ADDENDUM

ISHGG FIFTH ANNUAL OUTCOME (October 2022- September 2023)

(*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 and 2017 Batches completed their 5-year follow-up in 2021 and 2022 respectively, hence this compilation only includes the reports from Batches 2018 and 2019.*

A. Publications: (Total- 170; 2019- 85; 2018- 85)

2019 Batch

1. **Abad PJB**, **Tumulak M-AJR**, Guerbo R, de-Castro-Hamoy L, Bautista NG, Nuique R, Jacalan FI, Talapian GL, Felipe-Dimog EB, et al. Landscape of genetic counseling in the Philippines. J Genet Couns. 2023. (Submitted).
2. Abdel-Hamid MS, Hassib N, **Sayed ISM**, Elhossini R, Aglan M. A recurrent KCNK4 variant in a dominant pedigree with hypertrichosis and gingival fibromatosis syndrome: Variable phenotypic expressivity and insights. Clin Genet. 2023. (Submitted).
3. Afia E, Nwegbu MO, Inyang BA, Nwachukwu KC, **Alli LA**, Okoh MP. Evaluation of the antihypertensive activity of Pandiaka Heudelotii leaf Extract (PHE) on Wistar rats using biochemical parameters as indicators. Curr Res Compl and Alt Med. 2023. (Submitted).
4. Ahmed OH, Abd Alrahman N, **Alabid T**, Babekir SE, Mergani MM, Babiker NE. Serum ferritin level among Sudanese women with hypertensive disorders of pregnancy, Khartoum State 2023. Archives of Gynaecology and Women Health. 2023 Jul 14; 2(2). doi 10.58489/2836- 497X/018.
5. Ajala-Lawal R, Inyang BA, Nwachukwu KC, **Alli LA**, Okoh MP. Effects of 1, 3 dichloro-2-propanol oral gavage on male fertility parameters in male Wistar rats. 2023. (In preparation).
6. Ajiboye T, **Alli LA**. Computational analysis of fermented n-hexane extract of Pentaclethra macrophylla seed on glaucoma induced rats. 2023. (In preparation).
7. Akçay E, Delioğlu ENE, **Kars ME**, Demir P, Çağıl N. Evaluation of unilateral corneal collagen cross-linking on fellow untreated eyes of patients with keratoconus. Arq. Bras. Oftalmol. 2022 Nov 4; S0004-27492022005011209.
8. Alebiosu CO, Ayodele OE, Akinbodewa AA, **Okunola OO**. Chapter on 'Autosomal dominant polycystic disease'. Companion to Tropical Medicine. Obafemi Awolowo University Press, ISBN 978-978-136-105-0. 2023; pp 336-339.
9. Algader A, Alsiddig A, Abdallah EKA, Ibrahim WS, Hame SA, Gassoum A, Merghani MM, **Alabid T**, Ibrahim AE, Babiker NE. Prothrombin gene polymorphism (G20210A) among Sudanese patients with intracerebral hemorrhage, Khartoum State. Int J Med Sci Clin Invent. 2022 Nov 9; 9(11):6310-6317.
10. **Amarakoon G**, Rao S. A child with ADA2 deficiency- VAIH syndrome. 2023. (In preparation).
11. **Amarakoon G**, Rao S. Self-assessment. Paediatr Child Health. 2023 Aug; 33(8):250-252.
12. **Amarakoon GGGT**, Wijerathne NPKP, Chandrakumara WAJC, Wijayarathne HK. Clinical profile of Down syndrome in a tertiary care centre -Sri Lanka. 2023. (In preparation).
13. **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. 2023. (In preparation).

14. Babiker NE, Ahmad BAA, Mohammed FAA, Mohammed AYB, Abdelrhman AIH, Abdalrehim DA, Merghani MM, **Alabid T**. Coagulation parameters among Sudanese individuals vaccinated with Johnson and Johnson vaccine at Khartoum State, 2022. American Journal of Clinical and Experimental Medicine (AJCEM). 2023 Jan 13; 11(1):9-16.
15. Baynam G, Hartman A, Letinturier MCV, Bolz-Johnson M, Carrion P, Grady AC, …, **Wiafe SA**, Bjornson-Pennell H, Groft S, et al. Global Health for Rare Diseases through Primary Care. Lancet Glob Health. 2023. (Submitted).
16. Benjith Paul K, Bose S, **Mutreja D**. Lucio phenomenon or necrotic erythema nodosum leprosum in an untreated case of leprosy?? A diagnostic dilemma. Lepr Rev. 2023; 94(2):176-181.
17. Chan SC, Beh HC, Jeevajothi Nathan J, Sahadeevan Y, Engkasan JP, Chuah SY, Pek EW, Abdullah N, Wong CK, **Hussein N**, et al. Pulmonary rehabilitation capacity building through a Teach-the-Teacher Programme: A Malaysian experience. J Glob Health. 2023. (Accepted).
18. Chan SC, Patrick Engkasan J, Sekhon JK, Nathan JJ, Liew SM, **Hussein N**, Suhaimi A, Hanafi NS, Pang YK, et al. Home-pulmonary rehabilitation program for patients with chronic respiratory diseases in Malaysia: A mixed method feasibility study. J Glob Health. 2023. (Submitted).
19. Chandrakumara J, Wijesundara M, **Amarakoon G**. A child diagnosed with severe hemophilia A presenting with nephrotic syndrome: A case report. J Med Case Rep. 2023; 17(1):206.
20. Cismaru CA, **Pirlog R**, Calin GA, Berindan-Neagoe I. Stem cells in the tumor immune microenvironment–part of the cure or part of the disease? Ontogeny and dichotomy of stem and immune cells has led to better understanding. Stem Cell Rev Rep. 2022 Dec; 18(8):2549-2565.
21. Felipe-Dimog EB, **Tumulak M-AJR**, Garcia AP, Liang F-W, Silao CL, Hsu M-T, Saragih ID, Sia-ed AB. Caring behavior of Filipinos toward their elderly family members. Acta Med Philipp. 2023. (Submitted).
22. Freire LM Daza-Millone MA, Becerra-Hernández LV, **Paredes-Moscosso SR**. When reason is nourished by affections: reflections of young scientists on the challenges of science in Latin America. Horiz Med. 2023 May 30; 23(2):e2243. doi.org/10.24265/horizmed.2023.v23n2.14.
23. Goyal B, Manrai M, **Mutreja D**, Aggarwal R, Maurya P. Hepato Pancreatic Biliary (HPB) tuberculosis: Case series of six cases and literature review. Surg Case Reports. 2022; 6(3):1-4.
24. **Guven Y**, Saracoglu HP, Aksakal SD, Kalayci T, Altunoglu U, Uyguner ZO, Eraslan S, Borklu E, Kayserili H. Nance-Horan Syndrome: Characterization of dental, clinical and molecular features in three new families. BMC Oral Health. 2023 May 23; 23(1):314.
25. **Gyawali P**, Bhatt RD, Karmacharya RM, Pant V, Khadka A. High burden of vitamin B12 deficiency among adults and elderly visiting a tertiary care hospital. J Nepal Health Res Counc. 2023 Mar 10; 20(3):702-707.
26. **Gyawali P**, Shrestha R, Karmacharya RM, Baidya N, Sharma S. Holo-transcobalamin deficiency among type 2 diabetic patients under metformin therapy. 2023. (In preparation).
27. Hamad MN, Mohamed FI, Osman MM, Jadid AA, Abdalrhman IK, Yousif AM, **Alabid** T, Edris AMM, Mohamed NS, et al. Molecular detection of Epstein-Barr virus among Sudanese patients diagnosed with Hashimoto's Thyroiditis. Res Sq. 2023. (Submitted).
28. Haranguș A, Lajos R, Budisan L, Zanoaga O, Ciocan C, Bica C, **Pirlog R**, Simon I, Simon M, et al. Identification of potential microrna panels for male non-small cell lung cancer identification using microarray datasets and bioinformatics methods. J Pers Med. 2022 Dec 13; 12(12):2056.

29. Hassan Obaid SG, Mohamed RA, Abd Albagi RAA, Razan, Abdalla Tendil A, Babiker SE, **Alabid T**, Merghani MM, Babiker NA. The coagulation parameters among healthy menopausal Sudanese women. Int J Med Sci Clin Invent. 2023 May 19; 10(5):6750- 6759.
30. Hurtado-Salgado E, **Ortiz-Panozo E,** Salmerón J, Luna-Gordillo R, Allen-Leigh B, Saavedra-Lara N, Franco EL, Lazcano-Ponce E. Prevalence of cervical human papillomavirus in Mexico, 2010-2017: Analysis of 2.7 million women. Cancer Causes Control. 2023 Feb; 34(2):123-132.
31. **Hussein N**, Lee YK, Mohd Reza S, John P, Azmi A, Ng CJ. Exploring the experiences and beliefs on thalassaemia screening - A qualitative study among high-risk indigenous thalassaemia carriers in East Malaysia. 2023. (In preparation).
32. **Hussein N**, Liew SM, Hanafi NS, Lee PY, Cheong AT, Ghazali SS, Chinna K, Pang YK, Kassim A, et al. Asthma control and care among six public health clinic attenders in Malaysia: A cross-sectional study. Health Sci Rep. 2023 May 2; 6(5):e1021.
33. **Hussein N**, Ng CW, Ramli R, Liew SM, Hanafi NS, Lee PY, Cheong AT, Sazlina SG, Pinnock H, et al. Assessing catastrophic health expenditure and impoverishment in adult asthma care: a cross-sectional study of patients attending six public health clinics in Klang District, Malaysia. BMC Health Services Research. 2023. (Submitted).
34. **Hussein N**, Ramli R, Liew SM, Hanafi NS, Lee PY, Cheong AT, Sazlina SG, Mohd Ahad A, Patel J, et al. Healthcare resources, organisational support and practice in asthma in six public health clinics in Malaysia. NPJ Prim Care Respir Med. 2023 Mar 27; 33(1):13.
35. Ibrahim WS, Algader AAA, Abdallah EKA, Hasssan MS, Hamed SA, Gassoum A, **Alabid T**, Merghani MM, Babiker N. Factor XIII gene polymorphisms among Sudanese patients with intracerebral hemorrhage, Khartoum State 2022. J Drug Deliv Ther. 2022 Oct 15; 12(5-S):13-19.
36. Isacescu E, Chiroi P, Zanoaga O, Nutu A, Budisan L, **Pirlog R**, Atanasova AG, Berindan-Neagoe I. Melanoma cellular signaling transduction pathways targeted by polyphenols action mechanisms. Antioxidants (Basel). 2023 Feb 7; 12(2):407.
37. Ismail I, Ng CJ, Lee PY, **Hussein N**. Determinants of contraceptive practice among multi-ethnic Asian women with diabetes: A qualitative study. 2023. (In preparation).
38. Jiménez-Cardozo N, Mitsunami M, Mínguez-Alarcón L, **Ortiz-Panozo E**, Wang S, Souter I, Hauser R, Chavarro JE. Iron intake in relation to ovarian reserve among women seeking infertility treatment. Hum Reprod. 2023 Aug 1; 38(8):1613-1620.
39. **Kars ME**, Stein D, Bayrak CS, Stenson PD, Cooper DN, Itan Y. A comprehensive knowledgebase of known and predicted human genetic variants associated with COVID-19 susceptibility and severity. medRxiv. 2022 Nov 07. doi:10.1101/2022.11.03.22281867.
40. Khamis NM, Babiker NE, Khalil SH, Abdelraheem NE, **Alabid T**, Merghani MM. Protein C level among Sudanese Patients with ischemic stroke at Khartoum State. International Journal of Medical Science and Advanced Clinical Research (IJMACR). 2023 Apr; 6(2):375-380.
41. Khoo EM, Abdullah A, Liew SM, **Hussein N**, Hanafi NS, Lee PY, Abdullah KL, Vengidasan L, Abu Bakar AIB, et al. Psychological health and wellbeing of primary healthcare workers during COVID-19 pandemic in Malaysia: A longitudinal qualitative study. BMC Prim Care. 2022 Oct 13; 23(1):261.
42. Kirenga B, Mugenyi L, Sánchez-Rico M, Kyobe H, **Muttamba W**, Mugume R, Mwesigwa E, Kalimo E, Nyombi V, et al. Association of fluvoxamine with mortality and symptom resolution among inpatients with COVID-19 in Uganda: A prospective interventional open-label cohort study. Mol Psychiatry. 2023 Mar 3; 00:1-8. doi: 10.1038/s41380-023-02004-3.

43. Kyobe HB, Kariuki N, **Wayengera M**, Kirenga B, **Muttamba W**, Dawa J, Breiman RF, Osoro E, Ngere I, et al. Leveraging the structures of the COVID-19 pandemic response for successful control of Ebola in Uganda. Nat Med. 2023 Jun 23. doi: 10.1038/s41591-023-02395-4.
44. Lee PY, Salim H, Cheong AT, Sazlina SG, Harrun NH, Ho BK, Isa SM, **Hussein N**, Hanafi NS, et al. A self-management app to improve asthma control in adults with limited health literacy: A mixed-method feasibility study. 2023. (In preparation).
45. López-Star B. Review of ophthalmogenetics literature in Mexico. 2023. (In preparation).
46. Maceda EB, Asor BB, Cases RK, Abadingo M, **Tumulak M-AJR**, Anarna K, Supan RJ, Asuncion PC, Vesagas TD. Social media content analysis of public and private glucose-6-phosphate dehydrogenase deficiency (G6pD) Facebook Groups. J Community Genet. 2023. (Submitted).
47. Marwah V, Kumar N, Choudhary R, **Mutreja D**, Sharma A. A rare case of hemophagocytic lymphohistiocytosis associated with sarcoidosis. J Adv Lung Health. 2023. (Accepted).
48. McCarthy S, Nathan JJ, Khoo EM, Lim ZN, Hanafi NS, **Hussein N**. Health care professionals' challenges and solutions in providing palliative care to patients with severe COPD. 2023. (In preparation).
49. Mohamed SE, Khalil SH, Abdalrahim NE, **Alabid T**, Mergani MM, Babikir NE. Measurement of serum ferritin among Sudanese patients with ischemic stroke at Khartoum State, 2023 May 1. Int J Med Sci Clin Invention. 2023; 10(5):6697-6703.
50. **Monye HI**, Olawoye OO, Ugalahi M, Oluleye, Ashaye A. Burden and clinical profile of genetic eye diseases in children in Nigeria: A descriptive, cross-sectional study. Pan Afr Med J. 2023. (Accepted).
51. **Monye HI**, Olawoye OO, Ugalahi MO, Oluleye TS. Nigerian parents' perspectives on genetic testing in their children with genetic eye diseases. J Community Genet. 2023. (Accepted).
52. **Muttamba W**, O'Hare BA, Saxena V, Bbuye M, Tyagi P, Ramsay A, Kirenga B, Sabiiti W. A systematic review of strategies adopted to scale up COVID-19 testing in low-, middle- and high-income countries. BMJ Open. 2022; 12(11):e060838.
53. **Nair LS**, Dubey A, Mohan N, Vikraman SK, Desai J, Madadi M. Recurrent nonimmune fetal hydrops due to a novel pathogenic variant in PIEZO1 gene: A case report from South India. J Fetal Med. 2023; 10:69-72.
54. Nasser NA, Abd Allah MM, **Alabid** T, Babiker NE. Coagulation parameters pre and post vaccination with AstraZeneca among Sudanese population at Khartoum State, 2022. Int J Curr Res. 2023 Jan; 15(1):23479-23484.
55. Ng WL, Kee BP, **Hussein N**, Ng CJ, Mohd Zaidan FZ, Azmi SUF, Then S-M, Kwan Z, Qureshi N, et al. Prevalence of HLA-B*58:01 allele among patients with gout in Malaysia. Br J Clin Pharmacol. 2023. (Submitted).
56. Ng WL, Kee BP, **Hussein N**, Ng CJ, Mohd Zaidan FZ, Azmi SUF, Then S-M, Kwan Z, Qureshi N, et al. The views and opinions of implementing HLA-B*58:01 testing prior to allopurinol initiation in Malaysia: doctors' and patients' perspective. 2023. (In preparation).
57. Ohazurike EO, **Akinmola OO**, Olorunfemi G, Okunade KS. Association between beta-HCG and pre-eclampsia, a case-control study at the Lagos University Teaching Hospital. Hypertens Pregnancy. 2023. (Submitted).
58. Olayanju OA, Mba IN, **Akinmola OO**, Awah NE, Ofagbor E, Okonkwo O, Olasehinde OE, et al. Relationship between glycemic control and oral immunologic protein. West Afr J Med. 2022 Oct 20; 39(10):1062-1067.

59. Olayanju OA, Mba IN, **Akinmola OO,** Awah NE, Ofagbor E, Okonkwo O, Olasehinde OE, et al. Oral Innate immunity in patients with Type 2 diabetes mellitus in a tertiary hospital in Ibadan Nigeria: A cross sectional study. Pan Afr Med J. 2022 Nov 14; 43:134.
60. Onesmo A, Dismas M, Feredina J, Richard K, Elieza C, Bidiga S, **Mushi T**, Chami N, Kayange C, et al. A case of overt hypothyroidism: A forgotten cause of poor maternal-fetal outcome in low-resource setting. J Clin Images Med Case Rep. 2022 Nov 30; 3(11):2179.
61. **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. 2023. (In preparation).
62. **Otaify GA**, Elhossini RM, Abdel-Ghafar SF, **Sayed IM**, Abdel-Salam GMH, Aglan MS, Abdel-Hamid MS. CHST3-related skeletal dysplasia in 14 patients: Identification of 8 novel variants and further expansion of the phenotypic spectrum. Am J Med Genet A. 2023 Aug; 191(8):2100-2112.
63. Pirlog BO, Ilut S, **Pirlog R**, Chiroi P, Nutu A, Radutiu, DI, Cuc GD, Berindan-Neagoe, Nabavi SM, et al. New perspective on DNA response pathway (DDR) in glioblastoma, focus on classic biomarkers and emerging roles of ncRNAs. Expert Rev Mol Med. 2023 May 8; 25:e18.
64. **Pirlog R**, Chiroi P, Raduly L, Nutu A, Cismaru A, Berindan-Neagoe I. Chapter on 'Epithelial to mesenchymal transition in lung cancer'. Part of Interdisciplinary Cancer Research book series. Springer, Cham. 2023 Feb 10; pp 1-22. doi.org/10.1007/16833_2023_137.
65. Pozo-Palacios J, **Llamos-Paneque A**, Rivas Iglesias CR, Onofre Perez EJ. Experiences in the molecular diagnoses of Huntington's disease in Ecuador. 2023. (In preparation).
66. Quispe B, **Paredes-Moscosso SR**, Villegas-Llerena C. Retinitis pigmentosa: Gene therapy approaches. Revista Peruana de Biología. 2022. (Submitted).
67. Ramli R, Hanafi NS, **Hussein N**, Lee PY, Shariff Ghazali S, Cheong AT, Abu Bakar AI, Abdullah S, Abdul Samad A, et al. Barriers to and facilitators of asthma care for Malaysian hajj pilgrims: A qualitative study. Asia Pac J Public Health. 2023 Mar; 35(2-3):179-182.
68. Rusu I, **Pirlog R**, Chiroi P, Nutu A, Puia VR, Fetti AC, Rusu DR, Berindan-Neagoe I, Al Hajjar N. The implications of noncoding RNAs in the evolution and progression of nonalcoholic fatty liver disease (Nafld)-related hcc. Int J Mol Sci. 2022 Oct 15; 23(20):12370.
69. Sciascia S, Roccatello D, Salvatore M, Carta C, Cellai LL, Ferari G, …, **Wiafe SA**, Lumaka A, Groft S, et al. Unmet needs in countries participating in the Undiagnosed Diseases Network International: An international survey considering national health care and economic indicators. Front Public Health. 2023. (Submitted).
70. Shipra V, Baveja Puneet, Patnaik S, Thakur N, Tilak TVSVGK, **Mutreja D**. Young male with alleged snake bite and disseminated intravascular coagulation or a life threatening but potentially curable malignancy: Clinicopathologic correlation. Natl Med J India. 2022. (Accepted).
71. Sidharth KS, Rupesh R, Divya S, Sadaf A, **Khant AK**, Manoji M, Kalaivani G, Shraddha L, Rajiv S, et al. Assessment of South Asia Paediatric Acute kidney injury Epidemilogy and Risk Factors (ASPIRE): Prospective study on incidence and risk factors for Severe Pediatric AKI in SAARC nations with India, Myanmar, Bangladesh, Napal and Pakistan. 2023. (In preparation).
72. Singhal P, Venkatesan S, Chakraborthy B, **Mutreja D**, Sen A. Correlation between conventional karyotype, interphase FISH an immunophenotype in risk stratification of newly diagnosed plasma cell myeloma: An observational study. Medical Journal Armed Forces India (MJAFI). 2023. doi.org/10.1016/j.mjafi.2023.05.001.

73. Taft R, Thorpe E, Williams T, Shaw C, Chekalin E, Ortega J, Robinson K, Button J, …, **Wiafe SA**, et al. The impact of clinical genome sequencing in a global population of patients with suspected rare genetic disease. Nature. 2023. (Submitted).
74. Taft R, Thorpe E, Williams T, Shaw C, Chekalin E, Ortega J, …, **Milagros Duenas-Roque**, **Mario Cornejo Olivas**, **Wiafe SA**, et al. The impact of clinical genome sequencing in a global population of patients with suspected rare genetic disease. Res Sq. 2023 Aug 4. doi.org/10.21203/rs.3.rs-3117254/v1.
75. Tan HY, **Hussein N**, Lee YK, Malik TFA. Adolescents' experiences and views of the national school-based thalassaemia screening programme in Malaysia: A qualitative study. J Community Genet. 2023 Aug; 14(4):361-369.
76. Taruscio D, Salvatore M, Lumaka A, Carta C, Cellai LL, Ferrari G, Sciascia S, Groft S, …, **Wiafe SA**, et al. Undiagnosed diseases: Needs and opportunities in 20 countries participating in the Undiagnosed Diseases Network International. Front Public Health. 2023 Mar 2; 11:1079601.
77. Thao NP, **Long LH**, The HN, Hoang DN, Tran TN. ELN missense variant in patient with midaortic syndrome. BMC Cardiovasc Disord. 2022 Dec 3; 22(1):520.
78. Torrico-Lavayen R, Vargas-Alarcón G, Riojas-Rodriguez H, Sánchez-Guerra M, Texcalac-Sangrador JL, **Ortiz-Panozo E**, Gutiérrez-Avila I, De Vizcaya-Ruiz A, Cardenas A, et al. Long-term exposure to ambient fine particulate matter and carotid intima media thickness at bilateral, left and right in adults from Mexico City: Results from GEA study. Chemosphere. 2023 Sep; 335:139009.
79. Ubajaka CI, Onakoye A, Olawoye O, **Monye HI**, Adediran OA. Optical coherence tomography parameters of the optic nerve head and peripapillary nerve fibre layer in normal indigenous African eyes. 2023. (In preparation).
80. Ubom A, Adebayo O, Adeoye P, Kanmodi K, Salihu M, Umar SS, Tolani M., Ogunsuji O, **Monye HI**, et al. Health, well-being, and burnout amongst early career doctors in Nigeria. PLoS One. 2023 May 23; 18(5):e0285983.
81. Verma S, **Mutreja D**, Boruah D, Venkatesan S. Morphometric approach to angiogenesis in acute leukemia: An attempt to bring objectivity. Annals Pathol Lab Med. 2022 Oct 1; 9(9):A178-A182. doi:10.21276/apalm.3188.
82. Wu Y, Gettler K, **Kars ME**, Giri M, Li D, Bayrak CS, Zhang P, Jain A, Mafucci P, et al. Identifying high-impact variants and genes in exomes of Ashkenazi Jewish inflammatory bowel disease patients. Nat. Commun. 2023; 14(1):2256.
83. **Yadav S**, Potdar O. Fetal gall bladder duplication- A rare antenatal diagnosis. 2023. (In preparation).
84. **Yadav S**, Potdar O. Fetal rhabdomyoma- Implications in diagnosis and counselling. 2023. (In preparation).
85. **Yesilcinar İ,** Şahin E, Tutar SO. The decisional conflict and the genetic literacy of pregnant women regarding and prenatal screening tests. J Genet Couns. 2023. (Submitted).

2018 Batch

86. **Abdulkareem FB**. Recent advances in the molecular diagnosis of colorectal cancer makes personalised treatment possible even in resource-limited settings. Ann Trop Pathol. 2022; 13(2):61-68.
87. **Adewole OO**, Awopeju O, Komolafe A, Osigbeme A, Andero V, Erhabor G. Uptake and adherence to treatment for lung cancers in Nigeria: A single-centre experience. Niger J Health Sci 2022 Jul-Dec; 22(2):59-61.

88. **Adewole OO**, Omotoso BA, Ogunsina M, Aminu A, Odeyemi AO, Awopeju OF, Ayoola O, Adedeji T, Sogaolu OM, Adewole TO, Jiya E, Andero V, Obaseki D, Akintomide AO, Erhabor GE. Atorvastatin accelerates Mycobacterium tuberculosis clearance in pulmonary TB: A randomised phase IIA trial. Int J Tuberc Lung Dis. 2023 Mar 1; 27(3):226-228.
89. **Adeyemo TA**, Olatinwo AT, Akanmu AS. Effect of vitamin D levels on bone mineral density and its association with frequency of bone pains in sickle cell disease. 2023. (In preparation).
90. Afolabi BB, Babah OA, **Adeyemo TA**. Evidence-based obstetric management of women with sickle cell disease in low-income countries. Hematology Am Soc Hematol Educ Program. 2022 Dec 9; 2022(1):414-420.
91. Alfaro-Olivera M, Calle-Nuñez A, Uribe-León A, Araujo-Aliaga Il, Aguirre-Quispe W, Sarapura-Castro E, **Cornejo-Olivas M**. Friedreich's ataxia, review and literature update with a systematic search for cases in Latin America. Rev Neuropsiquiatr. 2023; 86(1):45-61.
92. Araujo-Aliaga I, Velásquez-Cruz F, Aguirre-Quispe W, López-Saavedra A, Sarapura-Castro E, **Cornejo-Olivas Mario**. Ataxia telangiectasia: A review from etiopathogenesis to current management with a description of reported cases in Peru. Rev Neuropsiquiatr. 2023 Apr; 86(1):18-29.
93. Asegaonkar P, Kotecha U, Dongre M, **Mistri M**, 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. 2023 Jan; 191(1):275-279.
94. Awolola NA, Akinjo D, Cookey KC, Haruna M, Arhere EP, Onyekwere CA, **Abdulkareem FB**. Concurrent Sarcina ventriculi infection in a patient with Helicobacter pylori-associated chronic gastritis: A case report. Nig. J. Gastroenterol. Hepatol. 2023 Jan 1; 15(1):20-22.
95. Awotoye W, Mossey PA, Hetmanski JB, Gowans LJJ, **Eshete MA**, Adeyemo WL, Alade A, Zeng E, Adamson O, et al: damaging mutations in afdn contribute to risk of nonsyndromic cleft lip with or without cleft palate. Cleft Palate Craniofac J. 2022 Nov 16:10556656221135926.
96. Babah OA, Akinajo OR, Beňová L, Hanson C, Abioye AI, Adaramoye VO, **Adeyemo TA**, Balogun MR, Banke-Thomas A, et al. Prevalence of and risk factors for iron deficiency among pregnant women with moderate or severe anaemia in Nigeria: a cross-sectional study. BMC Pregnancy Childbirth. 2023. (Submitted).
97. Balogun OS, Osinowo AO, **Abdulkareem FB**, Ajayi OO, Atoyebi OA, Da Rocha-Afodu JT. Clinicopathological features, risk profile assessment, and the surgical outcome of gastrointestinal stromal tumors in Lagos, Nigeria. Ann Afr Med. 2022 Oct; 21(4):432.
98. Bardaoui A, Hammami A, Elkarous R, Ali Aloui M, Oueslati R, **Messaoud O**, Santos DMF, Chtourou R. CVD graphene electrode for direct electrochemical detection of double-stranded DNA. Inorganics. 2023 Apr 20; 11(4):173.
99. Beriwal A, Chauhan L, Kumar A, **Dwivedi A**. A sib-pair with Al Kaissi syndrome caused by homozygosity for a novel CDK10 splice variant. Clin Dysmorphol. 2023 Jan 1; 32(1):32-35.
100. 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. (Submitted).
101. Chacaltana C, Ramirez-Pajares P; Manrique-Palomino A, Clause A, Chawla A, Thorpe E; Rivera-Valdivia A; Sarapura-Castro E; Bazalar-Montoya J; **Cornejo-Olivas M**. A novel variant in *SQSTM1* gene causing neurodegeneration with ataxia, dystonia, and gaze palsy in a Peruvian family. Mov Disord Clin Pract. 2023. (Submitted).
102. Chakraborty S, Mitra S, Ghosh A, Kumar K, Sanga S, **Dutta AK**, Mukherjee S, Biswas NK, Chakrabarti S, et al. A whole-exome analysis of non-syndromic hearing loss patients from India

reveals a wide spectrum of known and novel mutations. Res Sq. 2023 Feb 10. doi.org/10.21203/rs.3.rs-2549071/v1.
103. **Chowdhury EH**, **Roy SR**. The predilection to age, sex, laterally and treatment of retinoblastoma: a retrospective study at a tertiary eye care center in Bangladesh. Mathews J Pediatr. 2023 Jan 3; 8(1):28.
104. Conteh D, Amara R, **Edem-Hotah J**, Koroma M, Røssland A, Sekse RT, Giske T. Comparison of nursing students' experiences of learning nursing in Sierra Leone and Norway. 2023. (In preparation).
105. **Cornejo-Olivas M**, Solis-Ponce L, Araujo-Aliaga I, Milla-Neyra K, Ortega O, Illanes-Manrique M, Mazzetti P, Manrique-Enciso C, Cubas-Montecino D, et al. Machado Joseph-Disease is rare in the Peruvian population. Cerebellum. 2022 Nov 3. doi: 10.1007/s12311-022-01491-4.
106. **Cornejo-Olivas MR, Dueñas-Roque MM**, **Messaoud O**, **Kumar-Dutta A**, **Dwivedi A, Mistri M**, Nirmala S, Gyawali P, Sok-Kun T, et al. Global perspectives on becoming established as a next generation practitioner of genomic medicine. G2MC. 2023. (In preparation).
107. 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. Med J Armed Forces India. 2022. (Submitted).
108. **Das S**, Thomas M, Yoganathan S, Muthusamy K, Barney AM, **Dutta AK**, Rekha A, Mohan S, et al. Exploration of clinical and genetic findings in Ataxia-Telangiectasia (AT) patients from the Indian subcontinent. Eur J Med Genet. 2023 Jun; 66(6):104766.
109. Dutt R, Mishra NR, Singh R, Patel SK, Dem Dukpa R, Soniya C, **Dutta AK**. Feasibility of including hands-on microteaching in the medical faculty development program in India: A mixed-methods study. Cureus. 2023 Jun15; 15(6):e40470.
110. **Dutta AK**, Goswami K. Assessing cheating in online assessments during COVID-19. Med Educ. 2023 Aug; 57(8):770.
111. **Dutta AK**, Goswami K. Developing a blueprint for theoretical assessment of Biochemistry of phase I MBBS students. Med Sci Edu. 2023 Feb 16; 33(2):339-342.
112. Elebute OA, Komolafe MD, Akinjo AO, Alakaloko FM, Seyi-Olajide JO, Ladipo-Ajayi O, **Abdulkareem FB**, Ademuyiwa AO, Bode CO. Preputial cyst in the newborn: A case report from Africa. Afr J Paediatr Surg. 2023 Jan-Mar; 20(1):80-82.
113. **Gitaka J**, Mbugua S, Mwaura P, Gatungu D, Githanga D, Ndwiga C, Abuya T, K'Oduol K, Liambila W, et al. Devolved health system capacity in the provision of care for sick newborns and young infants in four counties serving vulnerable populations in Kenya. PLOS Glob Public Health. 2022 Oct 20; 2(10):e0000183.
114. Idowu EA, Ibiyemi O, **Taiwo OO**, Adedapo OA. Presentation and management of dental fluorosis in a resource limited setting in north-central Nigeria. Niger Dent J. 2022 Jul-Dec; 30(2): 31-45.
115. Khalid A, Afzal S, Abbas F, Afzal I, **Iqbal M**, Ahmad S, Ansari SA, Alkahtani HM, Chohan TA, et al. Phytochemical, biological, and in-silico analysis of Colute a armata Hemsl. & Lace: A possible source of bioactive natural compounds. S. Afr. J. Bot. 2023 Jul 23; 158:133-141.
116. Kiplimo J, **Mburu S**, Muriuki J. A metric scale-based assessment framework for risks categorization and associated harm for LMS in public universities of Kenya. 2023. (In preparation).

117. Kumar V, Kumar P, Chauhan L, **Dwivedi A**, Ramamurthy HR. Novel combination of *FLNC* (c.5707G>A; p. Glu1903Lys) and *BAG3* (c.610G>A; p.Gly204Arg) genetic variant expressing restrictive cardiomyopathy phenotype in an adolescent girl. J Genet. 2022 Nov 11; 101:54.
118. Leal TP, French-Kwawu JN, Gouveia MH, Borda V, Inca-Martinez M, Mason EA, Horimoto AR, Loesch DP, …, **Cornejo-Olivas MR**, et al. X-Chromosome association study in Latin American cohorts identifies new loci in Parkinson disease. medRxiv [Preprint]. 2023 Feb 2. doi.org/10.1101/2023.01.31.23285199.
119. **Lertwilaiwittaya P**, Tantai N, Maneeon S, Kongbunrak S, Nonpanya N, Hurst A, Srinonprasert V, Pithukpakorn M. BRCA1 and BRCA2 testing is cost-effective in high-risk breast cancer patients and family members in Thailand: An implication for public health policy in resource-limited countries. Front Public Health. 2023. (Submitted).
120. **Lertwilaiwittaya P**, Weber FH Jr. Tranexamic acid as a successful therapy in Turner Syndrome with recurrent overt gastrointestinal bleeding due to small intestinal venous anomalies. ACG Case Rep J. 2023 Jan 20; 10(1):e00961.
121. Li M, Olotu J, Buxo-Martinez CJ, Mossey PA, Anand D, Busch T, Alade A, Gowans LJJ, **Eshete M**, et al: Variant analyses of candidate genes in orofacial clefts in multi-ethnic populations. Oral Dis. 2022 Oct; 28(7):1921-1935.
122. Liambila W, Were F, Abuya T, Odwe G, Natecho A, Mungai S, Mwaura P, Githanga D, …, **Gitaka J**. Institutionalizing the management of sick young infants: Kenya's experience in revising national guidelines on integrated management of newborn and childhood illnesses. Glob Health Sci Prac. 2023 Apr 28; 11(2):e2200482.
123. Mata I, Salles P, **Cornejo-Olivas M**, Saffie P, Ross OA, Reed X, Bandres-Ciga S. LRRK2: Genetic mechanisms vs genetic subtypes. Handb Clin Neurol. 2023; 193:133-154.
124. Mbugua S, Mwaura P, Oyiengo L, Liambila W, Emuria A, Gatungu D, **Gitaka J**. Lessons learnt from community referral and follow up of sick young infants with possible severe bacterial infection in Turkana County, Kenya. Global Pediatrics. 2023 Jun; 4:100050.
125. **Mburu S**, Gitonga H, Ndung'u R. Making integrative, predictive data modeling approaches more actionable and localized for optimizing breast cancer treatment, management, and actualizing precision medicine for resource-limited settings. 2023. (In preparation).
126. **Mburu S**, Gitonga H, Ndung'u R. The efficacy and safety of adjuvant capecitabine treatment in TNBC patients with residual invasive disease: A meta-analysis. 2023. (In preparation).
127. **Mburu S**, Kimani K. Application of simple clustering data modeling for determining most cost-effective and rationalizing diagnostic testing in breast cancer management: a model for resource-limited settings (data mining, model building, testing, validation). 2023. (In preparation).
128. **Mburu** S. Digital e-Learning in the context of Covid-19 pandemic crisis: Leveraging on learning analytics. 2023. (In preparation).
129. McKnight D, Morales A, Hatchell KE, Bristow SL, Bonkowsky JL, Perry MS, Berg AT, ELEVIATE Consortium…**Dueñas-Roque MM**, et al. Genetic testing to inform epilepsy treatment management from an international study of clinical practice. JAMA Neurol. 2022 Dec 1; 79(12):1267-1276.
130. Miranda M, Díaz M, Hughes R, Barreto M, **Nakousi-Capurro N**, Campero M. CANVAS: A new etiology of adult taxia. The association with cough guides the diagnosis. Communication of 2 patients. Rev Med Chil. 2023. (Accepted).
131. **Nakousi-Capurro N**. Tracking uncertainty in germline genetic testing: Sources, attributes, and resolution of variants of uncertain significance in 1.6 million individuals. 2023. (In preparation).

132. Ndububa DA, **Abdulkareem FB**, Mustapha SK, Ekwunife CN, Omuemu CE. Soghin clinical practice guidelines for hepatocellular carcinoma. Nig. J. Gastroenterol. Hepatol. 2023 Jan 1; 15(1):1-5.
133. Nguyen Q, Woof W, Kabiri N, Sen S, **Varela MD**, Cabral De Guimaraes TA, Shah M, Sumodhee D, Moghul I, et al; Eye2Gene Patient Advisory Group. Can artificial intelligence accelerate the diagnosis of inherited retinal diseases? Protocol for a data-only retrospective cohort study (Eye2Gene). BMJ Open. 2023 Mar 20; 13(3):e071043.
134. Nwabudike VI, **Oluwarotimi AC**, Okoturo E, Ijogun A. Comparative study of the effect of single and multiple suture techniques on inflammatory complications following mandibular third molar surgery: a randomized prospective study at the Lagos state University Teaching Hospital (Lasuth), Ikeja. West Afr J Med. 2022. (Submitted).
135. Ojewunmi, OO, **Adeyemo TA**, Oyetunji AI, Inyang B, Akinrodoye A, Mkumbe BS, & Menzel, S. The genetic dissection of fetal haemoglobin persistence in sickle cell disease in Nigeria. Hum Mol Genet. 2023. (Submitted).
136. **Oluwarotimi AC**, Abah AA, Ogundana OM. Prevalence of Medical conditions among dental patients in a South-West hospital in Nigeria. African Journal of Oral Health (AJOH). 2022 Nov 30; 11(1):1-12.
137. Oyebade A, Adeola O, Akinmurele T, **Taiwo O**, Oginni A, Adekunle I, Gbadamosi M. Patients' perception of quality of Tuberculosis healthcare service in Southwest Nigeria. Int J Community Med Public Health. 2023 Apr 29; 10(4):1375-1382.
138. Painous C, Martí MJ, Graessner H, Camargo AP, El-Jaafary SI, Martínez-Ramírez D, Ojo OO, Taiwo FT, …**Cornejo-Olivas M**, et al. Management of rare movement diseases in different world regions. Parkinsonism Relat Disord. 2023 Mar; 108:105286.
139. Pranav Chand R, Vinit W, Vaidya V, Iyer AS, Shelke M, Aggarwal S, Magar S, Danda S, …, **Mistri M**, et al. Proband only exome sequencing in 403 Indian children with neurodevelopmental disorders: Diagnostic yield, utility and challenges in a resource-limited setting. Eur J Med Genet. 2023 May; 66(5):104730.
140. Quiat D, Timberlake AT, Curran JJ, Cunningham ML, McDonough B, Artunduaga MA, DePalma SR, **Duenas-Roque MM**, Gorham JM, et al. Damaging variants in FOXI3 cause microtia and craniofacial microsomia. Genet Med. 2023 Jan 30; 25(1):143-150.
141. Raha S, Kotecha U, **Mistri M**, Shah P, Sharda S. Familial infantile spasm syndrome due to biallelic variants in the gene encoding UFM1-specific peptidase 2 (UFSP2). Medical Reports. 2023. (Submitted).
142. Rauf B, Khan SY, Jiao X, Irum B, Ashfaq R, Zehra M, Khan AA, **Naeem MA**, Shahzad M, Riazuddin S, Hejtmancik JF, Riazuddin SA. Next-generation whole exome sequencing to delineate the genetic basis of primary congenital glaucoma. Sci Rep. 2022 Oct 14; 12(1):17218.
143. Rodriguez-Mateos P, Ngamsom B, Ameyo D, Wakaba P, Shiluli C, Iles A, **Gitaka J**, Pamme N. Integrated microscale immiscible phase extraction and isothermal amplification for colorimetric detection of Neisseria gonorrhoeae. Anal Bioanal Chem. 2023 Sep; 415(21):5129-5137.
144. Rossi M, Hamed M, Rodríguez-Antigüedad J, **Cornejo-Olivas M**, Breza M, Lohmann K, Klein C, Rajalingam R, Marras C, et al. Genotype-phenotype correlations for ATX-TBP (SCA17): MDSGene systematic review. Mov Disord. 2023 Mar; 38(3):368-377.
145. Rutayisire E, Mochama M, Ntihabose CK, **Utumatwishima JN**, Habtu M. Maternal, obstetric and gynecological factors associated with preterm birth in Rwanda: Findings from a national longitudinal study. BMC Pregnancy Childbirth. 2023; 23(1):365.

146. Saengphatrachai W, **Lertwilaiwittaya P**, Limwongse C, Rattanabannakit C, Pho-Iam T, Senanarong V. Case series of C9orf72 hexanucleotide repeat expansion among dementia patients in the Memory Clinics at Siriraj Hospital. BMC Geriatr. 2023. (Submitted).
147. Sagoe KO, Kyama MC, Maina N, Kamita M, Njokah M, Thiong'o K, Kanoi BN, Wandera EA, …, **Gitaka J**. Application of hybridization chain reaction/CRISPR-Cas12a for the detection of SARS-Cov-2 infection. Diagnostics (Basel). 2023 May 7; 13(9):1644.
148. Saha T, Sarma N, Dawn S, Dhar A, Chatterjee N, Hadem IKH, **Dutta AK**, et al. Serum triacylglycerol: A putative early biomarker of disease severity of Type 2 diabetes mellitus compared to microalbuminuria: TAG: Type 2 DM severity biomarker. Indian J Exp Biol. 2023 Jun 28; 61(7):499-504.
149. Sahoo S, Goswami K, Sarkar J, **Dutta AK**, Pal A, Tallapaneni S, Biswas S, Murugaiyan S. Significance of serial formative assessment on the teaching-learning process. Res Sq. 2023 Jan 3. doi.org/10.21203/rs.3.rs-2396435/v1.
150. Sahu PK, Sahoo S, Chatterjee N, Majhi J, **Dutta AK,** Goswami K, Pal A. Comparative evaluation of serum lithium estimation using plain glass vial and serum clot activator vacutainer by reflectance photometry. J Lab Physicians. 2023 Jun 13. doi.org/10.1055/s-0043-1769762.
151. Schlottmann PG, Luna JD, Labat N, Yadarola MB, Bainttein S, Esposito E, Ibañez A, Barbaro EI, …, **Varela MD**. Nationwide genetic analysis of more than 600 families with inherited eye diseases in Argentina. NPJ Genom Med. 2023 May 22; 8(1):8.
152. Shah MZ, Mustafa G, **Iqbal M**, Qasim M, Abbas K, Umair U, Baig HMA. Prevalence of gram-positive bacteria in the affected individuals of otitis media with effusion from the indigenous population of southern Punjab, Pakistan: first report. Braz. J. Biol. 2024; 84:e267874.
153. Sinha S, **Dutta AK**, Bhattacharya P, Bhattacharya S, Das MK. Spectrum of Rare and Novel Indel Mutations Responsible for β Thalassemia in Eastern India. Ind J Clin Biochem. 2023 Jan 3. doi.org/10.1007/s12291-022-01098-w.
154. Tan A, **Cornejo-Olivas M**, Okubadejo N, Kumar Pal P, Saranza G, Saffie-Awad P, Azlina Ahmad-Annuar A, Schumacher-Schuh A, Okeng'o K et al. Genetic testing for Parkinson's disease and movement disorders in less privileged areas: Barriers and opportunities. Mov Disord Clin Pract. 2023. (Submitted).
155. Thorpe E, **Cornejo-Olivas M**, Bazalar-Montoya J, **Duenas-Roque M**, et al. Compassive WGS genetic testing in the Peruvian population, iHope program experience. 2023. (In preparation).
156. Thorpe E, Taft R, **Cornejo-Olivas MR, Dueñas-Roque MM**, De La Torre CA, Purizaca N, Bazalar J, Milla K, Galarreta C, et al. Clinical whole genome sequencing as a first-tier test at three resource-limited genetics clinic in Peru. 2023. (In preparation).
157. Thorpe E, Williams T, Shaw C, Chekalin E, Ortega J, Robinson K, Button J, …, **Dueñas-Roque MM**, **Cornejo-Olivas MR**, et al. The impact of clinical genome sequencing in a global population of patients with suspected rare genetic disease. Nat. Med. 2023. (Submitted).
158. **Varela MD**, Bellingham J, Motta F, Jurkute N, Ellingford JM, Quinodoz M, Oprych K, Niblock M, Janeschitz-Kriegl L, et al. Multidisciplinary team directed analysis of whole genome sequencing reveals pathogenic non-coding variants in molecularly undiagnosed inherited retinal dystrophies. Hum Mol Genet. 2023 Jan 27; 32(4):595-607.
159. **Varela MD**, Duignan ES, De Silva SR, Ba-Abbad R, Fujinami-Yokokawa Y, Leo S, Fujinami K, Mahroo OA, Robson AG, et al. CERKL- Associated retinal dystrophy: Genetics, phenotype and natural history. Ophthalmol Retina. 2023 Oct; 7(10):918-931.

160. **Varela MD**, Georgiou M, Alswaiti Y, Kabbani J, Fujinami K, Fujinami-Yokokawa Y, Khoda S, Mahroo OA, Robson AG, et al. CRB1-Associated retinal dystrophies: Genetics, clinical characteristics, and natural history. Am J Ophthalmol. 2023 Feb; 246:107-121.
161. **Varela MD**, Laich Y, Hashem SA, Mahroo OA, Webster AR, Michaelides M. Prognostication in Stargardt disease using fundus autofluorescence: Improving patient care. Ophthalmology. 2023 Jun 17; S0161-6420(23)00428-1.
162. **Varela MD**, Sen S, De Guimaraes TAC, Kabiri N, Pontikos N, Balaskas K, Michaelides M. Artificial intelligence in retinal disease: clinical application, challenges, and future directions. Graefes Arch Clin Exp Ophthalmol. 2023 May 9:1-15.
163. **Varela MD**, Wong SW, Kiray G, Schlottmann PG, Arno G, Shams ANA, Mahroo OA, Webster AR, AlTalbishi A, et al. Detailed clinical, ophthalmic, and genetic characterization of ADGRV1-associated Usher syndrome. Am J Ophthalmol. 2023 Jul 7; 256:186-195.
164. Veturi YA, Woof W, Lazebnik T, Moghul I, Woodward-Court P, Wagner SK, Cabral de Guimarães TA, **Varela MD**, Liefers B, et al. SynthEye: Investigating the impact of synthetic data on artificial intelligence-assisted gene diagnosis of inherited retinal disease. Ophthalmol Sci. 2022 Nov 22; 3(2):100258.
165. Vishnevetsky A, **Cornejo-Olivas M**, Sarapura-Castro E, Inca-Martinez M, Rabinowitz D, Milla-Neyra K, Mazzetti P, Bird T. Juvenile-Onset Huntington's disease in Peru: A case series of 32 patients. Mov Disord Clin Pract. 2022 Dec 2; 10(2):238-247.
166. Vollstedt EJ, Schaake S, Lohmann K, Padmanabhan S, Brice A, Lesage S, Tesson C, Vidailhet M, …**Cornejo-Olivas M**, et al. Embracing monogenic Parkinson's disease: The MJFF global genetic pd cohort. Mov Disord. 2023 Feb; 38(2):286-303.
167. **Wangi KYW**, Berish D. Nursing care for patients with thalassemia-dependent-transfusion during the COVID-19 pandemic in Indonesia: from public health ethics to community nursing. J Radiol Nurse. 2023. (Submitted).
168. **Wangi KYW**, Birriel B, Smith C. Addressing the reproduction rights in the woman with thalassemia: a discursive paper. 2023. (In preparation).
169. **Wangi KYW**, Birriel B, Smith C. Psychosocial burden in transfusion-dependent beta-thalassemia patients and its impact on the quality of life and the problem of dignity. J Taibah Univ Med Sci. 2023 May 10; 18(6):1217-1219.
170. Waweru H, Kanoi BN, Kuja J, Maranga M, Kongere J, Maina M, Kinyua J, **Gitaka J**. Limited genetic variations of the Rh5-CyRPA-Ripr invasion complex in Plasmodium falciparum parasite population in selected malaria-endemic regions, Kenya. Front. Trop Dis. 2023 Mar 1; 4:1102265.

B. Grants: (Total- 37; 2019- 13; 2018- 24)

2019 Batch

1. **Bocoum A**. Visiting Training Grant. 2023. (Movement Disorders Society, Received).
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; Continued).
3. Covaliu B (PI), Neagoe I, **Pirlog R**. Healthy start-ups- SUS. 2022. (Ministry of Investments and European Projects; Continued).

4. **Gyawali P** (PI), Risal P. Capacity building training program on implementation of point of care testing. 2023. (Higher Education Institution Capacity Development Program- University Grant Commission; Received).
5. Khoo EM (PI), Hanafi NS, **Hussein N** (Co-I), Pang YK, Ahmad A, Ramdzan SNK, Wong CK, Wee C, Ramli R. NIHR Global Health Research Unit on Respiratory Health (RESPIRE-2). 2022. (NIHR, United Kingdom Funding; Received).
6. **Luong LH** (Co-I), 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; Accepted).
7. Nair A (PI), **Nair LS** (Co-PI), Nair MKC (Co-PI). Development of cost-effective ways in the molecular diagnosis of intellectual disability/ developmental delay in the population in Southern Kerala. 2023. (ICMR; Submitted).
8. 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; Continued).
9. **Okunola OO** (PI). Heritability and phenotypic characteristics in diabetic nephropathy. 2023. (International Society of Nephrology; Re-submitted).
10. Olawoye O, (PI), Fingert J, **Monye HI** (Co-I), Ashaye A, Hauser M. Genetic characterization of Juvenile Open Angle Glaucoma (JOAG) patients in Sub-Saharan Africa. 2023. (Shaffer grant-Glaucoma Research Foundation; Submitted).
11. Pinnock H (PI), Khoo EM, Hanafi NS, **Hussein N** (Co-PI), Cheong AT, Ghazali SS, Ramdzan SNR, Ramli R, Ng WL, et al. **R**each, **E**mpower, **A**ct: **C**ommunities for **H**ealth (REACH). 2023. (NIH-Global Alliance for Chronic Diseases; Submitted).
12. **Tumulak MJR** (PI), Maceda EB, Cases RK, Asor BB, Calalo-Magbanua C, **Abad PJ**. A content analysis of Facebook groups on congenital adrenal hyperplasia. 2023. (Newborn Screening Reference Center; Received).
13. Villegas-Llerena C, **Paredes-Moscosso SR** (Co-I), Yucra A, Cruz LA, Gutierrez G, Corredor F, Facundes FG. Transcriptomic analysis of fur to identify markers of genetic improvement in alpaca fiber. 2023-2025. (Concytec, World Bank, Universidad de San Martín de Porres; Received).

2018 Batch

14. Afolabi BB (CI), **Adeyemo TA** (Co-I), Abioye (Co-I), Oluwole EO (Co-I). Azithromycin in labour versus placebo for preventing clinical infections in Nigerian women having a vaginal birth (AZIN-V): A double blind, placebo-controlled, randomized trial alongside an implementation study. 2023-2026. (Bill and Melinda Gates Foundation; Submitted).
15. Alatise O (PI), Kingham P (Co-PI), **Abdulkareem FB** (Co-I). Nigerian Cancer Research Training Program. 2021. (OAK Foundation, Switzerland; Continued).
16. Basu A (PI), Sarkar S (Co- PI), **Dutta AK** (Co- PI). Pathways to Resilience And Mental Illness (PARAM). 2023. (Indian Council of Medical Research; Accepted).
17. Chinnaswamy S (PI), **Dutta AK** (Co-PI). Alterations in peripheral immune cell molecular phenotypes arise due to differences in socioeconomic status and environment. 2023. (Dept. of Science and Technology, Government of India - DST Supra Grant; Submitted).
18. **Cornejo-Olivas M** (PI), Araujo-Aliaga I, Milla-Neyra K, Sarapura-Castro E. Characterizing SCA10 in the Peruvian population, from genetics to deep phenotype profile. 2023. (National Ataxia Foundation Research Grants; In Preparation).

19. **Cornejo-Olivas M** (PI), Illanes-Manrique M, Castro-Suarez S, Sarapura-Castro E (PI). Capacity Building in International Dementia Research (CBIDR). 2023. (Alzheimer Association /Global Brain Health Institute; In Preparation).
20. Das S (PI), Basu A (Co-I), Sarkar S (Co-I), **Dutta AK** (Co-I), Sukumaran D (Co-I), Bagepalli BS (Co-I), Saha I (Co-I), Maurya PK (Co-I), et al. Clinical utility and cost-effectiveness of pharmacogenomics-assisted treatment versus standard of care in patients with schizophrenia attending a tertiary care hospital in eastern India. 2023. (Indian Council of Medical Research; Accepted).
21. Giske T, **Edem-Hotah J**, Amara R, Conteh D Koroma MJ, Hunskar I, Røssland A, Sekse RT. 'Fundamentals of Nursing', a textbook in a context relevant for Sierra Leone. 2023-2026. (EU ERASMUS+ Global Mobility, Approved).
22. **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. 2023. (NIH; Received).
23. **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).
24. Kingham P (PI), Alatise O (Co-PI), **Abdulkareem FB** (Site-PI). Expanding cancer research capacity in Nigeria with team science. 2022. (NIH-D43; Continued).
25. Kingham P (PI), Alatise O (Co-PI), Du (Co-PI), **Abdulkareem FB** (Co-I). Determining the risk factor profile and biology of colorectal cancer in Nigeria. 2020. (NIH-R01; Continued).
26. Kirenga B (PI), Byakiika P, Kiwanuka N, Atwine D, **Wayengera M**, **Muttamba W**, et al. A phase I/II randomized placebo-controlled trial to evaluate the safety and immunogenicity of Sudan ebolavirus vaccines in Uganda. 2022. (World Health Organization, Received).
27. 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).
28. **Mburu S** (PI), Gitonga H, Ndung'u R. Translational TMEs pathophysiological status in breast cancer? Implications as treatment effect modifier in TNBC. 2023. (NRF-Kenya; Submitted).
29. **Mburu S** (PI), Mutiso A, Muchangi D, Kiiru G, Kabata D. 2023. Bio innovation research project to recover health beneficial polyphenols from black tea (Camelia sinesis) leaves biowaste. 2023. (FID-Exchange; Submitted).
30. **Messaoud O** (Co-I), Haddad S (PI), Charfi-Kaddour S, Gargouri M, Drissi LB (PI), Hammami A, Bardaoui A. The use of advanced materials for molecular biology applications. 2020-2023. (The Tunisian Ministry of Higher Education and Scientific Research and the Moroccan Ministry of National Education, Vocational Training, Higher Education and Scientific Research Fund; Continued).
31. **Messaoud O** (PI), Elgaher W (Co-PI), Hammami A, Karyaoui M (Tunisian partners). Diamond synthesis using chemical vapor deposition process and assessment of biological properties of chemical molecules. 2023. (AGYA/German Federal Ministry of Education and Research Fund; Accepted).
32. **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. 2023. (AGYA/German Federal Ministry of Education and Research Fund; Continued).
33. 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).

34. Taramasco C, **Nakousi-Capurro N**, Martinez F, Ruete D. Diagnostic support platform for the identification of genetic- based pediatric neurological diseases through a recommender system based on machine learning: A before and after study. 2023- 2025. (Chilean National Agency for Research and Development).
35. Tosto G (PI), Pericak-Vance M, Rajabli F, Custodio N, **Cornejo-Olivas M** (Co-I), Sarapura-Castro E, Montesinos R, Illanes-Manrique M, Milla-Neyra K, et al. Global Latinos sequencing study for Alzheimer's disease. 2023. (NIA/NIH; Submitted).
36. **Wangi KYW** (PI), Birriel B. Predoctoral to postdoctoral transition award for a diverse genomics workforce. 2023. (NHGRI/NIH F99/K00; In preparation).
37. **Wangi KYW** (PI). Human dignity in thalassemia-dependent transfusion in Indonesia. 2024. (The American-Indonesian Cultural and Education Foundation & Penn State University; In preparation).

C. Collaborations (NIH and Other Institutions):

NIH: (Total- 11; 2019- 4; 2018- 7)

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; Amended protocol for IRB).
2. **Torres-Mejía G** (Site PI), Angeles-Llerenas A, Gomez-Flores-Ramos L, Sánchez- Zamorano LM, Flores-Luna L, Romieu I, **Ortiz-Panozo E** (Co-I), 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).
3. **Torres-Mejía G** (Site PI), Angeles-Llerenas A, Gomez-Flores-Ramos L, Rodríguez-Valentín R, Flores-Luna L, Sánchez- Zamorano LM, Fierros-Zarate G del S, **Ortiz-Panozo E** (Co-I), Fejerman L. LAGENO consortium. (LAGENO-BC & CONFLUENCE, Multicenter 2021, Instituto Nacional de Salud Pública, Mexico; NCI/NIH).
4. **Yesilcinar İ**, **Seven M**, Şahin E, Calzone K. Test/Retest reliability of a Turkish version of the genetics and genomics in nursing practice survey. (Izmir Katip Celebi University, Health Science Faculty Obstetrics and Gynecology Nursing, Turkey; University of Massachusetts Amherst College of Nursing, USA; NCI/NIH).

2018 Batch

5. Ginsburg G, Patrinos GP, Lopez-Correa C, **Cornejo-Olivas M**, Teri Manolio,…Global Genomic Medicine Consortium. Global leaders dedicated to advancing genomic medicine implementation in clinical care. (Instituto Nacional de Ciencias Neurologicas, Perú; Global Alliance for Genomics and Health; The Golden Helix Foundation; Australian Genomics Health Alliance; NHGRI/NIH).
6. **Gitaka J**, Niraj T, Kanoi B, Makokha F. Discovery of novel malaria vaccine targets levering on T cell receptor sequencing. (Directorate of Research and Innovation, Mount Kenya University, Kenya; NIAID/NIH).

7. 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, Perú; Undiagnosed Diseases Network International; NCATS/NIH).
8. Haines J, Griswold A, DeStefano A, Schellenberg G, Beecham G, Blue L, Peloso G, Vardarajan B,…, **Cornejo-Olivas M**, et al. PeADI study- Alzheimer´s Disease Sequencing Project (ADSP). (Instituto Nacional de Ciencias Neurologicas, Perú; University of Miami, FL; NIA/NIH).
9. 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).
10. 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).
11. Singleton A, Blauwendraat C, Brice A, Casey B, Fiske B, Bandres S, Mata IF, **Cornejo-Olivas M**, Rivera-Valdivia A, et al. GP2 Global Parkinson genetics program aiming to further understand the genetic architecture of Parkinson's disease through genotyping diverse patient groups. (Instituto Nacional de Ciencias Neurologicas, Perú; Cleveland Clinic, Ohio, USA; NIA/NIH).

Other Institutions: (Total- 54; 2019- 34; 2018- 20)

2019 Batch

1. **Abubakar S**. Trained staff of the Kano State Contributory Healthcare Management Agency on Healthcare Financing. (2023). (Bayero University Kano/Aminu Kano Teaching Hospital; Kano State Ministry of Health; Kano State Contributory Healthcare Management Agency, Nigeria).
2. **Chowdhury EH, Amarakoon GGGT, Mosema KBA, Iqbal M**. Study of mental health related to hemophilia in Bhutan, Sri Lanka, Democratic Republic of Congo and Pakistan. (Samtse General Hospital, Bhutan; Faculty of Medicine and Allied Sciences, Rajarata University of Sri Lanka, Sri Lanka; Institut National de Recherche Biomedicale, Biamba Marie Mutombo Hospital, Kinshasa, Democratic Republic of Congo; The Islamia University of Bahawalpur, Pakistan).
3. Cohen P, Itan Y, Plucinska K, **Kars ME**. Phenome-wide association studies to investigate the pleiotropic effects of variations in secreted proteome (Icahn School of Medicine at Mount Sinai, USA; The Rockefeller University, USA).
4. Cunnigham-Rundles C, Itan Y, Boisson B, **Kars ME**. Network-based heterogeneity clustering analysis of common variable immunodeficiency patients (The Charles Bronfman Institute for Personalized Medicine, Icahn School of Medicine at Mount Sinai; Division of Clinical Immunology, Icahn School of Medicine at Mount Sinai, USA).
5. Dunaif A, Itan Y, **Kars ME**, Cai J, Brewer K. Investigating the phenome-wide associations of genes implicated in polycystic ovarian syndrome (The Charles Bronfman Institute for Personalized Medicine, Icahn School of Medicine at Mount Sinai, USA; Division of Endocrinology, Diabetes and Bone Disease, Icahn School of Medicine at Mount Sinai, USA).

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, Perú; LAGENO-BC).
7. Gaillard E, Devani P, Pinnock H, Khoo EM, **Hussein N**, Daines L, Fowler S. What is the diagnostic value of repeated peak expiratory flow rate measurements in the diagnosis of asthma? (Universiti Malaya, Malaysia; University of Leicester, UK; University of Edinburgh, UK; University of Manchester, UK).
8. Gerzsten R, Itan Y, **Kars ME**. Phenome-wide association studies to investigate the genetic basis of proteome alterations in cardiometabolic traits. (Icahn School of Medicine at Mount Sinai, USA; Beth Israel Deaconess Medical Center, USA).
9. **Hussein N** (PI), Khoo EM, Lin C, Cheah W, Fong CS, Latif MT, Ooi MCG, Ho SB, Jenny S, Jackson K. Developing and evaluating a mobile phone-based early alert system using high resolution air quality forecast to improve asthma control in Malaysia. (Faculty of Medicine, Universiti Malaya; Institute of Ocean and Earth Sciences, Universiti Malaya; Dept. of Earth Sciences and Environment, Universiti Kebangsaan; Dept. of Environment, Ministry of Environment and Water; Malaysian Meteorological Dept.; Multimedia University, Malaysia; Cambridge Environmental Research Consultants, UK).
10. **Iqbal M**, Ansar M. Mutational analysis of DNA from consanguineous Pakistani families having ophthalmological disorders through whole exome sequencing (IUB, Pakistan; Jules-Gonin Eye Hospital, University of Lausanne, Switzerland).
11. **Iqbal M**, Iqbal Z. Mutational analysis of Pakistani families affected with Parkinson's disease through whole exome sequencing (Biotechnology-IUB, Pakistan; Oslo University Hospital, Norway).
12. **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, Pakistan).
13. **Iqbal M**, Serge N. Genetics studies of male infertility. (IBBB, IUB, Pakistan; University of Geneva, Switzerland).
14. **Iqbal M**, Tekin M. Whole exome or genome sequencing on 1000 families from all over the globe. (Biotechnology IUB, Pakistan; Miller School of Medicine, University of Miami, US).
15. Kantaputra PN, **Guven Y**. Consultation on genetic cases. (Chiang Mai University Faculty of Dentistry, Thailand; Istanbul University Faculty of Dentistry, Turkey).
16. Kristine M, Van Genderen, Collen F, **Mushi TL**. Pediatric Resident Global Health Education. (Northwestern University Feinberg School of Medicine, USA; Catholic University of Health and Allied Science, Tanzania)
17. **Llamos-Paneque A,** Anne Slavotinek. Genetic analysis of patients with eye anomalies and rare diseases (Genetics Service of Hospital No.1 FF. AA. Quito, Ecuador; Cincinnati Children's Hospital Medical Center, Ohio, USA).
18. **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).
19. **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).

20. **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).
21. **Llamos-Paneque A**, Randi Hagerman, Villarreal Jennifer, Flora Tassone. Fragile X Research and Treatment Program. (Genetics Service of Hospital No.1 FF. AA. Quito, Ecuador; MIND Institute, UC Davis, School of Medicine, CA, USA).
22. **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).
23. **Luong LH**, Hoang LT, Prof. Timothy J. Craig: Diagnosis, consultation and creating registry of hereditary angioedema patients in Vietnam. (Pennsylvania State University, USA; National E Hospital, Vietnam).
24. **Luong LH**, Olivieri L. Consultation on cardiovascular malformations and rare vascular disorders. (National E Hospital, Vietnam; Children's National Hospital, USA).
25. **Nair LS**, Nair MKC, Kumar S, Lukose R. Genetic evaluation of children with intellectual disability and autistic spectrum disorders. (Dept. of Medical Genetics; NIMS Spectrum-Child Development Research Center, NIMS Medicity, Trivandrum, India).
26. **Nair LS**, Thampi M. Genetic basis of chronic renal failure in adult population without any significant risk factors. (Dept. of Medical Genetics; Dept. of Nephrology, NIMS Medicity, Trivandrum, Kerala)
27. Olawoye O (PI), Olusanya B, Sarimiye T, **Monye HI** (Co-I). Effect of direct selective laser trabeculoplasty in reducing intraocular pressure in open angle glaucoma: An interrupted time series trial. (University of Ibadan, Nigeria; Eleta Eye Institute, Ibadan, Nigeria).
28. 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).
29. **Paredes-Moscosso SR**, Villegas-Llerena C, Guevara-Fujita ML, De León J, Rivera V, Davies DM, Fujita R. iCAR: Generation of CAR lymphocytes for cancer immunotherapy from iPSC lines (induced pluripotent stem cells). 2023-2025. (Universidad de San Martín de Porres, Perú; Leucid Bio, UK).
30. **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).
31. **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).
32. Rondon E, Gittens R, Anaya A, Daza MA, Vargas F, **Paredes-Moscosso SR** (Co-I), Enriquez E, Becerra L, Freire L. Comparitive anaylsis of scientific policies in Latin America. 2023. (multi-center, multi-countries in Latin America).
33. 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).

34. Valenzuela-Rubio N, Dahlgren A, **Ortiz-Panozo E**, Pérez-Gaxiola G. Mapping the ability to assess treatment claims: A survey of undergraduate students of nutrition in countries of Latin America. (Asociación Mexicana de Investigación en Nutrición y Salud, A.C., Cochrane México; Instituto Nacional de Salud Pública, Mexico; Norwegian Institute of Public Health).

2018 Batch

35. 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).
36. Awandare G, **Adeyemo TA**. Developing excellence in leadership training and science in Africa (DELTAS Africa). (Faculty of Clinical Sciences, College of Medicine, University of Lagos & Lagos University Teaching Hospital, Nigeria; University of Ghana, Ghana).
37. Bueleje JL, Acosta O, Huaman F, Obispo D, **Dueñas-Roque MM**, Purizaca N, Ledesma Y, Ochoa E, Guevara-Gil ML. Towards the study of the genome of the Peruvian inhabitant: Prevalence of germline mutations among various cohorts of probands suspected of predisposing hereditary cancer syndromes. (Universidad San Martín de Porres, Lima, Perú; Hospital Nacional Edgardo Rebagliati Martins, Lima, Perú; Hospital Ramiro Prialé, Huancayo, Perú; Hospital Regional de Cusco, Perú).
38. **Dueñas-Roque M**, **Cornejo-Olivas M,** Ortega J, Galarreta-Aima C. Ihope, an Ilumina project for genomic testing for rare diseases in Hospital Nacional Edgardo Rebagliati Martins, EsSalud, Lima, Perú. (Ilumina; Hospital Nacional Edgardo Rebagliati Martins, Perú; Instituto Nacional de Ciencias Neurologicas, Perú).
39. **Dutta AK**, Das S, Chakrabarti AK, Benegal V. Deciphering genetics of psychiatric disorders. (AIIMS, Kalyani; ICMR -Centre for Ageing and Mental Health, Bengaluru; National Institute of Mental Health and Neurosciences, Bengaluru, India).
40. **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).
41. **Gitaka J**, Pamme N. Development of molecular diagnostics for maternal infections using fiber mats. (Mount Kenya University, Kenya; Stockholm University, UK; Makerere University, Uganda).
42. **Lertwilaiwittaya P**, Vengoechea J. Review of Allgrove syndrome. (Center of Precision Animal Models, University of Alabama at Birmingham; Emory University, USA).
43. 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).
44. 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).
45. Mata IF, Inca-Martinez M, **Cornejo-Olivas M**, Cubas-Montecnio D, Manrique-Enciso C, Milla-Neyra K. Implementation of a Neurogenetics DNA Bank in Perú. (LARGE PD Consortium; GP2 and ASAP initiative; Instituto Nacional de Ciencias Neurologicas, Perú; San Marcos Foundation, Perú; Michael J. Fox Foundation, USA).
46. Mateo DiB, Alatise OI, Folaranmi L, Omoyiola T, **Abdulkareem FB**, Kingham TP. Effectiveness of a machine learning algorithm in diagnosing MSI-high cases through low-cost

H+E staining. (Obafemi Awolowo University, Nigeria; University of Ilorin, University of Lagos, Nigeria; Memorial Sloan Kettering Cancer Center, USA).

47. Mehmood A, **Naeem MA**, Qazi ZA, Butt H, Riazuddin S. Clinical Trial- Stem cell therapy for retinal diseases in patients. 2023. (CEMB, University of the Punjab, Lahore; Lyton Rehmatulla Benevolent Trust Eye Hospital, Lahore, Pakistan; In discussion).
48. 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. (Faculty of Clinical Sciences, College of Medicine, University of Lagos & Lagos University Teaching Hospital, Nigeria; Kings College London, UK).
49. **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).
50. 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).
51. **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).
52. Thorpe E, **Dueñas-Roque M**, **Cornejo-Olivas M**, Bazalar-Montoya J. iHope, an Ilumina project for genomic testing for rare diseases in Peru. (Ilumina, USA; Hospital Nacional Edgardo Rebagliati Martins, EsSalud; Instituto Nacional de Ciencias Neurologicas and Instituto Nacional de Salud del Niño San Borja, MINSA, Perú).
53. **Wangi KYW**, Shaleha RRA, Wijaya E. Psychosocial burden in patients with thalassemia: A systematic review. 2023. (College of Health and Human Development and Dept. of Public Health, Pennsylvania State University, USA; Hasanuddin University, Indonesia).
54. Xu J (PI), **Adeyemo TA**. Assessing Combination Hydroxyurea and Exogenous Erythropoietin in Sickle Cell Disease (ACHiEvE-SCD). (Faculty of Clinical Sciences, College of Medicine, University of Lagos & Lagos University Teaching Hospital, Nigeria; University of Pittsburgh, USA).

D. New Research Projects: (Total- 35; 2019- 22; 2018- 13)

2019 Batch

1. Alcausin MML, Lam H, **Tumulak MJR**. Cost-benefit analysis of cystic fibrosis in the Philippine Newborn Screening Program. (Revised per Ethics Review).
2. Bao M, **Kars ME**, Gettler K, Cho J, Itan Y. Phenome-wide association studies to investigate genes associated with very early onset inflammatory bowel disease.
3. Caneba JP, Maceda EB, **Tumulak MJR**, Alcausin MML. Clinical and molecular characteristics of Filipinos with confirmed Hemoglobin H Disease from 2019 to 2021. (Submitted to Technical Review Board).
4. **Chowdhury EH**, Dorji N, Lhendup K, Zangmo Y, Tobgay S, Phuntsho N. Incidence, and prevalence of congenital hypothyroidism in Bhutan. (Samtse General Hospital, Bhutan: Jigme Dorji Wangchuk National Referral Hospital, Thimphu, Bhutan).
5. Fakoya AOJ, **Akinmola OO**, Ramesh B. Stem cells for generating biological pacemaker tissue.

6. **Gyawali P**, Quinonez S. Current practice, and perspective for enhancing genetic services: An exploratory mixed-method study at Dhulikhel Hospital.
7. Hassib N, Abdel-Fattah M, **Sayed I, Mehrez M**, Ismail S, Abdel-Ghafar S, et al. Diagnosis of inherited disorders with oro-dental anomalies using recent advanced genetic approaches and impact of counseling and management.
8. **Hussein N**, Ramli R, Norimichi H, Lee PY, Lee CH, Beh HC. Revitalising Asthma Care and Treatment (ReACT): Developing and implementing asthma care kit for health care providers to improve asthma care in public health clinics.
9. **Kars ME**, Bao M, Gettler K, Cho J, Itan Y. Phenome-wide association studies of therapeutic targets of inflammatory bowel disease.
10. **Kars ME**, Gelb B, ItanY. Evaluation of genetic determinants of congenital heart disease using trio sequencing data.
11. **Kars ME**, Sevim Bayrak C, Wu Y, Gelb B, Itan Y. Investigation of genetic basis of pediatric food allergies.
12. **Kars ME**, Wu Y, Stenson PD, Cooper DN, Peter I, Itan Y. Investigating the genetic determinants of the comorbidity between Parkinson's disease and inflammatory bowel disease.
13. **Luong LH**, Hirofumi H. Generation and verification of EPO-GFP reporter iPS cell line for accurate monitoring of erythropoietin production in vitro and in vivo.
14. **Luong LH**, Hitomi H. Generation of human induced pluripotent stem cell (hiPSC) from patients with hereditary autoimmune disorders to study disease mechanism and potential therapy.
15. Okoh MP, **Alli LA**, Adejoh J, Madu C, Egua MO, Raheem D. A comprehensive review on the molecular and epigenetic mechanisms involved in the immune control of Plasmodium parasite infections. 2023.
16. **Okunola OO**. Heritability of chronic kidney disease in first degree relations in south-western Nigeria (Proposed).
17. **Sayed I**, Gamal El Din HM, **Mehrez M**, Abdel-Salam G, Ismail S,Abdel-Ghafar S et al. Advances in the delineation of genetic etiology of oro-dental anomalies in neurogenetic disorders.
18. **Tumulak MJR**, Abad PJ, Maceda EB. Knowledge on genetic services and family health history practices of medical doctors in the Philippines. (Proposed).
19. **Wiafe SA**, Aglozo EY, Thomford NE, Ali Naimatu. The burden and impact of rare diseases in Ghana- The Ghana Rare Impact Study.
20. **Wiafe SA**, Hammond C, Brew Y, Thomford NE, Maier U, Marbell M, Badoe E, Ashong J. Molecular and genetic diagnosis of patients suspected for metachromatic leukodystrophy plus related disorders in Ghana: The RDGI-Archimedlife- Orchard Therapeutics Project.
21. **Yesilcinar İ**, Şahin E. Determination of genetic literacy and level of genetic knowledge in nursing students.
22. Young B (PI), Everett L, **Monye HI** (Co-I). Choroideraemia atrophy progression kinetics in ultra-widefield fundus autofluorescence images. (Proposed).

2018 Batch

23. Abad-Murillo S, **Cornejo-Olivas M**. Comparison of epileptic seizures in spinocerebellar ataxia type 10 and other spinocerebellar ataxias diagnosed at the National Institute of Neurological Sciences during the 2018-2023 period.
24. **Abdulkareem FB**. CD70 expression in renal cell carcinoma.
25. **Abdulkareem FB**. PDL1 expression sin gastric and colorectal cancers.

26. **Adeyemo TA**, Adiat L, Osikomaiya B. Evaluation of vitamin D receptor polymorphism, bone mineral density and association with bone complications in patients with sickle cell disease.
27. Akinmurele T, **Taiwo O**, Oyebade O, Adeola O, Hollingsworth B. Systematic review on achieving sustainable health financing for Universal Health Coverage (UHC) through Mandatory Health Insurance Schemes in Low- and Middle-Income Countries (LMICs).
28. **Cornejo-Olivas M**, Illanes-Manrique M, Yrene-Cubas R. Factors associated with diagnostic delays in neurogenetic disease patients, treated at a specialized care institution in Lima, Perú.
29. Gupta A, Gupta A, **Dwivedi A**. Genomic characterization of dystrophin gene mutations in patients of Duchenne muscular dystrophy: A multicentric pilot study.
30. **Lertwilaiwittaya P**. Animal model to study the pathogenicity of variants of HMGB1 for variants reclassification.
31. MacLaren R, German G, Ruban A, Farkhat H, Kostiuk N, Parkhomchuk L, Kazak K, Lysytsia,… **Savina O**, et al. Ukraine Gene Therapy for hereditary eye disorders, at Okhmatdyt National Children's Specialized Hospital.
32. **Mburu S**, Gitonga, H, Ndung'u, R. Integrative comparative research approaches for combining information/data from multiple sources including into single analytical models.
33. Sultan A (Coordinator), Ismail T, Said L, Torre RM, Psychali C, Nako J, Calheiros C, **Messaoud O**, Hammami A, Brahmi K, Hamrouni B (Tunisian partners). Smart technique for treating sludge & sewage in Mediterranean countries. 2024-2025.
34. **Wangi KYW**, Birriel B, Smith C. Gene editing technology in people with genetic disorders: minus malum VS sumum bonum. (In discussion).
35. **Wangi KYW**, Birriel B, Smith C. The future of ethical, legal, and social implication in nursing genetics and genomics. (In discussion).

E. Ongoing Research Projects: (Total- 74; 2019- 45; 2018- 29)

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. (Knowledge and skills gained at the Summit helped mentor a specialist trainee to undertake this research to improve practice in primary care settings).
2. Alcausin MML, **Tumulak MJR**, Silao CL, Fabella TD. Clinical characterization, identification of CFTR gene mutations in newborns screening positive for cystic fibrosis in the expanded newborn screening program. (Summit lectures on Grant Writing helped write proposal).
3. Badoe E, **Wiafe SA**, Manu E, Adutum L, Adjetey DN, Thomford NE, Ashong J. Molecular and genetic diagnosis of MPS patients in Ghana: The RDGI-BioMarin-Archimedlife Project. (Knowledge from Summit and tailored training 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. Chagonda S, **Mhandire K**, Musarurwa C. High throughput sequencing towards optimization of African population specific cardiovascular gene panels. (Knowledge from Summit guided concept and methods).

6. Chakrabarty BK, Nagaraja M, Singhal P, Dagar Vikas, **Mutreja D**, Sen A. Evaluation of cytogenetic abnormalities and sperm FISH aneuploidy in infertile male. (Summit emphasized role of cytogenetic abnormalities in infertility).
7. 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).
8. **Chowdhury EH**, Lhendup K, Zangmo Y, Tobgay S, Phuntsho N. Incidence and prevalence of Turner syndrome in Bhutan. (Knowledge from Summit improved research quality).
9. Değirmenci T, **Yesilcinar İ**. Pregnant women's genetic literacy and affecting factors. (Summit inspired the project).
10. **Gyawali P**, Quinonez S. Medical genetics need assessment in Nepal: An online cross-sectional survey. (Summit emphasis on collaborations, research, and workshop on Grant Writing and Scientific Writing, helped write proposal).
11. Ibeh BO, Ifedilichukwu HN, Okoh MP, **Alli LA**. Modulation of cytokines and C-type lectin receptor expression in HIV infected population in Abuja, Nigeria. (Knowledge from Summit helped improve research design and execution).
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. Isuajah CE, Isuajah CC, **Akinmola OO**, Azinge EC. Fetal interleukin-6 and average plasma glucose concentration of obese pregnant women at term, in Lagos University Teaching Hospital. (Knowledge from Summit on prenatal diagnosis, complex relationship between maternal and fetal genes, placental activity, polygenic nature of obesity helped conceptualize study).
14. **Kars ME**, Itan Y. Genetic determinants and molecular mechanisms of obesity and cardiovascular disorders (Knowledge from Summit helped perform analyses).
15. **Kars ME**, Itan Y. Phenome-wide association studies and polygenic risk score calculations in sequencing cohorts from diverse ancestral origins (Knowledge from Summit helped perform analyses).
16. **Kars ME**, Unlu ES, Itan Y. Network-based heterogeneity clustering in primary open angle glaucoma (Knowledge from Summit helped write proposal).
17. **LLamos-Paneque A**, Christofolini DM. Clinical and genetic characterization of patients with Duchenne muscular dystrophy. (Summit lectures in Neurogenetics and emphasis on triplet expansion disorders, helped with execution of the study).
18. **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).
19. **Llamos-Paneque A**, Zambrano Ana Karina. VARIGEN: Genetic variants in rare and/or hereditary diseases in Ecuadorian individuals self-identified as mestizos who attend the Ecuadorian health system (Jan. 2022 – Dec. 2023). (Summit helped create partnerships, facilitate large genetic studies and clinical characterization of rare disease patients.)
20. **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).
21. **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).

22. **López-Star B,** Pérez- Serrano, Voourdain S. Gut microbiome in Latin people with diabetes mellitus with and without diabetic retinopathy. (Summit lectures provided tools to learn of most frequent genetic variation in Mexican population).
23. 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).
24. **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).
25. **Luong LH**, Hoang LT, Le NT, Tran TN. Study of rare immunodeficiency disorder. (Knowledge and credentials from the Summit initiated discussions and project).
26. **Mosema-Be-Amoti K**, Spencer D, Bramble MS, Likuba-Balali E, Ibanda-Matondo A, Kamate K. Differences of sex development (DSD) in Central Africa: Genetics, psychosocial adaptation and perceptions. (Summit attendance helped decide on this topic for a PhD thesis, in DR Congo).
27. **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 micro vessel density in angiogenesis of hematological malignancies. (Summit emphasized importance of gene-disease association in hematologic malignancies).
28. Myint YKS, **Khant AK**. Outcome of acute kidney injury in children by measuring the ratio of serum albumin and C reactive protein at Yangon Children Hospital (Knowledge, skills attained at Summit helped mentor specialist trainee for this research).
29. **Okunola OO**, Adekoya AA, Ojo EA, Adeoti PA, Adeyemi AA. Genetics study in autosomal dominant polycystic kidney disease in the tropics. (Used Summit resources and links to develop research questions).
30. **Okunola OO**, Ojo FA, Jones KP. Stigma related quality of life assessment scores in sickle cell disease patients in south-west Nigeria. (Used Summit materials and experience to design, write proposal).
31. Olabode SJ, Okunade KS, **Akinmola OO**, Ayorinde J, Akinsola T, Ajie O. Association between tumor necrosis factor alpha rs1800629 polymorphism and cervical cancer in Lagos State. (Knowledge from the Summit on GWAS, SNPs and variability of disease phenotypes played a major role in this study).
32. Olanrewaju B, Adegbola O**,** Okunowo AA, **Akinmola OO**. Association between serum fetuin A and occurrence of polycystic ovarian syndrome in women at the Lagos University Teaching Hospital. (Knowledge from Summit on polygenetic diseases helped conceptualize study).
33. **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).
34. **Paredes-Moscosso** SR, Villegas-Llerena C, Buleje S, Guevara-Fujita ML, Acosta O, **Dueñas-Roque 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 Perú. 2022-2023. (Summit lectures inspired and informed approach for project).

35. Sali BA, **Hussein N**, Ng WL. Exploring the views and experiences of primary care doctors in managing depression in primary care. (Knowledge and skills gained at the Summit helped mentor a specialist trainee for this research to improve practice in primary care settings).
36. **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).
37. Sondhi V, Mishra P, Sridhar G, Singhal P, **Mutreja D**, DBT- NIDAN Kendra. Genetic Center for Neonatal and Prenatal Screening. (Summit emphasized importance of mass neonatal screening).
38. 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 and phenotypes helped conceptualize study).
39. 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).
40. 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 helped lead the local research team).
41. 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. (Summit lectures informed approach for project).
42. **Wiafe SA**, Anyane-Yeboa K, Brew YA, Badoe E, Thomford NE, Ashong J, Maier U, Marbell M, Hammond C, et al. Clinical and genetic evaluation of undiagnosed genetic and rare disease patients in Ghana- The Ghana Undiagnosed Disease Program. (Knowledge from Summit helped improve project design and develop protocols).
43. **Wiafe SA**, Wiafe AA, Addo-Lartey E, Baynam G. Generative AI predictive models as tools for rare diseases diagnosis in Ghana, dxGPT. https://dx29.ai/https://dxgpt.app/ , https://wanglab.ml/clinical-camel.html. (Knowledge from Summit and tailored training helped design project).
44. Win TS, San CC, **Khant AK**, Lwin LW. Clinical profile of systemic lupus erythematosus children at Yangon Children Hospital (Summit lectures improved skills in reporting results).
45. **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).

2018 Batch

46. **Adewole OO**, Sogaolu OM, Aminu AA, Nwosu N, Adeyelu A. Clinicopathologic and genomics of lung cancers among Nigerians. (Leveraged on additional knowledge from Summit in clinical research and project implementation).
47. **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 from Summit in lung cancer genomics and biobanks).
48. 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).

49. Afolabi BB, Galadanci H, Balogun M, **Adeyemo TA**, Sam-Agudu N. Intravenous ferric carboxymaltose versus oral ferrous sulphate for the treatment of moderate to severe postpartum anemia in Nigerian women (IVON-PP): An open label, randomized controlled trial and implementation study. (Knowledge from Summit supported study design, implementation).
50. 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).
51. 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).
52. **Cornejo-Olivas M**, Araujo-Aliaga I, Isasi R, Sarapura-Castro E, Manrique-Enciso C. Addressing diagnostic gaps on inherited neuropathies and related disorders. (Summit lectures on Bioethics and Scientific Writing helped improve informed consent form and writing of proposal).
53. **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).
54. **DaichVarela M**. Long-read sequencing and transcript splicing analysis in RDH12 and PHYH. (Summit introduced genetics/genomics in ophthalmology, and its importance in precision medicine).
55. **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 Perú 2019-2021. (Summit encouraged research focus on rare diseases in Perú).
56. **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 conceptualize and execute project).
57. **Dwivedi A**, Kalra S. Genotype- phenotype correlations in children with renal tubular acidosis: Unravelling the mystery. (Knowledge from Summit helped conceptualize project).
58. **Dwivedi A**, Kalra S. Study the role of exome sequencing in children with congenital anomalies of kidney and urinary tract with extra renal manifestations. (Knowledge from Summit helped conceptualize project).
59. **Dwivedi A**, Kumar R, Singh AK, Dey M. Clinical utility, and diagnostic yield of exome sequencing in prenatal diagnosis of congenital anomalies. (Knowledge from Summit helped conceptualize project).
60. Ghosh T, **Dutta AK**. The association of TMPRSS6 polymorphism rs855791, rs2413450, and rs4820268 with iron and haematocrit parameters among women of reproductive age group in rural population of Nadia district, West Bengal. (Summit lectures, NCBI resources on genetics of hearing loss and variant prioritization were critical for this project).
61. Gidado, Agnes A, **Adewole OO**. Stigma reduction in tuberculosis. (Leveraged on additional knowledge gained at the Summit on project implementation and research collaboration).

62. Jaja C, **Edem-Hotah J**, Shepherd J. Expanding access to sickle cell disease care in Sierra Leone. The EASEL Pilot Implementation Project. 2021. (Summit initiated idea to expand access for sickle cell disease care).
63. Kirenga B, 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. (Summit lectures on Ethics in Research helped develop protocol, informed consent forms, protocols/forms for storage of biospecimens at a biorepository for future research).
64. 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. (Summit lectures helped develop protocol, informed consent forms, protocols/forms for storage of biospecimens at the biorepository for future research).
65. Kosiyo P, Otieno W, **Gitaka J**, Nyamuni J, Ouma C. Sickle cell genotypes in malaria. (Knowledge from Summit in molecular techniques enhanced sickle cell work, improved protocol development and data analyses).
66. **Lertwilaiwittaya P**, Rodriguez M, Callaway K, Korf B. University of Alabama, Birmingham's Undiagnosed Disease Program Review. (Knowledge from Summit helped in retrospective chart reviews).
67. 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).
68. **Mburu, S, Gitaka J**. A mixed predictive study design on breast cancer sub-types in Kenya and associated epidemiological risk factors, to identify population specific diagnostic and prognostic biomarkers as therapeutic targets. (Summit helped understand how to design mixed predictive studies using data analytics from multiple sources).
69. **Messaoud O**. Development of nano biosensors for molecular biology applications (Mentors, colleagues at Summit advised to and helped focus on a specific disease for research).
70. **Messaoud O**. Genetic investigation of DNA repair disorders in Fanconi anemia, Xeroderma pigmentosum and atypical forms of photogenodermatoses. (Knowledge gained through Summit's tailored training helped interpret incidental findings).
71. **Nakousi-Capurro N**, 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. (Summit-UDP program reinforced the need for genotype-phenotype studies in Latin American population, (Knowledge from Summit on GeneMatcher helped identify new phenotype-genotype associations).
72. **Nakousi-Capurro N**, Cavalcanti DP. Description of genotype-phenotype relations in patients with osteogenesis imperfecta in a Brazilian cohort. (Summit helped reinforce the need for genotype-phenotype association for Latin American population).
73. Onipede A, Awopeju OF, Obadire T, **Adewole OO**. TB gut microbiome. (Levaraged on additional knowledge gained at the Summit on research collaborations).
    Pericak-Vance M, **Cornejo-Olivas M**. Genetics of dementia in a Peruvian cohort. (Summit lectures on Bioethics and Scientific Writing helped improve informed consent form, writing of proposal).

74. **Taiwo OO**, Dhlakama DH, Jalo HP, Idowu EA, Ojukwu BT. A single blind, Randomized Controlled Trial (RCT) to assess oral health education intervention in 12-year-old Nigerian students. (Knowledge from Summit helped design, positioned me to lead the research).

F. Other Accomplishments/Initiatives:

2019 Batch

1. **Abubakar S**. Lectured on 'Ethical, Legal & Social Implication (ELSI) of genetics and genomics' in the course on 'Medical Ethics & Law' for MPH- Global Health, at College of Health Sciences (Apr. 2023), and the World Bank Funded Africa Center of Excellence for Population Health and Policy, Bayero University, Kano, Nigeria (Feb. 2023); Appointed, Examiner, included questions on genetics and genomics for Membership Exams, at the Faculty of Community Health, West African College of Physicians, Lagos/Abuja, Nigeria; Appointed, Qualitative Research Expert, on Service Delivery Innovations- a multi-country study (Jul. 2023), by Centre for Integrated Health Projects, Abuja, Nigeria; Appointed, Examiner, assessed dissertations (MPH-2) for Nigeria Field Epidemiology & Laboratory Program (Jul. 2023), Nigeria Centre for Disease Control & Ahmadu Bello University, Zaria, Nigeria.
2. **Akinmola OO**. Implementation of Genetic Counselling Unit in the Tertiary Hospital, Lagos University Teaching Hospital, under consideration by Management; Awarded, Virtual Summit Scholarship by Wellcome Connecting Science for attending events - Curating the Clinical Genome (Jul. 2023), Multi-omics in Metabolic Disease (Jun. 2023), Genomic Imprinting (Mar. 2023), for deepening and creating a platform for genomic medicine among other multi-omic approaches for training; Attended, Euro Lab International Federation of Clinical Chemistry (IFCC) Conference (Rome, 2023); 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, including resources and logistics for shipping proband samples.
3. **Alamin Alabid T**. Speaker on 'Factor XIII Gene Polymorphisms among Sudanese Patients with Intracerebral Hemorrhage', at the 4th International Symposium and Workshop on Functional Genomics and Structural Biology Kuala Lumpur, (Malaysia, Dec. 2022); Completed online training in Genomic Medicine offered by African Genomic Medicine Training Initiative 2023-H3Africa, sponsored by NIH (Jun. 2023); Selected, Member of International Advisory Board for 5th & 6th International Conference on Engineering Professional Ethics and Education, at International Islamic University, Malaysia (Aug. 2023).
4. **Alli LA**. Appointed, Associate of Innovation Hub, Innovation Centre, Abuja, Nigeria (Jul. 2023); Re-appointed, Visiting Lecturer, Medical Biochemistry and Molecular Biology for preclinical medical students, Nile University, Abuja, Nigeria (Jul. 2023); Appointed, External Examiner, Medical Biochemistry, for preclinical medical students, Baze University, Abuja (Mar. 2023); Supervised and mentored postgraduate research students (2 MSc); Used materials from Summit to lecture medical students on 'Basic Molecular Biology', 'Genetic Disorders', 'Inborn Errors of Metabolism'; Strengthened Biochemistry curriculum in Genetics and Genomics for postgraduate Medical Biochemistry students.

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; Selected for poster presentations on 'A structured home-based egg ladder can reduce the demand for hospital baked egg challenge in John Radcliffe Hospital, OUH', 'Anaphylactic reaction due to storage mites - pancake syndrome', 'Allergic reaction to cow's milk-based protein supplement in a child who is tolerant to cow's milk', 'Case series on tolerance to whole-grain wheat cereal products in three wheat allergy children', at the British Society of Allergy and Clinical Immunology (BSACI) Annual Conference (UK, Oct. 2023); Presented poster on 'A case report of a girl with congenital amegakaryocytic thrombocytopenia' at the 7th Annual Rare Disease Day Symposium Sri Lanka College of Paediatricians (Sri Lanka, Mar. 2023); Maintained database for recording syndromes, congenital malformations, genetic disorders at the University Pediatric Dept., Teaching Hospital Anuradhapura, Sri Lanka; Identified, diagnosed and treated genetic cases (20) - syndromes (12), hemoglobinopathies (5), ADA2 deficiency (1), hemophilia (5).
6. **Baatar N**. Helped establish a national level web and mobile telemedicine service, includes e-commerce in medicine, OTC drugs, consumer goods; Project continuation through membership service will include Dr. functions, corporate health checkup packages, booking in-clinic appointments, viewing of test results and medical records, referrals to specialist; Attended, Quality Standards in Telehealth, by IFC (Virtual, May 2023); Attended, Early Diagnosis of Newborn Screening Tests In Premature, Low Birth Weight And Sick Newborns, hosted by Screening Diagnostic Reference Center (Jan. 2023);
7. **Bocoum A**. Presented on 'First case of Huntington's disease like 2 in Mali, West Africa', at the 14th International Congress of Human Genetics (South Africa, Feb. 2023).
8. **Chowdhury EH**. Specialist in Dept. of Pediatrics, Samtse General Hospital (SGH), Royal Government of Bhutan (since 2020), Summit helped enhance CV and secure position; Initiated, continued Registry on Congenital Abnormalities, at 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 (Nov. 2022); Identified and managed pediatric genetic disorders (16) - Down's syndrome (4), hemophilia (2), congenital hypothyroidism (2), achondroplasia (2), neurofibromatosis (1), Turner's syndrome (3), Marfan's syndrome (2); Referred cases for cardiac surgery (2), 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 patients (28) - trichodentoosesous syndrome (1), ectodermal dysplasia (8), dentin dysplasia Type II (1), dentinogenesis imperfecta (1), amelogenesis imperfecta (2), isolated oligodontia cases (15); Referred trichodentoosesous syndrome and ectodermal dysplasia patients to the Genetics Dept. for molecular confirmation and medical referral; Patients referred from Genetics Dept. for dental examination (14) - Bardet-Biedl syndrome (4), Klinefelter syndrome (2), EVC syndrome (2), Hallerman Streiff syndrome (1), MPS syndromes (4), bone marrow failure syndrome (1); Provided dental prosthodontic, restorative therapies; Preventive and follow-up protocols, early diagnosis and surgical interventions prevented fatal consequences, contributed to social and psychological well-being of children, improved quality of life.
10. **Gyawali P**. Promoted to Assistant Professor, Kathmandu University School of Medical Sciences, Nepal (Jan. 2023); Awarded, Josip Matovinovic Endowed Clinical Medicine Fellowship, University of Michigan (Oct. 2023); Guest Speaker, on 'Early Career Investigators

- Career Development', at 7th G2MC meeting on Accelerating Implementation of Genomic Medicine for Global Equity & Access, Geneva (Switzerland, Oct. 2023); Member, Global Perspective on the Challenges of Establishing Genomic Medicine Practice- Young Investigators Writing Project.

11. **Hussein N**. Awarded, Certificate of Excellent Service from Universiti Malaya, Malaysia (Dec. 2022); Member, Academic Committee of Medical Humanities and Ethics Unit, Faculty of Medicine, Universiti Malaya, Malaysia (2022-2024); Member, Cochrane Collaboration, United Kingdom (2023-2028); Examiner, Coordinator for Master of Family Medicine Specialty, Universiti Malaya, Malaysia (Jun. 2023); Audited postgraduate Family Medicine curriculum to strengthen training in family history; Committee member for teleconsultation (Jan. 2023); Speaker, Facilitator for workshops on 'Evidence-based Medicine for Healthcare Providers' (Jul. 2023), 'Developing School Asthma Action Plan For Primary Schools' (Jul. 2023), 'ABC of Critical Appraisal for Laboratory Personnel' (Jun. 2023), 'Introduction to Research and Quality Improvement' (Jan. 2023); Chairperson, Facilitator for Thalassemia Camp for Patients and Parents/Caregivers (Mar. 2023); Grant Reviewer for 'Reliability and Construct Validity of Knowledge, Attitude and Practice of Doctors in Primary Healthcare Clinics Regarding E-cigarettes Cessation' (Apr. 2023); Attended Genetic Counselling Conference, Kuala Lumpur, Malaysia (Dec. 2022); Mentored Family Medicine trainee on prenatal genetic testing in primary care settings; Reviewer, PhD student thesis on 'Views of Tratuzumab Biosimilar Use in Patients with HERS Breast Carcinoma'; Journal Reviewer - npj Primary Care Respiratory Medicine-Nature (Apr. 2023) and Health Science Reports (Feb. 2023); Initiated, implemented a Revitalising Asthma Care and Treatment (ReACT) Registry to improve asthma documentation and audit.
12. **Iqbal M**. Chief Organizer, 1st International Conference on 'Innovations in Chemistry, Biochemistry, Biotechnology and Bioinformatics' (Dec. 2022); Delivered lectures on 'Advanced Molecular Genetics and Classical Genetics' to graduate & undergraduate students; Supervised graduate research students (5 PhD, 7 MS) on molecular and genetic basis of different hereditary disorders; Appointed, External Examiner, reviewed MS theses (2); Journal Reviewer- PLOS ONE.
13. **Kars ME**. Postdoctoral Fellow at Institute of Personalized Medicine at Icahn School of Medicine at Mount Sinai, NY (since 2022); Presented talk/poster on 'Application of network-based heterogeneity clustering for investigation of genotype-phenotype correlations in BioMe BioBank', at the Gordon Research Conference- Human Genetics and Genomics (New Hampshire, Jul. 2023), Cold Spring Harbor Laboratory-The Biology of Genomes meeting (New York, May 2023), 25th Annual Child Health Research Day (New York, Apr. 2023); Presented talk/poster on 'A comprehensive knowledgebase of known and predicted human genetic variants associated with COVID-19 susceptibility and severity', at Annual Clinical Immunology Society Meeting (Missouri, May 2023), Rocky 2022 Bioinformatics Conference (Colorado, Dec. 2022), 6th NYC Inborn Errors of Immunity Symposium (New York, Dec. 2022); Cold Spring Harbor Laboratory Biological Data Science meeting (New York, Nov. 2022), The Mindich Child Health and Development Institute Annual Retreat (New York, Nov. 2022); Shared knowledge acquired at Summit with graduate students (2) and supervised two research assistant interns (2); Journal Reviewer - Current Biology and BMC Genomics.
14. **Khant AK**. Senior Consultant Pediatrician, Pediatric Nephrology Dept., Yangon Children Hospital; Attended, World Congress of Nephrology (Virtual; Apr. 2023); Lectured undergraduate medical students, post graduate pediatric students, at University Pediatric Dept.,

Teaching Hospital, Yangon Children Hospital; Maintained database for recording acute kidney injury, chronic kidney disease, syndromes with renal abnormalities, cystic kidney disease, at Yangon Children Hospital; Examined, diagnosed and treated pediatric cases (500) in the emergency department - acute diarrhea (70), acute viral infection (200), dengue hemorrhagic fever (50), acute respiratory tract infection (60), pyrexia of unknown origin (30), syndromes (5); renal replacement therapy for acute kidney injury (25), chronic kidney disease children (30); Counselled patients with hereditary renal disease (30) - congenital nephrotic syndrome (1), congenital abnormalities of kidney urinary tract (20), meningomyelocele with neurogenic bladder (6), syndromic child with renal abnormalities (2), Lowe syndrome with renal tubular acidosis (1), Prune Belly Syndrome (1), Caroli syndrome (2), 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; Lectured Masters class on 'How to Approach Hereditary Amyloidosis Due to Transthyretin from Genetics' (Ecuador, Oct. 2022); Awarded travel scholarship for course on 'Basics in Human Genetics Diagnostics-VI' for CLGs* (Portugal, Sep. 2023); Participated as an expert Neuroeconomix S.A., in proposal evaluation on 'Evaluation of Effectiveness and Safety and Adaptation of the Economic Model of Ataluren for the Management of Duchenne Muscular Dystrophy due to Nonsense Mutation' (Ecuador, Jul. 2023); Presented on 'Genetics of Severe Hypertriglyceridemia and Familial Chylomicronemia Syndrome' at Army Hospital, Quito (Jul. 2023); Attended, Neonatal Screening Symposium, by Colombian Society of Human Genetics (May, 2023); Attended online continuous training courses and webinars: 'From Dysmorphology To Genetics in a Round Trip', by PTC Therapeutics (Jun. 2023), 'Course Panels, Exomes and Genomes in Clinical Practice', by Genotipia (May-Jun. 2023), 'Congenital Metabolic Diseases: Knowing to Diagnose', by AMGH (May- Jun. 2023), 'EIM Update: ASMD & Niemann Pick-C Disease', by SLEIMP (Online, Mar. 2023); Attended Euro-Latin American Summer School of Myology- EVELAM (Ecuador, Dec. 2022); Participated in 'Registry of Patients with Duchenne Muscular Dystrophy', sponsored by Ecuadorian Medical Federation, Manta (Ecuador, Nov. 2022); 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; Summit improved genetic counseling skills, implemented genetic counseling service at a private clinic's Maternal Fetal Unit (~40 cases/yr); Identified cases (11) - structural chromosomes anomalies including supermarket chromosome, confirmed by molecular cytogenetics, by German collaborator (Dr. T. Liehr) as this service is not available in Ecuador; Confirmed and evaluated genetic cases (211) - mono gene etiology via molecular tests (56), chromosome via cytogenetic studies (65), or multifactorial (90); Registered new cases (211) in the Registry of Genetic Diseases, Specialty Hospital No.1 FF. AA, Quito, Ecuador; Re-established the expanded metabolic screening for 27 diseases with 100 percent coverage for births (~200-300/yr); Enrolled an Ecuadorian patient with Dr. A. Slavotinek (Cincinnati Children's Hospital Medical Center, OH) for genetic analysis of patients with eye anomalies and rare disorders; Member, Editorial Board- Molecular Cytogenetics- BMC.
16. **López-Star B**. Knowledge acquired at Summit helped initiate an Ophthalmogenetics Dept., at Mexican Institute of Ophthalmology, with a telemedicine program in collaboration with other geneticists in Mexico City; Member, Pan American Inherited Retinal Diseases Group (since Feb. 2020); Trained junior staff in ophthalmogenetics; Continued to record, monitor cases (100) with inherited retinal disorders; Evaluated, diagnosed, treated opthalmogenetic cases (16) - congenital

cataract (4), congenital glaucoma (4), retinitis pigmentosa (8); Referred patients (10) for medical management to Children's and Women's Hospital, Mexico; Diagnosis improved quality of life of 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); Enrolled as PhD candidate in the International Postgraduate Program- iPS Cell Regenerative Medicine, at Kansai Medical University, Japan (Sep. 2022).
18. **Mhandire K**. Appointed, Genetic Variant Scientist- Review Analyst, GeneDx, MD, US (since Feb. 2022); Collaborated with genetic counselors and clinical geneticists to classify genetic variants according to the American College of Medical Genetics guidelines in neurodevelopmental disorders, whole exome/genome screening, prenatal/neonatal screening, targeted familial testing, metabolic disorders; Analyzed cases from the national newborn screening project GUARDIAN, https://guardian-study.org; Evaluated 50-60 cases/week; Co-wrote genetic test results for patients and doctors, applied knowledge acquired at Summit, including HGVS nomenclature, next-generation sequencing, and neonatal screening for variant interpretation.
19. **Monye HI**. Appointed, Director of Ophthalmic Research and Education, Eleta Eye Institute, Ibadan, Nigeria (Oct. 2022); Completed the '2023 African Genomic Medicine Training Initiative Course-Medical Doctors' (Jun. 2023); Completed course on 'Fundamentals Implementation Science' from University of Washington (Dec. 2022); Awarded the H3Africa Travel Fellowship, and Second best Fellows' presentation for 'The burden, manifestations and parental willingness to test for genetic eye diseases in children in Ibadan, Nigeria – Evidence for planning', at the 20th Consortium Meeting (Feb. 2023); Initiated a phenotypic and demographic database of patients with genetic eye diseases at Eleta Eye Institute; Identified, enrolled patients (17) - retinitis pigmentosa (9), juvenile open angle glaucoma (3), Stargardt disease (2), Marfan Syndrome (1), optic disc coloboma (1) and choroideraemia (1); Sensitized trainee ophthalmologists on evaluation of genetic eye diseases.
20. **Mosema KBA**. Lectured on 'Genetics' to pediatrics medical students at Université Protestante du Congo, Kinshasa and Université Uele, Isiro, Haut-Uele Province; Submitted abstract on 'Diagnosing rare conditions in low-resource settings: The experience of the differences of sex development clinic in the Democratic Republic of the Congo', to ASHG 2023 Annual Meeting (Nov. 2023); Speaker on 'Differences of sex development in Central Africa: Genetics, psychosocial adaptation and perceptions', 'Systematic screening of sickle cell in children and family centered care in the Biamba Marie Mutombo Hospital' and ' Treatment with hydroxyurea in children in the Biamba Marie Mutombo Hospital', at the 11th Meeting of the Society of Congolese Pediatricians, Kinshasa (DRC, Feb. 2023); Participated in the 7th Meeting of African Cancer Coalition (> 70 oncologists and physicians from 12 countries): Harmonization of Cancer Treatment Guidelines for use in Sub-Saharan (Tanzania, Oct. 2022); Continued establishment of Sickle Cell Disease Unit (screening, treatment, follow-up), at Biamba Marie Mutombo Hospital, Kinshasa City; Continued clinical training network (WhatsApp) in genetics and genomics for medical students and doctors; Diagnosed case of neurofibromatosis (1).
21. **Mushi TL**. Completed Masters in Medicine in Pediatrics and Child Health (MMED Pediatrics and Child Health (Sep. 2023); Attended training on 'The Changing Landscape of Meningococcal Meningitis and Vaccine Recommendations' (Virtual, Jul. 2023); Attended one-month observership in Pediatrics, at the Ann & Robert H. Lurie's Children Hospital of Chicago (Jun. 2023); Attended 3D-Echocardiography Conference in Chicago (Jun. 2023); Trained colleagues

in newborn screening; Established, continued to enroll in a Birth Defect Registry at Bugando Medical Centre, Tanzania; Diagnosed malformation cases (231), based on phenotype - congenital heart disease (37), anorectal malformation (65), omphalocele (12), gastroschisis (61) and others (56 - Down's syndrome, Patau' syndrome, Prune Belly syndrome, spina bifida); Offered counselling as part of routine management due to unavailability of a geneticist or genetics counselor; Medically managed congenital heart disorders with hemodynamic instability, others received surgical interventions.

22. **Mutreja D**. Appointed, Lab Director, Professor & Head, Dept. of Laboratory Medicine Command Hospital Air Force Bengaluru (Jan. 2023); Delivered lectures in 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; Presented poster on 'Rare presentation of an extra-nodal diffuse large B-cell lymphoma as a scalp nodule with marrow involvement', at MAPCON - local chapter (Apr. 2023); Speaker on 'Polymerase chain reaction', at IAPM APCON (Nov. 2022, Karnataka, India); Identified cases (14) with the help of Dr. B. Chakraborty, with cytogenetics and histopathologic/microscopic diagnosis - Trisomy 21 (1), mosaic turner syndrome (1); mosaic triple X syndrome (1); Edward syndrome/trisomy 18 (1); atypical fusion pattern in CML (1), complex karyotype in a case of AML (1), thalassemia (4), sickle cell disease (1), Gaucher disease (1); aided in diagnoses of infertility patients by demonstration of supernumerary marker chromosome (2); t(1;7)(q23;q32)(1); abnormal derivative Y chromosome (1); Klinefelter syndrome(1); Continued as Section Editor (Pathology) for Indian Journal of Medical Paediatric Oncology (2022-23).
23. **Nair L**. Conducted webinars for pediatricians including developmental pediatricians, therapists and nurses on 'Early Identification of Genetic Disorders Using a Community-Based Approach' (Jan.- May 2023, Aug.- Nov. 2022); Conducted OPD, evaluated, diagnosed patients with genetic disorders (211), identified cases (88) - intellectual disability/ developmental delay (22), recurrent pregnancy loss/ infertility (11), rare genetic disorders like MPS III (1), Coffin Siris syndrome (2), Angelman syndrome (1), hyper oxalosis (3) and others (48), cases undiagnosed or lost to follow-up (123); Provided genetic testing and counselling to patients.
24. **Okunola OO**. Used experience and materials from Summit to train, mentor medical students, Residents, colleagues in medical genetics and genomics, research grant applications; Liaison on working group with pediatric nephrologists, established congenital kidney disease registry with follow up; Diagnosed patients (20) with inherited kidney diseases (all autosomal dominant polycystic kidney disease, ADPKD), others (12) with congenital anomalies of the kidney and urinary tract (CAKUT)- ureteropelvic junction obstruction (8), renal agenesis (2) and horse shoe kidneys (2); 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 (Aug. 2022) to Harvard T.H. Chan School of Public Health for doctoral training in Reproductive, Perinatal and Pediatric Epidemiology, completed first year of doctoral training (Oct. 2022); Reviewer, for Agency for Public Health Education Accreditation, Brussels, Belgium (Oct. 2022).
26. **Paredes-Moscosso, SR**. Selected to participate in the Science Leadership Program for Latin America, organized by the Global Young Academy (Dec. 2022); Awardee, for course on 'Cancer Genomics and Transcriptomics', by EMBL-EBI, UK (Jul. 2023), presented poster on 'Overview of current and future projects'; Speaker on 'Bridging science to the world: Science communication principles and methodologies for impactful and well-structured messages', co-

author on ‘Leticia Declaration: Collaborative leadership for a sustainable CTI system with social impact in Latin America and the Caribbean’, at the Sustainability Research Institute Congress (Panama, Jun. 2023); Speaker on ‘In vitro modeling by CRISPR/Cas9 of a VUS mutation found in Peruvian patients with hereditary breast cancer’, at the Multidisciplinary International Seminar Cycle, by Universidad Autónoma de Tlaxacala, Mexico, and Universidad San Carlos, Guatemala (Online, May 2023); Invited peer reviewer for ‘Funding Research’ 2021-II call by Universidad Católica de Santa María, Perú (Nov. 2022).

27. **Pirlog R**. Completed fellowship in Breast Cancer Pathology, at Institute Curie Saint-Cloud, France (Nov. 2022- May 2023); Started fellowship in Hematopathology and Dermopathology at Henri Mondor Hospital, Creteil, France (May 2023); Accepted to the European Master of Molecular Pathology Program, France (2023) to advance knowledge in molecular pathology.
28. **Sayed I**. Associate Professor of Oro-dental Genetics; Participated in preparation of workshop ‘Basics of Oro-dental Genetics’ to introduce genetics to dentists working in Ministry of Health; Supervised, mentored PhD students (2); Identified cases (57) - neurogenetic manifestation (15), skeletal manifestation (20), ectodermal dysplasia patients (18); Referred patients (4) for prosthetic management of ectodermal dysplasia.
29. **Tiong SY**. Elected as Committee member, Genetic Counselling Society Malaysia (GCSM) (2022-2023); Presented poster on **‘**Double trouble in a child with global developmental delay and severe failure to thrive- importance of genetic counselling’, at the 4th BCM-CUHK-NUS Joint Symposium in Clinical Genetics (Singapore, Sep. 2023); Presented poster on ‘Case report: Two Malaysian patients with neurological-predominant CACNA1C-related disorder’, at the AOCCN (Thailand, Aug. 2023); Moderator for Malaysian Rare Diseases Society Patient Engagement Activity, at AGM & Outreach (Jun. 2023); Lectured on ‘Inherited metabolic disorders newborn screening’ for Antenatal Class (Feb. 2023); Implemented mandatory newborn screening for 33 types of inherited metabolic disorders (IEM) at Loh Guan Lye Specialists Centre (LSC), along with genetic counselling service (Jul. 2023); Continued to increase awareness among expectant and mothers on new born; Newborn screening uptake for IEM is about 56% (Oct. 2022 - Jun. 2023); Received new referrals (68) for genetic counselling (Oct. 2022 - Jun. 2023).
30. **Tumulak MJR**. Promoted to Research Assistant Professor I (Summit attendance was included as part of specialized training); Presented poster on ‘Psychosocial issues of Filipino parents with a child with maple syrup urine disease’, at Study of Inborn Errors of Metabolism (SSIEM) Conference, Jerusalem (Israel, Aug. 2023); 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 (renewed Jul. 2023); Thesis panel member for genetic counseling students (10); 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 (850) - breast cancer patients (10), cystic fibrosis patients (10), alpha and beta thalassemia patients (700), fatty acid disorders patients (100), InGeSt study participants (30).
31. **Wiafe SA**. Appointed as Co-Chair of the Africa Rare Disease Alliance (Jun. 2023); Organizer, speaker on ‘The Rare Disease Landscape in Africa’, at the 5th Symposium on Rare Disease and Orphan products, at the 11th African Pharmaceutical Symposium (Accra, Jul. 2023); Attended,

Undiagnosed Disease Hackathon, Karolinska Institute (Sweden, Jun. 2023); Speaker on 'The Ghana UDP and the Champions Initiative, at the Undiagnosed Disease Day Webinar hosted by Wilhelm Foundation (Virtual, Apr. 2023) and at the Re(ACT) Congress (Berlin, Mar. 2023); Speaker on 'Report on the Developing Nations Working Group', at the 11th UDNI Conference, Vienna **(**Austria, Nov. 2022); Member, IRDiRC-RDI Global Access Working Group (Jan.-May, 2023); Member, Funding Models Task Force, IRDiRC (Mar. 2023); Member, Taskforce on Impacts, IRDiRC (Mar. 2023); Established Congenital Anomalies, Undiagnosed and Rare Disease Registry in Ghana, enrolled 320 patients; Enrolled 3 patients in the Takeda Access Program; Enrolled 1 patient in MT1621 treatment for TK2 deficiency; Launched a quarterly case conference (Aug. 2023); Diagnosed cases (192) through the Undiagnosed Disease Program - G6PD (20), spinal muscular atrophy (7), syndromes without a name (5), HBB-related disorders (5), GRIN2A-complex neurodevelopmental disorder (4), MPS II/Hunter Syndrome (4), tuberous sclerosis complex (4), Bardet-Biedl syndrome (3), hereditary transthyretin amyloidosis (3), lymphedema-distichiasis syndrome (3), mitochondrial DNA depletion syndrome 2, myopathic type (3), MPS Iva/Morquo syndrome (3), osteogenesis imperfecta (3), SCN1A seizure disorders (3), 22q13.2-q13.33 duplication (2), COL2A1-related disorders (2), glycogen storage disease type II (2), MED12-related disorders (2), partial trisomy 3p syndrome (2), PLA2G6-associated neurodegeneration (2), Rett syndrome (2), X-linked intellectual disability, Cabezas type (2) and 1 each of 11q22.2-q25 duplication, 15q26.2-q26.3 duplication, 15q26.3 deletion, 16p12.2p11.2 microdeletion syndrome, 20p13 microdeletion, 2q13 microduplication, 3q duplication syndrome, 3q26.33q27.2 deletion, 8p duplication among other defects and syndromes.

32. **Yadav S**. Clinical Geneticist at Faith Diagnostic and Fetal Centre, Mohali, India; Used materials and knowledge from Summit to create awareness on genetic disorders among clinicians training for super-specialization in multiple clinical disciplines through Speed Institute, Chennai (online platform); Moderator, Panel discussion on 'Fetal Medicine and Genetics- Solving the Diagnostic Dilemma Of Fetal Abnormalities', at FERTICON, Chandigarh (May 2023); Panelist, webinar on 'National Fetal Growth Restriction', by IRIA, New Delhi (India, Dec. 2022) with focus on genetic causes of fetal growth restriction and identification, using relevant genetic tests; Conducted workshop on 'Genetic Diseases and Implications of Genetic Tests in Diagnosis and Management of Rare Diseases', at the Annual Symposium on Genetic Disease 'From Bench to Bed Side', New Delhi (India, Oct. 2022); Trained Fetal Medicine and Genetics Fellows (2); Involved in preconception and antenatal genetic counselling of patients (52) - Carriere of beta thalassemia (5 couples), previous baby with Meckel gruber syndrome (1), common aneuploidy on antenatal screening (10), couple carrier screening for previous neonatal deaths, babies with tyrosinemia and ichthyosis, 2 degree consanguinity (5), family history of deafness (2), recurrent abortions /bad obstetric history (5), multiple congenital malformations (17) were diagnosed using antenatal advanced 2D/3D ultrasonography and fetal ECHO, including facial clefts (1), arthrogryposis (2), neural tube defects, cardiac defects like right aortic arch (1), hypoplastic left heart syndrome (1), chondrodysplasia puncta (2), Blake's pouch cyst (2).
33. **Yesilcinar I**.

2018 Batch

34. **Abdulkareem FB**. Appointed, External Examiner, Pathology, Eko Medical University, Lagos, (Apr. 2023); Committee Chairman/Coordinator for Pathology Update/Revision Course of the National Postgraduate Medical College of Nigeria (Feb. 2023); Member, National Curriculum

Committee, completed prototype curriculum on BioRisk Management (BRM), for Nigerian Universities (Jul. 2023); Organized BRM orientation workshop for University of Lagos and University College Hospital Ibadan (Jul. 2023); Participated in BRM Advanced Trainer Workshop, Lagos (May 2023), and BRM Trainers Development Workshop, Lagos (Jan. 2023) after Certification (Sep. 2022); Member, Organizing Committee for the African Organization for Research and Training in Cancer (AORTIC) Genomics Conference, Lagos, Nigeria (May 2023); Nominee, University of Lagos to participate at the National University Commission Train the Trainers Workshop in Core Curriculum and Minimum Academic Standards, Abuja (May 2023); Presented posters on 'DNA methylation-based inflammation score is associated with hepatocellular carcinoma among people living with HIV' and 'Epigenetic signatures of virus-associated cervical cancer in women living with HIV', at the AACR Annual Meeting (Florida, Apr. 2023); Presented poster on, 'Unique microbial biomarkers identified in Nigerian patients with colorectal cancer', at the ASCO Gastrointestinal Cancer Symposium (California, 2023).

35. **Adewole OO**. Lectured postgraduate medical doctors on 'Tuberculosis and Respiratory Impairment'; Lectured Pulmonary Fellows on 'Genomics of Lung Cancer' at the West African College of Physicians and National Postgraduate Medical College of Nigeria (Jan. 2023); Contributed to the Genomics curriculum of a master's degree programme in Medicine; More medical students and residents developed interest in genomics and its relevance to modern practice; Teaching of genetics was strengthened at the undergraduate level.
36. **Adeyemo TA**. Appointed, Member, Scientific Working Group (SWG), Global Action Network for Sickle Cell and Other Inherited Blood Disorders (GANSID, Apr. 2023); Panelist, Guest speaker, at the World Sickle Cell Disease (WSCD) Sickle Cell Foundation, Nigeria, delivered lecture on 'Importance of early and accurate diagnosis of Sickle cell disease' (Nigeria, Jun. 2023); Attended a course on 'Clinical Trial Research Capacity-Building', by Nottingham Clinical Trials Unit, Nottingham University (UK, Oct. 2022); Initiated and continued work on sickle cell disease registry of Nigeria (SCDRN), enrolled 300 patients (target 5000 SCD cases), for accurate clinical and patient-reported data to support clinical research, interventions, management, and quality of care of patients.
37. **Cornejo-Olivas M**. Recipient of Annual Research Publication Award, Instituto Nacional de Ciencias Neurológicas (Dec. 2022); Principal Investigator, Coordinator of the first DNA Bank in the public system in Perú; President Council for Rare and Orphan Disorders by Peruvian Ministry of Health; Member, Executive of Pan American Section of the International Parkinson and Movement Disorder Society (2021-2023), and of Organizing Committee for the 5th Pan American Parkinson's Disease and Movement Disorder Congress (May 2024); Recognized as International Excellence Ataxia Center by National Ataxia Foundation (Mar. 2023); Presented posters on 'A new risk locus on chromosome 1 is suggested by genome-wide association study in Peruvians for Alzheimer disease', 'Deciphering the genomic regulatory architecture of iPSC derived oligodendrocytes from diverse ancestries for Alzheimer´s disease studies', 'Tau and amyloid plasma biomarker analysis in Alzheimer's disease cohorts from diverse genetic ancestries', at AAIC (Jul. 2023); Faculty for six international meetings and workshops, including Huntington Online Course series supported by MDS-EHDN (Jul. 2023), Fogarty Global Health Training Course 2023 (Jun. 2023), Brazilian Congress on Neurogenetics (Mar. 2023), Neurology WHO meeting (Online, Jan. 2023); Mentored trainees (2) from Northern Pacific Global Health Research Fellows Training Consortium (FIC/NIH) and Global Brain Health Institute, University of California, San Francisco; Diagnosed cases (109) - repeat expansion disorders (60) by PCR/RTPCR genetic testing, complex neurodegenerative disorders (45) by genome sequencing

through the iHope program, Huntington's disease at risk family members (4) through our predictive testing program.

38. **Daich Varela M**. Continued PhD at University College of London, London, UK; Continued work on gene therapy and pharmacotherapy clinical trials; Awarded, ARVO Foundation Early Career Clinician-Scientist Award (Apr. 2023); Presented, poster on 'Inherited eye diseases in Argentina' and moderated a session on 'Genetics and Gene Therapy'; Coordinator of Ophthalmic Genetics Module for post-graduate degree in Ophthalmology, University of Buenos Aires, Argentina; Speaker on 'Inherited retinal disorders', in Buenos Aires (Argentina, Jul. 2023); 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); Invited speaker on 'Genetic Disorders- Counselling', at BESTCON, organized by Dept. of Medicine, Guwahati, India (Apr. 2023); Volunteer Clinical Geneticist with International Office of Migrations, United Nations, Aruba.
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; Young investigator in Global Genomic Medicine Collaborative (G2MC); Established, continued to build on the Undiagnosed Disease Program at Hospital Edgardo Rebagliati Martins, EsSalud; Continued efforts with the Registry of Congenital Malformations at HNERM (ECLAMC); Enrolled cases (130 duo/trios) in iHope, Illumina Project for identification of genetic causes of rare diseases in Perú.
41. **Dutta AK**. Promoted to Additional Professor, Dept. of Biochemistry, AIIMS Kalyani (International collaborations established during and after the Summit helped get high-impact scientific publications, crucial for career progression); Started in-patient genetics consultations, and training of Psychiatry Residents (MD) and Nursing students (B.Sc.); Summit curriculum materials helped prepare talks and modules; Developed training modules on 'Genetic Testing' for the Indian Academy of Pediatrics; Resource person for a Guest lecture on 'Basics of Medical Genetics and Selection of Appropriate Genetic Tests for Infants', for DM Neonatology Residents of IPGME&R and SSKM Hospital Kolkata (Jun. 2023); Faculty for panel discussion on 'How to Counsel?' at the Society of Fetal Medicine Bengal Chapter Annual Conference (Mar. 2023); Procured equipment for the Molecular Genetics Laboratory at AIIMS.
42. **Dwivedi A**. Initiated next generation sequencing at Army Hospital R&R, New Delhi; Used materials from Summit to educate fellow Residents, Researchers; Conducted seminars/masters class on 'Prenatal Genetics', 'Oncogenetics', 'Lysosomal Storage Disorders', 'Ocular Genetics' etc.; Presented poster on 'Spectrum of hereditary cancer syndromes from a single tertiary care centre', 'Desanto Shinawi syndrome: First report from India, success story of IL10R deficiency with very early-onset inflammatory bowel disease', at the Society of Indian Academy of Medical Genetics, SGPIMS, Lucknow (India, Dec. 2022); Member, Society for the Study of Inborn Errors of Metabolism, India; Member, Society of Fetal Medicine, India; Continued to spread awareness on genetic disorders among armed forces personnel, and families; Diagnosed patients (1800) with various genetic disorders - chromosomal, Mendelian, disorders of genomic imprinting; Knowledge from Summit helped manage these patients.

43. **Edem-Hotah J**. Appointed, Member, Validation Committee, Sierra Leone Postgraduate College of Pharmacy curriculum, Freetown (Jun. 2023); Appointed, Chairman, for validation of the Sierra Leone National Fistula Strategy and Implementation Plans (May 2023), used materials from Summit for inclusion; Appointed, Assessor, by the Tertiary Education Commission, Sierra Leone for accreditation of BS in General Nursing Curricula for University of Makeni and Njala University, included genetics and research modules (May 2023); Appointed, Member, National Obstetric Fistula Taskforce and Obstetric Fistula Technical Working Group (Oct. 2022); Keynote speaker on 'Building and strengthening global sickle cell communities, formalizing new-born screening and knowing your sickle cell disease status', at the Sickle Smart Foundation Sickle Cell Disease Commemoration day (Jun. 2023).
44. **Eshete MA**.
45. **Gitaka J**. Earlier established and continued work on Sickle cell surveillance at Mary Help of the Sick Mission Hospital in Thika, Kenya; Undertaking development of a master's degree course in Bioinformatics at Mount Kenya University; Referred patients (8) for genetic counselling and medical management; Helped allay anxiety due to known pathway to care.
46. **Lertwilaiwittaya P**. Continued Residency in Combined Internal Medicine and Medical Genetics and Genomics Residency program at University of Alabama at Birmingham, USA (2021-2025); Initiated Internal Medicine and Medical Genetics Morning Report activity at the Tinsley Harrison Internal Medicine Residency Program, University of Alabama at Birmingham (Apr. 2023); Awarded, Excellence in 'Educational Conference Engagement and Attendance', and in 'Clinical Reasoning and Medical Knowledge' by Tinsley Harrison Internal Medicine Residency Program (2023); Awarded Champion of the American College of Physicians Alabama/Mississippi Chapter 2023 Doctor's Dilemma competition, Meridian (Mississippi, Jun. 2023); Presented talk on 'The health economics of gBRCA screening in high-risk breast cancer: How gBRCA became fully reimbursable in a middle-income country', at the ACMG Annual Clinical Genetics Meeting (Utah, Mar. 2023); Participated in the first GENEius challenge hosted by ACMG (Utah, Mar. 2023); Enriched genetics experience in Cardiology Fellows.
47. **Mburu S**. Designed, introduced medical genetics and genomics in undergraduate and graduate level (MS) courses in Medical Laboratory Sciences, Health Information Management and in Medical Microbiology, at the School of Health Sciences, Kirinyaga University, Kenya, based on knowledge, skills acquired at the Summit; Delivered lectures 'Research Methods and Proposal Development', at the Dept. of Clinical Medicine, School of Health Sciences, Kirinyaga University, Kenya (Sep- Dec. 2023); Presented on 'Limited potency of adjuvant capecitabine treatment in TNBC patients with residual invasive disease', at the Kirinyaga University 6th International Conference (Kenya, Mar. 2023); Supervised, mentored graduates from Pwani University on apoptotic gene expression in breast cancer and Mt. Kenya University on health promotion approaches and hygiene practices of pupils in primary schools; Helped integrate genetics in the Dept. of Clinical Medicine (clinicians, researchers), build capacity in genomics in the county and sub-county hospitals and strengthen county health care systems.
48. **Messaoud O**. Submitted a patent on 'New Nanotechnological Process for the Electrochemical Detection of Ascorbic Acid' (TN 2022/0331, Dec. 2022); Lectured on 'Field Report on the Dougga Project: an Example of an Interdisciplinary Research', at University of Mainz, (Germany. Feb. 2023); Lectured on 'Next Generation Sequencing and Applications in Genomics' at the College of Sciences and Technologies, Nouakchott (Mauritania. Feb. 2023); Used resources from Summit for lectures on 'Techniques for Gene Investigation' and 'Techniques for Genome Investigation', at the Faculty of Medicine of Tunis, 'Genomics of the

Living World', at the Higher Institute of Biotechnology Sidi Thabet, Tunisia (Dec. 2022); Moderator, plenary session on 'The Academy of Tomorrow: Many Routes to Many Futures', at the Annual Spring Conference, The Arab-German Young Academy of Sciences and Humanities, Leopoldina- German National Academy of Sciences, Halle (Germany, Mar. 2023); Organized an International Workshop on 'Introduction of Bioinformatics to Arab Education Systems', Arab-German Young Academy of Sciences and Humanities, Institut Pasteur de Tunis (Nov. 2022); Journal Reviewer - British Journal of Dermatology.

49. **Mistri M**. Senior Scientist (Inherited Genomics & Metabolism), at the Neuberg Centre for Genomic Medicine (Gujarat, India); Started comprehensive Newborn Screening Program at the Center; Initiated, whole exome sequencing based Express Newborn Genetic Screening Program, for treatable inborn errors of metabolism; Presented poster on 'Rare case of mosaic probable germline variant in CDKL5 gene: Challenges in discussing genotype-phenotype correlation', at the 8th Annual International Conference of Board of Genetic Counseling of India (Jul. 2023).
50. **Muttamba W**. Involved in discussions with University of Cincinnati on use of stored samples obtained from a cohort of asthmatic patients in East Africa, for genomic research; Mentored, supervised, PhD candidate for research on samples in the African Severe Asthma network.
51. **Naeem MA**. Lectured courses in 'Medical Genetics' and 'Pathophysiology of Human Diseases' for MS/Mphil/PhD; Member, Organizing Committee, for the International Conference on Trends and Challenges in Health Sciences, at the Centre of Excellence in Molecular Biology (CEMB), University of the Punjab, Lahore (Pakistan, Mar. 2023); Participated in an awareness seminar on 'GM Soybean and Potential of Soybean Cultivation in Pakistan' at CEMB (Pakistan, Jan. 2023); Participated in the International Conference on Plant Molecular Biology, at CEMB (Pakistan, Dec. 2022).
52. **Nakousi-Capurro N**. Clinical Geneticist at Hospital Carlos Van Buren of Valparaíso, Chile; Faculty at Universidad de Valparaíso as Associate Professor; Incorporated as Associate Professor at the Medical Faculty at Universidad Andrés Bello in Viña del Mar; Completed online course on 'Epigenetics in Medicine', by the Medical Faculty of the University of Valencia; Designed courses, lectured medical students in 'Genetics and Clinical Genetics'; Provided on-site Clinical Genetics training to neuropediatric, pediatrics, gyn/obs Residents and medical students; Established a Neurogenetics Clinic at the Carlos Van Buren Hospital, held weekly clinical meetings for evaluation of patients; Presented newly adjudicated grant, at the regional meeting of Pediatric Endocrinologists and the National Meeting of Clinical Geneticists (Valparaíso, Jul. 2023); Presented on 'Geneticists and Neurologists: Ways to work together', organized by the Neuropediatric Unit, University of Valparaíso, Valparaíso (May 2023); Drafted, distributed national surveys to pediatric neurologists to assess 'Self-perceived knowledge in genetics and level of genetics training in specialization' and to Clinical Geneticists to assess 'Basic aspects of their work and perceived level of insufficient clinical information provided by other healthcare providers for the evaluation of patients'.
53. **Oluwarotimi AC**. National Liaison Officer of Nigerian Dental Association (2019 - to date); Elected, Member, Membership Liaison and Support Committee, Federation Dental International (since Sep. 2022); Member, Antimicrobial Stewardship, Lagos University Teaching Hospital, Idi araba, Lagos; Alternate Delegate, representing Nigeria at the World Dental Parliament Meeting (Sep. 2023).
54. **Ottaru S**. Conducted, webinar on 'Sickle Cell Disease Awareness', in collaboration with Tanzania Medical Students Association of Hubert Kairuki Memorial University and State University of Zanzibar (Jan. 2023); Trained junior doctors on diagnosis and management of

sickle cell complications; Established, continued Sickle Cell Clinic for pediatrics patients at Kitengule Hospital in Dar es Salaam, Tanzania; Evaluated patients (~5/week) with sickle cell disease in outpatient clinic; Counseled parents during visits; Used newborn screening to identify SCD and congenital heart disease (CHD); Identified newborns (5 cases/month) with CHDs, referred them to Muhimbili National Hospital for ECHO and review by a cardiologist; Helped reduce their long term complications through early identification.

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; Delivered lecture on 'Aniridia and WAGR Syndrome Management', at Refractive Plener (Ukraine, Oct. 2022); Presented on 'Gene therapy in Ukraine is a new reality for the treatment of hereditary retinal diseases', at the IV Vitreoretinal Symposium- Retina Lviv-2023 (Ukraine, Jul. 2023); Presented on 'Genetic therapy of retinitis pigmentosa. Search and don't get lost. Analysis of a clinical case by ophthalmic geneticist', at the XI Scientific and Practical Conference of Children's Ophthalmologists and Optometrists of Ukraine- One must see one's childhood (Ukraine, Jun. 2023); Presented on 'Aniridia and WAGR syndrome- ophthalmogenetic and multidisciplinary approaches', at the 2nd Interdisciplinary Symposium of Opinion Leaders of Nutrition and Dietitians on Nutritional Imbalance in Today's Realities: Should We Panic or Act? (Ukraine, Feb. 2023); Lectured on 'Keratoconus- ophthalmogenetic aspects and therapeutic approaches', at the Scientific and Interdisciplinary Conference- Practical ophthalmology, medical and environmental problems of our days (Virtual, Feb. 2023); Implemented a National Registry for Hereditary Eye Disorders, enrolled, selected patients for clinical trials in gene therapy, with counseling and follow-ups at Okhmatdyt National Children's Specialized Hospital, Ukraine; Provided ophthalmogenetics consultation at Isida-IVF (private medical center) and at the Expert Ophthalmology of Dr. Maria Znamenska, Ukraine; Introduced ophthalmogenetic consultations at Top Clinic Denis (private medical center); Supervised, identified genetic eye disorders cases (52); Performed genetic testing on hereditary eye diseases; Identified mutations for accurate diagnosis in patients (19 cases) - aniridia (3), retinitis pigmentosa (2), Usher syndrome (1), Stargardt macular dystrophy (6), macular dystrophy (1), X-linked retinitis pigmentosa (3), cone-rod dystrophy (2), enhanced S-cone syndrome (1); Provided genetic counseling.
56. **Taiwo O**. Guest lecturer on 'The Dentist - A Researcher and a Scientist. How to Conduct Research and Publish Papers', 'Fundamental Ethical Principles and Ethical Requirements for Research Studies', 'Payment, Coercion and Inducement in Research and Vulnerable Groups in Research', Design of Oral Health Surveys', **'**Common Oral Health Indices for Epidemiological Surveys', and 'Implementing Oral Health Surveys on the Field', at the Nigeria Dental Association 5 weeks Intensive Research Methodology course (Online, Jan.- Feb. 2023); 'Development of Research Titles, Questions And Hypothesis', 'Research Approach and Research Designs (Part 1)', 'Research Approach and Research Designs (Part 2)', 'Sample Size Calculation and Sampling and Variables, Measurement Scales and Data Collection', at the Nigeria Dental Association Continuing Education and Practice Committee (CEPC) 2023 Regular Paced Research Methodology CME Series (Online, Apr. - Jul. 2023); Provided an in-house capacity training workshop on 'How to Write a White Paper', 'Writing Papers for Scientific Publications' for Xcene Research – a Clinical Research Organization, Nigeria (Online, Jun. 2023).
57. **Utumatwishima JN**. Received scholarship from Swedish International Development Agency for PhD, at Sahlgrenska Academy, University of Gothenburg, Sweden to 'Investigate the

Association Between Maternal Mental Health and Undernutrition of Children in the Northern Province of Rwanda' (since Oct. 2021); Appointed, Minister of Youth, in Rwanda.

58. **Wangi KYW**. Pursuing PhD (3rd year) at Penn State University, USA (Jul. 2023); Attended courses on 'Genetics Law', at Pennsylvania State University (Fall 2023); Presented on 'Social Issue, Stigma, and Human Dignity in the Thalassemia Community in Indonesia', at a seminar on 'Screening, Prevention, and Health Issues in Thalassemia' (Virtual, May 2023); Submitted abstract on 'Reproductive rights for women with thalassemia dependent transfusion: A qualitative study in West-Java, Indonesia', to the International Society of Nurses in Genetics (ISONG), Congress (Rhode-Island, Nov. 2023).