Medical Academy of Postgraduate Education; ISIDA-IVF Clinic; NGO- Aniridia; WAGR, Kyiv Ophthalmological Clinic; ISIDA-IVF Genetic Laboratory, Ukraine; In discussion).
16. Smith R, **Gitaka J**, Obimbo M. Role of oxidative stress and other factors in causing stillbirths. (Newcastle University, Australia; Mount Kenya University; University of Nairobi, Kenya).
17. **Utumatwishima JN**, Teuwen D, Dedeken P, Boon P. A Study in Rwandan Health Care System to Investigate the Quality of Life of People with Epilepsy: The MASS study (MASS: Medical Accessibility and Social Support). (UCB; Ghent University, Belgium; Ruhengeri Referral Hospital, Rwanda).
18. **Wangi KYW,** Agus ARS. Increase the Quality of Life of patients with rare disease in the community. (Dept. of Nurisng & Dept. of Pharmacy, Tarumanagara School of Health Sciences, Indonesia; Megawati S. Community Services, Indonesia; In discussion).
19. **Wangi KYW**, Saputri N, Ningsih DA. Human Dignity in Thalassemia Patient in Indonesia. (Tarumanagara University; Muhammadiyah University Pringsewu; Ibrahimy University, Indonesia; In discussion).
20. Yewubdar B, Getnet Y, Kirenga B, **Muttamba W**, et al. Pharmacogenomics of severe Asthma. (College of Health Sciences; University of Addis Ababa, Ethiopia; Makerere University Lung Institute; Uganda).

2017 Batch

21. **Abad, PJ** (Country Collaborator). Effectiveness of a Web-based Genomic Nursing Intervention. (University of the Philippines Manila, Philippines; University of Eastern Finland; South Eastern Finland University of Applied Sciences, Finland; University of Auckland, New Zealand; Don Mariano Marcos State University, Philippines; Clemson University).
22. Akpovi C, **Avogbe PH**, et al. Biobanking and Biomolecular Resources. (Faculty of Science and Techniques; Polytechnic School of Abomey-Calavi (EPAC)/ University of Abomey-Calavi, Benin).
23. **Avogbe PH**, Le Calvez-Kelm F, and McKay JD. Circulating tumor DNA biomarkers and early detection of hepatocellular carcinoma (University of Abomey-Calavi, Benin; International Agency for Research on Cancer/WHO).
24. **Casado PL**, World Dental Federation (FDI). Consensus in Peri-implant Disease Project. I (Fluminense Federal University, Brazil; World Dental Federation, 2018).
25. **Casado PL**, Vieira AR. Genetic basis of temporomandibular disorder/ peri-implant disease/ periodontal disease. (Fluminense Federal University, Brazil; University of Pittsburgh).
26. **Dhoro M**, Grimes KV. Role of epigenetics in severe acute malnutrition in children under 2 years. (University of Zimbabwe, Zimbabawe; Stanford University; In discussion).
27. **Petlichkovski A**, Barbov I, Sukarova-Angelovska E, Duma F**.** Establishment of genetic diagnosis for patients with muscular dystrophies and other neurological genetic diseases. (Institute for Immunobiology and Human Genetics, University Clinic of Neurology, University Pediatric Clinic, Medical Faculty Skopje, Macedonia).

28. **Petlichkovski A**, Sukarova-Angelovska E Brusco A, Rubeis SD. Analyzing families with a proband diagnosed with a neurodevelopmental disorder using next generation sequencing for whole-exome sequencing. (Institute for Immunobiology and Human Genetics, University Pediatric Clinic, Medical Faculty Skopje, Macedonia; University of Turin, Italy; Icahn School of Medicine, Mount Sinai).
29. **Roy S**. Kaliki S, Fabihan D. Lag time of Retinoblastoma presentation. (Chittagong Eye Infirmary, Bangladesh; London School of Hygiene, UK; L.V. Prasad Eye Institute, India; 14 other global institutions).
30. Valle D, **Mehrez MI**. Genome-wide Sequencing to Identify the Genes Responsible for Mendelian Disorders. (NRC, Egypt; Baylor-Hopkins Center for Mendelian Genetics-PhenoDB).
31. **Vishnopolska SA**, Camper S. Genetics of Hypopituitarism. Analysis of CNV mutations in Argentinian patients. (University of Buenos Aires, Argentina; University of Michigan).
32. **Vishnopolska SA**, Kitzman J. Development of saturation mutagenesis probes and protocol to test GL12 gene variants. (University of Buenos Aires, Argentina; University of Michigan).
33. **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, Jordan; Impact Genetics Lab, Canada).

2016 Batch

34. **Adeoye AM**, Bongani M, Connolly SJ. INVestIgation of rheumatiC AFTreatment Using vitamin K antagonists, rivaroxaban or aspirin Studies (INVITUS trial). (University of Ibadan, Nigeria; University of Cape Town, S. Africa; Population Health Research Institute, Canada).
35. **Adeoye AM**, Karaye KK, Loeb M, Yusuf S. A Randomized Controlled Trial of Influenza Vaccine to Prevent Adverse Vascular Events (IVVETrial). (University of Ibadan; Bayero University, Nigeria; McMaster University, Canada).
36. **Adeoye AM**, Tayo B, Ojo A, Cooper R. Cardiovascular And Renal Events In People With Chronic Kidney Disease. (University of Ibadan, Nigeria; University of Arizona; Loyola University Chicago Stritch School of Medicine).
37. **Adeyemo WL**, Sagay A, Ogunsola FT, Akanmu AS, Murphy M. Epigenomic Biomarkers of HIV-Associated Cancers in Nigeria. (University of Lagos; University of Jos, Nigeria; Northwestern University).
38. **Guio H**, Beraun J. Develop basic science research projects. (INBIOMEDIC; University of Huanuco, Peru).
39. **Guio H**, Loayza W. Develop genetic biomarkers in eye diseases. (INBIOMEDIC; Clinica Vista, Peru).
40. **Hussen DF**, Husien AA, Hammad SA, Refaat KM. Studying cases of Alzheimer disease and correlating cytogenetic biomarkers with severity of cognitive status. (NRC; Kasr El Aini Hospital, Cairo University; Al-Azhar University, Egypt).
41. Sjarif DR, **Ariani Y**, Agustina R, Shankar A. Epigenomic screening of Indonesian stunted toddlers compared to their amino acid profile. (Indonesian Medical Education and Research Institute; University of Indonesia; Indonesian Ministry of Health, Provincial Govt. of Jakarta, Indonesia; University of Oxford, UK).
42. **Tibrewal S**, Dubey S, Ratna R, Mathur U, Kumar V, Young TL. Genetic Evaluation of primary congenital glaucoma. (Dr Shroff Charity Eye Hospital; Guru Nanak Dev University, India; University of Wisconsin; In discussion).

43. **Tibrewal S**, Vanita V. Genetic analysis of pediatric cataract cases in north India. (Shroff Charity Eye Hospital; Guru Nanak Dev University, India).
44. **Uwineza A**, Boemer F, Debray G. R1 result: Training in inborn error of metabolism at the Center of Metabolic of Liege, Belgium. (ARES, Belgium; University of Rwanda, Rwanda).

D. New Research Projects: (Total- 38; 2018- 14; 2017- 15; 2016- 9)

2018 Batch

1. **Abdulkareem FB**, Badmos KB. Epigenetic profiles of colorectal carcinoma in Lagos, Nigeria.
2. **Abdulkareem FB**, Badmos KB, Sonusi, SE, Kolawole HF. Autopsy Prevalence of Congenital abnormalities recorded in LUTH.
3. **Cornejo-**Olivas, Cosentino C. Study for research on genetics of Parkinson disease in Peru, a LARGE PD study. (Approved by local IRB).
4. **Das S**, Thomas MM, Yoganathan S, Srivastava VM, et al. A descriptive study with molecular and cytogenetic analysis in patients of ataxia telangiectasia (AT) from the Indian subcontinent.
5. **Dueñas-Roque M**, Mayorga G, Prötzel A, Gamarra N et al. Birth defects registry in the Health Social Security of Peru. A pilot study at a reference hospital.
6. **Dueñas-Roque M**, Segura P, Soria M, Calderón M, et al. Epidemiology of lysosomal storage disorders in the Health Social Security of Peru.
7. **Gitaka J**, Obimbo M, Muuo S. Next generation sequencing of vaginal microbiome to combat infectious causes of prematurity and stillbirths.
8. **Kemevor-Asima D**, Danyo R, Oduro E. Spectrum of congenital anomalies seen at Korle Bu teaching Hospital.
9. **Mburu S**, Gitonga H. Sustainable Analytical and Predictive Approaches to Actualize Precision Medicine in Cancer Management for Resource-limited Settings.
10. **Messaoud O**, Kefi R, Abid A, et al. Identification of biomarkers for Type 2 Diabetes through multidisciplinary investigations.
11. Oluwole E, **Adeyemo TA**, Akinsete A. Feasibility and Acceptability of Newborn and Early Infant (6 weeks) screening for Sickle Cell Disease in Nigeria: A pilot study in a local government area in Lagos State, Nigeria.
12. Patnaik SK, **Dwivedi A**. Inherited disorders of Brain, Heart and Kidney (Army Hospital Research and Referral, India).
13. Sani B, **Taiwo O**, Basil O. The epidemiological survey of oral health status of Nigerians: A national study.
14. **Utumatwishima JN**, Makoneng P, Mancheski B. A Pilot-Study in Rwandan health care settings to examine the feasibility of a large clinical study to assess the value of paliperidone palmitate in Rwanda.

2017 Batch

15. Aglan MS, **Otaify GA**, AbdelHamid MS, Abdelrahman D, Hassib N, et al. Targeted next-generation sequencing in the diagnosis of osteogenesis imperfecta and Bruck syndrome.
16. **Avogbe PH**, Le Calvez-Kelm F et al. Whole transcriptional profiling and gene expression analysis in urothelial carcinoma.

17. Bayanova M, Kuantaeva M, **Tolegen N**. Clinical and Molecular Genetic Research in Auto-Inflammatory Diseases for Children.
18. **Casado PL**, Schimidlin P, Ramenzoni L. Associations between smoking and epigenetic modifications in periodontitis and peri-implantitis tissues.
19. **Fatima** SS, Arian F, Farhat S. Behavioral effects and neuronal changes associated with subclinical hypothyroidism on pregnant females and developing offsprings.
20. **Fatima SS**, Kanwal S, Anjum N. Markers of insulin resistance in Polycystic Ovary Syndrome in adolescents.
21. **Fatima SS**, Kumar W, Anjum N. Role of vascular derived growth factor in decreasing severity of retinopathy and Ischemia Modified Albumin Level.
22. **Fatima SS**, Zaidi AF, Khan GM. Sensitivity of Kidney Injury Molecule-1 Level In diagnosing Diabetic Nephropathy.
23. **Mahfoudh W**, Faleh R. Circulating tumor DNA as biomarkers for early diagnosis, treatment response and prognosis of breast cancer.
24. Meyle J, **Casado PL**. Structure and composition of a subgingival biofilm on titanium surfaces in patients with/without diabetes mellitus and periodontitis and in smokers versus non-smokers.
25. **Mgasa A**, **Otarru S**, **Malasa L**. Genetic counseling services in Tanzania- are we adequate? (Protocol submitted for ethical approval).
26. **Roy S**. Clinico-Pathological features of Retinoblastoma and treatment outcome in a Tertiary Eye Care Center of Bangladesh.
27. Serbest C K, **Tuncay FY**, Konuk O. Graves ophthalmopathy and FOXP3 polymorphisms.
28. **Vishnopolska SA**. Functional Studies of SNVs in Hypopituitarism and Diseritropoyetic Anaemia.
29. **Wayengera M**, Lewcynski D, Bongcam E, Vlachakis D, Paweska TJ, Jovelin F. Biochemical Adjustments of native EBOV Glycoprotein in Patient Sample to Unmask Target-epitopes for Rapid Diagnostic Testing.

2016 Batch

30. **Ekure EN**, Ifeorah I, Kruszka P, Adeyemo AA, Muenke M. Case reports on genetic diseases associated with congenital heart defects -Rubinstein Taybi syndrome.
31. **Ekure EN**, Sokunbi OJ, Kruszka P, Adeyemo AA, Muenke M. Case reports on genetic diseases associated with congenital heart defects -Hadju Cheney syndrome.
32. Elebute OA, Ojewumi O, Ademuyiwa AA, Bode C, **Adeyemo WL**. Association between hypospadias risk and genetic polymorphisms in steroid 5-alpha-reductase type 2 and fibroblast growth factor.
33. **Guio H.** Study genetic risk factors to metabolic diseases in individuals from Caral (5000 B.C.).
34. Mohamed AM, **Hussen DF**. Copy Number Variance and Gene Expression in Diagnosis of genetic causes of Intellectual Disability.
35. Mtalika A, **Malasa L**, Bukini D. Parental understanding on Sickle Cell Trait following Newborn Screening for SCD in Dar es salaam.
36. Nkya S, **Malasa L**, Maleto J, Christopher H, Ambroise E, et al. African Newborn Screening and Early Intervention Consortium: Tanzania Site.
37. Sjarif DR, **Ariani Y,** Priambodo R. The establishment of method to screen urine heparan sulphate and dermatan sulphate from Indonesian MPS type II patients.

38. **Tibrewal S**, Ratna R. Perceptions of Ophthalmologists regarding the need to genetic counseling in India, for hereditary eye diseases. (Submitted to IRB)**.**

E. Ongoing Research Projects: (Total- 49; 2018- 9; 2017- 18; 2016- 22)

2018 Batch

1. **Cornejo-Olivas M**, Smichdt T, Gordon C, Franca M. The EUSAge study: European-South American collaborative Project to identify and characterize age related genetic modifiers and biomarkers of neurodegenerative processes in SCA3/MJD. (Summit lectures in neurogenetics improved study execution).
2. Jaja C, **Edem-Hotah J**, Mustapha I, Bell N, Barrie N, et al. Incidence and Prevalence of Sickle Cell Disease (SCD) in Sierra Leone. (Summit reinforced idea of a registry for recording and tracking SCD population).
3. Kosiyo P, Otieno W, **Gitaka J**, Nyamuni J, Ouma C. Sickle cell genotypes in malaria. (Molecular techniques learnt during the Summit improved protocol development).
4. Margaret Pericak-Vance, **Cornejo-Olivas**. Genetics of dementia in a Peruvian cohort. (Summit lectures in Bioethics in genetic research and Scientific article writing improved informed consent form and writing of manuscript).
5. **Mburu S**. Tumor Microenvironment Heterogeneity and Individual Variabilities: Promising Precision Medicine Targets in Myeloid Neoplasms and Acute Leukemias; A Systematic Review (Meta-synthesis). (Summit helped strengthen concept of Precision medicine-response to treatment, prognosis, evolution of malignancies, targeted therapies in myeloid neoplasms and acute leukemias).
6. **Messaoud O**, Romdhane L, Naouali C, Belhaj O, et al. Genetic characterization of atypical forms of photogenosermatosis. **(**Knowledge gained from personalized trainings at Summit helped with interpretation of incidental findings, exome data analysis).
7. **Messaoud O**, Chimusa E.R, Ahmed A, Mainzer L, et al. Integrative Multi-omics-based Simulation Framework project. (Summit increased awareness; encouraged to be part of other consortia).
8. **Mistri M**, Sheth J. Multicentric collaborative study of lysosomal storage disorders (LSDs) in India. (Knowledge gained at Summit improved research quality).
9. **Nakousi-Capurro N**, Huserman J, Castillo S, Herrera L, et al. Knobloch Syndrome in a patient from Chile. 2019. (Observation of research and team work at NIH during the Summit, raised research standards).

2017 Batch

10. **Avogbe PH**, Le Calvez-Kelm F, et al. Whole transcriptional profiling and gene expression analysis in urothelial carcinoma. (Data analysis started).
11. **Benítez Cordero Y**. Characterization and development of an integrated system for management of major congenital defects in Cuba for the period 2007-2020. (Summit helped reaffirm usefulness of data registries for genetic epidemiological studies).
12. **Casado PL**. Analysis of 7 different techniques to isolate microRNA. (Summit Bioinformatics lecture provided clarity on relationship between miRNA, DNA and mRNA; improved execution of project).

13. **Casado PL**. Genetic profile of patients that underwent peri-implant maintenance therapy: relationship with peri-implant disease incidence. (Knowledge from Summit helped improve sample collection, medical history analysis, selection of the main related genes in multifactorial peri-implant diseases).
14. **Dhoro M**, Mujuru H, Kaisi D and Dingswayo P. Determine the status of congenital disorders at Parirenyatwa Hospital, Harare, Zimbabwe. (Summit helped develop proposal to fill data gap on prevalence and characterization of genetic disorders in sub-Saharan Africa).
15. Hammad SA, **Otaify GA**, **Ahmed DF**, Assem E, Aglan M, et al. Predictive Cytogenetic Biomarkers for non-disjunction disorders. (Knowledge gained from Summit improved reporting of results).
16. Kholoussi N, **Otaify GA**, Helwa E, Aglan MS, et al. Immunogenetic approach in diagnosis and prognosis of some genetic disorders. (Knowledge gained from Summit improved reporting of results).
17. 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 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).
18. **Otaify GA**, Aglan MS, AbdelHamid MS, Mehrez MI, et al. Clinical and molecular study of patients with inherited metabolic bone disorders. (Knowledge gained from Summit improved reporting of results).
19. Rinaldi S, **Torres-Mejía G**, Sánchez G, Garmendia Ma L. Molecular Subtypes of Premenopausal Breast Cancer in Latin American Women (PRECAMA): a multicenter population-based case-control study (IARC 2012- Continued).
20. **Roy S**. Effect of Early Treatment with Oral Propranolol on Periocular Capillary Haemangioma on Outpatient Basis. (Continued).
21. Sewankambo N, **Wayengera M**. Prevalence of polymorphic genes of Alzheimer's disease in nodding disease tauopathy. (NURTURE Supplementary fellowship).
22. Suarez Besil B, **Benítez Cordero Y**, et al. Community strategy for the promotion-prevention of genetic health in primary health care. (Continued).
23. Temtamy S, **Otaify GA**, Kholoussi N, Aglan M, Zaki MS. Recent advances in the diagnosis, management and research of genetic diseases. (Knowledge gained from Summit improved reporting of results).
24. **Tuncay FY**, Dinçer P. Evaluation of phenotypic effects of genome editing mediated TGFBI variation on zebrafish cornea. (Summit contacts helped resolve issues with project).
25. **Tuncay FY**, Ünlü M. Genetic analysis of a family with anterior segment dysgenesis. (Summit helped plan, explain importance of participation in project to patients, families, clinicians).
26. Vaca F, **Torres-Mejía G**, Díaz Velázquez CE, Martínez Razo G, Moreno Macías H. Identification of pathogenic variants of susceptibility to early-onset breast cancer in young women (Continued).
27. **Yousef YA**, Mehyar M, Mosallam M. The impact of RB1 type gene mutation in clinical presentation and management of outcome, in retinoblastoma patients. (Summit helped understand need and impact of types of mutations, presentation, response to management).

2016 Batch

28. **Adeoye AM**, Ayede AI. CARdiovascular rIsk Factors and diseases among pregnant women and their Foetal outcome in Ibadan (CARdIFF Study). (Continued).
29. **Adeoye AM**, Kuti M. Carotid intima media thickness and lipid markers of atherosclerosis among hypertensives. (Continued).
30. **Adeoye AM**, Owolabi MO. Exploring the phenomics, genomics and environmental determinants of left ventricular mass among offspring of hypertensive African Blacks: A family screening study. (Continued).
31. **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). (Continued).
32. **Deniz E**. Genome-wide screening with CRISPR/Cas9 and modelling of resistance mechanisms developed against cytotoxic drugs in cancer treatment. (Knowledge from Summit in personalized medicine and genomic information helped write proposal, analyze bioinformatics data).
33. **Ekure EN**, Bode-Thomas F, Sadoh WE, Orogade AA. Nigerian pediatric cardiac disease registry. (Summit reinforced relevance of registry for congenital heart disease).
34. Gakuru Angelique, **Uwineza A**. Patterns and risk factors for congenital abnormalities in tertiary health facilities in Rwanda. (Summit identified the need to implement a birth defect registry in Rwanda).
35. Helmy N, **Hussen DF**. Using of microarray technique in diagnosis of agenesis of corpus callosum. (Continued).
36. Mahmoud A, **Hussen DF**. Nicotine dependence as an environmental health problem, the efficacy of different approaches for its management. 3-year project. (Continued).
37. Nkya S, Mtei L, **Malasa L**, Soka D, Mdai V, Mwakale P, et al. Newborn screening for sickle cell disease in Dar es Salaam, Tanzania. (Knowledge gained from Summit supported implementation).
38. Olatosi O, **Adeyemo WL**, Butali A. Investigating the genetic causes of early childhood caries. (Continued).
39. **Roblejo HB**. Clinical-epidemiological surveillance of microcephaly and other congenital defects after infection by Zika in pregnant women, products of conception and Cuban newborns. (Continued).
40. **Roblejo HB**. Community strategy for the promotion-prevention of genetic health in primary health care. (Continued).
41. **Roblejo HB**. Location of genetic susceptibility markers associated with the origin of schizophrenia in the Cuban population. (Continued).
42. **Seven M**, Paşalak İŞ, Bağcivan G. Genetics, biomarkers and symptoms during the trajectory of breast cancer: Sytematic review of literature. (Summit helped focus on high-risk women/individuals with genetic pre-disposition to cancer and development of strategies for risk reduction; integrated genetics into behavioral research, biomarkers and strategies for management of patient's symptoms).
43. Sjarif DR, **Ariani Y,** Priambodo R. mRNA identification method to detect IDS gene defect in Indonesia type II MPS patients. (Continued, project was inspired by Summit lectures on Inborn errors of metabolism, Tools for molecular genetics, Diagnostic odyssey, Variant nomenclature and interpretation).
44. Sjarif DR, **Ariani Y**, Priambodo R. Variant analysis of galactosamine (N-Acetyl)-6-Sulfatase (GALNS) Gene in Mucopolysaccharidosis IVA patients in Indonesia. (Project was

inspired by Summit lectures on Inborn errors of metabolism, Tools for molecular genetics, Diagnostic odyssey, Variant nomenclature and interpretation).

45. Sjarif DR, **Ariani Y,** Priambodo R. Variants analysis of GBA gene in Indonesian Gaucher disease patients. (Continued, project was inspired by Summit lectures on Inborn errors of metabolism, Tools for molecular genetics, Diagnostic odyssey, Variant nomenclature and interpretation).
46. Sjarif DR, **Ariani Y**, Priambodo R**.** Variant analysis of IDS gene in 30 Mucopolysaccharidosis type II patients. (Continued, project was inspired by Summit lectures on Inborn errors of metabolism, Tools for molecular genetics, Diagnostic odyssey, Variant nomenclature and interpretation).
47. Sjarif DR, **Ariani Y**, Priambodo R**.** Variant analysis of PTS gene in PKU. (Continued, project was inspired by Summit lectures on Inborn errors of metabolism, Tools for molecular genetics, Diagnostic odyssey, Variant nomenclature and interpretation).
48. Sjarif DR, **Ariani Y**, Yuliarti K, Hafifah CN. Genotype-phenotype analysis of type II MPS patients from Indonesia. (Continued, project was inspired by Summit lectures on Inborn errors of metabolism, Tools for molecular genetics, Diagnostic odyssey, Variant nomenclature and interpretation).
49. **Uwineza A**, **Hitayezu J**. Genetic etiology of epilepsy in Rwandan patients with intellectual disability. (Knowledge gained from Summit helped include microdeletion, microduplication of genes; helped organize writing of scientific paper).

F. Other Accomplishments/Initiatives:

2018 Batch

1. **Abdulkareem FB**. Reviewer for NIH-sponsored Year 05 Mentored Research Applications for BRAINS (Building Research and Innovations in Nigeria's Science), University of Lagos (Nigeria, Jul. 2019); Abstract reviewer & facilitator for African Organization for Research & Training in Cancer Conference; Awarded 'Award of Excellence' by Faculty of Basic Medical Sciences, College of Medicine University of Lagos for contribution to research at the 5th Annual Scientific Conference (Oct. 2018); Attended Consortium of Universities in Global Health, Chicago (USA, Mar. 2019); Sponsored to attend Annual NCI/Fogarty International Program U54 Consortia Meeting on HIV-Associated Malignancies in Low-and Middle-income Countries (Tanzania, Mar. 2019); Invited speaker annual consortium meeting of the African Research Group for Oncology, presented on 'Update on the pathogenesis of colon cancer in Nigeria and future directions' (Nigeria, Apr. 2019); Presented on 'Colorectal cancer molecular subtypes by immunohistochemistry in a patient cohort from Nigeria', at ASCO Annual Meeting (USA, Jun. 2019); Invited speaker Global Oncology Summit (USA, Aug. 2019), presented on 'Point of care technologies in cancer diagnosis-the Nigerian experience'; Included cancer genetics/genomics in Pathology curriculum for MBBS/BDS; Founded, registered an NGO-FAJIM Medicare Foundation (https:fajimmf.org.ng) to improve health in underserved communities through health education, free medical checkups for non-communicable diseases; Initiated registry for congenital abnormalities; Identified patient with clinical, radiological features suggestive of Melorheostosis, managed by surgery, referred for counseling; Initiated documentation of cases of congenital abnormalities diagnosed at autopsy; Diagnosed cases (8) of congenital

anomalies at autopsy- tetralogy of Fallot (2) , VSD cases (2) with Eisenmenger's complex in one, tracheoesophageal fistula with malrotation (2) with ASD in one, Cleft lip and palate (1), multiple congenital anomalies in premature neonate (1).

2. **Adewole OO**. Lectured in genomics to medical students; Emphasized role of genomics in personalized care; Trained colleagues to establish research/clinical facilities.
3. **Adeyemo TA**. Facilitator at Genetic counseling course/workshop for Sickle Cell Disorder, Sickle Cell Foundation of Nigeria (May. 2019), delivered 4 lectures (Hemoglobinopathy: Definition and Types, Exchange Blood Transfusion, Basic Principles of Laboratory Diagnosis of Sickle Cell Disease and Practical Steps in Genetic Counseling); Mentored 7 junior faculty, College of Medicine University of Lagos and Lagos University Teaching Hospital; One awarded a seed Grant for 'Feasibility and Acceptability of New born and Early Infant (6 weeks) screening for Sickle Cell Disease in Nigeria; A pilot study in a local government area in Lagos State, Nigeria'; Established a Sickle Cell Disease Registry, Nigeria- targeted 5000 SCD entries for epidemiological studies; Facilitated World Bleeding Disorder Registry-enrolled 44 patients, improved access to replacement therapy and quality of life, established support and advocacy group meetings; Summit helped affirm the usefulness of data and registries for genetic epidemiological studies.
4. **Cornejo-Olivas M**. Part of Faculty team of the 10th MDS-PAS School for Young Neurologists (Peru, Nov. 2018); Invited to be reviewer for Neurogenetics chapter, Frontiers in Neurology Journal (opportunity due to improved scientific writing skills gained during the Summit); Organized with **M. Dueñas-Roque (**fellow Summiteer) and participated in I Curso Internacional de Genética Médica Genéticay Genómica Humana en Perú: Actualidad y perspectivas" (First Peruvian Congress on Medical Genetics, Peru, Jul. 2019); 2018-2019 LEAP Leadership fellow for International Society of Parkinson disease and other movement disorders (MDS)- an opportunity to improve leadership skills and share knowledge acquired during Summit on genetic variant nomenclature; Involved in MDSgene taskforce, an international effort to compile systematic review of every genetic disorder associated with abnormal movement disorder; Mentor for Northern Pacific Global Health Research Fellows Training consortium (FIC/NIH).
5. **Daich Varela M**. Awarded Clinical fellowship in Ophthalmic Genetics (NEI/NIH, 2019-2020, Summit influenced selection); Presented poster on 'Goldenhar Syndrome', Argentinian Ophthalmology Congress (Argentina, Jun. 2019).
6. **Das S**. Started as senior resident in Department of Medical Genetics, Christian Medical College Hospital, Vellore, India (Nov. 2018), selected as postdoctoral fellow (Clinical Genetics) for 2 years (Jul. 2019); Presented poster on 'A descriptive case series of ataxia telangiectasia (AT) from India', at 7th International Conference on Rare & Undiagnosed Disease (India, Apr. 2019); Awarded travel grant by The International Parkinson and Movement Disorder Society, presented poster on 'A descriptive study with molecular and cytogenetic analysis in patients of ataxia telangiectasia(AT) from the Indian subcontinent' (France, Sept. 2019); Identified, diagnosed and counselled patients (410) with various genetic disorders; Referred to specialty departments for interdisciplinary management; Involved in neuromuscular and perinatal clinic for patient management; Volunteered with Organization for Rare Diseases India (ORDI).
7. **Dueñas-Roque M**. President, Peruvian Society of Medical Genetics (2017-2019) and of Research and Innovation committee of Colegio Médico del Perú; Organized with **M. Cornejo-Olivas (**fellow Summiteer), first Medical Genetics meeting (Peru, Jul. 2019), first

course for young medical doctors of Lima (national broadcast via institutional Facebook page of the Colegio Médico del Perú, Peru, Jul. 2019; 5900 hits); Invited Dr. Muenke as speaker on 'Holoprosencephaly, Craniosynostosis syndromes, Genetics syndromes in diverse populations' and 'How to write a scientific paper'; Used materials and resources from the Summit to organize the event.

8. **Dutta AK**. Secured new position as Assistant Professor at All India Institute of Medical Education (AIIMS, Kalyani, Jul. 2019, Summit significantly impacted the selection); Used training materials from the Summit for presentations on 'Cell-free DNA in Prenatal Testing', organized by Association of Medical Biochemists, India; Summit helped design curriculum for graduate medical education programs at AIIMS; Summit instilled zeal to establish, maintain patient registries for rare inherited disorders at National Institute of Biomedical Genomics (NIBG), coincided with Indian National Rare Disease Registry launched by Indian Council of Medical Research (ICMR); NIBG to contribute to the National Registry and provide understanding on burden of inherited rare diseases in India, with focus on research and natural history studies; Achieved accreditation for molecular diagnostic services for NIBG from European Molecular Genetics Quality Network (EMQN) (lab id 2279; NCBI Gene Test Registry https://www.ncbi.nlm.nih.gov/gtr/labs/506786/); Diagnosed patients (578) with rare genetic disorders (chromosomal, neuromuscular and hematological), referred from 20 medical schools; Provided pre-test/ post-test counseling; Referred patients for management; Significantly impacted reproductive decision making, utilization of prenatal diagnostic services and ended 'diagnostic odyssey' for patients; Enrolled families in clinical trials for Duchenne Muscular Dystrophy (PTC therapeutics), charitable programs for enzyme replacement (Genzyme) and exon skipping therapy (Spinraza) in India; Measures permitted patient support groups (Organization of Rare Diseases India (ORDI) and PWS/ Angelman Society of India) to help patients.
9. **Dwivedi A**. Completed Doctorate in Medicine from SGPGIMS (India); Joined as Assistant Professor at Army Hospital Research and Referral (New Delhi, Jul. 2019); Presented on 'Outcome of Enzyme Replacement Therapy in Gaucher disease' (USA, Sept. 2019), at International Society of Inborn errors of Metabolism Conference; First Medical Geneticist at Armed Forces Medical Services in India; Initiated establishment of Genetics- clinic, OPD services, Counseling, Core Genetics lab (cytogenetic, molecular, NGS platforms); Knowledge from Summit helped conceptualize, design first Medical Genetics Centre for Indian Defense Services in management and counseling of patients with genetic disorders (holoprosencephaly, craniosynostosis syndromes, frontonasal dysplasia, lysosomal storage disorders etc.); Materials from Summit helped educate fellow residents and researchers.
10. **Edem-Hotah J**. Invited by **J. Gitaka** (fellow Summiteer), to attend African Academic of Sciences Maternal, Neonatal and Child Health (MNCH) Workshop (Kenya, Jun. 2019) to discuss research priorities, leadership, policy in Africa; Genomics research was emphasized to inform on reduction of maternal, neonatal and child mortalities in Africa; Attended the American Society of Hematology African New-born Screening Program and Early Intervention Consortium Meeting (S. Africa, Jul. 2019), to add Sierra Leone to the consortium for support of sickle cell disease research program; Presented posters on 'Model for early detection of sickle cell disease in rural African community healthcare settings' and 'Early diagnosis and genetic counseling for sickle cell disease across the care pathway in limited-resourced community health clinics', at ICM Regional Workshop Conference (Namibia, Sept. 2019); Included lectures from Summit in curriculum at University of Sierra

Leone (USL) College of Medicine of Medicine and Allied Health Sciences, Faculty of Nursing; Observed increased interest and awareness for genetics and genomics training among faculty and nursing students; Planned with faculty and discussed with administrators need to introduce a course in Diploma in Genetics and Genomics Counseling at the Faculty of Nursing.

11. **Eshete MA**. Chair, Surgical Department Research Ethics Committee, supported residents and faculty members in research; Presented on 'The role of Environmental Factors in the occurrence of orofacial clefts in the Ethiopian Population', at annual assembly of Ethiopian Society of Plastic and Reconstructive Surgeons; Used materials from Summit for seminars and lectures for faculty members and residents; Shared knowledge with fellows and residents from different universities who visited for short and long-term study; Upgraded cleft registry database, registered newborns and adults with orofacial clefts (>250); Cleft patients (> 300) received multidisciplinary cleft care in collaboration with Smile Train and Transforming Faces; Improved quality of life for cleft patients (100) and their parents because of full rehabilitation and ability to speak intelligibly and smile etc.
12. **Gitaka J**. Included genomics in Biomedical curriculum at Kerinyaga University with help of **S. Mburu** (fellow Summiteer); Developed the Mary Help Hospital Congenital Diseases System for recording, tracking and monitoring congenital birth defects, genetic diseases; Identified, diagnosed and treated cases (11)- Down syndrome (3), Congenital Talipes Equino Valus (CTEV, 8); Referred cases (5 CTEV) for counseling and medical management; Patients and families (11) reported improvement in quality of life due to correct diagnosis and pathway to care.
13. **Kemevor-Asima D**. Involved in Workshop in Genetics, Department of Child Health, Korle Bu Teaching Hospital (Dec. 2018); Organized symposium on 'Genomics and Rare Diseases', Rare Disease Ghana in collaboration with West African Genetic Medical Centre, to increase clinicians' awareness on genomics and relevance in patient care, presented on 'Non-invasive prenatal screening and newborn screening' (Feb. 2019); Invited to join a team of researchers and clinicians to start a registry on congenital anomalies, genetic disorders and rare diseases in Ghana.
14. **Lertwilaiwittaya P**. Completed internship in Internal medicine; Started as Attendant in Medical Genetics Clinic, Department of Medicine, and in Molecular Genetics Laboratory, Department of R&D, Faculty of Medicine Siriraj Hospital, Mahidol University, Thailand (Feb. 2019); Reviewed manuscript for Mol Genet Genomic Med.; Introduced, promoted usage, enrolled Mahidol University in OMIM and ClinVar databases; Interpreted dementia patients (64) using NGS panel (ACMG classification), for use in research and clinical utility; Involved in clinical whole exome sequencing interpretations in pediatric neurology (2), pediatric endocrinology (1), metabolic bone (1), adult neuro-ophthalmology (1)); Participated on Intersex Conference Board (disorders of sex development); Enrolled patients (300) with the Collaborative Aging and Dementia Research Society Thailand (CART, ,https://cogdementiathai.com/?locale=en), a web-based uniform data set for neurodegenerative dementia cases, analyzed samples (40) with NGS.
15. **Mburu S**. Included Medical Genetics and Genetic Counseling in graduate school (M.Sc.) curriculum (pending University's Senate approval, Sept. 2019); Introduced, strengthened, expanded medical genetics research and scope, and bioinformatics to include anti-inflammatory diseases and mitochondrial disorders etc., at School of Health Sciences, Kirinyaga University (KyU); Presented report on Summit to University faculty and

administrators, emphasized importance of medical genetics research, big data analysis, precision medicine; Made recommendations for path forward; Emphasized the need for computational tools in big data analysis and importance of electronic health records to undergraduates; Trained and mentored junior staff (2) on medical genetics and ethics in research; Established basic facilities for molecular techniques, cytogenetics, immunohistochemistry (IHC), cell cytotoxicity testing at KyU and KAVI-ICR.

16. **Messaoud O**. On jury for first PhD thesis in Biology, (Mauritania, Dec. 2018); Reviewed papers for new journals (Clinical genetics; Molecular Genetics and Genomic Medicine; Gene; Journal of European Academy of Dermatology Venereology); Member, Arab-German Young Academy of Sciences and Humanities Working Group 'Health and Society**'** **(**http://agya.info); Awarded Idea Competition Prize for 'Raising awareness on rare diseases' (Oct. 2018); Participated in Science Communication training on 'How to Integrate Story-telling in your Communication Style' (Egypt, Oct. 2018); Invited speaker International Symposium on Xeroderma Pigmentosum and other Nucleotide Excision Repair Disorders, presented on 'Clinical and genetic investigation of Xeroderma pigmentosum in Tunisia: current situation and perspectives' (UK, Mar. 2019); Organized Rare Disease Day (Tunisia, Mar. 2019); Participated in training on 'Intercultural team-building' (Germany, Apr. 2019); Participated in workshop on Maternal Neonatal Health, organized by African Academy of Sciences, the UK Academy of Medical Sciences and Bill & Melinda Gates Foundation (Kenya, Jun. 2019); Participated in workshop on 'Introduction to open science', Institut Pasteur de Tunis, (Tunisia, Jun. 2019); Participated in workshop on 'From Traditional to Innovative Active Methods in Higher Education' (Tunisia, Jul. 2019); Invited speaker at Phi's Research and Innovation Summit, presented on 'Biology in the era of Big Data' in plenary session on Bioethics of Gene editing, Stem Cells & Big Data in Research and Therapy (Jordan, Aug. 2019); Member, DNA Repair Interest Group (http://sigs.nih.gov/DNA-repair); Used resources from Summit for lecture on Human Genetics, at Faculty of Medicine of Tunis; Shared Summit lectures with researchers, post-docs and PhD students.
17. **Mistri M**. Scientist (Inherited Genomics) at Neuberg Centre for Medical Genetics (Gujarat, India); Awarded travel grant by Society for the Study of Inborn Errors of Metabolism (SSIEM) for symposium (Greece, Sept. 2018); Co-author on poster 'Mutations in PPA2 gene as a cause of sudden unexpected cardiac arrest in a Chilean family, with **N**. **Nakousi-Capurro** (fellow Summiteer), Chilean Genetics Society meeting, (Chile, Nov. 2018); Worked with NovaSeq NGS platform (first one in Gujarat, second in India); Involved in NGS based diagnostic services (Clinical exome, Whole exome study) for rare and common genetic disorders.
18. **Muttamba W**. Personalized/advanced training at Summit in 'Principles of Biobanking' at NHLBI, provided knowledge in shipment and biobanking of samples from other countries, for the African Severe Asthma Project (ASAP) and H3A Africa Biorepository; Assisted with DNA extraction at Medical and Molecular laboratory within the University.
19. **Naeem MA**. Member of organizing committee, participated in 3rd International Symposium on Advances in Molecular Biology of Plants and Health Sciences (Pakistan, Dec. 2018); Summit helped improve Medical Genetics curriculum; Trained undergraduate and graduate level students (M.Sc.) from Islamia University Bahawalpur and Sardar Bahadur Khan Women's University, Quetta, Pakistan, in human genotyping and Sanger sequencing based on advanced training received during the Summit.

20. **Nakousi-Capurro N**. Completed second year Medical Genetics resident training, University of Chile; Collaborator with the new Undiagnosed Diseases work group, Human Genetics Branch, Biomedical Sciences Institute, University of Chile; Presented poster on 'Mutations in PPA2 gene as a cause of sudden unexpected cardiac arrest in a Chilean family, at Chilean Genetics Society (Chile, Nov. 2018); Presented Clinical meetings on- CNVs in Inborn Errors of Metabolism, at National Institute of Food Technology (Chile, Apr. 2019), Turner Syndrome in Neonatology, at San Borja Arriarán Clinical Hospital (Chile, Jun. 2019), Genetic tests in the clinical setting, at San Borja Arriarán Clinical Hospital (Chile, Jul. 2019); Shared knowledge obtained at Summit with colleagues and students; Technical knowledge acquired during the Summit helped provide better clinical evaluations to patients, resulting in improved quality of life for patients and families.
21. **Olojede OC**. Presented poster on 'Odontogenic Tumours: Pattern seen at a Nigerian Tertiary Institution in relation to the latest W.H.O. Classification', at International Association for Dental Research, IADR/AADR/CADR (Canada, Jun. 2019).
22. **Ottaru S**. Committee Member of Tanzania Society of Human Genetics; Member of Genetic Society of America; Registered for Online self-paced course on Birth Defects Surveillance and Prevention organized by International Clearing House for Birth Defects; Submitted protocol for Genetic Counseling services to Catholic University of Health and Allied Sciences, Mwanza, Tanzania; Collaborated with **A. Mgasa**, **L. Malasa** (2018, 2016 Summiteers) to establish pilot Congenital Birth Defect registry in 3 regional referral hospitals in Dar es Salaam; Registered, diagnosed cases (13), referred them to Muhimbili National Hospital for management and surgeries.
23. **Savina O**. Completed specialization in Medical Genetics from Shupyk National Medical Academy of Postgraduate Education (Mar. 2019); Presented on ophthalmogenetics in Aniridia and WAGR syndrome management at round-table meeting of Ukrainian ophthalmologists (Ukraine, May. 2019); Presented on 'Genetic diagnostics of retinoblastoma', at International Applied Science Conference-Filatov's lections (Ukraine, May. 2019); Presented on 'Peculiarities of visual organs for improving quality of teaching children with eye pathologies', at Social and Pedagogical Partnership Conference (Ukraine, Jun. 2019); Honorary member of all-Ukrainian NGO 'Association of tiflopedagogues of Ukraine'; Initiated introduction of an ophthalmogeneticist position at ISIDA-IVF clinic (pending approval).
24. **Taiwo O**. Appointed member, Research and Training Committee of Intercountry Center for Oral Health for Africa (Nigeria, May. 2019); Invited speaker, presented on 'Maintaining Good Oral Hygiene in Crisis Situations' at Annual General Meeting of the Medical Women's' Association of Nigeria (Nigeria, Nov. 2018); Invited speaker, presented on 'Contingency Table Analysis' and 'Choice of Statistical Test for Data Analysis', at 15th Annual Scientific Conference of the International Association for Dental Research (Nigeria Division, Nov. 2018); Lectured on Research Appreciation at Department of Surgery, Jos University Teaching Hospital, Nigeria; Conducted two-day training on 'Research Methodology' for professional staff of Oral Health Advocacy Initiative, included courses on Introduction to Research Methodology, Development of a Research Title, Research Questions and Hypothesis, How to do a Literature Review, Research Designs, Sample Size Determination, Sampling Techniques, Variables and Measurement Scales, Data Collection and Questionnaire designs, Choice of Statistical Tests for Data analysis, Descriptive Statistics (Nigeria, Jun. 2019).

25. **Utumatwishima JN**. Initiated 'Face2gene Club', screened undetected congenital malformations in Rwanda (knowledge acquired on Face2gene from Summit); Transferred children (15) with Down syndrome to Rwanda Military Hospital.
26. **Wangi KYW**. Invited speaker on 'Prenatal and Newborn Genetics Screening', at Essential Neonatal Care Optimization Symposium & Workshop (Indonesia, Feb. 2019); Developed undergraduate nursing curriculum at Tarumanagara School of Health Sciences (2019), courses included Trend and Issues in Nursing; Mentored students (2) after completion of course; Introduced 3 topics for class meetings in genetics and genomics (Precision Medicine, Prenatal and Newborn Genetics Screening, Genetics Counseling in Nursing).

2017 Batch

27. **Abad PJ**. Organized forum 'Omics in Nursing' with Dr. L. Saligan (NINR/NIH) as resource speaker (Sept. 2018); Shared roles of genetic counselors in Asia at Summer school, Asia Pacific Society of Human Genetics (Nov. 2018); Served as resource person on genetic counseling for web-based resource for health professionals and parents in pediatric cancers (University of the Philippines Open University, funded by Philippines Department of Science and Technology).
28. **Avogbe PH**. Promoted to Associate Professor at University of Abomey-Calavi, Benin (Jul. 2019); Worked as Visiting Scientist, Genetic Cancer Susceptibility Group, International Agency for Research on Cancer/WHO, France; Invited speaker for Molecular Biomarkers and Cancer Research Symposium, University of Abomey-Calavi (Benin, Jun. 2019); Lectured on Genetic basis of disease, Next-generation sequencing methods, Cancer genetics and genomics to graduate level students, University of Abomey-Calavi; Used resources from Summit to include genomics in research curricula.
29. **Benítez Cordero YB**. Speaker at 50th Annual Meeting of ECLAMC and 3rd Annual Meeting of RELAMC (Argentina, Nov. 2018), presented on 'Cuban Registry of Congenital Malformations- General aspects of its operation and considerations for the Basic Manual of RELAMC'; Lectured in Genetic Counseling at graduate level (M.S.), National Center of Medical Genetics, Venezuela; Summit helped update topics in genetics and provided tools for genetic counseling; Mentored residents (2) in clinical genetics; Member of International Clearinghouse for Birth Defects Prevention & Research; Evaluated genetic disorders (52) of either monogenic, chromosomal or multifactorial etiology; Codified cases (3000) for the registry; Referred cases (12) for genetics analysis and medical management (2 Congenital Myopathy, 1 Erdheim-Chester disease, 9 infertile couples (3 trombopathy, 2 Tuner Syndrome, 4 with DNA fragmentation in spermatozoa)).
30. **Casado PL**. Reviewer for Archives of Oral Biology, International Journal of Growth Factors and Stem Cells in Dentistry, Brazilian Dental Journal and POS ONE (2018-2019).
31. **Dhoro M**. Updated graduate school curricula and research based on materials obtained at the Summit; Lectured on Genetic basis of disease and gene therapy, Diagnosis of genetic disorders, Genetic counseling, Genomic medicine and Pharmacogenetics to graduate level clinical pharmacology students (medical doctors, postgraduates); Reference materials and contacts from Summit increased knowledge, improved supervisory skills; Generated interest in genomics research among undergraduate and graduate students; Mentored graduate level students (M.Phil., D.Phil. Clinical pharmacology) on research projects; Used resources from the Summit (text book 'Smith's recognizable patterns of Human Malformation, 7th Edition'), to identify congenital disorders at Parirenyatwa Hospital, Zimbabwe and provide baseline

data for larger studies; Developed Biobank register and database on genomic studies to support genomics research at University of Zimbabwe, College of Health Sciences.

32. **Díaz C**. Graduated as a Clinical Geneticist (2019); Attended VI Latin American course on Lysosomal Storage Diseases organized by Genetic Institute For All and SHIRE (Chile, 2019); Coordinator of course 'Bridging the gap between Genomics and the Clinic-Experiences in Neurology and Psychiatry' (University of Chile, Santiago), presented on 'Experiences in diagnostic applications of Genomics in Chile'.
33. **Fatima SS**. Recipient of Dean's Award for 'Excellence in Research', Aga Khan University (Dec. 2018); Invited speaker on 'Influence of Perinatal Environment over fetal programming for metabolic syndrome', at 2nd Annual Research Talk (ART), University of Karachi (Pakistan, Sept. 2018); Presented on 'Molecular Basis of Nonalcoholic Fatty Liver Disease and Metabolic Syndrome in a South Asian Population', at 6th Biennial South Asian Association of Physiologists and 16th Biennial Pakistan Physiological Society Conference (Pakistan, Dec. 2018)- received Best Paper Award; Invited speaker on 'Health at Every Size; the Hidden Facts of Obesity Paradigms in Pakistan', SZABIST, BARS Symposium (Pakistan, Apr. 2019); Conducted research conference for undergraduate medical, nursing and biomedical students; Emphasized role of good protocol writing and need for inclusion of genetics and genomics in research and health care in low resource settings; Supervised graduate level students (4).
34. **Hitayezu J**. Invited speaker on 'Down syndrome: Medical approach' at World Down Syndrome Day organized by Rwanda Down Syndrome Organization (Rwanda, Mar. 2019)- Theme: 'Leave no one behind'.
35. **James O**. Lectured on Genetics of Craniofacial Malformations to head and neck surgeons at AOCMF Seminar on Congenital Deformities of the Craniofacial Region: Basic and Advanced Management (Nigeria, Jun. 2019); Performed cleft repair surgeries (25).
36. **Lotz-Esquivel S**. Started 3 month Clinical Clerkship in Medical Genetics and Metabolism Department, National Children's Hospital (Costa Rica, 2019); Attended 3rd PAC Summit Rare Diseases & Master Class XLSD – Sanofi Genzyme (Ecuador Apr. 2019); Attended 1st Expert Meeting in Phenylketonuria, Central America and Caribbean – Biomarín (Panamá, Jun. 2019); Aided in establishing first Multidisciplinary Adults' Clinic for Rare and Orphan Diseases in Latin America, Hospital San Juan de Dios (Costa Rica, 2018), a national reference center for adolescents and adults with genetic and metabolic conditions; Expanded database, managed new cases and follow-ups (160) with 60 different diagnosis (disorders of carbohydrate, mitochondrial energy, and protein metabolism, dyslipidemias, porphyrias, lysosomal storage diseases, and other miscellaneous conditions); Referred patients to other specialty departments; Trained hospital staff in different departments; Identified new cases (8) through newborn screening, diagnosed and treated with help from materials provided at the Summit; Helped transition patients to adult clinics; Promoted research and knowledge to colleagues; Consolidated multidisciplinary care, resulted in better patient management, improvement in quality of life of patients and families, and strengthened the public healthcare system.
37. **Mahfoudh W**. Selected to review abstracts for 12th International AORTIC Cancer Conference (Mozambique, Nov. 2019); Received financial support from CRDF Global to participate in 7th Annual Symposium, Global Cancer Research on Translation and Implementation for Impact in Global Cancer Research, organized by NCI, the University of

Chicago Cancer Center, and the Consortium of Universities for Global Health (CUGH) and to attend the 10th Annual CUGH Global Health Conference (USA, Mar. 2019).

38. **Maure Pizarro P**. Finished residency, started work at Hospital Exequiel González Cortés (Pediatric Hospital); Attended courses- III School of Clinical Genomics, University of Buenos Aires (Argentina, Aug. 2018), NGS in Clinical Genetics and research: Analysis and Interpretation of data, Catholic University of Chile (Chile, Nov. 2018); Participated in Scientific Day 'Improving the quality of life of patients with diagnosis of mucopolysaccharidosis in Chile' (Chile, Dec. 2018); Participated in National Registry of Congenital Anomalies (RENACH, Chile); Collaborated with countries in S. America on Latin American Collaborative Study of Congenital Malformations (ECLAMC), emphasized need to complete the implementation of the registry; Initiated a cytogenetics lab; Integrated learnings from the Summit to attain similar standards of practice in genetics to improve patient care, management of condition and quality of life.
39. **Mehrez MI**. Accepted for Continuing Education Fellowship Program in Craniofacial Genetics at University of Kentucky (USAID program, Jan. 2019); Presented on 'Early diagnosis of genetic disorders through oro-dental anomalies' in a secondary school to educate girls in basics of genetics (Mar. 2019); Presented on 'Research Cookbook' on Scientific Day of Oro-dental Genetics Department, National Research Centre (Egypt, Apr. 2019); Presented an e-poster 'Mandibulacral Dysplasia type A: An Oro-dental Perspective' at European Society of Human Genetics Conference (Sweden, Jun. 2019); Identified Blood disorders (45) and genodermatoses (15); Referred cases (4) for management of dental condition; Provided oral rehabilitation with prosthodontics to patients, resulting in improved quality of life and psychological well-being; Explained importance of genetic counseling as a profession based on Summit learnings; The Summit resulted in a collaboration with Baylor-Hopkins.
40. **Mgasa A**. Applied for early-career Scientist Leadership Program to Genetic Society of America; Submitted two abstracts for 9th International Conference on Birth Defect and Disabilities in the Developing World (Sri Lanka, Mar. 2020); Enrolled in Online Self-paced Course on Birth Defects Surveillance and Prevention organized by International Clearing House for Birth Defects; Collaborated with **S**. **Otarru**, **L**. **Malasa** (2018, 2016 fellow Summiteers) to establish a pilot Congenital Birth Defect registry in 3 regional referral hospitals in Dar es Salaam, Tanzania; Collaborated with pediatricians in regional hospitals for management of affected neonates and families, monitoring of registry; Trained Labor, Neonatal and Pediatric ward staff on identification of congenital birth defects, registration; Diagnosed cases (13), neonates referred to National Hospital for medical/surgical management; Resulted in improved quality of life of affected individuals and their families.
41. **Otaify GA**. Presented poster on 'Challenges in the Management of Metabolic Bone Disorders and Pitfalls to Avoid', at 15th Middle East Metabolic Group meeting (Lebanon, Nov. 2018); Presented on 'Genotype of Bruck syndrome with phenotype of Osteogenesis Imperfecta, separate syndromes or expansion of the spectrum', at European Society of Human Genetics (Sweden, Jun. 2019); Obtained scholarship to attend the 14th Goldrain course on Clinical Cytogenetics (Italy, Aug. 2019); Lectured in the 3rd Basic Course of Human Genetics on 'Phenotypic variation in relation to gene alteration at National Research Centre, Egypt; Lectured a course on Basics of Human Genetics, to undergraduates in Faculty of Physiotherapy, Cairo University, Summit helped structure lectures; Lectured nursing faculty and students, Cairo University on 'Genetic disorders- importance of early detection, management and preventive measures', as part of Community Awareness Program initiated

by Human Genetics and Genome Research Division and implemented goals of Summit; Improved Outpatient consultation, management and counseling for patients with different genetic diseases; Started a registry with few registrations (to be increased after training of personnel); Identified patients (425) with genetic diseases or congenital birth defects (345 skeletal malformations, 26 couples with previous affected births, 54 other genetic disorders); Referred these cases for counseling and management; Treated 40 of the 70 MPS cases with ERT, new cases of osteogenesis imperfecta (22) with bisphosphonate, and cases (15) of hereditary rickets.

42. **Owusu M**. Dr. Muenke's visit (Dec. 2018) initiated process for study on genetics of congenital heart disease at Komfo Anokye Teaching Hospital, Ghana.
43. **Petlichkovski A**. Presented on 'Possibilities and Challenges in Diagnosing Rare diseases' (Macedonia, Feb. 2019) organized by National Alliance for Rare Diseases, supported by the Ministry of Health; Included Summit lectures and data in curriculum at Medical Faculty; Proposed new program for specialization in clinical laboratory genetics (inheritance, diagnosis and treatment of genetic conditions) in cooperation with institutions in genetics, in Macedonia; Program would be recognized in Europe; Summit helped build picture of a functional entity- talks advanced in development of curriculum in genetics with other institutions, conducted preliminary talks with health authorities to merge testing, diagnosis, treatment and counseling under a single institution; Routinely used NGS platform for population-based HLA typing; Performed genetic testing on couples (170) diagnosed with recurrent spontaneous abortion; Referred these cases for genetic counseling.
44. **Nandal R**. Presented on 'Cardiovascular genetics – the new and evolving', at 13th Indian Society of Cardiology Conference (India, Dec. 2018); Conducted weekly congenital heart diseases clinics; Initiated, maintained a congenital heart disease registry; Initiated, streamlined use of facial dysmorphism recognition tools in dysmorphic patients with congenital heart diseases; Identified cases of Noonan syndrome (8), Marfan syndrome (3), William syndrome (3), Di Georgi syndrome (2); Referred these patients for genetic counseling; Referred other patients (~50) with congenital heart diseases and undiagnosed with facial dysmorphic recognition tools, to a clinical geneticist; Identified families (3) with autosomal dominant hypertrophic cardiomyopathy; Referred these families for genetic testing; Routinely established systems for detailed family history, pedigree analysis of patients with familial hyperlipidemia and young myocardial infarction.
45. **Roy S**. Speaker at 46th Annual Conference of Bangladesh Ophthalmological Society (Bangladesh, Mar. 2019) on 'Probing is more Effective as a Treatment Option in Complicated Congenital Nasolacrimal duct Obstruction'; Speaker at 34th Asia Pacific Academy of Ophthalmology (Thailand, Mar. 2019) on 'Establishing RB Center in a Tertiary Eye Care Center of Bangladesh – A New Hope for Retinoblastoma Patients'; Speaker at 6th Biennial conference of Bangladesh Oculoplastic Surgeon Society (Bangladesh, Jun. 2019) on 'Surprises in Pandora's Box- Atypical Cases of Orbit'; Introduced basics of Genetics and Genomics in B.Sc. (Optometry) curriculum; Introduced two CME programs (Basics of Ocular Genetic Diseases; Recent Management Protocol), at the hospital; Identified genetic disorder cases (75), treated patients (56), referred for medical management (19), patients (43) satisfied with their outcome.
46. **Thakur N**. Initiated curriculum in Pathology, Clinical genetics in residency program; Designed 15 day course in Clinical Genetics for faculty and residents at NAMS (pending approval from Academic Council); Established first sequencing center in Nepal; First tests

to be launched- Non Invasive Prenatal Testing, Aneuploidy screening in product of conception, Pre-implantation genetic screening, Cancer hotspot testing for targeted therapy, Germline mutation testing for BRCA1/BRCA2; Established double/triple/quad hormonal testing for prenatal screening; Worked with NAMS and Ministry of Health to establish a clinical geneticist position through examinations; Summit helped improve patient care along the care continuum, number of patients increased 2-fold; Diagnosed new cases (397- 247 new karyotypes with 30 new anomalies).

47. **Tolegen N**. Attended scientific and practical conference on Neuromorphology, at National Medical Research Center of Neurosurgery (Russia, May 2019); Karyotyped pregnant women (426) with suspected fetus abnormalities; Identified chromosomal abnormalities (45- 27 Down syndrome, 6 Patau syndrome, 5 Edwards syndrome, 2 Turner syndrome, 5 structural chromosomal abnormalities); Neuromuscular disease (Duchenne muscular dystrophy, muscular spinal amyotrophy, leukodystrophy) suspected in children (54) and fetuses (2); Molecular genetic testing of MLPA confirmed diagnosis in children (15) and fetus (1).
48. **Torres-Mejía G**. Invited speaker at APEC Conference on Smart Healthcare for Non-communicable Diseases and Their Risk Factors Prevention and Control, presented on 'Precision Prevention and Early Detection of Breast Cancer Using Genetic Markers Associated with Several Modifiable Established BC Risk Factors' (China, Apr. 2019); Presented poster on 'Genes regulating energy homeostasis modify the association between serum concentrations of IGF-1 and IGFBP-3 and breast cancer risk among premenopausal women: The breast health disparities study', at AICR Research Conference on Diet, Obesity, Physical Activity and Cancer: Beyond the Blueprint, (USA, May 2019); Invited speaker on 'Breast cancer in Mexican women, what have we learned?', in the Mammary gland pathology and Surgery course (Mexico, Jul. 2019); Identified (BRCA 1 or 2 mutations) in women (9) with breast cancer (PRECAMA study), sent results to geneticist; Due to importance of counseling emphasized at the Summit, added two geneticists to the staff at the hospital (PRECAMA study) and one at the local Institute.
49. **Tuncay FY**. Passed European Board of Ophthalmology and International Council of Opthalmology Visual Sciences, Optics and Refraction Examinations; Presented as speaker on 'Knockin with CRISPR/Cas9 in zebrafish' at Qatar International Zebrafish Conference and Workshop (Qatar, Apr. 2019); Member of Asian Eye Genetics Consortium, Turkey.
50. **Vishnopolska SA**. Lectured two postgraduate courses- Human Genetics and School of Clinical Genomics; Participated in writing a grant for Precision Medice from Agencia Nacional de Promoción Cientifica y Tecnologica (ANPCyT); Identified, diagnosed and treated a family with isolated growth hormone (GH) deficiency, newborn in family treated immediately with GH replacement therapy.
51. **Wayengera M**. Awarded PhD in Pathogen Omics (Jan. 2019); Selected finalist for WHO Innovation Challenge (Mar. 2019); Worked with Uganda Cancer Institute and partners for integrating genetic testing and counseling in cancer care through National Cancer Policy; Provided opportunity for integration of clinical and molecular genetics and genomics into the care of common diseases in Uganda and Africa; Identified, diagnosed treated patients (156) with congenital abnormalities; Involved in surveillance, control, prevention of the EVD outbreak in DR of Congo.
52. **Yousef YA**. Identified, diagnosed treated cases of Retinoblastoma (28); Referred patients (30) with familial and non-familial Retinoblastoma (genetic test positive) for counseling, management of condition; Improved quality of life of patients and families by informing

patients negative for germline disease, the extremely low risk of disease transmission to their off-springs.

2016 Batch

53. **Adeyemo AA**. Commencement of Universal Newborn Hearing Screening at University College Hospital, Ibadan.
54. **Adeoye AM**. Joint appointment with new Institute of Cardiovascular Diseases, College of Medicine, University of Ibadan, Nigeria; Head of the Molecular Cardiology and Preventive Cardiology Unit; Deputy Chairman, Medical Advisory Committee (Education and Training) at the University College Hospital, Ibadan, Nigeria, and the first Teaching Hospital in the country; As one of the World Heart Federation Emerging Leaders wrote application for inclusion of Dabigatran in WHO Emergency Medicine List (2019 EMLs), enlisted now as a new oral anticoagulant (NOAC) in prevention of stroke and thromboembolism for patients with atrial fibrillation and other hypercoagulable states.
55. **Adeyemo WL**. Awarded 2018 Nigerian Academy of Science (NAS) Gold Medal Prize in Life Sciences (highest honour bestowed on Nigerian Scientist, Jan. 2019); Coordinated Bioinformatics and Genomics Workshops for junior faculty at the College of Medicine University of Lagos, delivered lectures- Introduction to Genomics, Genomics and Other Omics, Techniques and Tools in Genomics, Introduction to Genome databases and SNPedia; Mentored junior faculty (7) awarded Seed Grant to execute research projects in genomics at the College of Medicine, University of Lagos.
56. **Ariani Y**. Presented on 'Identification of a Novel Mutation on Exon 6 of lduronate-2-Sulfatase Gene in an Indonesian Patient With Mucopolysaccharidosis Type ll', 2nd Physics and Technologies in Medicine and Dentistry Symposium (Indonesia, 2018); Developed Molecular Genetics, Population Genetics, Reproductive Genetics modules for graduate level (M.Sc.) Biomedical science students, Faculty of Medicine, University of Indonesia; Established Center of Excellence for Rare Diseases in Cipto Mangunkusumo National Referal Hospital, Jakarta; Promoted the MoU between Cipto Mangunkusumo Hospital and Human Genetic Research Center for free genetic testing for poor patients; Helped start registry for MPS I, Pompe, Gaucher disease, structural birth defects; Continued registry for MPS type II and type IVA; Documented, monitored enzyme replacement therapy for MPS II, MPS IVA, Gaucher and Pompe diseases; Empowered Indonesian Rare Disease Foundation to develop family support groups and patient monitoring.
57. **Deniz E**. Planned to establish a non-thesis introductory program in genomics at Department of Molecular Biology and Genetics, Acibadem University, Istanbul, Turkey, for medical doctors, biotechnologists and individuals curious in genomic data and its clinical utility; As a result of the Summit, supervised graduate level (M.Sc.) students (3) in genomics and genome editing projects.
58. **Ekure E**. Speaker at 1st Conference of the Special thematic Working Group on Maternal, Neonatal, Children and Women Health, on 'Managing Genetic Disorders in Childhood in Emerging Economies: Social, Ethical Financial, System and Family Implications', at University of Ibadan (Nigeria, Apr. 2019); Identified cases (318) of congenital heart defects at Lagos Teaching Hospital; All counseled and/or referred for medical management.
59. **Guio H**. Presented on 'Peruvian Genome Project: An opportunity to understand Peruvians and Latin People', at University of Chile (Chile, Dec. 2018) and Latinono American School

of Medical and Human Genetics (Brazil, May 2019); Presented on 'Genetics and Genomics in Peru' (Luxemburg, Jun. 2019).

60. **Hussen DF**. Selected as Faculty in Egyptian Committee of Pathology Training (ECPT); Lectured on Basic Genetics at The National Research Centre (NRC), in collaboration with ECPT, (Egypt, Mar. 2019); As member of Community Awareness Committee in Human Genetics and Genomics Research Division, NRC (estab. Jan. 2018), payed monthly visits to rural areas of Egyptian governorates and secondary schools to raise awareness in genetic diseases.
61. **Lallar M**. Faculty in DNB Medical Genetics super-specialty training program; Presented poster 'Exome sequencing identifies L1CAM mutation in fetus with ventriculomegaly and agenesis of corpus callosum', at 4th ICBD (India, Dec. 2018); Member organizing committee and session Chair on 'Turner Syndrome- Challenges across lifespan childhood, adolescence and adulthood- An Indo-US educational activity' (India, Jan. 2019); Presented poster 'Gaucher disease: Lessons learnt and insights in the natural history of treated and untreated patients', 5th ISIEM (India, Jan. 2019); Speaker on 'From Genetics to Genomics – An Obstetricians Perspective', at 25th Annual Conference NARCHI (India, Feb. 2019); Organizing Committee member and Speaker at 7th Conference of Undiagnosed Diseases Network International, presented on 'Mystery case- Severe failure to thrive, developmental delay and dysmorphism' and presented poster 'Noonan syndrome and related R A Sopathies: phenotypic and genotypic spectrum' (India, Apr. 2019); Involved in interdepartmental Medical Genetics teaching/training of physicians at SGRH, New Delhi; Identified new genetic cases (55), provided prenatal diagnosis (20), referred patients(25), for symptomatic treatment; Involved in follow up care of patients on ERT for LSD, and for improvement in quality of life; Started a Birth Defect Registry at the referral fetal medicine and diagnostic center.
62. **Malasa L**. Undertook Online training on Human Subject Research: Collaborative Institutional Training Initiatives by Baylor College of Medicine (Jan. 2019); Attended training on Research Electronic Data Capture (REDCap) at Muhimbili University of Health and Allied Sciences (Mar. 2019); Awarded Global Health Fellowship, Novartis Institutes for BioMedical Research (USA, Jun. 2019).
63. **Okafor F**. Promoted to Associate Professor of Nursing Science (back dated to 2017); Nominated, participated in the Accreditation of two Nursing Science Departments in Federal and State Institutions in Nigeria (Jun. 2019); Worked on Ph.D. dissertation 'Nursing Training Institutions' Lecturers' and Students' Reproductive Health Genetics and Genomics Knowledge, Beliefs and Practice in Edo State, Nigeria'; Enlisted as member of Global Genetic Nurses Alliance (G2NA) Group- participated in webinar conference (Jul. 2019); Presented on 'National Policy on Task Shifting and Sharing in Health Care Industry in Nigeria', at the National Association of Nigerian Nurses and Midwives (Aug. 2019).
64. **Roblejo Balbuena H**. Summit improved genetic counseling skills; Lectured on Genetic Counseling (graduate level) at National Center of Medical Genetics in Venezuela; Updated main topics in Human Genetics; Delivered lectures in Clinical genetics to residents, counselors and other professionals; Presented on 'Genetic bases of Schizophrenia', at IX Cuban Congress of Psychiatry (Cuba, Mar. 2019); Presented on 'Prenatal Screening of Sickle Cell Disease Carriers and Inborn errors of Metabolism in Cuba', at XX Anniversary Biolab (Panama, Mar. 2019); Recipient of The National Award of the Academy of Sciences of Cuba 2018 (co-author)- 'Clinical-molecular study of Wilson's disease in Cuba'; Evaluated genetic

cases (118) of monogenic, chromosomal or multifactorial etiology; Registered new cases (48) in the Genetic Diseases' Register of the Pediatric Hospital Center, Havana.

65. **Seven M**. Jury member for Ph.D. theses 'Evaluation of the Effectiveness of the Conseling Model Developed for Prenatal Screening and Diagnostic Tests' and 'Development of Genetic / Genomic Awareness for Pediatric Nurses Scale', Health Sciences University, Gulhane School of Nursing, Ankara, Turkey (2018, 2019); Organizing Commitee member for international conference 'Nursing, Genomics & Healthcare' (UK, Apr. 2020).
66. **Sirisena ND**. Received Korean Breast Cancer Foundation Scholarship for outstanding oral presentation on 'Pattern of Germline Genetic Variants Identified using Next-Generation Sequencing-based Testing in a Sri Lankan Cohort with Hereditary Breast Cancer', at Global Breast Cancer Conference (South Korea, Apr. 2019); Awarded Prof. N.D.W. Lionel Memorial Oration award, Sri Lankan Medical Association for research topic 'Molecular genetic determinants of sporadic breast cancer in Sri Lankan postmenopausal women' (Jul. 2019); Presented posters on 'Mouse Embryonic Stem Cell-Based Functional Analysis of Five Unclassified BRCA2 Variants Identifies One Deleterious and Four Neutral Variants' and 'Functional Studies Implicate XRCC2:rs3218550C>T as a Putative Functional Genetic Variant for Susceptibility to Sporadic Breast Cancer', at the Global Breast Cancer Conference (S. Korea, Apr. 2019); Presented poster on 'PPP1R16B is a Critical Gene in 20q11.2 Microdeletion Syndrome: Loss of Function Variant in PPP1R16B in a Girl with Multiple Congenital Anomalies', at 23rd Human Genome Meeting (S. Korea, Apr. 2019); Invited speaker at the Genomics Transforming Clinical Care Conference organized by the Board of Genetic Counseling, India (India, Jul. 2019); Planned to introduce Certificate course in Human Genetics & Genomics (2019), to improve the genomic literacy of medical health professionals; Provided diagnostic services at Human Genetics Unit, offered genetic counseling to patients (>500) with various genetic diseases and congenital birth defects, referred for appropriate supportive therapies (physiotherapy, speech therapy, occupational therapy), specialized medical management for improvement in quality of life.
67. **Tibrewal S**. Worked closely with appointed Genetic Counselor at Dr. Shroff Charity Eye Hospital (Oct. 2018), on genetic counseling and testing for hereditary eye disorders; Organized two-day workshop in genetics at Global Eye Genetics Consortium (India, Feb. 2019), chaired by Dr. T. Iwata (National Institute of Sensory Organs, National Hospital Organization Tokyo Medical Center, Tokyo, JAPAN) and Dr. G. Prakash (NEI/NIH); Speaker on 'Genetic and Systemic Aspects of Pediatric Cataract', at Annual Conference of Delhi Ophthalmological Society (Apr. 2019); Examined, diagnosed, treated children (~7442); Counselled with help of genetic counselor patients (232) with familial hereditary eye disorders (retinoblastoma, pediatric cataract, retinal dystrophies, corneal dystrophies, keratoconus, microphthalmos, coloboma, aniridia, albinism and several others); Operated patients (26) with pediatric developmental cataracts; Rehabilitated untreatable disorders with vision devices, resulted in significant improvement in quality of life.
68. **Uwineza A**. Lectured medical genetics and genomics in General Medicine and Graduate School (M.Med., Pathology/Pediatrics, Gynecology & Obstetrics), introduced Inborn Errors of Metabolism; Undertook training in NGS Analysis for Monogenic Disease in African Populations (Rwanda, Sept. 2018, funded by Wellcome Trust); Presented on 'Prevalence of epilepsy in Rwandan patients (Belgium, Mar. 2019); Out-patient consultations (~703) done weekly in Kigali Referral Teaching Hospital; Abnormal karyotypes found in patients (154, mostly Down Syndrome); Improved quality of life for patients with psychomotor

development delay through physiotherapy and nutrition, provided speech education for patients with learning difficulties, and psychosocial support for parents with children with genetic diseases.