

ADDENDUM

ISHGG FOURTH ANNUAL OUTCOME (October 2020- September 2021)

(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). The Covid-19 pandemic affected the work of some of our Summiteers.

A. Publications: (Total - 295; 2019- 86; 2018- 99; 2017- 66; 2016- 44)

2019 Batch

1. Abdel-Salam GMH, Abdel-Hamid MS, **Sayed ISM**, Zechner U, Jörn Bolz H. OTUD6B-associated intellectual disability: Novel variants and genetic exclusion of retina degeneration as part of a refined phenotype. *J Hum Genet.* 2021 Aug 5. doi: 10.1038/s10038-021-00966-2
2. Abdel-Salam GMH, Duan R, Abdel-Hamid MS, **Sayed ISM**, Jhangiani SN, Khan Z, Du H, Gibbs RA, Posey JE, et al. A SMG8 missense biallelic variant causing intellectual disability maps to an interacting domain and patients show a phenotype overlapping with SMG9-associated disease trait. *Am J Med Genet A.* 2021. (Submitted).
3. Adejoh J, Inyang BA, Egua MO, Nwachukwu KC, **Alli LA**, Okoh MP. In-vivo anti-plasmodial activity of phosphate buffer extract of *Calotropis procera* latex in mice infected with *Plasmodium berghei*. *J. Ethnopharmacol.* 2021. 277(6):154-159.
4. 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. *International Journal of Hematology and Oncology.* 2021. (Submitted).
5. Ahmed HA, El-Kamah GY, Rabie E, Mostafa MI, Abouzaid MR, Hassib NF, Abdel-Kader MA, **Mehrez MI**, **Sayed ISM**, et al. Gene mutations of the three ectodysplasin pathway key players (EDA, EDAR, and EDARADD) account for more than 60% of Egyptian ectodermal dysplasia: A report of seven novel mutations. *Genes.* 2021 Sep;12(9):1389.
6. **Akinmola OO**. The rare disease of Kallmann Syndrome in a 28yr old Male: Need for scrutiny of diagnostic tools. 2021. (In preparation).
7. **Alamin TA**, Komi OD, Ibrahim AI, Mohammed AN, Massad ER. Genetic and genomic medicine in Sudan, haematology prospectus review. 2021. (In preparation).
8. Ali AZ, Shalaei AA, Yousifl AK, Towfeek BA, Ahmed TA, Mohamed RA, **Alamin TA**. Polymorphisms of Glutathione S Transferase Mu 1 gene and susceptibility to sickle cell disease. *International Journal of Hematological Disorders.* 2021. (Submitted).
9. **Alli LA**, Nnodu OE. Anti-sickling Efficacy of medicinal plant extracts from Gwagwalada, Abuja, Nigeria. *West Afric J Med.* 2021; 37(7):790-795.
10. **Amarakoon GGGT**, Mendis D, Senadeera N, Thewarapperuma C. A case report of a child with congenital Amegakaryocytic thrombocytopenia. 2021. (In preparation).
11. **Amarakoon GGGT**, Wijesundara MR, Ranathunga PU, Wijesinghe AHNP, Rathnasiri KMSS, Wijayarathne HK. Patterns of congenital malformations and risk factors in newborn babies in Teaching Hospital - Anuradhapura. 2021. (In preparation).
12. Baig HMA, Ansar M, Iqbal A, Naeem MA, Quinodoz M, Calzetti G, **Iqbal M**, Carlo Rivolta C. Genetic analysis of consanguineous Pakistani families with congenital stationary night blindness. *Ophthalmic Res.* 2021. (Submitted).

13. Bica C, Ciocan CA, Braicu C, Harangus A, Nutu A, Pop L, **Pîrlog R**, Berindan-Neagoe I, Al Hajjar N. Massively parallel tumor multigene sequencing to assess the mutational landscape of lung cancer patients. *Cancers*. 2021 (In review).
14. Budisan L, Zanoaga O, Braicu C, **Pîrlog R**, Covaliu B. Links between infections, lung cancer, and the immune system. *Int J. Mol. Sci.* 2021 (In review).
15. Chandrakumara J, Fernando M, Rathnayake C, Muthukumarana O, **Amarakoon G**. Wilson disease presenting as acute haemolytic anaemia in a Sri Lankan child. *Ceylon Med J.* 2021. (Submitted).
16. Chori BS, Danladi B, Inyang BA, Okoh MP, Nwegbu MM, **Alli LA**, Odili AN. Hyperhomocysteinemia and its relations to conventional risk factors for cardiovascular diseases in adult Nigerians: the REMAH study. *BMC Cardiovasc Disord.* 2021; 21(102):1-8.
17. **Chowdhury EH**, Chowdhury MA, Samira HH. Study in relation between consanguinity of marriage and Beta Thalassemia Major at a tertiary children hospital in Bangladesh. *Bhutan Health Journal.* 2021. (Submitted).
18. Dimog EF, **Tumulak MJR**, Laurino MY, Daack-Hirsch S, Silao CLT, Conaco MCG, PADilla CD, Estacio LR. Beliefs on the causes of birth defects as perceived by mothers of children with birth defects in a tertiary care hospital in the Philippines. *J Community Genet.* 2021. (Submitted).
19. Drula R, **Pîrlog 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 Mol Basis Dis.* 2021; 1867(5):166045.
20. Duri K, Chimhuya S, Gomo E, Munjoma PT, Chandiwana P, Yindom LM, **Mhandire K**, Ziruma A, Mtapuri-Zinyowera S, et al. 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). *Virology J.* 2021 Jan;18(1):30.
21. Gamal El Din HM, El Kilani NS, Mostafa MI, **Sayed I**, Ismail S, Shoreibah E. Clinical and radiographic evaluation of ten congenital insensitivity to pain with anhidrosis patients. *Al-Azhar Dental Journal for Girls.* 2020 Oct; 7(4):577-783.
22. Ghosh S, Tilak TVSVGK, Venkatesan S, **Mutreja D**. The domino effect-treatment of superior vena cava obstruction. Triggering Tumor Lysis Syndrome: A Case Report. *Sci Med Jrl.* 2021; 3(1):44-50.
23. Gupta N, **Yadav S**. An approach to antenatally detected congenitally malformations. Chapter in, 'Protocols in perinatology', National Neonatology Forum, India. 2021. (In Review).
24. **Güven Y**, Saraçoğlu HP, Aksakal SD, Kalaycı T, Altunoğlu U, Uyguner ZO, Eraslan S, Börklü-Yücel E, Kayserili Karabey H. Clinical and Genetic Analysis of Three Turkish Families with Nance Horan Syndrome. 2021. (In preparation).
25. **Gyawali P**. Active Vitamin B12: what does the test mean and offer? *Medicos Next.* April 2021;12. ISSN NO. 2717-4778. <https://medicosnext.com/active-vitamin-b12-test-what-does-the-test-mean/>
26. **Gyawali P**. Utility of various biomarkers in COVID-19 disease management *Medicos next.* November 2020; 7. ISSN NO. 2717-4778. <https://medicosnext.com/utility-of-various-biomarkers-in-management-of-covid-19/>
27. **Gyawali P**. Glucometer: choosing diabetes technology well. *Medicos next.* January 2021; 9. ISSN NO. 2717-4778. <https://medicosnext.com/glucometer-choosing-diabetes-technology-well/>
28. Hanafi NS, Agarwal D, Chippagiri S, Brakema EA, Pinnock H, Sheikh A, Liew SM, Ng CW, Isaac R, Chinna K, Wong LP, **Hussein N**, et al. Chronic respiratory disease surveys in adults in

- low- and middle-income countries: A systematic scoping review of methodological approaches and outcomes. *J Glob Health*. 2021; 19(11): 04026.
29. Harris KH, Nakayama T, Lai J, Zhao B, Argyrou N, Gubbels CS, Soucy A, Genetti CA,... **Iqbal M**, et al. Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. *Genet Med*. 2021. 23; 1028–1040.
 30. Hawken S, Murphy MSQ, Ducharme R, Bota AB, Wilson LA, Cheng W, **Tumulak MJR**, Alcausin MML, Reyes ME, et al. External validation of ELASTIC NET regression models including newborn metabolomic markers for postnatal gestational age estimation in East and South-East Asian infants. *Gates Open Res*. 2021; 4:164.
 31. **Hussein N**, Henneman L, Kai J, Qureshi N. Preconception risk assessment for thalassaemia, sickle cell disease, cystic fibrosis and Tay-Sachs disease. A Cochrane systematic review. 2021. (Submitted).
 32. **Hussein N**, Lee YK, Qureshi N, John P, Azmi A, Ng CJ. Thalassaemia screening: Exploring the views and experiences of thalassaemia carriers in East Malaysia. 2021. (Submitted).
 33. **Hussein N**, Ramli R, Mohd Ahad A, Liew SM, Hanafi NS, Abu Bakar AI, Lee PY, Cheong AT, Ghazali SS, et al. Asthma care, asthma control and the associated factors: a cross sectional study from the baseline data of the Klang Asthma Cohort. 2021. (In preparation).
 34. Irmi ZI, Ng CJ, Lee PY, **Hussein N**. Factors influencing pregnancy planning of multi-ethnic Asian women with diabetes: A qualitative study. *PLoS ONE*. 2020; 15 (12):e0242690.
 35. Isokariari OM-A, Tolani MA, Igboke MC, Enebeli U, Babalola RN, **Monye HI**, Francis UW, Kpuduwei SPK, Uyilawa O, et al. Factors associated with participation in research by very early career doctors in a lower middle-income country in Africa: a multi-centre study. *Ibom Med J*. 2021;14(3):262–273.
 36. Juan Pozo-Palacios, **Llamos-Paneque A**, Rivas Iglesias CR, Onofre Perez EJ, Lopez Caceres A, Villarreal J. Experiences in the molecular diagnoses Fragile X syndrome in Ecuador. *Front Psychiatry*. 2021. (Submitted).
 37. **Kars ME**, Basak AN, Onat OE, Bilguvar K, Choi J, Itan Y, Caglar C, Palvadeau R, Casanova JL, et al. The genetic structure of the Turkish population reveals high levels of variation and admixture. *Proc. Natl. Acad. Sci. U. S. A*. 2021. (Accepted).
 38. Kaur J, Patnaik SK, Rahul G, **Mutreja D**, Tilak TVSVGK. Formidable challenges of establishing a diagnostic laboratory for a 1000-bedded makeshift COVID-19 hospital. *M J Armed Forces India*. 2021; 77 (S2):S517-S519.
 39. Kazahura P, **Mushi TL**, Pallangyo P, Janabi M, Kisenge R, Albaghdadi, Majani N, Kija E. Prevalence and risk factors for subclinical rheumatic heart disease among primary school children in Dar es Salaam, Tanzania: A community based cross-sectional study. *BMC Research Square*. 2021 Feb 9. (In Review). doi: 10.21203/rs.3.rs-197941/v1
 40. 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*. 2021; 71(3):816-821.
 41. Köhler S, Gargano M, Matentzoglou N, Carmody LC, Lewis-Smith D, Vasilevsky NA, Danis D, Balagura G,... **Wiafe SA**, et al. The human phenotype ontology in 2021. *Nucleic Acids Res*. 2021 Jan 8; 49(D1):D1207-D1217.
 42. 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. *Asia Pacific J Couns Psychother*. 2021. (Submitted).

43. Le TD, Vu HTT, Arunasiri I, Ito K, Makise T, Thi Le H, Nguyen TH, Nguyen LTH,...**Luong LH**, et al. Oral administration of salmon cartilage proteoglycan attenuates osteoarthritis in a monosodium iodoacetate-induced rat model. *Nat. Prod. Commun.* 2020; 15 (12):1-6.
44. Lee PY, Cheong AT, Ghazali SS, Hani S, Wong J, **Hussein N**, Ramli R, Pinnock H, Liew SM, et al. Barriers to implementing asthma self-management in Malaysian primary care: qualitative study exploring the perspectives of healthcare professionals. *npj Prim Care Respir Med.* 2021; 31 (38).
45. **Llamos-Paneque A**, Meneses S, Lamar-Segura E, Castro- Maribel N, Recalde H, Ñacato-karen L. Spontaneous chromosomal fragility in Fanconi anemia: Case presentation. *Biomedical J.* 2021. (Submitted).
46. **LLamos-Paneque A**, Pozo-Palacios J, Sancan-Byron Garzón-Castro M, Lamar –Segura E, Liehr T. A doble aneuploidy: an Ecuadorian case 45 X(7)/47XX+ 21 (3)/ 46, X, der (X)(: p11.1->q11.1)(1)/46,XX (1). 2021. (In preparation.)
47. **LLamos-Paneque A**, Rivas-Iglesias C, Onofre E. Molecular diagnosis of neurogenetic diseases in Ecuador: Experience in the Hospital of Specialties No.1 of the Armed Forces. 2021. (In preparation).
48. **López-Star B**, Pérez-Pérez JF., Sánchez-Moreno P, Ochoa-Ramírez LA. Evaluation by means of a survey of the degree of genetic knowledge in the clinical practise of ophthalmologists. *Rev. Mex. Oftalmol.* 2021 Jun; 95(3):101-106.
49. **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.* 2021. (Accepted).
50. Mohamed AE, Edris MA, Ahmed MM, Ali SA, **Alamin TA**. In silico analysis of single nucleotide polymorphism in human IL-4 gene. *Meta Gene.* 2021. (Submitted).
51. **Monye HI**, Hassan YF, Olajide LQ, Buowari DY, Enebeli U, Selowo TT, Ogbonna VI, Oomolawal O, Adesoye O, et al. Medical migration among Early Career Doctors (ECDs) in Nigeria - magnitude, causes and recommendations. *Nigerian Medical Practitioner.* 2021. (Accepted).
52. **Monye HI**, Olawoye O, Ugalahi MO, Onebunne EO, Oluleye TS. Genetic eye diseases and services - awareness, knowledge and perceptions of adult patients of the University college Hospital, Ibadan. 2021. (In preparation).
53. **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. 2021. (In preparation).
54. 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 Pharm. Rep.* 2021. (Accepted).
55. Mudawi OM, Ahmed B, Musa SL, Abbas AA, Ibrahim AM, Alamin **TA**. SCD in Sudan review. 2021. (In preparation).
56. Mukhongo HN, Kinyua JK, **Weldemichael YG**, Kasili RW. Screening for antifolate and artemisinin resistance in Plasmodium falciparum clinical isolates from three hospitals of Eritrea. *F1000Res.* 2021; 10:628.
57. Mukhopadhyay I, Aggarwal R, **Mutreja D**, Maheswari S. MRI findings in androgen insensitivity: Case series, Sultan Qaboos Univ Med J. 2021 Apr; 1(1). doi: 10.18295/squmj.4.2021.012

58. **Mutreja D**, Paliwal G, Mangal V, Biswas AK. Cold autoimmune hemolytic anaemia with Myelodysplastic Syndrome: Not just an "Open and Shut case". *Indian J Pathol Microbiol.* 2021. (Accepted).
59. **Mutreja D**, Paul K B, TVSVGK T. Fatal sickling-associated microvascular occlusive crisis in a young with sickle cell trait. *Autops Case Rep [Internet].* 2021; 11:e2021297.
60. **Mutreja D**, Singh S. Apple peel deformity and malrotation of gut: Autopsy findings of a rare cause of mortality in utero. *Turk Pataloji Derg.* 2021. (Accepted).
61. **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.* 2021 Jan-Mar; 64(1):189-191.
62. Nghiem TD, Do GT, **Luong LH**, Dang TH, Nguyen VA, Nguyen TT, Khanh TV, Ta TV, TH Tran. 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.* 2021; 9:e1648.
63. **Okunola O**. Genetics in Medicine. Chapter in Essential Textbook of Medicine, Section 22. In Alebiosu & Ayodele (eds) 1st edition. Osun State University Publishers. 2021. ISBN- 978-978-978-399-9.
64. Olawoye O, **Monye HI**, Onakpoya OH, Ibukun FA, Yeye-Agba OO, Echieh CI, Obuoforibo BR, Basse CI, Abdullahi MI, et al. The impact of COVID-19 pandemic on ophthalmology practice in Nigeria. 2021. (In preparation).
65. **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. 2021. (In preparation).
66. **Paredes-Moscossa SR**. Breaking tolerance to pancreatic cancer unresponsiveness to immunotherapy. 2021. Elsevier (Submitted).
67. **Pirlog R**, Cismaru A, Nutu A, Berindan-Neagoe I. Field cancerization in NSCLC: A new perspective on microRNAs in macrophage polarization. *Int J Mol Sci.* 2021; 22 (2):746.
68. Porras AR, Bramble MS, **Mosema KBA**, Spencer D, Dakande C, Many HB, Vashit N, Likuba EB, Mukau EJ, et al. Facial analysis technology for the detection of Down Syndrome in the Democratic Republic of the Congo. *Eur J Med Genet.* 2021 Jun; 64: 104267.
69. Raashid A, Hameed Y, Ejaz S, Khurshid M, **Iqbal M**. A variety of novel BRCA1 mutations detected in acute myeloid leukemia patients cause inactivation of BRCA1 protein. *Curr. Med. Sci.* 2020. (Accepted).
70. Rasool IG, Zahoor MY, Ahmad I, **Iqbal M**, Shafqat S, Anjum AA, Shehzad W. Description of novel variants in consanguineous Pakistani families affected with intellectual disability. *J. Neurogenet.* 2021. (Submitted).
71. Rasool IG, Zahoor MY, **Iqbal M**, Anjum AA, Ashraf F, Abbas HQ, Baig HMA, Bajwa TM, Shehzad W. Whole exome sequencing revealed novel variants in consanguineous Pakistani families with intellectual disability. *Genes & Genomics.* 2021; 43(5):503-512.
72. Sarimiye TF, **Monye HI**, Abiola VNI, Abo-Briggs JN, Ologundudu O. Awareness of glaucoma, and uptake and satisfaction of a free, targeted glaucoma screening programme in Ibadan, Oyo State. 2021. (In preparation).
73. Singh AR, Srinath R, Shankar S, **Mutreja D**, Ahmad FM, Nanda SK. An interesting case of chorea in a young male. *Neurol India.* 2021. (Accepted).
74. Tan HY, Lee YK, Malik TFA, **Hussein N**. School-based thalassemia screening programme in Malaysia. 2021. (In preparation).

75. **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. 2021. (In preparation).
76. Tran TD, Nguyen DH, **Luong LH**, Phan KN, Kieu QH, Le NT, Nguyen TT. Total femur replacement in a patient with chronic persistence osteomyelitis – A case report. *Int J Surg Case Rep.* June 2021; 84:106067.
77. **Tumulak MJR, Hussein N, López Star BD, Paneque AL**. Integrating genetics and genomics in the “new era”- Perspectives from low-middle income countries in Asia and South America. *J Community Genet.* 2021. (Submitted).
78. **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. 2021. (In preparation).
79. **Tumulak MJR**, Pascua AV, Jover EJ, Guerbo RJ, Canoy GM, Laurino MY. Genetic counseling in the time of COVID-19: The Philippine experience with telegenetics. *J Genet Couns.* 2021. (Submitted).
80. Van Pham AT, Luong MH, Dinh HTT, Mai TP, Trinh QV, **Luong LH**. Immunostimulatory effect of moringa oleifera extracts on cyclophosphamide-induced immunosuppressed mice. *J Herbs Spices Med Plants.* 2021 Jun 8; 1–9.
81. Verma S, Baveja P, Patnaik S, Thakur N, Tilak TVSVGK, **Mutreja D**. Young male with alleged snake bite and disseminated intravascular coagulation or a life threatening but potentially curable malignancy: Clinicopathologic correlation. *Natl Med J India.* 2021. (Accepted).
82. **Yadav S**, Madhumita RC, Gupta N, Chauhan S, Jana M, Kabra M. Molecular alterations at 11p15.5 locus in Isolated Hemihyperplasia. 2021. (In preparation).
83. **Yadav S**, Nanda S, Malhotra V. Role of chromohysteroscopy in patients of abnormal uterine bleeding. *Indian J Gynecol Oncol.* 2021; 19(15):1-6.
84. **Yeşilçınar I, Seven M**, Paşalak Şİ, Güvenç G. Interventions aiming to improve informed decision on prenatal screening and testing: A scoping review of the literature. *J Genet Couns.* 2021 Jun. doi: 10.1002/jgc4.1437
85. **Yesilcinar I, Seven M**, Sahin E, Calzone K. Genetics and genomic competency of Turkish nurses: a descriptive cross-sectional study. *Nurs Edu Today.* 2021. (In Review).
86. **Yesilcinar I, Seven M**, Sahin E, Calzone K. Test/Re-test Reliability of a Turkish version of the genetics and genomics in nursing practice survey (GGNPS). *J Nurs Meas.* 2021. (Accepted).

2018 Batch

87. **Adeyemo TA**, Bolarinwa AB, Olatinwo AT, Oyewole A, Akanmu AS. (2020). Blood transfusion service in a tertiary hospital in sub-Saharan Africa during the COVID 19 pandemic: Experience from Lagos University Teaching Hospital, Nigeria. *African Sanguine.* 2021; 23(1):30-35.
88. **Adeyemo TA**, Ojewunmi OO, Oyetunji, AI, Kalejaiye OO, Menzel S. Fetal-haemoglobin enhancing genotype at BCL11A reduces HbA2 levels in patients with sickle cell anaemia. *eJHaem.* 2021; 1–3.
89. Ahmed KS, Esbhani UA, Naseem Z, Lalani S, **Fatima SS**. Molecular basis of nonalcoholic fatty liver disease and metabolic syndrome in a subset of South Asians. *Int J Diabetes Dev.* 2021. doi: 10.1007/s13410-020-00906-4 I.F. 0.591
90. Akpogheneta O, Dicks S, Grant D, Kanneh Z, Jusu B, **Edem-Hotah J**, Kanneh L, Alhasan F, Gbokie M, et al. Boosting understanding of Lassa fever virus epidemiology: Field testing a novel

- assay to identify past Lassa fever virus infection in blood and oral fluids of survivors and unexposed controls in Sierra Leone. *PLoS Negl Trop Dis*. 2021; 15(3):e0009255.
91. Alam F, Rehman R, **Fatima SS**, Ashraf M, Khan TA. SIRT1 gene single nucleotide polymorphisms and female infertility: A cross sectional study in Pakistan. *Intern J Clin Pract*. e14132. 2021. doi: 10.22541/au.158880289.97250570
 92. Ali M, Khan SY, Rodrigues TA, Francisco T, Jiao X, Qi H, Kabir F, Irum B, ...**Naeem MA**, et al. A missense allele of PEX5 is responsible for the defective import of PTS2 cargo proteins into peroxisomes. *Hum Genet*. 2021 Apr; 140(4):649-666.
 93. Alva-Diaz C, Alarcon-Ruiz CA, Pacheco-Barrios K, Mori N, Pacheco-Mendoza J, Traynor BJ, Rivera-Valdivia A, **Lertwilaiwittaya P**, **Cornejo-Olivas M**, et al. C9orf72 hexanucleotide repeat in Huntington-like patients: Systematic review and meta-analysis. *Front Genet*. 2020 Nov 2; 11:551780.
 94. Baig HMA, Ansar M, Iqbal A, **Naeem MA**, Quinodoz M, Calzetti G, **Iqbal M**, Carlo Rivolta C. Genetic analysis of consanguineous Pakistani families with congenital stationary night blindness. *Ophthalmic Res*. 2021. (Submitted).
 95. Barney AM, Danda S, Abraham A, Fouzia NA, Gowdra A, Abraham SSC, Sony M, **Das S**, Korula S, et al. Clinicogenetic profile, treatment modalities and mortality predictors of Gaucher disease: A 15-year retrospective study. *Public Health Genomics*. 2021 Apr; 1-10. doi: 10.1159/000514507
 96. Bazalar-Montoya J, Bazalar-Montoya J, Sarapura-Castro E, Galarreta-Aima C, Ortega J, Galecio-Castillo M, Milla-Neyra K, Rodriguez R, **Duenas-Roque M**, **Cornejo-Olivas M**. Peruvian siblings with chronic neuropathy associated to IGHMBP2. *J Peripher Nerv Syst*. 2021 Mar. 26(1):139-140.
 97. Bazalar-Montoya J, Illanes-Manrique M, Inca-Martinez M, Marca V, Huaman-Dianderas F, Guevara-Fujita M, Fujita R, **Cornejo-Olivas M**. Genetic counseling to a DMD asymptomatic carrier: First case report in the Peruvian public healthcare system. *Rev Neuropsiquiatr*. 2020 Oct. 83(4): 278-283.
 98. Bello FO, Akanmu AS. **Adeyemo TA**, Idowu BM, Okonkwo P, Kanki PJ. Derangement of protein S and C4b binding protein levels as an acquired thrombophilia in HIV-infected adult Nigerians. *Sajhivmed*. 2021. (Accepted).
 99. Ben Haj Ali A, **Messaoud O**, Elouej S, Talmoudi F, Ayed W, Mellouli F, Ouederni M, Hadiji S, De Sandre-Giovannoli A, et al. FANCA gene mutations in North African Fanconi anemia patients. *Front Genet*. 2021 Feb 19; 12:610050.
 100. Bradley D, C Honeyman, Patel V, Zeolla J, Lester L, **Eshete M**, Demissie Y, Martin D, McGurk M. Smartphones can be used for patient follow-up after a surgical mission treating complex head and neck disfigurement in Ethiopia: Results from a prospective pilot study. *J Plast Reconstr Aesthet Surg*. 2021 Apr; 74(4):890-930.
 101. Bradley DR, Honeyman C, Patel V, Almas F, Walton G, **Eshete M**, Martin D, McGurk MJ. Improving smartphone follow-up after patient discharge from annual short-term head and neck missions in Ethiopia. *Plast Reconstr Aesthet Surg*. 2021 Mar; S1748-6815(21)00127-3.
 102. Cabral de Guimaraes TA, **Daich Varela M**, Georgiou M, Michaelides M. Treatments for dry age-related macular degeneration: therapeutic avenues, clinical trials and future directions. *Br J Ophthalmol*. 2021 Mar 19. doi: 10.1136/bjophthalmol-2020-318452
 103. Campus G, Diaz Betancourt M, Cagetti MG, Giacaman RA, Manton DJ, Douglas GVA, Carvalho TS, Carvalho JC, ... **Taiwo OO**, et al. The COVID-19 pandemic and its global effects on dental practice. An international survey. *J Dent*. 2021. (Accepted).

104. 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. *Pan Afr Med J*. 2021. (In review).
105. Castro V, Mattos D, Martinez-Carvalho F, Pontes-Cavalcanti D, **Duenas-Roque MM**, Llerena J, Cosentino V, Sayuri-Honjo R, Loguercio-Leite JC, et al. New SHH and known SIX3 variants in a series of Latin American patients with holoprosencephaly. *Mol Syndromol*. 2021; 12(4):219-233.
106. Chaudhuri J, Biswas S, Gangopadhyay G, Biswas T, Datta J, Biswas A, Datta AK, Mukherjee A, **Datta AK**, et al. A study of Mutation in ATP7B gene and its correlation with clinical phenotype and radiological features in Wilson Disease patients. *bioRxiv* 2021.01.21.427561. doi: 10.1101/2021.01.21.427561
107. Chaudhuri J, **Dutta AK**, Biswas T, Biswas A, Ray BK, Ganguly G. Charcot-Marie-Tooth disease type 4J with spastic quadriplegia, epilepsy and global developmental delay: a tale of three siblings. *Int J of Neurosci*. 2020 Oct; 1–4.
108. Cherif Ben Abdallah L, Mkaouar R, Naouali N, Lahbib S, Turki T, Elouej S, Bouyacoub S, Somai M, et al, **Messaoud O**. Oligogenism inheritance underlying incomplete penetrance of PROKR2 mutations in hypogonadotropic hypogonadism. *Front Genet*. 2021. (In press).
109. D'Antonio M, The COVID-19 Host Genetics Initiative (**Dutta AK**), Arthur TD, Nguyen P, Matsui H, Antonio-Chronowska A, Frazer KA. Insights into genetic factors contributing to variability in SARS-CoV-2 susceptibility and COVID-19 disease severity *medRxiv* 2021.05.10.21256423. doi: 10.1101/2021.05.10.21256423
110. **Daich Varela M**, Cabral de Guimaraes TA, Georgiou M, Michaelides M. Leber congenital amaurosis/early-onset severe retinal dystrophy: current management and clinical trials. *Br J Ophthalmol*. 2021 Mar 12. doi: 10.1136/bjophthalmol-2020-318483
111. **Daich Varela M**, Esener B, Hashem SA, Cabral de Guimaraes TA, Georgiou M, Michaelides M. Structural evaluation in inherited retinal diseases. *Br J Ophthalmol*. 2021 May 12. doi: 10.1136/bjophthalmol-2021-319228
112. **Daich Varela M**, Hufnagel RB, Guan B, Blain D, Sapp JC, Gropman AL, Alur R, Johnston JJ, Biesecker LG, Brooks BP. Clinical diagnosis of presumed SOX2 gonadosomatic mosaicism. *Ophthalmic Genet*. 2021 Mar; 15:1-6.
113. **Daich Varela M**, Ullah E, Yousaf S, Brooks BP, Hufnagel RB, Huryn LA. PDE6C: Novel mutations, atypical phenotype and differences among children and adults. *Invest Ophthalmol Vis Sci*. 2020 Oct 1; 61(12):1.
114. **Das S**, Godbole K, Abraham SSC, Ganesan P, Kamdar PP, Danda S. Alazami Syndrome: Report of three Indian patients with phenotypic spectrum from adolescence to adulthood. *Am J Med Genet*. 2021; 185A:1606-1609.
115. **Das S**, Thomas M, Yoganathan S, Muthusamy K, Barney A, Suneetha SCA, **Dutta AK**, Rekha A, Mohan S, et al. A descriptive study with molecular analysis in patients of ataxia telangiectasia (AT) from the Indian subcontinent. *Mov Disord Clin Pract*. 2021. (Submitted).
116. Deshpande D, Gupta SK, Sarma AS, Ranganath P, Nurul Jain SJ, Sheth J, **Mistri M**, Gupta N, Kabra M, et al. Functional characterization of novel variants in SMPD1 in Indian patients with acid sphingomyelinase deficiency. *Hum Mutat*. 2021 Jul 17. doi: 10.1002/humu.24263
117. Dharmasaroja PA, Assanasen J, Pongpakdee S, Jaisin K, Lolekha P, Phanasathit M, Cheewakriengkrai L, Chotipanich C, ...**Lertwilaiwittaya P**, et al. Etiology of dementia in Thai patients. *Dement Geriatr Cogn Dis Extra*. 2021; 11(1):64-70.

118. **Dutta AK**, Goswami K, Murugayan SB, 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.* 2021; 49(3):457–63.
119. **Dutta AK**, Goswami K. Host genomics of COVID-19: Evidence point towards alpha 1 antitrypsin deficiency as a putative risk factor for higher mortality rate. *Med Hypotheses.* 2021 Feb; 147:10485.
120. **Dwivedi A**, Beriwal A. Rare case of Al Kaissi syndrome in two siblings due to novel variant in CDK10 gene - first report from India. *Clin Dysmorphol.* 2021. (Submitted).
121. **Dwivedi A**, Bhat R, Bhat K. Changing indications of prenatal diagnosis in NGS era for 515 patients. 2021. (In preparation).
122. **Dwivedi A**, Chauhan L, Das G, Kumar A. A survey manuscript- knowledge, attitude, and practices towards genetic disorders amongst non-genetic clinicians of Armed Forces in India. 2021. (In preparation).
123. **Dwivedi A**, Mandal K, Moirangthem A, Pandey H, Phadke S, Gupta N. Yield of molecular testing in VHL and isolated VHL associated tumours in an Indian cohort. *Lab Med.* 2021. (Submitted).
124. **Dwivedi A**, Simalti A, Kalra S, Goswami J N, Negi V. Case series: Three cases emphasizing importance of genetics in PICU. *JPCC.* 2021; 8(3):153-156.
125. **Eshete M**, Abate F, Abera B, Hailu A, Demissie Y, Mossey P, Butali A. assessing the practice of birth defect registration at Addis Ababa health facilities. *Ethiop J Health Sci.* 2021; 31(3): 683- 687.
126. **Fatima SS**, Rehman R, Muhammad J, Martins R, Mohammed N, Khan U. Association of chemerin gene promoter methylation in maternal blood and breast milk during gestational diabetes. *J Dev Orig Health Dis.* 2021:1-7.
127. Guevara-Fujita M, Huaman F, Obispo D, Sánchez R, Barrenechea-Loli V, Rojas-Málaga D, Estrada-Cuzcano A, **Cornejo-Olivas M**, ... **Dueñas-Roque M**, et al. MLPA followed by target-NGS to detect mutations in the dystrophin gene of Peruvian patients suspected of DMD/DMB. *Mol Genet Genomic Med.* 2021; 00:e1759.
128. Hamdi Y, Mighri N, Boujemaa M, Mejri N, Ben Nasr S, Ben Rekaya M, **Messaoud O**, Bouaziz H, Berrazega Y, et al. Identification of eleven Novel BRCA mutations in Tunisia: Impact on the clinical management of BRCA related cancers. *Front Oncol.* 2021; 11: 674965.
129. Heike C, Hing A, Cunningham M, Chitayat D, Davis M, Doust S, Drake A, **Duenas-Roque M**, Griffin C, et al. Haploinsufficiency of SF3B2 causes craniofacial microsomia. *Nat. Commun.* 2021. (Accepted).
130. Honeyman C, **Eshete M**, Patel V, Martin D, McGurk M. Re: Use of the subclavian vessels for microvascular reconstruction in the vessel-depleted neck. A historical solution to a modern problem - the reverse flow, pedicled radial forearm flap as a back-up option in head and neck reconstruction. *Br J Oral Maxillofac Surg.* 2021 Jun; S0266-4356(21)00213-8.
131. Honeyman CS, Čebroň U, Bradley D, Patel V, **Eshete M**, McGurk M, Bertrand B, Martin D. High-tension closure of the submental flap donor site: Video and technical considerations. *Plast Reconstr Surg Glob Open.* 2020 Dec; 8(12):e3271.
132. Idowu EA, Fakuade BO, **Taiwo OO**, Majekodunmi JO, Alufohai OO, Sandabe FK. Dental caries prevalence, restorative needs and oral hygiene status in adult population: A cross sectional study among nurses in Jos University Teaching Hospital. *Nig J Basic Clin Sci.* 2021. (Accepted).
133. Jain G, Malhotra R, Ramchandran S, Nagaraj P, Appaswamy G, Sridharan U, Das G, **Dwivedi A**. Hypomagnesemia with secondary hypoparathyroidism and hypocalcemia due to novel

- variants in the transient receptor potential cation channel subfamily M member 6 (TRPM6) gene. Case based review. *J Pediatr Genet*. 2021. (Accepted).
134. Jaja C, Thurmond E, Ibemere S, **Edem-Hotah J**, Shepherd J. Diagnosing sickle cell disease in children in Sierra Leone with sickling test and SickleSCAN™ point-of-care test: A case series. *Oxf Med Case Reports*. 2021. (In review).
 135. Johns A, Luquetti D, Heike C, Hing A, Drake A, Cunningham M, Chitayat D, David M, **Dueñas-Roque MM**, et al. Parental reports of academic status and prevalence of teasing in a multinational craniofacial microsomia pediatric study. *Facial Plast Surg Aesthet Med*. 2021. (Submitted).
 136. Joseph AO, Li YH, Salako O, Doi S, Balogun OD, Awofeso OM, **Abdulkareem FB**, Onitilo AA. A review of breast cancer pathology reports in Nigeria. *Ecancermedicalscience*. 2021 Feb; 15:1190.
 137. Kamulegeya R, Kateete P, Bagaya B, Nasinghe E, **Muttamba W**. Biobanking: Strengthening Uganda's rapid response to COVID-19 and other epidemics. *Biopreserv Biobank*. 2021. (Accepted).
 138. Khan SY, Ali M, Lee MW, Ma Z, Biswas P, Khan AA, **Naeem MA**, Riazuddin S, Riazuddin S, et al. Whole genome sequencing data of multiple individuals of Pakistani descent. *Scientific Data*. 2020 Oct 13; 7(1):350.
 139. Kirenga B, Byakika-Kibwika P, **Muttamba W**, Kayongo A. Efficacy of convalescent plasma for treatment of COVID-19 in Uganda. *BMJ Open Resp Res*. 2021. (Accepted).
 140. Kolluri N, Kamath S, Lally P, Zanna M, Galagan J, **Gitaka J**, Kamita M, Cabodi M, Lolabattu SR, et al. Development and clinical validation of Iso-IMRS: A novel diagnostic assay for *P. falciparum* malaria. *Anal Chem*. 2021; 93(4):2097-2105.
 141. Kosiyo P, Otieno W, **Gitaka J**, Munde EO, Ouma C. Haematological abnormalities in children with sickle cell disease and non-severe malaria infection in western Kenya. *BMC Infect Dis*. 2021; 21:329.
 142. Kotecha U, **Mistri M**, Chand P, Shah P, Shah N. The diagnostic utility of exome-based carrier screening in families with a positive family history. *Am J Med Genet A*. 2021. (Revision Submitted).
 143. Kotecha U, **Mistri M**, Shah P, Nidhi N, Jain V, Goyal M. A novel mutation in KIF5C gene: Expanding the mutation spectrum and genotype-phenotype correlation of patients with and without hot-spot mutations. *Neuropediatrics*. 2021. (Submitted).
 144. Kumar R, **Dwivedi A**, Puri R. Prenatal diagnosis of radial ray defect associated with Fanconi anemia. *J Fetal Med*. 2021; 8:75-79.
 145. Ladipo-Ajayi O, Ihediwa G, Jolayemi E, Haruna M, Alakaloko F, Seyi-Olajide J, **Abdulkareem FB**, Kanu O. Enteromyeloschisis. A rare sacro - enteric mass in the newborn. *J Pediatr Surg Case Rep*. 2021; 64(1): 101707.
 146. **Lertwilaiwittaya P**, Roothumnong E, Nakthong P, Dungort P, Meesamaranpong C, Tansa-Nga W, Pongsuktavorn K, ...Limwongse C, Pithukpakorn M, et al. Thai patients who fulfilled NCCN criteria for breast/ovarian cancer genetic assessment demonstrated high prevalence of germline mutations in cancer susceptibility genes: Implication to Asian population testing. *Breast Cancer Res Treat*. 2021; 188(1):237-248.
 147. **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 RANKL osteopetrosis. *Mol Genet Genomic Med*. 2021 May 30:e1727. doi: 10.1002/mgg3.1727

148. Loesch DP, Horimoto ARVR, Heilbron K, Sarihan EI, Inca-Martinez M, Mason E, **Cornejo-Olivas M**, Torres L,...Latin American Research Consortium on the Genetics of Parkinson's Disease (LARGE-PD). Characterizing the genetic architecture of Parkinson's Disease in Latinos. *Ann Neurol*. 2021 Jul. doi: 10.1002/ana.26153
149. Marca-Ysabel MV, Rajabli F, **Cornejo-Olivas M**, Whitehead PG, Hofmann NK, Illanes Manrique MZ, Véliz-Otani DM, Milla Neyra AK, Castro Suarez S, et al. Dissecting the role of Amerindian genetic ancestry and the ApoE ϵ 4 allele on Alzheimer disease in an admixed Peruvian population. *Neurobiol Aging*. 2021 May; 101:298.
150. Mbugua S, **Gitaka J**, Gitau T, Odwe G, Mwaura P, Liambila W, Ndwiga C, K'Oduol K, Warren C, Abuya T. Family and provider perceptions of quality of care in the management of sick young infants in primary healthcare settings in four counties of Kenya. *BMJ Open Quality*. 2021 Jul 1; 10(3):e001125.
151. **Mburu S**, Mutuku I, Kimani K. Readiness of primary health care diagnostic laboratory services to support UHC programme in Kenya. A case study of three counties. *Journal of Health & Environmental Research*. 2020; 6(4):128-142.
152. McLellan A, Titualer P, Sidney D, Aden A, Lacroix A, **Edem-Hotah J**. Examining person-centered maternal care services at The Princess Christian Maternity Hospital, Freetown, Sierra Leone. 2021. (In preparation).
153. Musa J, Kim K, Zheng Y, Travis LB, Gursel B, Silas OA, **Abdulkareem FB**, Imade G, Akanmu A, et al. Accelerated epigenetic age among HIV-infected Nigerian women with invasive cervical cancer. *J Clin Oncol*. 2021; 39:15_suppl, e17504-e17504.
154. **Muttamba W**, Lusiba J, Namakulu O, Byakika-Kibwika P, et al. Feasibility of collecting and processing of COVID-19 Convalescent plasma for treatment of COVID-19 in Uganda. *Plos One* 2021 June; 16(6):e0252306.
155. Nabouli I, Chikhaoui A, Othman H, Elouej S, Jones M, Lagarde A, Rekaya MB, **Messaoud O**, Zghal M, et al. Case Report: Identification of novel variants in ERCC4 and DDB2 genes in two Tunisian patients with atypical Xeroderma Pigmentosum phenotype. *Front Genet*. 2021 May 31; 12:650639.
156. **Nakousi-Capurro N**, Alegría A, Cares C, Gainza-Lein M, Ojeda V, Irrarrázaval MJ. Congenital anomalies and comorbidities in Down Syndrome neonates (RCHPED 1518). *Rev Chil Pediatr*. 2020; 91(5):732–742.
157. 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, Ikeja. *Pan Afr Med J*. 2021. (In review).
158. Odeyemi AO, Odeyemi AO Awopeju OF, **Adewole OO**, Tanimowo MO, Erhabor GE. Pulmonary tuberculosis in a cohort of HIV patients in a tertiary hospital in Osogbo, Southwestern Nigeria. *Nig Del Med J*. 2021; 5(2):7-15.
159. Odeyemi AO, Odeyemi AO, Awopeju OF, **Adewole OO**, Tanimowo MO, Erhabor GE. Community-acquired lower respiratory tract infections in an adult population with HIV infection in a Nigerian tertiary hospital. *Res. J. of Health Sci*. 2021; 9(2):150-157.
160. Ojewunmi OO, **Adeyemo TA**, Oyetunji AI, Benn Y, Ekpo MG, Iwalokun BA. Association of alpha-thalassemia and Glucose-6-Phosphate Dehydrogenase deficiency with transcranial Doppler ultrasonography in Nigerian children with sickle cell anemia. *J Clin Lab Anal*. 2021 Jun; 35(6):e23802.

161. **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*. 2021. (Accepted).
162. **Oluwarotimi AC**, Emeka CI, Gbotolorun OM. Assault-related maxillofacial injuries in Nigeria: a review of the literature. *Nigerian Journal of Oral and Maxillofacial Surgery*. 2021. (Accepted).
163. Oluwole EO, **Adeyemo TA**, Osanyin GE, Odukoya OO, Kanki PJ, Afolabi BB. Feasibility and acceptability of early infant screening for sickle cell disease in Lagos, Nigeria—A pilot study. *PLoS ONE*. 2020; 15(12):e0242861.
164. Oluwole EO, Adeyemo TA. Predictors of knowledge of sickle cell disease among mothers of younger infants attending immunization clinic in an urban community in Lagos, Nigeria. *Journal of Community Medicine and Primary Health Care (JCMPHC)*. 2021. (Accepted).
165. Otoh EC, **Taiwo OO**, Adeleke OA, Majekodunmi JO, Ajike SO. Dental trauma in adults and elderly Nigerians. *West Afr J Med*. 2021; 38(4):313-320.
166. Pal A, Squitti R, Picozza M, Pawar A, Rongioletti M, **Dutta AK**, Sahoo S, Goswami K, Sharma P, et al. Zinc and COVID-19: basis of current clinical trials. *Biol Trace Elem Res*. 2020 Oct; 1-11.
167. Plasencia-Duenas EA, Diaz-Velez C, **Dueñas-Roque MM**. Factors associated to the occurrence of cleft lip and palate in newborns in a tertiary care Peruvian hospital. A case-control study. *Acta Med Peru*. 2020 Oct.7; 37(3):304-311.
168. 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. *Indian J Pediatr*. 2021; 88:836.
169. Rebecca R, **Edem-Hotah J**, Quaki IA. Factors influencing utilization of insecticide treated nets among children under-five years of age in Kpone-On-Sea in the Kpone Katamanso District in Ghana. *Sierra Leone J Biomed Res*. 2021. (Accepted).
170. Rodriguez RS, **Cornejo-Olivas M**, Bazalar-Montoya J, Sarapura-Castro E, Torres-Loarte M, Rivera-Valdivia A, Sulcahuaman-Allende Y. Novel compound heterozygous mutation c.3955_3958dup and c.5825C>T in the ATM Gene: Clinical evidence of ataxia-telangiectasia and cancer in a Peruvian family. *Mol Syndromol*. 2021 Jun. doi: 10.1159/000515696
171. Rodriguez-Mateos P, Ngamsom B, Walter C, Dyer CE, **Gitaka J**, Iles A, Pamme N. A lab-on-a-chip platform for integrated extraction and detection of SARS-CoV-2 RNA in resource-limited settings. *Anal Chim Acta*. 2021; 1177:338758
172. Romdhane L, Mezzi N, Dallali H, **Messaoud O**, Shan J, Fakhro KA, Kefi R, Chouchane L, Abdelhak S. A map of copy number variations in the Tunisian population: a valuable tool for medical genomics in North Africa. *NPJ Genom Med*. 2021 Jan 8; 6(1):3.
173. Sarapura-Castro E, Cosentino C, Landman J, Landman A, Torres L, Nuñez Y, Capellari S, Parchi P, **Cornejo-Olivas M**. Early sensory disturbances and seizures are common manifestations of familial Creutzfeldt-Jakob disease due to E200K PRNP mutation: Case report from two Peruvian families. *Clin Neurol Neurosurg*. 2021 Mar; 202:106490.
174. Sarihan EI, Pérez-Palma E, Niestroj LM, Loesch D, Inca-Martinez M, Horimoto ARVR, **Cornejo-Olivas M**, Torres L,...Latin American Research Consortium on the Genetics of Parkinson's Disease (LARGE-PD). Genome-wide analysis of copy number variation in Latin American Parkinson's disease patients. *Mov Disord*. 2021 Feb; 36(2):434-441.
175. Suárez C, Aranibar L, **Nakousi-Capurro N**, **Díaz C**, **Pizarro P**. Pigmentary mosaicisms and cytogenetic abnormalities: About 2 cases. *Piel. Formación continuada en dermatología*. 2021 May. Elsevier. doi: 10.1016/j.piel.2020.07.020

176. Sulaimon LA, Adisa RA, Samuel TA, Joel IY, Ayankojo AG, **Abdulkareem FB**, Olaniyi TO. Molecular mechanism of mitochinone mesylate in mitigating the progression of hepatocellular carcinoma-in silico and in vivo studies. *J Cell Biochem*. 2021 Apr 28. doi: 10.1002/jcb.29937
177. **Taiwo O.O.** Community pharmacists, pharmacy counter assistants and oral health promotion in Plateau State. *West Afr J Med* 2021; 38(6):434-438.
178. The COVID-19 Host Genetics Initiative (**Dutta AK**), Andrea Ganna. Mapping the human genetic architecture of COVID-19 by worldwide meta-analysis. medRxiv 2021.03.10.21252820.
179. Umeizudike KA, Osagbemi BB, Daramola OO, **Adeyemo TA**. Oral health related quality of life among HIV positive patients attending two HIV outpatient clinics in Nigeria - a cross sectional study. *African Health Sciences (AHS)*. 2021. (Accepted).
180. Véliz-Otani D, Cubas-Montecino D, Milla-Neyra K, Ashizawa T, Saraiva-Pereira ML, Jardim LB, **Cornejo-Olivas M**. Response to ATXN10 microsatellite distribution in a Peruvian Amerindian population. *Cerebellum*. 2021 Mar. doi: 10.1007/s12311-021-01258-3
181. **Wangi KYW**. Genome Editing: The Promise of a Technology. *Kompas*. 2021. (In preparation).
182. **Wangi KYW**. Health Professions in the era of precision medicine. Online newspaper: Kompasiana; May 2021.
183. Zghal M, **Messaoud O**, Mokni M. Xeroderma pigmentosum. *EMC Dermatologie*. 2020; 0(0):1-21 [Article 98-660-A-10]. doi:10.1016/S0246-0319(20)42977-2
184. Zheng Y, Hawkins C, Okeke E, Lesi OA, Qu Y, Roberts LR, Gursel D, **Abdulkareem FB**, ... **Adeyemo WA**, et al. Acceleration of blood-based circulating cell-free DNA epigenetic age among HIV-infected patients with hepatocellular carcinoma in Nigeria. *J Clin Oncol*. 2021; 39:15_suppl, e16137-e16137.
185. Zheng Y, Musa J, Joyce BT, Wang J, Gursel D, Silas OA, **Abdulkareem FB**, Imade G, Akanmu A, et al. Global line-1 hypomethylation as novel biomarker for cervical cancer in Nigerian women living with HIV. *J Clin Oncol*. 2021; 39:15_suppl, e17513-e17513.

2017 Batch

186. **Abad PJ**, Sur ALD. Genetics and genomics nursing in the Philippines. *Int Nurs Rev*. 2021. (In review).
187. Acheampong G, **Owusu M**, Nkrumah B, Obeng-Boadi P, Opare DA, Sambian DJ, Angra P, Walker C. Laboratory capacity in COVID-19 diagnosis and the need to enhance molecular testing in Ghana. *Global Security: Health, Science and Policy*. 2021; 6(1):10-17.
188. Adedokun B, Du Z, Gao G, Ahearn TU, Lunetta KL, Zirpoli G, Figueroa J, ... **Torres-Mejía G** ... Huo D. Cross-ancestry GWAS meta-analysis identifies six breast cancer loci in African and European ancestry women. *Nat Commun*. 2021 Jul; 7; 12(1):4198.
189. Adekunle AA, Olowo AY, Adetona M, **James O**, Adamson OO, Agbogidi FO, Oladega A, Ogulewe M, **Adeyemo WL**, et al. Variations in facial anthropometric measurements among major ethnic groups in Nigeria: A 3-dimensional stereophotogrammetry analysis. *FACE*. July 2021. doi: 10.1177/27325016211029013
190. Amin S, AlJboor M, Toro MD, Rejdak R, Nowomiejska K, Nazzal R, Mohammad M, 2 more.... **Yousef YA**. Management and outcomes of unilateral Group D tumors in retinoblastoma. *Clin Ophthalmol*. 2021; 15:65-72.
191. Amoo AT, **James O**, Adeyemi MO, Taiwo OA, **Adeyemo WL**. Does the initial width of cleft lip play a role in the occurrence of immediate local complications following primary cleft lip repairs? *J Craniofac Surg*. 2021; 32:670-674.

192. Anastacio T, Benevides N, Moraes E, Quinelato V, Calasans-Maia J, Aguiar T, Machado A, **Casado PL**. Analysis of Active Oral Tactile Sensitivity in Individuals with Complete Natural Dentition. *J Contemp Dent Pract* 2021; 22(3):268-272.
193. Ayisi-Boateng NK, **Owusu M**, Tawiah P, Ampah B, Augustina AS, Wusu-Ansah OK, Fred SS, Richard OP. Profile and outcomes of hospitalized patients with COVID-19 at a tertiary institution hospital in Ghana. *Ghana Medical Journal*. 2020; 54(4):39-45.
194. Biswas S K, M Alam, **Roy S**, Ahmed T, Bhuyan A M. A newer technique of ocular surface reconstruction in unilateral lime burn. *Journal of the Ophthalmological Society of Bangladesh*. 2021; 47(1):7-15.
195. Bravo-Ramírez M, Morales-Peralta E, Zúñiga-Rosales Y, **Benítez-Cordero Y**, González-Torres MA, **Roblejo-Balbuena H**, Marcheco-Teruel B. Covid-19: first outbreak in Cuban pediatric patients. *Rev Cubana Pediatría*. 2021. (Submitted).
196. Camilletti MA, Mayer JM, **Vishnopolska SA**, Perez-Millan MI. From pituitary stem cell differentiation to regenerative medicine. *Front Endocrinol*. 2021; 11:614999.
197. Castro JPG, Aguiar TRS, Tristão GC, Alves GG, Prado M, Quinelato V, **Casado PL**, Romanos GE. Peri-implant health after supportive mucositis therapy is associated with increased levels of FGF-2. *Braz Dent J*. 2021. (Accepted).
198. Ceylanoglu K, Guntekin Ergun S, **Yaylacioglu Tuncay F**, Tarlan B, Konuk O. Association of FOXP3 gene polymorphisms with the risk of Graves' disease and Graves' ophthalmopathy in Turkish population. 2021. (In preparation).
199. Cordeiro BQS, Carvalho WR, Fonseca EM, Martins CCP, Moreno MD, Quinelato V, Casado PL. Vertical discrepancy in height of Morse Cone pillars submitted to different torque forces. *Materials*. 2021. (Accepted).
200. Cordeiro PCF, Bonato LL, Quinelato V, Vieira AR, Granjeiro JM, Tesch R, **Casado PL**, Calasans-Maia J. Matrix metalloproteinase-2 gene polymorphisms decrease chances of muscle TMD development in Brazilian population. *J Dent & Oral Disord* 2021; 7(3):1163.
201. Egbunah UP, Adamson O, Fashina A, Adekunle AA, **James O**, **Adeyemo WL**. Comparing the treatment outcomes of absorbable sutures, non-absorbable sutures, and tissue adhesives in cleft lip repair: A systematic review. *Cleft Palate Craniofac J*. 2021 Mar 3;1055665621996107. doi: 10.1177/1055665621996107
202. Elfalah M, AlRyalat SA, Toro MD, Rejdak R, Zweifel S, Nazzal R, Abu-Ameerh M, Ababneh O,...**Yousef YA**. Delayed intravitreal anti-VEGF therapy for patients during the COVID-19 lockdown: An ethical endeavor. *Clin Ophthalmol*. 2021; 15:661-669.
203. Frimpong M, Amoako YA, Anim KB, Ahor HS, Yeboah R, Arthur J, Dakorah JS, Gborgblovor D, **Owusu M**, et al. Diagnostics for COVID-19: A case for field-deployable, rapid molecular tests for community surveillance. *Ghana Medical Journal*. 2020; 54(4):71-76.
204. Gergics P, Smith C, Bando H, Jorge AA, Rockstroh-Lippold D, **Vishnopolska SA**, Castinetti F, Maksutova M, Camper SA, et al. High-throughput splicing assays identify missense and silent splice-disruptive POU1F1 variants underlying pituitary hormone deficiency. *Am J Hum Genet*. 2021; S0002-9297(21)00237-8.
205. Global Retinoblastoma Study Group (**Roy S**, 278 retinoblastoma treatment centers). The role of maternal age and birth order on the development of unilateral and bilateral retinoblastoma: A multicentre study. *J Pediatr Haematol Oncol*. 2021. (Submitted).
206. Gokhroo RK, Kaul A, Mohammad TT, Chandra K, **Nandal R**, Kumar A, Goyal M, Chandra S. 1 year follow Up results of “ARTIM HF TRIAL” (angiotensin receptor neprilysin inhibitor effect on TEI index & left ventricular mass in heart failure). *Indian Heart J*. 2021; 73(2):205-210.

207. Gyanwali P, Bhattarai S, Dixit SM, Gautam B, **Thakur N**, Bhattarai NR, Sharma SK, Dhimal M. Deciphering whole genomic sequence of SARS-CoV-2 isolated from COVID-19 patients in Nepal. 2021. (In preparation).
208. Horn D, Fernández-Núñez E, Gomez-Carmona R, Rivera-Barahona A, Nevado J, Schwartzmann S, Ehmke N, Lapunzina P, **Otaify GA**, et al. Biallelic truncating variants in MAPKAPK5 cause a new developmental disorder involving neurological, cardiac, and facial anomalies combined with synpolydactyly. *Genet Med*. 2021 Apr; 23(4):679-688.
209. Hosen MI, Forey N, Durand G, Voegelé C, Bilici S, **Avogbe PH**, Delhomme TM, Foll M, Manel A, et al. Development of sensitive droplet digital PCR assays for detecting urinary TERT promoter mutations as non-invasive biomarkers for detection of urothelial cancer. *Cancers (Basel)*. 2020 Dec; 12(12):3541.
210. **James O**, Adekunle AA, Adamson OO, Agbogidi OF, **Adeyemo WL**, Butali A, Ladeinde AL, Ogunlewe MO. Management of orofacial cleft in Nigeria - A retrospective study. *Ann Maxillofac Surg*. 2021; 10(2):434-438.
211. **James O**, Adekunle AA, **Adeyemo WL**. How does Octyl –2- cyanoacrylate tissue adhesive compare with prolene sutures in cleft lip repair? *J Oral Maxillofac Surg*. 2021; 79(7):1540-1548.
212. **James O**, Akinboboye BO, Okunade KS, Adekunle AA, **Adeyemo WL**. Evaluation of the use and effectiveness of telemedicine among the health professionals during the Covid-19 lockdown period: A cross sectional study. *J Clin Sciences*. 2021; 18:117-122.
213. **James O**, Erinoso OA, 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; 23(11):1561-1565.
214. **James O**, Oyeneyin, AO, Adeyemi, MO, Erinoso OA, Adekunle AA, **Adeyemo WL**. Neurosensory deficits of inferior alveolar nerve following impacted mandibular third molar extraction: Comparison of one-stage complete extraction with two-stage partial coronectomy surgical technique. *J. Maxillofac Oral Surg*. 2021. doi: 10.1007/s12663-021-01601-5
215. Kadir M, Hasanuzzaman M, Gatmaitan Y, Mansurali V, Mitra M, Haider G, **Roy S**. Outcome of minimally invasive harvesting Fascia Lata (FL) in ophthalmic plastic surgery. *Eur J Ophthalmol*. 2021. (Submitted).
216. Kague E, Turci F, Newham E, Yang Y, Brown KR, Aglan MS, **Otaify GA**, Temtamy SA, Ruiz-Perez VL, et al. 3D assessment of intervertebral disc degeneration in zebrafish identifies changes to bone density that prime disc disease. *Bone Res*. 2021 Mar 15. (Accepted).
217. Khairat R, Elhossini R, Sobreira N, Wohler E, **Otaify G**, Mohamed A, Ragaa E, **Sayed I**, Aglan M, et al. Expansion of the phenotypic and mutational spectrum of Carpenter syndrome: two novel mutations in RAB23 in 2 Egyptian patients. *Mol. Syndromol*. 2021. (Submitted).
218. Kocova M, Anastasovska V, **Petlichkovski A**, Falhammar H. First insights into the genetics of 21-hydroxylase deficiency in the Roma population. *Clin Endocrinol (Oxf)*. 2021 Jul; 95(1):41-46.
219. Kunwar AJ, Thapa S, **Thakur N**, Joshi G, Shrestha R, Kafle S, Shrestha S, Shrestha K, Giri KR et al. Antibigram and comparison of Salmonella typhi detection in suspected patients by conventional blood culture, serology, and advanced PCR technique. *J Nepal Med Assoc*. 2021. (Submitted).
220. Lamptey J, Oyelami F.O, **Owusu M**, Nkrumah B, Oluwagbenga IP, Adu-Gyamfi EA, Czika A, El-Duah P, Yeboah R, et al. Genomic and epidemiological characteristics of SARS-CoV-2 in Africa. *PLoS Negl Trop Dis*. 2021; 15(4):e0009335.

221. Lomardo PG, Cardoso N, Motta L, Aguiar T, Neves FPG, Aguiar-Alves F, Calasans M, Quinelato V, **Casado PL**. Streptococcus pneumoniae prevalence in nasopharynx, oropharynx and gingival sulcus in Brazilian adults: A preliminary study. *Microb Pathogen* 2021; 156:104924.
222. **Lotz-Esquivel S**, Alvarado-Aguilar M, Saborío-Rocafort M. Genetic Services in Costa Rica. 2021. (In-preparation).
223. Madyadi A, **Dhoro M**, Shamu T, Washaya T, Kouamou V, Chimukangara B, Katzenstein D, Manasa J. HIV-1 genetic diversity and natural polymorphisms of the integrase gene in integrase inhibitor naive patients in Harare, Zimbabwe. *AIDS Res Hum Retroviruses*. 2021. (Submitted).
224. **Mahfoudh W**, Snoussi K, Gabbouj S, Bouaouina N, Chouchane L and Zakhama A. BRCA1 gene analysis in triple negative breast cancer in the Tunisian population. 2021. (In preparation).
225. **Mehrez MI**, ElBadry T, Nemat A, El Batran M, Lasheen N, AboulEzz E. Craniofacial anomalies and 3D imaging modalities with reference to the Egyptian experience; *Middle East J of Med Genet*. 2021. (Submitted).
226. Mendez R, Iqbal S, **Vishnopolska S**, Martinez C, Dibner G, Aliano R, Zaiat J, Biagioli G, Marti MA, et al. Oculocutaneous albinism type 1B associated with a functionally significant tyrosinase gene polymorphism detected with Whole Exome Sequencing. *Ophthalmic Genet*. 2021; 42(3): 291-295.
227. Moraes NB, Moraes EJ, Anastacio THS, Silva LE, Machado AN, Martins CCP, Schoichet JJ, Rangel B, Monte Alto RV, Romanos GE, **Casado PL**. Active tactile sensibility of Brånemark protocol prostheses: a double-blind randomized clinical study. *Materials*. 2021. (Accepted).
228. Nasrollahzadeh D, Roshandel G, Delhomme TM, **Avogbe PH**, Foll M, Malekzadeh R, Mckay J, Brennan P, Abedi-Ardekani B. TP53 targeted deep sequencing of cell-free DNA in esophageal squamous cell carcinoma using low-quality serum: Concordance with tumor mutation. *Int. J. Mol. Sci*. 2021 Jun; 22(11):5627.
229. Nuruddin M, **Roy S**, Hoque F. Combined subconjunctival pegylated (Peg) interferon alfa 2a injection and amniotic membrane graft for management of extensive conjunctival papillomatous neoplasia- A case report. *IP Int J Ocul Oncol Oculoplasty*. 2021; 7(1):94-96.
230. Nuruddin M, **Roy S**, Hoque F. Pegylated interferon alpha 2a for the treatment of ocular surface squamous neoplasia (OSSN)- A case report. *Oman J Ophthal*. 2021. (Submitted).
231. **Owusu M**, Sylverken AA, El-Duah P, Ayisi-Boateng NK, Yeboah R, Adu E, Asamoah J, Frimpong M, Senyo J, et al. Low risk of SARS-CoV-2 in blood transfusion. *PloS ONE*. 2021 Apr; 16(4):e0249069.
232. **Owusu M**, Sylverken AA, Twumasi SA, El-Duah P, Ayisi-Boateng NK, Yeboah R, Asamoah J, Binger T, Acheampong G et al. Epidemiological profile of SARS-CoV-2 among selected regions in Ghana: A cross-sectional retrospective study. *PloS ONE*. 2020;1(12):e0243711.
233. Palencia-Campos A, Aoto PC, Machal EM, Rivera-Barahona A, Soto-Bielicka P, Bertinetti D, Baker B, Vu L, ...**Otaify GA**, et al. Germline and mosaic variants in PRKACA and PRKACB cause a multiple congenital malformation syndrome. *Am J Hum Genet*. 2020; 107(5):977-988.
234. Palencia-Campos A, Aoto PC, Machal EMF, Rivera- Barahona A, Soto-Bielicka P, Bertinetti D, ...**Mehrez MI**, **Otaify GA**, Temtamy S, et al. Germline and mosaic variants in PRKACA and PRKACB cause a multiple congenital malformation syndrome. *Am J Hum Genet* 2020; 107(5):977-988.
235. Pasquale C, Gheno E, **Casado PL**, Benedicenti S. Acido 5-delta-aminolevulinico una nuova frontiera nella terapia fotodinamica. *Parodontologia e Stomatologia*. 2021 Jun; 60.

236. Pavinato L, Trajkova S, Grosso E, Giorgio E, Bruselles A, Radio FC, Pippucci T, Dimartino P, **Petlichkovski A**, et al. Expanding the clinical phenotype of the ultra-rare Skraban-Deardorff syndrome: Two novel individuals with WDR26 loss-of-function variants and a literature review. *Am J Med Genet A*. 2021 Jun; 185(6):1712-1720.
237. Radwan H, Rabie E, Mostafa M, Ahmed N, Abouzaid M, Hassib N, **Mehrez M**, El-Kamah Gh, Amr K, **Sayed I**. Targeted next generation sequencing identifies three genes (EDA, EDAR, EDARADD) to account for more than half of Egyptian ectodermal dysplasia with or without hypohidrosis. 2021. (In preparation).
238. Rodríguez-Valentín R, **Torres-Mejía G**, Martínez-Matsushita L, Angeles-Llerenas A, Gómez-Flores-Ramos L, Wolff R, Baumgartner KB, Hines LM,...**Ortiz-Panozo E**, et al. Energy homeostasis genes modify the association between serum concentrations of IGF-1 and IGFBP-3 and breast cancer risk: The Breast Cancer Health Disparities Study. *Cancer Epidemiol Biomarkers Prev*. 2021. (Submitted).
239. **Roy S**, Hoque F, Mudhar S, Nuruddin M. Diffuse large cell lymphoma of lacrimal sac may mimic as acute dacryocystitis - A case report. *HORIZON*. 2021. (In press).
240. **Roy S**, Kaliki S. Retinoblastoma- A major review. *Mymensing Med J*. 2021 Jul; 30(3): 881-895.
241. Schoichet JJ, Fonseca EM, Ramirez C, Villas-Boas R, Prazeres J, Quinelato V, Aguiar TR, Prado M, **Casado PL**. Epidermic Growth Factor is associated with loss of mucosae sealing and peri-implant mucositis development. *Materials*. 2021. (Accepted).
242. Sylverken AA, El-Duah P, **Owusu M**, Schneider J, Yeboah R, Ayisi-Boateng NK, Gorman R, Adu E, Kwarteng A et al. Transmission of SARS-CoV-2 in northern Ghana: insights from whole-genome sequencing. *Arch Virol*. 2021; 166(5):1385-1393.
243. **Thakur N**, Singh N, Shrestha S, Gyanwali P, Pokhrel S, Karki AR, Dhungana A, Acharya N, Neupane N. Hematological and biochemical biomarkers in COVID-19 patients. 2021. (In preparation).
244. Valentina Castillo J, **Catherine Díaz S**, Bustamante ML, Ferreira MG, Teive HAG, Miranda M. Autosomal recessive cerebellar ataxia 1: First case report depicting a variant in SYNE1 gene in a Chilean patient. *Cerebellum*. 2021 Mar. doi: 10.1007/s12311-021-01250-x
245. **Vishnopolska SA**, Mercogliano MF, Camilletti MA, Mortensen AH, Braslavsky D, Keselman A, Pérez-Millán MI. Comprehensive identification of pathogenic gene variants in patients with neuroendocrine disorders. *J Clin Endocrinol & Metab*. 2021; 106(&):1956-1976.
246. **Yaylacioglu Tuncay F**, Karaismailoglu E, Ozdek S. Practice Patterns and Needs Assessment of Ophthalmologists for Inherited Eye Disorders: Do we need a subspecialty as Ophthalmic Genetics? 2021. (In preparation).
247. **Yousef YA**, Al Jboor M, Mohammad M, Mehayar M, Toro MD, Nazzal R, Alzureikat QH, Rejdak M, Elfalah M, Sultan I, Rejdak R, Al-Hussaini M and Al-Nawaiseh I. Safety and efficacy of intravitreal chemotherapy (melphalan) to treat vitreous seeds in retinoblastoma. *Front. Pharmacol*. 2021; 12:696787. doi: 10.3389/fphar.2021.696787
248. **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*. 2021; 128(1):130-137.
249. **Yousef YA**, Mohammad M, Jaradat I, Shatnawi R, Mehayar M, Al-Nawaiseh I. Prognostic factors for eye globe salvage by external beam radiation therapy for resistant intraocular retinoblastoma. *Oman J Ophthalmol*. 2020 Nov; 13:123-128.

250. **Yousef YA**, Mohammad M, Mehyar M, Sultan I, Al-Hussaini M, Alhourani J, Halalshah H, Khzouz J, Jaradat I, et al. The predictive value of the eighth edition of the clinical TNM staging system for the likelihood of eye salvage for intraocular retinoblastoma by systemic chemotherapy and focal therapy. *J Pediatr Hematol Oncol*. 2021 Mar 23. doi: 10.1097/MPH.0000000000002144
251. Zaki MS, **Otaify GA**, Ismail S, Issa MY, El-Ruby MO, Sadek AA, Ashaat EA, El Saeidi SA, Aglan MS, et al. Blepharophimosis-ptosis-intellectual disability syndrome: A report of nine Egyptian patients with further expansion of phenotypic and mutational spectrum. *Am J Med Genet Part A*. 2020 Dec; 182 (12):2857-2866.

2016 Batch

252. **Adeyemo AA**, Ogunkeyede SA, Ogundoyin OA, Oyelakin OA. Evolving telemedicine practice: Experiences of health care workers during covid-19 pandemic. *Ann Ibd. Pg. Med*. 2021; 19 COVID-19 Supplement: S45-S49.
253. **Adeyemo WL, James O**, Bamigboye BA, Akinola MD, Adamson OO, Nkemjika BN, Ibikunle AA, Ogunlewe MO, Ladeinde AL, et al. Missense variants within GJB2 gene locus and the risk of hearing defects in nonsyndromic cleft lip and palate. *J Craniofac Surg*. 2021 Jun 30. doi: 10.1097/SCS.00000000000007921
254. **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*. 2021; 20:149-153.
255. Akpa OM, Okekunle AP, Ovbiagele B, Sarfo FS, Akinyemi RO, Akpalu A, Wahab KW, Komolafe M,...**Adeoye AM**, et al. Factors associated with hypertension among stroke-free indigenous Africans: Findings from the SIREN study. *J Clin Hypertens (Greenwich)*. 2021. doi: 10.1111/jch.14183
256. Borda V, Alvim I, Mendes M, Silva-Carvalho C, Soares-Souza GB, Leal TP, Furlan V, Scliar MO,...**Guio H**. The genetic structure and adaptation of Andean highlanders and Amazonians are influenced by the interplay between geography and culture. *Proc Natl Acad Sci USA*. 2020; 117(51):32557-32565
257. Dasanayaka NN, **Sirisena ND**, Samaranayake N. The effects of meditation on length of telomeres in healthy individuals: a systematic review. *Syst Rev*. 2021; 10:151.
258. Dissanayake R, Senanayake MP, Fernando J, Robertson SP, Dissanayake VHW, **Sirisena ND**. Frontometaphyseal dysplasia 1 in a patient from Sri Lanka. *Am J Med Genet A*. 2020; 185(4):1317-1320.
259. Egbunah UP, Adekunle AA, **Adeyemo WL**. Comparing the effectiveness of non-surgical and surgical treatment modalities in the management of oral cancer: A systematic review and meta-analysis. *FACE*. 2021; 2:210-220.
260. **Ekure EN**, Adeyemo A, Liu H, Sokunbi O, Kalu N, Martinez AF, Owosela B, Tekendo-Ngongang C, Addissie YA, et al. Exome sequencing and congenital heart disease in sub-Saharan Africa. *Circ Genom Precis Med*. 2021 Jan 15; 14:e003108.
261. **Ekure EN**, Adeyemo AA. Basic Sciences: Genetics and cardiovascular diseases. In: Omokhodion SI, Ogunkunle OO and Olowu AO (eds): *Essentials of paediatric cardiology in the tropics*. 2021. (In preparation).
262. El-Sergani AM, Brandebura S, Padilla C, Butali A, **Adeyemo WL**, Valencia-Ramirez C, Restrepo-Muneton CP, Moreno LM, Buxo CJ, et al. Parents of children with non-syndromic orofacial clefting show altered palate shape. *Cleft Palate Craniofac J*. 2021; 58:847-853.

263. Gould D, **Seven M**, Nurse Practitioners' Competency in Genetics and Genomics Related Healthcare Services. *J Contin Educ Nurs*. 2021. (Submitted).
264. Guimarães A, Sukow N, Cipolla G, Mendes M, Leal T, Petzl-Erlar M, Rodrigues de Souza R, **Guio H**, Arboleda-Bustos C, et al. Tracing the distribution of European lactase persistence genotypes along the Americas. *Front Genet*. 2021. (Accepted).
265. **Hussen DF**. Chromosome Therapy: different approaches and current challenges. *Middle East J Med Genet*. 2021. (In review).
266. Jaramillo-Valverde L, Levano K, Loarte T, Tarazona A, Sanchez C, Solari L, **Guio H**. NAT2 and CYP2E1 polymorphisms and adverse drug reactions to first-line antituberculosis drugs in Peruvian patients. *Open Forum Infect Dis*. 2021. (Submitted).
267. Jaramillo-Valverde L, Levano K, Sanchez C, Solari L, **Guio H**. Allelic and genotypic frequencies of NAT2, CYP2E1 and AADAC genes in a cohort of Peruvian tuberculosis patients. *Mol Genet Genomic Med*. 2021. (Accepted).
268. Kannam M, Sutraye J, Kapoor R, **Tibrewal S**, Kekunnaya R, Sachdeva V. Horizontal rectus muscle transplantation for recurrent and residual strabismus. *Strabismus*. 2021 Jan; 20:1-9.
269. Kantarama E, **Uwineza A**, Muvunyi JC. The central obesity and its association with other cardiometabolic risk factors among women of childbearing age at selected family planning centers in Kigali-Rwanda. *Int J Womens Health*. 2021. (Submitted).
270. **Lallar M**, Bijarnia-Mahay S, Verma IC, Mandal K, Puri RD. Mutation and phenotypic spectrum of patients with RASopathies. *Indian Pediatr*. 2021; 58:30-33.
271. **Lallar M**, Kaur L, Preet M, Singh D, Singh UP. Spectrum of congenital cardiac defects over a period of three years. *J Fetal Med*. 2021. (In review).
272. Morales-Peralta E, Zúñiga-Rosales Y, Collazo-Mesa T, Santos-González EN, Hernández-Pérez Y, González-Torres MA, **Roblejo-Balbuena H**, Marcheco-Teruel B, Sotomayor-Lugo F. A16974C Polymorphism of the IL-12 p40 gene in Cuban patients having recovered from COVID-19. 2021. (In preparation).
273. Moreira R, Saraiva-Duarte JM, Costa Pereira A, Sosa-Macías M, Galaviz-Hernandez C, Santollala M, **Guio H**, O'Connor T, Tarazona-Santos E. Acute lymphoblastic leukemia: translating pharmacogenetics from us-Hispanics to their more neglected native American ancestors. *J Genet Genomics*. 2021. (Submitted).
274. Mukhopadhyay N, Feingold E, Moreno-Uribe L, Wehby G, Valencia-Ramirez LC, Muneton CR, Padilla C, Deleyiannis F, ...**Adeyemo WL**, et al. Genome-wide association study of non-syndromic orofacial clefts in a multiethnic sample of families and controls identifies novel regions. *Front Cell Dev Biol*. 2021; 9:621482. doi: 10.3389/fcell.2021.621482
275. Naslavsky M, Scliar M, Yamamoto G, **Guio H**, Zatz M. Whole-genome sequencing of 1,171 elderly admixed individuals from the largest Latin American metropolis (São Paulo, Brazil). *Nat Commun*. 2021. (Submitted).
276. Naslavsky S, Scliar M, Nunes, Wang G, Yamamoto G, **Guio H**, Tarazona-Santos E. Biased pathogenic assertions of loss of function variants challenge molecular diagnosis of admixed individuals. *Am J Med Genet C Semin*. 2021; 1-7.
277. Ndayisaba MC, Rugwizangoga B, Musoni E, Manirakiza F, **Uwineza A**, Tuyisenge L, Nyundo M, Hategekimana T, Ntakirutimana G. Cause of death assignment using minimally invasive tissue sampling in low resource settings: a cross-sectional study from the University Teaching Hospital of Kigali, Rwanda. *Egypt J Forensic Sci*. 2021. (Submitted).
278. Nembaware V, Mazandu KG, Hotchkiss J, Safari SJM, Kent J, **Uwineza A**, Makani J, Mulder N, Wonkam A, et al. The sickle cell disease ontology: Enabling collaborative research and co-

- designing of new planetary health applications. *Omics*. 2020 Oct; 24(10):559-567.
279. Ntalindwa T, Nduwingoma M, Karangwa E, Rashid Soron T, Uworwabayeho A, **Uwineza A**. Development of a mobile application to improve numeracy skills of children with autism spectrum disorders in Rwanda. *JMIR Pediatr Parent*. 2021. (Accepted).
 280. Obi AI, Osula EI, Richmond RE, Obarisiagbon E O, Adayonfo E, Obi RU, Okonofua F, **Okafor FU**. Determinants of attitude towards gender-based violence among adult residents in an urban community in a developing country. *Trop J Health Sci*. 2021 Apr; 28(1):19-24.
 281. Ogunkeyede SA, **Adeyemo AA**, Ogundoyin OA, Oyelakin OA, Fawole OB. Readiness of Nigerian health-care workers to work during COVID-19 pandemic. *Niger J Med*. 2021; 30:246-251.
 282. Olatosi OO, Li M, Alade AA, Oyapero A, Busch T, Pape J, Olotu J, Awotoye W, ...**Adeyemo WL**, et al. Replication of GWAS significant loci in a sub-Saharan African cohort with early childhood caries: a pilot study. *BMC Oral Health*. 2021; 21(1):274.
 283. Rath S, Varma DR, **Tibrewal S**, Ganesh S, Gupta N. A flower in the brain: Planum sphenoidale meningioma. *Indian J Ophthalmol*. 2020 Nov; 68(11):2514-2516.
 284. **Roblejo-Balbuena H**, **Benítez-Cordero Y**, Álvarez-Gavilán Y, Bravo-Ramírez M, Pereira-Roche N, García-Gómez D, Sotomayor-Lugo F, Monzón-Benítez G, Zúñiga-Rosales Y, et al. Clinical-epidemiological characteristics of patients affected by COVID-19 residing in Havana. *Cuban J Biomed Res*. 2021; 40(2):e1566.
 285. **Roblejo-Balbuena H**, Marcheco-Teruel B, Monzón-Benítez G, Marín-Padrón L. Allelic variants *3, *4, *5 and *6 of gene CYP2D6 in a sample of Cuban patients with schizophrenia. *Cuban Journal of Community Genetics*. 2020; 13(1).
 286. Rugwizangoga B, Ndayisaba MC, Musoni E, Manirakiza F, **Uwineza A**, Tuyisenge L, Nyundo M, Hategekimana T, Ntakirutimana G. Exploring perceptions and acceptance of minimally invasive tissue sampling among bereaved relatives and healthcare professionals in Rwanda. *Forensic Sci Med Pathol*. 2021. (Submitted).
 287. Scliar M, Naslavsky S, Yamamoto G, Norris S, **Guio H**, Tarazona-Santos E. 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. *Int J Obes*. 2021; 45:1017–1029.
 288. **Sirisena ND**, Samaranyake UMJE, Neto OLA, Foley AR, Pathirana BAPS, Neththikumara N, Paththinige CS, Rathnayake P, Donkervoort S, et al. A novel variant in the COL6A1 gene causing Ullrich congenital muscular dystrophy in a consanguineous family: a case report. *BMC Neurol*. 2021; 21(1):105.
 289. **Sirisena ND**. Genomic Education - Updates from Sri Lanka. *Global Genomic Medicine Collaborative (G2MC) Quarterly Newsletter*. Oct. 2020.
 290. SKehdy F, Pita-Oliveira M, Scudeler M, Torres-Loureiro S, Zolini C, Moreira R, Michelin L, Alvim L, ...**Guio H**, 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. *Genet Mol Biol*. 2021. (Accepted).
 291. Sokunbi OJ, Okoromah CAN, **Ekure EN**, Olawale OA, Eke WS. Electrocardiographic pattern of apparently healthy African adolescent athletes in Nigeria. *BMC Pediatr*. 2021 Feb; 21(1):97.
 292. Sotomayor-Lugo F, **Roblejo-Balbuena H**, Marcheco-Teruel B. Roles of tumor necrosis factor alpha -308A>G polymorphism on the clinical evolution of SARS-CoV-2 infection. 2021. (In preparation).

293. **Tibrewal S**, Rath S, Ganesh S, Sangwan VS. Lateral rectus muscle injury during double-headed pterygium surgery: A rare complication. *J Pediatr Ophthalmol Strabismus*. 2020 Dec 11; 57:e96-e99.
294. Zúñiga-Rosales Y, **Roblejo-Balbuena H**, Monzón-Benítez G, Sotomayor-Lugo F, **Benítez-Cordero Y**, Suárez- Besil B, González-Torres MA, Torres-Tives B, Álvarez-Gavilán Y, et al. Epidemiological, clinical and immunological characteristics of cases from the first Covid-19 outbreak in Cuba. *Bulletin of the World Health Organization*. 2021. (Submitted).
295. Zureigat B, Gould D, **Seven M**. Educational interventions to improve nurses' competency in genetics and genomics: A Scoping Review. *J Contin Educ Nurs*. 2021. (Submitted).

B. Grants: (Total -62; 2019-13; 2018- 22; 2017- 11; 2016- 16)

2019 Batch

1. Al Hajjar N (PI), Berindan Neagoe I, **Pîrlog R**. Increasing the capacity, competitiveness and visibility, research of UMF Iuliu Hațieganu by forming and supporting multidisciplinary teams in research excellence. 2021. (Iuliu Hațieganu University of Medicine and Pharmacy, Received).
2. Badea R (PI), Covaliu B, **Pîrlog R**. Academy of Research Entrepreneurs in Medicine. 2020. (Iuliu Hațieganu University of Medicine and Pharmacy Cluj-Napoca, Renewed).
3. Berindan Neagoe I (PI), **Pîrlog R** (Co-PI). Development and support of the medical entrepreneurial culture among UMF Cluj-Napoca students through dissemination, training and mentoring. 2021. (Iuliu Hațieganu University of Medicine and Pharmacy, Received).
4. Chianumba R (PI), **Alli LA**. Patterns of liver function parameters among patients living with sickle cell disease on hydroxyurea therapy. 2021. (Institutional Research Fund grant; Submitted).
5. **Gyawali P** (PI), Quinonez S. Medical genetics need assessment in Nepal using a mobile health application. 2021. (Global Genomic Medicine Collaborative Seed Award; In preparation).
6. **Hussein N** (PI), Lee YK, Ng CJ, Ramli R, John P, Jafry N. Developing research capacity to improve the implementation of thalassaemia screening in Sabah, Malaysia using the Stanford design thinking framework. 2021. (University Malaya Specialist Centre (UMSC) Care Fund; Submitted).
7. **Hussein N** (PI), Lee YK, Ng CJ, Ramli R, John P, Jafry N. Developing research capacity to improve the implementation of thalassaemia screening in Sabah, Malaysia using the Stanford design thinking framework. 2021. (University Malaya Specialist Centre (UMSC) Care Fund; Submitted).
8. **Iqbal M** (PI), **Naeem MA** (Co-PI), Ansar M (Co-PI). Identification and characterization of novel DNA variants causing autosomal recessive inherited retinal dystrophies in consanguineous Pakistani families. 2021. (National Research Program for Universities, Higher Education Commission of Pakistan; Submitted).
9. **Iqbal M** (PI). Genetic analysis of retinitis pigmentosa in consanguineous families from southern Punjab, Pakistan. 2021. (Research Grant, ORIC, The Islamia University of Bahawalpur Pakistan; Received).
10. 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. 2021. (Tertiary Education Trust Fund, Accepted).

11. Nyamjav D, Osgonbaatar T, Tumenbayar A, **Baatar N**, Delgerduuren M, Alimaa A. Primary care approach in Adolescents. 2021. (Health Promotion Fund, MOH, Mongolia; Received).
12. Oo MTZ (PI), **Khant AK**. Fractional excretion of magnesium in children with nephrotic syndrome. 2020. (Department of Medical Research, Lower Myanmar Yangon; Received).
13. Vilain E. (PI), Linguraru M. (Co-PI), Mumba (local PI), Tshala DK, Bramble MS, **Mosema KBA**, Spencer D. Mobile diagnosis of congenital genetic conditions: A model for screening and surveillance in low-resource settings. 2021. (Children's National Medical Center, Institut National de Recherche Biomédicale; Received).

2018 Batch

14. **Adewole OO** (PI). Finding people with tuberculosis through strategic deployment of a non-sputum based (Sweat TB Test) in Nigeria. Grant challenges Africa Phase II. 2021. (African Academy of Science; Received).
15. **Adewole OO** (PI), Arun V(Co-PI). Statins as treatment adjunct in active pulmonary tuberculosis. 2020. (Cure Within Reach; Received).
16. Afolabi BB (PI), Galadanci H (Co-PI, Balogun M (Co-PI, **Adeyemo TA** (Co-PI), Sam-Agudu N (Co-PI). Intravenous versus oral iron for iron deficiency anemia in pregnant Nigerian women (IVON): an open label, randomized controlled trial. Clinical Research Protocol. 2020-2023. (Bill and Melinda Gates Foundation; Received).
17. Alatise O (PI), Kingham P (Co-PI), **Abdulkareem F** (Co-I). Nigerian cancer research training programme. 2021. (OAK Foundation, Switzerland; Received)
18. **Dutta AK** (PI). Estimation of birth prevalence and carrier frequencies of autosomal recessive inborn errors of metabolisms in South Asia. 2021. (Science and Engineering Research Board, Ministry of Science and Technology. Government of India; Start-up Research Grant; Submitted).
19. **Gitaka J** (PI), Suliman S, Arlehamn CL, Asiko O. Impact of SARS-CoV-2 infection on reactivation of latent Mtb infections in a Kenyan Cohort. 2021. (NIH; Submitted).
20. Jaja C (PI), **Edem-Hotah J** (Co- PI), Shepherd J. Integrating nurse champion model for group reproductive genetic counselling for sickle cell hemoglobinopathies into primary care: A pilot implementation science study. 2021. (NIH; Accepted).
21. 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; Continued).
22. Karina Milla-Neyra et al. **Cornejo-Olivas M** (co-investigator). Redefining the genetics of spinocerebellar ataxia type 10 in admixed and Amerindian population from Peru. 2020. (FONDECYT/CONCYTEC Peru; Received).
23. Kingham P (PI), Alatise O(Co-PI) **Abdulkareem FB** (Co-I). The efficacy and feasibility of fecal immunochemistry for colorectal cancer screening in Nigeria. 2020. (Prevent Cancer Foundation; Received).
24. Kingham P (PI), Olusegun a (Co-PI), Abdulkareem FB (Site-PI). Expanding cancer research capacity in Nigeria with Team Science. 2021. (NIH-D43; Submitted).
25. Kingham P(PI), Alatise O (Co-PI), Du (Co-PI), **Abdulkareem FB** (Co-I). Determining the risk factor profile and biology of colorectal cancer in Nigeria. 2020. (NIH-R01; Received).
26. Pericak-Vance M, **Cornejo Olivas M** (site PI). Genomic characterization of Alzheimer disease risk in admixed populations with native American and southern European genetic ancestry. 2021. (NIH R01 AG070864-0; Accepted).

27. Mehmood A (PI) **Naeem MA** (Co PI). Exploitation and development of stem cells and exosomes based replacements for treatment of damaged organs. 2021. (Grand Challenge Fund (GCF) of Higher Education Commission, Islamabad, Pakistan; Submitted).
28. **Messaoud O** (PI), Members and Alumni of working group Health and Society. Towards effective solutions for combating COVID-19: An interdisciplinary approach-part II. 2021. (AGYA/German Federal Ministry of Education and Research Fund; Accepted).
29. **Messaoud O** (PI), Rizk A (Co-PI), Abdel-Hafiez M, and Mirsky V, Bardaoui A, Hammami A, Chtourou R. Electrochemical biosensors for controlled sensing of biomolecules receptors. 2021. (AGYA/German Federal Ministry of Education and Research Fund; Accepted).
30. **Muttamba W** (PI), Kirenga B, Sabiiti W. A health systems analysis and modelling of methods to maximize uptake and utilization of advanced diagnostics in low- and middle-income countries- The case of COVID-19. 2020. (GCRF; Received).
31. **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. (Funded by Saval Laboratories; Continued).
32. Olopade OI (PI), Odunsi AO (Co-PI), Ojengbede O, Adejumo PO, **Abdulkareem FB** (Member-TAC. Global Scholars in Oncology Associated Research (Global-SOAR) training program. 2021. (NIH-D43; Submitted)
33. Olopade OI(PI), Ntekim A, Popoola AO, Arowolo OA, Sowunmi A, Olopade CS, Ibraheem A, **Abdulkareem FB** (Project Site Director), Daramola OA, et al. Assessing REsponse to neoadjuvant Taxotere and TrAstuzumab in Nigerian women with HER2-positive breast cancer (ARETTA), a multicenter study. 2019. (Universities of Lagos, Nigeria; University of Ibadan, Nigeria; Obafemi Awolowo University, Nigeria; Ile-Ife and Lagos State University, Nigeria; University of Chicago, USA; Continued).
34. Sabiiti W, **Muttamba W**, Woods C, Wolfgang P, Mmbaga B. Increasing capacity for biobanking and Whole Genome Sequencing (WGS) in low- and middle- income countries: The EES Project. 2021. (In preparation).
35. Wei JJ, **Abdulkareem FB** (Co-I), Silas Olugbenga (Co-I). Develop a telepathology program in promoting cervical cancer research and training in Nigeria. 2020. (Global Health Institute, Northwestern University; Continued).

2017 Batch

36. **Abad PJB**, Reyes ME, Sur ALD, Ngaya-an FV. Determinants of parental adherence to short-term follow-up in newborn screening. 2021. (Newborn Screening Reference Center, University of the Philippines Manila; Submitted).
37. **Abad PJB**. G2CN: Genetics and genomics in cancer nursing. 2021. (Education Grant 2021-International Society of Nurses in Genetics; Submitted).
38. Calalo CM, Cases RKC, Asor BBN, **Abad PJB**, Asuncion PC. A content analysis of Facebook groups on congenital adrenal hyperplasia. 2021. (Funded by the Newborn Screening Reference Center – University of the Philippines Manila; Under ethics review)
39. Guntekin Ergun S, **Yaylacioglu Tuncay F**, Talim B, Dincer P. Examination of ocular developmental pathologies by creating an Lrp5 mutant zebrafish model using CRISPR/Cas9. 2021. (TUBITAK 1002 Grant; Received).

40. Makani J (PI), **Mgasa A** (Co-PI), Luoga F, Lloyd W, Marco E, Konteh F, Chuwa H, Rifai A, Luzzatto L. Manual exchange transfusion project for sickle cell patients. 2021. (Funded by French Embassy in Tanzania; Received).
41. Mochly-Rosen D (PI), Manasa J (PI), **Dhoro M** (Co-PI), Masimirembwa C (Co-PI). Improving translational research in Zimbabwe with a focus on infectious diseases. 2021 (National Institutes of Health Global Infectious Disease Research Training Program; In preparation).
42. (Stanford University USA, University of Zimbabwe, African Institute of Biomedical Science & Technology, Zimbabwe; In preparation).
43. **Owusu M** (PI), Sambian D, Godfred A. Enhancing global health security: Expanding efforts and strategies to protect, diagnose and improve public health in Ghana. 2020. (CDC Fund through the Centre for Health System Strengthening; Accepted).
44. **Petlichkovski A**, Arsov T, Kirijas M, Brnjarchevska T, Milanovski G, Sibinovska O, Trajkov D, Efinska-Mladenovska O, Dobrevski B, et al. Immunogenetic profile of patients with severe form of COVID-19. 2021. (Ministry of Education and Science of Republic of North Macedonia; Received).
45. **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- extension applied for due to Covid).
46. **Torres-Mejía G** (PI), Rinaldi S, Ángeles-Llerenas A, Gómez Flores-Ramos L. Confluence uncovering breast cancer genetics research proposal. 2021. (National Institute of Public Health, Mexico; Submitted).

2016 Batch

47. Bozaykut Eker P (PI), Park T (PI), **Deniz E** (Consultant). Cardiac analysis on the superior hypoxia-tolerance through in vivo and in vitro approaches. 2021. (Joint grant from TUBITAK, Turkey and NFS, USA; Received).
48. Toklu Keskin N (PI), **Deniz E** (Consultant). Comparison of hot spot mutant p53 proteins' effects on glioblastoma. 2021. (TUBITAK, Turkey; Received).
49. **Seven M** (PI), Moratitis AM, Reid A, Sturgeon S, Pearlman J, Wenzel J, Hammer M. Factors influencing disparities in quality of life among people of color affected by cancer: A mixed-method study. 2021. (Oncology Nursing Foundation General Research Grant; Submitted).
50. Butali A (PI), **Adeyemo WL** (Co-investigator), **Eshete MA**. Refining the genetic and genomic architecture of non-syndromic orofacial clefts. 2020-2025. (NIH/NIDCR; Continued).
51. Hou L (PI), Sagay A (PI), Ogunsola FT, Akanmu AS, Murphy M (PI), **Adeyemo WL** (Co-I), **Abdulkareem FB** (Co-I). Epigenomic biomarkers of HIV-associated cancers in Nigeria. 2017-2022. (U54CA221205, NCI/NIH; Continued).
52. Hou, L, Sagay A, Ogunsola FT, Akanmu AS, Murphy M (PI), **Adeyemo WL** (Co-I), **Abdulkareem FB**. Nigeria research training grant for HIV and malignancies. 2019-2024. (D43TW009575-06, Northwestern University, USA; Continued).
53. Butali A, (PI) **Adeyemo WL** (Co-I). Orofacial clefts, whole genome sequencing and incidental findings: Ethical considerations. 2020-2025. (NIH/NIDCR; Received).
54. Babalola OE, (PI) **Adeyemo WL** (Co-I). A double blind randomized clinical trial assessing the efficacy and safety of ivermectin in COVID-19 (IVERCOVID STUDY). 2020-2021. (Central Bank of Nigeria; Received).

55. Salako B, (PI), Rotimi MK, **Adeyemo WL** (Co-I). Feasibility and efficacy of implementing a CPAP/oxygen helmet solution for non-invasive ventilation of COVID-19 patients in Nigeria. 2020-2021. (NMG-SA-VENT-06; Received).
56. Ntakirutimana G (PI), Masaisa F (Co-PI), Mukandekazi A (Co-PI). Uwizeyimana G (Co-PI). Rugwizangoga B(Co-PI) and **Uwineza A** (Co-PI). Assessment of molecular diagnosis and response of Chronic Myeloid Leukaemia (CML) using reverse transcriptase- polymerase chain reaction at CHUK. 2021. (University Teaching Hospital of Kigali (CHUK), Directorate of Research; Received).
57. **Guio H** (PI). Metagenomic analysis and antimicrobial resistance in urban and hospital's wastewater of Huánuco. 2021. (Universidad de Huánuco; Continued).
58. **Guio H** (PI). Association between ACE2 and TMPRSS2 genes polymorphisms and COVID-19 diagnosis. 2021. (Universidad de Huánuco; Submitted).
59. Gomez A (PI), **Guio H** (Co-I). Assessing the relationship between gut microbiome and intestinal function in diverse Peruvian populations affected with COVID-19. 2021. (University of Minnesota, Twin Cities, USA; Submitted).
60. **Adeyemo AA** (PI). Auditory evaluation of stroke survivors: validating a low-cost screening protocol. 2020. (UKRI/ARUA Centre of Excellence Mini Grants; Received).
61. **Ariani Y (PI)**, Yuridian R (Co-PI). The role of variant of COL1A1 gene in 7 major ethnic groups in Indonesia towards wrinkle appearance. 2021. (Paragon Technology and Innovation Corp. Indonesia; Accepted).
62. **Ariani Y (PI)**, Bawolaksono (Co-PI). Melasma and it's correlation to variant of MC1R gene in Indonesian women population. 2021. (Paragon Technology and Innovation corp. Indonesia; Accepted).

C. Collaborations (NIH and Other Institutions):

NIH: (Total- 10; 2019- 3; 2018- 4; 2017- 1; 2016- 2)

2019 Batch

1. **LLamos-Paneque A**, Gahl WA, Adams D, (Undiagnosed Diseases Network International (UDNI). Data Sharing Project. (Specialty Hospital No.1 FF. AA, Quito- Medical Genetics Branch; NHGRI/NIH).
2. **LLamos-Paneque A**, Stephen G, Tiffit C. ICORD's new initiative: Rare diseases in the Caribbean and Latin America (ERCAL). (Specialty Hospital No.1 FF. AA, Ecuador- ORDR; NHGRI/NIH).
3. **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. 2020. (Bayero University Kano/Aminu Kano Teaching Hospital; NHGRI/NIH). (Modification of workbook).

2018 Batch

4. Pasechnikova N, Zborovska O, **Savina O**. Molecular and clinical research in Ukrainian families with the inherited eye diseases. (V. Filatov Institute of Eye Diseases and Tissue Therapy; NEI/NIH).
5. Singleton A, Blauwendraat C, Brice A, Casey B, Fiske B, Bandres S, Mata IF, **Cornejo-Olivas M**, Rivera-Valdivia A, Sarapura -Castro E, et al. GP2 Global Parkinson genetics program aiming

to further understand the genetic architecture of Parkinson disease through genotyping diverse patient groups. (NIA/NIH; Cleveland Clinic, USA; Instituto Nacional de Ciencias Neurológicas, Peru).

6. Gloft S, Reichardt J, Posada M, Taruscio D, Repetto G, Giuliani R, **Dueñas-Roque M**, Bonilla C, **Cornejo-Olivas M**. Bridging the gaps in rare diseases and orphan products in Latin America and the Caribbean nations and territories (ERCAL). (NCATS-NIH; Undiagnosed Diseases Network International; Hospital Nacional Rebagliati Martins, Instituto Nacional de Ciencias Neurológicas, Peru).
7. Riazuddin S, **Naeem MA**, Hejtmancik JF, Gottsch JD, Riazuddin SA, Khan SY, Ali M, Qazi ZA, Butt NH, et al. Hereditary vision impairment in Pakistan (CEMB, University of the Punjab, Lahore-Pakistan; NEI/NIH; Wilmer Eye Institute, Johns Hopkins University, USA; Layton Rehmatulla Benevolent Trust Eye Hospital, Lahore; Allama Iqbal Medical College, University of Health Sciences, Lahore, Pakistan).

2017 Batch

8. **Yaylacioglu Tuncay F**, Hufnagel R. Investigation of the role of LRP5 mutations in hereditary eye diseases: EyeGene cohort and zebrafish model. (Gulhane Medical Faculty, Ankara; ICO-NEI/NIH Ocular Genetics Fellowship Program).

2016 Batch

9. **Ekure E**, Kruszka P, **Adeyemo A**. Atlas of human malformation syndromes in diverse populations. (University of Lagos/Lagos University Teaching Hospital, Nigeria; NHGRI/NIH).
10. **Ekure E**, Adeyemo A. Genetics of congenital heart diseases in Africa. (University of Lagos/Lagos University Teaching Hospital, Nigeria; NHGRI/NIH).

Other Institutions: (Total – 54; 2019- 16; 2018- 22; 2017- 9; 2016- 7)

2019 Batch

1. **Abubakar S**. HIV and AIDS Strategic Plan- Kebbi State, Nigeria (2021-2025). Technical Report. National Action Committee on AIDS & German Federal Ministry for Economic Cooperation and Development- GIZ BACKUP Project.
2. **Abubakar S**. HIV and AIDS strategic plan- Sokoto State, Nigeria (2021-2025). Technical Report. (National Action Committee on AIDS & German Federal Ministry for Economic Cooperation and Development- GIZ BACKUP Project).
3. Akay G, Posey JE, **Llamos-Paneque A**. Genome sequencing to elucidate causes and mechanisms of mendelian genetic disorders. (Baylor College of Medicine, USA; Genetics Service of Hospital No.1 FF. AA. Quito-Ecuador).
4. **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 new research (Centre Hospitalier Universitaire du Point "G", Bamako, Mali; Centre Hospitalier Universitaire Gabriel Touré, Bamako, Mali; UCL Queen Square Institute of Neurology, London, UK).
5. **Chowdhury EH**, **Amarakoon GGGT**, **Mosema KBA**, **Khant AK**. Associations/complications with Trisomy 21 in children. (Samtse Genral Hospital, Bhutan; Faculty of Medicine and Allied Sciences, Rajarata University of Sri Lanka, Sri Lanka; Instut National de Recherche Biomedical, Biamamarie Mutombo Hospital, Kinshasa, DR Congo; Yangon Children Hospital, Myanmar).

6. **Cornejo-Olivas M, Llamas-Paneque, A.** Collaboration in technical support regarding molecular diagnosis of neurogenetics diseases like spinocerebellar ataxias, Huntington disease, steinert Disease. (Neurogenetics Research Center in Perú; Genetics Service of Hospital No.1 FF. AA. Quito, Ecuador).
7. Fejerman L, Dominguez-Valentin M, Vianna-Jorge R, Agalliu I, **Paredes-Moscossa SR**, Carvallo P, Weitzel J, Bertoni B. The Latin American Genetics and Genomics of Breast Cancer Consortium (LAGENO-BC). (Universidad de San Martín de Porres; LAGENO-BC).
8. **Hussein N**, Quinonez SC. Introducing MiGene Family History App in Malaysia. (Family History Working Group, University of Malaya, Malaysia; Global Genomic Medicine Collaborative).
9. **Iqbal M**, Ahmed ZM. Analyses of DNA variants causing otitis media in Pakistani population. (Department of Biotechnology, The Islamia University of Bahawalpur, Pakistan; University of Maryland, School of Medicine, USA).
10. **Iqbal M**, Ansar M. Mutational analysis of DNA from consanguineous Pakistani families having ophthalmological disorders through whole exome sequencing (Department of Biotechnology, The Islamia University of Bahawalpur, Pakistan; Institute of Molecular and Clinical Ophthalmology Basel (IOB) Basel, Switzerland).
11. **Iqbal M, Naeem MA.** Identification, clinical evaluation, and genetic analysis of RP families (Department of Biotechnology, The Islamia University of Bahawalpur, Pakistan; National Centre of Excellence in Molecular Biology, University of the Punjab, Lahore, Pakistan).
12. **López-Star B, Llamas Paneque A.** Consultation on ophthalmogenetic case. (Mexican Society of Ophthalmology; Mexican Institute of Ophthalmology; Specialty Hospital No.1 FF. AA, Quito, Ecuador).
13. **Nair LS**, Nair MKC, Kumar S, Lukose R. Genetic evaluation of children with Intellectual disability and autistic spectrum disorders. (Department of Medical Genetics, NIMS Medicity; CDRC Spectrum, NIMS Medicity, Trivandrum, Kerala, India).
14. Olorunniji F (PI), **Alli LA**, Adeoye R. Development of a platform for rapid on-site testing of novel viral infections in Nigeria based on CRISPR, isothermal PCR amplification, and split G-quadruplex DNazyme technologies. (GCRF, Liverpool John Moores University, UK; University of Abuja, Nigeria; University of Ilorin, Nigeria).
15. 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).
16. Sidharth KS, Rupesh R, Divya S, Sadaf A, **Khant AK**, Manoji M, Kalaivani G, Shraddha L, Rajiv S, et al. Assessment of south Asian Paediatric Acute kidney injury Epidemiology and Risk Factors (ASPIRE): Design of prospective study on severe paediatric AKI. (SAARC nations with India, Myanmar, Bangladesh and Pakistan).

2018 Batch

17. Alatisse OI, Kingham TP, **Abdulkareem FB.** Colorectal cancer database in Nigeria. (Obafemi Awolowo University, Nigeria; University of Lagos, Nigeria; Memorial Sloan Kettering Cancer Center, USA).
18. **Cornejo-Olivas M, Dutta AK, Dueñas M, Dwivedi A, Mistri M.** Chapter on ‘Nutrigenomics’, Springer Nature book on Biochemical aspects of nutrients (micro and macro) and nutrients and

- therapeutic potential of nutrients along with nutrigenomics. (National Institute of Neurological Sciences, Peru; All India Institute of Medical Sciences Kalyani, India; Hospital Edgardo Rebagliati Martins, Peru, Army Hospital Research and Referral (AHRR), India, FRIGE's Institute of Human Genetics, India).
19. **Dutta AK**, Lemke J, Paul L, Riggs ER. ClinGen GRIN Variant Curation Expert Panel (VCEP). (AIIMS Kalyani, India; Institute of Human Genetics, Leipzig University Hospital, Germany; Geisinger Health System, Pennsylvania, USA).
 20. **Dwivedi A**, Kumar R, Singh AK, Dey M. To study the clinical utility and diagnostic yield of exome sequencing in prenatal diagnosis of congenital anomalies (AHRR, Delhi & Base Hospital Delhi Cantt, India).
 21. **Gitaka J**, Clark T. Molecular epidemiology of Plasmodium falciparum. (London School of Hygiene and Tropical Medicine, UK; Mount Kenya University, University of Nairobi).
 22. Jaja C, **Edem-Hotah J**. The insights into microbiome and environmental contributions to sickle cell disease and leg ulcers (The INSIGHTS Study). (University of Sierra Leone College of Medicine and Allied Health Sciences, Seirra Leone; University of South Carolina, USA).
 23. Kirenga B, **Muttamba W**, Kitaka S, Turyahabwe S. Strategies for resolving the gap in Adolescent TB care in Uganda: A pilot study (Design without Borders, Uganda; Makerere University Lung Institute, Uganda).
 24. Lopez-Cendes I, **Dueñas-Roque M**. Whole exome sequencing for identification of pathogenic genetics variants in early encephalopathy in infancy in Latin America. (Universidad Estadual de Campinas, Brasil; Hospital Nacional Edgardo Rebagliati Martins, Perú).
 25. Luquetti D, **Dueñas-Roque M**. Phenotypic and genomic characterization of microtia in the Andean Population. (Washington University, USA; Seattle Children's Hospital, USA; Hospital Nacional Edgardo Rebagliati Martins, Perú).
 26. Mata IF, Inca-Martinez M, **Cornejo-Olivas M**, Cubas-Montecnio D, Manrique-Enciso C, Milla-Neyra K. Implementation of the DNA Bank Neurogenetics in Peru. (LARGE PD Consotium; GP2 and ASAP initiative; Michael J. Fox Foundation, USA; San Marcos Foundation and Instituto Nacional de Ciencias Neurologicas, Peru).
 27. **Messaoud O**, **Dutta AK**, **Cornejo-Olivas MR**, Bhuiyan ZA, Banerjee K. Editing a special issue 'Monogenic vs Oligogenic Reclassification' <https://www.frontiersin.org/research-topics/13904/monogenic-vs-oligogenic-reclassification> in [Frontiers in Genetics](https://www.frontiersin.org/journals/genetics) (<https://www.frontiersin.org/journals/genetics>) (Institut Pasteur de Tunis, Tunisia; All India Institute of Medical Sciences, Kalyani, India; National Institute of Neurological Sciences, Lima, Peru; Centre Hospitalier Universitaire Vaudois, Lausanne, Switzerland; Institute White Plains, United States).
 28. **Messaoud O**, Bannerjee K, **Cornejo-Olivas M**, **Dutta AK**, Bhuiyan ZA. Special article series on 'Monogenic vs Oligogenic Reclassification' in Frontiers in Genetics/ Frontiers in Pediatrics (<https://www.frontiersin.org/research-topics/13904/monogenic-vs-oligogenic-reclassification>). (Institut Pasteur de Tunis, Tunisia; Weill Cornell Medicine, USA; National Institute of Neurological Sciences, Peru; All India Institute of Medical Sciences Kalyani, India; Centre Hospitalier Universitaire Vaudois (CHUV) Lausanne, Switzerland).
 29. **Messaoud O**, Haddad S, Charfi-Kaddour S, Gargouri M, Drissi LB, Hammami A, Bardaoui A. The use of advanced materials for molecular biology applications. (Institut Pasteur de Tunis, Tunisia; Faculty of Sciences of Tunis, Tunisia; Faculty of Sciences of Bizerte, Tunisia; Borj Cedria technopole, Tunisia; Faculty of Sciences, Rabat, Morocco).

30. **Messaoud O**, Khalil A, Nasr M, Azzab S, Sunoqrot S, Amro A. Screening of nanoparticles, biomaterials, natural extracts and disinfectants on coronavirus strain using several antiviral tests. (Institut Pasteur de Tunis, Tunisia; Egypt Japan University of Science and Technology (EJUST), Egypt; The Public Authority Ain Shams University, Egypt; Al-Zaytoonah University of Jordan, Jordan; Al-Quds University, Palestine)
31. **Naeem MA**, Ali M, Butt H, Qazi ZA, Mehmood A, Shahid AA, Riazuddin S. Clinical study on mesenchymal stem cell modality for management of Retinitis Pigmentosa in patients. (LRBT Hospital; Stem Cell Labs at Jinnah Burn & Reconstructive Surgery Center, Jinnah Hospital Complex, Lahore; National Centre of Excellence in Molecular Biology, University of the Punjab Lahore, Pakistan).
32. **Nakousi-Capurro N**, Cavalcanti DP. Description of genotype-phenotype relations in patients with osteogenesis imperfecta in a Brazilian Cohort. (Clinical Genetics Service, Carlos Van Buren of Valparaíso, Chile; Department of Clinical Genetics, Faculty of Medical Sciences at University of Campinas, Sao Paulo, Brazil).
33. Omisore AD, Sutton EJ, Omidiji O, **Abdulkareem FB**, Daramola AO. Tablet-based mobile health ultrasound for point-of-care breast cancer diagnosis in Nigeria. (Obafemi Awolowo University, Nigeria; University of Lagos, Nigeria; Memorial Sloan Kettering Cancer Center, USA).
34. 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).
35. Pasechnikova N, Zborovska O, **Savina O**, Kremenska U. Genetic diagnostics; medical and genetic consultation for families with retinoblastoma in an ISIDA-IVF genetical laboratory with a view to implementing pre-implantation diagnostics during the period of planning for the next pregnancy in the said families. (V. Filatov Institute of eye diseases and tissue therapy; ISIDA-IVF clinic, Ukraine).
36. **Savina O**, Rykov O, Kremenska U. Genetic testing; medical and genetic consultation for families with Aniridia and WAGR syndrome - implement pre-implantation diagnostics during family planning. (Department of Ophthalmology Shupyk's National Medical Academy of Postgraduate Education; ISIDA-IVF clinic; NGO - Aniridia. WAGR, Kyiv ophthalmological clinic, Ukraine; In discussion).
37. The COVID-19 Host Genetics Initiative (**Dutta AK**). A consortium of over 2000 scientists from over 54 countries. (<https://www.covid19hg.org/>).
38. **Utumatwishima JN**, Birungi F, Twagirayezu M. Leveraging artificial intelligence and data science techniques in harmonizing, accessing and analyzing SARS-COV-2/COVID-19 data in Rwanda. (LAISDAR Study). (IDRC, Canada; University of Rwanda, Rwanda).

2017 Batch

39. **Avogbe PH** (PI), Le Calvez-Kelm F, and McKay JD. Circulating tumor DNA biomarkers and early detection of hepatocellular carcinoma- sample collection ongoing. (University of Abomey-Calavi, Benin; International Agency for Research on Cancer/WHO).
40. **Casado PL**, Patrick Shmidlin. Frontier in Dental Medicine: Biomarkers in periodontology and peri-implant diseases. (Fluminense Federal University, Brazil; University of Zurich, Switzerland).

41. Finger PT, Tomar A, Kivela T, Krema H, **Yousef YA**. Treatment for radiation retinopathy, for patients with choroidal melanoma treated by radioactive plaque therapy. (King Hussein Cancer Center, Jordan; International multicenter study with New York Eye Cancer Center, USA).
42. **Mgasa A, Ottaru S**, Lloyd W, Luoga F, Chuwa H, Konteh F, Makani J, Luzzato L. Development of protocol for manual red cell exchange in patients with sickle cell disease in Tanzania. (National Blood Transfusion Services; Muhimbili University of Health and Allied Science; Aga Khan Hospital; Hindu Mandal Hospital- Mwanza, Tanzania).
43. **Petlichkovski A**, Sukarova-Angelovska E, Brusco A, Rubeis SD. Whole-exome sequencing of proband families with neurodevelopmental disorders. (Institute for Immunobiology and Human Genetics, Medical Faculty Skopje, Macedonia; Pediatric University Clinic, Medical Faculty Skopje, Macedonia, University of Turin, Italy; Icahn School of Medicine at Mount Sinai, New York, USA).
44. **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, Medical Faculty Skopje, Pediatric University Clinic, Medical Faculty Skopje, Macedonia.)
45. **Roy S**, Osmani M, Geary A, Dean W. Fundamental virtual reality simulation for manual small incision cataract surgery validity: Evaluation, efficacy and acceptability study in Bangladesh, China, Ethiopia, India, Mongolia, Togo, United Kingdom and United States of America. (Chittagong Eye Infirmary, Bangladesh; London School of Hygiene, UK; 13 centers from 8 countries).
46. Schneider T, Makani J, Delalandre C, **Mgasa A**, Lyimo M, Juma A, Luoga F, Chuwa H, Konteh F et al. Advanced therapy for sickle cell disease in Tanzania. (National Blood Transfusion Services, Muhimbili University of Health and Allied Science, Tanzania; Aga Khan Hospital, Tanzania; Etablissement Français du Sang –France).
47. **Thakur N**, Mayo C, Matt BJ. WHO Collective Global Network for Rare Disease (WHO CGN4RD).

2016 Batch

48. **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).
49. **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, Kano, Nigeria; McMaster University, Canada).
50. **Ariani Y**, Beben B, Khairunnisa S. Genomic variations of hair aging in 7 major ethnic groups in Indonesia. (Indonesian Medical Education and Research Institute (IMERI) - University of Indonesia; Biostatistics Department - University of South Australia; Paragon Technology and Innovation Corp. Indonesia).
51. **Guio H**, Bogi E. To develop personalized medicine in Latino America (INBIOMEDIC, Peru; Copenhagen Institute for future studies, Denmark).
52. Morales P, **Guio H**. Human Health Evolution in Latino America. (Universidad Tecnica de Marabi, Ecuador; INBIOMEDIC, Peru).
53. **Okafor FU**. Introduction of genetics and genomics in the nursing curriculum (The University of Benin; Benson Idahosa University, Nigeria; In discussion).

54. Okuyemi K, Ogunsola FT, **Adeyemo WL**. Collaboration for strengthening cancer population sciences research capacity in sub-Saharan Africa. 2021. (University of Utah, USA; University of Lagos, Nigeria).

D. New Research Projects: (Total- 46; 2019- 17; 2018- 8; 2017- 10; 2016- 11)

2019 Batch

1. **Amarakoon GGGT**, Wijayarathne HK. Awareness on existing neonatal screening among mothers in a tertiary care center-Sri Lanka.
2. **Amarakoon GGGT**, Wijayarathne HK. Study on congenital malformation detecting with anomalous scan during antenatal period in a tertiary care center-Sri Lanka.
3. Chaudhry A, **Bocoum A**, Rizig M. Identifying gaps in knowledge about Parkinson's disease amongst medical professionals in Africa.
4. **Chowdhury EH** (PI), Lhendup K, Zangmo Y, Tobgay S, Phuntsho N. Incidence and prevalence of Down Syndrome in Bhutan.
5. Dysted M, **Gyawali P**, Shrestha A, Gyawali B. Metabolic syndrome and insulin resistance among sub-urban adolescents with and without a family history of diabetes.
6. Foo JS, Ishak IH, **Hussein N**. The views and experiences of primary care doctors in managing children with learning disabilities.
7. Hoe HK, Sherina NS, **Hussein N**, Malik TF. Exploring the views and experiences of parents on school-based thalassemia screening in Klang district, Malaysia.
8. **LLamos-Paneque A**. Andean Duchenne Muscular Dystrophy Registry Initiative, coordinated by PTC Therapeutics.
9. Maceda EB, Abadingo M, **Tumulak MJR**, Cases RK, Asor BB. Social media content analysis of public Glucose-6-Phosphate deficiency (G6PD) Facebook Groups.
10. Malik TF, Ng CJ, Hanafi NS, Chiew TK, **Hussein N**, Hadi H. Covid 19: Developing, evaluating and implementing a teleconsultation system to follow-up patients with Type 2 Diabetes Mellitus during the Covid-19 Pandemic.
11. **Mhandire K**, Buxbaum NP. T-cell metabolism in graft-versus-host disease.
12. **Monye HI**, Kyari F, Ugalahi MO. Ophthalmologists' knowledge, perceptions, and practice patterns of ophthalmic genetics services in Nigeria (Submitted for ethical review).
13. Ng WL (PI), **Hussein N** (Co-PI), Lee YK, Ng CJ, Kee BP, Malik TFA, Qureshi N, Then SM, Kwan Z. HLA-b*58:01 Allele testing in primary care to prevent allopurinol induced-severe cutaneous adverse reaction (SCAR).
14. 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.
15. Sondhi V, Mishra P, Sridhar G, Singhal P, **Mutreja D**. DBT- NIDAN Kendra for Genetic Center for neonatal and prenatal screening. (Proposed).
16. **Wiafe SA**, Anyane-Yeboah K, Wiafe AA, Addo-Lartey E, Baynam G. Predicting diagnosis of congenital anomalies and rare diseases using artificial intelligence: A pilot of the <https://dx29.ai/> platform.
17. Win KPP, **Khant AK**. Thyroid hormone status and growth parameter in children with different stages of chronic kidney disease.

2018 Batch

18. Anorue EI, Gbotolorun MO, **Oluwarotimi AC**, Arotiba GT. Health related quality of life of patients presenting with orofacial Hard tissue injuries at Lagos University Teaching Hospital, Idi-araba Lagos. A prospective longitudinal study.
19. Castro Suarez S, **Cornejo-Olivas M**. Early identification of frontotemporal dementia in underserved settings through the LimaFTD network.
20. **Cornejo-Olivas M**. Closing the diagnostic gap in inherited neuropathies and related disorders.
21. **Dutta AK**, Goswami K, Sahoo S, Pal A. Early clinical exposure to inborn errors of metabolism through cinemeducation. (FAIMER educational project).
22. Kirenga B, Byakika P, **Muttamba W**, Namakula O, Kayongo A, Mugenyi L, Nakwagala F, Wayengera M, Kyobe H. COVID-19 Vaccine impact study.
23. Kirenga B, **Muttamba W**, Namakula O, Byanyima R, Nsereko C, Kisuule I, Mwesigwa E, Sekibira R. Data Quality Improvement project in COVID-19 treatment units in Uganda.
24. **Nakousi-Capurro N**. Clinical utility of NGS-based technologies: Experience in a public care service. (In discussion).
25. **Utumatwishima JN**, Nkeshimana M, Mutimura E. Associations of SARS-CoV-2 with CVD risks in Rwandan adults admitted at Tertiary Hospital, Kigali.

2017 Batch

26. **Casado PL**, Neves F, Aguiar T. Prevalence of periodontopathogens, Streptococcus pneumoniae and SARS-CoV-2 infection in patients under intensive therapy.
27. **Fatima SS** (supervisor). Methylation status of promoter region of FTO, POMC and IGF genes in familial Type 2 Diabetes.
28. **James O**, Clinical and demographic presentation of multiplex cleft families at Lagos University Teaching Hospital (LUTH) from 2016 -2021.
29. **Mahfoudh W**, Faleh R. Microbiome analysis in breast and ovarian cancers.
30. **Owusu M**, Boamah J, Ayisi-Boateng NK. Evaluating rapid diagnostic test kits for SARS CoV-2 detection.
31. **Owusu M**, Sylverken AA, Phillips R. Antibody responses to COVID-19 vaccines in Ghana.
32. **Roy S**, Hoque F. Cosmetic outcome of congenital ptosis treated with prang suture and without prang suture.
33. **Roy S**, Hoque F. Demographic profile, presentation and treatment outcome of ocular surface neoplasia treated in a tertiary eye hospital in Bangladesh.
34. Tosheska-Trajkovska K, **Petlichkovski A**. Dépistage sérologique de COVID-19 “chez les travailleurs de la santé en République de Macédoine du Nord“, envoyé par l'Agence Universitaire de la Francophonie le 10 avril 2020. (Serologic screening for COVID-19 in health-care workers from Republic of North Macedonia).
35. Yüksel M, Kayhan G, **Yaylacioglu Tuncay F**, Ergun MA, Ozdek Ş. Genotype and phenotype characteristics of Stargardt's Disease in Turkish population.

2016 Batch

36. Babalola OE, **Adeyemo WL**. Feasibility and efficacy of implementing a CPAP/Oxygen helmet solution for non-invasive ventilation of COVID-19 patients in Nigeria.
37. **Guio H**, Jaramillo L. Metagenomic analysis and antimicrobial resistance in urban and hospital's wastewater of Huánuco.
38. Jaramillo L, **Guio H**. Association between ACE2 and TMPRSS2 genes polymorphisms and COVID-19 diagnosis.

39. Krishnan R, **Sirisena ND**, Wickramasinghe D, Dissanayake VHW. Molecular characterization of somatic genomic variants, circulating tumour DNA and gut microbiota associated with colorectal cancers.
40. **Malasa L**. The magnitude of Glucose-6-Phosphate deficiency in Dar es Salaam, Tanzania. (Proposal development).
41. **Okafor FU**. Translate knowledge in genetics and genomes nursing to clinical settings, community, and primary health facility.
42. Sjarif DR, **Ariani Y**, Dewi BE. Development of iduronate 2-sulfatase enzyme using mRNA method as a treatment for mucopolysaccharidosis type II.
43. Sjarif DR, **Ariani Y**, Fadilah. Developing pipeline for detection of genes causing rare diseases using whole exome sequencing data.
44. Subhedar K, **Tibrewal S**, Sen P. Maternal vitamin A deficiency and MAC (Microphthalmos, Anophthalmos and Coloboma) disorders.
45. **Tibrewal S**, Ratna Ria, Das S. Genotype-phenotype correlation of retinoblastoma in north India.
46. **Uwineza A**. Qualitative assessment of facultys' perspectives toward precision medicine-Education innovation project: Integration of Precision Medicine in medical curriculum at the University of Rwanda.

E. Ongoing Research Projects: (Total- 87; 2019- 28; 2018- 19; 2017- 24; 2016- 16)

2019 Batch

1. Abuzaid M, Ahmed N, Mostafa M, Abdelfattah M, Zaki S, Abd-Allah M, **Sayed I**. Studying the role of serine proteases in periodontitis in Papillon Lefevre Syndrome. (Summit lectures improved skills in reporting results).
2. 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. (Summit lectures on proposal/grant writing helped with the proposal of this project).
3. **Amarakoon GGGT**, Wijerathne NPKP, Chandrakumara WAJC, Wijayarathne HK. Clinical profile of Down syndrome in a tertiary care centre -Sri Lanka (Knowledge from Summit improved study execution).
4. Badoe E, Ameyaw EK, **Wiafe SA**, Manu E, Asafo-Agyei SB, Thomford NE, Ashong J. Molecular diagnosis of MPS patients in Ghana: The RDGI-BioMarin Project. (Knowledge from Summit helped design project).
5. **Chowdhury EH**, **Roy S**, Hoque F. Epidemiological study of Retinoblastoma in a Tertiary Eye Care Center of Bangladesh. (Knowledge gained at Summit improved research quality).
6. Donohue J, Badoe E, **Wiafe SA**, Ahiabile E, Wojtowicz W. Screening for urea cycle disorders in children: A pilot of blood ammonia checker device in a tertiary facility. (Knowledge from Summit helped amend protocol).
7. Hassib N, **Sayed I**. An attempt to diagnose typical and atypical cases by detection genes causing tooth structure anomalies. (Summit lectures improved skills in diagnosis of rare genetic disorders).
8. **Hussein N**, Lee YK, Qureshi N, John P, Azmi A, Ng CJ. Identifying gaps in thalassaemia screening: Exploring the views, experiences, and beliefs among thalassaemia carriers in Sabah,

- Malaysia. (Awareness on challenges of underprivileged population gained at the Summit helped with better communication; improved health service).
9. **Kars ME**, Ozcelik T. Prevalence of actionable secondary findings in 3,599 whole exomes and genomes from Turkey. (Knowledge gained from Summit lectures, personalized training and field trips helped with the interpretation of genetic variants and sequencing data analysis).
 10. **Llamos-Paneque A**, Hernández-Iñiguez P, Rivas-Iglesias C, Sanchez-Salazar J, Onofre Perez EJ. Clinical characterization of pediatric patients with Steinert's myotonic dystrophy diagnosed at the Hospital of Specialties of the Armed Forces No. 1. (Knowledge from Summit lectures in neurogenetics emphasized on triplet expansion disorders; helped in execution of the study.)
 11. **Llamos-Paneque A**, Lamar-Segura, E, Ñacato-Pachacama K, Guzmán A. Cytogenetic variants in patients with Turner syndrome diagnosed at the Hospital of Specialties of the Armed Forces No. 1 of Quito, Ecuador. (Summit helped reaffirm usefulness of registries for epidemiological studies, clinical characterization of genetic disorders).
 12. **López-Star B**, Pérez- Serrano R, Ochoa L. Identification of individuals carrying the rs7677751 polymorphism of the PDGFRA gene in patients with Keratoconus. (Summit lectures on eye disorders provided knowledge for execution of study).
 13. **López-Star B**. Pérez- Serrano R. Identification of rs7677751 polymorphism in PDGFRA gene, in patients with corneal astigmatism. (Dr. Hufnagel's team (NEI), helped understand the best choice of genes associated with astigmatism in Mexican population).
 14. 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).
 15. **Luong LH**, Hoang LT, Le NT, Tran TN. Study of genetics component in cardiovascular malformation and rare vascular disorder. (Summit encouraged discussions, collaborations and initiation of this project).
 16. **Luong LH**, Hoang LT, Le NT, Tran TN. Study of rare immunodeficiency disorder. (Knowledge and credentials from the Summit initiated discussions and project).
 17. **Mhandire K**, Dandara C. Pharmacogenomic determinants of resistant hypertension in Africans: Towards a gene-based treatment outcome prediction tool. (Dr. Marino L, a Summit contact, shared online resources that helped me pursue training in programming and bioinformatics; helped in setting-up this machine learning based project; helped transition to dry lab).
 18. **Monye HI**, Olusanya BA, Tongo K. Normative values for ocular biometric parameters in preterm babies and associated socio-demographic and clinical factors at the University College Hospital, Ibadan. (Summit lectures and field trips stimulated interest in newborn screening-ophthalmic context, study will provide baseline data for monitoring preterm babies).
 19. **Mosema KBA**, Spencer D, Bramble MS, Likuba EB, Mumba-Ngoyi D, Tshala-Katumbay D, Vilain E, Délot E, Vilain E. Disorders/differences of sex development in Central Africa: Genetics, psychosocial adaptation and Perceptions. (New skill acquired at the Summit and lectures on genome sequencing and variants interpretation helped write research proposal, and conduct research for a PhD thesis).
 20. **Mutreja D**, Komala SB, Singh V, Sreenivasa S. A longitudinal prospective study on role of serum hepcidin as a biomarker for iron status in chronic kidney disease (CKD). (Summit reiterated personalized approach for diagnosis of CKD related anemia utilizing hepcidin).
 21. **Mutreja D**, Venkatesan S, Sharma S, Tilak TVSVGK, Boruah D. A pilot study to evaluate the role of vascular endothelial growth factor and its comparison with microvessel density in

- angiogenesis of hematological malignancies. (Summit emphasized importance of gene disease association in hematologic malignancies).
22. Nnodu O (PI), Isa H, Tanko Y, Otu T, **Alli LA**, Chianumba R, Agumadu UN, Ekong J, Elmi Z, Rooks H, Menzel S, Ekong R. A pilot study on sickle cell disease to identify an unusual haplotype at 1 the BCL11A locus in Nigeria. (Summit lecture on Genetics of sickle cell disease enhanced my contribution; improved research quality).
 23. **Paredes-Moscossa 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, awareness of VUS, particularly in underrepresented populations (Peruvian); informed approach for project).
 24. **Sayed I**, Abdel-Kader M. 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).
 25. 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).
 26. Villegas-Llerena C, **Paredes-Moscossa SR**, Guevara-Fujita ML, Acosta O, Cardenas B, Arber C, Wray S, 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 and discussions improved approach, execution of project).
 27. **Wiafe SA**, Anyane-Yeboah K, Brew YA, Badoe E, Ameyaw EK, Thomford NE, Ashong J. Clinical and genetic evaluation of undiagnosed genetic and rare disease patients through the IHOPE Program in Ghana. (Knowledge from Summit improved design, developed protocol).
 28. **Wiafe SA**, **Kemavor-Asima D**, Ameyaw EK, Dunyoh B, Appiah DY, Amisah H, Akyaw P, Addo-Lartey E, Amoako E, Asafo-Agyei SB. Screening for inherited disorders and rare diseases in babies: The PICU-NICU Project. (Knowledge, experience from Summit inspired development of project).

2018 Batch

29. Acharya M, **Dutta AK**, Mukherjee S. Understanding genetic architecture underlying hereditary non-syndromic hearing loss. (Summit lectures, NCBI resources on genetics of hearing loss and variant prioritization were critical).
30. **Adeyemo TA**, Bolarinwa AB, Olatinwo AT, Adiat AL, Akanmu AS. Clinical and laboratory phenotypes of persons living with haemophilia in Southwest Nigeria. (Knowledge from Summit improved case reporting).
31. **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 helped initiate registry).
32. Afolabi BB (PI), Babah O, **Adeyemo TA**. Low dose aspirin for preventing intrauterine growth restriction and preeclampsia in sickle cell pregnancy (PIPSICKLE): a randomised controlled trial. (Knowledge from Summit supported study design, implementation).
33. 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. (Knowledge from Summit helped design study).

34. **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 proposal).
35. **Cornejo-Olivas M**, Cubas-Montecino D, Bazalar-Montoya J. DNA biobank implementation in Neurogenetics Division, Instituto Nacional de Ciencias Neurológicas. (Summit lectures on broad informed consent, biobanking and ethics supported proposal and design of standardized procedures of our first biobank).
36. **Cornejo-Olivas M**, Schmidt 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 Neurogenetics lectures on modifying effect of some variants on phenotype of triplet expanded disorders, improved study execution).
37. **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. (Summit introduced me to researchers in congenital malformations, motivated me to continue this work).
38. **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 me to do more research in Peru).
39. Kosiyo P, Otieno W, **Gitaka J**, Nyamuni J and Ouma C. Sickle cell genotypes in malaria. (Molecular techniques learnt at Summit enhanced sickle cell work; improved protocol development).
40. **Lertwilaiwittaya P**, Jinda W, Senanarong V, Assanasen J, Muangpaisal W, Rattanabannakit C, Limwongse C. Next generation sequencing in dementia subgroup in Thailand. (Summit workshops on Variant Interpretation helped establish pipeline for dementia NGS panel).
41. **Lertwilaiwittaya P**, Pho-iam T, Soontrapa P, Boonyapisit K, Limwongse C. Prevalence of C9orf72 hexanucleotide repeat expansion in different neurodegenerative diseases in Thailand. (Summit connected me to Dr. Bryan Traynor who helped obtain a positive control sample to establish local test in Thailand).
42. **Lertwilaiwittaya P**, Roothumnong E, Thongnoppakhun W, Limwongse C, Pithukpakorn M. Next generation sequencing in breast-ovarian cancer in Thailand. (Summit lectures on writing manuscripts helped write the proposal and manuscript draft).
43. 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. (Dr. K. Fischbeck's (NINDS) Neurogenetics talk at the Summit, suggestions and comments helped improve recruitment strategy and MTA agreement).
44. Menzel Stephan, **Adeyemo TA**, Nnodu O. Identification of novel mechanisms of fetal-haemoglobin induction by common genetic variation in patients with sickle cell disease. (Summit encouraged research collaborations).
45. **Messaoud O**. Genetic investigation of Xeroderma pigmentosum (Knowledge gained from Summit personalized trainings helped interpret next generation sequencing data).
46. Negi V, **Dwivedi A**, Ravi H, Goswami JN, Kumar R, Behl A. Diagnosis of inherited genetic disorders of brain, heart and kidney, at Army Hospital Research & Referral, New Delhi. (Knowledge from summit helped in conceptualizing, execution of the project).

47. Ofori-Acquah SF, Makani J, Awandare GA, Sani MU, **Adeyemo TA** SickGenAfrica: Sickle Cell Disease Genomics Network of Africa. (Summit lectures and discussions improved approach, execution of project).

2017 Batch

48. 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).
49. **Avogbe PH**, Brun LVC, Sanni A. Comparison of HPV detection in urine and cervical samples collected from Beninese women. (Resources from the Summit helped study design).
50. **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).
51. 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 provided tools for research design in complex diseases, analysis and interpretation of NGS data).
52. Besil-Suárez B, **Benítez-Cordero Y**, Álvarez-Gavilán Y, **Roblejo Balbuena H**. Community strategy for the promotion-prevention of genetic health in primary health care. (Summit provided tools for research design in complex diseases, analysis and interpretation of NGS data).
53. **Casado PL**, Aguiar T, Jordao M, Quinelato V. Relationship of geographic tongue before and during COVID-19 pandemic in patients under treatment. (Summit helped understand research on multifactorial diseases).
54. **Casado PL**, Campello M, Aguiar T. The relationship between COVID-19 and the higher risk of development of Kawasaki disease in children: A Systematic review. (Summit helped understand associated risk with diseases; improved research and proposal).
55. **Casado PL**, Lomardo P, Aguiar T. Association between the use of ultrasound and the risk of aerosol contamination in the dental office: A systematic review for COVID-19 pandemic. (Summit helped understand associated risk with diseases; improved research and proposal).
56. **Casado PL**, Luiza Motta L, Aguiar T, Quinelato V. Analysis of alcohol consumption during the covid-19 pandemic in Brazilian students. (Knowledge from Summit provided research basis for associated risk in diseases).
57. **Casado PL**, Quinelato V, Aguiar T. Analysis of 7 different techniques to isolate microRNA. (Summit workshop on Bioinformatics helped understand the relationship of miRNA with DNA and messenger RNA; helped improve execution of project).
58. **Casado PL**, Quinelato V, Aguiar T. Genetic profile of patients underwent peri-implant maintenance therapy: relationship with peri-implant disease incidence. (Summit lectures helped understand characteristics to be considered when studying multifactorial diseases, such as peri-implant diseases; improved the project, sample collection, medical history analysis, selection of main related genes).
59. **Casado PL**, Santos M, Telma Aguiar T, Valquiria Quinelato V. Analysis of the development of bruxism during the covid-19 pandemic in Brazilian students. (Knowledge from Summit provided research basis for associated risk in diseases).
60. **James O**, Adekunle AA. Wound healing following palatoplasty: A randomized study using honey or warm saline mouth bath. (Summit gave insight in exploring better care options for subjects born with orofacial clefts).

61. Lloyd W, **Mgasa A**, Makani J. Blood transfusion for sickle cell patients attending Temeke Regional Referral Hospital in Tanzania. (Summit enhanced knowledge, focus on genetics of SCD).
62. **Lotz-Esquivel S**, Alvarado-Aguilar M. Acute intermittent porphyria in Costa Rica. (Knowledge from Summit provided tools for soliciting specialized treatment and laboratory tests for these patients).
63. **Mahfoudh W**, Faleh R. Circulating tumor DNA as biomarkers for early diagnosis, treatment response and prognosis of breast cancer. (Project was inspired by Summit lectures and tailored training).
64. 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).
65. **Nandal R**, Biswas S, Laller KS. A study of serum vitamin-d levels in young adults with ST Elevation Myocardial Infarction (STEMI). (Summit enhanced research methodology, protocol preparation and grant writing).
66. **Otaify GA**, Abdelhamid M, Elhousseini R and Temtamy S. Targeted next-generation sequencing in the diagnosis and prevention of skeletal dysplasia with joint deformities. (Summit helped in preparation of proposal, writing progress reports).
67. **Roy S**. Effectivity of interferon alfa 2a in treatment of ocular surface neoplasia. (Knowledge from Summit inspired exploration of alternative treatments).
68. **Roy S**. Presentation of genetic diseases in a Tertiary Eye Care Center of Bangladesh. (Summit inspired the diagnosis of other genetic diseases presented with retinoblastoma, the first of its kind in Bangladesh).
69. **Torres-Mejía G (PI)**, Fejerman L. Ángeles-Llerenas A, Gómez Flores-Ramos L. LAGENO-BC & CONFLUENCE research proposal. (Knowledge from Summit helped write manuscript).
70. **Yousef YA**, Mohammad M, Mehیار M, Sultan I, Al-Hussaini M, AlNawaiseh I. 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. (Summit helped execute the project).
71. Zaki SS, **Otaify G**, Zaki MS, Issa MY, Mahmoud MM, Abdelhamid MS. Analysis of dystrophin-associated glycoproteins (α -dystroglycan and Laminin subunit $\alpha 2$): A shortcut for biochemical screening of many types of congenital muscular dystrophies. (Summit helped in preparation of proposal, writing progress reports).

2016 Batch

72. Baloglu E, **Deniz E** (Consultant). Identification of protein interacting partners of Na⁺/K⁺-ATPase pump in ischemic heart disease model. (Knowledge from Summit in genomic analyses helped design experiments).
73. Bode-Thomas F, Yilgwan CS, Amusa GA, Isa MS, Uche U, Ugwu C, Udo P, Sani MU, ...**Ekure EN**, et al. Nigerian National Registry for rheumatic heart disease. (Summit emphasized on relevance of birth defects registry including congenital heart disease, which was translated to another cardiac disease of public health importance in our setting).
74. Butali A, **Adeyemo WL**. Refining the genetic and genomic architecture of non-syndromic orofacial clefts. (Knowledge from Summit of immense value in advancing this project).

75. Dasanayaka NN, **Sirisena ND**, Samaranyake N. Telomere length, gene expression profile and DNA methylation status of selected genes in a Sri Lankan group of practitioners of meditation and mindfulness techniques (Knowledge from Summit supported study design and implementation).
76. **Deniz E**. Genome-wide screening with CRISPR/Cas9 and modelling of resistance mechanisms developed against cytotoxic drugs in cancer treatment. (Knowledge from Summit in human genomic variations helped analyzing bioinformatics data).
77. **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, neurofibromatosis and Hajdu Cheney syndrome. (Summit's introduction to the electronic atlas and importance of highlighting genetic syndromes in diverse populations for diagnosis, led to the necessity of publishing findings related to children of sub-Saharan African descent).
78. **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. (Knowledge and contacts from the Summit helped in driving leadership for this research group).
79. Krawitz P, **Ekure EN**, Opoola ZA, Lesi FEA. Computer-assisted image analysis and exome sequencing for the diagnosis of genetic disorders. (Summit introduced utilization of the Face2Gene software; resulted in collaboration with German PI).
80. **Lallar M**, Kaur L, Singh UP. Attitude, expectation and perception towards genetic counselling in parents of children with genetic disease. (Genetic counselling sessions at the Summit helped modulate sessions for patients).
81. **Lallar M**, Kaur L. Diagnostic yield of fetal and perinatal autopsy. (Knowledge from Summit on molecular testing and web resources helped in analysis of genetic diseases).
82. Mekkiy MK, **Hussen DF**. Molecular cytogenetic characterization and breakpoint mapping of chromosomal rearrangements towards better management of patients with genetic disorders and carriers (Molecular cytogenetic techniques learned at Summit helped in accurate diagnosis).
83. **Okafor FU**. Analysis of data for 'Reproductive health genetics and genomics education among faculty and students in basic nursing programmes in default Old Western Region, Nigeria' (PhD Thesis). (Knowledge from Summit will guide action for inclusion of genetics and genomics in nursing).
84. **Roblejo-Balbuena H**, Marcheco-Teruel B, **Benitez Cordero Y**, Monzón Benítez G. Genetic factors associated with the clinical severity of COVID 19 in Cuban patients. (Summit provided tools for research design in complex diseases; analysis and interpretation of NGS data).
85. Sjarif DR, **Ariani Y**, Priambodo R, Prakoso NM, Hafifah CN, Yuliarti K. Lysosomal storage disease study: developing screening and diagnostic methods for MPS II, MPS IVA, Gaucher disease, Pompe disease and NCL (Neuronal Ceroid Lipofuscinosis) type 2. (Project was inspired by Summit lectures on Inborn Error of Metabolism, Tools for molecular genetics, Diagnostic odyssey, Variant interpretation, Variant nomenclature.)
86. **Tibrewal S**, Ratna R, Dubey S. Childhood glaucoma – inheritance pattern and role of consanguinity. (Summit gave confidence to embark on this project).
87. **Tibrewal S**, Ratna R, Ganesh S. Familial inheritance pattern in childhood cataract in north India. (Summit gave confidence to embark on this project).

F. Other Accomplishments/Initiatives:

2019 Batch

1. **Abubakar S.** Guest Researcher at Social Network Methods Section, NHGRI, Visiting Program, DHHS (2020); Lectured on ethical, legal & social implication (ELSI) of Genetics and Genomics in the Medical Ethics & Law Course for Masters programs (MPH; Global Health), at College of Health Sciences and the World Bank Funded Africa Center of Excellence for Population Health and Policy, Bayero University, Kano, Nigeria; Appointed as Reviewer, BMC Health Services Research, Springer Nature (Global Publishers, 2021); Supervised undergraduate medical student on knowledge of Family History Taking Tools among Health Professionals in Nigeria.
2. **Akinmola OO.** Advanced in dialogue with management for starting a genetic counselling unit in the Tertiary Hospital, Lagos University Teaching Hospital; Lectured on Management of Neonatal Diabetes, Genetics as a tool for precision medicine, to optimize outcome for patients, Newborn screening: Neonatal hypothyroidism, at Paediatric Endocrinology Training Centre For West Africa; Lectured on Human genetics to Chemical Pathologist Society of Nigeria; Member, G2MC Rare Disease Working Group for pilot study at different sites; Attended the virtual International conference on COVID-19, International Federation of Clinical Chemistry (Feb. 2021); Identified a rare disease of Kallmann syndrome, counselled and referred appropriately for treatment.
3. **Alamin TA.** Joined 2-year Master of Medical Education program (2021); Introduced genomics in undergraduate medical laboratory sciences curriculum; Participated in initiation of the Sudanese Human Genetic Association.
4. **Alli LA.** Appointed External examiner, first Professional Examination in Medical Biochemistry and Molecular Biology, Kaduna State University (Dec. 2020); Appointed, Visiting Lecturer, Medical Biochemistry and Molecular Biology for Preclinical Medical students, Nile University, Abuja Nigeria (Jun. 2021); Used materials from Summit to lecture on Basic molecular biology, Genetic disorders, and Inborn errors of metabolism; Strengthened the Biochemistry curriculum on genetics and genomics for preclinical medical trainees; Reviewed research articles for Pharmaceutical Biology and British Journal of Medicine and Medical Research; Supervised, mentored postgraduate research students (5 MSc, 1 PhD) on projects based on human genetics, molecular biology and ethnopharmacology management of SCA.
5. **Amarakoon GGGT.** Presented poster on 'A case report of a child with Southeast Asian Ovalocytosis', at 13th International Research Conference-KDU IRC (Sri Lanka, Oct. 2020), aPresented talk on 'Infections during the 'Remission-induction period' of acute lymphoblastic leukemia of children in Sri Lanka', at 134th Anniversary International Medical Congress, Sri Lanka Medical Association (Sri Lanka, Jul. 2021); Lectured on human genetics, hemoglobinopathies and other congenital hemolytic anemia to undergraduates, at Faculty of Medicine and Allied Sciences, Rajarata University of Sri Lanka; Maintained a database for recording syndromes, congenital malformations and genetic disorders in the University Pediatric Department, Teaching Hospital Anuradhapura, Sri Lanka.
6. **Baatar N.** Wrote a manual on 'Primary care approach in Adolescents' (Jun. 2021); Presented talk on 'Adolescent health issues' (virtual, Jun. 2021); Attended 6th Annual International Conference on 'Genomics and Genetic Counseling: Value in Health Care' (virtual, 2021); Attended Covid-19 webinars from Raphael International (2021), 'HPLC 1260 user training course' (Feb. 2021), 'Maccura's Cutting Edge Solution for COVID-19' (virtual, Sept. 2021).
7. **Bocoum A.** Joined International Parkinson's disease Genomic Consortium; Member, African Section Educational Committee; Awarded, GP2MSc scholarship; Established Malian National Registry of Parkinson's disease.

8. **Chowdhury EH.** Summit certificate helped secure position of a specialist in the Department of Pediatrics, Samtse General Hospital (SGH), Royal Government of Bhutan (2020); Previously was Associate Consultant, at Department of Pediatrics, Chittagong Eye Infirmary and Training Complex, Chittagong, Bangladesh; Initiated registry for congenital abnormalities in Department of Pediatrics, SGH; Emphasized importance of genetic counseling based on Summit learnings; Trained colleagues to establish research/clinical facilities in SGH; Emphasized on personalized care; Presented on 'Recent management protocol of common genetic diseases in children', CME program in SGH; Identified and managed pediatric genetic disorders (19) in Department of Pediatrics, SGH; Diagnosed cases Down's Syndrome (7), Hemophilia (3), Congenital Hypothyroidism (3), Achondroplasia (2), Neurofibromatosis (1), Turner's Syndrome (1), Duchenne Muscular Dystrophy (1), Marfan's Syndrome (1); Referred cases for cardiac surgery (4), Ophthalmic surgery (1), physiotherapy (2); Diagnosis improved quality of life of affected individuals and their families due to known prognosis and access to appropriate support.
9. **Guven Y.** Presented poster on 'Three Nance Horan syndrome families from Turkey: Three different approaches for molecular diagnosis', at ESHG Conference (virtual, Aug. 2021); Presented poster on 'Gorlin-Goltz Syndrome with a de novo variant in *PTCH1*: A case report', at 28th Congress of the IAPD (virtual, Jun. 2021); Diagnosed, referred patients (12) to genetics department for molecular confirmation and medical referral- Ectodermal dysplasia (4), Nance Horan syndrome (1), Primary Failure of eruption (1), Oligodontia cases (8); Early diagnosis, surgical interventions, dental prosthodontic and restorative therapies, preventive and follow-up protocols, contributed to socio-psychological well-being, improved quality of life of children.
10. **Gyawali P.** Young Investigator, Global Genomic Medicine Collaborative (G2MC); Member, Family Health History Flagship Project under G2MC; Corresponding Member, International Federation of Clinical Chemistry, Task Force in Global Newborn Screening; Master's thesis supervisor, Global Health, University of Copenhagen.
11. **Hussein N.** Coordinator, Postgraduate Family Medicine Specialty, University of Malaya, Malaysia; Audited postgraduate curriculum to increase awareness, strengthen Family Medicine in genetics and genomics; Received scholarship to attend RARD (S. Africa, Jun. 2020; postponed due to COVID-19 pandemic); Invited speaker, by **Tumulak MJR** (Summiteer) for an online session, on 'Cancer genetics in primary care' and 'Communicating genetic risk', University Malaya (Jun. 2021); Planned topics and workshops on 'Use of technology in patient care', 'Patient education and counselling', 'Chronic kidney disease' and 'Family planning'; Mentored Family Medicine trainees on research projects in 'Challenges among primary care doctors' on prenatal genetic counselling and testing', 'Parental consent in school-based thalassemia screening'; Developed guidelines, supervised trainees in tele-consultation of patients during COVID-19 pandemic; Identified genetic disorder- alpha thalassemia (1).
12. **Iqbal M.** Supervised graduate research students (4 PhD, 7 MS) on molecular and genetic basis of different ophthalmological disorders; Developed curriculum, delivered lectures on Advanced Molecular Genetics to graduate level students; Appointed as External Examiner, reviewed theses (1PhD, 7 MS/MPhil); Reviewed research articles (3) for Journal of Animal and Plant Sciences (JAPS) and Book Chapter for Frontiers in Molecular Pharming.
13. **Kars ME.** Presented seminar on 'Characterization of the fine-scale genetic structure of the Turkish Population', at Molecular Biology and Genetics Department, Bilkent University (Mar. 2021); Used materials from Summit for Teaching Assistantship on Human Genetics and Introduction to Human Biology (2020-2021); Shared knowledge with fellows and undergraduate

- students (7), during their rotations in the genetics laboratory; Used knowledge and resources gained from the Summit for data analysis and presentations to the Thesis committee (2020-2021).
14. **Khant AK.** Speaker on 'Acute Kidney Injury in Children', for MS students in Paediatric, organized by Myanmar Paediatric Society (Virtual, Oct. 2020); Examined, diagnosed, treated children (~200) with kidney disease- acute kidney injury due to viper bite/G6PD deficiency/drug overdose/sepsis, prolong shock (30), chronic kidney disease (20), recurrent urinary tract infection (10), nephrotic syndrome (40), steroid resistant nephrotic syndrome (30), rapidly progressive glomerulonephritis (10), systemic lupus erythromatosus (40), vasculitis (15), IgA nephropathy (10); Counselling patients (40) with hereditary renal disease- congenital nephrotic syndrome (5), congenital abnormalities of kidney urinary tract (11), meningomyelocele with neurogenic bladder (7), syndromic child with renal abnormalities (5), Lowe syndrome with renal tubular acidosis (2), focal segmental glomerulosclerosis (8) and several others (5).
 15. **LLamos-Paneque A.** Professor, Human Genetics (undergraduate level), School of Dentistry, International University of Ecuador; Updated topics in human genetics; Lectured on clinical genetics to medical students, professionals at Equinocial Technological University of Ecuador and Army Hospital of Quito; Selected Member, Editorial Board of Molecular Cytogenetics Journal; Summit provided access to research groups to drive medical genetics in low-income countries; Established contact with Ibero-American Network of Neurocutaneous Diseases (Drs. F. Ramos, MT. Acosta, NIH)- presented and attended discussions on neurocutaneous cases and therapeutic advances; Summit improved genetic counseling skills; Implemented genetic counseling service at Maternal Fetal Unit (50 cases/yr) for prenatal patients that tested positive for genetics conditions; Identified cases (2) with structural chromosomes anomalies including supermarket chromosome, detected through molecular cytogenetics studies with German collaborator (Dr. T. Liehr), as service is not available in Ecuador; Evaluated genetic cases (450) through molecular tests -mono gene etiology (100), chromosome (150) through cytogenetic studies, or multifactorial (200); Registered new cases (250) in the Registry of Genetic Diseases, Specialty Hospital No.1 FF. AA, Quito, Ecuador.
 16. **López-Star B.** Speaker on 'Experience as NEI/NIH Fellow', at Mexico-United States Symposium on Vision Health: Priorities and Opportunities for Bilateral and Regional Engagement in Scientific and Public Health Research (Jan. 2021); Trained junior staff in ophthalmogenetics; Established system to record, monitor patients (63) with inherited retinal Disease; Evaluated, diagnosed, treated ophthalmogenetic cases (8)- Marfan syndrome (4), pigmentary retinosis (4); Referred patients (3) for medical management to Children's and Women's Hospital, Mexico; Diagnosis improved quality of life for patients and their families due to known prognosis and access to appropriate support; None were candidates for treatment, but couples made informed decisions in family planning; Collaborated with the Asociación Para Evitar la Ceguera, Mexico to refer patients with congenital amaurosis of Leber (1).
 17. **Luong LH.** Affiliated with National E Hospital, Vietnam (2020); Helped establish lab with focus on genetics of immune disorders- immunodeficiency disorders, auto-inflammatory diseases, and autoimmune disorders.
 18. **Mhandire K.** Awarded, Postdoctoral fellowship at Roswell Park Cancer Institute, USA (Mar. 2021); Completed 54 hours of 'Lockdown Learning Bioinformatics' (<https://www.youtube.com/channel/UC7aizSyonJqZI3O3U4SDTiA>).
 19. **Monye HI.** Completed online courses on 'The genomic era: The future of genetics in healthcare', 'Genomic technology in clinical diagnostics: Next generation sequencing', offered by St George's, University of London (Jan-May 2021), 'Genomic scenarios in primary care', offered

by Wellcome Connecting Science (Jan. 2021); These positively influenced patient care; Awarded, Developing Country Eye Researcher Travel Fellowship (2021), by Association for Research in Vision and Ophthalmology; Awarded, Best abstract for ‘The impact of the COVID 19 pandemic on the ophthalmology residency training programme in Nigeria’, at the 52nd Annual Scientific Conference of the Association of Resident Doctors, University College Hospital, Ibadan; Member, Global Genomic Medicine Collaborative (G2MC) Family Health History and Evidence Workgroups, paired-reviewer for a systematic review on evidence of the clinical utility of genomic medicine in patient outcomes.

20. **Mosema KBA.** Lectured in genetics and pediatrics to medical students at Université Protestante du Congo, Kinshasa and Université Uele, Isiro, Haut-Uele Province; Continued establishment of Unit of Sickle Cell Disease (screening, treatment and follow-up), at Biamba Marie Mutombo Hospital, Kinshasa City; Created a WhatsApp group for clinical training in genetics and genomics for medical students of several universities in the DR Congo (UPC, UU, UK etc); Diagnosed cases- holoprosencephaly (1), achondroplasia (1) trisomy 13 (1).
21. **Mushi TL.** Founder, Executive Director of-INNOVATION FOR HEALTH DEVELOPMENT (iHD Foundation), an NGO, to promote community development through evidence-based research, advocacy and lifesaving initiatives that include advancements in genetics and genomics, precision medicine, empowerment of health workforce to reduce maternal-child mortality rates, respecting human rights, in Tanzania.
22. **Mutreja D.** Delivered online lectures on Genomics to faculty and colleagues, lectured post-graduate residents and medical students; Shared resource materials from Summit with medical students, postgraduate residents, colleagues from other specialties; Section Editor (Pathology), Indian Journal of Medical Paediatric Oncology (2020-21); Identified cases (18) by histopathologic /microscopic diagnosis- Lafora disease (1), solitary rectal ulcer syndrome(3), thalassemia (4), sickle cell disease (2), Gaucher disease (1); Improved quality of life by prompt diagnosis and medical management.
23. **Nair L.** Completed Medical Genetics Residency (Oct. 2021); Mentored Genetic Counselling students in basic genetics; Conducted webinars for Pediatricians, Pediatric Neurologists, Developmental Pediatricians, Genetic Counselling students on ‘Genetic disorders’, ‘Genetic tests and their clinical implications’; Conducted OPD, evaluated, diagnosed patients with various genetic disorders, provided genetic testing and genetic counselling; Identified cases- Down syndrome (6), unexplained intellectual disability/ developmental delay (32), recurrent pregnancy loss/ infertility (10); Evaluated and diagnosed rare genetic disorders like CLCN1 gene associated myotonic dystrophy (1), VPS51 gene associated Pontocerebellar hypoplasia type 13 (1), SCN3A gene associated neurodevelopmental disorder (1), pyruvate carboxylase deficiency (1), neurofibromatosis type 1 (1); Started Down syndrome registry at NIMS Medicity, Trivandrum, Kerala.
24. **Okunola O.** Attended Training Fellowship in Stigma research, FIC/NIH (virtual, Jun. 2021), Summit influenced selection; Used experience and materials from Summit to train, mentor medical students, resident doctors, colleagues in medical genetics and genomics, research grant applications; Liaison, working group with paediatric nephrologists to establish congenital kidney disease registry with follow up; Diagnosed patients (20) with inherited kidney diseases (all autosomal dominant polycystic kidney disease), screened, followed up on siblings; Patients and family reported improvement in quality of life due to accurate diagnosis and pathway to care.
25. **Ortiz-Panozo E.** Earned a Fulbright García-Robles scholarship for Doctoral training (starting 2022); Attended ‘Targeted Machine Learning for Big Data’, Summer Program, National Institute of Public Health, Mexico (2021).

26. **Paredes-Moscossa SR.** Submitted abstracts on ‘Abundance of Variants of Uncertain Significance (VUS) found using multi-gene panels in Peruvian patients with hereditary breast cancer’, ‘Modelling with CRISPR/Cas9 the BRCA1 gene ‘Variants of Uncertain Significance’ identified in Peruvian Patients’, and ‘Identification of a *TREM2* mutation (W44X) in a familial case of Alzheimer’s Disease from Lima (Peru) by Whole-Exome Sequencing’, to Asociación Latinoamericana de Genética (2021, Accepted); Selected, mentored a training program, ‘Introduction to Mentoring in Higher Education’, sponsored by CONCYTEC and The British Council (Nov. 2020); Invited reviewer by **Pirlog, R** (Summiteer), for ‘Innovation in Science’ call by Asaih, a Romanian Entrepreneurship Association connecting students from Iuliu Hatleganu University of Medicine and Pharmacy (Romania) with local and international stakeholders (Nov. 2020); Adviser, Multisectoral Working Group for National Policy for the Development of Science, Technology and Technological Innovation (Feb. 2021); Awarded, scholarship for Diploma in Science Communication, funded by CONCYTEC and organized by Universidad Peruana Cayetano Heredia (Mar. 2021); Awarded, training grants for ‘Innovation and Science Diplomacy’, sponsored and organized by The Sao Paulo Innovation and Science Diplomacy School (InnSciD SP) and The World Academy of Sciences (Jul. 2021) and AAAS (Aug. 2021).
27. **Pirlog R.** Awarded, accepted Fulbright scholarship at MD Anderson Cancer Center, Tx, USA (Aug. 2021) for research on role of tumor microenvironment in early development of lung cancer; Pursued an internship in molecular pathology at the University Hospital of Rouen, France (Nov. 2020- May 2021); Started a molecular pathology laboratory; Helped secure funds to sustain a local biobank and its infrastructure for alignment with international consortiums.
28. **Sayed I.** Promoted to Associate Professor of Oro-dental Genetics (Oct. 2020); Identified cases (61)- neurogenetic manifestation (32), skeletal manifestation (18), ectodermal dysplasia patients (8); Referred patients (3) for prosthetic management of ectodermal dysplasia.
29. **Tiong SY.** Integrated and implemented genetic counselling into mainstream prenatal service as part of antenatal and post-natal care (Jun. 2021); Increased awareness among expectant mothers on newborn screening, inherited metabolic disorders (IEM), at Loh Guan Lye Specialists Centre, Penang; Offered IEM genetic tests with an average of 7.5% uptake; Involved in integrating genetic counselling at Breast Cancer Centre, Loh Guan Lye Specialists Centre, Penang (Dec. 2020); Prepared bilingual (English, Chinese) flyers to increase awareness in newborn screening, metabolic disorders, hereditary breast and ovarian cancers; Committee member, Genetic Counselling Society Malaysia (2020-2021); Referred 74 new cases in cancer and paediatrics for genetic counselling.
30. **Tumulak MJR.** Thesis panel member for genetic counseling students (13 students); Genetic counselor for studies on ‘Determination of the prevalence of genetic mutation in high risk Filipino hereditary breast cancer patients’, ‘Genotypic and phenotypic variations of inherited retinal degenerations among Filipino patients in a tertiary hospital’; Invited, resource speaker on ‘The role of genetic counselors in newborn screening programs’ by various forums- Rare Disease Ghana Initiative (virtual, Oct. 2020), 18th National Newborn Screening Convention (virtual, Oct. 2020); 6th International Conference, Board of Genetic Counseling, India (virtual, Jul. 2021); Organized training on genetic counseling for hemoglobinopathies and fatty acid oxidation disorders, Food and Nutrition Research Institute InGest Study (Jul. 2021); Provided pre-test and post-test genetic counseling to patients – breast cancer patients (100), cystic fibrosis patients (50), alpha and beta thalassemia patients (600), and ophthalmology patients (3).
31. **Weldemichael YG.** Work impacted by COVID-19.

32. **Wiafe SA.** Co-Chair, Developing Nations Working Group- Undiagnosed Disease Network International (2020); Vice Chair, Patient Advocacy Constituent Committee, International Research Consortium for Rare Diseases (Jul. 2021); Panel member, WHO Collaborative Global Network for Rare Diseases; Organized, 3rd Symposium on Birth Defects and Rare Disease (virtual, Mar. 2021); Attended 9th Conference on Rare and Undiagnosed Diseases and Mayo Clinic Science Session (virtual, April, 2021); Speaker, on 'Improving the standards of care for rare disease patients in Ghana', IEEEPO Changemaker (Jun. 2021); Panelist on 'Unlocking the power of diversity in genomic data diversity', African Health Online (virtual, July 2021); Enrolled patients (30) with undiagnosed rare genetic disease in IHOPE Program, Ghana; Enrolled patients (3) with MPS in RDI-Bio Marin Project.
33. **Yadav S.** Fellow in Obstetrics and Gynecology (Advanced ultrasound course), at Mediscan, Chennai, India (Jan. 2021); Delivered lecture on 'Role of prenatal screening', for Genetic counsellors organized by Future Medicine Academy (Feb. 2021); Attended, 'Expert fetal ultrasound- one day extempore course' (virtual, Jul. 2021), organized by Ukrainian Evidence Based Medicine Association; Shared knowledge from Summit in genetics with students from different specialties; Performed antenatal scans and genetic sonograms (100); Identified fetuses with congenital malformations- congenital diaphragmatic hernia (1), multicystic dysplastic kidneys (2), congenital talipes (1); Provided preconception, antenatal counseling to patients (50).
34. **Yesilcinar I.** Presented talk on 'Post-test counselling: how to convey the gene test results to the patient & family?', at 4th Breast Cancer Virtual Conference, Turkey (Oct. 2020); Speaker on 'Test/Retest reliability of a Turkish version of genetics and genomics in nursing practice survey (GGNPS)', ISONG virtual Congress: Genomic Nursing: Fulfilling Nightingale's Legacy of Care (Nov. 2020). Presented posters on 'Genetics and genomic competency of Turkish nurses: a descriptive cross-sectional study' and 'Genetic knowledge and practice among Turkish nurses; A literature review', at Nursing Genomics and Healthcare Conference (virtual, Jul. 2021).

2018 Batch

35. **Abdulkareem F.** Obtained Doctor of Medicine (MD), from National Postgraduate Medical College of Nigeria (Jan. 2021); Obtained a certificate in Genomic Technologies in Clinical Diagnostics-Molecular Techniques, from St George's, University of London (Dec. 2020); Member, of Accreditation Panels at Eko Medical University of Medicine & Health Sciences and University of Nigeria Teaching Hospital, Enugu (Feb. 2021); Appointed, Curriculum review committee for Biorisk management in Nigerian universities, participated in workshops organized by Sandia National Laboratories, USA, in Abuja (Mar. 2021) and by Faculty of Pathology National Postgraduate Medical College (May 2021); Participated in Laboratory Assessors' Training Workshop Phase 3 on ISO/IEC for ISO 15189 organized by NINAS Abuja (May 2021); Attended Topics in Human Genetics Summer Course, University of Iowa (Jun. 2021); Did professional assessments for promotions at University of Nigeria Nzukka, Delta State University and University of Sokoto, Nigeria; Attended 15th Annual Scientific Conference of College of Nigerian Pathologists, (Nov. 2020); Attended, Genomic Frontiers Conference (virtual, Jan. 2021); Invited speaker on '-Unknown Case', at Pathology Symposium of African Research for Oncology (virtual, Feb. 2021); Accepted, oral presentation on 'Risk factors for abnormal cervical cytology in women living with HIV in Nigeria', virtual AORTIC (Nov. 2021); My NGO, FAJIM Medicare Foundation (<https://fajimmf.org.ng>) commemorated World Diabetes Day (Nov. 2020), World Cancer Day (Feb. 2021), Hypertension Day (May 2021); Raised awareness and medical outreach for free screening and treatment in colorectal and breast cancer through radio jingles,

television interviews, webinars; Due to COVID no cases of congenital abnormalities were recorded for this period.

36. **Adewole OO.** Appointed, Country Ambassador, Member of Trustee Royal Society of Tropical Medicine and Hygiene (2021); Lectured, trained residents, medical students in genomics and its implications, roles in clinical practice; Shared knowledge from the Summit.
37. **Adeyemo TA.** Mentored new junior faculty (3), College of Medicine University of Lagos and Lagos University Teaching Hospital; Expanded World Bleeding Disorder Registry; Identified, patients (80) with inherited bleeding disorders; Counseled and referred patients for care.
38. **Cornejo-Olivas M.** Completed 2-year Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Principal investigator, Coordinator of the First DNA Bank in the public system, Peru; Appointed member, Council for Rare and Orphan Disorders by Peruvian Ministry of Health; Nominated as Executive Committee member for the Pan American Section of the International Parkinson and Movement Disorder Society (MDS); As MDSgene Taskforce member completed systematic review on SCA17; Invited speaker, ERCAL Latin American initiative (Feb. 2021), and Telehealth program, by Ministry of Health (Jun. 2021); Faculty, XXVII Peruvian Congress in Neurology (Jun. 2021); Mentored trainees (3) from Northern Pacific Global Health Research Fellows Training Consortium (FIC/NIH), Global Brain Health Institute, University of California, and Young Members MDS Mentoring Program.
39. **Daich Varela M.** Started, PhD at University College of London, UK (Feb. 2021); Coordinator of Ophthalmic genetics module for post-graduate degree courses in Ophthalmology, University of Buenos Aires, Argentina; Presented talks on 'Inherited retinal disorders' at Buenos Aires (Nov. 2020) and in London (Feb. 2021); Continued working on gene therapy clinical trials; Evaluated ophthalmic genetics patients; Identified, diagnosed and managed cases (70)- CSNB (10), rod-cone dystrophy (20), cone-rod dystrophy (20), coloboma (5), albinism (5), other rarer syndromic diseases (10); Referred all for counseling and medical management.
40. **Das S.** Completed fellowship in Clinical Genetics, Dept. of Medical Genetics, Christian Medical College Hospital, Vellore, India (2021); Trained medical and nursing students in basic clinical genetics; Volunteered with Organization for Rare Diseases India; Involved in neuromuscular and perinatal clinic for patient management; Identified, diagnosed and counseled patients (~350) with various genetic disorders- juvenile amyotrophic lateral sclerosis 2 (1), autosomal recessive pontocerebellar hypoplasia with microcephaly (1), argininemia (1), ataxia telangiectasia (3), benign familial infantile seizures (2), familial infantile convulsions with paroxysmal choreoathetosis (1), 46XX,t(7;15)(q35;q26.2) (1), biotinidase deficiency (1), Charcot-Marie-tooth disease, axonal, type 2A2A (3), FSHD 1 (1), DMD (17), achondroplasia (2), cystinuria (1), homocystinuria (2), Glass syndrome (1), Coach syndrome (1), Coffin Lowry syndrome (1), Cohen syndrome (1) amongst others; Referred patients to specialty departments for interdisciplinary management.
41. **Dueñas-Roque MM.** Medical Geneticist, at Genetic Department, Hospital Nacional Edgardo Rebagliati Martin; Professor, Medical Genetics for undergraduate students, Medical School, Universidad San Ignacio de Loyola, Perú; Successfully completed the Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Shared knowledge from Summit with students; Submitted abstracts 'Deployment of clinical whole genome sequencing in support of more than 1,000 resource-limited patients: Four years of the iHope Program' to ASHG (2021) and 'Variable phenotype within dopa responsive dystonia, experience from two Peruvian cases' to Movement Disorders Meeting (2021); Participated in ICORD/ERCAL group, an international initiative to bridge the gap in rare diseases in Latin America (virtual, May 2021); Enrolled as

- young investigator, participated in Evidence Working Group and flagship projects in Global Genomic Medicine Collaborative (G2MC); Summit influenced, helped establish an Undiagnosed Disease Program (UDP) at Hospital Edgardo Rebagliati Martins, EsSalud; Enrolled cases (65 trios) in iHope, Illumina Project for identification of genetic causes of rare diseases in Peru.
42. **Dutta AK.** Established, Paediatric Genetics OPD, at AIIMS Kalyani (India); Summit inspired initiation of a quasi-experimental project for increasing medical graduates' interest in, Inborn Errors of Metabolism and as partial fulfilment for the Foundation for Advancement of International Medical Education and Research (FAIMER) Fellowship (May 2021); Faculty for the Society of Fetal Medicine Postgraduate Teaching Program, presented on 'What are aneuploidies, chromosomal abnormalities and non-disjunction' (Nov. 2020); Resource person for webinar series 'Genomics in clinical practice', organized by Bangladesh Physicians (Mar. 2021), presented on 'Multifactorial vs polygenic & monogenic disorders', 'Basic genetic testing and ethical issues', 'Genetic diagnosis-database and search engines', 'Inborn errors of metabolism case scenarios', 'Basic genetic counselling'; Summit curriculum, resource materials helped prepare for these talks; Joined the Education Working Group in Global Genomic Medicine Collaborative (2021).
 43. **Dwivedi A.** Created, designed various platforms to spread awareness on genetic disorders among armed forces personnel, families; Conducted webinars on cancer genetics, genetics of renal disorders & lysosomal storage disorders etc.; Used materials from Summit to educate fellow residents, researchers; Invited Speaker on World Rare Disease Day by Takeda, delivered lecture on 'Care for rare diseases' (virtual, Feb. 2021); Panelist on 'Lysosomal storage disorders', at 6th South Asian symposia (virtual, Apr. 2021); Presented poster on 'A cross sectional survey to assess the knowledge, attitude and practices towards genetic disorders and testing among non-geneticist clinicians of Indian Armed Forces', 6th Annual International Conference of the Board of Genetic Counseling India (virtual, Jul. 2021); Member of Global Genomic Medicine Collaboration; Initiated molecular testing including MLPA, Sanger sequencing for common genetic disorders (Thalassemia, Achondroplasia etc.), at AHRR, Delhi; Diagnosed patients (680) with various genetic disorders (Chromosomal, Mendelian and disorders of Genomic imprinting); Knowledge from Summit helped manage patients.
 44. **Edem-Hotah J.** Qualified as Foundation Fellow of the College of Nursing and Midwifery, Sierra Leone Postgraduate College of Nursing and Midwifery (May 2021); Invited by the Dean, Faculty of Basic and Health Sciences, Ernest Bai Koroma University of Science and Technology to revise Public Health curriculum (BSc Hons) for inclusion of genetics and genomics pedagogy, research and management of sickle cell disease (Mar-Jun. 2021)- used materials from the Summit; Observed increased awareness for genetics and genomics amongst COMAHS faculty and students, as well as political and technical leaderships of the Ministry of Health and Sanitation and related agencies; Initiated training of nurses and midwives and counselling at the Postgraduate College of Nursing and Midwifery.
 45. **Eshete MA.** Work impacted by COVID-19.
 46. **Gitaka J.** Established Sickle cell surveillance at Mary Help of the Sick Mission Hospital in Thika, Kenya; Developed, strengthened genomics content in Clinical Medicine Course curriculum, at Mount Kenya University; Referred patients (10) for genetic counselling and medical management; Helped allay anxiety due to known pathway to care.
 47. **Kemavor-Asima D.** Expanded, National Rare Disease Registry and Congenital Disease Registry, in collaboration with Mr. **S. Wiafe** (Summiteer) of Rare Disease Ghana Initiative; Increased

awareness to newborn screening and metabolic diseases, sensitized clinicians and organizers of clinical workshops.

48. **Lertwilaiwittaya P.** Started 4-year Combined Internal Medicine and Medical Genetics Residency Program at University of Alabama at Birmingham, USA (Jun. 2021); Prior physician, at Division of Medical Genetics, Department of Medicine and Siriraj Genomics Laboratory, and Department of R&D, Faculty of Medicine Siriraj Hospital, Mahidol University, Thailand (May 2021); Completed 2-year Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Presented posters on ‘Uptake and utilization of germline BRCA1/2 testing for Thai woman with breast cancer’ and ‘Unfavorable 5-year outcomes in the first familial amyloidosis polyneuropathy case that underwent liver transplantation in Thailand’, at American College of Medical Genetics (2021); Participated in Genomics Thailand (Whole Genome for 50,000 Thais) recruitment process; Co-founded Monday Oncogenetics Clinic, Siriraj Hospital; Increased the cohort of cancer patients (1940) for NGS testing.
49. **Mburu S.** Presented on ‘Digital learning in the context of Covid 19 crisis: Leveraging on big data analytics’ at 4th International Kirinyaga University Conference (Kenya, Mar. 2021).
50. **Messaoud O.** Completed 2-year Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Used resources from Summit for lecture on Human Genetics, at Faculty of Medicine of Tunis; Young investigator, Global Genomic Medicine Collaborative (<https://g2mc.org>); Reviewer, Medical Hypotheses journal and Topic Editor for the journal Frontiers in Genetics; Organized, participated, session moderator for an international event- Innovation-H Competition ‘Increasing mobility for people with handicaps’ (virtual, Oct. 2020); Participated in virtual trainings and workshops on ‘Facilitating online meetings and workshops’, ‘Integrating digital learning tools in higher education’, and ‘Leadership skills in academia’ (virtual, Nov. 2020); Participated in RDWG-ClinGen workshop, organised by H3Africa and ClinGen (virtual, Feb. 2021);
51. **Mistri M.** Received, Best Young Researcher Award, from International Academic and Research Excellence Committee for exceptional caliber, outstanding performance as academician, researcher, mentor, advisor and leader (2020); Published first largest series of Indian molecular data for various lysosomal storage disorders that include Niemann-Pick disease; As Member, Global Genomic Medicine Collaboration, implemented Undiagnosed and Rare Disease Flagship Project (March 2021).
52. **Muttamba W.** Summit introduced concept, importance of biobanking and sequencing; Involved in multi country initiative to increase biobanking and sequencing capacity in low- and middle-income countries as part of the COVID-19 pandemic response; Identified a case of sickle cell disease, referred for further management.
53. **Naeem MA.** Member, Entry Test Examination Committee for MPhil/PhD at CEMB (Dec. 2020); Supervised PhD students (2), who successfully graduated (2020); Trained MS/MPhil research internees (3) from University of Education (UE), University of the Central Punjab (UCP) and Kinnard College for Women (KCW), at Vision Impairment Lab of CEMB, Lahore, Pakistan.
54. **Nakousi-Capurro N.** Clinical Geneticist at Hospital Carlos Van Buren of Valparaíso, Chile (2020); Faculty at Universidad de Valparaíso as Associate Professor (May 2021); Designed courses, lectured medical students in genetics and clinical genetics; Provided on-site clinical genetics training to neuropediatric residents and medical students; Assisted in hosting 3 online courses (a) Skeletal dysplasias (first Latin American course), organized by Chilean Clinical Geneticists, (b) Genomic medicine, organized by the Society of Brazilian Geneticists, GeneOne and DASA educa., (c) Human and mammalian genetics and genomics: The McKusick Short

Course 202, organized by Johns Hopkins University and the Jackson Lab, USA (Apr.- Jul. 2021); Drafted proposal for acquisition of NGS technology for Carlos Van Buren Hospital; Presented online talks on ‘Prenatal testing techniques for chromosomal disorders’, for Regional and in-hospital gynecologists; Established protocols with breast cancer team for prioritizing patients for genetic testing; Collaborated with a telemedicine platform to deliver clinical care to patients and education/information to colleagues in genetics.

55. **Oluwarotimi AC.** Currently the sub-Dean, College of Medicine, University of Lagos; Chairman, Medical and Dental Consultant Association of Nigeria, Lagos University Teaching Hospital Chapter; Member, Dental Therapist Registration Board of Nigeria; National Liaison Officer, Nigerian Dental Association. (2019 –till date). Chairman, Constitution Amendment Committee, Nigerian Dental Association (2019/2020); Coordinator, Brush Twice Daily Programme organized by Unilever Global/ Nigeria. (2008- Till Date).
56. **Ottaru S.** Completed 2-year Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Invited, presented on ‘Predictors of Toxoplasma gondii IgG seropositivity and cranial ultrasound patterns among children with hydrocephalus in a tertiary hospital in Mwanza’, at 2nd International conference on Pediatrics and Pediatric Healthcare (France, Aug. 2021).
57. **Savina O.** Presented on ‘Labyrinths of ophthalmogenetics’, at International Applied Science Conference-You need to see your childhood (Ukraine, Jul. 2021); Participated in the TV show Doctor Knows on ‘Heterochromia’ (Ukraine, Jul. 2021); Introduced ophthalmogenetics consultation at Dobrobut Medical Center; Identified ophthalmic genetic cases (18)- aniridia (6), retinitis pigmentosa (4), Stargardt macular dystrophy (3), macular dystrophy with central cone involvement (1), autosomal dominant occult macular dystrophy (1), Leber congenital amaurosis, type 4 (1), Leber hereditary optic neuropathy (1), X-linked retinitis pigmentosa (1); Performed genetic testing, identified mutations, provided genetic counseling.
58. **Taiwo O.** Completed training on the ‘Basic applications of medical genetics and genomics’ (NHGRI, Jun. 2021); Presented abstracts on ‘ How professional experience influences risk perception to COVID-19 and practice of dentistry in Nigeria’, ‘Socio-demographic variations in infection control and practices among dentists in Nigeria during the COVID-19 pandemic’, ‘Pattern of patients’ attendance at the Dental Clinic, Regional Center for Oral Health Research and Training Initiatives, Jos, Nigeria’, at 17th Annual Scientific Conference of the International Association for Dental Research (Nigerian Division, Jan. 2021); Invited speaker on ‘Writing paper for publication and plagiarism’, seminar of the Oral Health Advocacy Initiative (Nigeria, May 2021); Invited speaker on ‘Writing paper for Publication’, at the 25th Anniversary Edition of the UAE International Dental Conference and Arab Dental Exhibition (Dubai, Jul. 2021).
59. **Utumatwishima JN.**
60. **Wangi KYW.** Enrolled for Ph.D. at Penn State University, USA; Applied for a fellowship at Trinity International University, Illinois, USA; Promotor, National virtual seminar on Development of Genetic Nursing in Indonesia, presented on ‘The Ethical challenges in genetics and genomics nursing in Indonesia’ (Indonesia, Oct. 2021); Awarded Nursing bursary in Genomics and Healthcare, by Wellcome Genome Campus, UK (virtual, Jul. 2021); Developed undergraduate nursing course (3 credits) ‘Trend & current issues in nursing’ includes- Introduction to genetics & genomics medicine, newborn genetic screening & prenatal genetic screening; and nursing role in genetic counseling (Jan - May 2021).

2017 Batch

61. **Abad PJ.** Member, Scientific Advisory Board on ‘Interventions using genomics-based strategies towards enhanced nutrition recommendations’, Department of Science and Technology, Food and Nutrition Research Institution (Mar. 2021-present); Served on Global Panel: Genomic practice and research during a pandemic, World Congress of the International Society of Nurses in Genetics (Nov. 2020); Served as abstract reviewer for Congress of the International Society of Nurses in Genetics (Nov. 2021); Served as external reviewer on ‘Investigation of dilated cardiomyopathy in Vietnamese patients using Next-generation sequencing’, Center for Genetics and Reproductive Health, Ho Ch Minh City, Viet Nam; Co-organized, moderated three webinars on telegenetics cancer counseling – focused on genetic counselors making a difference (Nov. 2020), peer supervision in genetic counseling (Dec. 2020), and genetics education for health professionals (Feb. 2021).
62. **Avogbe PH.** Used resources from Summit to design a new course on genetics and genomics databases (Masters’ program), Polytechnic School at the University of Abomey-Calavi, Benin; Delivered new courses in biochemistry and molecular biology; Lectured on Genetic basis of disease, Next-generation sequencing methods, Cancer genetics and genomics to Graduate level students, University of Abomey-Calavi, Benin.
63. **Benítez-Cordero Y.** Delivered lectures in Genetic counseling to graduate level (MS) students, at National Center of Medical Genetics, Venezuela and at ECLAMC Congenital Malformations Conference on Most frequent congenital defects in pregnant adolescents: 10-year study in Cuba; Member of International Clearinghouse for Birth Defects Prevention & Research; Mentored residents (5) in clinical genetics; Evaluated genetic cases (20) of monogenic, chromosomal or multifactorial etiology; Codified cases (2105) for the Registry; Referred cases (5) for genetics analysis and medical management- congenital myopathy (3), spinal muscular atrophy (2).
64. **Casado PL.** Presented talk on ‘Periodontal disease: a risk factor for Sars-Cov-2 infection’, for community engagement in dental practice (virtual, 2021); Trained and supervised graduate students (2 PhD) for research in molecular biology and implant dentistry, at Fluminense Federal University, Brazil; Invited, Editor for Brazilian Dental Journal.
65. **Dhoro M.** Completed 2-year Certificate program in Medical Genetics and Genomics (NHGRI, Jun. 2021); Lectured on genetic basis of disease and gene therapy, diagnosis of genetic disorders, genetic counselling, genomic medicine and pharmacogenetics to Clinical Pharmacologists (medical doctors, postgraduates); Mentored graduate level students (MPhil, DPhil, Clinical pharmacology) on research projects and proposals related to HIV genetic diversity and drug resistance in TB and HIV coinfections; Appointed on Panel, for review of applications for Africa Rapid Grant Fund, Covid-19 Programme Call (2020); Reviewed, edited abstracts for University of Zimbabwe Annual Medical Research Day.
66. **Fatima SS.** Work impacted by COVID-19.
67. **Hitayezu J.** Completed Leadership and Management in Health Course, University of Washington’s Department of Global Health, E-Learning Program (Dec. 2020); Completed 2-year sub-specialty fellowship in Pediatric Emergency & Critical Care, University of Nairobi, Kenya (Jun. 2021); Completed 2- year Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Invited panel discussant at the UConn 4th Annual Global Health Symposium on Theme of the year: ‘Decolonizing Global Health’ (Mar. 2021).
68. **James O.**
69. **Lotz-Esquivel S.** Completed 2- year Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Member of the Medical Genetics and Metabolism Department, National Children’s Hospital; Provided care for pediatric patients with inborn errors of metabolism and

genetic conditions (4 patients/week); Member, Rare and Orphan Disease Multidisciplinary Clinic, Hospital San Juan de Dios (8 patients/week); Aided in transition of adolescents to adults' hospital and with transfer of knowledge between hospitals; Expanded prospective patients database for ongoing and future publications (~275 patients); Requested, obtained specialized treatment and laboratory tests for these patients, previously not available in National Health System (San José, Costa Rica); Knowledge acquired through Summit provided opportunity to work at National Referral Centers; Improved quality of life for patients and families by providing optimized genetic services using multidisciplinary approaches.

70. **Mahfoudh W.** Supervised graduate (MS) student in breast cancer genetics; Participated in H3Africa Data Management Plans Workshop (virtual, Oct. 2020); Attended webinars: 'Natural experiments in human genetics'(Apr. 2021), 'From precise microbial genomics to precision medicine' (Jun. 2021), 'CRISPR: New techniques and complex models'(Jun. 2021), 'Socializing the genome: Genomics, public trust and engagement'(Jun. 2021); Reviewed manuscripts in genetics for Journal of Translational Medicine and Cancer Management and Research.
71. **Mehrez MI.** Promoted to Assistant Professor (Feb. 2021); Presented on '3D imaging modalities and craniofacial anomalies', National Research Centre (Cairo, Feb. 2021); Participated in an online Master's course 'Logical Reasoning in Human Genetics' (Jan. 2021- present), offered by Columbia Global Centers, Tunisia and Institut Pasteur de Tunis; Reviewed a paper 'Immunofluorescence studies to dissect the impact of Cockayne syndrome A alterations on the protein interaction and cellular localization' for Journal of Genetic Engineering and Biotechnology (May 2021); Identified cases (7) clinically- GAPO syndrome (2), Silver Russel (1), Sotos (1), Rubinstein-Taybi (2), Richieri-Costa-Periera (1); Referred cases (4) for oral rehabilitation- GAPO (2), Mandibulacr dysplasia (1), Silver-Russel (1); Dental management of conditions resulted in improved quality of life and psychological well-being of patients.
72. **Mgasa A.** Completed the 2-year Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021).
73. **Nandal R.** Appointed, Teaching faculty for DNB-SS Cardiology course at Pandit Bhagwat Dayal Sharma University of Health Sciences, Rohtak, lectured MBBS students and MDs; Conducted weekly congenital heart diseases clinics; Used facial dysmorphism recognition tools in dysmorphic patients with congenital heart diseases; Established three generation pedigree history as integral tool for patients with cardiomyopathy, sudden death suspected channelopathy, hyperlipidemia and congenital heart disease patients; Identified syndromic children (>50), of these- Noonan syndrome (5), Marfan syndrome (1), William syndrome (1), Di Georgi syndrome (1); Referred all identified and suspect syndromic and dysmorphic patients (20) for genetic counselling; Updated congenital heart disease registry.
74. **Otaify GA.** Completed the 2-year Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Introduced, lectured on clinical genetics to undergraduate students, NewGiza University, Egypt; Summit helped in preparation of lectures; Submitted abstract on 'Five novel mutations in *FKBP10* and *PLOD2* expanding the molecular and phenotypic spectrum in 10 new patients with rare Bruck syndrome', to ASHG (2021); Conducted outpatient consultations, management and counseling of patients with genetic diseases ,including hereditary bone disorders (8-15 new cases/week), with follow-up for treatment or continued investigation till diagnosis (30); Summit helped in better approach for diagnosis of genetic cases, management plans for patients with metabolic disorders; Lectured on 'Clinical interpretation of genes and variants for disease causality in WES data' at Generation Clinics and Labs to expand genetics education and training in the private sector.

75. **Owusu M.** Lead, for diagnostic testing for COVID-19, at the Kumasi Centre for Collaborative Research in Tropical Medicine; Engaged in media interactions on COVID-19.
76. **Petlichovski A.** Completed 2-year Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Included Summit lectures and data in medical school and specialization curricula; Implemented, new Genetic Counseling Service Center at the Institute for Immunobiology and Human Genetics (Macedonia); Performed genetic testing, provided genetic counseling for patients (>20) with rare genetic disorders- Noonan syndrome (3), Rett Syndrome (2), phenylketonuria (1), congenital glaucoma (1), neonatal severe encephalopathy (1), ichthyosis vulgaris (1), SRTD-6 (1), familial Mediterranean fever (4), Gaucher disease (1), Marfan Syndrome (3), Stickler Syndrome (1), Ehlers – Danlos Syndrome (1), hereditary angio edema (3) and for couples (200) with recurrent spontaneous abortion.
77. **Roy S.** Passed, Advanced exam in International Council of Ophthalmology (Mar. 2021); Member, Royal College of Surgeon of Edinburgh and Fellow of International Council of Ophthalmology (2021); Received Fazlul Hoque Memorial Award from Ophthalmological Society of Bangladesh for research paper on ‘Outcome of periocular capillary hemangioma treated with oral propranolol’ (Dec. 2020); Speaker on ‘Retinoblastoma – our endless journey’ and ‘Retinoblastoma today’, at international webinars, organized by Young ophthalmological Society of Bangladesh (Jan. 2021) and L.V. Prasad Eye Institute (May 2021); Speaker on ‘Childhood ocular cancer’ an awareness TV program (Mar. 2021); Arranged CME program on ‘Black fungus infection of eye and recent management protocol’ at the Hospital; Mentored graduate level research students (BSc in Optometry), Institute of Community Ophthalmology, Chittagong University; Identified genetic disorder cases (73)- Retinoblastoma (40), Neurofibromatosis (5), Blepharophimosis syndrome (3), Thyroid ophthalmopathy (25) etc.; Treated patients (55), some were referred for medical management; Patients (42) were satisfied with their outcome.
78. **Sanhueza Díaz C.** Assistant Professor, for MS course ‘Techniques and Methodologies in Genetics’, at Faculty of Medicine, University of Chile; Delivered lectures on Chromosomopathies at University of Chile.
79. **Thakur N.** Developed next generation sequencing for prenatal testing using amniotic fluid for the first time in Nepal; Knowledge gained from Summit was key in validation and analysis; Optimized, performed preimplantation genetic screening (9), clinical exome testing (23) helped diagnose new rare genetic disorders (13); BRCA1/BRCA2 tests were conducted (15) at Nova International Diagnostics Nepal; Conducted Symposium on ‘Rare diseases’ on the occasion of ‘World Rare Disease Day-2021’, featured on (<https://rarediseaseday.org/friend/2506>) and national newspapers; Conducted symposium, presented on ‘The current state of Down Syndrome, its challenges and future perspectives’, on World Down Syndrome Day (Mar. 2021), Co-organizer for International Brain Research Organization-Asia Pacific Regional Committee Nepal School in association with Neuroscience Society of Nepal, on ‘Understanding Neuroscience and the Spectrum of Neurogenetic Disorders’ (Aug. 2021); Lab coordinator for COVID-19 RT-PCR tests (45,000), trained laboratory scientists (40) on RT-PCR techniques at Bir Hospital; Provided genetic counselling, diagnostic services to patients (>200) with genetic disorders- recurrent abortion (56), Down syndrome (22), Turner syndrome (8), chromosomal anomaly (34), rare genetic diseases (14), beta thalassemia (12), sickle cell anemia (28) and congenital birth defects- Duchenne muscular dystrophy (17), spinal muscular atrophy (7), metachromatic leukodystrophy (4), hemophilia A (6), familial mental retardation syndrome (5), Crouzon syndrome (3), disorder of sexual dysfunction (8), cleft lip and palate (8), at Bir Hospital; Referred patients and their families for appropriate supportive therapies for improvement in quality of life.

80. **Torres-Mejía G.** Presented two talks on ‘History of breast cancer policy in Mexico’ to strengthen advocacy for ‘united’ civil society organizations, organized by the Salvati Foundation, A.C. and ‘Breast cancer: Risk factors and early detection’, to Chronic Diseases INSP staff (Oct. 2020); Mentored graduate students (2 MS) on gene-environment interactions in breast cancer.
81. **Tuncay Yaylacioglu F.** Accepted to the ICO-NEI Ocular Genetics Fellowship Program (August 2021-2022); Completed 2-year Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Presented talk on ‘Challenges in mimicking missense variants’ at a Zebrafish Workshop on creating rare disease specific CRISPR-Cas9 platforms in zebrafish and ensuring their sustainability at Hacettepe University Zebrafish Unit (Mar. 2021).
82. **Vishnuposka SA.** Taught graduate courses ‘Human Genetics’, ‘School of Clinical Genomics’, ‘Human Physiology’ to undergraduates; Presented abstract on ‘Novel pathogenic variants in LHX3, LHX4 and GLI2 identified in pediatric patients with congenital hypopituitarism: From variant calling to variant testing’, at ENDO (virtual, Mar. 2021).
83. **Yousef YA.** Identified, diagnosed, and treated patients with retinoblastoma (22); Referred genetically tested (14) with familial and non-familial retinoblastoma for counseling and medical management; Informed patients negative for germline disease about extremely low risk of transmission to offsprings; Improved quality of life by alleviating stress.

(2016 Batch)

84. **Adeoye AM.** Joint appointment with Institute of Cardiovascular Diseases, & College of Medicine, University of Ibadan, Nigeria- Head Molecular Cardiology and Preventive Cardiology unit; Currently Chairman, Medical Advisory Committee /Director of Clinical Services, Research, and Training at the University College Hospital, Ibadan, Nigeria, and the first Teaching Hospital in Nigeria.
85. **Adeyemo AA.** Presented on ‘Genomic analysis of profound childhood hearing loss in the Yoruba population of Nigeria’, at 44th Association of Research in Otolaryngology MidWinter (virtual, Feb. 2021).
86. **Adeyemo WL.** Awarded Fellowship of the Academy of Medicine Specialties of Nigeria (FACadMedS).
87. **Ariani Y.** Appointed Lecturer, in Pediatrics program for dysmorphology and genetic risk assessment; Supervised postgraduate students (3) on genetics research; Speaker on ‘Host genetic susceptibility towards COVID-19’, at 6th Annual International Conference and Exhibition, Indonesian Medical Education and Research Institute (Nov. 2020); Speaker on ‘Dysmorphology in Inborn error of metabolism’, at 4th Indonesian Pediatric Nutrition and Metabolic Update (Indonesia, Apr. 2021); Presented 4 papers on LSD research at Science and Mathematics International Conference (Indonesia, Oct. 2020), published in American Institute of Physics Conference Proceedings (Apr. 2021); Developed prenatal genetic tests for alpha and beta thalassemia, and spinal muscular atrophy at Morula International IVF Center, Jakarta; Managed cases (~200 post-natal, prenatal and preimplantation) in collaboration with other pediatric subspecialists and OB/GYN Department.
88. **Deniz E.** Secured a postdoctoral fellowship at the Dept. of Oncology, Georgetown University, Washington DC (Jun. 2021); Supervised graduate students (2 MS) in genomics and genomic editing projects (Turkey, 2021); Speaker on ‘Identifying novel gene products conferring drug resistance in cancer chemotherapy’, at 8th Multidisciplinary Cancer Research Congress (Turkey, Jan. 2021).

89. **Ekure EN.** Completed 2-year Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Speaker on ‘Rheumatic heart disease in Nigeria: Insights from a community based survey in Lagos, Nigeria’, at Nigerian Cardiac Society Quarterly Seminar (Oct. 2020); Identified, diagnosed, treated pediatric congenital heart defect cases (149), at Lagos University Teaching Hospital, Nigeria; Counselling patients (131) that needed surgical management; All received medical treatment but only few (42) were surgically managed.
90. **Guio H.** INBIOMEDIC Research Center was certified by the Ministry of Health to test COVID-19 patients. 2021; Completed the Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Started a master course in Precision Oncology (Spain, 2021).
91. **Hussen DF.** Promoted to Associate Professor (Jan. 2021); Collaboration with NIH on manuscript on Syndromes (Williams syndrome, Turner syndrome and Rubinstein Taybi syndrome in diverse populations) aided the promotion; Obtained Diploma in Psychiatric Medicine (May 2021), to assist in psychiatric evaluation of patients with genetic disorders.
92. **Lallar M.** Speaker on ‘Basics of genetics: Non-Mendelian inheritance’ and ‘Genetic techniques: Cytogenetic and molecular’, a class series in association with Bangladesh Society of Medical Genetics (Bangladesh, Oct. 2020); Faculty in workshop on ‘Decision making in Genetics and PGT’, at 16th annual Fertility Conference, Indian Fertility Society (India, Dec. 2020); Delivered talk on ‘Genetic testing in infertile couples’, FOGSI Webinar (India, Jan. 2021); Faculty in workshop on ‘Panel discussion on aneuploidy screening’ in North Zone YUVA FOGSI (India, Apr. 2021); Appointed faculty for Certificate Programme in Genetic Counselling, Future Medical Academy and Sri Ramachandra Institute of Higher Education and Research, Chennai; Trained students (40) over one year; Presented cases at Society of Foetal Medicine, India; Provided genetic counselling to antenatal patients (>500)- aneuploidy screen risk (200), fetal malformations (150), teratogenic exposure (10), family history /previous child with genetic disease (140); Provided prenatal diagnosis to patients (>50) with various single gene and chromosomal disorders-Thalassemia (25), balanced translocation carriers (5), lysosomal storage disorders (10), spinocerebellar ataxia (2), IEM (8); Continued to maintain birth defect registry for the institution; Mobilized parents and social workers to form support groups for pediatric patients with genetic diseases.
93. **Malasa L.** Completed, Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Appointed, Assistant Lecturer, Department of Paediatrics and Child Health, Hubert Kairuki Memorial University, Dar es Salaam, Tanzania (Feb. 2021); Attended training on ‘Teaching methodology and instructional assessment skills’ conducted by the Centre for Educational Development in Health, Arusha, Tanzania (Oct. 2020); Attended the Massive Open Online Course on ‘Implementation research with a focus on infectious diseases of poverty’, organized jointly by the University of Pennsylvania School of Nursing, the University of Pennsylvania Center for Global Health, USA, and the WHO Special Programme for Research and Training in Tropical Diseases (Oct. 2020).
94. **Okafor FU.** Completed promotion formalities for Professorship in Nursing Science- awaiting external assessment; Supervised nursing student projects.
95. **Robledo Balbuena H.** Obtained PhD (Apr. 2021); Lectured on Genetic counseling at graduate level (MS), at National Center of Medical Genetics, Venezuela; Mentored residents (2) in clinical genetics. Evaluated genetic cases (19) of monogenic, chromosomal or multifactorial etiology; Registered new cases (4) in the Genetic Diseases' Register of the Pediatric Hospital Center, Havana.

96. **Seven M.** Lectured on ‘Human development and theoretical basis of nursing practice’ in undergraduate nursing program, University of Massachusetts Amherst, College of Nursing (2020-2021); Dissertation advisor for PhD student on ‘The experiences and needs of individuals with a variant of uncertain significance (VUS) on genetic tests for hereditary cancer syndromes: A grounded theory study’, University of Massachusetts Amherst, College of Nursing (2021); Presented seminar on ‘Social network characteristics to improve cancer related health behaviors’, University of Massachusetts, Institute of Diversity Sciences Health Group Seminar (Nov. 2020). Organizing Committee Member, Virtual Congress Wellcome Genome Campus and presented on ‘Genetic knowledge and practice among Turkish nurses: A literature review, nursing, genomics and healthcare’ (virtual, Jul. 2021).
97. **Sirisena ND.** Awarded the Senate Award for Research Excellence at the Inauguration Ceremony of University Annual Research Symposium (Jan. 2021); Awarded first runner-up poster for ‘Design and implementation of a novel pharmacogenetic assay for the identification on CYP2D6*10 genetic variant in a cohort of oestrogen receptor positive breast cancer patients’, presented at Annual Research Symposium of the Faculty of Medicine, University of Colombo (Dec. 2020); Introduced Continuing Professional Development course titled ‘Training Program in Clinical Genetics’ for medical professionals, Faculty of Medicine, Colombo (Jan./Jul. 2021); Diagnosed patients (>250) with various genetic disorders- hereditary cancer (33), dysmorphism/developmental delay (48), thrombophilia (72), haemato-oncological diseases (47), thalassemia (10), and congenital birth defects- Down syndrome (35), Turner syndrome (10), ambiguous genitalia (22); Offered genetic counseling, diagnostic services at the Human Genetics Unit, and referred patients for appropriate supportive therapies for improvement in quality of life.
98. **Tibrewal S.** As Head, expanded the genetic clinic at Dr Shroff’s Charity Eye Hospital (SCEH); Conducted regular educational classes (18) for the faculty and trainees at SCEH; Initiated monthly case presentations of clinico-genetic correlation scenarios (Nov. 2020); Prepared one-page ready reckoner for different genetic disorders (I-GENES, Snapshots in Ophthalmology); Received and extended 2021 ARVO Developing Country Eye Researcher Travel Fellowship; Speaker on ‘The role of genetics in pediatric eye disorders’, at Delhi Ophthalmological Society Conference (Mar. 2021); Panelist for ‘Mainstreaming genetic counseling’, at Annual meeting of Board of Genetic Counsellors in India (Jul. 2021); Examined pediatric cases (3364); Counselling patients (328) with help of the genetic counselor during COVID pandemic; Performed genetic testing in patients (34) and provided definitive genetic diagnosis- retinoblastoma (15,) retinitis pigmentosa (4), Leber’s congenital amaurosis (4), Leber’s hereditary optic neuropathy (2), Down’s syndrome (4), Aniridia with lens subluxation (1), dominant optic atrophy (1), isolated foveal hypoplasia (1), megalocornea microspherophakia (1), Stargardt’s disease (1).
99. **Uwineza A.** Completed Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Joined the G2MC Young Investigator Committee (Sept 2020); Participated in Rare Disease Group-ClinGen Workshop, H3Africa (Feb 2021); Participated Pre-Congress Symposium (Mar. 2021), for International Congress of Human Genetics to be held in 2022; Examined suspect cases of genetic disorders (672) referred from other departments at CHUK; Identified genetic disorders (495)- sex development disorder (27), chromosomal abnormalities (433), monogenic syndrome (35); Offered pre and post- test counselling to patients that received genetic tests; These measures alleviated stress, helped improve quality of life of patients and their families.