ADDENDUM

ISHGG FIFTH ANNUAL OUTCOME (October 2021- September 2022)

(*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. The 2016 Batch completed their 5-year follow-up in 2021 and hence this compilation only includes Batches 2017-2019.*

A. Publications: (Total - 221; 2019- 85; 2018- 72; 2017- 64)

2019 Batch

1. Abdel-Salam G, Girgis M, Eid M, **Sayed I**, Abdel-Hamid M. A homozygous loss of function variant in BICD2 is associated with lissencephaly and cerebellar hypoplasia. J Hum Genet. 2022. (Accepted).
2. Abdel-Salam GM, Afifi HH, Saleem SN, Gadelhak MI, El-Serafy MA, **Sayed IS**, Abdel-Hamid MS. Further evidence of a continuum in the clinical spectrum of dominant PIEZO2-related disorders and implications in cerebellar anomalies. Mol Syndromol. 2022; 1-8. doi: 10.1159/000523956.
3. Abdel-Salam GMH, Duan R, Abdel-Hamid MS, **Sayed ISM**, Jhangiani SN, Khan Z, Du H, Gibbs RA, et al. Expanding the phenotypic and allelic spectrum of SMG8: Clinical observations reveal overlap with SMG9-associated disease trait. Am J Med Genet A. 2022; 188(2):648-657.
4. Agarwal D, Hanafi NS, Khoo EM, Parker RA, Ghorpade D, Salvi S, Abu Bakar AI, Chinna K,…**Hussein N**, et al. Predictors for detecting chronic respiratory diseases in community surveys: A pilot cross-sectional survey in four South and South-East Asian low- and middle-income countries. J Glob Health. 2021; 11:04065.
5. **Akinmola OO**. The rare disease of Kallmann Syndrome in a 28yr old male: Need for scrutiny of diagnostic tools. J Hu Genet. 2022. (Submitted).
6. Alebiosu CO, Ayodele OE, Adefuye B, Ugwu C, Chinenye S, Olasoji HO, **Akinmola OO**, **Alli LA**, **Okunola OO**, et al. Chapters on 'Prenatal diagnosis', 'Genetic counselling' 'Mitochondrial genetics'. Essential Textbook of Medicine. Published by Uniosun Publishing Ltd. 2022; 951-961.
7. Alebiosu CO, Ayodele OE, Adefuye B, Ugwu C, Chinenye S, Olasoji HO, **Alli LA**, **Akinmola OO**, **Okunola OO**, et al. Chapters on 'Types of inheritance', 'Diagnosis of genetic diseases'. Essential Textbook of Medicine. Published by Uniosun Publishing Ltd. 2022; 934-949.
8. **Amarakoon G**, Wedasingha S, Sandakelum I, Chandrakumara J, Silva A. Myocarditis and severe neuromuscular paralysis following a suspected common krait (Bungarus caeruleus) envenoming in a child: A case report. Anuradhapura Medical Journal AMJ. 2022; 16(1):26-30.
9. **Amarakoon GGGT**, Mendis D, Senadeera N, Thewarapperuma C. A case report of a child with congenital Amegakaryocytic thrombocytopenia. 2022. (In preparation).
10. **Amarakoon GGGT**, Mendis D, Senadeera N, Thewarapperuma C. Case report of a child presented as HLH with underlying X-linked lymphoproliferative disease type 2, Sri Lanka. BMJ. 2022. (Submitted).
11. **Amarakoon GGGT**, Wijerathne NPKP, Chandrakumara WAJC, Wijayarathne HK. Clinical profile of Down's syndrome in a tertiary care centre -Sri Lanka. 2022. (In preparation).

12. **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. 2022. (In preparation).
13. Ayala-García JC, Lagunas-Martínez A, Díaz-Benítez CE, Orbe-Orihuela YC, …**Ortiz-Panozo E**, Bermúdez-Morales VH, Bahena-Román M, Cruz M, Burguete-García AI. High relative abundance of *Staphylococcus aureus* and serum cytokines are associated with cardiometabolic abnormalities in children. Metab Syndr Relat Disord. 2022; 20(5):303-311.
14. 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. 2022; 65(1):104-110.
15. **Bocoum A**, Coulibaly T, Ouologuem M, Cissé L, Diallo SH, Maiga BB, Dembélé K, Diallo S, Coulibaly SDP, et al. Clinical and genetic aspects of Huntington's disease in the Malian population. J Huntington's Dis. 2022; 11(2):195-201.
16. Chediak L, Bedlington N, Gadson A, Kent A, Khalek AA, Rosen L, Rust M, Shaikh MF…. **Wiafe SA**, et al. Unlocking sociocultural and community factors for the global adoption of genomic medicine. Orphanet J Rare Dis. 2022; 17:191.
17. Cheong AT, Lee PY, Ghazali SS, Salim H, **Hussein N**, Pinnock H, Liew SM, Hanafi NS, Abu Bakar AI, et al. Implementing asthma management guidelines in public primary care clinics in Malaysia. NPJ Prim Care Respir Med. 2021; 31(1):47.
18. **Chowdhury EH**, Dorji N. A case report on osteogenesis imperfecta in Samtse General Hospital, Bhutan. BHJ. 2022. (Submitted).
19. Danso KA, Akuaku RS, Young FNA, **Wiafe SA**. Pfeiffer syndrome type 3 with FGR2 c.1052C>G (p. Ser351Cys) variant in West Africa: A case report. Pan Afr Med J. 2021; 40:136.
20. de Castro-Hamoy LG, **Tumulak MJ**, Cagayan M, Sy PA, Mira N, Laurino MY. Attitudes of Filipino parents of children with Down syndrome on noninvasive prenatal testing. J Community Genet. 2022; 13(4): 411-425.
21. Dinh HN, **Luong LH**, Trung TN, Minh TMT, Tran TD, Nguyen TT. Successful total hip arthroplasty for patient with TCIRG1-associated autosomal recessive osteopetrosis. Int J Surg Case Rep. 2022; 95:107262.
22. Felipe-Dimog EB, Liang F-W, **Tumulak M-AJR**, Hsu M-T, Sia-ed AB, Dumalhin YJB. Roles and functions of rural health midwives in Cordillera administrative region: A qualitative pilot study. Acta Med Philipp. 2022. doi: https://doi.org/10.47895/amp.vi0.5326
23. Felipe-Dimog EB, **Tumulak MJ**, Laurino MY, Daack-Hirsch S, Silao C, Conaco M, Padilla CD, Estacio L R Jr. 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. 2022; 13(2):183-191.
24. **Guven Y**, Saraçoğlu HP, Aksakal SD, Kalaycı T, Altunoğlu U, Uyguner ZO, Eraslan S, Börklü-Yücel E, Kayserili Karabey H. Three Nance–Horan syndrome families diagnosed with three different diagnostic approaches. Mol Syndromol. 2022. (Submitted).
25. **Gyawali P**, Shrestha H, Panta V, Risal P, Gautam S. C-Reactive protein to albumin ratio among patients admitted to intensive care unit of a tertiary care hospital: A descriptive cross-sectional study. JNMA J Nepal Med Assoc. 2021; 59(244):1247-1251.
26. **Gyawali P**, Tamrakar D, Shrestha A, Shrestha H, Karmacharya S, Mallik V, Mattei J, Spiegelman D, Shrestha A, et al. Consumer acceptance and preference for brown rice - A mixed method qualitative study from Nepal. Food Sci Nutr. 2022; 10(6):1864-1874.

27. **Hussein N**, Henneman L, Kai J, Qureshi N. Preconception risk assessment for thalassemia, sickle cell disease, cystic fibrosis and Tay-Sachs disease. Cochrane Database Syst Rev. 2021; 10(10):CD010849.
28. **Hussein N**, Lee YK, John P, Azmi A, Ng CJ. What does it mean to be a carrier? Exploring experiences of thalassemia carriers in East Malaysia. 2022. (In preparation).
29. **Hussein N**, Liew SM, Hanafi NS, Lee PY, Cheong AT, Ghazali SS, Chinna K, Pnag YK, Kassim A, et al. Asthma control and care in public health clinic attenders in Malaysia: a cross-sectional study. Health Sci Rep. 2022. (In review).
30. **Hussein N**, Ramli R, Liew SM, Hanafi NS, Lee PY, Cheong AT, Ghazali SS, Mohd Ahad A, Patel J, et.al. Are the healthcare resources, organizational support and practice in asthma management optimal in the Malaysian primary care setting? Prim Care Respir Med. 2022. (Submitted).
31. Ismail I, Ng CJ, Lee PY, **Hussein N**. What influence the lack of effective contraception practice among multi-ethnic Asian women with diabetes? A qualitative study. PloS One. 2022. (Submitted).
32. Kantaputra P, **Guven Y**, Aksu B, Kalayci T, Doğan C, Intachai W, Olsen B, Tongsima S, Ngamphiw C, Noppakun K. Distal renal tubular acidosis, autoimmune thyroiditis, enamel hypomaturation, and tooth agenesis caused by homozygosity of a novel double-nucleotide substitution in SLC4A4. J Am Dent Assoc. 2022; 153(7):668-676.
33. Kantaputra P, **Guven Y**, Kalayci T, Özer PK, Panyarak W, Intachai W, Olsen B, Carlson BM, Praditsap O, et al. Expanding genotypic and phenotypic spectrums of LTBP3 variants in dental anomalies and short stature syndrome. Clin Genet. 2022; 102(1):66-71.
34. Kantaputra PN, **Guven Y**, Tripuwabhrut K, Adisornkanj P, Hatsadaloi A, Kaewgahya M, Olsen B, Ngamphiw C, Jatooratthawichot P, et al. Mutations in LRP5 and BMP4 are associated with mesiodens, tooth agenesis, root malformation and oral exostoses. Clin Genet. 2022 Jun 26. doi: 10.1111/cge.14183.
35. **Kars ME,** Ozcelik T. Molecular underpinnings of Mendelian and complex traits in the Turkish population as determined from a whole exome/genome screen of 3,599 individuals. 2022. (In preparation).
36. Koh WM, Abu Bakar AI, **Hussein N**, Pinnock H, Liew SM, Hanafi NS, Pang YK, Ho BK, Mohamedd, Isa S, et al. Sociocultural influences on asthma self-management in a multicultural society: A qualitative study amongst Malaysian adults. Health Expect. 2021; 24(6):2078-2086.
37. Kuguyo O, Tsikai N, Muradzikwa SC, **Mhandire K**, Nhachi C, Magwal T, Madziyire MG, Matimba A, Dandara C. Clinical and demographic characteristics of cervical cancer patients presenting at Parirenyatwa Hospital, Zimbabwe. Afr J Reprod Health. 2021; 25(6):99-109.
38. Kumar S, Lukose R, **Nair LS**, Nair MKC. Chapter on 'GDD: Common neurometabolic and other conditions, lysosomal storage disorders (LSD): Sphingolipidoses. Developmental Pediatrics, 2nd ed. Noble Vision (Medical Book Publishers). 2022; 446-472.
39. Kumuthini, J, Zick B, Balasopoulou A, Chalikiopoulou C, Dandara C, El-Kamah G, Findley L, Katsila T, Lee R,…**Monye H**, et al. The clinical utility of polygenic risk scores in genomic medicine practices: A systematic review. Hum Genet. 2022; 1-8.
40. Lagarde JB, Cauyan JM, Laurino M, **Tumulak MJ**, San Juan M, Ventura E. Qualitative inquiry on the experiences of Filipino women living with a sister with breast cancer: Insights for psycho-oncology. Asia Pacific Journal of Counselling and Psychotherapy. 2022; 13(1):68-8.
41. **Llamos-Paneque A**, Ñacato-Pachacama K, Lamar-Segura E, Garzón-Castro, Recalde-Báez M, Román-Naranjo María, Vásquez G. Cytogenetic variants in patients with Turner syndrome

diagnosed in a tertiary care hospital in Ecuador. Chilean Journal of Obstetrics and Gynecology. 2022. (Submitted).
42. **LLamos-Paneque A,** Pozo-Palacios J, Sancan-Byron, Garzón-Castro M, Lamar –Segura E, Liehr T. A double aneuploidy: an Ecuadorian case 45 X (7)/47XX+ 21 (3)/ 46, X, der (X)(p11.1-> q11.1)(1)/46,XX (1). 2022. (In preparation).
43. **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. 2022. (In preparation).
44. **Long LH**, Dinh HN, Tuyen NNT, Tam MTM, Trinh LK, Tung PS, Tien DT, Tran TN. Case report of mild TCIRG1-associated autosomal recessive osteopetrosis in Vietnam. Am J Med Genet A. 2022. https://doi.org/10.1002/ajmg.a.62897
45. Lukose R, Kumar S, Kunju PAM, **Nair LS**, Nair MKC. Chapter on 'GDD: Objectives in evaluation'. Developmental Pediatrics, 2nd ed. Noble Vision (Medical Book Publishers). 2022; 368-391.
46. Mangal V, Khurana H, **Mutreja D**, Yadav NK, Chakilam PK. Sudden painless loss of vision as the initial manifestation of acute myeloid leukemia: a case report. Hematol Transfus Cell Ther. 2022; S2531-1379(22)00043-8.
47. **Mhandire K**, Saggu K, Buxbaum NP. Immunometabolic therapeutic targets of Graft-versus-Host Disease (GvHD). Metabolites. 2021; 11(11):736.
48. Mostafa MI, Abdelkader MA, Abdelrahman MA, Hassib NF, **Sayed I**, Ahmed NE, Ibrahim MA, **Mehrez MI**, Khalil YM, et al. Frequent genetic disorders associated with missing teeth and re-visiting classification of anodontia: A retrospective study Middle East J of Med Genet. 2021; 10(2):95-100.
49. **Mushi TL**, Kashaigili H, Koestner D, Masenga A, Aron, Tulla M, Bakalemwa R, Schroeder K, Kayange N. Multidisciplinary approach for precocious puberty secondary to adrenal tumor. A case report of 3-years-old female at Bugando Medical Centre, Tanzania. BMJ Case Rep. 2022. (Submitted).
50. **Mutreja D**, Paliwal G, Mangal V, Biswas AK. Cold autoimmune hemolytic anemia with myelodysplastic syndrome: Not just an "Open and Shut case". Indian J Pathol Microbiol. 2022; 65(1):195-197.
51. **Nair LS**, Kumar S, Lukose R, Nair MKC. Chapter on 'GDD: Genetic approach'. Developmental Pediatrics, 2nd ed. Noble Vision (Medical Book Publishers). 2022; 420-436.
52. **Nair LS**, Lukose R, Kumar S, MKC Nair. Chapter on 'GDD: Chromosomal disorders (Down Syndrome)' verified. Developmental Pediatrics, 2nd ed. Noble Vision (Medical Book Publishers). 2022; 407-419.
53. Nair MKC, Kumar S, Lukose R, Kunju PAM, **Nair LS**. Chapter on 'GDD: Introduction'. Developmental Pediatrics, 2nd ed. Noble Vision (Medical Book Publishers). 2022; 359-368.
54. Nerakh G, Vineeth V S, Tallapaka K, **Nair L**, Dalal A, Aggarwal S. Microcephalic primordial dwarfism with predominant Meier-Gorlin phenotype, ichthyosis, and multiple joint deformities-Further expansion of DONSON Cell Cycle-opathy phenotypic spectrum. Am J Med Genet A. 2022; 188(7):2139- 2146.
55. Ngo HT, Nguyen HC, Do TD, **Luong LH**, Phan NT, Nguyen HT, Ngo LTH, Nguyen TT, Le TN, et al. Aortic valve reconstruction surgery using autologous pericardium: The experience in Vietnam. J Card Surg. 2022; 37(5):1233–9.
56. Ofagbor ER, Okediji PT, Okua KO, Omoyele OO, **Akinmola OO**. Interleukin 2 and 6 levels in Nigerian men with prostate cancer. Afr J Biomed Res. 2022; 25:13-17.

57. Okoh MP, **Alli LA**. Chapter on 'Recent progress in drug repurposing using protein variants and amino acids in disease phenotypes/disorders'. Drug Repurposing, Molecular Aspects and Therapeutic Applications. ISBN978-1-83969-959-7. 2022; 1-24.
58. Okoh MP, Singla RK, Soremekun O, Adejoh J, **Alli LA**, Shen B. Phytomedicine in disease management: In-silico analysis of the binding affinity of artesunate and azadirachtin for malaria treatment. Front. Pharmacol. 2021; 12:751032.
59. Olawoye O, **Monye HI**, Ibukun FA, Onakpoya OH, Yeye-Agba OO, Echieh CI, Obuoforibo BR, Bassey CN, Abdullah MI, et al. The impact of the COVID-19 pandemic on eye care services and training in Niger J Med. 2022; 31(3):280-284.
60. **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).
61. **Pirlog R**, Calin GA. KRAS mutations as essential promoters of lymphangiogenesis via extracellular vesicles in pancreatic cancer. JCI Insight. 2022. (In preparation).
62. **Pirlog R**, Chiroi P, Rusu I, Jurj AM, Budisan L, Pop-Bica C, Braicu C, Crisan D, Berindan-Neagoe I, et al. Cellular and molecular profiling of tumor microenvironment and early-stage lung cancer. Int J Mol. Sci. 2022; 23(10):5346.
63. **Pirlog R**, Drula R, Nutu A, Calin GA, & Berindan-Neagoe I. The roles of the colon cancer associated transcript 2 (CCAT2) long non-coding RNA in cancer: A comprehensive characterization of the tumorigenic and molecular functions. Int J Mol. Sci. 2021; 22(22):12491.
64. **Pirlog R**, Sirbu OM, Laquerrière A, Billaud-Porte E, Curey S, Lozouet M, Marguet F, Derrey S. Tumor-to-tumor metastases: Latent renal cell carcinoma discovered after elective surgical resection of a convexity meningioma. Neurochirurgie. 2022; 68(2):196-201.
65. Pop-Bica C, Ciocan CA, Braicu C, Haranguș A, Simon M, Nutu A, **Pîrlog R**, Berindan-Neagoe, I. Next-generation sequencing in lung cancer patients: A comparative approach in NSCLC and SCLC mutational landscapes. J Pers Med. 2022; 12(3):453.
66. Pozo-Palacios J, **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. doi: 10.3389/fpsyt.2021.716311.
67. Quispe B, **Paredes-Moscosso SR**, Villegas-Llerena C. Retinitis pigmentosa: Gene therapy approaches. Revista Peruana de Biología. 2022. (Submitted).
68. 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. 2022. (Accepted).
69. Rabie EA, **Sayed ISM**, Amr K, Ahmed HA, Mostafa MI, Hassib NF, El-Sayed H, Zada SK, El-Kamah G. Confirmation of a phenotypic entity for TSPEAR variants in Egyptian ectodermal dysplasia patients and role of ethnicity. Genes (Basel). 2022; 13(6):1056.
70. Ramli R, Hanafi NS, **Hussein N**, Lee PY, Ghazali SS, Cheong AT, Abu Bakar AI, Samad AA, Abdullah S, et al. Hajj health examination for pilgrims with asthma in Malaysia: An ethnographic study. J Glob Health. 2022; 12:04023.
71. Rasool IG, Zahoor MY, Ahmad I, **Iqbal M**, Shafquat S, Anjum AA, Shehzad W. Description of novel variants in consanguineous Pakistani families affected with intellectual disability. Genes Genomics. 2022. doi: 10.1007/s13258-022-01219-y.
72. Shah Z, Mustafa G, **IqbalM**, Qasim M, Abbas K, Umair M, Baig HMA. Bacterial pathogens affect the otitis media independently the association of genetic factor in indigenous population. EXCLI. 2022. (Submitted).

73. Shipra V, Baveja Puneet, Patnaik S, Nachiketa Thakur, TVSVGK Tilak, **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. 2022. (Accepted).
74. 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).
75. Tan HY, **Hussein N**, Lee YK, Malik TFA. The National School-Based Thalassemia Screening Program in Malaysia: A qualitative study exploring the experiences and views of adolescents. APJPH. 2022. (Submitted).
76. Taruscio D, Posada M, Lumaka A, Salvatore M, Carta C, Cellai LL, Ferrari G, Sciascia S,…**Wiafe SA**, et al. Undiagnosed diseases: needs and opportunities in 20 Countries participating in the Undiagnosed Diseases Network International. Orphanet J Rare Dis. 2022. (Submitted).
77. Thaneefa MT, **Amarakoon G**, Mendis D, Jasinge E, Hooper AJ, Burnett JR. Incidental diagnosis of LPL deficiency in an infant presenting with an acute respiratory infection. Clinica chimica acta. 2022; 529:1-3.
78. Thao NP, **Long LH**, The HN, Hoang DN, Tran TN. ELN missense variant in patient with midaortic syndrome. BMC Cardiovascular Disorders. 2022. (Accepted).
79. **Tumulak M-AJR**, Pascua AV, Jover EJM, Guerbo RJ, Canoy G MR, Laurino MY. Genetic counseling in the time of COVID-19: The Philippine experience with telegenetics. J Genet Couns. 2021; 30(5): 1285-1291.
80. Tvsvgk T, Handa A, Kumar K, **Mutreja D**, Subramanian S. Chemotherapy-Associated Pulmonary Toxicity-Case Series from a Single Center. South Asian J Cancer. 2021; 10(4):255-260.
81. **Yadav S**, Jain A, Gupta N, Gupta S. A general approach to antenatally detected birth defects. Chapter in NNF Clinical Protocols in Perinatology. Jaypee Brothers Medical Publishers. 2022; Chapter 5:35-39.
82. **Yadav S**, Kabra M, Gupta N, RC Mahumita, Chauhan S, Puri RD, Phadke S, Jana M. Molecular alterations at 11p15.5 locus in cases of isolated hemihyperplasia- Series of 32 cases. J Pediatr Genet. 2022. (Submitted).
83. **Yeşilçinar 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; 30(6):1512-1521.
84. **Yesilcinar İ**, Seven M, Şahin E, Calzone K. Genetics and genomic competency of Turkish nurses: A descriptive cross-sectional study. Nurse Educ Today. 2022; 109:105239.
85. **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. 2022. (Accepted).

2018 Batch

86. **Abdulkareem FB**, Khramtsova G, Odukoya LA, Badmos KB, Adedokun T, Rotimi O, Ibraheem A, Khramtsova A, Sveen L, et al. Consensus molecular subtyping of colorectal cancer by immunohistochemistry, an imperative for a resource limited setting: Report of a Nigerian study. Niger J Gastroenterol Hepatol. 2021; 13(2):33-39.
87. **Adewole OO**, Awopeju OF, Onakpoya UU, ErhaborGE. Treatment pathway and uptake among lung cancer patients in Nigeria. Niger J Health Sci. 2022. (Submitted).

88. **Adewole OO**, Omotosho BA, Awopeju OF, Aminu AA, Adedeji TA, Jiya E. Safety and effectiveness of atorvastatin with standard antituberculosis therapy. Int J Tuberc Lung Dis. 2022. (Submitted).
89. Afolabi BB, Babah OA, **Adeyemo TA**. Evidence-based obstetric management of women with sickle cell disease in low-income countries. 2022. (In preparation).
90. Afolabi BB, Babah OA, Akinajo OR, Adaramoye VO, **Adeyemo TA**, Balogun MR, Banke-Thomas A, Quao R, Olorunfemi G, et al. Intravenous versus oral iron for iron deficiency anaemia in pregnant Nigerian women (IVON): Study protocol for a randomized hybrid effectiveness-implementation trial. Trials. 2022. (In review).
91. Agarwal M, Kumar V, **Dwivedi A**. Diagnosis of 22q11.2 deletion syndrome in children with congenital heart diseases and facial dysmorphisms. MJAFI. 2022. (Accepted).
92. Aguirre-Quispe W, Valdez-Taboada M, Urbina-Ramírez L, Rivera- Valdivia A, Sarapura-Castro E, Montoya J, **Cornejo-Olivas M**. MELAS in Latin America. Rev Neuropsiquiatr. 2021; 84(4): 316-332.
93. Alatise OI, Dare AJ, Akinyemi PA, **Abdulkareem FB**, Olatoke SA, Knapp GC, Kingham TP, Kingham PT,…African Research for Oncology Colorectal Screening Early Detection Group. Colorectal cancer screening with faecal immunochemical testing: A community-based, cross-sectional study in average-risk individuals in Nigeria. Lancet Glob Health. 2022; 10(7): e1012-e1022.
94. Awotoye W, Comnick C, Pendleton C, Zeng E, Alade A, Mossey PA, Gowans LJJ, **Eshete MA**, Adeyemo WL, et al. Genome-wide gene-by-sex interaction studies identify novel nonsyndromic orofacial clefts risk locus. J Dent Res. 2022; 101(4):465-472.
95. Azhar Baig HM, Ansar M, Iqbal A, **Naeem MA**, Quinodoz M, Calzetti G, **Iqbal M**, Rivolta C. Genetic analysis of consanguineous Pakistani families with congenital stationary night blindness. Ophthalmic Res. 2022; 65(1):104-110.
96. Baluku J, Nabwana M, **Muttamba W**, Bongomin F. Tuberculosis contact tracing yield and associated factors in Uganda. BMC Pulm Med. 2022; 22:64.
97. Bbuye M, **Muttamba W**, Nassaka L, Nakyomu D, Taasi G, Kiguli S, Mayega RW, Mukose AD. Factors associated with linkage to HIV care among oral self-tested HIV positive adults in Uganda. HIV/AIDS (Auckl) 2022; 14:61–72.
98. Botre A, Kotecha U, Inamdar P, **Mistri M**, Shah N, Shah P, Sharda S. A partial de-novo hemizygous deletion in WDR45 gene in a male with infantile onset epilepsy and static encephalopathy. Clin Genet. 2021. (Submitted).
99. 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. 2022. (In review).
100. Castro-Suarez S, Guevara-Silva E, Caparó-Zamalloa C, Osorio-Marcatinco V, Meza-Vega M, Miller B, **Cornejo-Olivas M**. Knowledge and attitudes for the management of behavioral variant of frontotemporal dementia. Front Neurol. 2022; 12:786448.
101. Castro-Suarez S, Vishnevetsky A, **Cornejo-Olivas M**. The next step for neurology education in Peru: A call for subspecialty training. Rev Neuropsiquiatr. 2022; 85(2):83-85.
102. Chaudhary AK, Gholse A, Nagarajaram HA, Dalal AB, Gupta N, **Dutta AK**, Danda S, Gupta R, Sankar HV, et al. Ectodysplasin pathogenic variants affecting the furin-cleavage site and unusual clinical features define X-linked hypohidrotic ectodermal dysplasia in India. Am J Med Genet A, 2022; 188(3):788- 805.

103. Chaudhuri J, Biswas S, Gangopadhyay G, Biswas T, Datta J, Biswas A, Pandit A, Datta AK, Mukherjee A, **Dutta AK**, et al. Correlation of ATP7B gene mutations with clinical phenotype and radiological features in Indian Wilson disease patients. Acta Neurol Belg. 2022; 122(1):181-190.
104. **Cornejo-Olivas M**, Wu L, Noyce A. Disruption of mitochondrial complex i induces progressive parkinsonism. Mov Disord. 2022; 37(3):478.
105. **Cornejo-Olivas MR**, **Dueñas-Roque MM**, **Messaoud O**, **Kumar-Dutta A**, **Dwivedi A**, **Mistri Mehul**, **Nirmala S**, **Gyawali P**, Sok-Kun T, et al. Global perspectives on becoming established as a next generation practitioner of genomic medicine. G2MC. 2022. (In preparation).
106. **Daich Varela M**, Brooks BP. Reply. Ophthalmology. 2021; 128(11):e214-e215.
107. **Daich Varela M**, Georgiou M, Hashem SA, Weleber RG, Michaelides M. Functional evaluation in inherited retinal disease. Br J Ophthalmol. 2021; bjophthalmol-2021-319994.
108. **Daich Varela M**, Michaelides M. RDH12 retinopathy: Clinical features, biology, genetics and future directions. Ophthalmic Genet. 2022; 2:1-6.
109. Das G, Chauhan L, Kumar A, **Dwivedi A**. A survey manuscript- Knowledge, attitude, and practices towards genetic disorders amongst non-genetic clinicians of Armed Forces in India. J Genet Couns. 2022. (Submitted).
110. **Dutta AK**, Goswami K. Association of alpha 1 antitrypsin deficiency with COVID-19 mortality: Basis for clinical trials. Chapter in Frontiers of COVID-19. Springer Nature. 2021. Accepted.
111. **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. 2022. (Accepted).
112. **Dwivedi A**, Bhatt R, Kishore K. Changing indications of prenatal diagnosis in molecular era: experience of single centre in North India. Int J Reprod Contracept Obstet Gynecol. 2021; 10(10):3744-3748.
113. **Dwivedi A**, Kanitkar M, Singh D, Sharma M, Kalra S. Correlation of renal length with anthropometric and demographic data in healthy children: A prospective cross-sectional study. MJAFI. 2022. (Accepted).
114. **Dwivedi A**, Mandal K, Moirangthem A, Pandey H, Gupta N, Kabra M, Srivastava P, Kumar P, Phadke S. Yield of molecular testing in VHL and isolated VHL associated tumors in an Indian cohort. Lab Med. 2022. (Submitted).
115. Elsayed I, Martinez-Carrasco A, **Cornejo-Olivas M**, Bandres-Ciga S. Mapping the diverse and inclusive future of Parkinson's Disease genetics and its widespread impact. Genes (Basel). 2021; 12(11):1681.
116. **Eshete M**, Alade A, Abate F, Mossey PA, Awotoye WA, Busch T, Hailu A, Demissie Y, Butali A. Mutations in Van Der Woude families from Ethiopia. J Craniofac Surg. 2021; 33(2):e138-e140.
117. **Eshete M**. Pattern of orofacial clefts at a tertiary care hospital in ETHIOPIA. Ethiop J Health Sci. 2021; 31(6):1175-1184.
118. Gama MTD, Braga-Neto P, Rangel DM, Godeiro C Jr, Alencar R, Embiruçu EK, **Cornejo-Olivas M**, Sarapura-Castro E, Saffie Awad P et al. Autosomal recessive cerebellar ataxias in South America: A Multicenter Study of 1338 Patients. Mov Disord. 2022. doi: 10.1002/mds.29046.
119. Georgiou M, Awadh Hashem S, **Daich Varela M**, Michaelides M. Gene therapy in X-linked retinitis pigmentosa due to defects in RPGR. Int Ophthalmol Clin. 2021; 61(4):97-108.
120. Hui P, Kanjilal S, Biswas T, **Dutta AK**, Bhattacharjee D, Das MK. Congenital chloride diarrhea presenting as dilated fetal bowel loops. Pediatr Neonatol. 2022. (Accepted).

121. Jardim LB, Hasan A, Kuo SH, Magaña JJ, França M Jr, Marques W Jr, Camejo C, Santana-da-Silva LC,…**Cornejo-Olivas M**, et al. An exploratory survey on the care for ataxic patients in the American Continents and the Caribbean. Cerebellum. 2022. doi: 10.1007/s12311-022-01442-z.
122. Johns AL, Luquetti DV, Heike CL, Drake AF, **Roque MMD**, Hurtado-Villa P, Pachajoa H, Porras-Hurtado GL, Zarante I, et al. Parental reports of intervention services and prevalence of teasing in a multinational craniofacial microsomia pediatric study. J Craniofac Surg. 2021; 32(8):2687-2691.
123. Jost ZT, Sang CJ 3rd, **Lertwilaiwittaya P**, Chapman GD. Coronary artery aneurysm in Loeys-Dietz syndrome: a case report. Eur Heart J Case Rep. 2022; 6(6):ytac204.
124. Joy P, Madhuri V, Palocaren T, **Das S**, Abraham SSC, Korula S, Koshy B, Jose J, Chandran M, et al. Case report of a rare purine synthesis disorder due to 5-aminoimidazole-4-carboxamide ribonucleotide formyl transferase (AICAR) deficiency. Brain Dev. 2022; S0387-7604(22)00085-7.
125. Kobia FM, Maiti K, Obimbo MM, Smith R**, Gitaka J**. Potential pharmacologic interventions targeting TLR signaling in placental malaria. Trends Parasitol. 2022; 38(7):513-524.
126. Kotecha U, Asegaonkar P, Dongre M, **Mistri M**, Shah N, Shah P, Sharda S. Expanding the spectrum of ADNP- related disorder- antenatally diagnosed congenital diaphragmatic hernia and a novel de novo mutation in ADNP gene. Am J Med Genet A. 2022. (Submitted).
127. Kotecha U, **Mistri M**, Rayabarapu 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. 2022; 188(4):1323-1333.
128. Kotecha U, **Mistri M**, Shah N, Shah PS, Gupta VA. Bi-allelic loss of function variants in GOLGA2 are associated with a complex neurological phenotype: Report of a second family. Clin Genet. 2021; 100(6):748-751.
129. **Lertwilaiwittaya P**, Srinonprasert V. Cost effectiveness analysis of *BRCA1* and *BRCA2* testing in high-risk Thai woman. 2022. (In preparation).
130. Loesch D, Horimoto ARVR, Sarihan EI, Inca-Martinez M, Mason E, **Cornejo-Olivas M**, Torres L, Mazzwetti P, Cosentino C, et al. Polygenic risk prediction and SNCA haplotype analysis in a Latino Parkinson's disease cohort. Parkinsonism Relat Disord. 2022; 102:7-15.
131. Ma Y, Wang X, Shoshany N, Jiao X, Lee A, Ku G, Baple EL, Fasham J,…**Naeem MA**, et al. CLCC1 c. 75C>A mutation in Pakistani derived retinitis pigmentosa families likely originated with a single founder mutation 2,000-5,000 years ago. Front Genet. 2022; 13:804924.
132. **Mburu S**. Statistical thinking for researchers. Volume 1 & 2, OfficeMax, Nairobi. ISBN:978-9966-59-018-3; ISBN:978-9966-59-019-0. 2022. (Final Proofread).
133. McKnight D, Morales A, Hatchell KE, Bristow SL, Bonkowsky JL, Scott Perry M, Berg AT, Borlot F, Esplin ED,...**Dueñas-Roque MM**, the ELEVIATE Consortium. Genetic testing to inform epilepsy treatment management: Results from an international study of clinical practice. JAMA Neurol. 2022. (In review).
134. 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. AJM. 2022; 16(2).
135. **Messaoud O**, Dajani R. Women in Science: What are the barriers for women in senior career positions. Adv Appl Sociol. 2021; 11(10):492-499.
136. **Messaoud O**, **Dutta AK**, **Cornejo-Olivas MR**, Bhuiyan ZA. Editorial: Monogenic vs. Oligogenic reclassification. Front Genet. 2021; 12: 821591.

137. Mezzi N, **Messaoud O**, Mkaouar R, Zitouna N, Romdhane S, Abdessalem G, Charfeddine C, Maazoul F, Ouerteni I, et al. Spectrum of genetic diseases in Tunisia: Current situation and main milestones achieved. Genes (Basel). 2021; 12(11):1820.
138. Mkaouar R, Riahi Z, Charfeddine C, Chelly I, Boudabbous H, Dallali H, Bonnet C, Marrakchi J,…**Messaoud O**, et al. Alpha-mannosidosis in Tunisian consanguineous families: Potential involvement of variants in GHR and SLC19A3 genes in the variable expressivity of cognitive impairment. PLoS One. 2021; 16:e0258202.
139. Mugenyi L, Nsubuga NR, Wanyana I, **Muttamba W**, Tumwesigye MN, Nsubuga SH. Feasibility of using a mobile app to monitor and report COVID-19 related symptoms and people's movements in Uganda. PLoSONE. 2021; 16(11):e0260269.
140. Musa J, Kim K, Zheng Y, Qu Y, Joyce BT, Wang J, Nannini DR, Gursel DB,…**Abdulkareem FB**, et al. Accelerated epigenetic age among women with invasive cervical cancer and HIV-infection in Nigeria. Front. Public Health. 2022; 10:834800.
141. Naidoo S, **Gitaka J**, Suliman S, Baptista S, Oyedemi BM, Nepolo E, Enany S. Coronavirus disease 2019 diagnostics: Key to Africa's recovery. DNA Cell Biol. 2022; 41(1):30-3.
142. Ngamsom B, Iles A, Kamita M, Kimani R, Rodriguez-Mateos P, Mungai M, Dyer CE, Walter C, **Gitaka J**, et al. An integrated lab-on-a-chip device for RNA extraction, amplification and CRISPR-Cas12a-assisted detection for COVID-19 screening in resource-limited settings. medRxiv. 2022. https://doi.org/10.1101/2022.01.06.22268835
143. Nidoi J, **Muttamba W**, Walusimbi S, Imoko JF, Lochoro P, Ictho J, Mugenyi L, Sekibira R, Turyahabwe S, et al. Impact of socio-economic factors on tuberculosis treatment outcomes in north-eastern Uganda: A mixed methods study. BMC Public Health. 2021; 21:2167.
144. Ntaganda E, Mugeni R, Harerimana E, Ngoga G, Dusabeyezu S, Uwinkindi F, **Utumatwishima JN**, Mutimura E, Davila-Roman VG, et al. High rates of undiagnosed and uncontrolled hypertension upon a screening campaign in rural Rwanda: A cross-sectional study. BMC Cardiovasc Disord. 2022; 22(1):197.
145. Nwabudike VI. **Oluwarotimi AC**, Okoturo E, Ijogun A. Comparative study of the effect of single and multiple suture techniques on inflammatory complications following mandibular third molar surgery: A randomized prospective study at the Lagos State University Teaching Hospital, Ikeja. Pan Afr Med J. 2022. (In review).
146. Nwagha TU, Okoye HC, Udo CE, Yuguda S, Korubo KI, **Adeyemo TA**. Clinical audit of low dose prophylaxis programme for Nigerian children with haemophilia. West Afr J Med. 2022; 39(1):11-15.
147. Ochola L, Ogongo P, Mungai S, **Gitaka J**, Suliman S. Performance evaluation of lateral flow assays for coronavirus disease-19 serology. Clin Lab Med. 2022; 42(1):31-56.
148. Odukoya O, Nenrot D, Adelabu H, Katam N, Christian E, Holl J, Okonkwo A, Kocherginsky M,…**Abdulkareem FB**, et al. Application of the research electronic data capture (REDCap) system in a low- and middle-income country- experiences, lessons, and challenges. Health Technol (Berl). 2021; 11(6):1297-1304.
149. **Oluwarotimi AC**, Abah AA, Ogundana OM. Prevalence of medical conditions among dental patients in a South-West Hospital in Nigeria. AJOH. 2022. (Accepted).
150. Oluwole EO, **Adeyemo TA**, Osinaike B, Akintan P. Sickle cell disease: Caregiver's awareness and phenotype distribution among children presenting to children emergency of a tertiary hospital in Lagos, Nigeria. Niger J Paediatr. 2022; 49(1):56–62.
151. Orolu A, **Adeyemo TA**, Akanmu AS. Elevated homocysteine and crises state in patients with sickle cell anemia. J Clin Sci. 2022. (Accepted).

152. Osborne A, Manko E, Takeda M, Kaneko A, Kagaya W, Chan C, Ngara M, Kongere J,…Gitaka J, et al. Characterizing the genomic variation and population dynamics of *Plasmodium falciparum* malaria parasites in and around Lake Victoria, Kenya. Scientific reports. 2021; 11(1):19809.
153. Ray-Offor E, **Abdulkareem FB**, Jebbin NJ. Pit pattern analysis of colorectal polyps using Storz Professional Image Enhancement System (SPIES) endoscopy: A pilot study. Journal of the West African College of Surgeons. 2022. (Accepted).
154. Saengphatrachai W., **Lertwilaiwittaya P**, Limwongse C., Rattanabannakit C., Pho-Iam T, Senanarong V. Case series of C9orf72 hexanucleotide repeat expansion among dementia patient in the Memory Clinics at Siriraj Hospital. 2022. (In preparation).
155. Ver-or N, Iregbu C, **Taiwo O**, Afeleokhai I, Aza C, Adaji J, Margima C. Retrospective characterization of Noma cases found incidentally across Nigeria during outreach programs for cleft lips from 2011- 2020. Am J Trop Med Hyg. 2022. (In Press).
156. **Wangi KYW**. Pandemic and a bag of blood. Penn State Viral Imagination Covid-19. 2022. https://viralimaginations.psu.edu/written-submissions/pandemic-and-a-blag-of-blood/.
157. Zelada-Ríos L, Sarapura-Castro E, Solórzano-Palacios K, La Serna-Infantes J, Aguirre-Quispe W, Cosentino-Esquerre C, Urbina-Ramírez L, Torres-Ramírez L,…**Cornejo-Olivas M**. Dopa-Responsive dystonia (DRD): Systematic search in Latin America. Rev Neuropsiquiatr. 2022; 85(1): 38-54.

2017 Batch

158. **Abad PJ**, Sur ALD. Nursing engagement in genetics and genomics: A developing country's perspective. Int Nurs Rev. 2022. doi: https://doi.org/10.1111/inr.12751
159. Abdel-Hamid MS, Elhossini RM, **Otaify GA**, Abdel-Ghafar SF, Aglan MS. Osteoporosis pseudoglioma syndrome in four new patients: Identification of two novel LRP5 variants and insights on patients' management using bisphosphonates therapy. Osteoporos Int. 2022; 33(7):1501-1510.
160. Abdullahi MAS, Olasoji HO, **James O**, **Adeyemo WL**. Comparison of the pattern of nasolabial measurements among complete unilateral cleft lip/palate and controls at a tertiary hospital in Northeast, Nigeria. Niger J Oral Maxillofac; 2022; 7 (1):26-31.
161. Abid F, Rubab Z, **Fatima SS**, Qureshi A, Azhar A, Jafri A. In-silico analysis of interacting pathways through KIM-1 protein interaction in diabetes nephropathy. BMC Nephrol. 2022. (Accepted).
162. Acquah-Gyan E, Acheampong PR, Mohammed A, Adjei TK, Agyapong E, Twumasi-Ankrah S, Sylverken A, **Owusu M**, Owusu-Dabo E. User experiences of a mobile phone-based health information and surveillance system (mHISS): A case of caregivers of children under-five in rural communities in Ghana. Plos One. 2022; 17(1): e0261806.
163. Adamson OO, **James O**, Adekunle A, Agbogidi FO, Osewa O, Babatunde A, **Adeyemo WL**, Ladeinde A Ogunlewe MO. Outcome of speech intervention in patients after cleft palate repair. Niger J Oral Maxillofac. 2022; 7(1):8-14.
164. Aiemjoy K, Seidman JC, Saha S, Munira SJ, Sajib MSI, Al Sium SM, Sarkar A, Alam N,…**Owusu M**. Estimating typhoid incidence from community-based serosurveys: a multicohort study. Lancet Microbe. 2022; 3(8):e578-e587.
165. Akhan Guzelcan E, **Yaylacioglu Tuncay F**. Chapter on 'The role of medical record systems and biobanks in hereditary eye diseases'. Türkiye Klinikleri Journals- Ocular Genetics. 2021; 103-110.

166. Akowuah E, Acheampong G, Ayisi-Boateng NK, Amaniampong A, Agyapong FO, Kamasah JS, **Owusu M**. Comparable detection of SARS-CoV-2 in sputum and oropharyngeal swab samples of suspected COVID-19 patients. COVID. 2022; 2:858-866.
167. Ali MK, Kadir MM, Gujral PA, **Fatima SS**, Iqbal R, Sun YV, Narayan KMV, Ahmad S Obesity associated metabolites in relation to type 2 diabetes risk: A prospective nested case-control study of the CARRS cohort. Diabetes Obes Metab. 2022. (Accepted).
168. **Avogbe PH**, Sanni A. Dyslipidemia and associated cardiovascular risk factors among non-diabetic taxi-motorbike drivers working in Cotonou, Benin. Biomed J Sci & Tech Res. 2022; 43(5):34952-34959.
169. **Avogbe PH**, Sanni A. Elevated white blood cell count is associated with an increased risk of insulin resistance in non-diabetic taxi-motorbike drivers working in Cotonou, Benin. Afr. J. Biochem. Res. 2022. (Accepted).
170. **Avogbe PH**, Sanni A. Hyperuricemia and associated cardiometabolic factors: A cross-sectional study in taxi-motorbike drivers working in Cotonou, Benin. Avicenna J Med Biochem. 2022; 10(1):46-51.
171. **Avogbe PH**, Sanni A. High-level circulating total calcium and low phosphate as predictors of insulin resistance among non-diabetic taxi-motorbike drivers living and working in Cotonou, Benin. Open Journal of Endocrine and Metabolic Diseases OJEMD. 2022; 12 (6):123–34.
172. Biswas S K, **Roy S**, Akbar US. Surgical management of a rare case of late onset capsular block syndrome (CBS). BIJCROO. 2022; 1(1):16-18.
173. Biswas SK, **Roy S**, Alam M. Neonatal herpes zoster ophthalmicus: Two rare cases. BIJCROO. 2022; 1(1):11-13.
174. Biswas SK, **Roy S**, Alam M. Outcome of chemical tattooing in corneal scars in a tertiary eye care center of Bangladesh. Arc Org Inorg Chem Sci. 2022; 5(5):768-772.
175. Biswas SK, **Roy S**, Khan SH, Akhter KT. Phacoemulsification in white cataract: Demographic profile, associated difficulties, and visual outcomes. International Journal of Innovative Science and Research Technology IJISRT. 2022; 7(4):22-26.
176. Brnjarchevska Blazhevska T, Babačić H, Sibinovska O, Dobrevski B, Kirijas M, Milanovski G, Arsov T, **Petlichkovski A**. A single dose of BNT162b2 vaccine elicits strong humoral response in SARS-CoV-2 seropositive individuals. Allergy. 2022; 77(1):296-298.
177. Byakika-Kibwika P, Sekaggya-Wiltshire C, Semakula JR, Nakibuuka J, Musaazi J, Kayima J, Sendagire C, Meya D,…**Wayengera M**, et al. Safety and efficacy of hydroxychloroquine for treatment of non-severe COVID-19 among adults in Uganda: A randomized open label phase II clinical trial. BMC Infect Dis. 2021; 21(1):1218.
178. 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; 32(5):55-66.
179. Cholakovska VC, Vlashki E, Kochova M, Stefanovska VV, **Petlichkovski A**. Leptin, obesity parameters, and atopy among children with asthma. Pril. Makedon Akad Nauk Umet Odd Med Nauki. 2021; 42(2):79-88.
180. Diakou KI, Mitsis T, Pierouli K, Papakonstantinou E, Bongcam-Rudloff E, **Wayengera M**, Vlachakis D. Ebola virus disease and current therapeutic strategies: A Review. Adv Exp Med Biol. 2021; 1339:131-137.
181. Elfalah M, AlNawaiseh T, Atoum D, AlKhassawneh A, Mohammad M, AlNawaiseh I, **Yousef YA**. Improving medical students' awareness about retinoblastoma: A practical strategy. Clin Ophthalmol. 2022; 16:1807-1814.

182. Elfalah M, Mohammad M, Toro MD, Abu-Yaghi N, Rejdak R, **Yousef YA**. Anterior ocular biometrics as measured by ultrasound biomicroscopy. Healthcare. 2022; 10 (7):1188.
183. Ertop M, **Yaylacioglu Tuncay F**, Aribas Y, Ozdemir HB, Gurelik G, Ozdek G. Evaluation of central and peripheral retinal vascular changes in the fellow eyes of patients with unilateral retinal vein occlusions. Turk J Ophthalmol. 2022. (Submitted).
184. Hoque F, Nuruddin M, **Roy S**. Bilateral congenital microblepharon associated with Down's Syndrome – A Case Report. BIJCROO. 2022; 1(1):8–10.
185. **James O,** Sabo VY, Adamson OO, Otoghile B, Adekunle AA, **Adeyemo WL**, Ladeinde AL, Ogunlewe MO. Presentation and management of atypical orofacial cleft clefts: A single-institution experience for a 13-year period. Cleft Palate Craniofac J. 2021; 10556656211055012.
186. Johnson DL, Korf BR, Ascurra M, El-Kamah G, Fieggen K, de la Fuente B, Mahmood S, ,…**Thakur N**, Vargas RP. Chapter on 'Preparing the workforce for genomic medicine: International challenges and strategies'. Genomic and Precision Medicine in Clinical Practice, vol- Medicine Skills and Competencies. Elsevier. 2022; 8:131-139.
187. Khairat R, Elhossini R, Sobreira N, Wohler E, **Otaify G**, Mohamed AM, Raouf ER, **Sayed I**, Aglan M, et al. Expansion of the phenotypic and mutational spectrum of Carpenter syndrome. Eur J Med Genet. 2022; 65(1):104377.
188. Kumar W, Martins RS, Anjum N, **Fatima SS**. Ischemia modified albumin as a useful marker for diagnosis and management of diabetic retinopathy. Pak J Med Sci. 2022; 38(4Part-II):1043-1047.
189. 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; 37(12):954-961.
190. **Mahfoudh W**, Bannour I, Gabbouj S and Zakhama A. Association of TNF-α-308 G/A polymorphism with severity of COVID-19. 2022. (In preparation).
191. Martin M, Fernanda, **Díaz S Catherine**, & Mira O, Magdalena. Tatton-Brown-Rahman Syndrome: Case report of a variant in the DNTM3A gene not previously described associated with the syndrome. Andes pediatrica. 2022; 93(4).
192. Mohamed AM, Kamel AK, Eid MM, Eid OM, Mekkawy M, Hussein SH, Zaki MS, Esmail S, …, **Otaify GA**. Chromosome 9p terminal deletion in nine Egyptian patients and narrowing of the critical region for trigonocephaly. Mol Genet Genomic Med. 2021; 9(11):e1829.
193. Morang'a CM, Ngoi JM, Gyamfi J, Amuzu DS, Nuertey BD, Soglo PM, Appiah V, Asante IA,…**Owusu M**. Genetic diversity of SARS-CoV-2 infections in Ghana from 2020-2021. Nat Commun. 2022; 13(1):2494.
194. Nuruddin M, **Roy S**, Hoque F. Pegylated interferon-alpha-2a for the treatment of ocular surface squamous neoplasia. Oman J Ophthalmol. 2022; 5:81-84.
195. Osei-Boakye F, Addai-Mensah O, **Owusu M**, Saasi AR, Appiah SK, Nkansah C, Wiafe YA, Debrah AY. Effect of pulmonary tuberculosis on natural anticoagulant activity in therapy-naïve Ghanaian adults: A case-control study. J Immunoassy Immunochem. 2022; 43(3):271-287.
196. **Otaify GA**, Abdel-Hamid MS, Hassib NF, Elhossini RM, Abdel-Ghafar SF, Aglan MS. Bruck syndrome in 13 new patients: Identification of five novel FKBP10 and PLOD2 variants and further expansion of the phenotypic spectrum. Am J Med Genet A. 2022; 188A(6):1815–1825.
197. **Owusu M**, Nkrumah B, Mensah EK, Lamptey J, Acheampong G, Sambian D, Sylverken A, Emery S, Robinson LM, et al. Surveillance and laboratory collaboration in response to an

outbreak of *Vibrio parahaemolyticus*, *Plesiomonas shigelloides*, and *Aeromonas hydrophila* in Sekondi-Takoradi, Ghana: A case series. J Med Case Rep. 2022;1 6(1):53.
198. Penon-Portmann M, Westbury SK, Ling L, Pluthero FG, Liu RJY, Yao HHY, Geng RSQ, Warner N, …**Lotz-Esquivel S**, et al. Platelet VPS16B is dependent on VPS33B expression, as determined in two siblings with arthrogryposis, renal dysfunction and cholestasis (ARC) syndrome. J. Thromb Haemost. 2022; 20(7):1712-1719.
199. Quinelato V, Bonato LL, Vieira AR, Granjeiro JM, Menezes K, Borojevic R, **Casado PL**, Calasans-Maia JA, Tesch R. PAX7 gene polymorphism in muscular temporomandibular disorders as potentially related to muscle stem cells. BMC Musculoskelet Disord. 2021; 22:959.
200. 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: Sci Rep. 2022; 12(1):1837.
201. Rosman SL, Daneau BC, Rutare S, McCall N, Monuteaux MC, Unyuzumutima J, Uwamaliya A, **Hitayezu J**. The impact of pediatric early warning score and rapid response algorithm training and implementation on interprofessional collaboration in a resource-limited setting. PLoS ONE. 2022; 17(6):e0270253.
202. **Roy S**,…Global retinoblastoma study group. Global retinoblastoma outcome study: prospective analysis of 4,064 patients from 149 countries. Lancet Glob. Health. 2022. (Submitted).
203. **Roy S**, Kader S. Phenotypic analysis of a rare case of '3 MC Syndrome'. International Journal of Ophthalmology and Optometry. 2022. (Submitted).
204. **Roy S**, Nuruddin M, Hoque F Nuruddin M. A rare case series of ankyloblepharon filiforme adnatum. International Journal of Innovative Science and Research Technology (IJISRT). 2022; 5(5):1279-1281.
205. **Roy S**, Nuruddin M, Hoque F. Adult-onset retinoblastoma- Two rare cases. Mymensingh Medical Journal. 2022. (Accepted.)
206. **Roy S**, Nuruddin M, Osmani M. Community screening and community participation along with hospital based multidisciplinary treatment can be the key factors for both early detection, life and globe salvage of retinoblastoma patients in developing countries- Our experience. BIJCROO. 2022; 1(1):3-7.
207. **Roy S**, Nuruddin M. Periocular capillary hemangioma treated with low dose oral propranolol - presentation and outcome of 30 patients. Arch. Pathol. Clin. Res. 2021; 5:037-041.
208. **Roy S**, Osmani M, Huque F. Field level activity along with hospital-based management of retinoblastoma and its impact on community. Horizon. 2022. (Submitted).
209. **Roy S**. Generating awareness and a planned multidisciplinary treatment approach can save both the sight and life in retinoblastoma in developing countries. Arch Clin Ophthalmol [Commentary]. 2021; 1(1):27-29.
210. **Roy S**,…Global Retinoblastoma study group. Risk factors associated with abandonment of care in retinoblastoma: Analysis of 692 patients from 10 countries. Br J Ophthalmol. 2022. (Accepted).
211. Sabo VY, **James O**, Adamson OO, Otoghile B, Adeyemo WL, Ladeinde AL, Ogunlewe MO. The impact of COVID-19 pandemic on cleft care services at a tertiary health facility in Nigeria. FACE. 2021; 2(4):386-392.
212. Sedó-Mejía G, **Lotz-Esquivel S**, Monge-Ortega O, Pino-García C. Clinical profile of patients with chronic urticaria in a tertiary care hospital of the social security system in Costa Rica. Rev Alerg Mex. 2021; 68(4):225-32.

213. Shahriar NA, **Roy S.** Timely diagnosis of Weill-Marchesani syndrome can preserve vision and prevent complication: A Case Report. BIJCROO. 2022; 1(1):26-28.
214. Suleiman IK, **James O**, Olasoji HO. Effect of dexamethasone, ketoprofen and cold compress on postoperative sequelae in mandibular third molar surgery: A randomized clinical trial. Advances in Oral and Maxillofacial Surgery. 2022; 5(January- March):100211.
215. Tomar AS, Finger PT, Gallie B, Kivelä TT, Mallipatna A, Zhang C, Zhao J, Wilson MW, Brennan RC,…**Yousef YA**. Metastatic death based on presenting features and treatment for advanced intraocular retinoblastoma: A multicenter registry-based study. Ophthalmology. 2022; 129(8):933-945.
216. Tomar AS, Finger PT, Gallie B, Kivelä TT, Mallipatna A, Zhang C, Zhao J, Wilson MW, Brennan RC,…**Yousef YA**. High-risk pathologic features based on presenting findings in advanced intraocular retinoblastoma: A multicenter, international data-sharing American Joint Committee on cancer study. Ophthalmology. 2022; 129(8):923-932.
217. **Wayengera M**, Severity of adverse events following COVID-19 vaccination as a predictive test for extent of hypersensitivity to future break through SARS-CoV-2 infections. Editorial. J Antivir Antiretrovir. 2022. (Accepted).
218. **Yaylacioglu Tuncay F**, Reeves M, Guan B, Ullah E, Tumminia S, Hufnagel R. Variant spectrum of Norrin/β-catenin signaling pathway genes in the eyeGENE® Familial Exudative Vitreoretinopathy cohort. IOVS. 2022. (Submitted).
219. **Yousef YA**, Mohammad M, Al-Nawaiseh I, Mahafza H, Halalsheh H, Mehyar M, AIJabari R, AlRawashdeh K, Jaradat I, et al. Retinoblastoma and uveal melanoma in Jordan: Incidence, demographics, and survival (2011-2020). Ophthalmic Genet. 2022. (Accepted).
220. **Yousef YA**, Mohammad M, AlNawaiseh I, AlJabari R, Toro MD, Gharaibeh A, Rejdak R, Nowomiejska K, Zweifel S, et al. Ultrasound biomicroscopy measurements of the normal thickness for the ciliary body and the iris in a Middle East population. Clin Ophthalmol. 2022; 16:101-109.
221. **Yousef YA**, Qaddoumi I, Al-Nawaiseh I, Mohammad M, AlRimawi D, Toro MD, Zweifel S, Rejdak R, Nazzal R, et al. A proposal for future modifications on clinical TNM staging system of retinoblastoma based on the American Joint Committee on Cancer Staging Manual, 7th and 8th Editions. J Cancer. 2022; 13(4):1336-1345.

B. Grants: (Total- 58; 2019- 16; 2018- 25; 2017- 17)

2019 Batch

1. **Alli LA** (PI), Mokuolu OA, Olorunniji F, Nnodu OA, Oyakanmi N, Na'Allah A. Artificial Intelligence-Guided identification, and experimental characterization of *Plasmodium* targets for development of novel antimalarial drugs. 2022. (Nigerian Tertiary Education Trust Fund, Thematic Area: Science, Engineering, Technology and Innovation (SETI) Research Category; Submitted).
2. Buzoianu AD (PI), Neagoe I, **Pirlog R**. Increasing the performance of scientific research, supporting excellence in research and innovation in medicine-progress. 2021. (Ministry of Research, Innovation and Digitalization; Received).
3. Covaliu B, Neagoe I, **Pirlog R**. Healthy Start-ups – SUS. 2022. (Ministry of Investments and European Projects; Received).

4. Değirmenci T (PI), **Yesilcinar İ**. Pregnant women's genetic literacy and affecting factors. 2022. (The Scientific and Technological Research Council of Turkey, TUBITAK; Accepted).
5. **Gyawali P** (PI), Basnet S. Diagnostic utility of highly sensitive cardiac troponin in detecting acute coronary syndrome. 2022. (University Grant Commission; Accepted).
6. **Gyawali P** (PI), Shrestha R. Prevalence of holo-transcobalamin deficiency among type 2 diabetic patients under metformin therapy. 2021. (Nepal Health Research Council Provincial Research Grant; Received).
7. Halkoaho A (PI), Salomaa M, Smolander N, Tonkin E, Limoges J, Calzone K, Angelo D, Hegarty J, **Yesilcinar İ**, et al. Genomic nursing competence network. 2022. (COST Action-European Cooperation in Science and Technology; Submitted).
8. **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. 2022. (University Malaya Specialist Centre (UMSC) Care Fund; Submitted).
9. **Luong LH**, Lam HT, Dan VB, Duong QS. Increase awareness and establish a core laboratory in diagnosis of hereditary angioedema (HAE) and other hereditary auto-inflammatory disorders. 2022. (Ministry of Health, Vietnam; Submitted).
10. Maceda EB, Abadingo M, **Tumulak MJR**, Cases RK, Asor BB. Social media content analysis of public Glucose-6-Phosphate deficiency (G6PD) Facebook Groups. 2022. (Newborn Screening Reference Center; Received).
11. Neagoe I (PI), **Pirlog R** (Co-PI). Strategic inter-university cooperation to achieve higher educational quality by improving research abilities of Ph.D. students. 2022. (EEA and Norway Grants; Received).
12. Neagoe I (PI), **Pirlog R**. Increasing the research capacity of UMF Iuliu Haţieganu Cluj Napoca, by developing a CLOUD-type infrastructure connected to global information resources. 2022. (Ministry of Research, Innovation and Digitalization; Received).
13. **Okunola OO**. Heritability and phenotypic characteristics in Diabetic nephropathy. 2022. (International Society of Nephrology; Submitted).
14. **Paredes-Moscosso SR** (PI), Villegas-Llerena C, Buleje S, Guevara-Fujita ML, Acosta O, Dueñas M, Ledesma Y, **Tumulak MK**, Padilla C, et al. Ancestry and interpretation of genetic variants found from whole exome sequencing in Peruvian patients with hereditary cancer: Advances in genomic medicine in Peru. 2022. (Universidad de San Martín de Porres; Received).
15. Torres-Atencio I, Islas P, Labraña J, **Paredes-Moscosso SR**, Luchessi L, Echeverría-King L. Latin American perspectives on science diplomacy. 2022. (CYTED; Submitted).
16. 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

17. Afolabi BB (CI), **Adeyemo TA** (Co-I), Babah OA (Co-I), Balogun M (Co-I). Intravenous ferric carboxymaltose versus oral ferrous sulphate for the treatment of postpartum anaemia in Nigerian women (IVON-PP): An open labelled randomized controlled trial alongside an implementation study. 2022-2025. (Bill and Melinda Gates Foundation; Received).

18. Alatise O (PI), Kingham P (Co-PI), **Abdulkareem FB** (Co-I). Nigerian cancer research training program. 2021. (OAK Foundation, Switzerland; Continued).
19. Chinnaswamy S (PI), **Dutta AK** (Co-PI). A molecular epidemiological study to understand trained innate immunity in the context of COVID-19 pandemic in India. 2022. (Welcome Trust - Dept. of Biotechnology, Government of India; Submitted).
20. Ghosh T (PI), **Dutta AK** (Co-PI). The association of TMPRSS6 polymorphism rs855791, rs2413450 and rs4820268 with iron and hematocrit parameters among women of reproductive age group in rural population of Nadia district, West Bengal. 2022. (Indian Council of Medical Research; Accepted).
21. **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. 2022. (NIH; Received).
22. **Gitaka J** (PI), Vatish M, Sadovsky Y, Smith R. The interactome defining *Plasmodium falciparum* and components of the maternal-placental interface that underlie placental malaria. 2022. (Wellcome Trust; Submitted).
23. Giuseppe Tosto, Margaret Pericak-Vance, Nilton Custodio, **Cornejo Olivas M** (Co-I). Global Latinos Sequencing Study for Alzheimer's disease. 2022. (NIA/NIH; Submitted).
24. Jaja C (PI), **Edem-Hotah J** (Co-PI), Shepherd J. Expanding access to sickle cell disease care in Sierra Leone. The EASEL Pilot Implementation Project. 2021. (Formabridge; Approved).
25. 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; Continued).
26. Kingham P (PI), Olusegun a (Co-PI), **Abdulkareem FB** (Site-PI). Expanding cancer research capacity in Nigeria with Team Science. 2022. (NIH; Received).
27. Kirenga B (PI), Byakiika P, Kiwanuka N, Ocan M, Nakibuuka J, Okia J, **Muttamba W**, Kyosiimire J, Bakamutumaho B, et al. A multicenter, multiple arms, multiple stage omni adaptive, randomized trial to evaluate the safety and efficacy of natural/herbal investigational therapeutics for the treatment of acute respiratory viral infections including SARS-CoV2 in Uganda. 2022. (Government of Uganda, Ministry of Science, Technology and Innovation; Received).
28. Kirenga B, Byakiika P, Kyobe H, **Muttamba W**, **Wayengera M**, Kayongo A, Mugenyi L. Evaluation of effectiveness and determinants of effectiveness of China manufactured and other COVID-19 vaccines in Uganda. 2021. (China Centers for Disease Control and Prevention; Received).
29. Klapperich C (PI), **Gitaka J** (Co-PI), Srinivas R. Development of molecular point of care diagnostics for placental malaria, *Neisseria gonorrhoea* and *Chlamydia trachomatis*. 2022. (NIH; Submitted).
30. **Mburu S** (PI), **Gitaka J** (Co-PI). Improving patient's care, health outcomes through tailored, medical genetics and genomics capacity building educational programs for county as well as sub county hospitals in Kenya. 2022. (National Research Fund (NRF)- Kenya; NIH-H3 Africa; Submitted).
31. **Mburu S** (PI), **Gitaka J** (Co-PI). Understanding breast cancer patient population, subtype distribution in Kenya, their epidemiological risk factors and identification of population specific potential diagnostic, prognostic biomarkers as well as therapeutic targets: A mixed predictive study design. 2022. (Pfizer, AstraZeneca, GlaxoSmithKline Wellcome NCD Open Lab, NRF-Kenya; Submitted).

32. **Mburu S** (PI), Waweru D. Breaking community transmission in Covid-19: Innovations in contact tracing approaches suitable for resource-limited settings. 2022. (NRF-Kenya; Submitted).
33. Mehmood A (PI), **Naeem MA** (Co-PI). Development of cell replacements for ocular repair. 2022. (Pakistan Science Foundation; Submitted).
34. **Messaoud O** (PI), Henning B (Co-PI), Hammami A, Hassan Eissa S, Lipps J, Burton E, Varlık N. NiAl2O4 nanostructures for high-enhanced detection of DNA traces extracted from ruins of the archaeological site of Dougga. 2022. (AGYA/German Federal Ministry of Education and Research Fund; Accepted).
35. **Messaoud O** (PI), Scerri E (Co-PI), Kefi R, Dallali H, Jmel H, Touj F, AounALLAH S, Becher D, Ajlani I, et al. Multidisciplinary approaches to ancient DNA from the archaeological site of Dougga in Tunisia. 2022. (AGYA/German Federal Ministry of Education and Research Fund; Accepted).
36. Murphy M (PI), Hou, L, Sagay A, Ogunsola FT, Adeyemo WL (Co-I), **Abdulkareem FB** (Co-I). Northwestern/Nigeria Research Training Grant for HIV and Malignancies. 2019. (NIH/NCI; Continued).
37. **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. (Saval Laboratories; Continued).
38. **Nakousi-Capurro N** (PI). Acquisition of NGS technology for Carlos Van Buren Hospital. 2021. (Ministry of health, Servicio de Salud Valparaíso/San Antonio; Submitted).
39. Olopade OI (PI), Ntekim A, Popoola AO, Arowolo OA, Anthonia S, Olopade CS, Abiola I, **Abdulkareem FB** (Project Site Director). Assessing REsponse to neoadjuvant Taxotere and TrAstuzumab in Nigerian women with HER2-positive breast cancer (ARETTA)- A multicenter study. 2019. (University of Chicago Center for Global Health; Continued).
40. **Peter** Kingham (PI), Olusegun Alatise (Co-PI), Du (Co-PI), **Abdulkareem FB** (Co-I). Determining the risk factor profile and biology of colorectal cancer in Nigeria. 2020. (NIH; Continued).
41. **Wangi KYW** (PI), Birriel B, Smith C. 'Reproductive rights for women with thalassemia' Interdepartmental Graduate Student Grants (IGSGs). 2022. (Pennsylvania State University; Submitted).

2017 Batch

42. Calalo CM, Cases RKC, Asor BBN, **Abad PJB**, Asuncion PC. A content analysis of Facebook groups on congenital adrenal hyperplasia. 2021. (Newborn Screening Reference Center-University of the Philippines, Manila; Approved).
43. **Fatima SS** (PI), Fatima A (Co-PI). Mobile health (m-Health) intervention to reduce the epigenetic signature of Advance Glycation end Products (AGE) as a risk biomarker in metabolically healthy and unhealthy obese adults. 2021. (Pakistan Science Foundation; Concept paper approved).
44. **Fatima SS** (PI), Palla A (PI). Epigenetic signature of adipokines, Advanced Glycation End products (AGE) and inflammatory cytokines as a risk predictor in metabolically healthy and unhealthy obese adults. 2022. (Health Research Institute, National Institute of Health Pakistan; Received)

45. **Fatima SS** (PI), Thobani H. Epigenetic signature of the mental health burden of COVID-19 and beyond: SLC6A4 and its response to SSRI antidepressant therapy. 2021 (Student and Trainee Initiated Research (STIR)- Aga Khan University, Received).
46. **Mgasa A** (PI). Empowering community leaders to effectively engage with community health workers to improve community engagement in blood donation for sustainable blood supply to cancer patients in Dar es Salaam. 2022. (French Embassy in Tanzania; Submitted).
47. Moyes D (PI), Chirenda J (Co-PI), Challacombe S, **Dhoro M**, Mugadza G, Shoaie S. Post-COPARIZ: Post-COvid Pandemic Antimicrobial Resistance Profile In Zimbabwe. 2022. (National Institute for Care and Health Research: Research and Innovation for Global Health Transformation Call; Submitted).
48. **Owusu M** (PI), Sambian D, Godfred A. Enhancing global health security: Expanding efforts and strategies to protect, diagnose and improve public health in Ghana. 2021-2022. (Centre for Health System Strengthening/CDC; Accepted).
49. Palla A (PI), **Fatima SS** (Co-PI). Cardiovascular disease (CVD) risk stratification in people visiting a health camp in Gilgit-Baltistan, Pakistan before and after intervention using Framingham and Atherosclerotic cardiovascular disease (ASCVD) risk scoring system and subsequent validation of the tool. 2022. (Foundations for Health and Empowerment (F4HE); Concept paper approved).
50. Perez Millan MI, **Vishnopolska SA**. Neurodevelopmental delay with congenital anomalies in pediatric patients from South America. 2022. (End the Diagnostic Odyssey Grant, 3billion; Received).
51. **Petlichkovski A** (PI). Real-world effectiveness of COVID-19 vaccines in preventing symptomatic disease, hospitalization and death in the Macedonian population. 2022. (Chinese Center for Disease Control, China; Received).
52. **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. 2019-2022. (International Rotary Club, Global Grant; Continued).
53. Tae SK, Thong MK, Mazlan RA, Sulaiman S, Maceda EB, **Abad PJB**, **Tumulak MJ**, Calalo CM, Abadingo M. Development, and acceptability testing of a patient decision aid for prenatal testing amongst Asian women and their spouses in the ASEAN Region. 2021. (Global Genomics Medicine Collaborative SEED Award; Submitted).
54. **Wayengera M** (PI) Orem J, Kitaka S, Muhwezi WW. Use familial cancer-risk among adolescents in Uganda as an impetus to promote critical life stage interventions that prevent NCDs. 2022. (NIH; Submitted).
55. **Wayengera M** (PI), Delaporte E, Bongcam-Rudolf E, Plewcynski D, Weyer. Genomic epidemiology surveillance for poverty and emerging, re-emerging diseases in east, central & southern Africa. 2022. (HORIZON GH-EDCTP3 JU-01-03; Submitted).
56. **Wayengera M** (PI), Munier-Lehmann H, van Calenbergh S, Parzy D. Promoting uptake of mobile laboratory and paper-strip point of care diagnostics technologies in Africa: Uganda-pilot. 2022. (HORIZON GH-EDCTP3 JU-01-01; Submitted).
57. **Wayengera M** (PI), N Jillani N(PI). Pre-clinical testing of bacteria derived synthetic enzymes (ZFN & CRISPER-Cas9) in non-human primates as an HIV-1/AIDS cure. 2022. (ICIPE/Bio-Innovate; Submitted).
58. **Wayengera M** (PI). S-Phase enzymatic DNA synthesis biomarkers for rapid detection of *Mycobacterium tuberculosis* and all TB drug resistance testing. 2022 (NIH; Submitted).

C. Collaborations (NIH and Other Institutions):

NIH: (Total- 10; 2019- 2; 2018- 5; 2017- 3)

2019 Batch

1. **Abubakar S**, Koehly L. Assessment 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, Nigeria; NHGRI/NIH, Finalizing protocol).
2. **Yesilcinar İ**, **Seven M**, Şahin E, Calzone K. Genetics and genomic competency of Turkish nurses: A descriptive cross-sectional study. (Izmir Katip Celebi University, Health Science Faculty Obstetrics and Gynecology Nursing, Turkey; University of Massachusetts Amherst College of Nursing; NCI/ NIH).

2018 Batch

3. Ginsburg G, Patrinos GP, Lopez-Correa C, **Cornejo-Olivas M**, Teri Manolio, et al. Global Genomic Medicine Consortium G2MC. Global leaders dedicated to advancing genomic medicine implementation in clinical care. (Instituto Nacional de Ciencias Neurologicas, Peru; Global Alliance for Genomics and Health; The Golden Helix Foundation; Australian Genomics Health Alliance; NHGRI/NIH).
4. 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). (Hospital National Rebagliati Martins, Instituto Nacional de Ciencias Neurologicas, Peru; Undiagnosed Diseases Network International; NCATS/NIH).
5. 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, Ukraine; NEI/NIH).
6. 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; Layton Rehmatulla Benevolent Trust Eye Hospital, Lahore; Allama Iqbal Medical College, University of Health Sciences, Lahore, Pakistan; Wilmer Eye Institute, Johns Hopkins University, USA; NEI/NIH).
7. 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. (Instituto Nacional de Ciencias Neurologicas, Peru; Cleveland Clinic, Ohio; NIA/NIH).

2017 Batch

8. **Torres-Mejía G** (Site PI), Angeles Llerenas A, Gomez-Flores-Ramos L, Rodríguez-Valentín R, Flores-Luna L, Sanchez-Zamorano LM, Fierros-Zarate G del S, **Ortiz-Panozo E**, Fejerman L. LAGENO Consortium. (Multicenter 2021; Instituto Nacional de Salud Pública, Mexico; LAGENO-BC & CONFLUENCE; NCI/NIH).
9. **Torres-Mejía G** (Site PI), Angeles-Llerenas A, Gomez-Flores-Ramos L, Sánchez- Zamorano M, Flores-Luna L, Romieu I, **Ortiz-Panozo E**, Fierros-Zarate G del S, Rodríguez-Valentín R,

Rinaldi S. CONFLUENCE. Uncovering breast cancer genetics. (Multicenter 2022; Instituto Nacional de Salud Pública, Mexico; NCI/NIH).

10. **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, Turkey; ICO-NEI/NIH).

Other Institutions: (Total- 57; 2019- 27; 2018- 20; 2017- 10)

2019 Batch

1. **Abubakar S**. Development of the Jigawa State cholera outbreak preparedness, prevention and response plan-Technical Report. 2021. (Bayero University Kano/Aminu Kano Teaching Hospital; Jigawa State Ministry of Health; Jigawa State Primary Healthcare Development Agency, Nigeria; Nigeria/Save the Children International).
2. Albaghadi M, **Mushi TL,** Khuzeima K, Mujunia E, Katende A, Rohacek M, O'Brien P. Echocardiographic training and screening program for rheumatic heart disease, congenital heart disease, and cardiomyopathies in northern Tanzania. (Jakaya Kikwete Cardiac Institute, Tanzania; Madaktari Africa-USA; Naples Heart Institute-NCH Healthcare System, Italy; University of Iowa, USA).
3. **Bocoum A**, Koita A, Rizig M, Maiga Y, Guinto CO. Parkinson's disease in Africa: Developing a clinically characterized DNA resource for Genome-Wide Association (GWAS) and other collaborative genetic studies. (Centre Hospitalier Universitaire du Point "G", Bamako, Mali; Centre Hospitalier Universitaire Gabriel Touré, Bamako, Mali; UCL Queen Square Institute of Neurology, London, UK).
4. **Chowdhury EH**, **Amarakoon GGGT**, **Mosema KBA**, **Khant AK, Muhammad Iqbal**. 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; The Islamia University of Bahawalpur, Pakistan).
5. Dysted M, **Gyawali P**, Shrestha A, Shrestha R, Gyawali B, Christensen D. Cardiometabolic risk factors among Nepalese adolescents with and without the family history of diabetes. (Kathmandu University School of Medical Sciences; Dhulikhel Hospital, Nepal; University of Copenhagen, Denmark; World Diabetes Foundation).
6. Fejerman L, Dominguez-Valentin M, Vianna-Jorge R, Agalliu I, **Paredes-Moscosso 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, Peru; LAGENO-BC).
7. **Gyawali P**, G2MC. Global perspective on the challenges of establishing genomic medicine practice. Young Investigators writing project. (Kathmandu University School of Medical Sciences, Nepal; G2MC).
8. **Iqbal M**, Ansar M. Mutational analysis of DNA from consanguineous Pakistani families having ophthalmological disorders through whole exome sequencing (Dept. of Biotechnology, The Islamia University of Bahawalpur, Pakistan; Institute of Molecular and Clinical Ophthalmology Basel (IOB) Basel, Switzerland).
9. **Iqbal M**, **Naeem MA**. Identification, clinical evaluation, and genetic analysis of RP families (Dept. of Biotechnology, The Islamia University of Bahawalpur; National Centre of Excellence in Molecular Biology, University of the Punjab, Lahore, Pakistan).

10. **Iqbal M**, Serge N. Genetics studies of male infertility. (Dept. of Biotechnology, IBBB, IUB, Pakistan; Dept. of Genetic Medicine & Development, University of Geneva, Switzerland).
11. **Iqbal M**, Waryah AM. Identification, and genetic analysis of CC & congenital glaucoma families. (Dept. of Biotechnology, IBBB, IUB, Pakistan; Liaqat University of Medical & Health Sciences, Pakistan).
12. **Iqbal M**, Zafar MI. Mutational analysis of Pakistani families affected with Parkinson's disease through whole exome sequencing (Dept. of Biotechnology-IUB, Pakistan; Dept. of Neurology, Oslo University Hospital, Norway).
13. Kantaputra PN, **Guven Y**. Consultation on patients' cases related to genetics. (Chiang Mai University Faculty of Dentistry, Thailand; Istanbul University Faculty of Dentistry, Turkey).
14. Kennedy J, Dawes M, Smith H, Bryan S, Goldsmith L, **Hussein N**, Ng WL, Qureshi N, Winther M. Evaluation of pharmacogenomics in primary care. (Universiti Malaya, Malaysia; University of Toronto and University of British Columbia, Canada; University of Nottingham and University of Liverpool, United Kingdom; Nanyang Technological University, Singapore).
15. **Llamos-Paneque A**, Andrade M. Enzymatic and molecular studies for Fabry, Pompe, MPSI, Niemann Pick and Gaucher diseases in Ecuadorian patients treated at the Genetics Service of the Quito Military Hospital. (Genetics Service of Hospital No.1 FF. AA. Quito-Ecuador; Sanofi Ecuador).
16. **Llamos-Paneque A**, Giugliani R. Metabolic and genetic study of Ecuadorian suspect DLD disorder cases at Army Hospital. (Genetics Service of Hospital No.1 FF. AA. Quito-Ecuador; Hospital de Clinicas de Porto Alegre, Brazil).
17. **Llamos-Paneque A**, Giugliani R. Metabolic and genetic study of Ecuadorian suspect MPS disorder cases at Army Hospital. (Genetics Service of Hospital No.1 FF. AA. Quito-Ecuador; Hospital de Clinicas de Porto Alegre, Brazil).
18. **Llamos-Paneque A**, Liehr T. Genetic study of marker chromosome. (Genetics Service of Hospital No.1 FF. AA. Quito-Ecuador; Jena University Hospital, Institute of Human Genetics, Germany).
19. **López-Star B**, **LLamos Paneque A**. Consultation on ophthalmogenetic cases. (Specialty Hospital No.1 FF. AA, Quito, Ecuador; Mexican Society of Ophthalmology; Mexican Institute of Ophthalmology, Mexico).
20. **Luong LH**, Olivieri L. Consultation on cardiovascular malformations and rare vascular disorders. (National E Hospital, Vietnam; Children's National Hospital, USA).
21. **Mosema-Be-Amoti K**, Fu Y. Genetics of neurofibromatosis in Central Africa. In discussion. (Institut National de Recherche Biomédicale Biomédicale, DR Congo; University of Alabama at Birmingham, USA).
22. **Nair LS**, Nair MKC, Kumar S, Lukose R. Genetic evaluation of children with Intellectual disability and autistic spectrum disorders. (Dept. of Medical Genetics, NIMS Medicity; NIMS-Spectrum-Child Development Research Center, NIMS Medicity, Trivandrum, India).
23. 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. (University of Abuja; University of Ilorin, Nigeria; GCRF, Liverpool John Moores University, UK).
24. **Pirlog R**, Calin GA. Role of non-coding RNAs in response to DNA damage response pathway inhibitors in cancer. (Luliu Hatieganu University of Medicine and Pharmacy, Romania; MD Anderson Cancer Center, USA).

25. **Pirlog R**, Calin GA. Role of ultra-conserved elements in cellular defense in primordial organisms. (Luliu Hatieganu University of Medicine and Pharmacy, Romania; MD Anderson Cancer Center, USA).
26. Santra MK, Rapole S, Tripathi V, Shridhar PR, Shanamugham D, Tilak TVSVGK, **Mutreja D**, Deshmukh C, Tamhankar AS, Chatterjee S. Mechanism and 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, India).
27. Sidharth KS, Rupesh R, Divya S, Sadaf A, **Khant AK**, Manoji M, Kalaivani G, Shraddha L, Rajiv S, et al. Assessment of south Asian Pediatric Acute Kidney Injury Epidemiology and Risk Factors (ASPIRE): Design of prospective study on severe pediatric AKI. (SAARC nations with India, Myanmar, Bangladesh, Pakistan).

2018 Batch

28. **Adewole OO**, Gidado I, Agnes B, Onipede A. Connecting diaspora for capacity building in tuberculosis care. (Obafemi Awolowo University Teaching Hospital, Nigeria; International Organization for Mobility International; KNCV, Netherlands).
29. **Adewole OO**, Liu G, Komolafe A. Lung cancer genomics among Nigerians. (Obafemi Awolowo University, Nigeria; Princess Margaret Cancer Center, Canada).
30. **Adeyemo TA**, Xu JZ. Assessing Combination Hydroxyurea and Exogenous Erythropoietin in Sickle Cell Disease (ACHiEvE-SCD). (University of Lagos, Nigeria; University of Pittsburgh Medical Centre, USA.
31. Alatise OI, Kingham TP, **Abdulkareem FB**. Colorectal cancer database in Nigeria. (Obafemi Awolowo University, Nigeria; University of Lagos, Nigeria; Memorial Sloan Kettering Cancer Center, USA).
32. **Dueñas-Roque M**, **Cornejo-Olivas M**, Ortega J, Galarreta-Aima C. Ihope, an Ilumina project for genomic testing for rare diseases. (Hospital Nacional Edgardo Rebagliati Martins, EsSalud; Instituto Nacional de Ciencias Neurologicas, Peru; Ilumina, USA).
33. **Dwivedi A**, Kapoor S. Newborn screening as part of Delhi government's mission- Neonatal early evaluation vision (NEEV). (Army Hospital R&R (AHRR); Maulana Azad Medical College, Lok Nayak Hospital, New Delhi).
34. **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).
35. **Eshete M**, Hailu A, Abate F, Alamnie G, Butali A. Investigating the genetics and environmental causes of orofacial clefts in the Ethiopian population. (Addis Ababa University, Ethiopia; College of Dentistry, University of Iowa, USA).
36. **Gitaka J**, Pamme N. Development of molecular diagnostics for maternal infections using fiber mats. (Mount Kenya University, Kenya; Stockholm University, UK; Makerere University, Uganda).
37. Lopez-Cendes I, **Dueñas-Roque M**. Whole exome sequencing for identification of pathogenic genetics variants in early encephalopathy in infancy in Latin America. (Hospital Nacional Edgardo Rebagliati Martins, Perú; Universidad Estadual de Campinas, Brasil).
38. López-Köstner F, Alvarez K, De la Fuente M, Dominguez-Valentin M, **Dueñas-Roque MM**,…LA-GETH group. Implementation of a Latin American genetic counselling network in

hereditary colorectal cancer. (Hospital Nacional Edgardo Rebagliati Martins, Perú; Clínica Universidad Los Andes, Chile, LA-GETH group).

39. Luquetti D, **Dueñas-Roque MM**, Zarante I, Porras L, Timberlake AT, Hurtado P, Griffin C, Heike CL, Hing AV, et al. Phenotypic and genomic characterization of microtia in the Andean population. (Hospital Nacional Edgardo Rebagliati Martins, Perú; Washington University, USA; Seattle Children's Hospital, USA).
40. Mata IF, Inca-Martinez M, **Cornejo-Olivas M**, Cubas-Montecnio D, Manrique-Enciso C, Milla-Neyra K. Implementation of a Neurogenetics DNA Bank in Peru. (LARGE PD Consortium; GP2 and ASAP initiative; Instituto Nacional de Ciencias Neurologicas, Peru; San Marcos Foundation, Peru; Michael J. Fox Foundation, USA).
41. **Messaoud O**, Alkuraya FS. Investigating unsolved cases with undiagnosed phenotypes (Institut Pasteur de Tunis, Tunisia; King Faisal Specialist Hospital and Research Centre, Kingdom of Saudi Arabia).
42. **Messaoud O**, Hassan Eissa S. Developing innovative biosensors for trace DNA detection (Institut Pasteur de Tunis, Tunisia; Alfaisal University, Kingdom of Saudi Arabia).
43. **Nakousi-Capurro N**, Cavalcanti DP. Description of genotype-phenotype relations in patients with osteogenesis imperfecta in a Brazilian cohort. (Hospital Carlos Van Buren of Valparaíso, Chile; Faculty of Medical Sciences, University of Campinas, Brazil).
44. 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).
45. **Ottaru S**. Initiated congenital birth defect registry at the Pediatrics Dept. (Nkinga Hospital, Tabora, Tanzania; Multicenter).
46. Pasechnikova N, Zborovska O, **Savina O**, Kremenska U. Medical and genetic diagnosis, and consultations for families with a history of retinoblastoma considering pre-implantation. (V. Filatov Institute of Eye Diseases and Tissue Therapy; ISIDA-IVF Clinic, Ukraine).
47. **Savina O**, Rykov O, Kremenska U. Genetic testing, medical and genetic consultation for families with Aniridia and WAGR syndrome- pre-implantation diagnostics for family planning. (Dept. of Ophthalmology, Shupyk's National Medical Academy of Postgraduate Education; ISIDA-IVF clinic; NGO- Aniridia WAGR; Kyiv Ophthalmological Clinic, Ukraine).

Batch 2017

48. **Casado PL**, Patrick Shmidlin. Frontier in Dental Medicine: Biomarkers in periodontology and peri-implant diseases. (Fluminense Federal University, Brazil; University of Zurich, Switzerland).
49. 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 in collaboration with New York Eye Cancer Center, USA).
50. Joloba M, Kateete DP, Wampande E, Kyobe S, **Wayengera M**. Integrated biorepository of H3Africa in Uganda. (Makerere University, Uganda; Multi-center).
51. Kekitiinwa A, Kateete D, Wampande E, **Wayengera M**, Brown CW, Hanchard NA, Mardon G, Joloba M, Anabwani G, et al. Collaborative African Genomics Network-CAfGEN- build capacity for genetics and genomic training among collaborating African institutions through research on genetic determinants of pediatric HIV and TB disease- progression. (Baylor Pediatric

CoEs- Uganda; Makerere University, Uganda; Baylor College of Medicine, Texas; University of Botswana).

52. **Mahfoudh W**, Olufunmilayo I. Olopade. Breast cancer genomics- Exome sequencing of Tunisian women with triple negative breast cancer. (Faculty of Medicine of Monastir, Tunisia; Dept. of Medicine, The University of Chicago).
53. **Petlichkovski A**, Sukarova-Angelovska E, Brusco A, Rubeis SD. Whole-exome sequencing and analyses of proband families with neurodevelopmental disorders. (Institute for Immunobiology and Human Genetics, Medical Faculty Skopje; Pediatric University Clinic, Medical Faculty Skopje, Macedonia; University of Turin, Italy; Icahn School of Medicine at Mount Sinai, USA).
54. **Petlichkovski A**, Tofoski G. Analysis of KIR - HLA combinations in couples with infertility problems. (Institute for Immunobiology and Human Genetics, Faculty of Medicine, Skopje; University Clinic for Gynecology and Obstetrics, Medical Faculty Skopje, Macedonia).
55. **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, U and USA. (Chittagong Eye Infirmary, Bangladesh; London School of Hygiene, UK; 13 other centers in 8 countries).
56. **Thakur N**, Saxena AK. Identification and characterization of new gene variants of sex chromosome between two different populations in male infertility based on exome sequencing. (National Academy of Medical Sciences, Nepal; AIIMS, Patna, India).
57. **Wayengera M**, Paweska J, Plewcynski D, Bongcam-Rudlof E, Vlachakis DP. Biochemical adjustments of native EBOV glycoprotein in patient sample to unmask target- epitopes for rapid diagnostic testing (Makerere University, Uganda; The National Institute of Communicable Disease, South Africa; University of Warsaw, Poland; Swedish University of Agricultural Sciences, Sweden; Agricultural University of Athens, Greece).

D. New Research Projects: (Total- 53; 2019- 28; 2018- 10; 2017- 15)

2019 Batch

1. Abd Aziz NS, **Hussein N**, Ng WL. Prenatal genetic testing in primary care: Exploring the experiences and views of primary care doctors in Klang Valley, Malaysia.
2. **ALamin TA**. Use of early erythropoietin injection versus tonic supplementation in preterm neonates in multicentre in Khartoum State.
3. Alcausin MML, Lam H, **Tumulak MJR**. Cost-benefit analysis of cystic fibrosis in the Philippine Newborn Screening Program (Proposed).
4. **Amarakoon GGGT**, Silva A. Clinico-epidemiological profile of children with snake bite admitted to a Teaching Hospital, Anuradhapura, Sri Lanka: A prospective study.
5. Babalola Y, **Monye H**. Clinical and demographic characterization of adult patients with retinal diseases at the University College Hospital Ibadan: a two-year review. (In discussion).
6. Caneba JP, Maceda EB, **Tumulak MJR**, Alcausin MML. Clinical and molecular characteristics of Filipinos with confirmed Hemoglobin H Disease from 2019 to 2021. (Proposed).
7. Chagonda S, **Mhandire K**, Musarurwa C. High throughput sequencing towards optimization of African population specific cardiovascular gene panels.
8. Chakrabarty BK, Nagaraja M, Singhal P, Dagar Vikas, **Mutreja D**, Sen A. Evaluation of cytogenetic abnormalities and sperm FISH aneuploidy in infertile male (Proposed).

9. **Chowdhury EH** (PI), Dorji N, Lhendup K, Zangmo Y, Tobgay S, Phuntsho N. Incidence and prevalence of Turner's Syndrome in Bhutan.
10. **Guven Y**, Altunoğlu U, Eraslan S, Kayserili Karabey H. Elucidating the molecular etiopathogenesis of HED/EDA cases. (In discussion).
11. **Gyawali P**(PI), Quinonez S. Medical genetics need assessment in Nepal: an online cross-sectional survey.
12. Hassib N, Abdel-Fattah M, **Sayed I**, **Mehrez M**, Ismail S, Abdel-Ghafar S. Diagnosis of inherited disorders with oro-dental anomalies using recent advanced genetic approaches and impact on counseling and management. (Submitted).
13. Ibeh BO, Ifedilichukwu HN, Okoh MP, **Alli LA**. Modulation of cytokines and C-type lectin receptor expression in HIV infected population in Abuja, Nigeria.
14. Isuajah CE, Isuajah CC, **Akinmola OO**, Azinge EC. Foetal interleukin-6 and average plasma glucose concentration of obese pregnant women at term in Lagos University Teaching Hospital.
15. **Kars ME**, Itan Y. Genetic determinants and molecular mechanisms of obesity and cardiovascular disorders.
16. **Kars ME**, Itan Y. Phenome-wide association studies and polygenic risk score calculations in sequencing cohorts from diverse ancestral origins.
17. **Kars ME**, Stein D, Stenson P, Cooper D, Itan Y. A comprehensive knowledge base of known and predicted genetic variants associated with COVID-19 severity.
18. **Kars ME**, Unlu ES, Itan Y. Network-based heterogeneity clustering in primary open angle glaucoma.
19. **LLamos-Paneque A**, Christofolini DM. Clinical and genetic characterization of patients with Duchenne muscular dystrophy.
20. **LLamos-Paneque A**. Ecuadorian Duchenne Muscular Dystrophy Registry Initiative, coordinated by PTC Therapeutics.
21. **López-Star B**, Pérez- Serrano, Macías J. Evaluation of changes in lacrimal microbiota after treatment with therma eye plus in the treatment of dry eye disease due to meibomian gland dysfunction.
22. **López-Star B,** Pérez- Serrano, Voourdain S. Gut microbiome in Latin people with Diabetes Mellitus with and without Diabetic Retinopathy.
23. **Okunola OO**. Heritability of chronic kidney disease in first degree relations in southwestern Nigeria. (Proposed).
24. **Ortiz-Panozo E**, Melo-Zurita M, Mendoza-Benitez S. Congenital birth defects in a reference hospital in Cuernavaca, Mexico, 2005-2015. (In discussion).
25. **Sayed I**, Gamal El Din HM, **Mehrez M**, Abdel-Salam G, Ismail S, Abdel-Ghafar S. Advances in the delineation of genetic etiology of oro-dental anomalies in neurogenetic disorders. (Submitted).
26. **Tumulak MJR**, Maceda EB, Cases RK, Asor BB, Calalo-Magbanua C, **Abad PJ**. A content analysis of Facebook groups on congenital adrenal hyperplasia (Submitted for ethics approval).
27. Villegas-Llerena C, **Paredes-Moscosso SR**, Guevara-Fujita ML, Obispo D, Acosta O, Flores O, Parodi J, Montesinos R, Custodio N, et al. Mutational spectrum of Peruvian families affected by Parkinson's disease and other early-onset dementias.
28. **Wiafe SA**, Anyane-Yeboah K, Hamoud W. Whole exome sequencing for the RDGI-54Gene pilot project (100 patients). (Agreement signed).

2018 Batch

29. Araujo-Aliaga I, **Cornejo-Olivas M**. Alellic distribution of ATXN10 gene in Mestizo and Amerindian populations.
30. **Daich Varela M**. Long-read sequencing and transcript splicing analysis in RDH12.
31. **Dwivedi A**, Kalra S. Study the role of exome sequencing in children with congenital anomalies of kidney and urinary tract with extra renal manifestations.
32. Gidado, Agnes A, **Adewole OO**. Stigma reduction in tuberculosis.
33. **Lertwilaiwittaya P**, Rodriguez M, Callaway K, Korf B. University of Alabama, Birmingham's Undiagnosed Disease Program Review (Data analysis).
34. **Mburu S**, Kimani H. Developing integrated clustering models for laboratory testing in breast cancer, suitable for resource limited settings such as Kenya.
35. Mutesa L, Uwimana A, **Utumatwishima JN**. A multicentre randomized controlled non-inferiority trial to compare the efficacy, safety and tolerability of triple artemisinin-based combination therapies versus first-line ACTs + placebo for the treatment of uncomplicated *Plasmodium falciparum* malaria in Africa.
36. Onipede A, Awopeju OF, Obadire T, **Adewole OO**. TB gut microbiome.
37. Ordinola-Calle D, **Cornejo-Olivas M**. Clinical characteristics of CADASIL.
38. **Ottaru S**. Prevalence of *Toxoplasma* infection in asymptomatic infants (Submitted for review).

2017 Batch

39. **James O**, Adekunle AA. Evaluation of the effectiveness of community awareness programs by the cleft care team of the Lagos University Teaching Hospital.
40. Jilala E, **Mgasa A**. Status of medical genetics and genomics services and training in Tanzania (Submitted for ethics approval).
41. **Mahfoudh W**, Bannour I. Immunogenetics of COVID-19: Identification of genetic risk factors associated with severity and mortality of COVID-19 in the Tunisian population.
42. **Nandal R**, Laller KS. Genotypic and phenotypic spectrum of children with congenital heart disease clinic: A three year case series.
43. **Nandal R**, Laller KS. Role of 2D echo and global longitudinal strain in identifying sub-clinical LV dysfunction in patients with diabetes mellitus Type 2.
44. **Owusu M**, Boamah J, Ayisi-Boateng NK. Evaluation of novel rapid diagnostic test kits for detection of respiratory viruses.
45. **Petlichkovski A**. Estimating COVID-19 vaccine effectiveness against severe acute respiratory infections (SARI) hospitalizations associated with laboratory-confirmed SARS-CoV-2 in north Macedonia.
46. **Roy S**, Hoque F. Correlation of congenital ocular anomalies with intrauterine TORCH infection.
47. **Roy S**, Hoque F. Presentation of choroidal melanoma and treatment outcome in Chittagong Eye Infirmary.
48. **Roy S**, Kaliki S, Berry J, Fabian D, Nathalie C, Coronado DR, Reddy A, Parulekar M, Suzuki S et al. Histopathological high-risk factors in primary enucleated cases of retinoblastoma.
49. **Uwineza A**, **Hitayezu J**. Genetic etiology of neurodevelopmental disorders in Rwandan children.
50. **Vishnopolska SA**. Structure-function studies directed at identifying and understanding the functional impacts of variation in human disease genes using cutting-edge high-throughput functional assays.

51. **Wayengera M**, Moses J, Nantulya V. Development and laboratory validation of TB rapid diagnostic tests MakRIF.
52. **Wayengera M**. Development and validation of alternative point of care diagnostics platforms for easy detection of COVID-19 suited for sub-Saharan Africa.
53. **Yousef YA,** Mohammad M, Mehyar M, Sultan I, Al-Hussaini M, AlNawaiseh I. Mutational analysis for RB1 gene for patients with unilateral non-familial retinoblastoma.

E. Ongoing Research Projects: (Total- 86; 2019- 42; 2018- 21; 2017- 23)

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 write proposal).
3. 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).
4. **Bocoum A**, Cissé L, Cissé AKC, Ouologuem M, Maiga AB, Guinto CO, Landouré G, Krause A. Huntington's disease like 2: The first case in Mali, supposing the widespread of the disease. (Knowledge from Summit helped design study).
5. Chianumba R, **Alli LA**. Patterns of liver function parameters among patients living with sickle cell disease on hydroxyurea therapy. (Knowledge from Summit helped research execution, improved study quality).
6. **Chowdhury EH**, Lhendup K, Zangmo Y, Tobgay S, Phuntsho N. Incidence and prevalence of Down's syndrome in Bhutan. (Knowledge from Summit improved research quality).
7. Değirmenci T, **Yesilcinar İ**. Pregnant women's genetic literacy and affecting factors. (Summit inspired the project).
8. Foo JS, Ishak IH, **Hussein N**. The views and experiences of primary care doctors in managing children with learning disabilities. (Knowledge, skills attained at Summit helped mentor specialist trainee to undertake this research).
9. **Gyawali P**, Karmacharya R, Pant V. High burden of vitamin B12 deficiency among adults and elderly visiting a tertiary care hospital. (Summit lecture on Scientific Writing helped write manuscript).
10. Hassib N, **Sayed I**. An attempt to diagnose typical and atypical cases by identification of genes causing tooth structure anomalies. (Summit lectures improved skills in diagnosis of rare genetic disorders).
11. Hoe HK, Sherina NS, **Hussein N**, Malik TF. Exploring the views and experiences of parents on school-based thalassemia screening in Klang district, Malaysia. (Knowledge, skills gained at Summit helped mentor specialist trainees to undertake this research).
12. **Iqbal M**. Genetic analysis of retinitis pigmentosa in consanguineous families from southern Punjab, Pakistan. (Summit workshop on Grant Writing helped write grant proposal).

13. **Khant AK**, Khin YY. Clinical study of nephrotic syndrome in children at Yangon Children Hospital (Knowledge from Summit improved reporting research results).
14. **Khant AK**, San CC. Clinical study on Takayasu arteritis at Yangon Children Hospital. (Knowledge from Summit improved reporting research).
15. **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 in neurogenetics emphasized on triplet expansion disorders, helped in execution of the study.)
16. **Llamos-Paneque A**, Tekin M**,** Rivas-Iglesias C. Clinical-molecular research of hereditary deafness in Ecuadorian families of different origins at the Medical Genetics Service, Specialty Hospital FF.AA. No. 1 of the City of Quito. (Summit reaffirmed usefulness of registries for epidemiological studies, clinical characterization of genetic disorders).
17. **López-Star B**, Pérez- Serrano R, Ochoa Luis. Identification of individuals carrying the rs7677751 polymorphism of the PDGFRA gene in patients with Keratoconus. (Summit lectures provided tools to learn about the most frequent genetic variations in Mexican populations).
18. **López-Star B**, Pérez- Serrano R. Identification of rs7677751 polymorphism in PDGFRA gene, in patients with corneal astigmatism. (Summit lectures provided tools to learn of most frequent genetic variation in Mexican population).
19. 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 DR Congo. (Summit lectures on Bioethics and Variant Interpretation helped develop human subject consent protocols).
20. **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).
21. **Luong LH**, Hoang LT, Le NT, Tran TN. Study of rare immunodeficiency disorder. (Knowledge and credentials from the Summit initiated discussions and project).
22. Malik TF, Ng CJ, Hanafi NS, Chiew TK, **Hussein N**, Hadi H. Developing, evaluating and implementing a teleconsultation system to follow-up patients with type 2 diabetes mellitus during the Covid-19 pandemic. (Summit lectures on Telemedicine/remote learning, built confidence to develop, implement a teleconsultation module during the pandemic).
23. **Mhandire K**, Buxbaum NP. Bulk and single cell RNA sequencing of syngeneic and allogeneic cells in graft-versus-host disease. (Knowledge from Summit enhanced the bioinformatics input).
24. **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 to provide baseline data for monitoring preterm babies).
25. **Mushi TL,** Kabiligi J, Hokololo A. Prevalence, factors associated and diagnostic utility of differential oxygen saturation in the diagnosis of persistent pulmonary hypertension among newborn (PPHN) at Bugando Medical Centre, Tanzania. (Knowledge from Summit on NBS helped understand diversity in neonatal disease, abnormality during embryogenesis, fetal-to-neonatal transition failure and need for early detection, influenced exploring alternatives to echocardiography such as oxygen saturation for PPHN in low-resource settings).
26. **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).

27. Ng WL, **Hussein N**, 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). (Knowledge, skills in pharmacogenetics from Summit initiated the project).
28. **Okunola OO**, Adekoya A, Ojo EA, Adeoti PA, Adeyemi AA. Genetics study in autosomal dominant polycystic kidney disease in the tropics. (Used Summit resources and links).
29. **Okunola OO**, Ojo FA, Jones k, Enitan BA, Adeyemo TY. Stigma related quality of life assessment scores in sickle cell disease patients in south-west Nigeria. (Used Summit materials, experience).
30. **Paredes-Moscosso SR**, 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. (Knowledge from Summit deepened understanding, awareness of VUS, particularly in underrepresented populations-Peruvian, informed approach for project).
31. Rathnasiri KMSS, **Amarakoon GGGT**, Fernando MGUS. Knowledge and attitudes on *Varicella zoster* infection and its vaccine among parents of children presenting to a pediatric unit in a tertiary care hospital in rural Sri Lanka. (Knowledge from Summit helped design project).
32. **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).
33. Sondhi V, Mishra P, Sridhar G, Singhal P, **Mutreja D**. DBT- NIDAN Kendra for Genetic Center for Neonatal and Prenatal Screening. (Summit provided insight into NBS programs in the US which helped formulate policy guidelines for our own screening program for genetic, endocrine, metabolic disorders).
34. Vanlare TO, Emokpae MA **Akinmola OO**. Single nucleotide polymorphism in renalase and KCNQ1 genes among women investigated for infertility in Lagos, Nigeria. (Knowledge from Summit on polymorphisms in disease phenotype helped conceptualize study).
35. 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).
36. Vilain E, Linguraru M, Mumba-Ngoyi, Tshala-Katumbay D, Bramble MS, **Mosema-Be-Amoti K**, Spencer D. Mobile diagnosis of congenital genetic conditions: A model for screening and surveillance in low-resource settings. (Summit attendance positioned me to lead the local research team).
37. **Wiafe SA**, Ameyaw EK, Dunyoh B, Appiah DY, Amissah 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).
38. **Wiafe SA**, Anyane-Yeboa 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 helped design protocol).
39. **Wiafe SA**, 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. (Knowledge from Summit and training helped design this project).
40. **Yadav S**. Retrospective study on diagnostic accuracy of absent superimposed line (vomer and palate) in mid sagittal view for the detection of isolated posterior cleft palate. (Summit's Tailored

Training in Fetal Medicine at Medstar Washington Hospital Center (Dr Malissa Fries), provided insight into a structured approach to antenatal ultrasonography and detection of congenital malformations- NT/NB scans and Target scan).

41. **Yesilcina**r **İ**, Bektaş Pardes B, Güvenç G. Development of the health belief model scale for prenatal genetic screening and diagnostic tests. (Knowledge gained at Summit in prenatal diagnosis helped design research).
42. **Yesilcinar İ**, Şahin E, Tutar SO. The decisional conflict and the genetic literacy of pregnant women regarding and prenatal screening tests. (Knowledge from Summit helped design project).

2018 Batch

43. 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 for this project).
44. **Adewole OO**, Sogaolu OM, Aminu AA, Nwosu N, Adeyelu A. Clinicopathologic and genomics of lung cancers among Nigerians. (Leveraged on additional knowledge gained at the Summit in clinical research and project implementation).
45. **Adewole OO**. Finding people with tuberculosis through strategic deployment of a non-sputum based (Sweat TB Test) in Nigeria. Grant challenges Africa Phase II. (Leveraged on additional knowledge gained at the Summit on lung cancer genomics and biobanks).
46. **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, continued implementation).
47. Afolabi BB, 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).
48. Afolabi BB, Galadanci H, Balogun M, **Adeyemo TA**, Sam-Agudu N. Intravenous versus oral iron for iron deficiency anemia in pregnant Nigerian women (IVON): An open label, randomized controlled trial. (Knowledge from Summit supported study design, implementation).
49. Ajoloko EA, **Oluwarotimi AC**, Adeyemi MO. Efficacy of sodium bicarbonate buffered lidocaine hydrochloride in reduction of injection, onset of action and depth of anesthesia intra-alveolar extraction of mandibular first and second molars: A randomized controlled double-blind study. (Knowledge from Summit helped design study).
50. 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. (Knowledge from Summit helped design study).
51. **Cornejo-Olivas M**, Cubas-Montecino D, Bazalar-Montoya J. DNA biobank implementation in Neurogenetics Division, Instituto Nacional de Ciencias Neurologicas. (Summit lectures on Broad Informed Consent, Biobanking and Ethics, supported proposal, design of standardized procedures for the first biobank).
52. **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 reinforced epidemiological research on congenital malformations).
53. **Dueñas-Roque MM**, Segura P, Mendiola L, Acuña I, De La Torre C, Purizaca N, Prötzel A, Ledesma Y. Clinical and epidemiological profile of lysosomal storage diseases in the Social

Security of Health of Peru 2019-2021. Collaborative Study. (Summit emphasized the need to lead research on rare diseases in Peru).

54. **Dwivedi A**, Gupta A, Choubey M, Kalra S, Kumar A, 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).
55. **Dwivedi A**. Genotype-phenotype correlations in children with renal tubular acidosis: Unravelling the mystery. (Knowledge from Summit helped in conceptualizing the proposal).
56. 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).
57. **Lertwilaiwittaya P**, Roothumnong E, Thongnoppakhun W, Limwongse C, Pithukpakorn M. Next generation sequencing in breast-ovarian cancer in Thailand (Data analysis). (Knowledge from Summit helped design bioinformatics pipeline).
58. 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. (Summit lecture on Neurogenetics (Dr. K. Fischbeck, NINDS), suggestions and comments helped improve recruitment strategy and MTA agreement).
59. **Mburu S**, Gitongam H. Sustainable analytical approaches, and predictive modelling to actualize precision medicine in breast cancer management for resource-limited settings. (Knowledge and skills from the Summit in Big Data Analytics and its potential generated interest/passion in data science, which culminated in this research project).
60. **Mburu, S, Gitaka J**. Understanding breast cancer sub-types and their epidemiological risk factors by a mixed predictive study design. (Summit helped understand importance of genetic diversity).
61. Menzel Stephan (P1), **Adeyemo TA**, Nnodu O. Identification of novel mechanisms of fetal-haemoglobin induction by common genetic variants in patients with sickle cell disease. (Summit encouraged research collaboration).
62. **Messaoud O**. Genetic investigation of DNA repair disorders (Fanconi anemia, Xeroderma pigmentosum and atypical forms of photogenodermatoses). (Knowledge gained through Summit's Tailored Training helped interpret incidental findings).
63. Pericak-Vance M, **Cornejo-Olivas M**. Genetics of dementia in a Peruvian cohort. (Summit lectures in Bioethics and Scientific Writing helped improve the informed consent form, writing of proposal).

2017 Batch

64. Aglan MS, **Otaify G**. Targeted next-generation sequencing in the diagnosis of osteogenesis imperfecta and Bruck syndrome. (Knowledge gained at Summit helped improve reporting of results).
65. **Avogbe PH**, Brun LVC, Sanni A. Comparison of HPV detection in urine and cervical samples collected from Beninese women. Approved by Ethical Committee. 150 samples collected. (Resources from Summit helped design study).
66. **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).

67. 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).
68. **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).
69. **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).
70. **Casado P**L, 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).
71. **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).
72. **Casado PL**, Quinelato V, Aguiar T. Analysis of 7 different techniques to isolate miRNA. (Summit's Bioinformatics Workshop helped understand relationship of miRNA with DNA and messenger RNA, improved project execution).
73. **Casado PL**, Quinelato V, Aguiar T. Genetic profile of patients who underwent peri-implant maintenance therapy: Relationship with peri-implant disease incidence. (Summit lectures helped understand characteristics to be considered when studying multifactorial diseases (peri-implant), improved the project).
74. **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).
75. **Casado PL**. Prevalence of periodontopathogens, *Streptococcus pneumoniae* and SARS-CoV-2 infection in patients under intensive therapy. **(**Summit provided basis for correlation of different diseases and their genetic contribution to phenotype).
76. **James O**. Clinical and demographic presentation of multiplex cleft families at Lagos University Teaching Hospital (LUTH) from 2016 -2022. (Summit gave insight for exploring better patient care options for treatment).
77. Jjingo D, Kateete D, **Wayengera M**. Nurturing genomics and bioinformatics research training in Africa (BreCA): High-throughput sequencing technologies. (Summit offered curriculum for the training integrated in this program).
78. **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 patients).
79. **Mahfoudh W**, Snoussi K Faleh R. Breast cancer genetics- identification of genetic risk factors associated with the early onset and poor prognosis characterizing the disease, in the Tunisian population. (Knowledge, Tailored Training acquired at the Summit, helped initiate project, secure funding from NIH and other international funding agencies for implementation of NGS).
80. **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, grant writing).
81. **Otaify GA**, Abdelhamid M, Elhusseini R, Aglan M. Targeted next-generation sequencing for diagnosis and prevention of joint deformity skeletal dysplasias. (Summit helped in proposal preparation, writing progress reports).

82. **Roy S**. Demographic profile, presentation and treatment outcome of ocular surface neoplasia treated in a tertiary eye hospital of Bangladesh. (Knowledge from Summit inspired search for alternative methods and treatments).
83. **Roy S**. Presentation of genetic diseases in a tertiary eye care center of Bangladesh. (Summit inspired diagnosis and creation of awareness on ophthalmological genetic diseases, the first of its kind in Bangladesh).
84. **Thakur N**, Brutsaert TD, Bigham A. Role of EGLN1 gene in Sherpa highlanders. (Summit inspired research).
85. **Torres-Mejía G**, Vaca-Paniagua F (PI), Rodriguez-Valentin R, Romieu I. Identification of pathogenic variants of susceptibility to early-onset breast cancer in young women (CONACyT, 000000000285879) (CONACyT UNAM 2018-2022). (Knowledge gained at Summit allowed expansion of research networks with other Mexican institutions).
86. Zaki SS, **Otaify G**, Zaki MS, Issa MY, Mahmoud MM, Abdelhamid MS. Analysis of dystrophin-associated glycoproteins (α-dystroglycan and laminin subunit α2): A shortcut for biochemical screening of congenital muscular dystrophies. (Summit helped in proposal, writing progress reports).

F. Other Accomplishments/Initiatives:

2019 Batch

1. **Abubakar S**. Lectured on 'Ethical, Legal & Social Implication (ELSI) of genetics and genomics' in the Medical Ethics & Law Course for 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 Examiner (Feb. 2022), included questions on genetics and genomics for Membership Exams, at the Faculty of Community Health, West African College of Physicians, Lagos/Abuja, Nigeria (Apr. 2022); Appointed as Supervisor/Reviewer, Network on Behavioral Research for Child Survival in Nigeria (NETBRECSIN); Reviewed proposals and dissertations on Maternal & Child Health at NETBRECSIN meeting (Dec. 2021), sponsored by Federal Ministry of Information in collaboration with Federal Ministry of Health and the United Nations Children's Fund (UNICEF), Nigeria; Journal Reviewer- BMC Health Services Research, Springer Nature (Global Publishers, 2021).
2. **Akinmola OO**. In the advanced stages of dialogue for starting a Genetic Counselling Unit in the Tertiary Hospital, Lagos University Teaching Hospital, implementation is in the offing including workshops for sensitization; Lectured on 'Management of neonatal diabetes', 'Genetics as a tool for precision medicine, to optimize outcome for patients', 'Newborn Screening: Neonatal hypothyroidism', at Pediatric Endocrinology Training Centre For West Africa; Lectured on 'Polygenic risk scores assessment utility pros and cons' to Residents in Pathology, Lagos University Teaching Hospital; Attendee, Virtual Cancer and Immunology Research Summit by SelectScience (Jul. 2022); Virtual Summit Scholarship Awardee, Genomics of Rare Disease (Mar. 2022), by Wellcome Connecting Science; Member, G2MC Rare Disease Working Group for pilot study on rare diseases, chose 6 clinical sites in 6 countries (Malaysia, Sri-Lanka, Chile, Mexico, South Africa and Nepal) out of 16 countries including Nigeria (Sept. 2021); Identified cases (3) at each site for sequencing at Gentogene, case conference on results and diagnosis is pending; Expansion to include more sites is contingent on funding.

3. **Alamin TA**. Selected member, International Advisory Board, at the 5th International Conference on Engineering Professional Ethics and Education (Aug. 2022).
4. **Alli LA**. Re-appointed, Visiting Lecturer, Medical Biochemistry and Molecular Biology for Preclinical Medical students, Nile University, Abuja, Nigeria (Jul. 2022); Completed online course on 'Genomic variant analysis and interpretation' (Jun. 2022); Supervised and mentored postgraduate research students (4 MSc); Lectured on 'Health implications of polychlorinated biphenyls and persistent organic pollutants', at UNDP/Ministry of Environment, Abuja, Nigeria (May 2022); Used materials from Summit to lecture medical students on 'Basic molecular biology', 'Genetic disorders', 'Inborn errors of metabolism'; Strengthened Biochemistry curriculum on Genetics and Genomics for postgraduate Medical Biochemistry students; Journal Reviewer - British Journal of Pharmaceutical Research, International Journal of Biochemistry Research and Review (May 2022).
5. **Amarakoon GGGT**. Lectured on 'Human genetics', 'Hemoglobinopathies', 'Congenital hemolytic anemia' to undergraduates in Faculty of Medicine and Allied Sciences, Rajarata University of Sri Lanka; Successfully completed a course 'Genomic variant analysis and clinical interpretation', held by CSIR Institute of Genomics & Integrative Biology (CSIR-IGIB) as part of the Genomics for Understanding Rare Diseases India Alliance Network (GUaRDIAN) (Jun. 2022); Maintained database for recording syndromes, congenital malformations and genetic disorders at the University Pediatric Dept., Teaching Hospital Anuradhapura, Sri Lanka; Identified, diagnosed and treated genetic cases (26)- syndromes (16), hemoglobinopathies (6), Wilson disease (1), paroxysmal cold hemoglobinuria (1), G6PD deficiency (1), choledochal cyst (1).
6. **Baatar N**. Helped establish a national level web and mobile based telemedicine service, includes booking in-clinic appointments, viewing of test results and medical records, referring patients to specialist, ordering health packages; Further developments will include ordering laboratory tests, medicines covered by national health insurance and diagnostic radiology tests; Attended 7th Annual International Conference by Board of Genetic Counseling India (Virtual, Jul. 2022); Attended Human Genomic Epidemiology-Asia (Virtual, Jun. 2022); Attended 1st Southeast Asia Rare Disease Summit (Virtual, Jan. 2022).
7. **Bocoum A**. Work impacted by Covid.
8. **Chowdhury EH**. Initiated and continued to register cases in Registry for Congenital Abnormalities, Dept. of Pediatrics, SGH, Bhutan; Emphasized importance of genetic counseling based on Summit learnings; Continued to train 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 at SGH; Identified and managed pediatric genetic disorders (17)- Down's syndrome (5), hemophilia (2), congenital hypothyroidism (3), achondroplasia (1), neurofibromatosis (2), Turner's syndrome (3), Marfan's syndrome (1); Referred cases for cardiac surgery (3), 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 systems.
9. **Guven Y**. Diagnosed, referred patients (11) to genetics department for molecular confirmation and medical referral- Ectodermal dysplasia (4), Papillon LeFevre (1), isolated oligodontia cases (6); Early diagnosis, surgical interventions, dental prosthodontic and restorative therapies, preventive and follow-up protocols prevented fatal consequences; Contributed to socio-psychological well-being of children and improved their quality of life.

10. **Gyawali P**. Completed courses in 'Introduction to epidemiology for global health', (Mar. 2022) and 'Leadership and management in health' (Dec. 2021), University of Washington, USA; Invited resource person, speaker on 'Sample management', at workshop on 'Good clinical laboratory practice', organized by Nepal Medical College (Jun. 2022); Invited resource person, speaker on 'Study designs', at 'Introduction to clinical research and good clinical research practice' workshop, organized by Nepal Eye Hospital (May 2022); Invited speaker, presented on 'Encouraging HbA1c rather than fasting and PP blood sugar', at Indo-Nepal-UK Diabetes Meeting and Annual Professional Conference (Nepal, Mar. 2022).
11. **Hussein N**. Shortlisted for Seed Award from G2MC for Feasibility of using MiGene Family History App (FMHA) in primary clinics in Malaysia (Oct. 2021) for purposes of promoting genetics in low-resource settings; Speaker, at 'Teach our students' workshop (Jan. 2022); Auditor/Coordinator, postgraduate curriculum in Family Medicine Specialty, University of Malaya, Malaysia, to increase awareness and strengthen genetics and genomics (Jan. 2022); Mentored Family Medicine trainees on research projects in prenatal genetic testing in primary care and parental consent in school-based thalassemia screening; Member, Academic Committee, Medical Humanities and Ethics Unit, Faculty of Medicine, University Malaya, Malaysia (Jun. 2022); Member, Scientific Committee for 'Breakfast at UMHealth' webinar series for healthcare providers, University Malaya, Malaysia (Jan. 2022); Presented on 'Asthma in Malaysia: Bridging the gaps' and 'Evaluating environmental triggers of asthma in Asian countries', at NIHR Global Health Research Unit on Respiratory Health (RESPIRE), The University of Edinburgh (UK, May 2022); Presented proposal on 'Feasibility of using MiGene Family History App (MFHA) at primary care clinic in Malaysia', at 6th G2MC International Conference (Virtual, Oct. 2021); Reviewer, PhD students (2) thesis and defence; Journal Reviewer- Public Health Challenges, European Journal of Contraception and Reproductive Health Care, Malaysian Family Physician.
12. **Iqbal M**. Awarded Training Grant by University of Geneva (Oct. 2021); Focal person for hands-on training for 'IBBB- HRD capacity building workshop'organized by The Islamia University of Bahawalpur, Pakistan (2022); Delivered lectures on 'Advanced molecular genetics and classical genetics' to graduate & undergraduate level students; Supervised graduate research students (4 PhD, 7 MS) on molecular and genetic basis of different hereditary disorders; Appointed as External Examiner, reviewed MS theses (5); Reviewed Research Grants (5) for funding by ORIC, IUB & HEC, Pakistan; Journal Reviewer- PLOS ONE.
13. **Kars ME.** Obtained PhD in Molecular Biology and Genetics, Summit resources and knowledge helped write thesis (Jan. 2022); Joined the Itan Lab, Institute of Personalized Medicine at Icahn School of Medicine at Mount Sinai, NY (USA) as a postdoctoral fellow (Mar. 2022); Teaching Assistant for 'Human genetics' and 'Introduction to human biology' lectures (2021-2022), shared knowledge from Summit
14. **Khant AK**. Promoted to Senior Consultant Pediatrician, Pediatric Nephrology Dept., Yangon Children Hospital (July 2022); Attended the Genomic England Research Summit (Virtual, May 2022); Examined, diagnosed and treated pediatric cases (~2100) in the emergency department-acute diarrhea (500), acute viral infection (1000), dengue hemorrhagic fever (100), acute respiratory tract infection (500), pyrexia of unknown origin (30), renal replacement therapy for acute kidney injury (12), chronic kidney disease children (13); Counselled patients with hereditary renal disease (30)- congenital nephrotic syndrome (2), congenital abnormalities of kidney urinary tract (10), meningomyelocele with neurogenic bladder (4), syndromic child with

renal abnormalities (3), Lowe syndrome with renal tubular acidosis (1), Prune Belly Syndrome (1), focal segmental glomerulosclerosis (7) and several others.

15. **LLamos-Paneque A**. Professor, Human Genetics (undergraduate level), School of Dentistry, International University of Ecuador; Updated topics in human genetics; Applied for travel scholarships to attend a course on 'Lysosomal storage diseases', Panama (Oct. 2022), and 5th Summit on Rare Disorders, Colombia (Aug. 2022); Summit provided access to research groups to drive medical genetics in low-income countries; Continued to work with Ibero-American Network of Neurocutaneous Diseases (Drs. F. Ramos, MT. Acosta, NIH)- presented and attended discussions on neurocutaneous cases and therapeutic advances; Pursued educational activities, increased awareness on genetic disorders using social networks (Facebook), radio/TV media on Campaign Day-Batten Disease Awareness Event (Jun. 2022), and with patient associations such as Embracing Hopes Foundation, on Day of Trisomies (Mar. 2022); Participated in 'Registry of patients with Duchenne muscular dystrophy', sponsored by Ecuadorian Medical Federation, Manta, Ecuador (May 2022); Attended webinars- 'Update on EIM: Hyperammonemia: A diagnostic-therapeutic challenge', by Spanish Society of Inborn Errors of Metabolism, 'MLD-S2-M2: Identification of pre-symptomatic patients - The importance of family screening', by Association Excellence In Pediatrics Institute, 'Matching ultra-rare cases in the clinic', by Face 2gene team, 'CentoArray: opening the door to early and accurate diagnosis' , by Centogene Lab, 'Approach to genetic diagnosis in ophthalmology', by Invitae Lab (May-Jun. 2022); Attended course on 'Genetic counseling in prenatal diagnosis', by the Fetal Medicine Chapter, Ecuadorian Society of Gynecology (Mar. 2022); Summit improved genetic counseling skills, implemented genetic counseling service at a private clinic's Maternal Fetal Unit (~80 cases/yr); Identified cases (4)- structural chromosomes anomalies including supermarket chromosome, confirmed by molecular cytogenetics by German collaborator (Dr. T. Liehr), as service is not available in Ecuador; Confirmed and evaluated genetic cases (400)- mono gene etiology (120) via molecular tests, chromosome (100) via cytogenetic studies, or multifactorial (100); Registered new cases (320) in the Registry of Genetic Diseases, Specialty Hospital No.1 FF. AA, Quito, Ecuador; Editorial Board- Molecular Cytogenetics- BMC.
16. **López-Star B**. Knowledge acquired at Summit helped initiate an Ophthalmogenetics Dept., at Mexican Institute of Ophthalmology; Invited to lecture on 'Basic Genetics' to medical students from Autonomous University of Querétaro; Trained junior staff in ophthalmogenetics; Member of Pan American Inherited Retinal Diseases group (since Feb. 2020); Continued to record, monitor cases (72) with inherited retinal disorders; Evaluated, diagnosed, treated opthalmogenetic cases (15)- congenital glaucoma (5), retinitis pigmentosa (5), congenital cataract (5); Referred patients (8) for medical management to Children's and Women's Hospital, Mexico; Diagnosis improved quality of life for patients and families due to known prognosis and access to appropriate support systems; None were candidates for treatment, but couples made informed decisions in family planning.
17. **Luong LH**. Affiliated with National E Hospital, Vietnam (2022); Recipient of the KMU Scholarship (PhD candidate) for the International Postgraduate Program, iPS Cell Regenerative Medicine, at Kansai Medical University, Japan (Sep. 2022); Organized a workshop on 'Diagnosis and management of hereditary angioedema (HAE)' (Apr. 2022); Raised awareness and established core labs for diagnosis of HAE; Invited speaker, presented on 'Update on the diagnosis and management of hereditary immune disorders', at Annual Meeting of HCMC Society of Asthma Allergy and Clinical Immunology (Vietnam, Jul. 2022); Diagnosed cases (~100), counseled patients and families affected by genetic disorders- immune deficiency (20),

hereditary angioedema (10), familial autoimmune disorders (10), cardiovascular malformation (50), rare vascular disorders (10); Implemented Redcap (Vanderbilt University) in E Hospital for research data management and registry of patients/families with genetic disorders; Journal Reviewer -Journal of Clinical Rheumatology (3 manuscripts) and Gynecologic and Obstetric Investigation (1 manuscript).

18. **Mhandire K**. Appointed Genetic Variant Scientist-Review Analyst, GeneDx, MD, US (Feb. 2022); Analyzed cases (10 patients/ day); Classification guided patient therapy and intervention; Learned variant analysis at GeneDx, during a Summit field trip; Networking with experts at the Summit helped undertake self-training in high throughput genetic analysis and bioinformatics; Skills being acquired at GeneDx, will be used to train other geneticists and clinicians in Africa on variant interpretation.
19. **Monye HI**. Completed residency in Ophthalmology (Apr. 2022); Awarded Fellowship of the West African College of Surgeons (FWACS), Fellowship of the National Postgraduate Medical College of Nigeria (FMOph, May 2022); Completed a short course in 'Cancer clinical genetics in Africa', by African Oncogenetics Network (Nov. 2021); Nominated member, Scientific Committee of the Ophthalmological Society of Nigeria (Jul. 2022); Awarded H3Africa Travel Fellowship for 19th H3Africa Consortium meeting (Jun. 2022), attended 'Scientific Writing and Publishing' workshop; Appointed member of the H3Africa Rare Disease Working group (May 2022).
20. **Mosema KBA**. Lectured on 'Genetics' to pediatrics medical students at Université Protestante du Congo, Kinshasa and Université Uele, Isiro, Haut-Uele Province; Speaker on, 'Basics of the genomics', for medical doctors and health professionals at the Biamba Marie Mutombo Hospital, Kinshasa City; Presented poster on 'Differences of sex development (DSD) in Central Africa: Genetics, psychosocial adaptation and perceptions', at ACMG Annual Meeting, Nashville (USA, Mar. 2022); Continued establishment of Sickle Cell Disease Unit (screening, treatment and follow-up), at Biamba Marie Mutombo Hospital, Kinshasa City; Continued clinical training network (WhatsApp) in genetics and genomics for medical students and doctors; Diagnosed familial cases (5)- Crouzon syndrome (2), Sotos syndrome (3).
21. **Mushi TL**. Used knowledge from Summit to train undergraduate medical students, interns, general practitioners in basic genetics, newborn screening and precision medicine; Attended training on 'Acute pediatric care', by Stanford Centre for Innovation in Global Health (Virtual, 2022-2023); Completed Pan African Society of Cardiology and Pan African Society of Cardiology (PASCAR) training on 'Cardiac CT in congenital heart disease (Virtual, Jun. 2022); Completed training on 'Morphology of ASD, VSD, PDA and other complex lesions', conducted by Congenital Heart Academy's (Jun. 2022); Attended 1st Tanzania International Conference on 'Multidisciplinary approach towards cancer care' (Tanzania, Jul. 2022); Attended PASCAR, Hypertension and Preventive Task force conference (Virtual, Jul. 2022); Established a Birth Defect Registry at Bugando Medical Centre, Tanzania; Diagnosed malformation cases (256) based on phenotype- congenital heart disease (47), anorectal malformation (68), gastrointestinal malformation (omphalocele, gastroschisis, duodenal atresia, trachea-esophageal fistula), Down's syndrome, Beckwith Weidman syndrome, Patau' syndrome, Prune Belly syndrome; Offered counselling as part of routine management; Patients with congenital heart disease with hemodynamic instability (36) were medically managed, others received surgical interventions.
22. **Mutreja D**. Delivered online lectures on 'Genomics' to faculty and colleagues, lectured post graduate residents and medical students; Shared resource material from Summit with medical students, postgraduate residents and colleagues from other specialties; Identified cases (13) by

cytogenetics and histopathologic/microscopic diagnosis- de la Chapelle syndrome (1), atypical fusion pattern in CML (1), atypical fusion pattern in APML (1), additional cytogenetic abnormality in CML trisomy 8 (1), Robertsonian translocation(14; 21) in a patient with recurrent miscarriage (1), thalassemia (4), sickle cell disease (2), Gaucher disease (1) translocation (7;14) in a child with congenital heart disease (1); Section Editor (Pathology)- Indian Journal of Medical Pediatric Oncology (2020-2021).

23. **Nair L**. Examiner, Level 1/Level 2 certification examination, Board of Genetic Counselling, India; Speaker on 'Troubleshooting with VOUS, at Trivandrum Neurocon, Annual Conference of the Kairali Neurosciences Society (India, May 2022); Speaker on 'Identification and management of genetic disorders' in Kairali television channel (May 2022); CME Speaker on 'Stem cell therapy - the genetic aspects' in the Deccan Conference (Virtual, Feb. 2022); Conducted webinars for pediatricians, developmental pediatricians, therapists and nurses on 'Early identification of genetic disorders' (Jan - May 2022); Conducted OPD, evaluated, diagnosed patients with genetic disorders; Identified cases (73)- intellectual disability/ developmental delay (15), recurrent pregnancy loss/ infertility (18), rare genetic disorders like *PEX2* gene associated peroxisome biogenesis disorder (1), SMARCA4 associated Coffin Siris syndrome type 4 (1), *INF2* gene associated focal segmental glomerulosclerosis type 5 (1), *SLC19A3* associated biotin-thiamine responsive encephalopathy type 2 (1), *SLC12A3* gene associated Gitelman syndrome (1), *BRAF* associated cardiofaciocutaneous syndrome (1); Provided genetic testing and counselling to patients.
24. **Okunola OO**. 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 pediatric nephrologists to establish congenital kidney disease registry with follow up; Diagnosed patients (15) with inherited kidney diseases (all autosomal dominant polycystic kidney disease) and others with congenital anomalies of the kidney and urinary tract (CAKUT)- ureteropelvic junction obstruction (7), renal agenesis (1) and horse shoe kidneys (1), screened, followed up on siblings (especially in ADPKD); Patients and family reported improvement in quality of life due to accurate diagnosis and pathway to care.
25. **Ortiz-Panozo E**. Admitted to Harvard T.H. Chan School of Public Health for doctoral training in Reproductive, Perinatal and Pediatric Epidemiology (Aug. 2022); Provided consultancy to Dr. **G**. **Torres-Mejía** (Summiteer) graduate students, on statistical analysis of gene-environment interactions for their master theses; Applied for Accreditation as a Continued Professional Development Reviewer, Agency for Public Health Education Accreditation, Brussels, Belgium (Jul. 2022); Associate Editor for Journals- Frontiers in Global Women's Health (Women's Health Section, 2022); Frontiers in Pediatrics (Children and Health Section, 2021), and Frontiers in Public Health (2021).
26. **Paredes-Moscosso SR**. Awarded Secondment, at StabVida (Portugal) as part of the EU-funded LungCard Project (Aug. 2022); Selected facilitator for course on Science Diplomacy, AAAS, USA and The World Academy of Sciences (Aug. 2022); Attended the 10th Annual Symposium on Global Cancer Research, organized by the NCI/NIH (Virtual, Mar. 2022); Invited peer reviewer for 'Funding Research' 2021-II call by Universidad Católica de Santa María (Peru, Nov. 2021).
27. **Pirlog R**. Completed 9 month Fulbright scholarship at MD Anderson Cancer Center, Tx, USA, on role of tumor microenvironment in early development of lung cancer (May 2022); Established a molecular pathology laboratory; Helped secure funds to sustain a local biobank and its infrastructure for alignment with international consortiums; Selected as one of the future leaders

in pathology by the European Society of Pathology at ESPA- ESP Academy for rising stars in pathology (Jun. 2022); Started a collaboration with Oslo University Hospital, funded by EEA to establish a joint Romanian-Norwegian curriculum for PhD students and young researchers working on 'Involvement of epigenetic dysregulations and non-coding RNAs in the development of neoplastic diseases' (Mar. 2022).

28. **Sayed I**. Associate Professor of Oro-dental Genetics; Supervised, mentored PhD students (2); Presented poster on 'Likely pathogenic and known variants in EDA, EDAR and NECTIN4 in Egyptian families with different forms of ectodermal dysplasia', at European Society of Human Genetics Conference (Austria, Jun. 2022); Abstract on 'Clinical overlap between *NECTIN1* and *NECTIN4* related ectodermal dysplasia' accepted for ASHG annual meeting (to be held Oct. 2022); Identified cases (76)- neurogenetic manifestation (34), skeletal manifestation (19), ectodermal dysplasia patients (15); Referred patients (8) for prosthetic management of ectodermal dysplasia.
29. **Tiong SY**. Elected, Committee member, Genetic Counselling Society (GCSM), Malaysia (2021-2022); Organising Committee member (Publication & Website), speaker on 'Cancer genetic testing', at 2nd Genomic & Genetic Counselling Conference organised by GCSM in partnership with Genetics Society of Malaysia (Nov. 2021); Expanded, mandatory new born screening for 33 types of inherited metabolic disorders (IEM) at Loh Guan Lye Specialists Centre (LSC), along with genetic counselling service (May 2022); Continued to increase awareness among expectant mothers on new born screening (IEM), at Loh Guan Lye Specialists Centre, Penang; New born screening for IEM increased from 7.5% (before Oct. 2021) to 11.7% (Oct. 2021-Jun. 2022); Received new referrals (48) for genetic counselling (Oct. 2021-Jun. 2022).
30. **Tumulak MJR**. Thesis panel member for genetic counseling students (10); Organized training on genetic counseling for hemoglobinopathies, cystic fibrosis, and fatty acid oxidation disorders; Member, Scientific Advisory Board for the Food and Nutrition Research Institute InGest Study (Jul. 2022); Genetic counselor for studies on 'Determination of the prevalence of genetic mutation in high risk Filipino hereditary breast cancer patients', 'Interventions using Genomics-based Strategies (InGeSt) towards enhanced nutrition recommendations Phase III: A proof-of-concept randomized controlled trial for the DNA-based nutrition and lifestyle recommendation in overweight and obese Filipino adults'; Provided pre-test and post-test genetic counseling to patients – breast cancer patients (50), cystic fibrosis patients (10), alpha and beta thalassemia patients (700), InGest study participants (20).
31. **Wiafe SA**. Submitted abstract on 'Lyfe languages and health equity' for 45th Human Genetics Society of Australia Annual Scientific Meeting (to be held Nov. 2022); Organized 4th Symposium on Birth Defects and Rare Disease (Hybrid, Apr. 2022); Speaker on 'Report on the Developing Nations Working Group', at the 10th UDNI Conference, Italy (Hybrid, Feb. 2022); Hosted, First African Summit on Rare Diseases, Accra, Ghana (Hybrid, Dec. 2021); Started a congenital anomalies, undiagnosed and rare disease registry, enrolled 150 patients; Enrolled patients (7) in the RDGI-BioMarin MPS Project, diagnosed cases (6)- mucopolysaccharidosis type 2 (3), mucopolysaccharidosis type 3 (3); Enrolled 82 patients in the IHOPE Program, diagnosed cases (68)- glucose-6-phosphate dehydrogenase deficiency (5), Treacher Collins syndrome (5), HBB-related disorders (4), primary microcephaly (3), COL4A1-related disorders (3), spinal muscular atrophy (3), Rubinstein-Taybi syndrome (3), hereditary transthyretin amyloidosis (3), ataxia-telangiectasia (2), COL2A1 related disorders (2), glycogen storage disease type II (2), achondroplasia (2), GRIN2A-complex neurodevelopmental disorder (2), Marfan syndrome (2), osteogenesis imperfecta (2), and 1 each of SLC6A8-related creatine

transporter deficiency, BRWD3-related X-linked syndromic intellectual disability, GJB1-related disorders, Witteveen-Kolk syndrome, LAMA2-related congenital muscular dystrophy, 16p12.2p11.2 deletion syndrome, KCNQ2-related disorders, neuro-ocular syndrome, STXBP1-related neurodevelopmental disorder, AP1S2-related X-linked syndromic intellectual disability, Progeria, SCN1A- related seizure disorders, partial trisomy 13, INSR-related severe syndromic insulin resistance, TAB2-related syndrome, Aicardi-Goutières syndrome, WAC-related intellectual disability, 3q26.33q27.2 deletion, KAT6A syndrome, cytochrome P450 oxidoreductase deficiency with or without Antley-Bixler syndrome, LMNA-related muscular disorders, TCIRG1-related osteopetrosis, TAF1-Related X-linked syndromic intellectual disability, fumarate hydratase deficiency, partial trisomy 3p syndrome, 22q13.3 deletion syndrome, 3p deletion syndrome, 22q13.2-q13.33 duplication.

32. **Yadav S**. Completed Fellowship in Advanced Ultrasound in Obstetrics and Gynecology, from Mediscan, Chennai, India (Jan. 2022); Used materials and knowledge from Summit to create awareness among patients on prenatal screening and its utility; Lectured colleagues and fellow gynecologists on 'Utility of genetics in reproductive issues- recurrent abortions, infertility, prenatal diagnosis' (2022); Conducted CME, lectured on 'Early fetal growth restriction and role of genetic testing in its early onset' (Apr. 2022); Involved in antenatal diagnosis of congenital malformations, regularly performing genetic sonogram (500) and diagnosed rare malformations - transverse limb defects (2), cardiac structural defects(2), omphalocele with increased NT (confirmed as Down's syndrome by invasive testing), chondrodysplasia punctate (1), spine abnormality (3) and facial clefts (1); Involved in preconception and antenatal counselling of patients (20).
33. **Yesilcinar I**. Invited speaker, G2NA webinar on 'Genetics and gnomic competency of Turkish Nurses' (Jan. 2022); Invited Panelist, presented talk on 'Promoting Diversity, Inclusion, and Health Equity in Genomic Nursing', at ISONG (Virtual, Nov. 2021).

2018 Batch

34. **Abdulkareem F**. Completed certificate course in Leadership and Management in Health, School of Public Health, University of Washington, USA (Jul. 2022); Awarded a fellowship by Nigerian Academy of Medicine, FNAMed (Nov. 2021); Member, National Curriculum Committee, drafted Biological Risk Management (BRM) curriculum for Nigerian Universities (May 2022); Course Coordinator for revision of courses in Pathology (Feb. 2022); Chairperson Organizing Committee, Sensitization Public Lecture and Workshop on Biosafety and Biosecurity Management for Staff of College of Medicine and Lagos University Teaching Hospital in Collaboration with Sandia Laboratories, USA (Feb. 2022); Guest Lecturer on 'Recent advances in the molecular diagnosis of colorectal cancer makes personalised treatment possible even in resource-limited settings', at the Annual All Fellows Conference of the Faculty of Pathology, National Postgraduate Medical College of Nigeria (Aug. 2022); Attended BRM Systems Implementation and Material Control Workshop (Mar. 2022); Attended Stakeholders Consultative Forum for a National Pathogen Control and Reporting System in Abuja, Nigeria (Dec. 2021); Attended Emory-Nigeria HIV Research Training Program (EN-RTP)- Mentor-Mentee training, Lagos (Dec. 2021); Attended, 16th Annual General meeting & National Scientific Conference of College of Nigerian Pathologists (Nov. 2021); Invited plenary speaker on 'Pathology of *Helicobacter pylori* associated gastric cancer' at African Assembly of International Academy of Pathology Divisions, Abidjan (Nov. 2021); Invited plenary speaker on 'Pathology biobanking', at African Organization for Research & Training in Cancer (AORTIC,

Nov. 2021); Attended, scientific conference, Faculty of Basic Medical Science, College of Medicine University of Lagos (Oct. 2021); My NGO, FAJIM Medicare Foundation (https: fajimmf.org.ng) continued to positively impact through its programs; Held free community health outreaches (3), screened patients (3661), provided free treatment for patients (1198); Increased awareness, contributed to dissemination of knowledge and education, awarded 32 new scholarship grants to medical undergraduate (Dec. 2021) students.

35. **Adewole OO**. Visiting scientist to Lung Cancer Unit, Princess Margaret Cancer Center, Toronto, Canada (Nov. 2021- Feb. 2022); Lectured postgraduate medical doctors on 'Tuberculosis and respiratory impairment'; Lectured Pulmonary Fellows on 'Genomics of lung cancer' at The Nigerian Thoracic Society meeting (Nov. 2021); More medical students and residents developed interest in genomics and its relevance to modern practice; Teaching of genetics was strengthened at the undergraduate level and will be updated at the next review of curriculum; Identified a case of tracheoesophageal fistula, referred for surgical management; Provided family with education, support and assistance through medical social workers.
36. **Adeyemo TA**. Established the Centre for Clinical Trials, Research and Implementation Science (CCTRIS), at the University of Lagos, for clinical research, training, networking, and to provide support to the clinical research community in Nigeria; Facilitator, and lecturer on 'Hemoglobinopathy: Definition and types', 'Exchange blood transfusion', 'Basic principles of laboratory diagnosis of sickle cell disease' and 'Practical steps in genetic counselling' at the Genetic Counselling workshop for Sickle Cell Disorder, at Sickle Cell Foundation of Nigeria (Feb. 2022); Facilitated at the Workshop for Biological Risk Management at the College of Medicine, University of Lagos in collaboration with Sandia National Laboratories USA (Feb. 2022); Delivered lectures to sensitize the university and hospital community about practices and principles in biosafety and biosecurity and to create awareness of decontamination practices.
37. **Cornejo-Olivas M**. Principal Investigator, Coordinator of the first DNA Bank in the public system in Peru; Appointed President Council for Rare and Orphan Disorders by Peruvian Ministry of Health; Elected, Executive Committee member for the Pan American Section of the International Parkinson and Movement Disorder Society (2021-2023); Presented posters on 'Exploring effect of known Alzheimer disease genetic loci in the Peruvian population', 'The Peruvian Alzheimer Disease Initiative (PeADI)- An international effort model to increase diversity in Latinos AD research' and on 'Admixed ancestral composition with Amerindian predominance' at AAIC meeting (Jul. 2022); Faculty, at 4th Pan American Parkinson´s Disease and Movement Disorders Congress (May 2022); Awarded, Annual Research Publication Award, Instituto Nacional de Ciencias Neurológicas (Dec. 2021); Mentored trainees (3) from Northern Pacific Global Health Research Fellows Training Consortium (FIC/NIH), Global Brain Health Institute, University of California, San Francisco and Young Members MDS Mentoring Program.
38. **Daich Varela M**. Continued PhD at University College of London, London, UK; Continued working on gene therapy and pharmacotherapy clinical trials; Coordinator of ophthalmic genetics module for post-graduate degree in Ophthalmology, University of Buenos Aires, Argentina; Speaker on 'Inherited retinal disorders', in London (Feb. 2022) and in Palestine (Mar. 2022); Evaluated ophthalmic genetics patients; identified, diagnosed and managed cases (50)- CSNB (10), cone-rod dystrophy (20), coloboma (5), albinism (5) and other rarer syndromic diseases (10); Referred all for genetic counseling and support.
39. **Das S**. Assistant professor, Biochemistry and Medical Genetics at Xavier University School of Medicine, New York (Aruba Campus); Completed Clinical Prenatal Genetics training, from BC

Natal, Faculty of Medicine, University of Barcelona (Mar. 2022); Invited speaker on 'Introduction to genetic disorders', organized by the United Nations, International Office of Migrations, Aruba (Aug. 2022).

40. **Dueñas-Roque MM**. Medical Geneticist at The Genetics Dept., Hospital Nacional Edgardo Rebagliati Martin; Professor, Medical Genetics for undergraduate medical students at Medical School, Universidad San Ignacio de Loyola, Perú; President of the Postgraduate Residency Program of Medical Genetics at Universidad Nacional Mayor de San Marcos, Perú; Member, IRB at HNERM and National IRB for COVID-19 clinical trials; Collaborated with other Summiteers on research and global educational efforts; Enrolled in Genomic Variant Analysis & Clinical Interpretation Course (GUaRDIAN Consortium, 2022); Enrolled as young investigator in Global Genomic Medicine Collaborative (G2MC, 2022); Summit influenced, helped establish an Undiagnosed Disease Program (UDP) at Hospital Edgardo Rebagliati Martins, EsSalud; Continued efforts with the registry of congenital malformations at HNERM (ECLAMC); Enrolled cases (78 trios) in iHope, Illumina Project for identification of genetic causes of rare diseases in Peru.
41. **Dutta AK**. Started e-Sanjeevani Telemedicine Pediatric Genetics OPD at AIIMS Kalyani; Faculty for Silchar Medical College Multidisciplinary Research Unit Seminar Series, presented on 'How Genomics is transforming clinical medicine' (Jun. 2022); Resource person for Jagadis Bose Scholars Professional Development Forum Expert Talk Series on ' The journey of genetic diagnostics: From Holmes to AI' (Apr. 2022); Joined the Global Genomic Medicine Collaborative (G2MC) Young Investigator Forum Writing Project (2022).
42. **Dwivedi A**. Initiated a Bioinformatics and Computational Biology Laboratory at Army Hospital R&R, New Delhi, equipped with next generation sequencing (NGS) analysis; Created, designed various platforms to spread awareness on genetic disorders among armed forces personnel, and families; Conducted webinars/seminars on prenatal genetics, oncogenetics, lysosomal storage disorders, ocular genetics etc.; Used materials from Summit to educate fellow Resident researchers; Member of Global Genomic Medicine Collaboration and Organization of Rare Diseases, India; Presented poster on 'Yield of whole exome sequencing in patients with abnormal head size' and 'Novel pathogenic variants causing TBCK syndrome in three patients of Indian origin', at the Annual ACMG (Nashville, Mar. 2022); Presented poster on 'To evaluate the spectrum of single gene disorders in prenatal cohort' and 'Can chromosomal microarray analysis in fetuses with structural anomalies replace karyotype: Our initial experience', at the World Congress of The International Society of Ultrasound in Obstetrics and Gynecology (Virtual, Oct. 2021); Diagnosed patients (3200) with various genetic disorders- chromosomal, Mendelian, disorders of genomic imprinting; Knowledge from Summit helped manage these patients; Created support groups for Down's syndrome and Angelman syndrome for patients of Indian Armed Forces personnel.
43. **Edem-Hotah J**. Elected, Dean of the Faculty of Nursing and Midwifery at the College of Medicine and Allied Health Sciences, University of Sierra Leone (Jun. 2022); Appointed Coordinator, Integrated Disease Surveillance Response Project to train faculty at Eastern University of Technology and University of Sierra Leone (Jun. 2022); Appointed Coordinator, used materials from the Summit to develop curriculum for MSc in Pediatric and Neonatal Nursing Program, College of Medicine and Allied Health Sciences (COMAHS), included genetics, research and management of babies and critically sick children (Mar. 2022); Observed increased awareness for genetics amongst COMAHS faculty and students, as well as technical and political leadership of the Ministry of Health and Sanitation; Facilitated the faculty

mentorship training for nurses and midwives in Pediatric and Neonatal Nursing at COMAHS, Princess Christian Maternal Hospitals and Ola During Children's Hospital in Freetown.

44. **Eshete MA**. Work impacted by COVID-19.
45. **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 (12) for genetic counselling and medical management; Helped allay anxiety due to known pathway to care.
46. **Lertwilaiwittaya P**. Started Residency in Combined Internal Medicine and Medical Genetics Residency Program at University of Alabama at Birmingham, USA (2021-2025); Awarded Thomas N. James Award for Excellence in Research by a Resident for poster on 'Cost effectiveness analysis of BRCA1 and BRCA2 testing in high-risk Thai woman', at the University of Alabama at Birmingham Trainee Research Symposium (Apr. 2022); Consultant to Health Economics Team and successfully enrolled BRCA1 and BRCA2 as a fully reimbursable genetics test under Universal Health Care Coverage for Thai women with breast/ovarian cancer (one of the first in the country); Participated as a translator for the Exposure Notification System for COVID-19 in Thailand with Thailand's Ministry of Public Health, Apple and Google (2021-2022 https://g.co/ENS/r/th).
47. **Mburu S**. Designed 2 new graduate level (MS) courses in Clinical Medicine 'Oncology & Palliative Care' and 'Forensic Medicine' at Dept. of Clinical Medicine, School of Health Sciences, Kirinyaga University, Kenya, based on knowledge and skills acquired at the Summit; Helped build research capacity, conducted a workshop on grantsmanship titled 'Writing a successful grant application', at the School of Health Sciences, Kirinyaga University (Kenya, Apr. 2022); Presented poster on 'Vitamin A immunization uptake, awareness and childhood infections: A case study of pregnant women visiting antenatal clinic at Kerugoya Hospital' at Kirinyaga University 5th International Conference (Mar. 2022); Speaker on 'Precision medicine in the context of breast cancer', at the School of Health Sciences, Kirinyaga University (Kenya, Nov. 2021); Helped integrate genetics in the Dept. of Clinical Medicine (clinicians, researchers), build capacity in genomics in the county and sub-county hospitals; Strengthened health care systems for the county and at the national level.
48. **Messaoud O**. Visiting Scientist at Translational Dept., Genomic Centre, King Faisal Specialist Hospital and Research Centre, Saudi-Arabia (Jul. 2022); Used resources from Summit for lectures on 'Basic concepts in genetics', at Faculty of Medicine of Tunis, 'Genetics and omics techniques' at Higher Institute of Medical Technologies of Tunis, 'Genomics of the living world' at Higher Institute of Biotechnology of Sidi Thabet, Tunisia; Lectured on 'Next generation sequencing and examples of genomic applications' at Institut Pasteur de Tunis (May 2022); Principal organizer for 'Interdisciplinary investigations of the archaeological site of Thugga in Dougga', Arab-German Young Academy of Sciences and Humanities, Institut Pasteur de Tunis, National Heritage Institute, Carthage National Museum and The Archaeological Site of Thugga (May 2022); Principal organizer for 'Campaign targeting the problem of antibiotic resistance', Arab-German Young Academy of Sciences and Humanities at Institut Pasteur de Tunis and Children Hospital, Bechir Hamza, Tunis (Mar. 2022); Reviewed abstracts for posters and oral presentations as Young Investigator in the Global Genomic Medicine Collaborative (https://g2mc.org); Journal Reviewer for- Frontiers in Genetics, Journal of European Academy of Dermatology Venereology.
49. **Mistri M**. Promoted to Senior Scientist (Inherited Genomics & Metabolism) at Neuberg Centre for Genomic Medicine (Gujarat, India); Involved in NGS based diagnostic services (clinical

exome, whole exome, whole genome sequencing and newborn screening); Awarded 2nd prize, best poster on 'Importance of paternal family history in ascertaining risk assessment in breast cancer', and 3rd prize, poster on 'Importance of post-test genetic counseling in establishment genotype-phenotype correlation: A case study based rare findings in ZNF699 gene', at the 7th Annual International Conference of Board of Genetic Counseling of India (Jul. 2022); Journal Reviewer for-The Cureus Journal of Medical Science.

50. **Muttamba W**. Involved in preliminary discussions with University of Cincinnati on use of stored samples obtained from a cohort of asthmatic patients in East Africa for genomic work.
51. **Naeem MA**. Lectured on 'Advanced human molecular genetics' as part of curriculum to graduate level students (2021-2022); Participated in NEI-GEGC Educational Workshop 'International Collaboration, NEI Grant Writing & NIH Review Process' (Virtual; Jun. 2022); Member, Organizing Committee, 4th International Symposium on 'Advances in molecular biology of plants and health sciences', National Centre of Excellence in Molecular Biology (CEMB), University of the Punjab, Lahore (Pakistan, Dec. 2021); Supervised, trained graduate school students (2 MS/MPhil); Trained MS/MPhil researchers in collaboration with Dr. Z. Hussain at Dept. of Zoology, University of Education, Lahore, Pakistan.
52. **Nakousi-Capurro N**. Clinical Geneticist at Hospital Carlos Van Buren of Valparaíso, Chile; Faculty at Universidad de Valparaíso as Associate Professor; Summit provided tools and motivation to train others, set up research/clinical facilities and introduce genomics in the curriculum; Established sequencing capabilities at Basic Sciences Dept., Valparaíso University, completed pilot testing, started services for the public (Sept. 2022); Established a Neurogenetics Clinic at the Carlos Van Buren Hospital, included specialists in Pediatric Neurology and Clinical Genetics who see patients (~2/week); Held weekly meetings, advised Neuropediatric Specialists from Carlos Van Buren and Claudio Vicuña Hospitals, Valparaíso on suspected genetic conditions, helped long distance patients (>70 kms); Provided on-site training to postgraduate residents (1each in Pediatrics, Pediatric Neurology, Neurology, Neonatology, 2 Obs/Gyn), from Valparaíso University at the Carlos Van Buren Hospital; New curriculum, introduce one-month rotation in Clinical Genetics for Ob/Gyn, Pediatric Neurology Residents from the University of Valparaiso (Jan. 2023); Diagnosis improved lives of patients and families, allowed for better healthcare; Genetic counselling helped with informed reproductive decisions, directed patients to preimplantation/prenatal diagnostic testing to reduce risk of recurrence of genetic conditions.
53. **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; Chairman, Committee on Clinical Governance, Nigeria Medical Association, Lagos State Chapter; Member, Dental Therapist Registration Board of Nigeria; Delegate representing Nigeria at the World Dental Parliament Meeting (Sep. 2022); Keynote speaker at the annual workshop of the Dental Therapist Registration Board of Nigeria, on 'Management of oral cancer: The role of dental therapist and dental surgery technician', organized by the Dental therapist Registration Board of Nigeria (May 2022); National Liaison Officer of Nigerian Dental Association (2019 –to date); Coordinator Brush Twice Daily Programme organized by Unilever Global/ Nigeria. (2008- to date).
54. **Ottaru S**. Trained in 'Introduction and history of vaccines in Africa' organized by East Africa Center for Vaccine and Immunization, 8th Vaccinology Course for Health Professionals, University of Makerere (Uganda, Jun. 2022); Attended, course on 'Fundamentals of early childhood development for pediatricians', organized by Kenyan Pediatric Association and American Academy of Pediatrics (Feb. 2022).

55. **Savina O**. Member-Consultant, European Union Cooperation in Science and Technology Action Aniridia- network (EU COST-NET) to address unmet medical, scientific, and societal challenges (Feb. 2022); Held webinars on 'Introduction to opthalmogenetics' and 'Types of inheritance of ophthalmic diseases- What they don't teach in school' (Ukraine, Feb.- Jun. 2022); Presented on 'Ophthalmogenetics, complications' at Khmelnytskyi City Children's Hospital (Ukraine, Apr. 2022); Provided ophthalmogenetics consultation at Dobrobut Medical Center, Kiev, Ukraine; Introduced ophthalmogenetics consultation at Altris Inc. with Dr. Maria Znamenska; Supervised, identified genetic eye disorders cases (31): aniridia (12), retinitis pigmentosa (8), Usher syndrome (2), Stargardt macular dystrophy (1), vitelliform macular dystrophy (2), Leber congenital amaurosis, type 7 (1), X-linked retinitis pigmentosa (2), family exudative vitreoretinal degeneration (1), colobomas of the retina and optic nerve (2); Performed genetic testing, identified mutations, provided genetic counseling.
56. **Taiwo O**. Guest lecturer on 'What evidence? Identifying good evidence for research', at International Association for Dental Research (Nigeria Division) Mid-Year Conference (Online, Jun. 2022), 'Introduction to research methodology (Part I - V), at Nigeria Dental Association Continuing Medical Association Series (Online, Nov. 2021- Jun. 2022); Provided 9-week training to Nigeria Dental Association Plateau State Branch (Online, Jan.- Mar. 2022); Delivered lecture on 'Choice of statistical tests for data analysis', at Africa Health Exhibition and Conferences, Cairo (Egypt, Jun. 2022); Speaker on 'Role and training needs of community pharmacists for improving oral health awareness in Nigeria', at International Pharmaceutical Federation (Collaboration with the International Dental Federation) Webinar on The Roles of Pharmacists in Oral Health (Online, May 2022); Speaker on 'Dental caries and periodontal diseases', at National Postgraduate Medical College of Nigeria Virtual Revision Course (Jan. 2022); Trained supervisors, enumerators on COVID-19 mitigation, provided overview of SMC implementation, study design, household selection, data collection for seasonal malaria chemoprevention (SMC) execution of coverage survey for Plateau State, Nigeria (Nov. 2021).
57. **Utumatwishima JN**. Received scholarship from Swedish International Development Agency for PhD on 'Investigate the association between maternal mental health and undernutrition of children in the Northern Province of Rwanda' at Sahlgrenska Academy, University of Gothenburg, Sweden (Oct. 2021).
58. **Wangi KYW**. Pursuing PhD (2nd yr) at Penn State University, USA (Jul. 2022); Presented on 'Social issues, stigma, and human dignity in Indonesian thalassemia community' at the Indonesian National Seminar Series on 'Screening, prevention, and health issues in Thalassemia' at Duquesne University, PA (Jul. 2022); Attended online Genomic Competencies Course for Nurses From Theory to Application, at School of Nursing Duquesne University (Jan.- Mar. 2022); Submitted proposal on 'Eugenics, ethics, and nursing', for Nursing Philosophy Fellowship to University of CA, Irvine (2022); Lecturer on 'Midwife care in women and children with vulnerabilities- emphasis on genetics in thalassemia', undergraduate Midwife Program at School of Midwifery, Faletehan University, Banten-Indonesia (Online, Feb.-Jun. 2022); Presented talks on 'This life would be meaningful human dignity in a beta-Thalassemia major patient with dependence transfusion' (May 2022), 'Ethical issue in human gene editing research' (Apr. 2022), 'Gene editing technology and utilitarianism' (Dec. 2021), at Pennsylvania State University; Wrote a proposal on 'Public health ethics attitude in gene editing technology' for PhD program at Pennsylvania State University (Nov. 2021).

2017 Batch

59. **Abad PJ**. Accepted for PhD at The University of Iowa College of Nursing (Aug. 2022); Member, Scientific Advisory Board on 'Interventions using genomics-based strategies towards enhanced nutrition recommendations', Dept. of Science and Technology, Food and Nutrition Research Institution (Mar. 2021- Feb. 2022); Abstract reviewer for Congress of the International Society of Nurses in Genetics (USA, Nov. 2022); Resource person on 'Genetic counseling for childhood cancer', webinar organized by the Philippine Oncology Nurses Association (Jul. 2022); Resource person on 'Genetic counseling in the Philippines: Opportunities and future directions', at the 46th Annual Meeting of the Japanese Society for Genetic Counseling (Jul. 2022).
60. **Avogbe PH**. Used resources from Summit to design new courses in Cancer Genetics and Genomics (undergraduate, graduate level) at Polytechnic School at the University of Abomey-Calavi, Benin, included Next-generation sequencing methods, biomarkers and molecular diagnostics, sequences data retrieval and manipulations in databases (NCBI, UCSC, Ensembl); Delivered new courses in biochemistry, molecular biology, cytogenetics.
61. **Benítez-Cordero Y**. Member, International Clearinghouse for Birth Defects Prevention & Research; Mentored residents (7) in clinical genetics; Evaluated genetic cases (18) of monogenic, chromosomal, or multifactorial etiology; Codified cases (1258) for the Registry; Referred cases (4) for genetics analysis and medical management- congenital myopathy (2), overgrowth syndrome (1).
62. **Casado PL**. Trained, supervised graduate students (2 PhD, 2 MS) for research in molecular biology and implant dentistry, at Fluminense Federal University, Brazil; Editor for- Brazilian Dental Journal.
63. **Dhoro M**. Lectured on 'Genetic basis of disease and gene therapy', 'Diagnosis of genetic disorders', 'Genetic counselling', 'Genomic medicine and pharmacogenetics' to graduate level students (medical doctors, postgraduates) in Clinical Pharmacology and undergraduates pursuing Medical Analytics and Informatics; Mentored graduate level students (MPhil, Clinical Pharmacologists) on research projects and proposals related to HIV genetic diversity and natural polymorphisms of the integrase gene. Appointed member, African Tuberculosis Bioinformatics Training Program (NIH D71/D43), a collaborative initiative to strengthen bioinformatics training in Africa for infectious diseases; Attended, planning meeting and faculty development workshops on research proposal and grant writing; Attended 4 workshops held by the Southern African Training & Research Network for Genomic Surveillance of Antimicrobial Resistance Project (SAGESA); Appointed Panel Member and Reviewer for Africa Rapid Grant Fund, Covid-19 Programme Call (since 2020); Reviewed, edited abstracts for University of Zimbabwe Annual Medical Research Day and the African Forum for Research and Education in Health (2022).
64. **Fatima SS**. Completed 2- year Medical Genetics and Genomics Certificate Program (NHGRI, Oct. 2021); Completed 2-year Fellowship in Health Professional Education and Leadership, from the Foundation for Advancement of International Medical Education and Research (FAIMER-ECFMG, Jun. 2022); Introduced, developed curriculum for Fetal or Prenatal Programs for undergraduate and graduate studies; Delivered lectures on Advanced Molecular Genetics to graduate level students; Appointed External Examiner, reviewed theses (2PhD, 7 MPhil); Supervised graduate research students (4 PhD, 3 MPhil) on molecular and genetic basis of metabolic syndrome and its related disorders; Member, Epigenetics Society and the Developmental Origins of Health and Disease.

65. **Hitayezu J**. Faculty member at University of Rwanda, Coordinator/Chair of teaching modules; Instructed Pediatric Emergency Medicine (PEM) and Pediatric Critical Care modules to Pediatrics & Child Health Residency Program, University of Rwanda, Kigali (2021-2022); Co-course Director, Pediatric Basic Course & Point-of-Care Ultrasound workshop at The University Teaching Hospital of Kigali, CHUK (Jul. 2022).
66. **James O**. Impacted by Covid.
67. **Lotz-Esquivel S**. Admitted to Clinical Genetics and Rare Diseases Master Program in Universitat de Barcelona – Hospital Sant Joan de Déu, Spain (start Oct. 2022); Held joint positions at Medical Genetics and Metabolism Dept., National Children's Hospital (NCH) and Rare and Orphan Disease Multidisciplinary Clinic, Hospital San Juan de Dios (ODMC); Provided care for pediatric patients with inborn errors of metabolism and genetic conditions at NCH (4 patients/week) and at ODMC (8 patients/week); Aided in smooth transition of adolescent patients and their informaton to the adult hospital; Developed and updated a self-sustaining database, facilitated extraction of relevant patient data for diagnosis, treatment and for analysis and publications (~320 patients); Attended Latin American Society of Inborn Errors of Metabolism and Neonatal Screening (SLEIMPN) XII Congress (Dominican Republic, May 2022), promoted exchange of ideas, experiences, and scientific advances among specialists in the region; Knowledge, experience attained at the Summit (2017) and MGGCP (2021) advanced my academic training in genetics and helped contribute to developing genetics and genomics in Costa Rica; This helped improve lives of patients with rare genetic diseases and their families through appropriate diagnosis, treatment, management and support.
68. **Mahfoudh W**. Selected for Fulbright Visiting Scholar Program (2022-2023) at The University of Chicago (May- Aug. 2023); Obtained 'Certificate in Oncogenetics' from Faculty of Medicine of Sousse, Tunisia (May 2022); Supervised graduate students (MS) on immunogenetics of COVID-19.
69. **Mehrez MI**. Attended online webinar, 'The ins and outs of sustaining a longitudinal cohort for genetic studies', Columbia University (Mar. 2022); Attended an online webinar, 'Genetic updates in management of beta-thalassemia', Arab Association of Genetic Research (Feb. 2022); Presented 'Biological effects of ionizing radiations' on the Science Day for Radiation Hazards and Safety in Healthcare (Cairo, May 2022); Presented poster on 'Oculo-dento-digital syndrome revealed as cause of rootless teeth by WES', at Genetics of Rare Diseases Conference (Virtual, Mar. 2022); Presented 'A glimpse at psychosocial counseling in Egypt- A pilot study', at the First Annual Conference of UNESCO Chair in Bioethics: Biotechnology: Ethical Challenges and Opportunities (Virtual, Mar. 2022); Identified cases (3)- Goldenhar syndrome (1), Oculo-dento-digital (1), Escobar syndrome (1); Referred cases (4) for dental management- mandibuloacral dysplasia (2), oculo-dento-digital (1), Progeria (1); Dental management of conditions resulted in improved quality of life and psychological well-being of patients; Journal Reviewer for- Orphanet Journal of Rare Diseases (Feb. 2022).
70. **Mgasa A**. Attended a course on Cancer Clinical Genetics in Africa, organized by The African Oncogenetics Network (Nov. 2021); Trained in Vaccinology in Africa, a virtual Master Level course, University of Oxford (Oct. 2021); Working on establishing a Medical Genetics Clinic at PCMC Polyclinic, Kigamboni Branch, Dar es Salaam (Nov. 2022).
71. **Nandal R**. Teaching Faculty for DM Cardiology and DNB-SS Cardiology courses at Pandit Bhagwat Dayal Sharma University of Health Sciences, Rohtak, India; Lectured MBBS students and MDs; Conducted weekly congenital heart diseases clinics; Used pedigree charting, facial dysmorphism recognition tools in dysmorphic patients with congenital heart diseases at the

Coronary Heart Disease Clinic (CHD); Maintained pedigree charting, genetic testing, counseling records for patients with cardiomyopathy, sudden death suspected channelopathy, hyperlipidemia and congenital heart disease; Identified syndromic children (30) in collaboration with pediatrician and geneticist in CHD clinic and NICU/PICU- Noonan syndrome (5), Marfan syndrome (1), William syndrome (2), Di Georgi syndrome (2); Referred all identified and suspect syndromic and dysmorphic patients (20) for genetic counselling; Updated congenital heart disease registry.

72. **Otaify GA**. On secondment as Medical Genetic Consultant, at Sultan Qaboos University Hospital, Oman (Sep. 2021); Instructor for newly established Omani Fellowship Program of Medical Genetics; Trained Residents, instructed undergraduate students in medical genetics; Summit materials helped in preparation of lectures on disorders of sex development, craniosynostosis, syndromic and non-syndromic deafness, Down's syndrome, approaches to dysmorphic child, hereditary musculoskeletal disorders; Knowledge acquired at Summit helped in developing better approaches for diagnosis and counseling of genetic cases at the outpatient genetic clinic (8-12 patients/week) and for inpatients admitted in different hospital departments; Identified syndromes included- Achondroplasia (2), Marfan syndrome (1), Neurofibromatosis 1 (3), Prader-Willi (4), Sotos (1), Short rib thoracic dysplasia (1), McCune-Albright (1), Noonan syndrome (1).
73. **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.
74. **Petlichovski A**. Established, The Genetic Counseling Service Center at the Institute for Immunobiology and Human Genetics (Macedonia) which now operates full-time; Collaborated with genetic testing centers like Invitae, Cegat for testing rare conditions; In-house testing for common disorders was developed to include inherited metabolism disorders, cardiomyopathies, immunodeficiencies etc.; Performed genetic testing, provided genetic counseling for patients (>70) with rare genetic disorders - Usher syndrome (1), X-linked recessive agammaglobulinemia (1) Autosomal dominant myopathy and Charcot-Marie-Tooth disease (1), Joubert syndrome (2), MPZ spectrum of autosomal dominant neuropathies (1), Lowe syndrome and Dent disease (1), GNE AD Sialuria (1) NF1 AD, Neurofibromatosis-Noonan syndrome and Watson syndrome (1), CLCN1 autosomal dominant myotonia congenita (1), CHEK2 cancer risk (1), Alport syndrome (1), IL10RB autosomal recessive early onset inflammatory bowel disease (1), Cherubism (2), PRRT2 AD neuropathies (2), Osteogenesis imperfecta (3), CHD2 AD childhood onset epileptic encephalopathy (1), ZIC2 AD holoprosencephaly (1), RANBP2 AD infection induced necrotizing encephalopathy (1), Rett syndrome (2), Epidermolysis bullosa (1), Helsmoortel-van der AA syndrome (1) Acrofacial dysostosis (1), Ehlers-Danlos syndrome (1), CYP1B1 congenital glaucoma (1), Marfan syndrome (1), OTX2 Syndromic microphthalmia (1), CM-AVM syndrome (1), Metatropic dysplasia (1), PAH phenylketonuria (1), WNT10A selective dental agenesis type (4), CONSDIAS neuropathy (1), GRIN2B encephalopathy (1), PDHA1 pyruvate dehydrogenase E1-alpha deficiency (1), Turner Syndrome mosaicism (1), Huntington disease (1), FMF (4), Singelton Merten syndrome (1), Aicardi Goutières syndrome (1) Noonan spectrum RASopathies (5), and for couples (200) with recurrent spontaneous abortion.
75. **Roy S**. Speaker on 'Fight against Retinoblastoma- Hospital and community based activity of CEITC and its impact' and 'New technique of treatment in ocular surface neoplasia', at 9th Ophthalmological Society Bangladesh (Bangladesh, Sep. 2022); Mentored graduate level research students (BSc in Optometry), Institute of Community Ophthalmology, Chittagong University & BSMMU; Identified genetic disorder cases (>90)- retinoblastoma (47),

neurofibromatosis (4), Blepharophimosis Syndrome (5), thyroid ophthalmopathy (35) among others; Treated patients (57), referred some for medical management; Patients (38) were satisfied with their outcome.

76. **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.
77. **Thakur N**. Pursuing PhD dissertation on 'Identification and characterization of new gene variants of sex chromosome responsible for male infertility in two different populations based on exome sequencing at Shobhit University, Meerut, India (since Jul. 2021); Chaired as Panelist in CME program on Molecular Oncology (Khatmandu, Jul. 2022); Speaker on 'Clinical genetics services in Nepal: Current scenario, challenges and future aspects', at National Annual Conference of Nepalese Society of Obstetrics and Gynecologists (Kathmandu, Apr. 2022); Continued prenatal testing by NGS of amniotic fluids at Nova International Diagnostics, Nepal; Knowledge from Summit's tailored training helped in analysis and validation of cancer genetics test results obtained by real time PCR and NGS; Started clinical investigations for preimplantation genetic screening in collaboration with six IVF clinics using WES for rare diseases; Performed non-invasive prenatal testing (200), WES (23), preimplantation genetic screening (300), BRCA1/BRCA2 testing (45) at Nova International Diagnostics, Nepal; Diagnosed- new rare genetic disorders (23) through clinical exome sequencing; Provided genetic counselling, diagnostic services to patients (>600) with genetic diseases - Recurrent pregnancy loss (156), Down's syndrome (42), Turner syndrome (18), chromosomal anomaly (86), rare genetic diseases (53), beta thalassemia (68), sickle cell anemia (32) and congenital birth defects- Duchenne muscular dystrophy (27), spinal muscular atrophy (19), metachromatic leukodystrophy (8), hemophilia A (12), familial mental retardation syndrome (12), Crouzon syndrome (7), disorder of sexual dysfunction (18), cleft lip and palate (17), cleidocranial dysplasia (5), Bartter Syndrome (1), skeletal dysplasia (6), congenital seizure disorder (13), macular dystrophy(7), retinitis pigmentosa (7) at the Medical Genetics Unit, Bir Hospital; Referred patients and their families for appropriate support for improvement in quality of life.
78. **Tolegen N**. Cytogenetic studies on bone marrow of children (164, <18yrs) hospitalized at the Oncohematology Dept., National Research Center for Maternal and Child Health University Medical Center, Astana (Kazakhstan, Jan.- Jun. 2022), helped diagnose acute leukemia cases (164)- acute lymphoblastic leukemia (124), acute myeloid leukemia (39), Biphenotypic AL (1) with the help of expression markers for myeloid and lymphoid lines; Quantitative chromosomal rearrangements included,- mostly trisomy in chromosomes 8, 9, 10, and 21, monosomy mostly in chromosomes 5,7,12, and Y; Structural chromosomal abnormalities were observed in 85 cases- rearrangements, translocations (67), deletions (8), other structural rearrangements (10); Most frequent detected translocations were: t(12;21)(p13;q22), t(8;21)(q22;q22), translocations involving chromosome 11 in region 11q23 with others; Observed relatively higher percentage of cases with tetraploidy (12%), hyperdiploidy (47-50 chromosomes, 7.6%), and high hyperdiploidy (> 50 chromosomes, 6.5%).
79. **Torres-Mejía G**. Supervised graduate students (3 MS) on projects related to gene-environment interaction in breast cancer using Mexican data from Women's Health Clinical Research, in collaboration with Dr. Elad Ziv from UCSF; Two students hosted as Visiting Scientists (3 months) at the UCSF, CA.

80. **Tuncay Yaylacioglu F**. Presented poster on 'Practice patterns and needs assessment of ophthalmologists for inherited eye diseases: Do we need a subspecialty as ophthalmic genetics?', at ARVO (Colorado, May 2022).
81. **Vishnoposka SA**. Taught courses 'Human Physiology' and 'Genome Analyses' to undergraduates and 'Human Genetics' to post-graduates; Presented abstract on 'Argentinian multicentric genetic study of pituitary hormonal deficiencies using a custom panel based on single molecule molecular inversion probes', at ENDO (Jun. 2022).
82. **Wayengera M**. Pursuing MSc in Genetics and Genomics; Chair-Ministerial COVID19 Scientific Advisory Committee (Mar. 2020- to date); In-charge, Unit of Genetics & Genomics, Mulago National Referral Hospital (new super specialty reference center for rare Mendelian disorders); Developed capacity and infrastructure for experimental gene therapy for Mendelian diseases such as sickle cell anemia, modeled on the HIV-1 gene therapeutic model; Worked on establishing fully fledged clinical and molecular genetics laboratory for patient care, research and training; Worked with Uganda Cancer Institute and partners on integrating genetic testing and counseling in cancer care, through implementation of the National Cancer Policy- provides opportunity for integration of clinical and molecular genetics for common diseases in Uganda; Regularly examined patients (2-3/week); Provided diagnosis, options for treatments, prevented drain on funds on witch-doctors; Provided counseling, helped patients and families cope with diagnosis.
83. **Yousef YA**. Received the 2021 King Hussein Cancer Research Award for 'Young Investigator' best research project on 'Retinoblastoma genetics, prognostics, and management outcomes'; Identified, diagnosed, treated patients with retinoblastoma (24); Referred genetically tested with familial and non-familial retinoblastoma (21) for counseling and medical management; Informed patients negative for germline disease about extremely low risk of transmission to offspring; Improved quality of life by alleviating stress.