

ADDENDUM

ISHGG FOURTH ANNUAL OUTCOME (October 2019 - September 2020)

(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 it may not appear in the other three categories).

A. **Publications:** (Total - 304; 2019- 82; 2018- 92; 2017- 56; 2016- 74)

2019 Batch

1. Abdel-Salam GMH, **Sayed ISM**, Afifi HH, Abdel-Ghafar SF, Abouzaid MR, Ismail SI, Aglan MS, Issa MY, El-Bassyouni HT, et al. Microcephalic Osteodysplastic Primordial Dwarfism Type II: Additional Nine Patients with Implications on Phenotype and Genotype Correlation. *Am J Med Genet A*. 2020 Jun; 182(6):1407-1420.
2. Ahmed AE, Ebrahim EA, Mustafa HM, Altohami HI, Gasm Allah LT, Mohammed RA, **Alamin TA**. Polymorphisms in the Glutathione S Transferase Theta 1 Gene and Susceptibility to Sickle Cell Disease. 2020. (In Preparation).
3. **Akinmola OO**, **Okunola O**, **Alli O**. Genetics Section- ‘Types of Inheritance’, ‘Autosomal Inheritance’, ‘Diagnosis of Genetic Diseases’. Textbook of Medicine. 2020. (Submitted).
4. **Akinmola OO**, Okusanya B, Olorunfemi G, Okpara HC, Azinge E. Insulin and Insulin-Like Growth Factor-1 Levels in the Cord Blood of Macrosomic Neonates in Lagos, Nigeria. 2020. (In Preparation).
5. **Alamin TA**, Komi OD, Ibrahim AI, Mohammed AN, Massad ER. Genetic and Genomic Medicine in Sudan, Haematology Prospectus Review. 2020. (In Preparation).
6. **Alamin TA**. Narrative Review Article on Thrombosis in Covid-19 Patients. 2020. (In Preparation).
7. **Alamin TA**. Promising Precision Medicine Targets in Myeloproliferative Neoplasms in Sudan; a Review. 2020. (In Preparation).
8. Ali AZ, Shalaei AA, Yousif AK, Towfeek BA, Ahmed TA, Mohamed RA, **Alamin TA**. Polymorphisms of Glutathione S Transferase Mu 1 Gene and Susceptibility to Sickle Cell Disease. 2020. (In Preparation).
9. **Alli LA**, Nnodu OE. Anti-Sickling Efficacy of Medicinal Plant Extracts from Gwagwalada, Abuja, Nigeria. *West Afric J Med*. 2020. (Submitted).
10. **Amarakoon GGGT**, Mendis D, Senadeera N, Thewarapperuma C, Yoganathan S. a Case Report of a Child with Congenital Lymphoproliferative Disease Type 2, Presented with Hemophagocytic Lymphohistiocytosis Following an EBV Infection. 2020. (In Preparation).
11. **Amarakoon GGGT**, Mendis D, Senadeera N, Thewarapperuma C. a Case Report of a Child with Congenital Amegakaryocytic Thrombocytopenia. 2020. (In Preparation).
12. **Amarakoon GGGT**, Somathilaka M. Infections During Induction Treatment Period in Children with Acute Lymphoblastic Leukemia in Sri Lanka. 2020. (In Preparation).
13. **Baatar N**, Batmunkh B, Tumenbayar B, Baasanjav S. Diabetes Mellitus’s Glucokinase of Gene Sequenced in Mongolian People. *Healthcare Innovation Intermed*. 2019; 34:8.
14. Badoe E, Ameyaw EK, Ghansah A, Amoako E, **Wiafe SA**, Thomford NE, Ernestine Kubi EA, Akyaw PA. Systematic Review of Congenital, Genetic and Rare Disorders Reported in Ghana. 2020. (In Preparation).
15. Baynam G, Chediak L, Bilkey G, Gratton D, **Wiafe SA**. Digit-All: Rare Diseases. *Eur Med J*. 2020; 5(3):11-16.

16. Bramble MS, **Mosema KBA**, Many HB, Likuba EB, Dakande CK, Vilain E, Linguraru M. Using of a Facial Recognition Software for The Diagnosis of Genetic Disorders in the Democratic Republic of Congo. 2020. (In Preparation).
17. Colwell B, **Mosema KBA**, Bramble MS, Maddock J. Comparisons of Social and Demographic Determinants of Tobacco Use in the Democratic Republic of Congo. *Global Health*. 2020 Jul; 16(1):66.
18. Diaz-Gonzalez F, Gayarre-Navarro J, **Llamos-Paneque A**. Rare Event of Two Variants in FGFR2 and FGFR3 in a Child with Syndromic Craniosynostosis. *Eur J Med Genet*. 2020. (Submitted).
19. Diep QM, **Luong LH**, Tran TH, Dinh OTL, Nguyen HQ, Bui TH, Ta TV, Tran VK. a Case of Self-Improving Collodion Ichthyosis in Vietnam. *Brief Report. Pediatr Dermatol*. 2020; 7(3):574-575.
20. Drula R, **Pirlog R**, Trif M, Slaby O, Braicu C; Berindan-Neagoe I. Circfoxo3: Going Around the Mechanistic Networks in Cancer by Interfering with Mirnas Regulatory Networks. *Biochim Biophys Acta Bioenerg*. 2020. (Submitted).
21. Duri K, Chimhuya, Gomo E, Munjoma PT, Chandiwana P, **Mhandire K**, Ziruma A, UZBCS the UZ Birth Cohort Study Team. Role of Antenatal Plasma Cytomegalovirus DNA Levels on Pregnancy Outcome and HIV-1 Vertical Transmission Among Mothers in the University of Zimbabwe Birth Cohort Study (UZBCS). *Virol J*. 2020. (Submitted).
22. Elouej A, Harhour K, Le Mao M, Baujat G, Nampoothiri S, **Llamos-Paneque A**,...Christian Kubisch, Davor Lesser, Annachiara De Sandre- Giovannoli et al. Loss of MTX2 Causes Mandibuloacral Dysplasia and Links Mitochondrial Dysfunction To Altered Nuclear Morphology. *Nat Commun*. 2020; 11:4589.
23. Gamal El Din HM, El Kilani NSE, Mostafa IM, **Sayed I**, Ismail S, Shoreiba EA. Clinical, Radiographic and Histopathologic Evaluation of Ten Congenital Insensitivity to Pain with Anhidrosis Patients. *Al-Azhar Dental Journal for Girls*. 2020. (Accepted).
24. Goel J, Anadure R, Gupta S, Wilson, Saxena R, Sahu S, **Mutreja D**. a Study of the Clinical Profile, Radiologic Features, and Therapeutic Outcomes in Neurosarcoidosis from Two Tertiary Care Centers in Southern India. *Neurol India*. 2020; 68(3):609-616.
25. Gupta N, **Yadav S**, Gurramkonda VB, VI R, Sg T, Kabra M. First Report of THOC6 Related Intellectual Disability (Beaulieu Boycott Innes Syndrome) in Two Siblings from India. *Eur J Med Genet*. 2020; 63(3):103742.
26. **Gyawali P**. Cystatin C: a Better Surrogate for Creatinine. *Medicos Next*. 2020 Mar; ISSN NO. 2717-4778. <https://Medicosnext.com/cystatin-c-a-better-surrogate-for-creatinine>.
27. Harris KH, Nakayama T, Lai J, Zhao B, Argyrou N,...**Iqbal M**, Uguen K, Audebert-Bellanger S, Ferec C, et al. Loss-Of-Function Variants in RFX Family Members Cause Autism, Attention Deficit Hyperactivity Disorder, Intellectual Disability, and Dysregulated Behavior. 2020. (In Preparation).
28. Hendriksz C, Bradley C, Wong-Reiger D, **Wiafe SA**, Du Plessis K, Selebatso E, Mutena C. Challenges Facing the Rare Disease Community Across Africa: the African Task Force for Rare Rare Diseases Report- Executive Summary. 2020. (In preparation).
29. **Hussein N**, Malik TFA, Salim H, Samad AA, Qureshi N, Ng CJ. Is Family History Still Underutilised? Exploring the Views and Experiences of Primary Care Doctors in Malaysia. *J Community Genet*. 2020; 11:413-420.

30. **Hussein N, Tumulak MJR, Lopez Star B, Llamos Paneque A.** Integrating Genetics and Genomics in the 'New Era'. Perspectives from Low-Middle Income Countries in Asia and South America. 2020. (In Preparation).
31. Jois DS, **Mutreja D**, Handa A, Moorchung N. Correlation Between Transbronchial Lung Biopsy and Lung Cytology. *Revista Española De Patología.* 2020 Apr-Jun; 53(2):75-78.
32. **Kars ME**, Toklu Y, Arikan Yorgun M, Neselioglu S, Erun F. Electrolyte, Nitric Oxide and Oxidative Stress Levels of Aqueous Humor in Patients with Retinal Detachment and Silicone Oil Tamponade. *Curr Eye Res.* 2020 Apr; 1-8.
33. Khan AA, Waryah YM, **Iqbal M**, Baig MA, Rafique M, Warya AM. p.Arg102Ser Is a Common PDE6a Mutation Causing Autosomal Recessive Retinitis Pigmentosa in Pakistani Families. *J. Pak. Med. Assoc.* 2020. (Submitted).
34. Köhler S, Gargano M, Matentzoglou N, Carmody CL, Vasilevsky NA, ... **Wiafe SA**, Haendel MA, Mungall CJ, Robinson PN . the Human Phenotype Ontology in 2021. 2020. (In Preparation).
35. Kunz F, Kayserili H, Midro A, De Silva D, Basnayake S, **Güven Y**, Borys J, Schanze D, Stellzig-Eisenhauer A, et al. Characteristic Dental Pattern with Hypodontia and Short Roots in Fraser Syndrome. *Am J Med Genet A.* 2020; 182(7):1681-1689.
36. Lagarde JBL, Laurino MY, Cauyan JML, **Tumulak MJR**, San Juan MD, Ventura ER. It All Began with a Life-Threatening Diagnosis: Experiences of Filipino Women Living with a Sister with Breast Cancer. *J Psychosoc Oncol.* 2020. (Submitted).
37. Landouré G, Dembélé K, Diarra S, Cissé L, Samassékou O, **Bocoum A**, Yalcouyé A, Traoré M, Fischbeck KH, Guinto CO, H3Africa Consortium. a Novel Variant in the Spatacsin Gene Causing SPG11 in a Malian Family. *J Neurol Sci.* 2020; 411:116675.
38. **Llamos-Paneque A**, Gómez-García Ariel O, Rivas-Iglesias C, Garzón-Castro M, Hernández-Iñiguez M, Recalde-Baez MA. Schaaf-Yang Syndrome: An Example of Genomic Imprinting and Expanding Phenotype. *J Mol Genet Med.* 2020; 14:2.
39. **Llamos-Paneque A**, Recalde-Baez MA, Garzón-Castro M, Montúfar S, Rivas-Iglesias C, Lamar-Segura E, Román-Naranjo M, Tambaco- Jijón N, Hernández-Iñiguez P, et al. Supernumerary Marker of Chromosome 15 Associated with Paternal Uniparental Disomy in a Case with Angelman Syndrome. *J Mol Genet Med.* 2019; 13(4):1000439.
40. **López-Star B**, Pérez-Pérez J, Sánchez Moreno P. Evaluation Survey of Genetics Knowledge in Daily Practice of Pediatric Ophthalmologists. *Revista Mexicana De Oftalmología.* 2020. (Submitted).
41. **Luong LH**, Cao MH, Linh KT, Anh HT, Phuong TL, Dung CV, Ha TN, The-Hung Bui, Khanh TV, et al. IGHMBP2- Related Neuromuscular Disorders in Patients from Vietnam. *J Clin Neuromuscul Dis.* 2020. (Submitted).
42. **Luong LH**, Cao MH, Van AP, Oanh TLD, The-Hung Bui, Khanh TV, Ta TV. Microcephaly Primary Hereditary (MCPH): Report of Novel ASPM Variants and Prenatal Diagnosis in a Vietnamese Family. *Taiwanese J Obstet Gynecol.* 2020. (Submitted).
43. **Luong LH**, Nghiem TD, Do GT, Dang TH, Nguyen VA, Nguyen TT Khanh TV, Ta TV. Association of the STAT4, CDKN1A and IRF5 Variants with Risk of Lupus Nephritis and Renal Biopsy Classification in Patients in Vietnam. *Mol Genet Genomic Med.* 2020. (Submitted).
44. Mhandire D, Morse G, Maponga C, **Mhandire K**, Dandara C. Plasma Efavirenz Concentration Inversely Correlates with Increased Risk of Cytomegalovirus Infection in HIV-Infected Pregnant Women. *SAMJ.* 2020 Jan; 110(1):10-5.

45. Mhandire D, Rowland-Jones S, **Mhandire K**, Kaba M, Dandara C. Epidemiology of Cytomegalovirus Among Pregnant Women in Africa. *J Infect Dev Ctries*. 2019 Oct; 3(10):865-876.
46. Mhandire DZ, **Mhandire K**, Magadze M, Wonkam A, Kengne AP, Dandara C. Genetic Variation in Toll Like Receptors 2, 7, 9 and Interleukin-6 Is Associated with Cytomegalovirus Infection in Late Pregnancy. *BMC Med Genet*. 2020 Dec; 21:1-9.
47. Mohamed AE, Edris MA, Ahmed MM, Ali SA, **Alamin TA**. in Silico Analysis of Single Nucleotide Polymorphism in Human IL-4 Gene. 2020. (In Preparation).
48. **Monye HI**, Ekuaze E, Tolani MA, Jolayemi J, Abdulrahman H, Omololu A, Sokomba A, Adebayo O, Ilesanmi OS. a Systematic Approach to the Prevention of COVID-19 in Clinical Settings in Nigeria. *Ibom Medical Journal*. 2020; 13(3):149-155.
49. Morar T, **Pirlog R**, Vlaicu S, Bintintan V, Crisan D. the Dark Side of the Moon in Necrotizing Soft Tissue Infections: a Fulminant Case of Necrotizing Myositis. *Med and Pharm Reports*. 2020. (Submitted).
50. Mostafa MI, El-Badry TH, Abdelrahman MA, Abouzaid MR, Hassib NF, **Sayed ISM**, Ahmed N, Ibrahim MA, **Mehrez MI**, Et Al, Frequent Genetic Disorders Associated with Missing Teeth and Revisiting the Classification of Anodontia: a Retrospective Study. 2020. (In Preparation).
51. Mudawi OM, Ahmed B, Musa SL, Abbas AA, Ibrahim AM, **Alamin TA**. SCD in Sudan; Review. 2020. (In Preparation).
52. **Mutreja D**, Verma S, Venkatesan S, Sharma S, Das SR. Isolated CNS Relapse in FLT 3 Mutation Positive CMML Post Allogeneic Stem Cell Transplant: Report of a Rare Case. *Indian J Pathol Microbiol*. 2020. (Accepted).
53. Nemekhee O, **Baatar N**, Baasanjav S. Clinical and Epidemiological Features of Coronavirus. *Journal of Health Sciences*. 2020; 16(53):83.
54. Okafor L, **Abubakar S**, Anayo E. Partners' Landscape Analysis in Nigeria. Development Activities International (DAI). Technical Report. Bill and Melinda Gates Foundation (BMGF). 2020. (Submitted).
55. **Okunola O**, Alebiosu C, Oyebisi O. Chapters on 'Infections and the Sickle Cell', 'Sickle Cell and the Kidneys'. in Alebiosu Co (Ed). *Sickle Cell Disease: From Laboratory to Clinical Practice*, 1st ed. Cambridge Scholars Publishers. 2020. ISBN-13: 978-1-5275-4260-0.
56. **Okunola O**. Genetics of Kidney Diseases in the Tropics- Review Article. *Annals of Tropical Medicine*. 2020. (Submitted).
57. Onaciu A, Munteanu R, Munteanu VC, Gulei D, Raduly L, **Pirlog R**, Atanas A, Schuyler KS, et al. Spontaneous and Induced Animal Models for Cancer Research. *Diagnostics*. 2020; 10 (9):660.
58. Onat OE, **Kars ME**, Gul S, Bilguvar K, Wu Y, Özhan A, Aydin C, Basak AN, Trusso MA, et al. Human CRY1 Variants Associate with Attention Deficit/Hyperactivity Disorder. *J Clin Invest*. 2020 Jul; 130(7):3885-3900.
59. **Ortiz-Panozo E**, Gómez-Abraján M, Gómez-Flores-Ramos L, Ángeles A, **Torres-Mejía G**. the Role of Aldehyde Dehydrogenase, Alcohol Dehydrogenase and Cytochrome P450 Variants on the Association Between Ethanol Consumption and Breast Cancer Risk. 2020. (In Preparation).
60. Paz-Y-Miño C, Yumiceba V, Moreta G, Paredes R, Ruiz M, Ocampo L, **Llamos-Paneque A**, Ochoa Pérez C, Ruiz-Cabezas JC, et al. Multi-Institutional Experience of Genetic Diagnosis in Ecuador: National Registry of Chromosome Alterations and Polymorphisms. *Mol Genet Genomic Med*. 2020; 8:e1087.
61. **Pirlog R**, Drula R, Berindan-Neagoe I. the Role of Mirna and Macrophage Polarization in NSCLC Field Cancerization. 2020. (In Preparation).

62. Raashid A, Hameed Y, Ejaz S, Khurshid M, **Iqbal M**. Variety of Novel BRCA1 Mutations Detected in Leukemia Patients Inactivate BRCA1. *Curr. Med. Sc.* 2020. (Submitted).
63. Ramdzan SN, Khoo EM, Liew SM, Cunningham S, Kendall M, ...**Hussein N**, Hanafi NS, Mohd Ahad A, Pinnock H. How Young Children Learn Independent Asthma Self-Management: a Qualitative Study in Malaysia. *Arch Dis Child.* 2020; 105(9):819-824.
64. Ranganath P, Sreeja P, **Nair L**, Pamu P K, Shankar A, Murugan S, Dalal A. a Newly Recognized Multiple Malformation Syndrome with Caudal Regression Associated with a Biallelic C.402G>A Variant in TBX4. *European Journal of Human Genetics.* 2020; 28:669–673.
65. Rasool IG, Zahoor ZM, **Iqbal M**, Mahmood T, Anjum AA, Shehzad W. Genetic Variation Analysis of PRSS12, CC2D2A and TRRAP in Pakistani Families Having Children with Intellectual Disability with a Novel Variant. *Fetal. Pediatr. Pathol.* 2020. (Submitted).
66. Romero Aguilar V, Pozo J, Saenz S, **Llamos-Paneque A**, Liehr T, Hosomichi K, Tajima A. Toddler with Whirl-Pooled-Type Pigmentary Mosaicism and Ambiguous Genitalia Resulting from Trisomy 14, Der(Y)T(Y;14). *Human Genome Variation.* 2020 Jul. (Accepted).
67. Sarimiye T, **Monye HI**, Hughes SJ. Impact of COVID-19 Pandemic on Ophthalmology Residency Training in Nigeria. 2020. (In Preparation).
68. **Sayed I**, Rabie E, Mostafa M, Ahmed N, Abouzaid M, Hassib N, **Mehrez M**, El-Kamah Gh, Amr K. Mutational Analysis of WNT10A Gene in Ten Egyptian Patients with Ectodermal Dysplasia. 2020. (In Preparation).
69. **Sayed ISM**, Abdel-Hamid MS, Abdel-Salam GMH. KBG Syndrome in Two Patients from Egypt. *Am J Med Genet A.* 2020; 182(6):1309-1312.
70. Susman S, **Pirlog R**, Leucuța D, Mitre AO, Padurean VA, Melincovici C, Moldovan I, Crișan D, Florian SI. the Role of P-Stat3 Y705 Immunohistochemistry in Glioblastoma Prognosis. *Diagnostic Pathology.* 2019 Dec; 14(1):1-8.
71. Tan HY, Lee YK, Malik TFA, **Hussein N**. School-Based Thalassemia Screening Program in Malaysia. 2020. (In Preparation).
72. Thapa S, **Gyawali P**, Bhatta R, Risal P. Comparison Between Direct Assay and Popular Equations for Low Density Lipoprotein-Cholesterol Estimation in Nepalese Population. *Journal of Pathology of Nepal.* 2020; 10(1):1618-1624.
73. Tilak TVSVGK, **Mutreja D**, Subramaniam S, Handa A. Chemotherapy Associated Pulmonary Toxicity- Case Series from a Single Centre. *South Asian J Cancer.* 2020. (Accepted).
74. **Tiong SY**, Latiff ZA, Abdul Aziz BBS, Winnie Ong PT, Shuib S, Yakub Y, Sulong S. Estimating Growth in the Demand for Genetic Testing As An Indicator for The Demand of Genetic Counselling in Malaysia. 2020. (In Preparation).
75. Tran TH, Luu BT, Pham AD, **Luong LH**, Nguyen LTN, Nguyen BN, Nguyen HV, Bui TH, Khanh TV, et al. Diagnosis and Management of a Patient with Primary Hypertrophic Osteoarthropathy with SCLO2A1 Pathogenic Variants in Vietnam. *J Clin Rheumatol.* 2020 Aug. <https://doi.org/10.1097/rhu.0000000000001524>. (Accepted).
76. **Tumulak MJR**, Padilla CD, Laurino MY, Regalado ES, Legaspi AV, Ventura ER. Psychosocial Issues of Filipino Parents with a Child with Maple Syrup Urine Disease. 2020. (In Preparation).
77. **Tumulak MJR**, Pascua AV, Jover EJ, Guerbo RJ, Canoy GM, Laurino MY. Counseling in the Time of COVID-19: Experiences of Genetic Counselors in the Philippines. 2020. (In Preparation).
78. Verma S, Walia G, Kulkarni SV, **Mutreja D**. Isolated Hydatid Cyst of Pancreatic Tail Masquerading as Serous Cystadenoma. *Archives of International Surgery.* 2019; 9(1):21-23.

79. **Yadav S**, Thakur S, Kohlhase J, Bhari N, Kabra M, Gupta N. Report of Two Novel Mutations in Indian Patients with Rothmund-Thomson Syndrome. *J Pediatr Genet.* 2019 Sep; 8(3):163-167.
80. **Yesilcinar I, Seven M**, Paşalak SI, Guvenc G. Interventions Aiming to Improve Informed Decision on Prenatal Screening and Testing: A Scoping Review of the Literature. 2020. *J Genet Couns.* 2020. (Submitted).
81. **Yesilcinar I, Seven M**, Sahin E, Calzone K. Test/Retest Reliability of a Turkish Version of the Genetics and Genomics in Nursing Practice Survey (GGNPS). 2020. (In Preparation).
82. Zaki MS, Eid OM, Eid MM, Mohamed AM, **Sayed ISM**, Abdel-Hamid MS, Abdel-Salam GMH. Bilateral Calcification of Basal Ganglia in a Patient with Duplication of Both 11q13.1q22.1 and 4q35.2 with New Phenotypic Features. *Cytogenet Genome Res.* 2019; 159(3):130-136.

2018 Batch

83. **Adewole OO**. Impact of COVID-19 on TB Care: Experiences of a Treatment Centre in Nigeria. Letter to the Editor. the Union. *Int J Tuberc Lung Dis.* 2020. <https://www.Theunion.Org/News-Centre/News/Impact-Of-Covid-19-On-Tb-Care-Experiences-Of-A-Treatment-Centre-In-Nigeria>.
84. **Adeyemo TA**, Ojewunmi OO, Oyetunji A, Menzel S, Akinsulie AO. Co-Inheritance of Alpha Thalassaemia and BCL11A in Nigerian Patients with Sickle Cell Anaemia. 2020. (In Preparation).
85. **Adeyemo TA**, Olude E, Bello F, Ogbenna AA, Akanmu AS. Frequency of Intron 1 and 22 Inversion and the Clinical Characterization of Haemophilia a in Nigeria. 2020. (In Preparation).
86. Akpogheneta O, Dicks S, Grant D, Kanne Z, Jusu B, **Edem-Hotah J**, Ijaz S, Tedder R, Bower H. a Novel Approach to Boost Understanding of Lassa Fever Virus Epidemiology: Field Validation of a New Assay to Identify Past Lassa Fever Virus Infection in Blood and Oral Fluids of Survivors and Unexposed Controls in Sierra Leone, 2020. (In Preparation).
87. Alosaimi S, Bandiang A, Van Biljon N, Awany D, Thami PK, Tchamga MSS, Kiran A, **Messaoud O**, Hassan RIM, et al. a Broad Survey of DNA Sequence Data Simulation Tools. *Brief Funct Genomics.* 2020; 19:49-59.
88. Alva-Diaz C, Alarcon-Ruiz C, Pacheco-Barrios K, Mori N, Pacheco-Mendoza J, Traynor BJ, Rivera-Valdivia A, **Lertwilaiwittaya P**,...**Cornejo-Olivas M**. C9orf72 Hexanucleotide Repeat in Huntington-Like Patients: Systematic Review and Meta-Analysis. *Frontiers in Neurology.* 2019. (Submitted).
89. Anata D, **Wangi KYW**, Sumartiningsih MS. Fortitude in Teenager Thalassemia Patient in Jakarta. 2020. (In Preparation).
90. Babalola OC. Gbotolorun OM, **Oluwarotimi OC**, Adesina AOS. Impacted Third and Fourth Molars: a Case Series. *Nig. J Dent Res.* 2020; 451:30-34.
91. Castano BO, **Oluwarotimi AC**, Gbotolorun OM, Obisesan B, Ayodele AOS, Adesina AO. Effects of Prednisolone and Kotase on Post-Disimpaction Sequelae of Mandibular Third Molars in General Hospital, Lagos: a Comparative Study. *Niger Postgrad Med J.* 2020. (Submitted).
92. Cavilah AN, **Wangi KYW**, Sumartiningsih MS. Mental Vulnerability in Adult Thalassemia Patient in Jakarta. 2020. (In Preparation).
93. **Cornejo-Olivas M**, Alva-Diaz C, Custodio N. Evidence Based Guidelines: Development and Application in Clinical Neurology in Peru. *Revista De Neuro-Psiquiatria.* 2019 Dec; 82(4):231-233.

94. **Cornejo-Olivas M**, Custodio N, Mazzetti P. Neurologic Healthcare During COVID Emergency. *Rev. Neuropsiquiatr.* 2020 Mar; 83(2):69-71.
95. **Cornejo-Olivas M**, Inca-Martinez M, Castilhos RM, Furtado GV, Mattos EP, Bampi GB, Leistner-Segal S, Marca V, Mazzetti P, et al. Genetic Analysis of Hereditary Ataxias in Peru Identifies SCA10 Families with Incomplete Penetrance. *Cerebellum.* 2020 Apr; 19(2):208-2156.
96. **Daich Varela M**, Huryn LA, Hufnagel RB, Zein WM, Blain D, Brooks BP. Ocular and Systemic Findings in Adults with Uveal Coloboma. *Ophthalmology.* 2020. <https://doi.org/10.1016/j.ophtha.2020.05.028>.
97. **Daich Varela M**, Moya R, Schlottmann PG, Hufnagel RB, Arberas C, Fernández FM, Eugenia, Inga M, Lores J, Pachajoa H, et al. Ophthalmic Genetics in South America. *Am J Med Genet C Semin Med Genet.* 2020 Aug; 1-9.
98. **Daich Varela M**, Zein WM, Toro C, Grodon C, Johnston J, Huryn LA, d'Azzo A, Tiffet CJ, Fitzgibbon E. a Sialidosis Type I Cohort and a Quantitative Approach to Multimodal Ophthalmic Imaging of the Macular Cherry-Red Spot. *Br J Ophthalmol.* 2020 Aug; *Bjophthalmol-2020-316826*.
99. Danda S, Sony M, Devaraj P, **Dutta AK**, Nampoothiri S, Yesodharan D, Phadke SR, et al. Founder Effects of the Homogentisate 1,2-Dioxygenase (HGD) Gene in a Gypsy Population and Mutation Spectrum of the Gene Among Alkaptonuria Patients from India. *Clin Rheumatol.* 2020; 39(9):2743-2749.
100. **Das S**, Godbole K, Abraham SSC, Ganesan P, Kamdar PP, Danda S, et al. Alazami Syndrome: Report of Three Indian Patients with Phenotypic Spectrum from Adolescence to Adulthood. *Am J Med Genet.* 2020. (Submitted).
101. **Dutta AK**, Goswami K, Murugaiyan S, Sahoo S, Pal A, Paul C, Thallapaneni S, Biswas S. the Transition from Objectively Structured Practical Examination (OSPE) To Electronic OSPE in the Era of COVID-19. *Biochem Mol Biol Educ.* 2020. <https://doi.org/10.1002/Bmb.21410>.
102. **Dutta AK**, Goswami K, Murugaiyan S, Sahoo S, Pal A, Paul C, Thallapaneni S, Biswas S. Evaluation of E-OSPE As Compared to Traditional OSPE: a Pilot Study. *Biochem Mol Biol Educ.* 2020. (Submitted).
103. **Dutta AK**. Variable Expressivity of Malan Syndrome in An Indian Family. 2020. (In Preparation).
104. **Dwivedi A**, Kumar V, Ravi H. Partial Trisomy 16q Associated with Left Ventricular Noncompaction Cardiomyopathy- Novel Cardiac Finding. *J Pediatr Genet.* 2020. (Accepted).
105. **Dwivedi A**, Moirangthem A, Pandey H, Sharma P, Mandal K, Saxena D, Yadav P, Dabadghao P, Phadke S, et al. Yield of Molecular Testing in VHL and Isolated VHL Associated Tumours in an Indian Cohort. *South Asian J Cancer.* 2019. (Accepted).
106. **Dwivedi A**, Simalti A, Kalra S, Goswami J N, Negi V. Role of Including Geneticist in Daily Pediatric Intensive Care Unit (PICU) Rounds: Changing Paradigms. *Turk J Pediatr.* 2020. (Submitted).
107. Eboji OK, Sowemimo AA, Ogunkunle OO, Sofidiya MO, Badmos KB, **Abdulkareem FB**. Toxicological Evaluation of the Stem Bark of *Burkea africana* Hook. (Caesalpinaceae) in Wistar Rats. *Niger. J. Nat. Prod. Med.* 2019; 23(1):33-40.
108. **Eshete M**, Butali A, Abate F, Hailu T, Hailu A, Degu S, Demissie Y, Gravem PE, Deressa WJ, et al. the Role of Environmental Factors on the Etiology of Nansyndromic Orofacial Clefts. *Craniofac Surg.* 2020; 31(1):113-116.

109. **Eshete M**, Muhidin AB, Fikre A, Abiye H, Yohannes D, Abeje B, Mary B, Tamara B, ...Butali A. Amniotic Band Syndrome (ABS) Is Associated with Extremely Severe Atypical Clefts of the Orofacial Region. *Plast Reconstr Surg Global Open*. 2020. (Submitted).
110. Fernanda YA, **Wangi KYW**, Sumartiningsih MS. the Quality of Life Children with Haemophilia in Amarylis Kirana Foundation Tangerang. 2020. (In Preparation).
111. Gelanew M, Fikre A, Abiye H, Yohannes D, **Eshete M**. Indications for Reconstructive Surgery at a Major Public Referral Hospital in Addis Ababa, Ethiopia: a Retrospective, Descriptive Study. *East and Central African Journal of Surgery*. 2020 Jun; 25(2).
112. Gonzales-Sáenz C, Cruz-Rodriguez C, Espinoza-Huertas K, Véliz-Otani D, Marca V, Ortega O, Milla-Neyra K, Alvarez-Tejada J,... **Cornejo-Olivas M**. Distribution of the CAG Triplet Repeat in ATXN1, ATXN3, and CACNA1A Loci in Peruvian Population. *Cerebellum*. 2020 Aug; 19(4):527-535.
113. Haj Ali AB, **Messaoud O**, Elouej S, Talmoudi F, Ayed W, Mellouli F, Ouederni M, Hadiji Mseddi S, De Sandre Giovannoli S, et al. FANCA Gene Mutations in North African Fanconi Anemia Patients. *Am J Med Genet A*. 2020. (Submitted).
114. Hall EG, Wenger LW, Wilson NR, Undurty-Akella SS, Standley J, Augustine-Akpan EA, Kousa YA, Acevedo DS,...**Eshete MA**, et al. SPECC1L Regulates Palate Development Downstream of IRF6. *Hum Mol Genet*. 2020 Mar; 29(5):845-858.
115. Idowu EA, Agbara R, **Taiwo OO**, Nwathor S. Early Childhood Caries and the Role of Parents in Its Primary Prevention: a Report of Three Cases in Nigerian Children. *Nigeria Journal of Dental Research*. 2019; 4(2):75-80.
116. Idowu EA, **Taiwo OO**, Fakuade BO, Afolabi AO. Tooth Loss Awareness and Reasons for Non-Replacement of Missing Teeth Among Nurses at Jos University Teaching Hospital, Jos, Nigeria. *Nigeria Journal of Dental Research* 2020; 5(2):136-144.
117. Iqbal H, Khan SY, Zhou L, Irum B, Ali M, Ahmed MR, Shahzad M, Ali MH, **Naeem MA**, et al. Mutations in FYCO1 Identified in Families with Congenital Cataracts. *Mol Vis*. 2020 Apr; 26:334-344.
118. Isa H, Adegoke S, Madu A, Hassan AA, Ohiaeri C,...**Adeyemo TA**, Kuliya-Gwarzo A, Dogara L, Lawal H, et al. Sick Cell Disease Clinical Phenotypes in Nigeria: a Preliminary Analysis of the Sick Cell Pan Africa Research Consortium Nigeria Database. *Blood Cells Mol Dis*. 2020 Jul 29:102480.
119. Jaja C, **Edem-Hotah J**, Gibson R. Communicating Sick Cell Disease Point-Of-Care Testing Results: We Need Culturally Appropriate Genetic Counseling and Therapeutic Education for Parents. *Lancet Haematol*. 2020. (Accepted).
120. Jaja C, **Edem-Hotah J**, Shepherd J, Patel N, Hongyan Xu H, Gibson R. Analytic Characteristics and Performance of Novel Immunoassay Point-Of-Care Tests for Early Diagnosis of Sick Cell Disease: a Systematic Review. *the Journal of Near-Patient Testing and Technology*. 2020. (Accepted).
121. Jaramillo-Valverde L, Levano KS, Villanueva I, Hidalgo M, Cornejo M, **Cornejo-Olivas M**, Sanchez C, Poterico JA, ...**Guio H**. Guillain-Barre Syndrome Outbreak in Peru: Association with Polymorphisms in IL-17, ICAM1, and CD1. *Mol Genet Genomic Med*. 2019 Oct; 7(10):e00960.
122. Jiao X, Khan SY, Kaul H, Butt T, **Naeem MA**, Riazuddin S, Hejtmancik JF, Riazuddin SA. Autosomal Recessive Congenital Cataracts Linked to HSF4 in a Consanguineous Pakistani Family. *Plos One*. 2019 Dec; 14(12):e0225010.
123. Johnson A, Ayuk A, **Adewole OO**. COVID-19 Handbook (Consensus Statement on Management of COVID-19) Published by the Nigerian Thoracic Society. 2020 May.

124. Kagaya W, **Gitaka J**, Chan CW, Kongere J, Idris ZM, Deng C, Kaneko A. Malaria Resurgence After Significant Reduction by Mass Drug Administration on Ngodhe Island, Kenya. *Sci Rep Nature*. 2019 Dec 13; 9:19060. <https://doi.org/10.1038/S41598-019-55437-8>.
125. Kay C, Collins JA, Caron NS, Agostinho LA, Findlay-Black H, Casal L, Sumathipala D, Dissanayake VHW, **Cornejo-Olivas M**, et al. a Comprehensive Haplotype-Targeting Strategy for Allele-Specific HTT Suppression in Huntington Disease. *Am J Hum Genet*. 2019 Dec; 105(6):1112-1125.
126. Khan R, Shabbir RMK, Raza I, Abdullah U, **Naeem MA**, Ahmed A, Malik S, Hu Z, Xia K. a Founder RDH5 Splice Site Mutation Leads to Retinitis Punctata Albescens in Two Inbred Pakistani Kindreds. *Ophthalmic Genet*. 2020 Feb; 41(1):7-12.
127. Khera S, Pramanik S K, Kalra S, **Dwivedi A**. Type B Lactic Acidosis Due to Warburg Effect in a Child Presenting with T Cell Acute Lymphoblastic Leukaemia: a Milder Phenotype. *BMJ Case Rep*. 2020; 13(3):e233941.
128. Kirenga B, Chakaya J, Getnet Y, Nyale G, Tewodros H, **Muttamba W**, Mugenyi L, Katagira W, Worodria W, et al. Phenotypic Characteristics and Asthma Severity in An East African Cohort of Adults and Adolescents with Asthma: Findings from the African Severe Asthma Project. *BMJ Open Respir Res* 2020; 7(1):e000484.
129. Kirenga B, **Muttamba W**, Kayongo A, Nsereko C, Siddharthan T, Lusiba J, Mugenyi L, Byanyima R, Worodria W, et al. Characteristics and Outcomes of Admitted Patients Infected with SARS-Cov-2 in Uganda. *BMJ Open Respir Res*. 2020; 7(1):e000646.
130. Kosiyo P, Otieno W, **Gitaka J**, Nyamuni J, Ouma C. Correlation Between Haematological Parameters and Sickle Cell Genotypes in Children with P. Falciparum Malaria. *BMC Infect Dis*. 2020. (Submitted).
131. Kumar R, **Dwivedi A**, Puri R. Prenatal Diagnosis of Radial Ray Defect Associated with Fanconi Anemia. *Journal of Fetal Medicine*. 2020. (Accepted).
132. **Lertwilaiwittaya P**, Roothumnong E, Dungort P, Meesamarnpong C, Tansa-Nga W, Pongsuktavorn K, Wiboonthanasarn S, Tititumjariya W, Thongnoppakhun W, et al. High Prevalence of Germline Pathogenic/Likely Pathogenic Variants in Cancer Susceptibility Genes in Thai Breast-Ovarian Cancer Spectrum. 2020. (In Preparation).
133. **Lertwilaiwittaya P**, Suktitipat B, Khongthon P, Pongsapich W, Limwongse C, Pithukpakorn M. Identification of Novel Mutation in RANKL By Whole Exome Sequencing in a Thai Family with Osteopetrosis; a Case Report and Review of Osteopetrosis Cases in Thailand. *Mol Genet Genomic Med*. 2020. (Submitted).
134. Luquetti DV, Heike C, Zarante I, Timms AE, Gustafson J, Pachajoa H, Porrás-Hurtado GL, Ayala P, **Dueñas-Roque M**, et al. MYT1 Role in the Microtia-Craniofacial Microsomia Spectrum. *Mol Genet Genomic Med*. 2020 Sept. <https://doi.org/10.1002/Mgg3.1401>.
135. Mathew M, Tinuola FR, Goel SR, **Taiwo O**, Jalo P. Effects of Peer Education Intervention for Hepatitis B on Level of Knowledge and Beliefs of School Adolescents in Jos, Plateau State Nigeria. *Acta Scientific Nutritional Health*. 2019; 3(11):53-60.
136. **Mburu S**, Gitonga H. An Integrated, Multiple Dimensional, Analytical and Predictive Modelling Approaches to Facilitate Clinical Decision Making: a Breast Cancer Management Model Suitable for Resource-Limited Settings. 2020. (In Preparation).
137. **Mburu S**, Kimani K, Mutuku I. Readiness of Primary Health Care Laboratory Diagnostic Services to Support UHC Programme in Kenya: a Case Study of Three Counties. 2020. (In Preparation).

138. **Mistri M**, Mehta S, Solanki D, Kamate M, Gupta N, Kabra M, Puri R, Girisha K, Hariharan S, et al. Identification of Novel Variants in a Large Cohort of Children with Tay-Sachs Disease: An Initiative of a Multicentric Taskforce on Lysosomal Storage Disorders by Government of India. *J Hum Genet.* 2019; 64:985–994.
139. **Muttamba W**, Tumwebaze R, Mugenyi L, Batte C, Sekibira R, Nkolo A, Katamba A, Kasasa S, Majwala R. Households Experiencing Catastrophic Costs Due To Tuberculosis in Uganda: Magnitude and Cost Drivers. *BMC Public Health.* 2020. (Accepted).
140. Nadeem R, Kabir F, Li J, Gradstein L, Jiao X, Rauf B, **Naeem MA**, Assir MZ, Riazuddin S, et al. Mutations in CERKL and RP1 Cause Retinitis Pigmentosa in Pakistani Families. *Hum Genome Var.* 2020 May; 7(1):1-4.
141. Naifar M, Kallel F, Hadjkacem F, Boudabous H, Kallel R, Boudawara T, **Messaoud O**, Tbib N, Charfi N, et al. Homozygous Parg610del Mutation Unusually Associated with Severe Delay of Growth in 2 Acid Sphingomyelinase Deficiency-Affected Sibs. *J Pediatr Hematol Oncol.* 2020; 42:e499-e502.
142. **Nakousi-Capurro N**, Alegría A, Cares C, Gainza-Lein M, Ojeda V, Irrarázaval MJ. Congenital Anomalies and Comorbidities in Down Syndrome Neonates (RCHPED 1518). *Revista Chilena De Pediatría.* 2020. (Accepted).
143. **Nakousi-Capurro N**, Huserman J, Castillo S, Herrera L, Romero P, Pizarro F, Quezada C. Knobloch Syndrome in a Patient from Chile. *Am J Med Genet Part A.* 2020; 182(10).
144. Ngamson B, Wander A, Iles A, Kimani R, Muregi F, **Gitaka J**, Pamme N. Rapid Detection of Group B Streptococcus (GBS) from Artificial Urine Samples Based on IFAST and ATP Bioluminescence Assay: from Development to Practical Challenges During Protocol Testing in Kenya. *Analyst.* 2019 Oct. 10;144(213):6889-6897.
145. Nwabudike VI, **Oluwarotimi AC**, Okoturo E, Ijogun A. Comparative Study of the Effect of Single and Multiple Suture Techniques on Inflammatory Complications Following Mandibular Third Molar Surgery: a Randomized Prospective Study at the Lagos State University Teaching Hospital (Lasuth), Ikeja. *J West Afr Coll Surg.* 2020. (Submitted).
146. Obiajulu FJ, Ikeri NI, Anunobi CC, Daramola AO, **Abdulkareem FB**, Banjo AAF. the Diagnostic Utility of Cell Block in Fine Needle Aspiration Cytology of Palpable Breast Lesions in a Nigerian Tertiary Health Institution. 2020. *Diagn. Cytopath.* 2020. (Accepted).
147. Ocholla IA, Agutu NO, Ouma PO, Gatungu D, Makokha FO, **Gitaka J**. Geographical Accessibility in Assessing Bypassing Behaviour for Inpatient Neonatal Care, Bungoma County-Kenya. *BMC Pregnancy Childbirth.* 2020 May; 20:287. <https://doi.org/10.1186/S12884-020-02977-x>.
148. Oguntunde OA, Ikeri NZ, Igbokwe U, **Abdulkareem FB**, Banjo AF. Histopathologic Spectrum of Lymph Node Disease in a Lagos Facility. *Ann. Trop. Path.* 2019; 10(2):114-118.
149. Oguntunde OA, Sonusi S, Ikhisimoje S, **Abdulkareem FB**, Banjo AF. Testicular Schistosomiasis Mimicking Hydrocele in a Child: a Case Report. *Pan Afr Med J.* 2020; 35:1-556.
150. **Oluwarotimi AC**, Abah AA, Ogundana OM. Prevalence of Medical Conditions Among Dental Patients in a South-West Hospital in Nigeria. *African Journal of Oral Health.* 2020. (Submitted).
151. **Oluwarotimi AC**, Ogundana OM, Gbotolorun OM, James OA, Adebule O. Odontogenic Tumours: Pattern Seen at a Nigerian Tertiary Center by the 2017 World Health Organization Classification. *East Afr Med J.* 2020. (Submitted).
152. Oluwole E, **Adeyemo TA**, Osanyin EG, Odukoya OO, Afolabi BB, Kanki P. a Pilot Study of the Feasibility and Acceptability of Newborn and Early Infant Screening for Sickle Cell Disease in Nigeria. 2020. (In Preparation).

153. **Ottaru S**, Mirambo M, Kabyemera R, Kidenya B, Seugendo M, Msanga D, Ngoya P, Morona D, Mshana S. Predictors of Toxoplasma Gondii Igg Seropositivity and Cranial Ultrasound Patterns Among Children with Hydrocephalus in a Tertiary Hospital in Mwanza, Tanzania. *International Journal of Pediatrics*. 2020. (Submitted).
154. Oyetunji IA, Amoo AJ, **Adeyemo TA**, Ojewunmi OO. Asymptomatic Malaria Infection Among Children with Sickle Cell Anaemia: the Role of IL-10 and Possible Predisposing Factors. *PAMJ - One Health*. 2020; 2:7.
155. Pasumarthi D, Gupta N, Sheth J, Nurul Jain SJ, Rungsung I,...**Mistri M**, Sankar VH, Gowrishankar K, Agrawal D, et al. Identification and Characterization of 30 Novel Pathogenic Variations in 69 Unrelated Indian Patients with Mucopolidosis Type II and Type III. *J Hum Genet*. 2020; 65:971-984.
156. Ranjan M, Kalra S, Gupta A, **Dwivedi A**. Stumped by Potassium: a Rare Case of Familial Pseudohyperkalemia. *Clinical Case Letters. Indian Pediatrics*. 2020; 57(5):472-473.
157. Ranjan R, **Dwivedi A**, Khera S. Delayed Diagnosis Due to Cognitive Bias and Diagnostic Labelling in a Child with Hypertrophic Cardiomyopathy- a Rare Presentation of Neuroblastoma. *BMJ Case Rep*. 2020. (Submitted).
158. Rauf B, Irum B, Khan SY, Kabir F, **Naeem MA**, Riazuddin S, Ayyagari R, Riazuddin SA. Novel Mutations in LTBP2 Identified in Familial Cases of Primary Congenital Glaucoma. *Mol Vis*. 2020 Feb; 26:14-25.
159. Rekaya MB, Hamdi Y, Labidi S, Mejri N, Jaidane O, Ayari J, Nasr SB, Dallali H, **Messaoud O**, et al. Whole Exome Sequencing Reveals a Combination of Rare High and Low Penetrance Variants That Correlates with Familial Breast Cancer Relative Risk. Preprint from Research Square. 2020 Apr. <https://doi.org/10.21203/Rs.3.Rs-22989/v1>.
160. Rico EMG, Kikuchi A, Saito T, Kumondai M, Hishinuma E, Kaneko A, Chan CW, **Gitaka J**, Nakayoshi T, et al. 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.*, 2020 Feb; 35(1):89-101.
161. Romdhane L, Nessrine M, Dallali H, **Messaoud O**, Shan J, Fakra K, Yosri N, Kefi R, Romdhane S, et al. A Map of Copy Number Variations in the Tunisian Population: a Valuable Tool for Medical Genomics in North Africa. *Genomic Med*. 2020. (Submitted).
162. Sarapura-Castro E, Ramirez-Quñones J, **Cornejo-Olivas M**. Panda with "Bright Eyes": a Rare Sign in Wilson Disease. *Arq Neuropsiquiatr*. 2020 Jun; 28(8):525.S0004-282X2020005014107.
163. Sayeb M, Riahi Z, Laroussi N, Bonnet C, Romdhane L, Mkaouar R, Zaouak A, Marrakchi J ...**Messaoud O**, 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; 58:1439-1443.
164. Schiff ER, **Daich Varela M**, Robson AG, Pierpoint K, Ba-Abbad R, Nutan S, Zein WM, Ullah E, Huryn LA, et al. a Genetic and Clinical Study of Individuals with Nonsyndromic Retinopathy Consequent Upon Sequence Variants in HGSNAT, the Gene Associated with Sanfilippo C Mucopolysaccharidosis *Am J Med Genet C Semin Med Genet*. 2020 Aug; 10.1002/Ajmg.C.31822.
165. Silva-Paredes G, Urbanos-Garrido RM, Inca-Martinez M, Rabinowitz D, **Cornejo-Olivas M**. Economic Burden of Huntington's Disease in Peru. *BMC Health Serv Res*. 2019 Dec 30; 19(1):1017.

166. Suárez C, Aranibar L, **Nakousi-Capurro N, Díaz C, Pizarro P**. Pigmentary Mosaicisms and Cytogenetic Abnormalities: About 2 Cases. *Piel. Formación Continuada En Dermatología*. 2020. (Accepted).
167. Tanimowo MO, Abudu EK, Udo IA, **Abdulkareem FB**. Histopathological and Immunohistochemical Characteristics of Breast Carcinomas in Uyo, Subtropical Region of Africa. *Med. J. Zambia*. 2019; 46(2):100-108.
168. Torres-Romucho CE, Uriondo-Ore VG, Ramirez-Palominio AJ, Arroyo-Hernandez H, Loo-Valverde M, Protzel-Pinedo A., **Dueñas-Roque M**. Factors Associated with Survival at One Year of Life in Neonates with Severe Congenital Cardiopathy in a National Hospital in Peru. *Rev Peru Med Exp Salud Publica*. 2019; 36(3):433-441.
169. **Utumatwishima JN**, Karara G, Sonja S, Carlos G. Aliniq-CDS (Clinical Decision Support) Implementation Initiative in Rwanda. 2020. (In Preparation).
170. **Utumatwishima JN**, Makoneng P, Mancheski B. Assessment of the Value of Paliperidone Palmitate to Treat Schizophrenia in Rwandan Healthcare System. 2020. (In Preparation).
171. **Utumatwishima JN**, Thomas T, Smithswintosky V. Proposal To Use Electronic Medical Records in Health Economic Modelling Research on Schizophrenia. 2020. (In Preparation).
172. 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 Oct; 18(5):841-848.
173. Vishnevetsky A, **Cornejo-Olivas M**, Sarapura-Castro E, Inca-Martinez M, Rabinowitz D, Milla-Neyra K, Mazzetti P, Bird TB. Juvenile Huntington Disease in Peru: Review of 32 Patients and Challenges of Classification. *Mov Disord*. 2020. (Submitted).
174. Zevallos-Morales Alejandro, Murillo Alexis, **Dueñas-Roque M**, Prötzel A, Venegas-Tresierra L, Angeles-Villalba V, Guevara-Cruz M, Chávez-Gil A, Fujita R, Et Al. Novel Mutation in ENG Gene Causing Hereditary Hemorrhagic Telangiectasia in a Peruvian Family. *Genet. Mol. Biol*. 2020; 43(1):e0190126.

2017 Batch

175. **Abad PJ**, Sibulo MSK, Sur ALD. Role of the Nurse in Newborn Screening: Integrating Genetics in Nursing Education and Practice. *Phil J Nurs*. 2019; 89(1):16-21.
176. Acheampong G, **Owusu M**, Owusu-Ofori A, Osei I, Sarpong N, Sylverken A, Kung HJ, Cho ST, Kuo CH, et al. Chromosomal and Plasmid-Mediated Fluoroquinolone Resistance in Human Salmonella Enterica Infection in Ghana. *BMC Infect Dis*. 2019 Oct; 19(1):898.
177. Adhikari S, **Thakur N**, Shrestha U, Shrestha M, Shrestha MM, Thapa B, Poudel M, Kunwar AJ. Genetic Analysis of Children with Congenital Anomalies in Three Ecological Regions of Nepal: a Phase-II Nepal Pediatric Ocular Diseases Study. *BMC Med Genet*. 2020. (Accepted).
178. Biswas S K, M Alam M, **Roy S**, Ahmed T, Bhuyan AM. a Newer Technique of Ocular Surface Reconstruction in Unilateral Lime Burn. *Ophthalmic Horizon*. 2019; 14:7-15.
179. Buda G, **Vishnopolska SA**, Biagioli G, Marti MA, Turjanski A. Genomic Techniques Applied to Reproductive Biology. *SAEGRE*. 2019 Jan-Jun; XXVI(1):59- 65.
180. Burimuah V, Sylverken A, **Owusu M**, El-Duah P, Yeboah R, Lamptey J, Frimpong YO, Agbenyega O, Folitse R, et al. Sero-Prevalence, Cross-Species Infection and Serological Determinants of Prevalence of Bovine Coronavirus in Cattle, Sheep and Goats in Ghana. *Vet Microbiol*. 2020 Feb; 241:108544.
181. Calandra CR, Buda G, **Vishnopolska SA**, Oliveri J, Olivieri FA, Millán MIP, Biagoli G, Miquelini LA, Pellene AL, et al. Spastic Ataxia with Eye-Of-The-Tiger-Like Sign in 4 Siblings

- Due to Novel Compound Heterozygous AFG3L2 Mutation. *Parkinsonism & Related Disorders*. 2020 Apr; 73:52-54.
182. De Souza TR, Ladeira BL, Quinelato V, **Casado PL**, Rezende VA, Granjeiro JM, Góes, C. Evaluation of Genetic Risk Related to Catechol-O-Methyltransferase (COMT) and B2-Adrenergic Receptor (ADRB2) Activity in Different Diagnostic Subgroups of Temporomandibular Disorder in Brazilian Patients. *Int J Oral Maxillofac Surgery*. 2020; 49(2):237-243.
 183. Delhomme TM, **Avogbe PH**, Gabriel AAG, Alcalá N, Leblay N, Voegelé C, Vallée M, Chopard P, Chabrier A, et al. Needlestack: An Ultra-Sensitive Variant Caller for Multi-Sample Next Generation Sequencing Data. *NAR Genom Bioinform*. 2020 Jun; 2(2):Lqaa021. <https://doi.org/10.1093/nargab/lqaa021>.
 184. **Dhoro M**, Dingswayo P, Kaisi D, Mujuru HA. Structural Congenital Birth Defects in New-Born Babies at Parirenyatwa Group of Hospitals, Harare, Zimbabwe. *Plos Genet*. 2020. (Submitted).
 185. Dobrevski B, Shukarova- Angelovska E, Kirijas M, Milanovski G, Brnjarchevska Blazhevaska T, Boceska F, **Petlichkovski A**. Detection of a Rear Mutation in a Noonan Syndrome Suspected Patient: a Case Report. *MEBM*. 2020; 3(1):44-47.
 186. Dumo AM, Laing B, Lim AG, Palaganas EC, **Abad PJB**, Valdehueva O, Palovaara M, Saunders H, Estola M, Et Al. Randomized Controlled Trial on the Effectiveness of Web-Based Genomics Nursing Education Intervention for Undergraduate Nursing Students: a Study Protocol. *J Adv Nurs*. (In Press).
 187. Fachal L, Aschard H, Beesley J, Barnes DR, Allen J, **Torres-Mejía G**, Troester MA, Truong T, Tung N, et al. Fine-Mapping of 150 Breast Cancer Risk Regions Identifies 191 Likely Target Genes. *Nat Genet*. 2020; 52(1):56-73.
 188. Farhat S, Zafar MU, Sheikh MA, Qasim CM, Urooj F, **Fatima SS**. Association of Resolvin Level in Pregnant Women with Preeclampsia and Metabolic Syndrome. *Taiwan J Obstet Gynecol*. 2020; 59:105-108.
 189. GBD Chronic Kidney Disease Collaboration (**Fatima SS**). Global, Regional, and National Burden of Chronic Kidney Disease, 1990-2017: a Systematic Analysis for The Global Burden of Disease Study 2017. *Lancet*. 2020; 395:709-733.
 190. Global Retinoblastoma Study Group (**Roy S, Yousef YA**). Global Retinoblastoma Presentation and Analysis by National Income Level. *JAMA Oncol*. 2020; 6(5):685-695.
 191. Gokhroo RK, Chandra K, **Nandal R**, Bisht DS, Gupta S, Kishor K, Avinash A, Pandey S, Roy R, et. al. the Initial Experience of 2495 Cases of the Ulnar Artery as Default Access for Coronary Diagnostic and Interventional Procedures at a Single Center: An Observational Study. *Indian Heart J*. 2020; 72(3):184-88.
 192. Hosen MI, Sheikh M, Zvereva M, Scelo G, Forey N, Durand G, Voegelé C, Poustchi H,... **Avogbe PH**, et al. Urinary TERT Promoter Mutations are Detectable Up to 10 Years Prior to Clinical Diagnosis of Bladder Cancer: Evidence from the Golestan Cohort Study. *Ebiomedicine*. 2020 Mar; 53:102643.
 193. **James O**, Erinoso OA, Ogunlewe AO, **Adeyemo WL**, Ladeinde AL, Ogunlewe MO. Parental Age and the Risk of Cleft Lip and Palate in a Nigerian Population: A Case-Control Study. *Ann Maxillofac Surg*. 2020. (Accepted).
 194. Kaul A, Gokhroo RK, Tarik MT, Chandra K, **Nandal R**, Ashish K, Goyal M, Chandra S. 1 Year Follow Up Results of ARTIM HF Trial. *J Am Coll Cardiol*. 2020; 75:11.

195. LBD Double Burden of Malnutrition Collaborators (**Fatima SS**). Mapping Local Patterns of Childhood Overweight and Wasting in Low- and Middle-Income Countries Between 2000 and 2017. *Nat Med*. 2020; 6(5):750-759.
196. Liu J, Prager-Van Der Smissen WJC, Collée JM, Bolla MK, Wang Q, **Torres-Mejía G**, Dörk T, Swerdlow AJ, Hamann U, et al. Germline HOXB13 Mutations P.G84E and P.R217C Do Not Confer an Increased Breast Cancer Risk. *Sci Rep*. 2020 Jun; 10(1):9688.
197. Lucas RRS; Martins CCP, Oliveira HE, Cordeiro BQS, Machado A, **Casado PL**, Aguiar T, Tristao GC. Evaluation of the Success Rates of Immediate Implant Placed in Anterior and Posterior Regions: a Retrospective Study. *Revista Científica Do CRORJ*. 2019; 4:2 -9.
198. **Mahfoudh W**, Snoussi K, Gabbouj S, Remadi Y, Bouaouina N, Chouchane L and Zakhama A. FAS -670 A/G polymorphism: Association with breast cancer risk, progression and survival in the Tunisian population. 2020. (In preparation).
199. **Mahfoudh W**, Snoussi K, Gabbouj S, Remadi Y, Bouaouina N, Chouchane L and Zakhama A. FASL-844 T/C polymorphism: a biomarker of good prognosis of breast cancer in the Tunisian population. 2020. (In preparation).
200. **Mehrez MI**, Hassib N, **Sayed I**, Aboul-Ezz E, Ramzy MI, ... **Otaify G**, El-Husseini R, Esmail AM, El-Desouky D, et al. Genetic Syndromes with Premature Loss of Teeth: a Retrospective Study and a Suggested Classification. *Middle East J Med. Genet*. 2019; 8:100-106.
201. **Mehrez MI**, Magdi P, Morford L, Hartsfield J. a Glimpse at Genetic Counseling in Egypt. 2020. (In Preparation).
202. Mehyar M, Mosallam M, Tbakhi A, Saab A, Sultan I, Deebajah R, Jaradat I, Aljabari R... **Yousef YA**. Impact of RB1 Gene Mutation Type in Retinoblastoma Patients on Clinical Presentation and Management Outcome. *Hematol Oncol Stem Cell Ther*. 2020; 13(3):152–159.
203. Mohammad M, Nazzal RM, Alhussaini M, Abdullah N, Saleh A, Muhsen S, Rejdak R, **Yousef YA**. Primary Apocrine Adenocarcinoma of the Orbit with Lacrimal Sac Invasion. *Ophthalmol J*. 2020; 5:38–41.
204. Mustafa A, Tekka FS, **Hitayezu J**, Reel B, Kumar R. Severe Impact of Covid-19 Among Children in Sub-Saharan Africa? *J Intensive Care*. 2020. (Submitted).
205. Nuruddin M, **Roy S**, Hoque F. Management of Centurian Syndrome: An Unexplained Cause of Epiphora. *Ophthalmic Horizon*. 2019; 14:27-30.
206. Oliver J, Quezada Urban R, Franco Cortés CA, Díaz Velásquez CE, Montealegre Paez AL, Pacheco-Orozco RA, Castro Rojas C, Garcia-Roblesr, ... **Torres Mejía G**, et al. Latin American Study of Hereditary Breast and Ovarian Cancer LACAM: a Genomic Epidemiology Approach. *Front Oncol*. 2019 Dec; 9:1429.
207. **Otaify GA**. Bone-Specific Therapeutic Modalities for Genetic Skeletal Diseases. *Middle East J Med. Genet*. 2019; 8:69-82.
208. **Owusu M**, Acheampong G, Annan A, Acheampong G, Annan A, Marfo KS, Osei I, Amuasi J, Sarpong N, et al. Ralstonia Mannitolilytica Sepsis: a Case Report. *J Med Case Rep*. 2019 Oct; 13(1):318.
209. Owusu-Dabo E, Acheampong G, **Owusu M**, Owusu-Ofori A, Osei I, Sarpong Nimako S, Park SE, Marks F. Et Al., Identification of Multidrug-Resistant Salmonella Plasmid Variants Using Polymerase Chain Reaction (PCR) Based Replicon Typing Technique in Ghana. *Africa Health Agenda International Journal*. 2019 Dec; 1:6.
210. Özdek S, Altıntop M, **Tuncay Yaylacioglu F**, Arıbaş Y, Ozdemir HB, Gürelik G. Evaluation of Central and Peripheral Retinal Vascular Changes in the Fellow Eyes of Patients with Unilateral Retinal Vein Occlusions. 2020. (In Preparation).

211. Palencia-Campos A, Martínez-Fernández ML, Altunoglu U, Soto-Bielicka P, Torres A, Marín P, Aller E, Şentürk L... **Otaify GA**, et al. Heterozygous Pathogenic Variants in *GLI1* are a Common Finding in Isolated Postaxial Polydactyly A/B. *Human Mutation*. 2020; 41:265-276.
212. Park SE, Toy T, Cruz Espinoza LM, Panzner U, Mogeni OD, Im J, Poudyal N, Pak GD... **Owusu M**. the Severe Typhoid Fever in Africa Program: Study Design and Methodology to Assess Disease Severity, Host Immunity, and Carriage Associated with Invasive Salmonellosis. *Clin Infect Dis*. 2019 Oct; 30:69(Suppl6):S422-S434.
213. Penon-Portmann M, **Lotz-Esquivel S**, Chavez Carrera A, Jiménez-Hernández M, Alvarado-Romero D, Segura-Cordero S, Rimolo-Donadio F, Hevia- Urrutia F, Mora-Guevara A, et al. Wilson Disease in Costa Rica: Pediatric Phenotype and Genotype Characterization. *JIMD Reports*. 2020; 52:55-62.
214. Rodríguez Martínez K, Ramírez Blanco D, **Benítez Cordero Y**, Gutierrez Gutierrez R. Sperm DNA Fragmentation in Couples Treated By in Vitro Fertilization. *Rev Cubana Endoc* 2019; 309(3):e194867.
215. **Roy S**, Nuruddin M. Outcome of Early Treatment with Oral Propranolol on Periocular Capillary Haemangioma on Outpatient Basis. *Ophthalmic Horizon*. 2020. (Submitted).
216. Rutagumba D, Kalimba E, **Hitayezu J**. Predictors of Exclusive Breastfeeding in Urban Kigali, Rwanda. a Cross-Sectional Study. *Rwanda Med J*. 2020. (Accepted).
217. Shieh Y, Fejerman L, Lott PC, Marker K, Sawyer SD, Hu D, Huntsman S, Torres J... **Torres-Mejía G**, et al. a Polygenic Risk Score for Breast Cancer in US Latinas and Latin American Women. *J Natl Cancer Inst*. 2020 Jun; 112(6):590-598.
218. Sotomayor Lugo F, Corbacho Padilla JM, Valiente Linares AM, **Benítez Cordero Y**, Viera González T. General Aspects About the Structure of Severe Acute Respiratory Syndrome Coronavirus 2 (SARS- Cov-2). *Rev Cubana Invest Bioméd* 2020; 39(3):e867.
219. Tomar AS, Finger PT, Gallie B, Mallipatna A, Kivelä TT, Zhang C, Zhao J, Wilson MW,..., **Yousef YA**, et al. a Multicenter, International Collaborative Study for AJCC-Staging of Retinoblastoma Part I: Metastasis-Associated Mortality. *Ophthalmology*. 2020. <https://doi.org/10.1016/J.Ophtha.2020.05.050>.
220. Tomar AS, Finger PT, Gallie B, Mallipatna A, Kivelä TT, Zhang C, Zhao J, Wilson MW,..., **Yousef YA**, et al. a Multicenter, International Collaborative Study for AJCC-Staging of Retinoblastoma: Treatment Success and Globe Salvage. *Ophthalmology*. 2020; S0161-6420(20):30524-30528.
221. **Tuncay Yaylacioglu F**, Güntekin Ergün S, Öner A, Turan A, Ozmert E, Ali Ergun M, Ozdek S. Inherited Eye Diseases in Turkey: Current Approaches and Future Directions. *Am J Med Genet Part C: Seminars in Medical Genetics*. 2020. (Accepted).
222. **Tuncay Yaylacioglu Y**, Talim B, Kayman Kürekçi G, Dinçer PR. Evaluation of Phenotypic Effects of Genome Editing Mediated TGFBI Variation on Zebrafish Cornea. 2020. (In Preparation).
223. **Vishnopolska SA**. *Journal of Science Humanities and Art (JOSHA, an Open Book)*. 2020 Jun; 7:3. <https://doi.org/10.17160/josha.7.3.676>.
224. **Yousef YA**, Al-Nawaiseh I, Mehayar M, Sultan I, Al-Hussaini M, Jaradat I, Mohammad M, Aljabari R, Abu-Yaghi N, et al. How Telemedicine and Centralized Care Changed the Natural History of Retinoblastoma in a Developing Country: Analysis of 478 Patients. *Ophthalmology*. 2020. (In Press).
225. **Yousef YA**, Alkhoms A, Aljabari R, Aljboor M, Mohammad M, Lahlouh M, Deebajah R, Halalsheh H, Al-Hussaini M, et al. Programmed Screening for Retinoblastoma Enhances Early

Diagnosis and Improves Management Outcome for High-Risk Children_ Ophthalmic Genet. 2020; 41(4):308-314.

226. **Yousef YA**, Elrimawi AH, Nazzal RM, Qaroot AF, Alaref AH, Mohammad M, Abureesh O, Rejdak R, Nowomiejska K, et al. Coats' Disease: Characteristics, Management, Outcome, and Scleral External Drainage with Anterior Chamber Maintainer for Stage 3b Disease. Case Report. Medicine (Baltimore). 2020; 99(16):e19623.
227. **Yousef YA**, Mohammad M, Jaradat I, Shatnawi R, Banat S, Mehyar M, Al-Nawaiseh I. the Role of External Beam Radiation Therapy for Retinoblastoma After Failure of Combined Chemoreduction and Focal Consolidation Therapy. Ophthalmic Genet. 2020; 41(1):20-25.
228. **Yousef YA**, Noureldin AM, Sultan I, Deebajah R, Al-Hussaini M, Shawagfeh M, Mehyar M, Mohammad M, Jaradat I, et al. Intravitreal Melphalan Chemotherapy for Vitreous Seeds in Retinoblastoma. J Ophthalmol. 2020 Jan; 2020:8628525.
229. Yüksel E, Akçam HT, **Tuncay Yaylacioglu Y**, Yüksel N, Akata FR. A New Surgical Technique for Severe Ocular Surface Burns: Full Ocular Surface Transplantation. Indian J Ophthalmol. 2020. (In Press).
230. Zavala M, González M, **Sanheuz Díaz C**. Hallazgo Incidental En Estudio De Secuenciación Exómica. (Incidental Finding in Exomic Sequencing Study). Genética Médica Y Genómica. 2020; 4(4):13-17.

2016 Batch

231. Adekile A, Anie K, Ben Hamda C, Brown B, Bukini D,... **Malasa L**, Masekoameng T, Mazandu G, Mnika K, Et Al. the Sickle Cell Disease Ontology: Enabling Universal Sickle Cell-Based Knowledge Representation. Database. 2019; 2019:1-12.
232. Adekunle AA, Adamson OO, **James O**, Ogunlewe MO, Butali A, **Adeyemo WL**. Breast Feeding Practices Among Mothers of Children with Oro-Facial Cleft in an African Cohort. Cleft Palate Craniofac J. 2020; 57(8):1018-1023.
233. Adekunle AA, **James O**, **Adeyemo WL**. Health Information Seeking Through Social Media and Search Engines by Parents of Children with Orofacial Cleft in Nigeria. Cleft Palate Craniofac J; 2020; 57:444-447.
234. **Adeoye AM**, Fakunle A, Aderonmu O, Tayo B. Short-Term Exposure to Household Air Pollution and Risk of Hypertension Among Adults: a Pilot Study in Ibadan. Journal of Health and Environmental Research. 2020; 6(2):37-43.
235. **Adeyemo WL**, **James O**, Oladega AA, Adamson OO, Adekunle AA, Olorunsola KD, Busch T, Butali A. Correlation Between Height and Impacted Third Molars and Genetics Role in Third Molar Impaction. J. Maxillofac Oral Surg. 2020. <https://doi.org/10.1007/S12663-020-01336-9>.
236. **Ariani Y**, Priambodo R, Hafifah CN, Sjarif DR. Identification of a Deletion Variant in Exon 9 of Iduronate 2-Sulfatase Gene in Patients with Type II. J Natural Sc Biol Med. 2019; 10(3):38-42.
237. **Ariani Y**, Priambodo R, Sjarif DR. Identification of a Novel Variant in Exon 5 of Galactosamine (N-Acetyl)-6-Sulfatase (GALNS) Gene in Mucopolysaccharidosis IVA Patients in Indonesia. J Natural Sc Biol Med. 2019; 10(3):99-102.
238. **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 Natural Sc Biol Med. 2019; 10(3):109-112.
239. Asgari S, Luo Y, Belbin Y, Bartell E, Slowikowski K, Contreras C, O Connor T, Dietz H, **Guio H**, et al. a Positively Selected, Common, Missense Variant in FBN1 Confers a 2.2 Centimeter Reduction of Height in the Peruvian Population. Nature. 2020; 582 (7811):234-239.

240. Baldev V, **Tibrewal S**, Rath S, Ganesh S. Isolated Abducens Nerve Palsy Following Temporal Bone Fracture in a Child. *Taiwan J Ophthalmol*. 2020. https://doi.org/10.4103/Tjo.Tjo_75_19.
241. Baldev V, **Tibrewal S**. Anomalous Orbital Structure Mimicking Fracture of Orbital Floor. *J AAPOS*. 2020; S1091-8531(20)30105-1.
242. Borda V, Alvim I, Aquino MM, Silva C, Soares-Souza GB, Leal TP, Scliar MO, Zamudio R,...**Heinner G**. the Genetic Structure and Adaptation of Andean Highlanders and Amazonian Dwellers Is Influenced by the Interplay Between Geography and Culture. *Biorxiv*. 2020. <https://doi.org/10.1101/2020.01.30.916270>.
243. Brown P, RELISH Consortium (**Adeyemo AA**), Zhou Y. Large Expert-Curated Database for Benchmarking Document Similarity Detection in Biomedical Literature Search. *Database*. 2019; 1–67.
244. Bukini D, Mbekenga C, Nkya S, **Malasa L**, Mccurdy S, Manji K, Makani J, Parker M. Influence of Gender Norms in Relation to Child's Quality of Care: Follow-up of Families of Children with SCD Identified Through NBS in Tanzania. *J Community Genet*. 2020 Aug. <https://doi.org/10.1007/s12687-020-00482-4>.
245. Casavilca-Zambrano S, Cancino-Maldonado K, Jaramillo-Valverde L, **Guio H**. Epigenetics: the Relationship of the Environment and Mental Health. *Rev Neuropsiquiatr*. 2019; 82(4):266-273.
246. **Ekure EN**, Adeyemo AA, Sokunbi O, Kalu N, Martinez AF, Tekendo-Ngongang C, Olusegun-Joseph A, Ikebudu D, Berger SI, et al. Next Generation Sequencing and Congenital Heart Disease in Sub-Saharan Africa. *Circulation: Genomic and Precision Medicine*. 2020. (Submitted).
247. **Ekure EN**, Sokunbi O, Kalu N, Olusegun-Joseph A, Kushimo O, Amadi C, Hassan O, Ikebudu D, Onyia S, et al. Congenital Heart Disease in School Children in Lagos, Nigeria: Prevalence and the Diagnostic Gap. *Am J Med Genet C Semin Med Genet*. 2020; 184(1):47-52.
248. Feoktistova YF, Domech CR, **Roblejo Balbuena H**. Clinical and Molecular Studies of Wilson's Disease in Cuban Patients. *Rev. Annals of the Cuban Academy of Sciences. Ciencias Biomédicas*. 2019; 3.
249. Hall EG, Wenger LW, Wilson NR, Undurty-Akella SS, Standley J, Augustine-Akpan EA, Koussa YA, Acevedo DS, ...**Adeyemo WL**. SPECC1L Regulates Palate Development Downstream of IRF6. *Hum Mol Genet*. 2020; 29:845-858.
250. **Hussen DF**, Hammad SA, Refaat KM, Ashaat EA, Aglan MS, **Otaify GA**, El-Bassyouni HT, Temtamy SA. Chromosomal Aberrations and Chromosomal Heteromorphisms Among Young Couples with Recurrent Spontaneous Abortion. *Middle East J Med. Genet*. 2019; 8:48-54.
251. **Hussen DF**, Hammad SA, Refaat KM, Fayez AG, Aglan MS, **Otaify GA**, Temtamy SA. De Novo Mutation of NIPBL Gene in an Egyptian Patient with Cornelia De Lange Syndrome. 2020. (In Preparation).
252. **Hussen DF**, Kamel AK, Mekkawy M, El Ruby MO, Ashaat EA. Phenotypic and Molecular Cytogenetic Analysis of a Case of Monosomy 1p36 Syndrome Due to Unbalanced Translocation. *Mol. Syndromol*. 2020. (Accepted).
253. **Hussen DF**, Kamel AK, Mekkawy M, Mohamed AM, Zaki MS, Issa MY, El Ruby MO, Ashaat EA, Ismail S, et al. Cytogenetic Study of a Large Cohort of Patients with Corpus Callosum Abnormalities. *Middle East J Med Genet* 2019; 8:158-68.
254. **James O**, Erinoso O, Adamson OO, Sokunbi OJ, Agbogidi FO, Adekunle AA, Ogunlewe AO, **Ekure EN**, **Adeyemo WL**, et al. Risk of Congenital Cardiovascular Anomalies in Patients with Non-Syndromic Orofacial Cleft: A Preliminary Case-Control Study. *Niger J Clin Pract*. 2020. (Accepted).

255. Jaramillo-Valverde L, Levano K, Villanueva I, Mazzetti P, **Guio H**. Guillain-Barre Syndrome Outbreak in Peru: Association with Polymorphisms in IL-17, ICAM-1 and CD1. *Mol Genet Genomic Med.* 2019; 7(10):e00960.
256. Jaramillo-Valverde L, Vásquez-Domínguez A, Levano KS, Novoa-Bellota P, Machaguay-Romero M, Cano RJ, Solis RS, **Guio H**. Peruvian Mobile Laboratory for Ancient DNA Extraction. 2020. (In Preparation).
257. Jatto ME, **Adeyemo AA**, Ogunkeyede SA, Lagunju IA and Nwaorgu OG. Pediatric Hearing Thresholds Post- Bacterial Meningitis. *Front. Surg.* 2020; 7:36. <https://doi.org/10.3389/fsurg.2020.00036>.
258. Joseph ADD, **Sirisena ND**, Kumanan T, Sujanitha V, Strelow V, Yamamoto R, Wieczorek S and Dissanayake VHW. Hypoparathyroidism, Sensorineural Deafness and Renal Disease (Barakat Syndrome) Caused by a Reduced Gene Dosage in GATA3: a Case Report and Review of Literature. *BMC Endocr Disord.* 2019; 19:111.
259. Kehdy FSG, Pita-Oliveira M, Scudeler M, Torres-Loureiro S, Zolini C,...**Guio H**, Tarazon-Santos E, Leal TP, Rodrigues- Soares F, et al. Human-SARS-Cov-2 Interactome and Human Genetic Diversity: Tmprss2-Rs2070788, Associated with Severe Influenza, and Its Population Genetics Caveats in Native Americans. *Journal of Human Genetics.* 2020. (Submitted).
260. Kruszka P, Addissie YA, Tekendo-Ngongang C, Jones KL, Savage SK,...**Sirisena ND**, **Uwineza A**, **Duenas-Ropque M**, **Ekure EN**, et al. Turner Syndrome in Diverse Populations. *Am J Med Genet A.* 2020; 182(2):303-313.
261. **Lallar M**, Arora V, Saxena R, Puri RD, Verma IC. Complete Labyrinthine Aplasia: a Unique Sign for Targeted Genetic Testing in Hearing Loss. *J Pediatr Genet.* 2020; <https://doi.org/10.1055/S-0040-1708052>.
262. **Lallar M**, Mahay SB, Mandal K, Verma IC, Puri R. Clinical and Genetic Profile of RASopathies. 2020. (Submitted).
263. Massa JD, Arora V, **Lallar M**, Bijarnia-Mahay S, Puri RD. Current Status of Noninvasive Prenatal Testing and Counselling Considerations: An Indian Perspective. *J. Fetal Med.* 2020; 7:9–16.
264. Mateus V, Gouveia H, Borda V, Leal T, Moreira R, Aquinon M, **Guio H**, Yeager R, Tarazona-Santos E. Origins, Admixture Dynamics and Homogenization of the African Gene Pool in the Americas. *Mol Biol Evol.* 2020; 37(6):1647-1656.
265. Musanabaganwa C, Jansen S, Fatumo S, Rutembesa E, Mutabaruka J, Gishoma D, **Uwineza A**, Wildman D, Uddin M, et al. Burden of Post-Traumatic Stress Disorder in Postgenocide Rwandan Population Following Exposure To 1994 Genocide Against the Tutsi: a Meta-Analysis. *J Affect Disord.* 2020; 275:7–13.
266. Mutesa L, Ndishimye P, Butera Y, Souopgui J, **Uwineza A**, Rutayisire R, Musoni E, Rujeni N, Nyatanyi T, et al. a Strategy for Finding People Infected with SARS-Cov-2: Optimizing Pooled Testing at Low Prevalence. 2020. <https://arxiv.org/abs/2004.14934>.
267. Nwaze CE, Adebayo O, **Adeoye AM**, Akinmoladun V. Orofacial Clefts and Cardiovascular Risk and Diseases: the Causal Relationship and Associations: *Ann Ibd. Pg. Med.* 2020; 18 Cleft Supplement S28 - S34.
268. Ogun GO, Olusanya AA, Akinmoladun VI, **Adeyemo AA**, Ogunkeyede SA, Daniel A, Awosusi BL, Fatunla EO, Fasunla AJ, et al. Nasopharyngeal Carcinoma in Ibadan, Nigeria: a Clinicopathologic Study. *Pan Afr Med J.* 2020; 36:82.
269. Ogunkeyede SA, **Adeyemo AA**, Daniel A, Yaro PJ, Ogundoyin OA. Pharyngo-Cutaneous Fistula Post Total- Laryngectomy: a Local Experience. *Nig J Med.* 2020; 29(1):69-72.

270. Ogunsola FT, Odukoya OO, Banigbe B, Caleb-Adepoju SO, Folarin O, Afolabi BB, Okubadejo NU, **Adeyemo WL.** a Preprogram Appraisal of Factors Influencing Research Productivity Among Faculty at College of Medicine, University of Lagos. *Ann Afr Med.* 2020; 19:124-130.
271. **Okafor FU**, Imai OZ. Tramadol Use and Associated Predictors Among Adolescents Attending Tertiary Health Facility in Benin City, Edo State South-South Nigeria. *Ilorin Journal of Health, Physical Education and Recreation (IJOHPER).* 2020. (Accepted).
272. **Okafor FU**, Omorogbe CE, Edobhiye VE. Knowledge, Attitude and Practice of Food Hygiene Among Food Vendors in a Selected Tertiary Educational Institution in Benin City, Edo State Nigeria *Journal of Education, Health, and Technology Research (NJEHETR)* 2020. (Accepted).
273. **Okafor FU**, Omorogbe CE, Eweka A. Perception of Nursing Care Received in Tertiary Health Institution Among Ante-Natal and Post-Natal Women in Benin Metropolis, Edo State. *Journal of Science and Technology, John Harris Library University of Benin.* 2020. (Accepted).
274. **Okafor FU**, Omorogbe CE, Osian EA, Aniekan JE. Ocular Trauma Knowledge Among Public Primary School Teachers in Egor Local Government Area, Benin City, Edo State. *Lautech Journal of Nursing.* 2020. (Accepted).
275. Omorogbe CE, **Okafor FU**, Ekiomoado ET. Nurses' Perception of Asepsis and the Practice of Aseptic Techniques in the Tertiary Health Facility, Benin City, Edo State. *Nigeria Journal of Nursing.* 2020. (Accepted).
276. Oyelakin OA, **Adeyemo AA.** Audit of Open Pediatric Tracheostomies at the University College Hospital, Ibadan. *Niger J Med.* 2020; 29:256-60.
277. Prabodha LBL, Paththinige CS, Jayawardana SMA, **Sirisena ND**, Dissanayake VHW. Host Genetic and Epigenetic Factors Affecting the Susceptibility and Outcome of Severe Coronavirus Infections. *Front. Genet.* 2020. (Submitted).
278. Rai A, Mandal K, Saxena D, **Lallar M**, Phadke SR. Distal Arthrogryposis: a Clue To the Etiology of Neonatal Cholestasis. *Scientific Letter, Indian J Pediatr.* 2020; 87(10):869-870.
279. Ratna Ria, **Tibrewal S.** Pedigree Charting in Ophthalmology. *Nepal Journal of Ophthalmology.* 2020. (Submitted).
280. **Robledo Balbuena H**, Marcheco Teruel B, Monzón Benítez G, Marín Padrón L. CYP2D6*3, *4, *5 and *6 Allelic Variants in a Sample of Cuban Patients with Schizophrenia. *Revista Cubana De Genética Comunitaria.* 2020. (Submitted).
281. Sahin E, İnciser Paşalak Ş, **Seven M.** Consanguineous Marriage and its Effect on Reproductive Behavior and Uptake of Prenatal Screening. *J Genet Couns.* 2020; 29(5):849-856.
282. Sarfo FS, Ovbiagele B, Gebregziabher M, Akpa O, Akpalu A,...**Adeoye AM**, Olugbo O, Ogunjimi L, Osaigbovo G, Et.Al. Unraveling the Risk Factors for Spontaneous Intracerebral Hemorrhage Among West Africans. *Neurology.* 2020; 94(10):e998-E1012.
283. Scliar MO, Sant Anna HP, Santolalla ML, Leal TP, Araújo N,...**Guio H**, Hsing AW, Mbulaiteye SM, Mensah J, et al. Admixture/Fine-Mapping in Brazilians Reveals a West African Associated Potential Regulatory Variant (Rs114066381) with a Strong Female-Specific Effect on Body Mass- and Fat Mass-Indexes. *Biorxiv.* 2019. <https://doi.org/10.1101/827311>.
284. **Seven M**, Shah LL, Daack-Hirsch S, Yazici H. Experiences of BRCA1/2 Gene Mutation-Positive Women with Cancer in Communicating Genetic Risk To Their Relatives. *Cancer Nurs.* 2020; 10.1097/NCC.0000000000000796.
285. **Seven M**, Shah LL, Daack-Hirsch S, Yazici H. From Proband To Relatives; Communication of Genetic Risk for Hereditary Breast-Ovarian Cancer and Its Influence on Subsequent Testing, *Cancer Nurs.* 2020. (Accepted).

286. Sharma M, Ganesh S, **Tibrewal S**, Sabharwal S, Sachdeva N, Adil M, Chaudhary J, Siddiqui Z. Accuracy of Noncycloplegic Photorefractive Error Using Spot Photoscreener in Detecting Amblyopia Risk Factors in Preschool Children in An Indian Eye Clinic. *Indian J Ophthalmol*. 2020; 68:504-509.
287. **Sirisena ND**, Biswas K, Sullivan T, Stauffer S, Cleveland L, Southon E, Dissanayake VHW, Sharan SK. Functional Evaluation of Five BRCA2 Unclassified Variants Identified in a Sri Lankan Cohort with Inherited Cancer Syndromes Using a Mouse Embryonic Stem Cell-Based Assay. *Breast Cancer Res*. 2020; 22:43.
288. **Sirisena ND**, Dissanayake VHW. Genomic Medicine Training - Updates from Sri Lanka. *Global Genomic Medicine Collaborative (G2MC) Quarterly Newsletter*. 2020 Apr.
289. **Sirisena ND**, Dissanayake VHW. Implementing Genomic Medicine in Resource-Constrained Settings: An Update Report from Sri Lanka. *Global Genomic Medicine Collaborative (G2MC) Quarterly Newsletter*. 2019 Dec.
290. **Sirisena ND**, Dissanayake VHW. Navigating Breast Cancer in the Post-Genomic Era. *SLMA NEWS - Official Newsletter of the Sri Lanka Medical Association*, 2019 Oct: 12(10):6-8.
291. **Sirisena ND**, Dissanayake VHW. Strategies for Genomic Medicine Education in Low- and Middle-Income Countries. *Front. Genet*. 2019; 10:944.
292. **Sirisena ND**, Samaranyake UMJE, Malalasekera AP, Dissanayake VHW. High Frequency of Klinefelter Syndrome in a Cohort of Sri Lankan Males with Non-Obstructive Azoospermia and Oligozoospermia. *Sri Lanka Journal of Surgery*. 2019; 37(4):11–13.
293. **Sirisena ND**. Genetic Testing to Help Prevent and Reduce Breast Cancer Risk - *Mediscene Section, Sunday Times Newspaper*. 2020 Feb.
294. Sokunbi OJ, Okoromah CAN, **Ekure EN**, Olawale OA, Eke WS. Electrocardiographic Pattern of Apparently Healthy African Adolescent Athletes in Nigeria. *J Cardiovasc Electrophysiol*. 2020. (Submitted).
295. Subasinghe CJ, Gunatilake SSC, **Sirisena ND**, Berge KE, Leren TP, Bulugahapitiya U, Dissanayake VHW. Hypokalaemic Paralysis - a Double Trouble from Concurrent Thyrotoxicosis and Gitelman Syndrome: a Report of Two Cases. *Sri Lanka Journal of Diabetes Endocrinology and Metabolism*. 2020; 10(1):26–29.
296. Sweat YY, Sweat M, Mansaray M, Cao H, Eliason S, **Adeyemo WL**. Six2 Regulates Pax9 Expression, Palatogenesis and Craniofacial Bone Formation. *Dev Biol*. 2020; 458:246-256.
297. Tekendo-Ngongang C, Owosela B, Addissie YA, **Lotz-Esquivel S**, **Hussen DF**, **Uwineza A**, **Sirisena ND**, **Llamos-Paneque A**, **Ekure EN**, et al. Rubinstein-Taybi Syndrome in Diverse Populations. *Am J Med Genet A*. 2020. (Submitted).
298. **Tibrewal S**, Sharma M, Rath S, Ganesh S. Extra-Large V Pattern in Exotropia: a Rare Case and Its Management. *Strabismus*. 2020; 28(2) 91-96.
299. **Tibrewal S**, Subedar K, Sen P, Mohan A, Shivanand S, Shah C, Nischal KK, Ganesh S, Bodhya Eye Consortium. Clinical Spectrum of Non-Syndromic Microphthalmos, Anophthalmos and Coloboma (MAC) in the Pediatric Population – a Multicentric Study from North India. *BJO*. 2020. (Accepted).
300. **Uwineza A**, **Hitayezu J**, Murorunkwere S, Ndinkabandi J, Dushimirimana G, Kwihangana F, Niyirera S, Niyomugabo O, Uwacu B, et al. Clinical Characterization of Cytogenetic Abnormalities in Children with Global Developmental Delay/Intellectual Disability with Epilepsy. *Rwanda Journal of Medicine and Health Sciences (RJMHS)*, 2019. (Submitted).

301. Valdivia-Silva J, Perez L, Flores L, Malaga M, **Guio H**. Development of an Accessible and Low-Cost Micro-Fluidic System (Lab-On-A-Chip) for Detecting Circulating Breast Cancer Tumor Cells. *Acta Med Peru*. 2020; 37(1):40-47.
302. Vásquez-Domínguez A, Jaramillo-Vaverde L, Levano K, Novoa-Bellota P, Machaguay-Romero M, Solis R, **Guio H**. Comparative Analysis of Gut Microbiome in Individuals of the Old Civilization of Caral-Supe Based on Data from 16S Rrna and ITS Region. *Biorxiv*. 2020. <https://doi.org/10.1101/2020.04.24.060582>.
303. Verma IC, **Lallar M**, Arora V. Looking Back at Fetal Medicine in India in 2018 and Looking Forward to 2019. *J. Fetal Med*. 2019; 6:47–50.
304. Yareta J, Galarza M, Capristano S, Pellón O, Sánchez C, Ballon C, **Guio H**. Differential Expression Analysis of Circulating Micrnas in Patients with Active Tuberculosis and Infected with Latent Tuberculosis. *Rev Peru Med Exp Salud Publica*. 2020; 37(1):51-56.

B. Grants: (Total - 89; 2019- 26; 2018- 33; 2017- 15; 2016- 15)

2019 Batch

1. **Abubakar S** (PI). Improving Genomic Literacy and Promoting Considerations for The Ethical, and Social Implications (ELSI) of Genomic Research in Nigeria. 2019. (African Postdoctoral Training Initiative of the African Academy of Sciences; Submitted).
2. Abuzaid M (PI), Ahmed N (Co PI), Mostafa M, Abdelfattah M, Zaki S, Abd-Allah M, **Sayed I**. Studying the Role of Serine Proteases in Periodontitis in Papillon Lefevre Syndrome. 2019. (National Research Centre; Received).
3. Alcausin MML, **Tumulak MJR**, Silao CL, Fabella TD. Clinical Characterization and Identification of CFTR Gene Mutations in Newborns with Positive Screen for Cystic Fibrosis in the Expanded Newborn Screening Program. 2020. (Newborn Screening Reference Center, Received).
4. Badea R (PI), Covaliu B, **Pirlog R**. Academy of Research Entrepreneurs in Medicine. 2020. (Iuliu Hatieganu University of Medicine and Pharmacy; Renewed).
5. Berindan Neagoe I (PI), **Pirlog R** (Co-PI). Prioritizing Oncologic Patient Access to Precision Therapy Through Digital Technologies in the COVID19 Pandemic - Oncoaccess. 2020. (Iuliu Hatieganu University of Medicine and Pharmacy; Submitted).
6. Buzoianu AD, Berindan Neagoe I, **Pirlog R**. MEDU-Learn: Adapting Medical Educational Tools and Practices to New Frameworks Generated by COVID -19. 2020 (Iuliu Hatieganu University of Medicine and Pharmacy; Submitted).
7. Chatterjee G (PI), **Mutreja D** (Co-PI). Comparison of Hematologic Scoring System with Serologic Markers in Cases of Sepsis in Infants & Children. 2020-2021. (MUHS Short Term Research Grant; Received).
8. Chennath AC (PI), **Mutreja D** (Co-PI). To Measure and Study the Blood Monocyte to Lymphocyte Ratio (MLR) in HIV Patients with and Without Active Tuberculosis (TB) and Establish a Relationship if Any to Use it as an Indirect Diagnostic Marker for TB. 2019-2020. (AFMC Alumni Research Grant; Received).
9. Ferracin M (PI), Berindan Neagoe I, **Pirlog R**. Molecular Characterization and Isolation of Circulating Tumor Cells in Metastatic Cancers: Diagnostic and Therapeutic Opportunities. 2020. (Iuliu Hatieganu University of Medicine and Pharmacy; Submitted).

10. Hassib N (PI), **Sayed I** (Co PI). An Attempt to Diagnose Typical and Atypical Cases by Detection Genes Causing Tooth Structure Anomalies. 2019. (National Research Centre; Received).
11. **Iqbal M** (PI), Khursheed M, Shahzad M. Identification, Characterization and Expression Analyses of DNA Variants Causing Otitis Media in Pakistani Population. 2020. (National Research Program for Universities/Higher Education Commission of Pakistan; Submitted).
12. Irimie a (PI), Berindan Neagoe I, **Pirlog R**. Development of Entrepreneurial Skills Among UMF Cluj-Napoca Students Through the Development of Experiential Educational Modules. 2020. (Iuliu Hatieganu University of Medicine and Pharmacy; Received).
13. **López-Star B** (PI), Hurn LA, Hufnagel RB. Ophthalmic Genetics Fellowship. 2020. (NEI/NIH; Submitted).
14. **Mutreja D** (PI), Venkatesan S (Co-PI), Sharma S (Co-I), Tilak TVSVGK (Co- I), Boruah D (Co-I). a Pilot Study to Evaluate the Role of Vascular Endothelial Growth Factor and its Comparison with Microvessel Density in Angiogenesis of Hematological Malignancies. 2019-2022. (Armed Forces Medical Research Grant; Received).
15. Ng WL (PI), **Hussein N** (Co-PI), Lee YK, Ng CJ, Kee BP, Malik TFA, Qureshi N, Then SM, Kwan Z. HLA-B*5801 Testing Among Patients with Gout to Prevent Allopurinol-Induced SCAR in Primary Care. 2020. (University Malaya Specialist Centre Care Fund; Received).
16. Nnodu OE (PI), Menzel S, Adekile AD, Agumadu UN, **Alli LA**. the Genetic and Clinical Variability of Sickle Cell Disease in Nigeria. Thematic Area: Science, Technology and Innovation. 2019. (Tertiary Education Trust Fund; Submitted).
17. Ogedengbe JO (PI), Odili AN, Mojiminiyi F, Sabo AM, Nwegbu MM, **Alli LA**, Agida T, Singh S, Isiguzo G. Urinary Proteomics in Predicting Preeclampsia in Nigeria. 2020. (National Research Fund; Submitted).
18. **Okunola OO**, Adedeji A. Pilot Initiative for Newborn Screening in Nigeria. 2020. (Tertiary Education Trust Fund; Submitted).
19. Olawoye O (PI), Ashaye a (Co-PI), Ajunwon a (Co-PI), **Monye HI** (PM). a Novel Community Engagement Program to Improve Glaucoma Screening and Management in High Risk Populations in Nigeria. 2019. (Novartis Pharma AG- XOVA Excellence in Ophthalmology Vision Award 2019; Submitted).
20. **Paredes-Moscossa SR** (PI), Villegas-Llerena C, Guevara-Fujita ML, Buleje J, **Tumulak MK**, Carmencita P, Fujita R, Sandoval JR, Dueñas-Roque M, et al. Ancestry and Interpretation of Genetic Variants from Peruvian Patients with Hereditary Breast Cancer Using Whole Exome Sequencing (WES): Advances in Genomic Medicine in Peru. 2020. (Universidad De San Martín De Porres; Received).
21. **Paredes-Moscossa SR** (PI), Villegas-Llerena C, Ibanez AJ, Acosta O, De Leon J, Yabar C, Kim YH, Menon M, Fujita R. Multiomic Research Platform for Detection of Immune Prognostic Biomarkers Based on Immune Profiling and Clinical Outcome of Peruvian COVID-19 Patients. 2020. (UK Research & Innovation- Global Effort on COVID-19 Health Research; Submitted).
22. Pop L (PI), Berindan Neagoe I, **Pirlog R**. Emergency Development of Molecular Tools for Emergency Assessment and Reappearance COVID-19. 2020. (Iuliu Hatieganu University of Medicine and Pharmacy; Received).
23. Sandoval JR (PI), Estrada A, Villegas-Llerena C, Guevara-Fujita ML, Acosand O, **Paredes-Moscossa SR** (Co-I), Riva L, Castro-Gamero MA, Fujita R, et al. Project 1000 Peruvian Genomes for The Bicentennial: Phase I - 200 Genomes in Cancer and Rare Diseases. 2019. (CONCYTEC; Submitted).

24. Teiosanu a (PI), **Pirlog R** (Co-PI). Supporting Vulnerable Groups in Botoşani County Through Information Actions, Clinical-Psychological Evaluation and Provision of Hygienic-Sanitary Materials. 2020. (Asociatia Nord; Received).
25. **Wiafe SA** (PI), Baynam G (Co-PI), Yarlalu T (Co-I), Addo-Lartey EO (Co-I). Implementing Lyfe Language Project in Ghana; Translating the Human Phenotype Ontology into Indigenous Ghanaian Languages. 2020. (Genetics Services of Western Australian Health Department; Received).
26. **Wiafe SA** (PI). the Undiagnosed Disease Program in Ghana. 2020. (Illumina Laboratories; Received).

2018 Batch

27. Abou-Elenein M (PI), Nasr M (Co-PI), Khalil A, **Messaoud O**. Exploring the Antiviral Properties of Nanoparticles on Corona Virus Strain. 2020. (German Federal Ministry of Education and Research Fund- AGYA; Accepted).
28. **Adewole OO** (PI), Awopeju FO (Co-PI), Adewole TO (Co-PI), Adedigba M (Co-PI). Cytokine Profile and Kinetics in COVID-19 Among Nigerians. 2020. (Tertiary Education Trust Fund; Submitted).
29. **Adewole OO** (PI), Vellore a (Co-PI). Statins as Treatment Adjunct in Active Pulmonary Tuberculosis. 2020. (Cure Within Reach; Submitted).
30. **Adewole OO** (PI). Innovative Approaches to Improving TB Case Detection. Grant Challenges Africa Phase II. 2020. (African Academy of Sciences; Submitted).
31. Afolabi BB (PI), Babah O, **Adeyemo TA** (Co-I). Low Dose Aspirin for Preventing Intrauterine Growth Restriction and Preeclampsia in Sickle Cell Pregnancy (PIPSICKLE): a Randomised Controlled Trial. 2020-2022. (Tertiary Education Trust Fund NR; Received).
32. Alatise Olusegun a (PI), Peter Kingham P (Co-PI), **Abdulkareem FB** (Site-PI), Sullivan R, Vanderpuye V, Ramesar R, Aggarwal A, Mutebi MC, Parham G. Advancing African Leadership in Cancer Training & Research: Prevention, Early Diagnosis and Treatment. 2020. (DELTAS; Submitted).
33. **Dutta AK** (PI), Goswami K (Co-PI), Biswas N (Co-I), Basu a (Co-I). Estimation of Birth Prevalence and Carrier Frequency of Autosomal Recessive Inborn Errors of Metabolism in India from Publicly Available Next-Generation Sequence Data. 2020. (Science & Engineering Research Board, Ministry of Science and Technology, Government of India; Submitted).
34. **Gitaka J** (PI), Pamme N, Klapperich C, **Hotah JE**, Kimotho J, Kirira P, Mungai S, Onono M, Raju S, Wade D, Ntinginya NE, Gatungu D, et al. Afropoc – African Network for Point-Of Care Diagnostics to Tackle Endemic and Emerging Infectious Diseases. 2020. (African Academy of Sciences- Deltas II Program; Submitted).
35. Guizani I (PI), **Messaoud O** (Task Leader). Assess, Predict, Act: Federated Strategy for The Fight Against COVID-19. 2020-2022. (Tunisian Ministry of Higher Education and Scientific Research- Federated Research Projects; Accepted).
36. Haddad S (PI), Charfi-Kaddour S (Co-PI), **Messaoud O** (Team Lead), Gargouri M, Drissi LB (PI), El Hassan S (Co-PI), Khalid R, Rachid Ahll, et al. Advanced Materials: a Strategic Choice for Energy, Environment and Health. 2020-2022. (Tunisian-Moroccan Bilateral Fund- Research and Development Projects; Accepted).
37. Hou, L (PI), Sagay A, Ogunsola FT, Murphy M, Adeyemo WL (Co-I), **Abdulkareem FB** (Co-I). Epigenomic Biomarkers of HIV-Associated Cancers in Nigeria. 2019. (NIH-U54; Continued).

38. Kalra S (PI), **Dwivedi a** (Co-PI). Conduct Research Project Entitled Genotype- Phenotype Correlations in Children with Renal Tubular Acidosis. 2020. (Indian Council of Medical Research; Accepted).
39. Kefi R (PI), **Messaoud O**. Genomic Investigation of the SARS-Cov-2-Host Interaction in the Tunisian Population. 2020-2022. (The Tunisian Ministry of Higher Education and Scientific Research- Federated Research Projects; Accepted).
40. Kingham P (PI), Olusegun a (Co-PI), **Abdulkareem FB** (Site-PI), Barton-Burke M, Adesina F, Fitzgerald DW, **Adeyemo WL**, Ogunsola FT, Hosgood D. Building Cancer Research Capacity in Nigeria. 2020. (NIH-D43; Submitted).
41. Kirenga B, Ngadaya E, Zawedde S, **Muttamba W** (Co-PI), Sayoki M, Wandiga S, Walusimbi S, Mmbaga B, Turyahabwe S, et al. Trial of Expanded Tuberculosis Prevention Therapy and its Adherence Using Multiple Patient Care Approaches and New Drugs, to Improve Population Coverage in Africa. 2020. (European & Developing Countries Clinical Trials Partnership; Submitted).
42. Margaret Pericak-Vance et al. **Cornejo Olivas, M** (Sub-Contract PI Site Peru). Genomic Characterization of Alzheimer Disease Risk in Admixed Populations with Native American and Southern European Genetic Ancestry. 2020. (NIH- R01, Submitted).
43. **Mburu S** (PI), Anzala O (Co-PI), Waithaka, P (Co-PI), Heselmeyer K Reid T, Webale M, Mwaura P. Climate Change and Increased Prevalence of Non-Communicable Diseases. 2020. (Fogarty International Research Training Award for LMICs/NIH K43; in Preparation).
44. **Mburu S** (PI), **Gitaka J** (Co-PI). Improving Patient's Care, Health Outcomes Through Tailored, Medical Genetics and Genomics Capacity Building Educational Programmes for County as well as Sub-County Hospitals in Kenya. 2020. (Takaedi- Seed Global Health Programme and H3 Africa; in Preparation).
45. **Mburu S** (PI), **Gitaka J** (Co-PI). Understanding Breast Cancer Patient Population, Subtypes Distribution in Kenya, Their Epidemiological Risk Factors and Identification of Population Specific Potential Diagnostic, Prognostic Biomarkers as Well as Therapeutic Targets: a Mixed Predictive Study Design. 2020. (Pfizer-Glaxo Wellcome NCD Open Lab; NRF Kenya, in Preparation).
46. **Messaoud O** (Member in Charge), Members and Alumni of the Working Group Health and Society. Towards Effective Solutions for Combating COVID-19: An Interdisciplinary Approach. 2020. (German Federal Ministry of Education and Research Fund- AGYA; Accepted).
47. **Messaoud O** (PI), Abdel-Hafiez M (Co-PI). for a Better SARS-Cov-2 Diagnosis. 2020. (German Federal Ministry of Education and Research Fund- AGYA; Accepted).
48. **Messaoud O** (PI), Jones M, Abdelhak S. Clinical and Genetic Characterization of Atypical Forms of Photo-Genodermatoses. 2020-2021. (Tunisian Ministry of Higher Education and Scientific Research- Young Researchers Incentive Project; Accepted).
49. **Messaoud O** (PI), Rizk a (Co-PI). Phi's Research and Innovation Summit. 2020. (German Federal Ministry of Education and Research Fund- AGYA; Accepted).
50. 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; Continued).
51. **Nakousi-Capurro N** (PI), Bustamante L, Miranda M, Pardo RA, Varela D, Diamantino C, De Gracia C, López M. Functional Study of Genetic Variants Associated with Isolated Congenital Anosmia in a Chilean Family: An Alternative to the Study of Central Nervous System Development. 2019-2020. (Saval Laboratories; Continued).

52. Ofori-Acquah SF (PI), Makani J (PI), Awandare GA (PI), Sani MU, **Adeyemo TA** (Co-I). Sicklegenafrica: Sickle Cell Disease Genomics Network of Africa. 2018-2022. (NIH-U54; Continued).
53. Olopade OI (PI), **Abdulkareem FB** (Site- PI), Sowumi A(Co-I), Daramola AO. U. Chicago Interdisciplinary Cancer Health Disparities SPORE in Response to the NOSI Announcement: Administrative Supplement Opportunity to Stimulate or Strengthen Global Cancer Health Disparities Research. 2020. (University of Chicago; Submitted).
54. Olopade OI (PI), Wickrema a (Co-PI), Brown J (MPI/PD), Phiri M (PI/PD), **Abdulkareem FB** (Member-TAC), Adejumo PO, Babalola C, Nnodu OE, Aribisala B. Global Scholars in Oncology Associated Research (SOAR)- Program to Strengthen Institutional Capacity for Collaborative Global Cancer Research in LMIC Countries. 2020. (NIH-D43; Submitted).
55. Oyediran K, Kirenga B, Turyahabwe S, Davis N, Chauffour J, **Muttamba W** (Co-PI), Muzoora A, Muyinda H. Quality of Tuberculosis Services Assessment in Uganda. 2020. (USAID-MEASURE Evaluation; Received).
56. Romdhane L (PI), **Messaoud O**, Abdelhak S, Ghedira K, Ben Hamda C, M'rad R, Mezzi N, Romdhane S. Development and Establishment of an Information Portal on Genetic Diseases in Tunisia. 2020-2021. (Tunisian Ministry of Higher Education and Scientific Research- Young Researchers Incentive Project; Accepted).
57. **Utumatwishima JN** (PI), Uwimana A, Gunilla K. Investigating the Association Between Gender-Related Psychosocial Factors and Undernutrition of Mothers and Children in Rwanda. 2020. (Swedish International Development Cooperation Agency, PhD scholarship, Gothenburg University; Received).
58. **Wangi KYW**. Genetics Newborn Screening: Sharing a Benefit? Ethical Challenges in Developing Country. 2020. (NIMHD/NIH-R03; in Preparation).
59. Wei JJ (PI), **Abdulkareem FB** (Co-I), Silas Olugbenga S (Co-I). Develop a Telepathology Program in Promoting Cervical Cancer Research and Training in Nigeria. 2020. (Northwestern University; Received).

2017 Batch

60. **Abad PJ**, Tumalak MJR, Laurino MYL. Establishment of a Telegenetics Program to Improve Cancer Genetic Counseling Service Delivery. 2020. (Philippine-NIH; in Preparation).
61. Chimusa ER (PI), Moller M, Dandara C, Mulder N, Sangeda R, **Mahfoudh W** (Co-I), Vilhjalmsson B, Bah B, Cathal S, et al. African Genomic Risk Assessment. 2020. (NIH-U01; Submitted).
62. Dalle H (PI), Makani J (Co-PI), **Mgasa A**, Brethon D, Fahd M, Leblanc T, Seror E, Lachenaud J, Ray Lunven, a et al. French-Tanzanian SCD Program : Hematopoietic Stem Cell Transplantation. Jan. – Mar. 2020. (French Embassy in Tanzania- Internship Grant; Received).
63. **Dhoro M** (PI), Bwakura M (PI). Nutrigenomics Study-Establishment of a Biobank for samples from a Cohort Hospitalized for Severe Acute Malnutrition. 2020. (University of Zimbabwe Department of Dentistry-NORHED Faculty Development Grant; Received).
64. Dhungel S, **Thakur N**, Kunwar AJ, Mahat B, Thapa S. Understanding of Neuroscience and Spectrum of Neurogenetic Disorders. IBRO-APRC Nepal School. 2020. (Conference Grant, International Brain Research Organization; Received).
65. Ergün SG (PI), **Tuncay Yaylacioglu F**, Talim B, Dincer PR. Evaluation of Phenotypic Effects of CRISPR/Cas9 Mediated LRP5 Variations on Zebrafish. 2020. (TUBITAK 1002 - Short Term R&D Funding Program; Submitted).

66. **James O**, Akinboboye BO, Okunade KL, Adekunle AA, **Adeyemo WL**. Evaluation of Use and Effectiveness of Telemedicine Among the Health Professionals at Lagos University Teaching Hospital During the COVID-19 Lockdown Period. 2020. (College of Medicine, University of Lagos- Da Boyz COVID-19 Research Grant; Received).
67. **Owusu M** (PI), Acheampong G, Owusu-Dabo E, Adu-Sarkodie Y. Chromosomal and Plasmid Mediated Fluoroquinolone Resistance in Human Salmonella Enterica Infection. 2020. (KNUST Research Excellence Fund; in Preparation).
68. **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; Received).
69. Serbest KC, **Tuncay Yaylacioglu F** (Sub-I), Konuk O (PI). Are Foxp3 Polymorphisms Risk Factor for Development of Ophthalmopathy in Patients with Graves' Disease? 2019-2020. (Gazi School of Medicine; Continued).
70. Sylverken A, Owusu Dabo E, **Owusu M** (Co-PI). the Burden of Respiratory Pathogens Among Motor Tricycle Operators in Kumasi Metropolis, Ghana. 2020. (KNUST Research Fund; Accepted).
71. **Thakur N** (PI). Medians for Maternal Serum Markers of Fetal Abnormalities: Progress in Prenatal Screening. 2020. (NHRC Provincial Grant; Received).
72. **Torres-Mejía G** (PI), Fejerman L. Ángeles-Llerenas A, Gómez Flores-Ramos L. LAGENO-BC & CONFLUENCE Research Proposal. 2020. (National Institute of Public Health; Submitted).
73. **Tuncay Yaylacioglu F** (PI), Ergün SG, Talim B, Dincer PR. Evaluation of Phenotypic Effects of CRISPR/Cas9 Mediated CHST6 Knock-Out on Zebrafish Cornea. 2020. (Güven Medical Group- New Investigator Research Grant; Received).
74. **Tuncay Yaylacioglu F** (PI), Ozmen MC. Macular Corneal Dystrophy as a Part of TUSEB Collaboration Program in Personalized and Translational Medicine. 2020. (Health Institutes of Turkey; Accepted).

2016 Batch

75. **Adeyemo AA** (PI). DELTAS Africa CPE Seed Fund: The Silent Plague. 2019. (African Academy of Sciences; Received).
76. Bsglu E (PI), **Deniz E** (Consultant). Identification of Protein Interacting Partners of Na⁺/K⁺-ATPase Pump in Ischemic Heart Disease Model. 2019. (TUBITAK; Received).
77. Butali A (PI), **Adeyemo WL** (Co-I), **Eshete MA**. Refining the Genetic and Genomic Architecture of Non-Syndromic Orofacial Clefts. 2020-2025. (NIDCR/NIH; Received).
78. Dasanayaka HMNN, **Sirisena ND** (Co-PI), Samaranayake TN (PI). Genetic and Epigenetic Profile of Practitioners of Meditation and Mindfulness Techniques in A Sri Lankan Cohort. Assessment of the Effects of Meditation and the Developed Protocols on Genetics in the Meditation, Mindfulness & Health Project. 2020. (World Bank-AHEAD Grant, University of Colombo; Received).
79. **Deniz E** (PI). Investigation of Molecular Mechanisms of the Genes That Have the Potential to Affect Drug Resistance. 2020. (TUBITAK, Submitted).
80. **Guio H** (PI), Jarmillo L, Palmar D. Evaluation of Three Methods (LFIA, CLIA and FIA) for the Detection of Anti-SARS-Cov-2 Antibodies in the Peruvian Population, in the Year 2020. 2020. (Universidad De Huanuco; Submitted).
81. 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). Phase II. 2020. (University of Rwanda-Directorate of Research and Innovation; Received).
82. Hou L (PI), Sagay A (PI), Ogunsola FT (PI), Akanmu AS, Murphy M (PI), **Adeyemo WL, Abdulkareem FB** (Co-I). Epigenomic Biomarkers of HIV-Associated Cancers in Nigeria. 2017-2022 (NCI/NIH-U54; Continued).
 83. Kantarama E (PI), Gahutu JB (Co-PI), **Uwineza A** (Co-PI). Cardiometabolic Risk Factors Among Users of Injectable Hormonal Contraceptives in Rwanda. 2020. (CARTA; Received).
 84. Munezero Louise (PI), Musoni Grant (PI), Rugwizangiga B (PI), **Uwineza A** (Co-PI). Genexpert Assay on Fine Needle Aspiration Samples for Diagnosis of Tuberculous Lymphadenitis. 2020. (University of Rwanda-Directorate of Research and Innovation; Received).
 85. Murphy M (PI), Hou, L, Sagay A, Ogunsola FT, Akanmu AS, **Adeyemo WL** (Co-I), **Abdulkareem FB** (Co-I). Nigeria Research Training Grant for HIV and Malignancies. 2019-2024. (Northwestern University, USA; Continued).
 86. **Seven M** (PI), Pachucki M (Co-PI), Gould D, Bae Y, Leblanc R, Walker R. Evaluation of Family and Community Social Network Characteristics Among High-Risk Family Members To Improve Cancer-Related Health Behaviors. 2020- 2021. (University of Massachusetts, USA; Received).
 87. Sjarif DR (PI), **Ariani Y** (Co-PI). Developing of GALNS Enzyme Activity Method and Gags Urine Concentration Screening from Mucopolysaccharidosis Type IV Patients in Indonesia. 2019. (Ministry of Higher Education; Accepted).
 88. Sjarif DR (PI), **Ariani Y** (Co-PI). Variant Analysis of CLN2 Gene from Neuronal Ceroid Lipofuscinosis Type 2 Patients in Indonesia. 2019. (Ministry of Higher Education; Submitted).
 89. Sjarif DR (PI), **Ariani Y** (Co-PI). Variant Analysis of GAA Gene from Pompe Disease Patients in Indonesia. 2019. (Ministry of Higher Education; Submitted).

C. Collaborations (NIH and Other Institutions):

NIH: (Total- 20; 2019- 8; 2018- 7; 2017- 1; 2016- 4)

2019 Batch

1. **Abubakar S**, Koehly L. Implementation of a Family Health History Taking Tool for Improving Family Health History & Genomic Literacy in Nigeria. a Pilot Feasibility Study. (Bayero University Kano/Aminu Kano Teaching Hospital, Nigeria; NHGRI/NIH, USA; Proposed).
2. **Chowdhury E**, Hufnagel R (PI). Study of Relation Between Consanguinity of Marriage and Retinitis Pigmentosa. (Samtse General Hospital, Bhutan; NEI/NIH, USA).
3. Fejerman L (PI), Dominguez-Valentin M, Vianna-Jorge R, Agalliu I, **Paredes-Moscossa SR** (Co-I), Carvallo P, Weitzel J, Bertoni B, Latin American Genetics and Genomics of Breast Cancer Consortium (LAGENO-BC). Resource to Study Latino Breast Cancer Architecture Through GWAS. (Universidad De San Martin De Porres, Peru; NCI/NIH, USA).
4. Gahl W, Baynam G, **Wiafe SA**. Clinical and Genetic Evaluation of Undiagnosed Genetic and Rare Disease Patients Through the Undiagnosed Disease Program. (Rare Disease Ghana Initiative, Ghana; Western Australian Health Department, Australia; NHGRI/NIH, USA).
5. **Iqbal M, Naeem MA**, Hufnagel R. Identification, Clinical Evaluation, Genetic Analyses of Consanguineous Pakistani Families Affected with Syndromic Retinitis Pigmentosa. (The Islamia University of Bahawalpur, Pakistan; University of the Punjab, Pakistan; NEI/NIH, USA).

6. **Llamos-Paneque A**, Groft S, Tiffit C. ICORD's New Initiative: Rare Diseases in the Caribbean and Latin America (ERCAL). (Specialty Hospital No.1 FF. AA, Ecuador; NIH Office of Rare Diseases Research, USA).
7. **Wiafe SA**, Groft S, Baynam G. Clinical and Research Networks for Undiagnosed, Genetic and Rare Disorders in Ghana. 2020. (Rare Disease Ghana Initiative, Ghana; Western Australian Health Department, Australia; NCATS/NIH, USA).
8. **Yesilcinar I, Seven M**, Calzone K. Determining Genetics and Genomics Competency of Turkish Nurses. (Izmir Katip Celebi University, Turkey; Health Science Faculty Obstetrics and Gynecology Nursing, Turkey; University of Massachusetts Amherst College of Nursing, USA; NCI/NIH, USA).

2018 Batch

9. **Adewole OO**, Landhi M. Genetic Epidemiology of Lung Cancers in Never Smokers (OAUTHC, Nigeria; NCI/NIH, USA).
10. **Adewole OO**, Levin S. Defining the Role of Apolipoprotein Pathways in African Asthmatics (OAUTHC, Nigeria; NHLBI/NIH, USA).
11. **Cornejo-Olivas M, Lertwilaiwittaya P**, Traynor B, Alva-Diaz C, Alarcon-Ruiz C, Pacheco-Barrios K, Mori N, Pacheco-Mendoza J, Rivera-Valdivia A. Systematic Review Related To C9orf72, Huntington Like Disorders and HLD in Peru and Mexico (Instituto Nacional De Ciencias Neurologicas, Peru; Mahidol University, Thailand; NIA/NIH, USA).
12. **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, USA).
13. **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, Tunisia; Charles Nicolle Hospital, Tunisia; Farhat Hached Hospital, Tunisia; NCI/NIH, USA).
14. 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, USA).
15. Riazuddin S (PI), **Naeem MA** (Co- I), Husnain T, Sieving PA, Fielding JH, John D. Gottsch, Riazuddin SA, Qazi ZA, Butt NH, et al. Hereditary Vision Impairment in Pakistan (CEMB, University of the Punjab, Pakistan; NEI/NIH, USA; Wilmer Eye Institute, Johns Hopkins University, USA; Layton Rehmatulla Benevolent Trust Eye Hospital, Pakistan; Allama Iqbal Medical College, University of Health Sciences, Pakistan).

2017 Batch

16. **Mgasa A**, Lyimo M, Nkya S, Makani J, Sheehan V, Tisdale J, Gibbons G. Advanced Therapy for Management of Sickle Cell Disease Project (Muhimbili University of Health and Allied Science Tanzania; National Blood Transfusion Service Tanzania; Baylor College of Medicine, USA; NHLBI/NIH, USA).

2016 Batch

17. **Ekure EN**, Kruszka P, Adeyemo A. Genetics of Congenital Heart Diseases in Africa. (University of Lagos/Lagos University Teaching Hospital, Nigeria; NHGRI/NIH, USA).

18. **Ekure EN, Lotz-Esquivel S, Dueñas-Roque M**, Kruszka P, Adeyemo A. Atlas of Human Malformation Syndromes in Diverse Populations. (University of Lagos/Lagos University Teaching Hospital, Nigeria; NHGRI/NIH, USA).
19. Ogunsola FT, Alatisé OI, Kingham TP, **Adeyemo WL**. Building Cancer Research Capacity in Nigeria. (Obafemi Awolowo University, Nigeria; University of Lagos, Nigeria; Memorial Sloan Kettering Cancer Center, USA).
20. Okuyemi K, Ogunsola FT, **Adeyemo WL**. Collaboration for Strengthening Cancer Population Sciences Research Capacity in Sub-Saharan Africa. (University of Lagos, Nigeria; University of Utah, USA).

Other Institutions: (Total - 42; 2019- 8; 2018- 18; 2017- 10; 2016- 6)

2019 Batch

1. **Alamin TA**, Elzubeir AM, Elsheikh SH. Association of Hypercoagulable State with Clinical Severity of Sickle Cell Disease in Sudanese Patients. (University of Khartoum, Sudan; University of Colorado, USA).
2. **Bocoum A**, Koita A, Rizig M, Maiga Y, Guinto CO. Parkinson's Disease in Africa: Developing a Clinically Characterized DNA; Resource for Genome-Wide Association (GWAS) and Other Collaborative Genetic Studies. (Centre Hospitalier Universitaire Du Point 'G', Bamako, Mali; Centre Hospitalier Universitaire Gabriel Touré, Bamako, Mali; UCL Queen Square Institute of Neurology, London, UK).
3. Gulsen A, **Llamos-Paneque A**. Genome Sequencing to Elucidate Causes and Mechanisms of Mendelian Genetic Disorders. (Baylor College of Medicine, USA; Genetic Service of Hospital No.1 FF. AA., Quito, Ecuador).
4. **Gyawali P**, Delaney M, Koju S. International Perspectives Research Survey on Transfusion Services in Low- and Middle-Income Nations. (Dhulikhel Hospital, Nepal; Pathology & Transfusion Medicine, Children's National Hospital, Washington DC, USA).
5. Isaac LD, Guay-Woodford LM, San CC, **Khant AK**. Redcap Data Collection for Genetic of Cystic Kidney Disease in Asia. (National University Hospital, Singapore; Yangon Children Hospital, Myanmar).
6. **López-Star B, Llamos Paneque A**. Consultation on Ophthalmogenetic Cases. (Mexican Society of Ophthalmology, Mexico; Mexican Institut NA, Ophthalmology, Mexico; Specialty Hospital No.1 FF. AA, Ecuador).
7. Santra MK, Rapole S, Tripathi V, Shridhar PR, Shanamugham D, Tilak TVSVGK, **Mutreja D**, Deshmukh C, Tamhankar AS, Chatterjee S. Mechanism & Therapeutic Application of Neuroimmune Communication in Colorectal Cancer (Dept of Biotechnology, Ministry of Science & Technology, India; NCCS, Pune; NCL Pune; DMH Pune; School of Chemistry, University of Hyderabad; Submitted).
8. Sidharth KS, Azmeri S, **Khant AK**, Sadaf A, Syed SH, Iftikhar I, South Asia Association for Regional Cooperation -Pediatric Acute Kidney Injury Consortium. Prospective Study on Incidence and Risk Factors for Severe Pediatric AKI in SAARC Nations. (Yangon Children Hospital, Myanmar; India; Bangladesh; Pakistan; Sri Lanka; Myanmar; Nepal; Bhutan; Maldives).

2018 Batch

9. **Abdulkareem FB**, Colorectal Management and MSK's African Research Group for Oncology. Monthly Virtual ECHO Conference Project (MSKCC; Multiple Institutions; Nigeria).

10. **Adewole OO**, Abdou M, Oyedeji M. COVID -19 and TB Co-Infection (Obafemi Awolowo University Teaching Hospital Complex, Lome Teaching Hospital, Nigeria; Federal Medical Center, Katsina; University College Hospital, Nigeria).
11. **Dwivedi A**, Kalra S, Chand K, Bose T, Dey SK. Diagnostic Yield of Exome Sequencing in Children with CAKUT (Congenital Anomalies of Kidney & Urinary Tract) with Extra Renal Manifestations. (AHRR, Delhi; Command Hospital, Lucknow, India).
12. **Dwivedi A**, Kumar R, Singh AK, Dey M. Clinical Utility and Diagnostic Yield of Exome Sequencing in Prenatal Diagnosis of Congenital Anomalies. (AHRR, Delhi, India; Base Hospital Delhi Cantt, India).
13. **Eshete MA**, Project Harar Ethiopia. Provide Safe Cleft Surgery and Complex Craniofacial Surgery (Addis Ababa University, Ethiopia; Project Harar, Ethiopia).
14. **Gitaka J**, Mutapi F. Molecular Epidemiology of Plasmodium Falciparum. (University of Edinburgh, UK; Mount Kenya University, Kenya; University of Nairobi, Kenya).
15. Lopez-Cendes I, **Dueñas-Roque M**. Whole Exome Sequencing- Identification of Pathogenic Genetic Variants in Early Infancy Encephalopathy, in Latin America. (Universidad Estadual De Campinas, Brasil; Hospital Nacional Edgardo Rebagliati Martins, Perú).
16. **Mburu S**, Anzala O, Waithaka P, Webale M, Mwaura P. Low-Dose Microbial Endotoxins Exposure and Quantification of Specific Cancer Characteristics Response. (Kirinyaga University; Kenya Aids Vaccine Initiative -University of Nairobi, Kenya; Murang'a University of Technology, Kenya).
17. **Mburu S**, Waithaka P. Tailored Containment Measures and Innovative County Specific Contact Tracing Approaches for Breaking Community Transmission of Covid-19. (Kirinyaga University; Kenya Aids Vaccine Initiative - University of Nairobi, Kenya).
18. Menzel Stephan, **Adeyemo TA**, Nnodu O. Identification of Novel Mechanisms of Fetal-Haemoglobin Induction by Common Genetic Variation in Patients with Sickle Cell Disease. (Kings College, London, UK; University of Lagos, Nigeria; University of Abuja, Nigeria).
19. **Messaoud O**, Banerjee K, **Cornejo-Olivas M**, **Kumar Dutta A**, Bhuiyan Z. Monogenic Vs. Oligogenic Reclassification. Topic Section at Frontiers in Genetics (Laboratoire De Génomique Biomédicale Et Oncogénétique, Institut Pasteur De Tunis, Tunisia; Burke Medical Research Institute White Plains, USA; Instituto Nacional De Ciencias Neurológicas, Peru; All India Institute of Medical Sciences, Kalyani, India; Centre Hospitalier Universitaire Vaudois, Switzerland).
20. **Messaoud O**, Khalil A, Nasr M, Abou-Elenein M, Sunoqrot S, Amro A, Romdhane L, Adm M, Alkatan M, et al. Screening of Nanoparticles, Biomaterials, Natural Extracts and Disinfectants on Coronavirus Strain Using Several Antiviral Tests and Assessment of the Susceptibility of T-Lymphocytes to SARS-Cov-2 Infection (Institut Pasteur De Tunis, Tunisia; Faculty of Sciences of Bizerte, Tunisia; Egypt Japan University of Science and Technology (EJUST), Egypt; Charité Universitätsmedizin Berlin, Germany; Al-Zaytoonah University of Jordan, Jordan; the Public Authority for Applied Education and Training in Kuwait, Kuwait; Ain Shams University, Egypt; University of Sharjah, UAE; Palestine Polytechnic University, Palestine; Al-Quds University, Palestine).
21. Morgan R, Ssali S, **Muttamba W**, Kirenga B, Buregyeya E. Piloting the Intersectional Gender Analysis Toolkit in Tuberculosis Research: a Case Study on Outcomes of Intensified Passive TB Case Finding (John Hopkins University, USA; Makerere University Lung Institute, Uganda).
22. Olopade OI, Ntekim A, Popoola AO, Arowolo OA, Sowunmi A, Olopade CS, Ibraheem A, **Abdulkareem FB**, Daramola OA, et al. Assessing Response to Neoadjuvant Taxotere and

- Trastuzumab in Nigerian Women with HER2-Positive Breast Cancer (ARETTA); a Multicenter Study. (Universities of Lagos, Nigeria; University of Ibadan, Nigeria; Obafemi Awolowo University, Nigeria; Ile-Ife and Lagos State University, Nigeria; University of Chicago, USA).
23. Ortega J, Galarreta-Aima C, **Dueñas-Roque M**, **Cornejo-Olivas M**. Ihope, An Illumina Project for Genomic Testing for Rare Diseases in Peru. (Illumina; Hospital Nacional Edgardo Rebagliati Martins; Instituto Nacional De Ciencias Neurológicas, Peru).
 24. Pasechnikova N, Zborovska O, **Savina O**, Kremenska U. Medical and Genetic Diagnostics and Consultation for Families with Retinoblastoma, for Options of Pre-Implantation for The Next Pregnancy. (V. Filatov Institute of Eye Diseases and Tissue Therapy; ISIDA-IVF Clinic, Ukraine).
 25. Sabiti W, **Muttamba W**, Kirenga B, Devesh JD. Exacerbation of Chronic Bronchial Sepsis (ECBS): a Study of the Role of Key Bacteria in Exacerbations of COPD and Bronchiectasis (Makerere University Lung Institute, Uganda; St Andrews University Scotland, UK).
 26. **Savina O**, Rykov O, Kremenska U. Medical and Genetic Diagnostics and Consultations for Families with Aniridia and WAGR Syndrome for Pre-Implantation Diagnostics During Family Planning. (Department of Ophthalmology, Shupyk's National Medical Academy of Postgraduate Education, Ukraine; ISIDA-IVF Clinic, Ukraine; NGO – Aniridia; Ukraine; WAGR, Kyiv Ophthalmological Clinic, Ukraine; in Discussion).

2017 Batch

27. **Abad, PJ**, Dumo AM, Laing B, Lim AG, Valdehueza O, Ward LD, Carlberg C, Vehvilainen-Julkunen K. Effectiveness of a Web-Based Genomic Nursing Intervention. (University of the Philippines, Philippines; University of Eastern Finland, Finland; South Eastern Finland University of Applied Sciences, Finland; Clemson University, USA; University of Auckland, New Zealand; Don Mariano Marcos State University, Philippines).
28. **Avogbe PH**, Durand G, Le Calvez-Kelm F. Whole Transcriptional Profiling and Gene Expression Analysis in Urothelial Carcinoma. Data Available for Analyses. (International Agency for Research on Cancer/WHO; the University of Abomey-Calavi, Benin).
29. **Avogbe PH**, Le Calvez-Kelm F, and Mckay JD. Circulating Tumor DNA Biomarkers and Early Detection of Hepatocellular Carcinoma. Sample Collection Underway. (University of Abomey-Calavi, Benin; the International Agency for Research on Cancer/WHO).
30. **Casado PL**, Patrick Shmidlin.: Frontier in Dental Medicine. Research Topic: Biomarkers in Periodontology and Peri-Implant Diseases. (Fluminense Federal University, Brazil; University of Zurich, Switzerland).
31. **Petlichkovski a** Sukarova-Angelovska E. Analysis of Genes Involved in Folate Metabolism in Children with Congenital Anomalies Associated with Palate Cleft (Institute for Immunobiology and Human Genetics, Neurology University Clinic & Pediatric University Clinic, Medical Faculty Skopje, Macedonia).
32. **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 & Pediatric University Clinic, Medical Faculty Skopje, Macedonia; University of Turin, Italy; Icahn School of Medicine at Mount Sinai, USA).
33. **Petlichkovski A**, Trpceviski T. Introduction of First Preimplantation Genetic Testing of Aneuploidies (PGT-A) Using the NGS Technology in Macedonia. (Institute for Immunobiology

and Human Genetics, Medical Faculty Skopje, Macedonia; Fertility Clinic ‘Plodnost’ Bitola, Macedonia).

34. **Roy S**, Kaliki S, Fabihan D. Retinoblastoma Management During the Covid-19 Pandemic Situation. (Chittagong Eye Infirmary, Bangladesh; London School of Hygiene, UK; L.V. Prasad Eye Institute, India; 193 Centers from 94 Countries).
35. **Thakur N**, Akhundi RS, Kunwar AJ. Sickle Cell Anemia and Its Coexistence with Alpha and Beta Thalassemia in a Cohort of Indigenous Tharu Population of Nepal: Implications for The South Asian Region. (Kathmandu Center for Genomics and Research Laboratory, Nepal; South Asian University, India).
36. **Vishnopolska SA**, Camper S, Kitzman J. Development of Saturation Mutagenesis Probes and Protocol to Test GLI2 Gene Variants and Sensor for GLI2 Function. (Universidad De Buenos Aires, Argentina; University of Michigan, USA).

2016 Batch

37. **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).
38. **Adeoye AM**, Karaye KK, Loeb M, Yusuf S. a Randomized Controlled Trial of Influenza Vaccine to Prevent Adverse Vascular Events (Ivvetrial). (University of Ibadan, Nigeria; Bayero University, Nigeria; McMaster University, Canada).
39. **Ariani Y** (PI), Yuridian R, Muizzuddin N. Genomic Study of Skin Aging in Indonesian Population. (Indonesian Medical Education and Research Institute, Indonesia; University of Indonesia, Indonesia; Paragon Technology and Innovation Corp., Indonesia; Saint John’s University, USA).
40. **Guio H**, BGI Group. Develop Genetic Analysis in Peru. (INBIOMEDIC, Peru; BGI Group, China).
41. **Guio H**, Tarazona E. Collect Samples to Study Genetics in Patients Infected with SARS-Cov2. (INBIOMEDIC, Peru; University of Mina Gerais, Brazil).
42. Manirakiza F, Sugimura H, **Uwineza A**. Comparative Molecular Epidemiology: Environment, Genomics and Phenotype Interaction in Solid Tumors in Japan and Rwanda. (School of Medicine and Pharmacy, University of Rwanda, Rwanda; Hamamatsu University School of Medicine, Japan).

D. New Research Projects: (Total- 72; 2019- 26; 2018- 11; 2017- 21; 2016- 14)

2019 Batch

1. **Amarakoon GGGT**, Wijayarathne HK. Clinical Presentations and Phenotype – Genotype Correlation in Children with Acute Lymphoblastic Leukaemia in Sri Lanka.
2. **Amarakoon GGGT**, Wijayarathne HK. Clinical Presentations of Retinoblastoma in Sri Lankan Children.
3. Badoe E, Ameyaw EK, **Wiafe SA**, Manu E, Asafo-Agyei SB, Thomford NE, Arthur BE. Molecular Diagnosis of MPS Patients in Ghana: the RDGI-Biomarin Project.
4. Chaudhry A, **Bocoum A**, Rizig M. Identifying Knowledge Gaps in Parkinson’s Disease Amongst Medical Professionals in Africa.
5. **Chowdhury E**, Lhendup K, Zangmo Y, Tobgay S, Phuntsho N. Incidence and Prevalence of Down Syndrome in Bhutan.

6. Donohue J, **Wiafe SA**, Ahiable E, Wojtowicz W. Screening for Urea Cycle Disorders in Ghana: a Pilot of Blood Ammonia Checker Device.
7. **Kars ME**, Ozcelik T. Prevalence of Actionable Secondary Findings in 3,599 Whole Exomes and Genomes from Turkey.
8. Lam HT, **Luong LH**. Stem Cell Transplant for Patient with Refractory and Treatment Resistant Auto Immune Disease.
9. **Llamos-Paneque A**, Mata V. Study on Genetics of Parkinson Disease in Ecuador; a LARGE PD Study. (Pending IRB Approval).
10. **Llamos-Paneque A**, Vilain, E. Molecular Characterization of Sexual Developmental Disorders in a Population from Ecuador. (Pending IRB Approval).
11. **López-Star B**, Pérez- Serrano R, Ochoa Luis. Identification of rs7677751 Polymorphism in PDGFRA Gene, in Patients with Keratoconus.
12. **Mhandire K**, Dandara C. Pharmacogenomic Determinants of Resistant Hypertension in Africans: Towards a Gene-Based Treatment Outcome Prediction Tool.
13. **Monye HI**, Oluleye TS, Olawoye O, Ugalahi MO. Proportion, Clinical Profile and Pedigrees of Children with Presumed Genetic Eye Diseases and Parental Willingness to Test at the University College Hospital, Ibadan. (Pending Ethic Approval).
14. **Mosema KBA**, Bramble MS, Vilain E. Disorders of Sex Development in Central Africa: Genetics and Psychosocial Adaptation. (Proposal in Preparation).
15. **Mushi TL**, Nkya D, Kija E. Magnitude and Etiology of Persistent Pulmonary Hypertension Among Newborns in Tertiary Setting, Tanzania. a Prospective Study.
16. **Mushi TL**. Newborn Screening for Congenital Heart Disease at Mnazi Mmoja Hospital, Zanzibar. Submitted Proposal).
17. **Okunola OO**, Odeyemi A. Inheritance Pattern in Diabetic Nephropathy in Lagos, Nigeria.
18. **Okunola OO**. Health Related Quality of Life Assessment Scores in Sickle Cell Patients.
19. Osgonbaatar O, Nyamjav D, Tumenbayar A, **Baatar N**, Duuren M, Sukhbaatar B, Alimaa A. Assess Pediatric Sex Development Disorders Among School Children in Mongolia.
20. Osgonbaatar T, Nyamjav D, Tumenbayar A, **Baatar N**, Duuren M, Sukhbaatar B, Alimaa A. Implementation of Griffith's Assessments to Determine Mental Development of Mongolian Children.
21. Rawal S, **Gyawali P**. Microbiota in Pregnancy.
22. Talukdar S, Dhani AK, **Mutreja D**. Evaluation of Human Epidermal Growth Factor Receptor 2 (HER2) Expression in Endometrial Cancers in a Tertiary Care Teaching Hospital in Western Maharashtra: a Cross Sectional Study. (Approved by IRB).
23. Tekle S, Mussa M, Okbe Z, Asrat K, Abdu N, Mosazghi A, Tesfamariam EH, **Weldemichael YG**. Incidence and Risk Factors of Neural Tube Defects (NTDs) in Three Selected Hospitals, Eritrea.
24. Thomford NE, **Wiafe SA**, Asafo SM, Agblozo YE, Akyaw P, Mangortey D. Assessment of the Needs and Quality of Care for Undiagnosed, Genetic and Rare Diseases in Ghana.
25. **Wiafe S**, **Kemevor-Asima D**. a Pilot Study on Newborn Screening for Inherited Metabolic Diseases in a Tertiary Institution, Accra, Ghana.
26. **Wiafe SA**, Ameyaw EK, Dunyoh B, Appiah DY, Amissah H, Gyening-Yeboah A, Akyaw P, Addo-Lartey EO, Amoako E, et al. Screening for Inherited Metabolic Disorders Among Newborns in Ghana.

2018 Batch

27. **Adeyemo TA**, Bolarinwa AB, Olatinwo AT, Adiat AL, Akanmu AS. Clinical and Laboratory Phenotypes of Persons Living with Haemophilia in Southwest Nigeria.
28. Ajoloko EA, **Oluwarotimi AC**, Adeyemi MO. Efficacy of Sodium Bicarbonate Buffered Lidocaine Hydrochloride in Reduction of Injection, Onset of Action and Depth of Anaesthesia Intra-Alveolar Extraction of Mandibular First and Second Molars: a Randomized Controlled Double-Blind Study.
29. **Cornejo-Olivas M**, Cubas-Montecino D, Bazalar-Montoya J. DNA Biobank Implementation in Neurogenetics Division Instituto Nacional De Ciencias Neurologicas.
30. Kirenga B, Lusiba J, Byakika P, **Muttamba W**, Kayongo A, Ssali F, Siddharthan T, Ssali F, Dzungu H, et al. Assessment of Safety and Efficacy of COVID-19 Convalescent Plasma for Treatment of COVID-19 in Adults in Uganda; a Randomized Controlled Trial.
31. **Lertwilaiwittaya P**, Jinda W, Senanarong V, Assanasen J, Muangpaisal W, Rattanabannakit C, Limwongse C. Next Generation Sequencing in Dementia Subgroup in Thailand.
32. **Lertwilaiwittaya P**, Pho-Iam T, Soontrapa P, Boonyapisit K, Limwongse C. Prevalence of C9orf72 Hexanucleotide Repeat Expansion in Different Neurodegenerative Disease in Thailand.
33. **Lertwilaiwittaya P**, Roothumnong E, Thongnoppakhun W, Limwongse C, Pithukpakorn. Next Generation Sequencing in Breast-Ovarian Cancer in Thailand.
34. Mazzetti-Soler P, **Cornejo-Olivas M**, Cosentino C, Torres L, Illanes-Manrique M. Enroll-HD: a World Observational Study for Huntington's Disease Families, a CHDI Foundation Project.
35. **Messaoud O**, Adm M. Simulation of Genomes from Arab and German Datasets: a Valuable Tool for Variant Detection Prioritization.
36. **Messaoud O**, Mahmoud AH, Rami C, Drissi B. Functional Materials for Health.
37. Pithukpakorn M, Suktitipat B, **Lertwilaiwittaya P**. Molecular Genetics Study of a Thai Family with Skeleton Dysplasia.

2017 Batch

38. **Avogbe PH**, Brun LVC, Sanni A. Comparison of HPV Detection in Urine and Cervical Samples Collected from Beninese Women. (Pending Ethical Approval).
39. **Casado PL**, Luiza Motta L, Telma Aguiar T, Valquiria Quinelato V. Analysis of Alcohol Consumption During Covid-19 Pandemic in Brazilian Students.
40. **Casado PL**, Mariana Campello M, Telma Aguiar T. the Relationship Between COVID-19 and High Risk of Development of Kawasaki Disease in Children: a Systematic Review.
41. **Casado PL**, Mylena Santos M, Telma Aguiar T, Valquiria Quinelato V. Analysis of the Development of Bruxism During Covid-19 Pandemic in Brazilian Students.
42. **Casado PL**, Priscilla Lomardo P, Telma Aguiar T. Association Between the Use of Ultrasound and Risk of Aerosol Contamination in the Dental Office: a Systematic Review for COVID-19 Pandemic.
43. **Casado PL**, Telma Aguiar T, Miriam Jordao M, Valquiria Quinelato V. Relationship of Geographic Tongue Before and During COVID-19 Pandemic in Patients Under Treatment.
44. **Fatima SS**. Association of C3D with Carotid Intima Media Thickness in Diabetic Patients. (Graduate Student Supervision).
45. Gyanwali P, Dhimal M, Jha R, Pandey B, Bastola A, Yadav B, Sharma SK, Khanal B,... **Thakur N**. Deciphering Whole Genomic Sequence of Sarscov-2 and Analysis of Immunological Parameters in COVID-19 Patients in Nepal.

46. **James O**, Otoghile B. Comparative Study of the Risk of Obstructive Sleep Apnoea Between Children with Orofacial Clefts and Children without Orofacial Cleft at the Lagos University Teaching Hospital.
47. Lloyd W, **Mgasa A**, Makani J. Blood Transfusion for Sickle Cell Patients Attending Temeke Regional Referral Hospital in Tanzania.
48. Marcheco Teruel B, **Roblejo Balbuena H**, **Benitez Cordero Y**, Monzón Benítez G. Genetic Factors Associated with Clinical Severity of COVID 19 in Cuban Patients.
49. Marco E, **Mgasa A**, Makani J. Effectiveness of Manual Exchange Blood Transfusion on Chronic Leg Ulcers Outcome to Patients with Sickle Cell Disease in Dar Es Salaam, Tanzania.
50. **Mgasa A**, Dalle H, Gluckman E, Maeda D. Mapping Human Leucocyte Antigen Diversity and Establishment of Hematopoietic Stem Cell Donor Registry in Tanzania.
51. **Nandal R**, Aggarwal S, Kumar A, Taxak S. Cardiac MRI – Utility Above Routine Echocardiography.
52. **Otaify GA**, Abdelhamid MS, Elhousseini R and Temtamy S. Targeted Next-Generation Sequencing in the Diagnosis and Prevention of Skeletal Dysplasias with Joint Deformities.
53. **Roy S**, Nuruddin M. Effectivity of Interferon Alfa 2a in Treatment of Ocular Surface Neoplasia.
54. **Roy S**, Nuruddin M. Outcome of Evisceration by Three Layer Scleral Coat and Orbital Implant.
55. **Torres-Mejía G**, Neuhausen S, Ziv E, Angeles-Llerenas A, Sánchez-Zamorano LM, Flores-Luna L. Genomics and Breast Cancer in Hispanic Women.
56. **Tuncay Yaylacioglu F**, Özdek Sengül. Survey of Practice Patterns for the Management of Genetic Eye Disorders Among Turkish Ophthalmologists.
57. **Yousef YA**, Qaddoumi I, Alrimawi D, Mohammad M, Mehyar M, Jaradat I, Sultan I, Al-Nawaiseh I, Al-Hussaini M. a Proposal for a New Clinical TNM Staging System of Retinoblastoma Based on the American Joint Committee on Cancer Staging Manual, 7th and 8th Editions.
58. Zaki SS, **Otaify GA**, Zaki MS, Issa MY, Mahmoud MM, Abdelhamid MS. Analysis of Dystrophin-Associated Glycoproteins (A-Dystroglycan and Laminin Subunit A2): a Shortcut for Biochemical Screening of Many Types of Congenital Muscular Dystrophies.

2016 Batch

59. **Adeyemo WL**. Refining the Genetic and Genomic Architecture of Non-Syndromic Orofacial Clefts.
60. Bode-Thomas F, Yilgwan CS, Amusa GA, Isa MS, Uche U,...**Ekure EN**, Mbakwem A, Olusegun-Joseph A, Sokunbi O, et al. Nigerian National Registry for Rheumatic Heart Disease.
61. **Ekure EN**, Oguche S, Esezobor C, Jalloh I, Garba BI, Oguonu T, Hassan-Hanga F, Osarogiagbon W, Umar L, et al. An Assessment of the Burden, Risk Factors and Management of Pneumonia Among Under- Five Children Presenting in Health Facilities in Nigeria.
62. **Guio H**, Jarmillo L, Vasquez A. 1000 Peruvian Genome Project.
63. Iramfasha J, Mbanza S., Ndayayisenga S, Ndayishimiye JB, Nduwayezu Rand, **Uwineza A**. Prevalence of Congenital Hypothyroidism, Congenital Adrenal Hyperplasia and Glucose-6-Phosphate Dehydrogenase Deficiency in Rwanda.
64. Kholoussi NM, **Hussen DF**. New Genetic and Immunologic Algorithm for Children with Immune Dysregulation.
65. Krawitz P, **Ekure EN**, Opoola ZA, Lesi FEA. Computer-Assisted Image Analysis and Exome Sequencing for the Diagnosis of Genetic Disorders.

66. Krishnan R, **Sirisena ND**, Dissanayake VHW. Comprehensive Genetic Characterization of Colorectal Adenocarcinoma and Correlation with Gut Microbiome.
67. **Lallar M**. Diagnostic Yield of Fetal and Perinatal Autopsy.
68. Muhawenimana A, Hagenimana S, Mukundwa P, Rhamadan N, Nduwayezu R, **Uwineza A**. Prevalence and Pattern of Sick Cell Disease in Rwandan Pediatric Patient.
69. **Robledo Balbuena H**, Marcheco Teruel B, Monzón Benítez G. Genetic Factors Associated with Clinical Severity of COVID 19 in Cuban Patients.
70. Sjarif DR, **Ariani Y**, Priambodo R, Hafifah CN. Correlation of N-Acetylgalactosamine-6-Sulfatases (GALNS) Gene Mutation and GALNS Enzyme Activity in Mucopolysaccharidosis Type IVA (MPS IVA) Patients in Indonesia.
71. **Tibrewal S**, Ratna R, Dubey S. Childhood Glaucoma – Inheritance Pattern and Role of Consanguinity.
72. **Tibrewal S**, Ratna R, Ganesh S. Familial Inheritance Pattern in Childhood Cataract in North India.

E. Ongoing Research Projects: (Total- 60; 2019- 18; 2018- 17; 2017- 17; 2016 - 8)

2019 Batch

1. **Amarakoon GGGT**, Dehigama D, Wijayarathne HK, Wijesinghe AHNP. Distribution of Congenital Malformations in Newly Born Babies in a Tertiary Care Center in Sri Lanka. (Summit lectures emphasized awareness in congenital malformations).
2. Batmunkh B, **Baatar N**, Baasanjav S. ATP6V0A2 Gene Sequenced in Rare Cutis Laxa Patient (Bioinformatics Workshop at Summit helped find, interpret gene mutation).
3. Cherry PA, **Khant AK**. Correlation of Serum Ferritin Level with Disease Activity and Renal Involvement in Systemic Lupus Erythematosus at Yangon Children Hospital. (Knowledge from Summit improved reporting of research).
4. **Chowdhury E, Roy S**, Hoque F. Epidemiological Study of Retinoblastoma in a Tertiary Eye Care Center of Bangladesh. (Knowledge gained at Summit improved research quality).
5. Espín, S, **Llamos-Paneque A**, Hernández-Iñiguez P. Genetic Study of Two Ecuadorian Families with Clinical Suspicion of Hereditary Spastic Paraplegia. (Summit lectures in neurogenetics improved study execution).
6. **Hussein N**, Lee YK, Qureshi N, John P, Azmi A, Ng CJ. Identifying Gaps in Thalassemia Screening: Exploring the Views, Experiences, and Beliefs Among Thalassemia Carriers in Sabah, Malaysia. (Knowledge gained at Summit helped pursue discussions with authorities to improve carrier screening in population with health literacy challenges).
7. **Kars ME**, Ozelik T. Characterization of the Genetic Structure of the Turkish Population. (Knowledge gained from summit lectures in bioinformatics, personalized training and field trips to NISC and JHU, helped interpret genetic variants and sequencing data analysis).
8. **López-Star B**, Pérez- Serrano R. Identification of Rs7677751 Polymorphism in PDGFRA Gene, in Patients with Corneal Astigmatism. (Summit lectures provided tools to learn of most frequent alleles in Mexican population).
9. Lotempio J, Bramble MS, **Mosema KBA**, Kamangu EN, Mumba-Ngoyi D, Tshala-Katumbay D, Vilain E. Building High-Quality Genome-Enabled Reference Sets to Address Genetic Diversity in Congolese Ethnic Groups in the Democratic Republic of Congo (Summit lectures on Bioethics and Variant Interpretation helped develop human subject consent protocols).

10. May TZO, **Khant AK**. Fractional Excretion of Magnesium in Children with Nephrotic Syndrome. (Knowledge from summit improved reporting of research).
11. **Mutreja D**, Komala SB, Singh V, Sreenivasa S. a Longitudinal Prospective Study on Role of Serum Hepsidin As a Biomarker for Iron Status in Chronic Kidney Disease (CKD). (Summit reiterated facts for personalized approach to diagnosis of CKD related anemia utilizing hepcidin).
12. Nnodu O (PI), Isa H, Tanko Y, Otu T, **Alli A**, Chianumba R, Nnebe-Agumadu U, Ekong J, Elmi Z, et al. Prevalence of Known Fetal Haemoglobin Modifiers and Co-Inheritance of Alpha-Thalassemia in Nigeria: the Abuja/Gwagwalada Pilot Study. (Summit lecture on Genetics of Hematological Disorders improved research quality).
13. Omosule B, **Okunola O**, **Sanusi AA**, Arogundade FA. Gene Expression in Autosomal Dominant Polycystic Kidney Disease (ADPKD) in Ile-Ife. (Summit reinforced faster, cost-effective strategies for next generation sequencing of large cohorts, identification of mutation/s of adpkd3 undetected in the tropics).
14. **Paredes-Moscosso SR** (PI), Villegas-Llerena C, Buleje S, De León J, Guevara-Fujita ML, Acosta O, Fujita R. Generation of in Vitro Models Using CRISPR/Cas9 Technology: Modelling BRCA-1 ‘Variants of Undetermined Significance’ (VUS) Identified in Peruvian Patients with Breast Cancer. (Summit lectures deepened understanding, raised awareness in studying vus, particularly in underrepresented populations (peruvian); informed approach of project).
15. **Sayed I**. (PI), Abdel-Kader M (Co PI). Clinical and Molecular Characterization of Syndromic and Non-Syndromic Tooth Agenesis with Suggested Management in Cases with Severely Atrophied Alveolar Ridges. (Summit lectures improved skills in diagnosis of rare genetic disorders).
16. Temtamy S (PI), **Otaify GA**, Kholoussi N, **Sayed I**, Abdel-Salam G, El-Kamah G, El Ruby M, Aboul-Ezz E, **Mehrez M**, et al. Recent Advances in the Diagnosis, Management and Research of Genetic Diseases. (Summit lectures improved skills in diagnosis of rare genetic disorders).
17. Venkatesan S, **Mutreja D**, Sharma S. Flow Cytometric Detection of Minimal Residual Disease in Cases of B-Acute Lymphoblastic Leukemia. (Summit emphasized importance of gene-disease association with types of mutations in acute lymphoblastic leukemia).
18. Villegas-Llerena C (PI), **Paredes-Moscosso SR** (Co-I), Guevara-Fujita ML, Acosta O, Wray S, Cardenas B, Arber C, Fujita R. Molecular Platform for Corrective Gene Therapy in Retinitis Pigmentosa, Using CRISPR/Cas9-Modified Induced Autologous Stem Cells (Ipsc): Multifamily Case of Parán. (Summit lectures, discussions improved approach, execution of project).

2018 Batch

19. **Abdulkareem, FB** (PI), Badmos KB, Sonusi, SE, Kolawole HF. Autopsy Prevalence of Congenital Abnormalities Recorded in LUTH Carcinoma. (Summit highlighted need to document and maintain a register of congenital abnormalities).
20. Acharya M, **Dutta AK**, Mukherjee S. Understanding Genetic Architecture Underlying Hereditary Non- Syndromic Hearing Loss (NSHL). (Summit lectures and resources (ncbi) in genetics of hearing loss and variant prioritization were critical for this project).
21. **Adeyemo TA**, Akinsete AM, Ojewunmi OO, Akinsete A. Sickle Cell Disease Registry of Nigeria (SCDRN). Targets 5000 SCD for Accurate Clinical and Patient-Reported Data to Support Clinical Research, Interventions, Management and Quality of Care of Patients. (Summit initiated a registry).

22. Adisa RA, **Abdulkareem FB**. Quantification of Free Circulating Mitochondrial DNA Content and Detection of Alterations in the Plasma in HIV-Associated HCC or Cervical Cancer. (Summit provided insight and understanding of molecular research).
23. Ajayi Gbenle O, **Abdulkareem FB**, Odubanjo MO. CD70 Expression Patterns in Renal Cell Carcinoma (Summit lecture on cancer genomics influenced choice of project).
24. **Cornejo-Olivas M**, Cosentino C. Study for Research on Genetics of Parkinson Disease in Peru, a LARGE PD Study. (Summit lectures in bioethics and scientific writing helped improve informed consent form and writing of proposal).
25. **Cornejo-Olivas MM**, 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-effect of variants on phenotype of triplet expanded disorders, improved study execution).
26. **Dueñas-Roque M**, Mayorga G, Prötzel A, Gamarra N, Ota A. Birth Defects Registry in the Health Social Security of Peru. a Pilot Study at a Reference Hospital. (Through Summit met researchers who motivated work in congenital malformations).
27. **Dueñas-Roque M**, Segura P, Soria M, Calderón M, Rondón P. Epidemiology of Lysosomal Storage Disorders in the Health Social Security of Peru. (Summit Encouraged Research in Peru).
28. Hallowell M, **Edem-Hotah J**, Falisse J-B, Sridhar D, Saikat S. RE_EMERGE: Research to Sustain Universal Health Coverage in Guinea, Liberia and Sierra Leone in the Covid-19 Era. (Knowledge from Summit on human skin microbiome in health and disease, helped in the design and implementation of project).
29. **Kemever-Asima D**, **Samuel Wiafe S**, Akyaw P, Kubi EA, Thomford NE, Gyening-Yeboah A, Adu-Gyamfi E, Okusaga D, Ahiable EA. Establishing a Registry for Undiagnosed, Congenital and Rare Disorders in Ghana (Testing Phase). (Summit helped understand link between congenital diseases and undiagnosed rare diseases, improved design and initiation of registry).
30. Kosiyo P, Otieno W, **Gitaka J**, Nyamuni J, and Ouma C. Sickle Cell Genotypes in Malaria. (Summit enhanced sickle cell work through learnt molecular techniques; improved protocol development).
31. **Messaoud O**, Romdhane L, Naouali C, Belhaj O, Abdelhak S, Zghal M, Jones M, Maazoul F, Mezzi N. Genetic Characterization of Atypical Forms of Photogenosermatosis. (Knowledge gained from personalized trainings at summit helped interpret incidental findings, exome data analysis).
32. Negi V, **Dwivedi A**, Ravi H, Goswami JN, Kumar R, Behl A. Diagnosis of Inherited Disorders of the Brain, Heart and Kidney at Army Hospital Research & Referral, New Delhi. (Knowledge gained at summit helped conceptualize, execute project).
33. Pericak-Vance M (PI), **Cornejo-Olivas M** (PI-Site Peru). Genetics of Dementia in a Peruvian Cohort. (Summit lectures in bioethics and scientific writing helped improve the informed consent form and writing of proposal).
34. **Utumatwishima JN**, Ingabire A. the Effectiveness of Community Public Health Response on COVID19 in Rural Area. (Summit training in leadership in research, understanding data and communication skills facilitated my selection as a team lead, to guide scientific evidence and epidemiological response to COVID-19).
35. **Utumatwishima JN**, Lassalle D, Mancheski B. Clinical Trial to Assess the Treatment of Schizophrenia with Paliperidone Palmitate in Rwandan Healthcare Settings (CASPAR). (Skills gained at summit through genetics conferences on good clinical practice, bioethics, data quality and clinical research, facilitated selection as a PI).

2017 Batch

36. Aglan MS, **Otaify GA**. Targeted Next-Generation Sequencing in the Diagnosis of Osteogenesis Imperfecta and Bruck Syndrome. (Knowledge gained from summit helped improve reporting of results).
37. **Benítez Cordero Y**, Suárez Besil B. 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 registries for genetic epidemiological studies).
38. **Casado PL**, Telma Aguiar T, Valquiria Quinelato T. Analysis of 7 Different Techniques to Isolate Microna. (Summit Bioinformatics lectures helped understand relationship of miRNA with DNA and mRNA, improved execution of project).
39. **Casado PL**, Telma Aguiar T, Valquiria Quinelato V. Genetic Profile of Patients That Underwent Peri-Implant Maintenance Therapy: Relationship with Peri-Implant Disease Incidence. (Summit highlighted characteristics to be considered in studying multifactorial diseases; helped improve sample collection, medical history analysis, selection of related genes).
40. Ceylanoğlu KS, **Tuncay Yaylacioglu F**, Konuk O. Are Foxp3 Polymorphisms Risk Factor for Development of Ophthalmopathy in Patients with Graves' Disease? (Summit improved my knowledge in bioinformatics and genetic data analysis).
41. **Dhoro M**, M. Bwakura. Establish a Biobank for Cohort of Children Hospitalised for Severe Acute Malnutrition (SAM); Study the Role of Genetics. (Summit helped develop Nutrigenomics study).
42. Dinçer P, Kürekçi GK, Kural E, **Tuncay Yaylacioglu F**, Önal G, Ünsal S. Preparation and Maintenance of CRISPR/Cas9 Platforms in Zebrafish Specific to Rare Diseases. (Summit helped meet researchers interested in zebrafish, shared knowledge and experience).
43. **James O**, Adekunle AA. a Comparison of 2-OCTYL Cyanoacrylate (DERMABOND®) Tissue Adhesive and Sutures in the Repair of Cleft Lip at the Lagos University Teaching Hospital - a Randomized Controlled Clinical Trial. (Summit gave insight in exploring better care options for subjects born with orofacial cleft).
44. **James O**, Amoo AT. Assessment of Early Surgical Site Complications Following Primary Cleft Lip Repairs: a Prospective Study (Summit gave insight in exploring better care options for subjects born with orofacial cleft).
45. **Lotz-Esquivel S**, Alvarado-Aguilar M. Acute Intermittent Porphyrria. (Knowledge from Summit provided tools for soliciting specialized treatment and laboratory tests for patients).
46. **Mahfoudh W**, Faleh R. Circulating Tumor DNA as Biomarkers for Early Diagnosis, Treatment Response and Prognosis of Breast Cancer. (Summit lectures and tailored training inspired the project).
47. Marcheco Teruel B, **Benítez Cordero Y**, Suarez Besil B, Rojas Betancourt IA, Lantigua Cruz PA. Evaluation of the Impact of the National Program for Diagnosis, Handling and Prevention of Genetic Diseases and Congenital Defects. (Summit reaffirmed usefulness of data for personalized medicine, clinical validity of gene-disease associations, registries for genetic epidemiological studies).
48. **Roy S**. Clinico-Pathological Features of Retinoblastoma and Treatment Outcome in a Tertiary Eye Care Center of Bangladesh (Summit provided contacts with ocular oncologists and researchers; exchange of ideas facilitated first study in Bangladesh).
49. Suarez Besil B, **Benítez Cordero Y**, Álvarez Gavilán Y, **Roblejo Balbuena H**, Rojas Betancourt IA. Community Strategy for The Promotion-Prevention of Genetic Health in Primary Health

Care. (Personalized training at Summit provided elements for identification, management of risk factors, design of investigation, tools for genetic counseling).

50. **Torres-Mejía G**, Rinaldi S, Romieu I, Lazcano-Ponce E, Angeles-Llerenas A, Ortega-Olvera C, Sánchez-Zamorano LM. Molecular Subtypes of Premenopausal Breast Cancer in Latin American Women (PRECAMA): a Multicenter Population-Based Case-Control Study. (Knowledge from Summit permitted inclusion of genomic studies).
51. **Torres-Mejía G**, Vaca-Paniagua F, Rodriguez-Valentin R, Romieu I. Identification of Pathogenic Variants of Susceptibility to Early-Onset Breast Cancer in Young Women (Knowledge from Summit allowed expansion of research networks with Mexican institutions).
52. **Tuncay Yaylacioglu F**, Ergün SG, Talim B, Dincer PR. Evaluation of Phenotypic Effects of CRISPR/Cas9 Mediated CHST6 Knock-Out on Zebrafish Cornea. (Through Summit met researchers interested in zebrafish; shared knowledge and experience).

2016 Batch

53. Besil Suárez B, **Benítez Cordero Y**, **Roblejo Balbuena H**. Clinical-Epidemiological Surveillance of Microcephaly and Other Congenital Defects After Infection by Zika in Pregnant Women, Products of Conception and Cuban Newborns. (Summit strengthened concept of congenital birth defects and medical history analysis).
54. **Deniz E**. Genome-Wide Screening with CRISPR/Cas9 and Modelling of Resistance Mechanisms Developed Against Cytotoxic Drugs in Cancer Treatment. (Knowledge from Summit in genomics and personalized medicine helped analyze bioinformatics data).
55. **Ekure EN**, Adeyemo AA, Sokunbi OJ, Muenke M, Kruszka P. Case Reports on Genetic Diseases Associated with Congenital Heart Defects -Emery-Dreifuss Muscular Dystrophy Syndrome. (Knowledge from summit improved case reporting).
56. Mekkawy MK, **Hussen DF**. Molecular Cytogenetic Characterization and Breakpoint Mapping of Chromosomal Rearrangements Towards Better Management of Patients with Genetic Disorders and Carriers. (Cytogenetic techniques learnt at the Summit helped diagnose genetic disorders).
57. Olatosi O, **Adeyemo WL**, Butali A. Investigating the Genetic Causes of Early Childhood Caries. (A side talk during the Summit by Dr. Guverder (NIAMS), on genetic aetiology of dental caries influenced project).
58. Ranadeva NDK, Noordeen N, Wetthasinghe TK, **Sirisena ND**, Dissanayake VHW. Design and Implementation of Novel Pharmacogenomic Assays for CYP2D6*10 and CYP2D6*41 Variants to Predict Response to Tamoxifen Therapy in Breast Cancer Patients. (Knowledge from Summit supported study design and implementation).
59. **Roblejo Balbuena H**, Marcheco Teruel B, Monzón Benítez G. Location of Genetic Susceptibility Markers Associated with the Origin of Schizophrenia in the Cuban Population. (Summit provided tools to analyze results of WES, using NGS).
60. Sjarif DR, **Ariani Y**, Priambodo R. Lysosomal Storage Disease Study: Developing Screening and Diagnostic Methods for MPS II, MPS IVA, Gaucher Disease, Pompe Disease and Neuronal Ceroid Lipofuscinosis Type 2. (Summit lectures on Inborn Errors of Metabolism, Tools for Molecular Genetics, Diagnostic Odyssey, Variant Interpretation and Variant Nomenclature, inspired this project).

F. Other Accomplishments/Initiatives:

2019 Batch

1. **Abubakar S.** Appointed, Guest Researcher, Social Network Methods Section (NHGRI), Visiting Program, U.S. Department of Health and Human Services (2020); Developed, integrated, ethical, legal & social implication of Genetics and Genomics into Medical Ethics & Law Course for graduate programs (MPH; Global Health), College of Health Sciences and the World Bank Funded Africa Center of Excellence for Population Health and Policy, Bayero University, Nigeria; Delivered lectures on 'Introduction to Genomics', 'Genetics and ELSI' (2019); Registered NGO - Cigaba Health Empowerment Research & Development Foundation with Corporate Affairs Commission of Nigeria (2020), for community/public engagement in genetics and genomics (counseling services, advocacy for sickle cell disease and newborn screening) in Nigeria; Approved by Management of Aminu Kano Teaching Hospital, to establish a Preventive & School Health Promotion Clinics Rural Complex at Comprehensive Health Centre, Kumbotso, Kano for newborn screening, genetic counseling, community & school health outreach programs; Reviewer for BMC Health Services Research, Springer Nature (Global Publishers, 2019).
2. **Akinmola OO.** Lectured on 'Management of neonatal diabetes', 'Genetics as a tool for precision medicine to optimize outcome for patients', 'Newborn screening: Neonatal hypothyroidism', to Pediatric Endocrinology Training Centre for West Africa; Introduced genetics and genomic medicine in Residency training program, Department of Clinical Chemistry, Lagos University Teaching Hospital, Idi-Araba Lagos State.
3. **Alamin TA.** Presented, poster on 'Polymorphisms in GSTM1 and GSTT1 among Sudanese SLE patients', at 2nd Conference of the Arab Association of Genetic Research (Egypt, Nov. 2019); Submitted abstract on 'Genetic Polymorphism of the Glutathione S-Transferase Tp1 Gene in Sudanese patients with Sickle Cell disease', to 15th Annual Sickle Cell and Thalassemia Conference (virtual, Oct. 2020); Used materials, resources from Summit to redesign, update curriculum in cytogenetic tools and applications for graduate school (M.Sc.); Facilitated inclusion of Faculty of Medical Laboratory Sciences, University of Khartoum, in the 'H3Africa Introduction to Bioinformatics' training course (2020).
4. **Alli LA.** Lectured in genetics and genomics to medical and graduate level students (MSc); Completed online courses in 'Genetic counselling', 'The genomics era: the future of genetics in medicine' (Apr. 2020); Attended, 5th Global Genomic Medicine Collaborative Conference on Genomic Medicine Implementation in Low Resources Settings (virtual, May 2020).
5. **Amarakoon GGGT.** Lectured on 'Human genetics and hemoglobinopathies' to undergraduates, Faculty of Medicine and Allied Sciences, Rajarata University of Sri Lanka; Participated in workshop on Congenital Malformations, 9th International Conference on Birth Defects and Disabilities in the Developing World (Sri Lanka, Feb. 2020); Submitted case reports on 'Child with 'Southeast Asian Ovalocytosis' and 'An Infant with Congenital Urachal Sinus', to 13th International Research Conference of General Sir John Kotelawala Defense University (Sri Lanka, Oct. 2020); Initiated database for syndromes, congenital malformations, genetic disorders, University Pediatric Department, Teaching Hospital Anuradhapura; Initiated establishment of Genetics Clinic and Multidisciplinary Care Center; Identified, diagnosed and treated cases (21) – syndromes (13), biliary atresia (2), congenital neutropenia (1), hemoglobinopathies (4), southeast Asian ovalocytosis (1).
6. **Baatar N.** Implemented, 'School Health Promotion Project' in Mongolia; Co-founded Mongolian Society of Pediatric Research and Study; Signed agreement on scientific, technical cooperation

- between (a) Mongolian Society of Pediatric Research & Study (b) Children's Hospital SPPH, Shaanxi Provincial Center & Research of Pediatric Clinic; Attended 7th Asia-Pacific Gastroenterology Cancer Summit (virtual, Apr. 2020); Attended Summer School of Clinical Genetics, Asia Pacific Society of Human Genetics (virtual, Aug. 2020); Attended 4th International BRCA Forum (virtual, Aug. 2020); Referred leukemic pediatric cases (3) to China and Turkey for diagnosis, treatment due to limitations in Mongolia.
7. **Bocoum A.** Provided training on 'Grant writing', at Youth Association, Djenné (Mali, Aug. 2020); Registered with International Parkinson Disease Genomic Consortium, Africa section; Attended 'Journées de Neurologie de l'association Sénégalaise de Neurologie' (Senegal, Feb. 2020); Sponsored, attended MDS training on 'Parkinson's disease and atypical Parkinsonism: Neuroimaging and Emerging Concepts' (S. Africa, Mar. 2020); Presented on 'Clinical and etiological aspects of movement disorders in low income country: case of Mali', at MDS Congress (virtual, Sept. 2020); Initiated recruitment of patients with movement disorders at Teaching Hospital of Gabriel Touré, Bamako, Mali.
 8. **Chowdhury E.** Joined as Specialist, Department of Pediatrics, Samtse General Hospital, Royal Government of Bhutan (Feb. 2020); Previously, Associate Consultant at Department of Pediatrics, Chittagong Eye Infirmary and Training Complex (CEITC), Chittagong, Bangladesh; Lectured on 'Human genetics and genomics' to medical students at CEITC; Trained colleagues to establish research/clinical facilities in CEITC, emphasized on personalized care, genetic counseling; Presented on 'Common genetic diseases in children and recent 'Management protocol', for CME program, Bangladesh Medical Association, Chittagong; Initiated registry for congenital abnormalities in Department of Pediatrics, CEITC; Identified, managed pediatric genetic disorders cases (47) at CEITC; Diagnosed cases- Thalassemia (15), Hemophilia (12), Down's Syndrome (9), Turner's Syndrome (5), Duchenne Muscular Dystrophy (3), Achondroplasia (2), Neurofibromatosis (1); Referred cases for cardiac surgery (7), ophthalmic surgery (3), physiotherapy (1), general surgery (1); Improved quality of life of affected individuals and families, due to known prognosis and access to appropriate support systems.
 9. **Guven Y.** Invited speaker, presented on 'Ectodermal dysplasias and treatment approaches: a dentist's perspective', at 1st Bursa International Genetics Days: Dermatogenetics Symposium; Diagnosed, referred patients (6) to genetics department for molecular confirmation and medical referral- Ectodermal dysplasia (3), Papillon Lefevre syndrome (2), Gorlin-Goltz syndrome (1); Contributed to social and psychological well-being of children, improved quality of life, prevented fatal consequences through early diagnosis, surgical interventions, dental prosthodontic therapies, and preventive and follow-up care.
 10. **Gyawali P.** Lectured on 'Basic molecular biology', 'Molecular diagnostic technology' and 'Genetic disorders (single gene and chromosomal)', to MBBS and BDS students using synchronous and asynchronous online teaching strategies; Attended, Global Alliance for Chronic Disease (GACD) and Health System Research Institute (HSRI) Implementation Science School (Thailand, Nov. 2019); Member, Project Management Committee, for 'A cluster randomized trial of a community-based lifestyle intervention for diabetes management, in Kavrepalanchowk and Nuwakot district in Nepal' (GACD/Agency for Medical Research and Development project); Attended workshop on Designing Program in Open and Distance Learning, Indira Gandhi Open National University (India, May 2020); Completed COURSERA certificate courses on- 'From Disease to Gene and Back (Novosibirsk State University, May 2020)', 'Introduction to Genomic Technology' as part of 'Genomic Data Science Specialization' (John Hopkins University, Jul. 2020), 'Understanding Medical Research' (Yale University, Jul. 2020); Completed certification

on COVID-19, AACC Learning Lab for Laboratory Medicine on NEJM Knowledge+, Massachusetts Medical Society (2020); Facilitator, trainer for faculty training in Moodle Learning Management System (LMS).

11. **Hussein N.** Coordinator for Postgraduate Family Medicine Specialty, University of Malaya, Malaysia; Strengthened genetics and genomics curriculum in Family Medicine postgraduate courses; Lectured on ‘Genetics and genomics in primary care’; Led discussions on ‘Psychosocial issues in genetics’; Planned topics for ‘Cancer genetics’ and workshop on ‘Communicating genetic risk’; Mentored early-career lecturers, graduate students (Master of Family Medicine) in genetic and health research; Received scholarship to attend RARD (S. Africa, Jun. 2020).
12. **Iqbal M.** Supervised, graduate research students (3 PhD, 8 MS) on molecular and genetic basis of different ophthalmological disorders; Developed curriculum, delivered lectures on ‘Human molecular genetics’ to graduate level students; Reviewed research articles (3) for The Journal of Animal and Plant Sciences (JAPS).
13. **Kars ME.** Qualified, PhD exam in Molecular Biology and Genetics (Jan. 2020), presented thesis proposal to committee (Jun. 2020), knowledge and resources from Summit helped in preparation; Shared knowledge from Summit with fellows, undergraduate students during their internship in Human Genetics Laboratory; Used materials from Summit for teaching assistantship in ‘Human Genetics and Molecular Genetics’ (2019-2020).
14. **Khant AK.** Provided CME for doctors, nurses in pediatric acute kidney injury and kidney health, at Taungoo, Pyay (Nov. 2019), and at HPa-An, Mawlamine General Hospital (Feb 2020); Awarded travel grant, presented poster on ‘Systemic lupus erythematosus presenting as hemolytic uremic syndrome’, at 18th Congress of International Pediatric Nephrology Association (Italy, Oct. 2019); Speaker on ‘Pediatric renal transplantation at Yangon Children Hospital’, at 4th Myanmar Nephro-Uro Conference Yangon (Myanmar, Oct. 2019); Examined, diagnosed and treated children (~500); Counseled patients with hereditary renal disease (congenital nephrotic syndrome, congenital abnormalities of kidney urinary tract, meningomyelocele with neurogenic bladder, syndromic child with renal abnormalities, Lowe syndrome with renal tubular acidosis, focal segmental glomerulosclerosis and others); Performed renal biopsy of nephrotic syndrome, SLE, and renal transplant patients.
15. **LLamos-Paneque A.** Professor, Human Genetics, School of Dentistry, International University of Ecuador; Updated major topics in human genetics; Lectured on ‘Clinical genetics’, at University and Army Hospital of Quito; Developed basic genetics content with Drs. **López-Star B** and **Ortiz-Panozo E** (Summiters), for course in ‘Genetics in Ophthalmology’, at Mexican Institute of Ophthalmology; Summit improved genetic counseling skills, provided access to research groups and funding sources; Collaborated with Dr. Karen Heath, Institute of Medical and Molecular Genetics, La Paz University Hospital, Spain for bone dysplasia (20 cases) genetic panel studies unavailable in Ecuador; Selected Clinical Geneticist, for ‘Train the trainers’ by BioMarin, for CLN2 and MPS disorders; Applied for CME, Mexican Association of ‘Human Genetic: News and Challenges in Inborn Errors of Metabolism and Neonatal Screening’ (Jul. 2020); Awarded travel grant by CENTOGENE to present on ‘Recent advances in rare diseases’, at RARD 2020 (S. Africa; postponed due to COVID-19); Evaluated genetic cases (800) of monogenic etiology, confirmed cases by molecular studies (200), cytogenetic studies (138), others were multifactorial (462); Registered new cases (338) in the ‘Registry of Genetic Diseases’ of the Specialty Hospital No.1 FF.AA, Quito, Ecuador.
16. **López-Star B.** Started, Ophthalmo-genetics Department at Mexican Institute of Ophthalmology; Trained junior staff in ophthalmo-genetics; Developed basic genetics content with Drs. **LLamos-**

- Paneque a and Ortiz-Panozo E** (Summiteers), for course in ‘Genetics in Ophthalmology’, at Mexican Institute of Ophthalmology; Member of Pan American Inherited Retinal Diseases Group (Feb. 2020); Speaker, on ‘Survey of practice patterns for management of ophthalmogenetic disorders among Mexican pediatric ophthalmologists’, at Ophthalmopediatric Mexican Society (Mexico, Jul. 2020); Established system to record, monitor patients with inherited retinal diseases; Enrolled cases (118); Evaluated, diagnosed, treated ophthalmogenetic cases (20); Referred cases (10) for medical management to Children’s and Women’s Hospital, Mexico; Improved quality of life of affected individuals and families due to known prognosis, access to appropriate support systems, and informed decisions related to family planning.
17. **Luong LH.** Lectured on ‘Clinical genetics’ at hospitals to increase awareness, integrate genetics in clinical practice; Invited speaker, presented on ‘Clinical genetics in Vietnam, challenges and opportunities in a developing country’, at 64th Annual Meeting of Japan Society of Human Genetics (Japan, Nov. 2019); Invited speaker, presented on ‘Clinical genetics- current trend and challenges, the need for interdisciplinary collaboration’, at 2nd Vietnam- North American Conference of Medical Genetics and Genomics, Hanoi (virtual, Nov. 2020); Invited reviewer, for Journal of Clinical Rheumatology and Natural Product Communication; Diagnosed cases (~50), counselled patients and families affected by neuromuscular disorders and inherited immune disorders -Primary Hypertrophic Osteoarthropathy, Job’s Syndrome.
 18. **Mhandire K.** Awarded, Harry Crossley Postdoctoral Fellowship by University of Cape Town (S. Africa, Jul. 2020); Lectured on ‘Molecular pathology’ and ‘Inborn errors of metabolism’ modules to third year MBChB students, University of Zimbabwe (2019); Presented, seminar on ‘International Summit in Human Genetics & Genomics: My experience’ to Division of Human Genetics, University of Cape Town (Oct. 2019); Completed online course in ‘Statistics and Data Science Specialization’, Coursera (Apr. 2020); Learnt programming languages R, Python 3 and Unix for Bioinformatics (2019-2020); Co-supervised an undergraduate Honors and graduate level (2 MSc) on Pharmacogenetics and HIV drug resistance projects (S. Africa; Zimbabwe, 2020).
 19. **Monye HI.** Elected Member, Royal College of Surgeons of Edinburg, MRSCEd (Ophthalmology, Feb. 2020); Speaker, on ‘A second chance at sight - a twelve year review of penetrating keratoplasties at the University College Hospital, Ibadan’, at World Ophthalmology Congress (virtual, Jun. 2020); Upgraded to electronic record keeping for genetic eye disorders at the clinic, University College Hospital Ibadan; Summit lectures, field trips, online courses improved genetic counseling skills; Used social media to create awareness on important causes of blindness (glaucoma, childhood cataracts, retinoblastoma) through a non-profit, EyeHubNigeria Initiative; Identified, diagnosed, treated/referred for medical and/or surgical management patients (58) with genetic eye diseases -primary open angle, juvenile open angle, childhood glaucoma (40); retinoblastoma (3); pediatric cataracts (15).
 20. **Mosema KBA.** Participated, Co-chaired ‘Genetics and genomics solutions in DRC’ session, at International Congress of Infectious and Parasitological Pathologies (DR. Congo, Nov. 2019); Trained medical students, young medical doctors on use of pedigree for suspected genetic disorders, Biamba Marie Mutombo Hospital, Kinshasa City; Trained health workers (100, included 50 maternities) for detection of visible malformations during child birth in Kinshasa City; Established systematic screening procedures for SCD in children, with follow-up care (<5yrs) at Biamba Marie Mutombo Hospital, Kinshasa City; Provided guidance to General Practitioners in remote areas (Kwilu and Tshuapa provinces) on diagnosis of genetic disorders, primarily in sexual development.

21. **Mushi TL.** Enrolled in Master's Degree in Pediatric and Child Health, at Muhimbili University of Health and Allied Science, Tanzania (2019); Presented, poster on 'Outcome and challenges of transcatheter procedures in low-income countries: a case of Jakaya Kikwete Cardiac Institute, Tanzania' (Dec. 2019); Attended, focused on 'Current practices and role of geneticists in medical and surgical management', at Clinical Cardiogenetics: Myth and Truth conference, organized by Congenital Heart Academy (virtual, Jun. 2020); Worked on utility of genetic testing in the pediatric cardiac intensive care unit (Jun. 2020).
22. **Mutreja D.** Lectured on genomics to residents, medical students, nursing graduates, paramedical trainees; Shared resource materials from Summit with medical students, residents, colleagues from other specialties; Suggested revision of competency based medical school curriculum; Initiated and applied for Newborn Screening Program for Indian Armed Forces; Identified cases (18) by histopathologic/microscopic diagnosis- Acute Leukemia in Down's syndrome (2), thalassemia (12), sickle cell disease (2), Gaucher disease (1), odontogenic keratocyst in Gorlin Goltz syndrome (1); Section Editor (Pathology) for Indian Journal of Medical Pediatric Oncology (2019-20).
23. **Nair L.** Completed Residency in Medical Genetics; Awarded Best Oral Paper, presented on 'A new multiple malformation syndrome with sacroccygeal agenesis and lower limb aplasia associated with bi-allelic variant in the TBX4 Gene', at 6th Annual Conference of the Society for Indian Academy of Medical Genetics (India, Nov. 2019); Completed online courses on 'Epigenetic control of gene expression', University of Melbourne (May 2020), 'From disease to genes and back', Novosibirsk State University, Russia (May 2020).
24. **Okunola O.** Awarded, Training Fellowship in Stigma Research, Fogarty International Centre/NIH (2020), Summit influenced selection; Member, Faculty Curriculum Review Committee for integration of genetics/ genomics under Fulbright Fellowship Curriculum Development Pathway (2021); Trained, mentored medical students, resident doctors, colleagues in medical genetics, genomics, research, grant applications; Used experience and materials from Summit; Liaison, study group with pediatric nephrologists to establish congenital kidney disease registry and follow up; Team lead, pioneered Newborn Screening Consortium in SW Nigeria with resources from the Summit; Diagnosed patients (15) with inherited kidney diseases, screened, followed up on siblings; Patients and families reported improvement in quality of life due to accurate diagnosis and pathway to care.
25. **Ortiz-Panozo E.** Joined new research group in breast cancer genomics, presented detailed information on Summit; Developed basic genetics content with Drs. **LLamos-Paneque A, Lopez B** (Summitteers), for course in 'Genetics in Ophthalmology', at Mexican Institute of Ophthalmology, Mexico; Attended 'Genomic sciences data analysis with R', Summer Program, National Institute of Public Health, Mexico (2020).
26. **Paredes-Moscoso SR.** Participated as NGS user, in First International seminar on 'Technological perspectives in the Americas' (Peru, Oct. 2019); Discussed impediments in widespread use of NGS, information referred to CONCYTEC (Peruvian scientific agency) for review; Guest at round table session, First symposium Henrietta Lacks – 'Bioethical aspects on the handling of biological samples' (Peru, Oct. 2019); Participated in clinical bioinformatics course organized by Univ. de San Martín de Porres and CABANA, UK to strengthen bioinformatics in Latin America (Peru, Nov. 2019); Speaker on 'Highlights and insights from the International Summit in Human Genetics and Genomics, NHGRI' at Encuentro Científico Internacional (Peru, Jan. 2020); Speaker on 'Genetics in hereditary breast cancer: From variant analysis to functional genetics', at V Jornada Científica Internacional en Biociencias (Peru, Jan.

- 2020); Invited reviewer by Dr. **Pîrlog R** (Summiteer), for ‘Innovation in Science’ call by Asaih, a Romanian entrepreneurship association, connects students from Iuliu Hatieganu University of Medicine and Pharmacy (Romania) with local and international stakeholders; Selected member, UKRI International Development Peer Review College, part of Global Challenges Research Fund (GCRF)/Official Development Assistance (ODA) in research (Jun. 2020).
27. **Pîrlog R**. Presented Summit highlights in two open scientific sessions at Romanian Society of Bioinformatics (2019), and to medical residents of Iuliu Hatieganu University (2020); Participated in educational module on cytogenetics and immunohistochemistry for pathologists, organized by Bucharest Oncologic Institute, Romania (2019); Attended AACR Annual Meeting (virtual, Apr. 2020); Attended European Association for Cancer Research Congress (virtual, Jun. 2020); Started Molecular Pathology Research Laboratory as an integral part of Research Center for Functional Genomics, Biomedicine and Translational Medicine at Iuliu Hatieganu University of Medicine and Pharmacy PhD School; Developed training courses in research and entrepreneurship for PhD students, post-docs to facilitate technology transfer from bench to market.
 28. **Sayed I**. Helped prepare course on ‘Basics of oro-dental genetics’ to introduce genetics to dentists; Organized, presented poster on ‘WNT10A gene mutational analysis in ten Egyptian ectodermal dysplasia patients’, at 2nd Conference of the Arab Association of Genetic Research (Cairo, Nov. 2019); Identified cases (125)- neurogenetic manifestation (88), skeletal manifestation (25), ectodermal dysplasia patients (12); Referred patients (7) for prosthetic management of ectodermal dysplasia.
 29. **Tiong SY**. Elected, Committee Member, Genetic Counselling Society Malaysia (Dec. 2019); Involved in planning/discussions for establishing molecular laboratory for NIPT service at Loh Guan Lye Specialists Centre, Penang (Malaysia, Oct 2019); Presented on ‘IEM Screening’, at Loh Guan Lye Specialists Centre (Dec. 2019); Helped establish IEM screening service (optional) for newborns at Loh Guan Lye Specialists Centre (2019) - previously done by appointed private labs only (avg. uptake 4%); Team member for planning Genetic Counsellor Registration and Credentialing Pathway, Malaysia (Aug. 2020); Referred new cases (28) for genetic counselling.
 30. **Tumulak MJR**. Served on Organizing Committee and resource speaker on ‘Genetic counseling of thalassemia’ at pre-conference events- ‘Medical Biotechnology and Molecular Biosciences’ and ‘Genetic Counseling Workshop’, at Asia Pacific Conference on Human Genetics (Philippines, Nov. 2019); Invited speaker, on ‘Genetic counseling in cancer’, Asian Hospital and Medical Center (Jan. 2020); Genetic counselor on studies ‘Determination of the prevalence of genetic mutation in high risk Filipino hereditary breast cancer patients’ and ‘Genotypic and phenotypic variations of inherited retinal degenerations among Filipino patients in a tertiary hospital’; Moderated ‘Congenital diseases and newborn screening’, Newborn Screening Society of the Philippines (Jul. 2020); Moderated ‘Prenatal genetics’, Institute of Human Genetics (Aug. 2020); Organized monthly webinars in tele-genetics counseling; Thesis panel member for genetic counseling students (9); Provided pretest, post-test genetic counseling to patients – breast cancer (100), cystic fibrosis (50), alpha and beta thalassemia (300), ophthalmology (3), X-linked dystonia parkinsonism(15), Duchene muscular dystrophy (5).
 31. **Weldemichael YG**. Delivered seminars on ‘Genetics in health and disease’, at Mai Nefhi College of Science, Eritrea and Orotta College of Medicine and Health Sciences Community, Eritrea; Used Summit materials and resources; Shared knowledge, increased genomic literacy among healthcare providers, counselors managing patients with autism, Down syndrome, other

genetic disorders, and cared for by National Association for Intellectual and Developmental Disability of Eritrea.

32. **Wiafe SA.** Completed online course in ‘Genetic counseling’, Wellcome Genome Campus Advanced Courses and Scientific Conferences (Sept. 2020); Represented Rare Disease Ghana Initiative at Rare Disease International (RDI, Oct. 2019); Speaker on ‘How to serve patients living with Rare Diseases’, Ted-Talk (Dec. 2019); Member, International Rare Disease Research Consortium; Appointed on Workgroup for Treatment Access, and on Patient Advocacy Constituent Committee (Jan. 2020); Attended, 8th Conference of Undiagnosed Diseases Network International (The Netherlands, Feb. 2020); Organized, 2nd Symposium on Rare Disease and Orphan Products (Ghana, Mar. 2020); Attended, 6th Annual Meeting -RDI General Assembly, served as a panelist (virtual, May 2020); Member, Global Genomic Medicine Collaboration, implementing Undiagnosed and Rare Disease Flagship Project (May 2020); Member, Undiagnosed Disease Network, Co-chair Developing Nations Working Group (Aug. 2020); Participated in focus group on Collaborative Global Network, Rare Diseases, shared perspective from WHO, Africa region (virtual, Jun. 2020); Moderated webinar on ‘Indigenous populations’ health and launched Lyfe language project’ (virtual, Aug. 2020); Panelist on ‘Increased access to diagnostics and treatment in emerging rare disease markets’, World Orphan Drug Congress (virtual, Aug. 2020); Keynote speaker, on ‘Rare disease and mental health: Focus on indigenous populations and developing nations’, at the Rare Fair (virtual, Sept. 2020); Nominated in 3 categories for 2020 WEGO Health Awards.
33. **Yadav S.** Submitted Dissertation on ‘Evaluation of molecular alterations at 11p15.5 locus in isolated hemihyperplasia’ (Jan. 2020); Completed Doctorate in Medicine (Medical Genetics) from All India Institute of Medical Sciences (AIIMS), New Delhi (India, Jul. 2020); Speaker on ‘Cystic fibrosis- genetics and prenatal diagnosis’, at 20th Education Program for Parents of Children with Cystic Fibrosis, AIIMS (India, Nov. 2019); Delivered lecture at a workshop ‘Application of MLPA in genetic diseases’, to increase awareness among graduate students at AIIMS (Jan. 2020); Referred patients (~250) from various specialities to genetic clinic; Diagnosed patients with rare genetic disorders (45), provided antenatal counselling (74), and comprehensive management of hospitalized patients with inborn error of metabolism and storage disorders (15); Helped maintain a Birth Defect Registry at AIIMS.
34. **Yesilcinar I.** Enrolled in online certificate course in ‘Pharmacogenomics in Practice’, Precision Medicine Academy (Apr. 2020); Speaker, on ‘Genetic and Nursing in Turkey’, at 1st International, 2nd National Oncology Nursing Congress (Turkey, Dec. 2019); Invited speaker, on ‘Post-test ‘Counselling: how to convey the gene test results to the patient & family’, at 4th Breast Cancer Virtual Conference (Oct. 2020).

2018 Batch

35. **Abdulkareem F.** Resource person on pathogenesis of colorectal cancer and pathology of viral hepatitis for ‘Advanced Pathology Course’, National Postgraduate Medical College of Nigeria (Feb. 2020); Performed faculty assessments for promotion, University of Kano, Ahmadu Bello University, Nigeria and University of Ghana Medical School, Ghana (2020); Member, Planning & Implementation Committee for new University of Medicine and Medical Sciences Abeokuta, Nigeria (2020); FAJIM Medicare Foundation (my NGO), gave scholarships to medical students (14; Nov. 2019); Invited facilitator, for session ‘Women in Oncology’, and presented abstract on ‘Annona senegalensis stem bark extracts orally pre-administered to wistar strain albino rats attenuated doxorubicin induced hepatotoxicity and oxidative stress’, at AORTIC (Mozambique,

- Nov. 2019); Abstract author, on ‘Mitoubiquinol mesylate (MitoQ) attenuated DEN-induced hepatocellular carcinoma through modulation of mitochondrial antioxidant defense systems’, at 8th Annual Symposium on Global Cancer Research (Apr. 2020); Chairperson/Moderator, parallel session on ‘Entrepreneurship and women empowerment in a global environment’, at International Conference of the Criterion, Madinah Institute of Science & Technology (Ghana, Aug. 2020); Invited panelist, on ‘Pathology of musculoskeletal tumors-Successes & challenges’, at Lagos Nigerian Medical Association Scientific Conference (Aug. 2020); Hosted webinar, ‘Hepatitis-free future: Taking responsibility’ on World Hepatitis Day, (Aug. 2020); Reviewer, ARGO sponsored pilot research grants applications (Aug. 2020); Provided free breast and cervical cancer screening for women on World Cancer Day (Feb. 2020); Diagnosed, documented congenital abnormalities cases (11) at autopsy (7 males, 4 females)- VSD (2) with Eisenmenger’s complex in one, tetralogy of Fallot (1), bilateral renal cysts (1), multiple congenital abnormalities (7).
36. **Adewole OO.** Lectured, trained resident doctors, medical students, in aspects of genomics, its implications and role in clinical practice; Shared knowledge from the Summit.
 37. **Adeyemo TA.** Course facilitator, ‘Genetic counselling workshop on SCD’, for Sickle Cell Foundation of Nigeria (Mar. 2020), delivered 4 lectures- ‘Hemoglobinopathy: Definition and types’, ‘Exchange blood transfusion’, ‘Basic principles of laboratory diagnosis of SCD’, ‘Practical steps in genetic counseling’; Mentored junior faculty (3), College of Medicine University of Lagos and Lagos University Teaching Hospital; Registered, Haemophilia Treatment Centre (HTC), Lagos University Teaching Hospital with the World Bleeding Disorder Registry; Identified, registered, counselled, managed care for patients (30) with inherited bleeding disorders.
 38. **Cornejo-Olivas M.** Appointed, Head, Department of Neuropathology and Delegate for Rare Disorders, Instituto Nacional de Ciencias Neurológicas; Mentored trainees (4) for the Northern Pacific Global Health Research Fellows Training Consortium (FIC/NIH), Global Brain Health Institute, University of California, San Francisco and for Young Members MDS Mentoring Program; Enrolled in Medical Genetics and Genomics Certificate Program (NHGRI, 2019); Review Editor and Guest Associate Editor, for ‘Frontiers in Neurogenetics’ and ‘Frontiers in Genetics of Common and Rare Diseases’ journals (participation due to skills gained at Summit in scientific writing and reviewing); Faculty staff, 3rd Pan American Parkinson’s Disease and Movement Disorders Congress (USA, Feb. 2020).
 39. **Daich Varela M.** Awarded, Clinical Research Fellow position in Moorfields Eye Hospital (UK, Jul. 2020); Presented talks on ‘SOX2 microphthalmia’ and ‘CHARGE syndrome’ at Ophthalmic Genetics Study Club (USA, Oct. 2019); Identified, diagnosed, treated cases (20)- CSNB, CHARGE, Coloboma, RP, Usher; Referred patients for counseling and medical management.
 40. **Das S.** Post-doctoral Fellow (Clinical Genetics), Department of Medical Genetics, Christian Medical College Hospital, Vellore, India (2019); Trained medical and nursing students in basic clinical genetics; Presented, poster on ‘Novel variant identified in LARP7 in an Indian patient with Alazami syndrome with expanded phenotype of buphthalmos’, at 61st McKusick short course, the Jackson Laboratory (virtual, Jul. 2020); Identified, diagnosed, counselled patients (472) 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).
 41. **Dueñas-Roque MM.** Professor, Genetics and Molecular Biology, Health Sciences Faculty, Universidad Tecnológica del Perú; Shared knowledge from Summit with undergraduate students; Co-authored conference report, 40th Annual David W Smith Workshop on ‘Malformations and

Morphogenesis: Abstracts of the 2019 Annual Meeting’, Am J Med Genet a (Mar. 2020); Enrolled in Medical Genetics and Genomics Certificate Program (NHGRI, 2019); Summit influenced establishment of Undiagnosed Disease Program (UDP) at Hospital Edgardo Rebagliati Martins, EsSalud; Enrolled cases (21 trios) in iHope Illumina Project to identify genetic causes of rare diseases in Peru.

42. **Dutta AK.** Summit helped understand importance and promotion of medical genetics in medical students at AIIMS, Kalyani, India, and instilled zeal for defining population burden of treatable rare disorders in India; Organized workshop on Next Generation Phenotyping, at Facial Dysmorphology Novel Analysis (FDNA) Inc. (India, Nov. 2019); Awarded, second prize for poster on ‘CEDNIK syndrome’, at 6th Annual Conference of SIAMG and the Indo-US Symposium on Genetic Neuromuscular Disorders (India, Nov. 2019); Presented poster on ‘Host genomics of COVID-19: Alpha 1 antitrypsin deficiency is a putative risk factor’ in Human and Mammalian Genetics and Genomics: the 61st McKusick Short Course (virtual, Jul. 2020); Speaker, on ‘How genomics is transforming clinical medicine’, at International webinar on Basic Research for Clinical Implications, Department of Zoology, Ranaghat College (India, Jul. 2020); Contributed to hospital’s Rare Disease Registry through determination of autosomal recessive IEMs (200), from publicly available NGS data (GenomeAsia 100K, IndiGenomes, gnomAD), preliminary data on Pompe Disease (unpublished); Volunteered for the ClinGen Community Curation Database.
43. **Dwivedi A.** Assistant Professor, Army Hospital Research and Referral (AHRR), New Delhi (India); First Clinical Geneticist, Indian Armed Forces; Used materials from Summit to educate fellow residents, researchers; Developed genetic OPD manuals for clinicians; Established Bioinformatics Lab for NGS analysis at AHRR; Conducted webinars on genetic testing, dysmorphology, spinal muscular atrophy, lysosomal storage disorders etc.; Attended, Annual Conference of Society of Indian Academy of Medical Genetics (India, Nov. 2019); Attended, European Study Group on Lysosomal Diseases WORLD Symposium (virtual, Feb. 2020); Invited Speaker, presented on ‘Care for Rare Disease Patients’, on World Rare Disease Day by Takeda (virtual, Feb. 2020); Member of Scientific Advisory Board of Sanofi Genzyme and Organization for Rare Diseases India; Created, designed various informational materials (pamphlets etc.) to increase awareness among armed forces personnel and families in genetic disorders, prenatal diagnosis, newborn screening and Down syndrome; Diagnosed patients (486) with various genetic disorders- chromosomal, Mendelian, Genomic imprinting disorders; Knowledge from Summit helped manage patients.
44. **Edem-Hotah J.** Invited by Dr. Mark Hallowell, University of Edinburgh, to present on ‘Insight into organizational expertise on emergency preparedness/outbreak management and university health coverage (UHC)- Status of the COVID-19 outbreak in Guinea, Liberia and Sierra Leone; and research priorities for COVID-19 in West Africa’, discussed genetics and genomics research for treatment of COVID-19 and other emerging infectious diseases (virtual, Jul. 2020); Included materials from Summit in undergraduate curriculum, University of Sierra Leone (USL) College of Medicine and Allied Health Sciences (COMAHS) Faculty of Pharmaceutical Sciences; Observed increased awareness for genetics and genomics among pharmacy faculty and students at COMAHS/USL and Technical staff of Sierra Leone Government Ministry of Health and Sanitation.
45. **Eshete MA.** Chair, Surgical Department Research Ethics Committee; Used materials from Summit to lecture residents, faculty members in research, graduate and undergraduates, Addis Ababa University; Presented on ‘The Amniotic Band Associated with severe and complicated

- clefts in the orofacial region’, at Annual General Assembly of the Ethiopian Society of Plastic and Reconstructive Surgeons; Provided support to families with cleft children during COVID.
46. **Gitaka J.** Established sickle cell surveillance at Mary Help of the Sick Mission Hospital, Thika, Kenya; Helped develop, strengthen genomics content in Biomedical curriculum, Murang’a University; Referred patients (8) for genetic counseling and medical management, helped allay anxiety through pathway to care.
 47. **Kemevor-Asima D.** Increased clinicians’ awareness on genomics in patient care and management; Nominated on work team in conjunction with West African Genetic Medical Centre, for inclusion of genetics in graduate Maternal Fetal Medicine programs; Worked on including genetics in Ob/Gyn clinical practice; Participated, helped organize 2nd Symposium on Rare Disease and Orphan Products, Rare Disease Day (Feb. 2020); Enrolled in Medical Genetics and Genomics Certificate Program (NHGRI, 2019); Initiated record keeping for congenital anomalies to track and monitor cases; Identified cases (10) antenatally, followed till delivery- AV septal defects (4), fetal hydronephrosis (3), bilateral cleft lip (1), fetal hydrocephalus (2); This facilitated multidisciplinary management at new Feto-Maternal Subspecialty Unit, Department of Obstetrics Korle-Bu; Collaborated with clinical psychologists for antenatal management of cases (3), counselling of women carrying babies diagnosed with anomalies, and follow-up care post-delivery.
 48. **Lertwilaiwittaya P.** Physician at, Division of Medical Genetics, Faculty of Medicine Siriraj Hospital, Mahidol University, Thailand; Presented poster on ‘Next Generation Sequencing in Dementia Subgroup in Thailand’, at American Society of Human Genetics Annual Meeting (Texas, Oct. 2019); Presented poster on ‘Identification of Novel Mutation by Whole Exome Sequencing in a Thai Family with Osteopetrosis’, at American College of Medical Genetics Annual Meeting (virtual, Mar. 2020); Lectured (online) in neurogenetic field; Enrolled in Medical Genetics and Genomics Certificate Program (NHGRI, 2019); Initiated new local NGS panel and Molecular Tumor Board for sex development disorders (SDD), cancer and dementia; Analyzed cases- SDD (43), cancer (332), dementia (88); Reported a variant to ClinVar; Initiated registry for breast-ovarian cancer, enrolled patients (306); Registered patients with neurogenetic disorders (317) and dementia (175) with Collaborative Aging and Dementia Research Society, Thailand.
 49. **Mburu S.** Mainstreamed teaching of Data Science and Machine Learning at School of Health Sciences, Kirinyaga University; Speaker, presented on ‘Targeting the seven cancer hallmarks by amelioration of oxidative stress-induced inflammation’, at National Research Fund Board Meeting, Naivasha (Nov. 2019); Presented on ‘Sustainable analytical and predictive modeling approaches for cancer care in resource-limited settings’, at the third KyU International Conference, Kirinyaga University (Nov. 2019).
 50. **Messaoud O.** Used materials from Summit for lectures on ‘Human genetics’ and ‘Genetic Variant Nomenclature’ for undergraduates, at Faculty of Medicine of Tunis; Member-in-charge, Working Group ‘Health and Society’, Arab-German Young Academy of Sciences and Humanities; Participated in ‘Engaging citizens in genomics research: the first steps for the establishment of a participatory action research in omics in Tunisia and North Africa’, at the 2nd AAGR Conference (Egypt, Nov. 2019); Helped organize Rare Disease Day (Tunisia, Feb. 2020); Invited speaker, presented on ‘Next Generation Sequencing and Related Challenges’ at Phi’s Research and Innovation Summit (virtual, Jul. 2020); Invited speaker, presented on ‘Women in science: What are the barriers for women in senior career positions’ and ‘The Future of Genomic Application’, Arab Science Week (virtual, Aug. 2020); Enrolled in Medical Genetics and

- Genomics Certificate Program (NHGRI, 2019); Reviewer, Informatics in Medicine and Frontiers in Cell and Developmental Biology and for a book chapter.
51. **Mistri M.** Published first largest series of Indian molecular data for Tay-Sach's, Gaucher, Batten disease (Type-I & II) and Mucopolysaccharidosis-II & III; Identified founder disease causing mutations in Tay-Sach's, Batten disease and Morquio-A disease in Gujarat; Presented, poster on 'Effectiveness of NGS based multi-gene approach for diagnosis of neuromuscular and movement disorders', at Indo-US Symposium on Genetic Neuromuscular Disorders & 6th Annual Conference of the Society for Indian Academy of Medical Genetics (India, Nov. 2019); Presented, poster on 'Identification of missense mutation c.230C>G (P77R) as the founder mutation for Morquio-A disease in Indian Gujarati patients', at 9th European Symposium on Lysosomal Storage Disorders (Spain, Dec. 2019).
 52. **Muttamba W.** Summit helped initiate work on identifying genes implicated in treatment resistant asthma; Invited Reviewer, for Plos ONE, BMC Public Health, Hindawi, African Health Sciences and Dove Medical Press journals; Completed course in 'Mixed methods modular research', supported by Pulmonary Complications of AIDS Research Training (PART) Program, Makerere University (Oct. 2019).
 53. **Naeem MA.** Modified medical genetics curriculum for graduate school (MS/MPhil, PhD, 2019-2020); Member, Organizing Committee for Scientific Symposium on Brain, Neurogenetics & Regenerative Medicine (Pakistan, Oct. 2019); Participated in International Conference in Molecular Sciences, University of the Punjab, Lahore (Pakistan, Nov. 2019); Invited by Dr. **Tibrewal S.** (Summiteer) to participate in webinar on 'Genetics in ophthalmology' (India, Jul. 2020); Supervised, graduate level students (2 PhD, 3 MS/MPhil); Trained researchers (MS/MPhil) in collaboration with Dr. **Iqbal M** (Summiteer), Chairman, Department of Biochemistry & Biotechnology, the Islamia University of Bahawalpur, Pakistan.
 54. **Nakousi-Capurro N.** Clinical Geneticist at Hospital Carlos Van Buren of Valparaíso, Chile (Apr. 2020) after completion of final year Medical Genetics resident training from University of Chile, Santiago, Chile; Lectured on 'Dysmorphology' to pediatric neurology residents, Universidad de Valparaíso; Shared knowledge from Summit with colleagues and students; Summit lectures and clinics improved clinical evaluations, performance standards, and success in research; Presented on clinical genetics at multiple meetings in hospitals; Developed patient referral guidelines for hospital; Joined Chilean initiative to improve access to Clinical Genetics through telemedicine; Completed several short courses- Exome Analysis (Universidad del Desarrollo, Sept. 2019), Medical Bioinformatics (Universidad de Chile, Oct. 2019), NF1, NF2 and Schwannomatosis (ACMG, Jul. 2020); Visiting researcher on Genotype-Phenotype correlations in COL1A1/2-related Osteogenesis Imperfecta, at Skeletal Dysplasia Group, Campinas University, Sao Paulo, Brazil (Feb. 2020); Poster abstract accepted for presentation on 'Public Health: Blood folate concentration detected by microbiological assay in women of childbearing age in the metropolitan region', at the XXXIV Chilean Conference (Chile, June 2020); Contact established during Summit with Dr. N. Sobreira (JHU), helped initiate dialogue for collaboration on anosmia at a conference in Chile (2019).
 55. **Oluwarotimi CO.** External examiner, resource person for faculty of Family Dentistry and Dental Surgery, National Postgraduate Medical College of Nigeria; Member, Animal Care and Use Research and Ethics Committee, College of Medicine, University of Lagos (2019-2020); Assistant Secretary General and Medical/Dental Consultant, Lagos University Teaching Hospital Chapter, Nigerian Dental Association (2019); Board Member, Dental Therapist Registration Board of Nigeria; Former President, current Chairman, Constitution Amendment Committee and

- National Liaison Officer, Nigerian Dental Association (2019-2020); Keynote speaker, on 'Management of Noma diseases: the role of dental therapist and dental surgery technician', at Annual Workshop organized by Dental Therapist Registration Board of Nigeria (Aug. 2020); Coordinator, Brush Twice Daily Program organized by Unilever Global, Nigeria.
56. **Ottaru S.** Enrolled in Medical Genetics and Genomics Certificate Program (NHGRI, 2019); Established pediatric sickle cell clinic at Hindu Mandal Hospital –Mwanza and enrolled new cases (30); Provided genetic counseling to patients and parents, access to support groups, and medications and management of complications; Registered new cases (32) with Congenital Birth Defect Registry (estab. 2019 with Summiteer Dr. **Mgasa A**); Surgically managed cases (21), others (11) were referred to Muhimbili National Hospital, for additional management.
 57. **Savina O.** Presented, on 'Aniridia and WAGR syndrome management; Modern approach from the point of view of ophthalmogenetics', at International Applied Science Conference-Refractive Planer (Ukraine, Oct. 2019); Presented, on 'Stargardt's disease; Modern ophthalmogenetic approaches to diagnosis and treatment', at International Applied Science Conference-You need to see your childhood (Ukraine, Jun. 2020); Participated in the TV show 'Doctor Knows', on 'Genetics- a guarantee of eye health. Fantastic reality' (Ukraine, Jul. 2020); Diagnosed, identified mutations in patients (6) with genetic eye diseases; Referred patients for genetic testing and counseling; Recommended, pediatric neurologists for the 2020 Summit.
 58. **Taiwo O.** Appointed, President, International Association for Dental Research, Nigeria Division (Nov. 2019); Chairman, Intercountry Center for Oral Health (ICOH) for Africa, Health Research Ethics Committee (Dec. 2019) and Acting Director/CEO, Intercountry Center for Oral Health (ICOH) for Africa (Dec 2019 – Jan 2020); Lectured on 'Breaking the dentistry medicine barrier, overcoming the historical separation between oral and general health', at Annual General Meeting of the Nigeria Medical Association, Plateau State Branch (Aug. 2019); Invited speaker, presented on 'Introduction to the International Association for Dental Research', at the 16th Annual Scientific Conference of the International Association for Dental Research, Nigeria Division (Nov. 2019); Enrolled in Medical Genetics and Genomics Certificate Program (NHGRI, 2019).
 59. **Utumatwishima JN.** Initiated 'Face2gene Club', with members (8), including pediatricians (2) and a gynecologist; Knowledge on Face2gene from Summit helped screen previously undetected congenital malformations in Rwandan children; Transferred children (45) with suspected genetic malformations to Rwanda Military Hospital; Established Genetic Counseling Department at Neonatology Unit, Rwamagana Hospital (Summit emphasized its need and importance).
 60. **Wangi KYW.** Completed research on 'The use of gene editing technology (CRISPR-Cas9) in maternity care: Ethical perspective' and applied for PhD Program with Australian Awards Scholarship (AAS, 2020); Applied with research proposal on 'Preserving human dignity in thalassemia community' for the International Bioethics Scholar, Center for Bioethics and Human Dignity, Trinity International University (2020-2021); Developed content for 'Health law and bioethics' course in 'Bioethics perspective in genetics/genomics technology', for a law student (Dec. 2020); Guest lecturer, presented on 'Newborn genetics screening and ethical issues in reproductive genetics', Bachelor Midwifery Program; Member, Ethics and Public Policy Committee, International Society of Nurses in Genetics; Participated in Policy Working Group, Young Investigator Initiative, Global Genomics Medicine Collaborative.

2017 Batch

61. **Abad PJ.** Resource person for Basic Genetics at the National Newborn Screening Convention (Nov. 2019); Co-organized, moderated, 3rd Pre-conference in Genetic Counseling, 13th Asia

- Pacific Conference in Human Genetics (Nov. 2019); Elected Secretary, Professional Society of Genetic Counselors in Asia (Nov. 2019); Resource person for workshop on ‘Cultural beliefs of disease causation in hereditary disorders’, Newborn Screening Reference Center (Feb. 2020); Co-organized, moderated two webinars on tele-genetics cancer counseling, ‘Focusing on value of genetics in clinical oncology practice’ (Philippines, Jun. 2020), and ‘Hereditary Breast and Ovarian Cancer’ (Philippines, Jul. 2020).
62. **Avogbe PH.** Delivered new courses in Biobanking and Molecular biology; Lectured on ‘Genetic basis of disease’, ‘Next-generation sequencing methods’, ‘Cancer genetics and genomics’ to graduate level students (MSc), University of Abomey-Calavi, Benin; Included materials from Summit for genomics research curricula; Awarded, Developing Country Travel Award for American Society of Human Genetics Annual Meeting (USA, Oct. 2019); Selected reviewer, for abstracts submitted to Annual Conference of the University of Abomey-Calavi, Benin (Dec. 2019).
 63. **Benítez-Cordero Y.** Lectured on ‘Genetic Counseling’, at National Center of Medical Genetics, Venezuela; Summit helped update topics in genetics, provided tools in counseling; Speaker, presented on ‘Teenage pregnancy and congenital defects in America region’, at 51st Annual Meeting of ECLAMC and 4th Annual Meeting of RELAMC (Nov. 2019); Mentored residents (5) in clinical genetics; Member of International Clearinghouse for Birth Defects Prevention & Research; Evaluated genetic disorders (38) of either monogenic, chromosomal or multifactorial etiology; Codified cases (2035) for Registry; Referred cases (8) for genetics analysis and medical management- Congenital Myopathy (1), infertile couples (7; 2 trombopathy, 1 Turner syndrome, 4 DNA fragmentation in spermatozoa).
 64. **Casado PL.** Reviewer, Brazilian Dental Journal (2019-2020); Supervised graduate students (3; MScD, PhD); Presented online on ‘Peri-implant Disease: Main aspects to be considered under COVID-19 pandemic’, Fluminense Federal University (Brazil, May 2020).
 65. **Dhoro M.** Lectured on ‘Genetic basis of disease and gene therapy’, ‘Diagnosis of genetic disorders’, ‘Genetic counseling’, ‘Genomic medicine and pharmacogenetics’, to medical doctors, postgraduates in Clinical Pharmacology; Mentored graduate level students (MPhil, DPhil, Clinical Pharmacology) on research projects in pharmacogenetics/genomics, and on implementation of pharmacogenetics among clinical practitioners in Harare, Zimbabwe; Enrolled in Medical Genetics and Genomics Certificate Program (NHGRI, 2019); Reviewed research article for Journal of Pharmacogenomics; Reviewed, edited abstracts for University of Zimbabwe Annual Medical Research Day.
 66. **Fatima SS.** Conducted research conference for undergraduate medical, nursing, and biomedical students, emphasized role of good protocol writing, and need for including genetics and genomics in research and health care, especially in low resource settings; Presented paper on ‘Association of chemerin in blood and breast milk with gene methylation in gestational diabetes’, at 11th International DOHaD Congress (Australia, Oct. 2019).
 67. **Hitayezu J.** Started as Fellow, Pediatric Emergency & Critical Care, 2 year sub-specialty program at University of Nairobi, Kenya (Jun. 2019- May 2021); Completed, ‘Fundamentals of implementation science in global health’, University of Washington, Department of Global Health, E-Learning Program (Dec. 2019); Enrolled in Medical Genetics and Genomics Certificate Program (NHGRI, 2019); Attended, Med-Lysosomal Conference (virtual, Jun. 2020).
 68. **James O.** Lectured on ‘Genetics of Craniofacial and Dental Anomalies’ to resident doctors, Faculty of Dental Surgery, National Postgraduate Medical College of Nigeria (Feb. 2020).

69. **Lotz-Esquivel S.** Completed Clinical Clerkship joined Medical Genetics and Metabolism Department, National Children's Hospital; Participated in, 'Society for the study of inborn errors of metabolism inherited metabolic disease adult course' (France, Oct. 2019); Enrolled in Medical Genetics and Genomics Certificate Program (NHGRI, 2019); Sensitized, educated medical professionals on Rare Disease Day through presentations, activities, patient participation (Feb. 2020); Provided care for pediatric patients with inborn errors of metabolism (4 patients/week); Aided in transition of adolescents to adults' at Hospital San Juan de Dios; Consolidated Rare and Orphan Disease Multidisciplinary Clinic (8 patients/week); Expanded prospective database (~220 patients); Requested specialized treatment, laboratory tests for patients, not previously available in National Health System (San José, Costa Rica); Knowledge acquired through Summit provided opportunity to work at children and adult National Referral Centers; Improved quality of life for patients and families by providing optimized genetic services.
70. **Mahfoudh W.** Promoted to Assistant Professor (Jun. 2020); Used materials and lectures from Summit for graduate students in Neuroscience at Faculty of Medicine of Monastir; Supervised graduate student (MS), in breast cancer genetics; Participated in graduate course on 'Innovative educational technologies' (Tunisia, Jul. 2020); Reviewed manuscripts for Journal of Translational Medicine and BMC Women's Health.
71. **Mehrez MI.** Attended, Continuing Education Fellowship Program in 'Craniofacial genetics', College of Dentistry, University of Kentucky (Mar. 2020) and presented on 'Genetic counseling in Egypt' and 'Risk assessment calculation'; Accepted as speaker on 'Genodermatoses with oriental features, a retrospective study', for International Conference on Dentistry and Oral Health (France, Sept. 2020); Reviewer for Special Care in Dentistry journal (Apr. 2020); Referred cases (2) for management of dental condition; Resulted in improved quality of life and psychological well-being of patient.
72. **Mgasa A.** Enrolled in Medical Genetics and Genomics Certificate Program (NHGRI, 2019); Presented on 'Donor registry' and 'Transfusion for transplantation & gene therapy' during gene editing workshop.
73. **Nandal R.** Speaker, on 'Xanthomas as a diagnostic phenotypes', at 1st International Conference of Indian Science Congress Association (India, Mar. 2020); Routinely performed pediatric echocardiography (~5-10 /day); Identified (20) cardiac genetic disorders (William syndrome, Noonan syndrome etc.); Routinely involved in hypertension and hyperlipidemia clinics, performed detailed pedigree analysis; Referred patients with family history for genetic testing and counseling; Identified families (2) with LDLR mutations, managed them with statins.
74. **Otaify GA.** Promoted to Associate Professor of Clinical Genetics (2019); Helped organize 2nd Arab Association of Genetic Research Conference (Egypt, Nov. 2019); Used Summit materials to introduce new clinical genetics courses for the pediatric curriculum in Faculty of Medicine, New Giza University; Enrolled in Medical Genetics and Genomics Certificate Program (NHGRI, 2019); Involved in outpatient consultation, management and counseling for genetic disorders including hereditary bone disorders (10-15 new cases/week), followed-up on treatment and/or investigation (20); Summit helped develop better approaches for diagnosis and management of patients with metabolic disorders.
75. **Owusu M.** Lead, for diagnostic testing for COVID-19 at Kumasi Centre for Collaborative Research in Tropical Medicine; Presented webinars on 'COVID-19 in Africa' to the Ghana Biomedical Convention.
76. **Petlichovski A.** Enrolled in NHGRI Medical Genetics and Genomics Certificate Program (NHGRI, 2019); Included Summit lectures and data in medical school curricula and for

specialization trainees; Genetic Institutions in Macedonia adopted new curriculum in clinical laboratory genetics; Program recognized by European Society for Human Genetics, covers inheritance, diagnosis and treatment of genetic conditions; Implemented new genetic tests for Noonan syndrome, autoinflammatory syndrome, immune deficiencies etc.; Performed genetic testing on couples (200) diagnosed with recurrent spontaneous abortion; Referred these cases for genetic counseling.

77. **Roy S.** Qualified as Fellow of College of Physician and Surgeon, Bangladesh College of Physicians and Surgeons (Jan. 2020); Arranged CME program on ‘Common ocular genetic diseases and recent management protocols’; Speaker, on ‘Establishing RB department in a tertiary eye care center of Bangladesh – a new hope for retinoblastoma patients’, at Annual CME, Bangladesh Eye Hospital (Dec. 2019); Speaker, on ‘Childhood ocular cancer and common blinding diseases under 6 years’, at International Rotary Club, Chittagong Division of Bangladesh; Mentored undergraduate optometry research students, Institute of Community Ophthalmology, Chittagong University; Identified genetic disorder cases (60), treated patients (43), referred others for medical management; Patients (37) were satisfied with outcomes.
78. **Sanhueza Díaz C.** Professor, Clinical Genetics specialty program, University of Chile; Head, Genetics unit, Roberto del Río Hospital and Cytogenetics Laboratory of the Clinical Hospital of the University of Chile; Taught postgraduate curriculum and clinical evaluation of patients at both institutions.
79. **Thakur N.** First Consultant Clinical Geneticist under Ministry of Health and Population, Nepal (2020); Awarded for crisis management (COVID-19) by Minister of Health and Vice Chancellor, National Academy of Medical Sciences on 131st Annual day of Bir Hospital, Kathmandu, Nepal (Jul. 2020); Used training materials from Summit to conduct Symposium on Reproductive Genetics at National Maternity Hospital, Kathmandu (Dec 2019); Initiated whole exome sequencing (2020); Established, supervised 19 labs for COVID-19 testing; Coordinator, COVID-19 PCR lab at Bir Hospital; Advisor, COVID-19 pathology to Ministry of Health and Population, Nepal; Scientific Committee Member, 9th International Conference on Birth Defects and Disabilities in the Developing World (Sri Lanka, 2020); Used materials and guidelines from Summit to set up, optimize, validate first clinical based next generation sequencing lab (15-20 patients/ per week) and to design protocols for patient selections; Validated for the first time in Nepal, Non-invasive prenatal testing and Pre-implantation genetic screening.
80. **Tolegen N.** Speaker, on ‘The role of the standard cytogenetic study of bone marrow cells in the diagnosis of acute lymphoblastic leukemia in children’, at the International Conference on Biomedicine (Kazakhstan, May 2020); Karyotyped pregnant women (95) with suspected fetus abnormalities; Identified 15 chromosomal abnormalities -Down syndrome (6), Edwards syndrome (2), Turner syndrome (1), structural chromosomal abnormalities (6); Examined children (152, <18yrs) with diagnosis of acute lymphoblastic leukemia, linearity determined by cytological examination of bone marrow; Majority patients (80%) had normal diploid set of chromosomes, 60% had hyper-diploid set (≥ 47) and a more favorable prognosis; 20% had aberrations- t(6;16)(q13;p13.1), t(8;21)(q21.3;q22), t(4;12)(p14;q13), t(1;11)(p36.1;q23), del(16)(p11), i(17)(q10), t(9;10)(q34;q22), der(9)(q32) and presence of marker chromosomes, 16% had hypodiploid set of chromosomes (≤ 44) and a poor prognosis due to 30% survival rate.
81. **Torres-Mejía G.** Summit played a significant role in meeting researchers in breast cancer and establishing collaborations inside and outside of Mexico.
82. **Tuncay Yaylacioglu F.** Graduated from MD-PhD program of Hacettepe Medical School (2019); Promoted to Assistant Professor in Medical Biology Department, University of Health Sciences,

Gülhane Medical Faculty, Ankara, Turkey (Jul. 2020); Awarded, ICO-NEI Fellowship in Ophthalmic Genetics (starts Jul. 2021); Enrolled in Medical Genetics and Genomics Certificate Program (NHGRI, 2019); Awarded Travel Grant to attend 6th Course in Eye Genetics (Italy, Mar. 2019); Initiated a registry for inherited ophthalmic eye diseases.

83. **Vishnuposka SA.** Lectured two graduate level courses ‘Human genetics’ and ‘School of clinical genomics’; Submitted abstract on ‘MON-717 novel GLI2 mutations identified in pediatric patients with combined pituitary hormone deficiency: One gene, various genotypes’ to ENDO ONLINE (Apr. 2020).
84. **Yousef YA.** Identified, diagnosed and treated patients with retinoblastoma (31); Referred genetically tested (22) with familial and non-familial retinoblastoma for counseling and medical management; Informed patients negative for germline disease about extreme low risk of transmission to offsprings, hence improved quality of life by alleviating stress.

(2016 Batch)

85. **Adeoye AM.** Joint appointment with new Institute of Cardiovascular Diseases, College of Medicine, University of Ibadan as Head Molecular Cardiology and Preventive Cardiology unit; Currently Deputy Chairman, Medical Advisory Committee (Education and Training) and Chairman, Medical Advisory Committee (Elect), University College Hospital, Ibadan, Nigeria, and the First Teaching Hospital.
86. **Adeyemo AA.** Speaker, presented on ‘Randomized controlled clinical trial of intra-tympanic dexamethasone in prevention of cisplatin ototoxicity’, at the 60th International Conference of the West African College of Surgeons (Nigeria, Feb. 2020).
87. **Adeyemo WL.** Coordinated, Bioinformatics workshop for junior faculties, College of Medicine University of Lagos (Dec. 2019), delivered lectures on ‘Introduction to genome databases’ and ‘SNPedia’; Co-ordinated, Responsible Conduct of Research Workshop for junior faculty, College of Medicine University of Lagos (Feb. 2020), delivered lectures on ‘Introduction to mentoring’, ‘Mentoring competencies’ and ‘Responsibilities of mentors and mentees’ ;Presented lecture on ‘Etiology of facial clefts: Role of genetics’ to global audience in the Tele-Cleft Online Series of Lectures organized by Smile Train (Jul. 2020); Mentored awardees (9), awarded Seed Grant for execution of genomic research at College of Medicine University of Lagos.
88. **Ariani Y.** Obtained PhD (Medical Genetics, 2019); Appointed, Genetics Consultant, Morula Bunda International Clinic (IVF), Jakarta (2019); Elected, Head of Stunting and Prematurity Research Group, Indonesian Medical Education and Research Institute (2019); Presented poster on ‘Mutation identification of unreported and reported mutation profile in exon 7 of N-acetylgalactosamine-6-sulfatases (GALNS) gene of mucopolysaccharidosis type IVA (MPS IVA) patients in Indonesia’, at 6th International Conference on Mathematics, Science and Education (Indonesia, Oct. 2019); Presented poster on ‘Variants of iduronate 2-sulphatase (IDS) gene from Indonesian mucopolysaccharidosis type II patients : a preliminary study’, at Asia Pacific Conference on Human Genetics (Philippines, Nov. 2019).
89. **Belhassan K.** Graduated from Laboratory Genetics and Genomics Fellowship Program, Washington University, Saint Louis; Planning for American Board Certification (Medical Genetics); Presented posters on ‘A novel de novo RAC1 frameshift likely pathogenic variant in a patient with developmental delay and epilepsy suggests haplo-insufficiency as a new mechanism of disease in RAC1-related disorders’ and on ‘Mitochondrial-processing peptidase subunit beta (PMPCB) rare compound heterozygous variants in a patient with developmental delay, neurodegeneration, cerebellar vermis agenesis and cerebellar atrophy’, at ASHG annual

- meeting (USA, Oct. 2019); Presented poster on ‘Lysine acetyltransferase 6B (KAT6B) gene copy number loss: a likely pathogenic variant in 2 patients with failure to grow and speech delay’, at ACMG annual meeting (virtual, Mar. 2020).
90. **Deniz E.** Received Best Young Researcher Award, Medical Biology and Genetics Congress (Turkey, Oct. 2019); Introduced new course ‘Research techniques and ethics in natural sciences’, for graduate students in molecular and translational biomedicine (2019); Supervised 2 graduate students (MS), on genomics projects (2020).
 91. **Ekure EN.** Speaker on ‘Role of genetics in childhood cardiovascular disease’, at Nigerian Society for Pediatric Cardiology Webinar (Jun. 2020); Identified, diagnosed, treated pediatric congenital heart defects cases (244), at Lagos University Teaching Hospital; All cases were counselled and necessary ones (200) were medically managed.
 92. **Guio H.** Enrolled in Medical Genetics and Genomics Certificate Program (NHGRI, 2019); Ambassador, Conference on Precision Medicine Program in Peru (2020); Attended, ‘Genetics and Genomics in Peru’, Instituto Nacional de Medicina Genomica (Mexico, Oct. 2019); Founder, Latino American Initiative to develop genetic and genomics (Ecuador, Jan. 2020).
 93. **Hussen DF.** Presented poster on ‘Combined markers for preclinical identification of Alzheimer's disease’, at 2nd Arab Association of Genetic Research Conference (Egypt, Nov. 2019).
 94. **Lallar M.** Speaker, on ‘Genetic testing in preimplantation genetic diagnosis’, at 2nd ISAR Summit (India, Feb. 2020); Panelist on ‘Provision of Enzyme Replacement Therapy to LSD patients by Government’ and speaker on, ‘Genetics of lysosomal storage disease’, for International Rare Disease Day, organized by Department of Health and Family Welfare, Punjab (India, Feb. 2020); Presented webinar, on ‘Genetics of recurrent pregnancy loss’, Medgenome India (Apr. 2020); Provided with genetic counseling to patients (>250) and prenatal diagnosis to individuals (30) with various genetic disorders; Maintained birth defect register for the institute.
 95. **Malasa L.** Awarded Global Health Fellowship, Novartis Institutes for BioMedical Research (USA, Jun. 2019); Presented poster on ‘Basic molecular techniques for SCD diagnosis and genomics research’ (USA, 2019); Enrolled in Medical Genetics and Genomics Certificate Program (NHGRI, 2019); Completed online training on ‘Disease screening in public health’, University of Geneva & University of Lausanne (Apr. 2020); Co-founded organization, Tanzania for Better Health (Jan. 2020).
 96. **Okafor FU.** Defended, PhD dissertation on ‘Nursing training programmes’ lecturers’ and students’ reproductive health genetics and genomics knowledge, beliefs and practice in Edo State, Nigeria’; Pending, promotion to Professor of Nursing Science; Participated in TeleECHO on ‘Prevention of community transmission of COVID 19’.
 97. **Roblejo Balbuena H.** Lectured on ‘Genetic counseling’ to graduate students (MS), at National Center of Medical Genetics, Venezuela; Mentored residents (3) in clinical genetics; Speaker, on ‘CYP2D6*3, *4, *5 and *6 allelic variants in a sample of Cuban patients with schizophrenia’, at XVII Latin American Congress of Genetics (Argentina, Oct. 2019); Presented on ‘CYP2D6 genotype, genetic ancestry and antipsychotic-induced extrapyramidal side effects in Cuban patients with schizophrenia’, at 5th ESPT Congress Precision Medicine and Personalized Health (Spain, Oct. 2019); Evaluated genetic cases (88) of monogenic, chromosomal or multifactorial etiology; Registered new cases (33) in the Genetic Diseases' Register, Pediatric Hospital Center, Havana.
 98. **Seven M.** New Faculty position as Assistant Professor at University of Massachusetts, Amherst, MA, USA (2019); Organizing Committee Member, Nursing, Genomics and Healthcare for 2021 meeting, Welcome Genome Campus, UK.

99. **Sirisena ND.** Awarded PhD, from University of Colombo, Sri Lanka (Feb. 2020); Received, Senate Award for Research Excellence at Inauguration Ceremony, University Annual Research Symposium (Sri Lanka, Nov. 2019); Awarded, Best Oral Presentation for ‘High frequency of Klinefelter syndrome in a cohort of Sri Lankan males with azoospermia and oligozoospermia’, at 14th Annual Academic Sessions of the Sri Lanka Association of Urological Surgeons (Sri Lanka, Nov. 2019); Elected, Founder President, Human Genetics Society (Jan. 2020); Appointed Steering Committee member of the Global Genomic Medicine Collaborative (G2MC) initiative (Feb. 2020); Co-ordinated Pre-Congress Workshop on ‘Implementing genomic medicine in the clinical setting’, and presented on ‘Genetics and genomics of breast cancer in Sri Lanka’, at the Symposium on Genetics, 150th Anniversary Colombo Medical Congress (Sri Lanka, Feb. 2020); Appointed, Founder Editor-in-Chief, GeneNews, official e-Newsletter, Human Genetics Society (Mar. 2020); Appointed, Co-Chair Young Investigator Sub-Committee, Global Genomic Medicine Collaborative (G2MC) initiative (Jul. 2020); Provided genetic counseling, diagnostic services to patients (>300) with genetic diseases and congenital birth defects, at the Human Genetics Unit; Referred patients for appropriate supportive therapies for improvement of quality of life.
100. **Tibrewal S.** Awarded, ARVO Developing Country Eye Researcher Travel Fellowship (2020); Organized two-day training program for optometrists on ‘Pedigree Charting’ with help of Genetics Counselor (India, Sept. 2019); Organized meeting for geneticists and ophthalmologists at Dr. Shroff’s Charity Eye Hospital, Delhi, under aegis of Global Eye Genetics Consortium (India, Feb. 2020), focused on collaborations and grant writing (knowledge obtained at Summit); Organized webinar on ‘Genetics for Ophthalmologists’ (Jul. 2020); Involved in archiving of samples, function and maintenance of Biobank; Continued to guide genetic counsellor, and the function and growth of genetic clinic; Examined children (5648), provided appropriate care, including low vision rehabilitation; Attended pediatric genetic cases (181); Some patients (10) received genetic tests, pre and post-test counselling; Identified patients (31) with systemic abnormalities, referred to specialists.
101. **Uwineza A.** Senior Lecturer, Medical Genetics and Medical Genomics, School of Medicine and Pharmacy (General Medicine), and Graduate School (MMED Pathology/Pediatrics and OB/Gyn); Introduced ‘Testing for inborn errors of metabolism’ in General Medicine (Level 2); Participated in 4th Sickle Cell Disease Ontology Workshop (S. Africa, Nov. 2019); Enrolled in Medical Genetics and Genomics Certificate Program (NHGRI, 2019); Provided outpatient consultation for patients (825) with with genetic disorders, at CHUK.