ADDENDUM

ISHGG SIXTH and FINAL ANNUAL OUTCOME (October 2023- September 2024)
(*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 2016, 2017 and 2018 Batches completed their 5-year follow-up reports in years 2021, 2022 and 2023 respectively, hence, this compilation only includes the reports from the last Batch- 2019.*

A. Publications: (Total- 98; 2019- 98)

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 Oct 25. doi: 10.1002/jgc4.1804.
2. Abdel-Hamid M, Hassib N, Elhossini R, **Sayed I**, Aglan M. COL1-related overlap disorder: An emerging phenotype linked to mono- and bi-allelic COL1A1/2 variants. Calcif Tissue Int. 2024. (Submitted).
3. Abdel-Kader MA, NEMB Ahmed, **Sayed ISM**. Implant-supported oral rehabilitation in patients with ectodermal dysplasia. 2024. (In preparation).
4. Abdel-Kader MA, NEMB Ahmed, **Sayed ISM**. Ridge augmentation in patients with ectodermal dysplasia using a regenerative approach. 2024. (In preparation).
5. Abdelhai A, Gurashi A, Mohamed HA, Babiker H, Ali KM, Nasir EA, **Alamin TA**. Prevalence of Factor XIII polymorphisms among Sudanese sickle cell disease patients. 2024. (In preparation).
6. Abdellatif A, Sayedahmed AA, Ibrahim HA, Mohammed RA, Osman SF, **Alamin TA**. The influence of *L1CAM* gene polymorphism in chronic myeloid leukemia in Sudanese patients. 2024. (In preparation).
7. Abdul Aziz NS, **Hussein N**, Ng WL. The views and experiences of primary care doctors on prenatal genetic screening test and counselling in Malaysia. 2024. (In preparation).
8. Adejoh J, Madu C, Egua MO, **Alli LA**, Raheem D and Okoh MP. Molecular and epigenetics mechanisms for the immune control of Plasmodium parasites infection: A comprehensive review. Asian J. Biol. Sci. 2023 Dec 31; 16(4):636-669.
9. 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).
10. Ahmed HA, El-Kamah GY, Rabie E, Mostafa MI, Abouzaid MR, Hassib NF, Amr KS, **Sayed ISM**. Targeted mutational analysis prior to WES for genetic diagnosis of non-X-linked ectodermal dysplasias in Egyptian patients. 2024. (In preparation).
11. Alebiosu CO, Ayodele OE, Akinbodewa AA, **Okunola OO**. Chapter on 'Autosomal Dominant Polycystic Disease'. In: Concise Textbook of Medicine. Obafemi Awolowo University Press, ISBN 978-978-136-131-9. 2024; pp. 336-339.
12. Altunoglu U, Palencia-Campos A, Güneş N, Turgut GT, Nevado J, Lapunzina P, Valencia M, …, **Güven Y**, …, Ruiz-Pérez V. Variant characterization and clinical profile in a large cohort of patients with Ellis-van Creveld syndrome and a family with Weyers acrofacial dysostosis. J Med Genet. 2024 Jun 20; 61(7):633-644.

13. **Amarakoon G**, Rao S. A child with ADA2 deficiency- VAIH syndrome. 2024. (In preparation).
14. **Amarakoon G**, Suriapperuma T, Umasunthart. A structured home-based egg ladder can reduce the demand for hospital baked egg challenge in John Radcliffe Hospital, OUH. Arch Dis Child. 2024. (Submitted).
15. **Amarakoon G**, Wijayarathne K, Rathnasiri KMSS, Wijesundara MR, Ranathunga PU, Wijesinghe AHNP. Congenital malformation patterns in newborn: An insight from a tertiary care centre in Sri Lanka. Arch Dis Child. 2024. (Submitted).
16. **Amarakoon GGGT**, Wijerathne NPKP, Chandrakumara WAJC, Wijayarathne HK. Clinical profile of Down syndrome in a tertiary care centre -Sri Lanka. 2024. (In preparation).
17. **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. 2024. (In preparation).
18. Babalola YO, **Monye HI**, Oluleye TS, Majekodunmi OI, Ijaduola MA. Evolution in the trend of vitreoretinal diseases post sub-specialty training in an African subpopulation. 2024. (In preparation).
19. Bărar AA, Pralea IE, Maslyennikov Y, Munteanu R, Berindan-Neagoe I, **Pîrlog R**, Rusu I, Nutu A, ..., Kacso IM. Minimal change disease: Pathogenetic insights from glomerular proteomics. Int J Mol Sci. 2024 May 21; 25(11):5613.
20. Baru O, Pop L, Raduly L, Bica C, Mehterov N, **Pirlog R**, Baduru S, Braicu C, ..., Badea M. The evaluation of a 5-miRNA panel in patients with periodontitis disease. JDR Clin Trans Res. 2024 May 31:23800844241252395.
21. Baynam G, Hartman AL, 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. 2024 Jul; 12(7):e1192-e1199.
22. **Bocoum A**, Ouologuem M, Cissé L, Essop F, Coulibaly SP, Botha N, Cissé CAK, Maiga AB, …, H3Africa Consortium. The first case of Huntington's disease like 2 in Mali, West Africa. Tremor Other Hyperkinet Mov (NY). 2024 Apr 2; 14:15.
23. Chan SC, Patrick Engksan J, Jeevajothi Nathan J, Sekhon JK, **Hussein N**, Suhaimi A, Hanafi NS, Pang YK, Mohamad Yatim S, et al. Developing a home-based pulmonary rehabilitation program for patients with chronic respiratory diseases in Malaysia: A mixed-method feasibility study. J Glob Health. 2023 Oct 27; 13:04099.
24. Değirmenci T (PI), **Yesilcinar İ**. Pregnant women's genetic literacy and affecting factors. 2024. (In preparation).
25. Delgado-Vega AM, Cederroth H, Taylan F, Ekholm K, Marlene Ek, …, Buske OJ, **Wiafe SA**, Cederroth M, Nordgren A, et al. The first undiagnosed hackathon - Pushing the boundaries for undiagnosed rare diseases. Nat Genet. 2024. (Submitted).
26. Ejaz S, Raashid A, Hameed Y, Imtiaz N, Khurshid M, **Iqbal M**, Khurshid M, Malik WN. A report of novel inactivating BRCA1 missense mutations detected in acute myeloid leukemia patients. J Med Case Rep. 2024. (Submitted).
27. Elhossini RM, **Sayed IM**, Hellal US, Mahmoud SAM, Aglan MS, Hassib NF, Abdel-Hamid MS. A recurrent KCNK4 variant in a dominant pedigree with hypertrichosis and gingival fibromatosis syndrome: Variable phenotypic expressivity and insights on patients' dental management. Am J Med Genet A. 2024 Jan; 194(1):39-45.
28. Felipe-Dimog EB, **Tumulak M-AJR**, Dacquigan ELA, Padilla JA. Coping strategies of Filipino mothers of children with congenital heart disease in a tertiary hospital in the

Philippines. Acta Med Philipp. 2024 Apr 12. https://actamedicaphilippina.upm.edu.ph/index.php/acta/article/view/9043.
29. 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 Oct 25. https://actamedicaphilippina.upm.edu.ph/index.php/acta/article/view/6880.
30. Felipe-Dimog EB, **Tumulak MJ**, Yu CH, Liang FW. Dedication of professional midwives in providing healthcare services in selected rural communities in the Philippines: A descriptive qualitative study. Phil J of Sci 2024 Feb; 153(1):469-478.
31. Galván-Valencia O, Sanders AP, Ariza AC, Burris HH, **Ortiz-Panozo E**, Svensson K, Mercado-García A, Téllez-Rojo MM, Wright RO, et al. Associations of salivary aldosterone levels during pregnancy with maternal blood pressure and birth weight-for-gestational age in a Mexico City birth cohort. J Perinatol. 2024 May; 44(5):643-649.
32. Gargano MA, Matentzoglu N, Coleman B, Addo-Lartey EB, Anagnostopoulos AV, Anderton J, …, **Wiafe SA**, Wiggins LD, Williams AE, et al. The human phenotype ontology in 2024: Phenotypes around the world. Nucleic Acids Res. 2024 Jan 5; 52(D1): D1333-D1346.
33. **Gyawali P**, Shrestha B, Quinonez S. Medical genetics need assessment in Nepal: An online cross-sectional survey. 2024. (In preparation).
34. **Gyawali P**, Shrestha R, Karmacharya RM, Baidya N, Sharma S. Holo-transcobalamin deficiency among type 2 diabetic patients under metformin therapy. 2024. (In preparation).
35. Hartman AL, Monaco L, Letinturier MCV, Aketa N, Athanassiou D, …, McMaster C, Parker S, Scherman D, **Wiafe SA**, et al. The complexity of funding rare disease research: An IRDiRC assessment of the landscape. Rare Disease and Orphan Drugs Journal, RDOD. 2024 (Submitted).
36. Hassan MO, Aurogundade FA, Osasan SA, Gbadegesin BA, Omotoso BA, **Okunola OO**. Clinicopathologic study of sickle cell-associated kidney disease: A Nigerian Experience. Niger Postgrad Med J. 2024 Jan 1; 31(1):53 -61.
37. **Hussein N**, Chun L, Cheah W, Fong CS, Abdullah A, Ng WL, Lee PY, Teo CH, Liew CS. The challenges in accessing air quality data from a middle-income country for the development of a street scale-resolution air quality forecast system using the Atmospheric Dispersion Modelling System (ADMS) urban model. 2024. (In preparation).
38. **Hussein N**, Hanafi NS. Chapter on 'Community Engagement to Improve Community Health'. In: The Handbook of Public Health in the Asia-Pacific. Publisher Springer Nature. 2024. (In preparation).
39. **Hussein N**, Lee YK, Mohd Reza S, John P, Azmi A, Ng CJ. Uncovering the experiences and insights of thalassemia carriers - A qualitative study in a rural community in Sabah, Malaysia. Aust J Prim Health. 2024. (Submitted).
40. **Hussein N**, Ng CW, Ramli R, Liew SM, Hanafi NS, Lee PY, Cheong AT, Ghazali SS, 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 Serv Res. 2024 Mar 12; 24(1):327.
41. Isachesku E, Braicu C, **Pirlog R**, Kocijancic A, Busuioc C, Pruteanu LL, Pandey DP, Berindan-Neagoe I. The role of non-coding RNAs in epigenetic dysregulation in glioblastoma development. Int J Mol Sci. 2023 Nov 14; 24(22):16320.
42. Ismail I, Ng CJ, Lee PY, **Hussein N**. Exploring the determinants of family planning practices among women with diabetes with no pregnancy intention: The women's voice. Korean J Fam Med, KJFM. 2024. (Submitted).

43. 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. 2024. (In preparation).
44. **Kars ME**, Stein D, Stenson PD, Cooper DN, Gelb B, ItanY. Deciphering the digenic architecture of congenital heart disease using trio exome sequencing data. 2024. (In preparation).
45. **Kars ME**, Wu Y, Stenson PD, Cooper DN, Burisch J, Peter I, Itan Y. The landscape of rare genetic variation associated with inflammatory bowel disease and Parkinson's disease comorbidity. Genome Med. 2024 May 14; 16(1):66.
46. Lim ZN, Liew SM, Khoo EM, Pinnock H, McCarthy S, Pang YK, Hanafi NS, **Hussein N**, Abu Bakar AI. Exploring the disease experience and supportive care for people with very severe chronic obstructive pulmonary disease in Malaysia: A multi-perspective qualitative study. 2024. (In preparation).
47. **LLamos-Paneque A**, Liehr Thomas, Echavarria-Frutos I, León-Sosa A. Duchenne muscular dystrophy in a man with Klinefelter syndrome: A rare association. OBM Genetics. 2024. (Submitted).
48. **Long LH**, Fujioka T, Craig TJ, Hitomi H. Long term outcome of C1-esterase inhibitor deficiency. Asian Pac J Allergy Immunol. 2024 Jun 15. doi: 10.12932/AP-220224-1792.
49. **López-Star B**, Pérez- Serrano R. Identification of rs7677751 polymorphism in PDGFRA gene, in patients with corneal astigmatism. 2024. (In preparation).
50. **López-Star B**. Review of ophthalmogenetics literature in Mexico. 2024. (In preparation).
51. Maceda EBG, Abadingo ME, Asor BBN, Cases RKC, Supan RJ, Anarna KS, Libo-on PCA, Vesagas TDC, **Tumulak M-AJR**. Social media content analysis of public and private glucose-6-phosphate dehydrogenase (G6PD) deficiency Facebook Groups. Acta Med Philipp. 2024 Jan 29. https://actamedicaphilippina.upm.edu.ph/index.php/acta/article/view/8737.
52. Magdy P, Mohamed AM, Mostafa MI, Abdel Latif SH, **Sayed ISM**, Amr K, El-kamah G. Exploring the impact of utilizing different approaches for feature selection on the identification of crucial features in phenotype classification: An investigation conducted on a cohort of individuals affected with ectodermal dysplasia. 2024. (In preparation).
53. Makura A, **Mhandire K**, McCarty K, Mutsvangwa J, Nhidza A. Human Immunodeficiency Virus Drug Resistance (HIVDR) and baseline characteristics among Antiretroviral Therapy (ART) experienced children and adolescents under the care of Chidamoyo Christian Hospital in Hurungwe, Zimbabwe. J Clin Res in HIV AIDS and Prev 4. 2023 Dec 12; 4(2):32-42.
54. Malka S, Biswas P, Berry AM, Sangermano R, Ullah M, …, **Iqbal M**, …, **Naeem MA**, Akram SJ, Akram J, et al. Substitution of a single non-coding nucleotide upstream of TMEM216 is a common cause of non-syndromic retinitis pigmentosa and is associated with reduced TMEM216 expression. Am J Hum Genet. 2024. (Submitted).
55. Marwah V, Kumar N, Choudhary R, **Mutreja D**, Sharma A. A rare case of hemophagocytic lymphohistiocytosis associated with sarcoidosis. J Adv Lung Health. 2023 Sep-Dec; 3(3):125-127.
56. 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. J Glob Health. 2024. (Submitted).
57. Mitsunami M, Wang S, Soria-Contreras DC, Mínguez-Alarcón L, **Ortiz-Panozo E,** Stuart JJ, Souter I, Rich-Edwards JW, Chavarro JE. Pre-pregnancy plant-based diets and risk of hypertensive disorders of pregnancy. Am J Obstet Gynecol. 2024 Mar; 230(3): 366.e1-366.e19.

58. **Nair LS**, Jain JMN, Dalal A, Ranganath P. Etiologic spectrum of pediatric onset leukodystrophies and genetic leukoencephalopathies: The five-year experience of a tertiary care center in Southern India. Pediatr Neurol. 2024 Mar; 152:130 -152.
59. **Nair LS**. Chapter on 'Orthopedic Management and Genetic Interventions in NMI. Part B: Newer Therapies in Neuromuscular Disorders'. In: Neurodisabilities Rehabilitation Therapy. Noble Vision (Medical Book Publishers), ISBN 978-81-957204-9-1. 2023; pp. 391- 395.
60. Nathan JJ, Ramakrishnan L, Abdullah A, Fong CS, **Hussein N**. Policy measures and their effectiveness in addressing air pollution in Malaysia: a scoping review. 2024. (In preparation).
61. Ng WL, **Hussein N**, Ng CJ, Qureshi N, Lee YK, Kwan Z, Kee BP, Then SM, Abdul Malik TF, et al. Implementing HLA-B*58:01 testing prior to allopurinol initiation in Malaysian primary care setting: A qualitative study from doctors' and patients' perspectives. PLoS One. 2024 Jan 11; 19(1):e0296498.
62. 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. Singapore Med J. 2024. (Submitted).
63. Odunlami GJ, Ajibade A, Omotoso BA, Hassan MO, Adefidipe AA, Olanrewaju FO, Enitan AO, Adetunji TA,…**Okunola OO**. Clinical and laboratory profile of systemic lupus erythematosus patient in a rheumatology clinic in southwestern Nigeria. The experience so far. Rheumatologia. 2024 May; 62(2):83-93.
64. 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. 2024. (Submitted).
65. Okunade KS, Adebola AA, Olumodeji AM, Olowe A, Oyedeji OA, Ademuyiwa IY, Adelabu H, Toks-Omage E,…**Akinmola OO**, et al. Prenatal anemia and risk of postpartum hemorrhage: a cohort analysis from the Predict-PPH study. BMC Public Health 2024; 24:1028.
66. Okunade KS, **Akinmola OO**. Impact of obesity on survival outcomes of women with advanced epithelial ovarian cancer in Lagos, Nigeria. Ecancer. 2024. (Submitted).
67. 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. 2024. (In preparation).
68. Ooi NZM, **Hussein N**, Abdul Malik TF. Health seeking behavior for menopausal symptoms: a qualitative study in primary care clinics. J Menopausal Med, JMM. 2024. (Submitted).
69. **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. 2024. (In preparation).
70. Osman OM, Sidahmed RA, Mohammed JS, **Alamin TA**. Detection of *L1CAM* gene mutations in Sudanese sickle cell anemia patients. 2024. (In preparation).
71. Padilla-González E, Orzua-de la Fuente WM, Quezada-Sanchez AD, Doubova SV, **Ortiz-Panozo E**. Social determinants of hypertensive disorders of pregnancy trends in Mexican municipalities. Pregnancy Hypertens. 2024 Mar; 35:55-60.
72. **Paredes-Moscosso SR**, Nathwani A. 10 years of BiTE Immunotherapy: An overview with a focus on pancreatic cancer. Frontiers in Oncology. 2024. (Submitted).
73. **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. 2024. (In preparation).

74. Potdar O, Suresh I, Raja V**, Yadav S**. Diagnostic dilemmas in fetal scalp cystic lesions- A case series. J Fetal Med. 2024. (Submitted).
75. Pozo-Palacios J, **Llamos-Paneque A**, Rivas Iglesias CR, Onofre Perez EJ. Experiences in the molecular diagnoses of Huntington disease in Ecuador. 2024. (In preparation).
76. Quispe B, **Paredes-Moscosso SR**, Villegas-Llerena C. Retinitis pigmentosa: Gene therapy approaches. Rev Peru Med Exp Salud Publica. 2024. (Submitted).
77. Ramdzan SN, Khoo EM, Cunningham S, **Hussein N**, Ramli R, Senawi SA, Sukri N, Nathan JJ, Kassim A, …, RESPIRE collaborators. Qualitative research influencing guideline and policy: An exemplar of the development of a national school asthma guideline in Malaysia. J Glob Health. 2024 May 17; (14):03027.
78. Ramírez-Silva I, Ariza AC, Barragán-Vázquez S, Mendoza Jiménez M, **Ortiz-Panozo E**, Batis C, Burguete-García A, Ávila-Jimenez L, Zambrano E, et al Longitudinal patterns of breastfeeding and its association with adiposity and subjective indicators of satiety/appetite in the first 2 years of life. Appetite. 2023 Nov 1; 190:107030.
79. Rehman N, Kumar MK, Nikunj V, Manikandan A, **Mutreja D**. An intriguing case of diffuse sclerosing variant of papillary thyroid carcinoma: A Case Report. Asian Oncology Research Journal, AORJ. 2024 Jul 13; 7(1):22-27.
80. Romitan M, Zanoaga O, Budisan L, Jurj A, Raduly L, Pop L, **Pirlog R**, ..., Brerindan-Neagoe, I. MicroRNAs expression profile in chemotherapy-induced cardiotoxicity in non-small cell lung cancer using a co-culture model. Biomol Biomed. 2024 Jan 3; 24(1):125-137.
81. Rondon-Jara E, Chiriboga Morales X, Enríquez ME, Bernales M, López-Ríos JM, Guarderas P, Flórez-Cuadros M, Daza MA, …, **Paredes-Moscosso SR**. Leticia Declaration: Collaborative leadership for a sustainable CTI system with social impact in Latin America and the Caribbean. Zenodo. 2024 May 2. doi :10.5281/zenodo.11104916.
82. Saraswat A, **Mutreja D**, Singh A, Raut V. Choroidal Pseudomelanoma: Haemorrhagic retinal detachment masquerading as choroidal melanoma. IOSR Journal of Dental & Medical Sciences. 2023 Nov; 22(11):40-42.
83. Sarfo J O, Doe-Yo Tawiah E, Hammond CK, **Wiafe SA**. "I Just Cry My Heart Out"- Coping strategies of mothers living with children with spinal muscular atrophy. BMC Womens Health. 2024. (Submitted).
84. Sidharth KS, Rupesh R, Amard S, Sadaf A, **Khant AK**, Manoji M, Kalaivani G, Shraddha L, Sinha R, et al. Assessment of South Asia Paediatric Acute kidney Injury: Risk factors and Epidemilogy (ASPIRE): A prospective study on 'Severe dialysis dependent paediatric AKI', Pediatr. Nephrol. 2024; Mar 08. doi:10.1007/s00467-024-06324-6.
85. Simone B, Sciascia S, Carta C, Salvatore M, …, **Wiafe SA**, Bodamer O, Posada M, Taruscio D et al. A global survey about Undiagnosed rare diseases: Perspectives, challenges, and solutions. Lancet Public Health. 2024. (Submitted).
86. Stein D, **Kars ME**, Wu Y, Sevim Bayrak C, Stenson PD, Cooper DN, Schlessinger A, Itan Y. Genome-wide prediction of pathogenic gain-and loss-of-function variants from ensemble learning of a diverse feature set. Genome Med. 2023; 15(1):103.
87. Suriapperuma T, **Amarakoon G**, Umasunthar T. Oral food challenge outcomes in children and adolescents at Oxford University Hospitals NHS Foundation Trust: A 2-year retrospective review. Arch Dis Child. 2024. (Submitted).
88. Tahir U, Barber J, Cruz D, **Kars ME**, Deng S, Tuftin B, Gillman M, Benson MD, Robbins JM, et al. Leveraging diverse ancestries in the integration of genetics and proteomics to inform clinical disease biology. JCI. 2024. (Submitted).

89. **Tumulak MJ**, Padilla CD, Ongchangco JC, Laurino MY, Lagarde JB, Regalado ES, Legaspi AV, Ventura EV. Living with a child with MSUD: Psychosocial issues of Filipino parents with a child with maple syrup urine disease. Gen in Med Open. 2024 Apr 13. doi.org/10.1016/j.gimo.2024.101847.
90. 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. 2024. (In preparation).
91. Umair M, **Iqbal M**, Mustafa G, Bhinder MA, Shah MZ, Khan PR, Ejaz A, Baig HMA. A novel missense mutation (c.1982A>C) FCHSD1 gene causes autosomal recessive early onset Parkinson's disease in a consanguineous Pakistani family. J Popul Ther Clin Pharmacol. 2024 May 8; 31(5):329-337.
92. Wang S, Mitsunami M, **Ortiz-Panozo E**, Leung CW, Manson JE, Rich-Edwards JW, Chavarro JE. Pre-pregnancy healthy lifestyle and adverse pregnancy outcomes. Obstet Gynecol. 2023 Dec 1; 142(6):1278-1290.
93. **Yadav S**, Kabra M, Gupta N, RC Mahumita, Chauhan S, Puri RD, Phadke S, Jana M. Isolated lateralized overgrowth – Phenotypic spectrum and molecular alteration at 11p15.5 locus. Indian J Pediatr, IJP. 2024. (Submitted).
94. **Yesilcinar İ**, Bektaş Pardes B, Güvenç G. Development of the health belief model scale for prenatal genetic screening and diagnostic tests. 2024. (In preparation).
95. **Yesilcinar İ**, Şahin E, Tutar SO. The decisional conflict and the genetic literacy of pregnant women regarding and prenatal screening tests. J Genet Couns. 2024. (Accepted).
96. **Yesilcinar İ**, Şahin E. Determination of genetic literacy and genetic knowledge levels of nursing students. 2024. (In preparation).
97. Zhen Xin, Betti MJ, **Kars ME**, Patterson A, Medina-Torres EA, Mendoza SCS, Sánchez DAH, Lopez-Herrera G, Svyryd Y, et al. Molecular and clinical characterization of a founder mutation causing G6PC3 deficiency. medRxiv. 2024 May 14. doi: 10.1101/2024.05.13.24307299.
98. Zimta AA, Cenariu D, Tigu AB, Moldovan C, Jurj A, **Pirlog R**, Pop C, Gurzau ES, …, Berindan-Neagoe I. Differential effect of the duration of exposure on the carcinogenicity of cadmium in MCF10A mammary epithelial cells. Food Chem Toxicol. 2024 Apr; 186:114523.

B. Grants: (Total- 14; 2019- 14)

2019 Batch

1. **Abubakar S**, Koehly L, Bishop R. Training and learning on- Families Sharing Health Assessments and Risk Evaluations (SHARE) – Nigeria Project. 2024. (NIH Mini Sabbatical Grant; Received).
2. **Alli LA** (PI), Ajiboye TA, Kyari F, Okoh MP, Aliyu NO. Experimental and computational validation of Ginkgo biloba leaves extract on high intraocular pressure reduction in Wistar rats for glaucoma intervention. 2024. (University of Abuja, Nigeria; Accepted).
3. Bender A, Berindan-Neagoe I, **Pirlog R**. Lung squamous cell carcinoma therapeutic targets using systems level machine learning based on single-cell RNA-sequencing. 2023. (Romania's National Recovery and Resilience Plan- PNRR-III-C9-2022 – I8; Received).
4. Campbell H (PI), Fernandes G, Khoo EM, **Hussein N** (Co-I), Abdullah A, Nathan JJ. Identifying policy priorities for tobacco control and improving air quality (IPPTA):

Stakeholder engagement in seven countries. 2023-2024. (National Institute of Health and Care Research, United Kingdom; Received).

5. Florez-Cuadros M (PI), **Paredes-Moscosso SR** (Co-I), Bethancourt D, Rondon E, Arboleda J, Vargas F, Eunice M, Becerra-Hernández L, Balboa L, et al. One Health network for science-informed public policies. 2024. (Cyted- Ibero-American Program for Science and Technology for Development; Submitted).
6. Hisham R (PI), **Hussein N** (Co-I), Saupi Udin Z. Rural primary care physicians' information needs and seeking behaviour in Sabah, East Malaysia. 2024-2025. (Faculty of Arts and Social Sciences Early Career Researcher Grant, Universiti Malaya; Accepted).
7. 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). 2023-2026. (National Institute of Health and Care Research, United Kingdom; Received).
8. Miño S (PI), Perez-Guedes A, Cevallos-Gil B, Pantovic B, Garcia-Cienfuegos B, Moscote M, **Paredes-Moscosso SR** (Co-I), Barraza C, Rodrigues C, et al. Ibero-American science diplomacy network for climate action. 2024. (Cyted; Submitted).
9. Miño S (PI), Perez-Guedes A, Gochez A, Rodrigues M, Florez-Cuadros M, Pantovic B, **Paredes-Moscosso SR** (Co-I), Figueroa P, Barraza C, et al. 2024. Empowering communities for climate resilience: Food sovereignty as a catalyst for socioeconomic and gender justice. 2024. (International Science Council Pilot Missions for Sustainability; Submitted).
10. Mohsen Atroosh WM (PI), Lau YL, Bee PC, **Hussein N** (Co-I), Sarhan Almekhlafi HM. Elucidating Plasmodium knowlesi malaria parasite preference to β-thalassemic erythrocytes in Malaysia. 2024-2026. (Fundamental Research Grant Scheme, Ministry of Higher Education, Malaysia; Accepted).
11. Netea R, Berindan-Neagoe I, **Pirlog R**. Decoding the immuno-inflammatory axis in rare non-medullary thyroid cancer as an innovative approach for novel combinatory therapeutic approaches. 2023. (Romania's National Recovery and Resilience Plan- PNRR-III-C9-2022 – I8; Received).
12. **Okunola OO** (PI). Heritability and phenotypic characteristics in diabetic nephropathy. 2024. (International Society of Nephrology; Re-submitted).
13. **Ortiz-Panozo E**, Chavarro JE (PI). Maternal polycystic syndrome and offspring's growth trajectories and epigenetic changes in early childhood. 2024. (NICHD/NIH; In preparation).
14. Young B, (PI), Everett L, **Monye HI** (Co-I). Choroideraemia atrophy progression kinetics in ultra-widefield fundus autofluorescence images. 2024. (Randy Wheelock Research Award – Submitted).

C. Collaborations (NIH and Other Institutions):

NIH: (Total- 3; 2019- 3)

2019 Batch

1. **Abubakar S**, Koehly L. Assessment of a family health history taking tool, for improving family health history & genomic literacy in Nigeria. A pilot feasibility study. 2023. (Bayero University Kano/Aminu Kano Teaching Hospital, Nigeria; NHGRI/NIH; Amended protocol for IRB-approved).

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).

Other Institutions: (Total- 28; 2019- 28)

2019 Batch

1. **Abubakar S**. Trained staff of the Kano State Contributory Healthcare Management Agency on Healthcare Financing. 2024. (Bayero University Kano/Aminu Kano Teaching Hospital; Adamawa State Ministry of Health; Adamawa State Contributory Healthcare Management Agency, Nigeria).
2. **Chowdhury EH**, **Amarakoon GGGT**, **Mosema KBA**, **Iqbal M**. Study of complications of iron overload in thalassemia patients in Bhutan, Sri Lanka, Democratic Republic of Congo, Myanmar 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. 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; Division of Endocrinology, Diabetes and Bone Disease, Icahn School of Medicine at Mount Sinai, USA).
5. Fakoya AOJ, **Akinmola OO**. An attempt to make bio-pacemaker by CRISPR/CAS9 gene editing of human mesenchymal stem cells. (Lagos University Teaching Hospital, Nigeria; Louisiana State University Health, 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. **Hussein N**, 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).

9. **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).
10. **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).
11. **Iqbal M**, **Naeem MA**. Identification, clinical evaluation, and genetic analysis of RP families (Dept. of Biotechnology, The Islamia University of Bahawalpur, Pakistan; National Centre of Excellence in Molecular Biology, University of the Punjab, Pakistan).
12. **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, USA).
13. Kantaputra PN, **Guven Y**. Consultation on genetic cases. (Istanbul University Faculty of Dentistry, Turkey; Chiang Mai University Faculty of Dentistry, Thailand).
14. **Llamos-Paneque A**, Aguilar VR, Diaz Manera J, Töpf A, Gonzalez-Chamorro A. Latin-SEQ Project. (International Centre for Life: San Francisco University of Quito, Ecuador; John Walton Muscular Dystrophy Research Centre, Institute of Translational and Clinical Research, Newcastle University, UK).
15. **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).
16. **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).
17. **Llamos-Paneque A**, Hagerman R, Villarreal J, Tassone F. 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).
18. **Llamos-Paneque A**, Liehr T. Genetic study of marker chromosome. (Genetics Service of Hospital No.1 FF. AA. Quito, Ecuador; Jena University Hospital, Institute of Human Genetics, Germany).
19. **Llamos-Paneque A**, Slavotinek A. 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, OH, USA).
20. **Luong LH**, Hoang LT, Craig JT: Diagnosis, consultation and creating registry of hereditary angioedema patients in Vietnam. (National E Hospital, Vietnam; Pennsylvania State University, USA).
21. Martinez-Barricarte R, Itan Y, **Kars ME**. Investigating the role of ARPC1B variation in immunodeficiency. (Icahn School of Medicine at Mount Sinai, USA; Vanderbilt University, USA).
22. **Nair LS**, Nair MKC, Kumar S, Lukose R. Genetic evaluation of children with intellectual disability and autistic spectrum disorders. (Dept. of Medical Genetics; NIMS Spectrum-Child Development Research Center, NIMS Medicity, Trivandrum, Kerala, India).
23. **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, India).

24. Olawoye O, Olusanya B, Sarimiye T, **Monye HI**. Effect of direct selective laser trabeculoplasty in reducing intraocular pressure in open angle glaucoma: An interrupted time series trial. Data Analysis. (University of Ibadan, Nigeria; Eleta Eye Institute, Ibadan, Nigeria).
25. **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).
26. Ramdzan SN, Satav A, Kartasasmita C, Khan F, Mahmood H, Hanafi NS, **Hussein N**, Schwarze J, Khoo EM, Pinnock H. Adapting school-based Asthma Programme: a mulTicountry (AdAPT) study. (Universiti Malaya, Malaysia; Mahatma Gandhi Tribal Hospital (MAHAN) Trust, India; Universitas Padjadjaran, Bandung, Indonesia; Aga Khan University, Pakistan; Fasiuddin Khan Research Foundation, Bangladesh; University of Edinburgh, UK).
27. 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).
28. 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).

D. New Research Projects: (Total- 12; 2019- 12)

2019 Batch

1. 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.
2. **Amarakoon G**, Wijayarathne K. Complex clinical presentation and multidisciplinary management of Mowat-Wilson Syndrome: A case report.
3. **Chowdhury EH**, Dorji N, Lhendup K, Zangmo Y, Tobgay S, Phuntsho N. Clinical and genetic association studies of children with mucopolysaccharidosis in Bhutan.
4. Ekpe LE, **Akinmola OO**. Correlating the diagnostic accuracy of imaging and histological findings in the diagnosis of non-alcoholic fatty liver disease among patients in a tertiary hospital.
5. **Gyawali P**, Quinonez S. An assessment of newborn screening readiness in Nepal.
6. **Kars ME**, Stein D, Itan Y. The phenomic landscape of gain- and loss-of-function genetic variants across diverse human populations.
7. **Llamos-Paneque A**, Romero-Aguilar V. Analysis of the frequencies of molecular variants in patients with cystic fibrosis treated in private clinic in Quito, Ecuador 2023-2024.
8. **Luong LH**, Hitomi H, Craig TJ. Expand the natural history and long-term clinical effect of hereditary angioedema patients with focus on novel aspects- neurological effect/myopathies-developed secondary to C1-INH deficiency, utilizing the newly developed disease-specific iPS cell lines.
9. **Mhandire K**, Gadbery J. Developing polygenic risk scores for detection of neurodevelopmental disorders – beyond GWAS. (Pending ethical approval).

10. **Mushi T**, Kubhoja S, Tarimo T, Lyimo F, Janab M. Epidemiology of parasitic infection causing cardiovascular diseases. A collaborative multidisciplinary approach for risk stratification, diagnosis and treatment.
11. Okunade KS, **Akinmola OO**. Diagnostic validity of HPV E6/E7 mRNA in HIV infected women with genital high risk HPV infections.
12. Villegas-Llerena C, **Paredes-Moscosso SR**, Yucra A, Cruz LA, Gutierrez G, Corredor F, Facundes FG. Transcriptomic analysis of fur to identify markers of genetic improvement in alpaca fiber.

E. Ongoing Research Projects: (Total- 40; 2019- 40)

2019 Batch

1. 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).
2. **Amarakoon G**, Rao S. Integrating artificial intelligence in undergraduate clinical education: Enhancing teaching and learning outcomes. (Knowledge from Summit helped literature review).
3. 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. (Knowledge from Summit helped perform analyses).
4. Chagonda S, **Mhandire K**, Musarurwa C. High throughput sequencing towards optimization of African population specific cardiovascular gene panels. Approved by IRB, sample collection underway in Zimbabwe. (Knowledge of cardiovascular disease panel sequencing gained at the Summit helped guide concept and methods).
5. 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).
6. **Chowdhury EH**, Lhendup K, Zangmo Y, Tobgay S, Phuntsho N. Incidence and prevalence of congenital hypothyroidism in Bhutan. (Knowledge from Summit improved research quality).
7. Fagbemi OY, Okunade KS, Oluwole A, **Akinmola OO**. Evaluation of urinary soluble endoglin as a biomarker for pre-eclampsia at the Lagos University Teaching Hospital (Knowledge from Summit on cell free DNA of fetus in maternal circulation helped in conceptualizing this project).
8. **Gyawali P**, Quinonez S. Current practice, and perspective for enhancing genetic services: An exploratory mixed-method study at Dhulikhel Hospital.
9. Hassib N, Abdel-Fattah M, **Sayed I**, **Mehrez M**, Ismail S, Abdel-Ghafar S, Elhossini R, Hellal U, Gamal El Din H. Diagnosis of inherited disorders with oro-dental anomalies using recent advanced genetic approaches and impact on counseling and management (Summit lectures improved skills in diagnosis of rare genetic disorders).
10. **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. (Knowledge, skills, networking at the Summit built confidence to lead, develop interventions to improve practice in primary care setting).
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 and results).
13. **Kars ME**, Bao M, Gettler K, Cho J, Itan Y. Phenome-wide association studies of therapeutic targets of inflammatory bowel disease. (Knowledge from Summit helped to perform analyses).
14. **Kars ME**, Gelb B, ItanY. Evaluation of genetic determinants of congenital heart disease using trio sequencing data. (Knowledge from Summit helped to perform analyses).
15. **Kars ME**, Itan Y. Genetic determinants and molecular mechanisms of obesity and cardiovascular disorders (Knowledge from Summit helped to perform analyses).
16. Lingao MD, Artiaga JCM, Espiritu DNV, Cabrera P, Collantes ER, Uy HS, **Tumulak** MJR. Genotypic and phenotypic variations of inherited retinal degeneration among Filipinos. (Summit lectures on proposal/grant writing helped write project proposal).
17. **Llamos -Paneque A**, Guapi, V, Guayasamin P, Nieto, B**.** Experiences in cytogenetic prenatal diagnosis at the Maternal-Fetal Unit, Luz Arizmendi Maternity: Genetics Service of Hospital No.1 FF. AA, Quito-Ecuador (2020-2024). (Knowledge from Summit in cytogenetics, prenatal diagnosis helped improved diagnosis).
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. 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. Submitted amendment to Ethics Board for development of immortal cell lines. (Summit lectures on Bioethics and Variant Interpretation helped develop human subject consent protocols).
22. **Luong LH**, Hitomi H. Generation and verification of EPO-GFP reporter iPS cell line for accurate monitoring of erythropoietin production in vitro and in vivo. (Knowledge of gene editing, genetic reprogramming acquired at the Summit helped in discussion and completion of project).
23. **Luong LH**, Hitomi H. Generation of human induced pluripotent stem cell (hiPSC) from patients with hereditary andioedema disorders to study disease mechanism and potential therapy. (Knowledge from Summit helped initiate discussions for this project).
24. **Mosema KBA**, 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).
25. **Mushi T**, Kabirigi J, Tulla M, Chami N, Kayange N, Adolfine AH. Prevalence, factors associated with persistent pulmonary hypertension and diagnostic utility of differential oxygen saturation among newborn babies in Mwanza, Tanzania. (Summit provided key knowledge on newborn screening, use of pulse oximeter- a recommended tool for identification of critical congenital heart diseases and persistent pulmonary hypertension in newborns).

26. 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).
27. **Okunola OO**, Adekoya AA, Ojo EA, Adeoti PA, Adeyemi AA. Genetics study in autosomal dominant polycystic kidney disease in the tropics. (Used Summit resources to counsel families on genetic testing).
28. **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 for patient counselling, new options for therapies focused on gene editing).
29. 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).
30. 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. (Summit increased awareness, interest in ocular genetic diseases such as JOAG prevelant in Nigeria).
31. **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. 2022-2024. (Summit lectures inspired and informed approach for project).
32. 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).
33. **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 and reporting of the results).
34. **Sayed I**, Gamal El Din HM, **Mehrez M**, Abdel-Salam G, Ismail S, Abdel-Ghafar S, Hassib N, Magdy P, AbuZaid M, et al. Advances in the delineation of genetic etiology of oro-dental anomalies in neurogenetic disorders (Summit lectures improved skills in diagnosis of rare genetic disorders).
35. 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).
36. **Tumulak MJR**, Maceda EB, Cases RK, Asor BB, Calalo-Magbanua C, **Abad PJ**. A content analysis of Facebook groups on congenital adrenal hyperplasia. 2023. (Summit lectures on proposal/grant writing helped write project proposal).
37. 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).
38. Vilain E, Linguraru M, Mumba-Ngoyi, Tshala-Katumbay D, Bramble MS, **Mosema KBA**, Spencer D. Mobile diagnosis of congenital genetic conditions: A model for screening and surveillance in low-resource settings. Phase II- innovative exploratory and development research. (Summit lectures on Telemedicine helped advance the project).
39. 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).

40. 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).

F. Other Accomplishments/Initiatives:

2019 Batch

1. **Abubakar S**. Submitted memo to Leadership/Management Committee, Aminu Kano Teaching Hospital for the establishment of the Rural and School Health (RuSH Lab) at Kumbotso LGA, Kano, Nigeria- awaiting approval (Jun. 2024); Lectured on health advocacy in the course on 'Health Advocacy & Policy' for MPH- Global Health, at College of Health Sciences and the World Bank Funded Africa Center of Excellence for Population Health and Policy, Bayero University, Kano, Nigeria (Apr. 2024); Appointed as Sabbatical Staff in Public Health (Feb. 2024), lectured at the Department of Community Medicine, College of Medical Sciences, Federal University, Dutse/Jigawa, Nigeria; Appointed as a Member of the Health Research Ethics Committee by Aminu Kano Teaching Hospital, Kano Nigeria (Jun. 2024); Appointed as Faculty, at the Vanderbilt University funded Masters in Research Ethics by Bayero University Kano (Apr. 2024).
2. **Akinmola OO**. Established a Genetic Counselling Unit in the Tertiary Hospital, Lagos University Teaching Hospital with consent from Management; Planned workshops to increase awareness and uptake of hereditary and genetic based diagnosis in different specialties; Awarded, Virtual Summit Scholarship by Wellcome Connecting Science to attend events - Human Immunology- Genes and Environment (May 2024); Antimicrobial Resistance- Genomes, Big Data and Emerging Technologies (Mar. 2024), Mitochondrial Medicine- Therapeutic Development (Mar. 2024), Epigenomics of Common Disease (Nov. 2023); Certified in AAI Clinical Trial Regulation and Ethics digital course by BIO Ventures for Global Health (BVGH, Feb. - May 2024); 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 (since 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.
3. **Alamin Alabid T**. Served as a tutor for an online learning platform Learna | Diploma MSc (diploma-msc.com) for post-graduate students enrolled in haematology, coordinated activities, scored students.
4. **Alli LA**. Promoted to Professor of Medical Biochemistry, University of Abuja (Oct. 2023); Re-appointed, Visiting Professor, Medical Biochemistry and Molecular Biology for Preclinical Medical students, Nile University, Abuja Nigeria (Jul. 2024); Appointed External Examiner, Medical Biochemistry, Federal University of Health Sciences, Azare, Bauchi state, Nigeria (May 2024); Appointed Visiting Professor of Medical Biochemistry, IBB University Lapai, Niger state, Nigeria (Jan. 2024); Supervised and mentored postgraduate research students (2 PhD, 4 MSc); Used materials from Summit to lecture medical students on 'Basic Molecular Biology', 'Genetic Disorders', 'Inborn Errors of Metabolism'; Strengthened postgraduate Biochemistry curriculum in Genetics and Genomics for biochemistry medical 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; Submitted abstracts on 'Evaluation of home-based milk ladder introduction for children under 24 months in a pediatric allergy clinic', 'Epidemiological and clinical characteristics of pediatric patients with spontaneous urticaria/ angioedema at John Radcliffe Hospital' to the The British Society of Allergy and Clinical Immunology (BSACI) Annual Conference (UK, Oct. 2024); Presented posters on 'Anaphylactic reaction due to storage mites - pancake syndrome', 'Case series on pollen food syndrome', 'Structured dietary management of egg allergy in padiatrics: Insights from a case study', 'Home based milk ladder (MAP) in the management of IgE mediated cow's milk protein allergy – A case report', at the South Asian Pediatric Association & Sri Lanka College of Pediatricians International Congress (Jun. 2024); Presented posters on 'A structured home-based egg ladder can reduce the demand for hospital baked egg challenge in John Radcliffe Hospital, OUH', 'Oral food challenge outcomes in children and adolescents at Oxford University Hospitals NHS Foundation Trust: A 2 year retrospective review', 'Congenital malformation patterns in newborn : An insight from a tertiary care center in Sri Lanka' at the RCPCH Conference (Mar. 2024); Presented posters 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);); Identified, diagnosed and treated genetic cases (18) – syndromes (9), hemoglobinopathies (4), Inborn errors of metabolism (5).
6. **Baatar N**. Continued to develop hospital information system (HIS) at the national level by adopting an OpenEHR framework, using SNOMED CT, HL7FHIR, and openEHR; Attended, A Decade Toward Healthcare Excellence (Mongolia, Jun. 2024); Attended, The 4th International Congress of Organ Transplantation (Mongolia, May 2024); Organized, attended, presented posters on 'Glucokinase gene sequencing in Mongolian patients' and 'Study of polymorphism of *HNF1A, HNF4A* genes in diabetes', at Digital Advanced Solutions for Healthcare Industry-2024 (Mongolia, Apr. 2024); Attended, She Loves Tech 2023 Global Startup Competition, Mongolia Round (Oct. 2023); Attended, Promoting and Upscaling Digital Solutions for Enhancing Women Enterprise Growth and Resilience Program, held by UN ESCAP (Mongolia, Oct. 2023).
7. **Bocoum A**. Presented talk on 'Clinical and Eiological Aspects of Movements Disorders', at the 4th Congress of African Academy of Neurology (Senegal, Jul. 2024).
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 every semester (3 months) on 'Updates in Inborn Error of Metabolism in Children', CME program at SGH; Identified and managed pediatric genetic disorders (12) - Down's syndrome (5), Turner's syndrome (2), Thalassemia (2), congenital hypothyroidism (1), Marfan's syndrome (1), hemophilia (1); Referred cases for physiotherapy (4), cardiac surgery (1), ophthalmic surgery (1); 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 (24) - enamel renal gingival syndrome (1), papillon lefevre (1), ectodermal dysplasia (6), dentinogenesis imperfecta (1), amelogenesis imperfecta (3), isolated oligodontia cases (12); Ectodermal dysplasia patients were referred to the genetics department for molecular confirmation and medical referral; Dental prosthodontic and restorative therapies, preventive and follow-up protocols, early diagnosis and surgical interventions contributed to social and psychological well-being of children, prevented fatal consequences and improved quality of life for children and their families; Section Editor, Orphanet Journal of Rare Diseases.
10. **Gyawali P**. Assistant Professor, Kathmandu University School of Medical Sciences, Nepal (since Jan. 2023); Awarded, Josip Matovinovic Endowed Clinical Medicine Fellowship, University of Michigan (Dec. 2023 - Nov. 2024); Completed a capacity building training program on implementation of point of care testing, financed by the Higher Education Institution Capacity Development Program- University Grant Commission (2023).
11. **Hussein N**. Featured in Top Universiti Malaya Researcher Highlight – Top 10% Web of Science indexed journals (May. 2024); Member, Cochrane Collaboration, United Kingdom (2023-2028); Member, Academic Committee of Medical Humanities and Ethics Unit, Faculty of Medicine, Universiti Malaya, Malaysia (2022-2024); Programme Coordinator, Postgraduate Family Medicine Specialty, Universiti Malaya, Malaysia (2023-2024); Committee Member, National Family Medicine Specialty (2023-2024); Committee Member for Teleconsultation in clinics (2023-2024); Postgraduate Examiner for Master of Family Medicine (Nov. 2023, Feb. 2024); Appointed Coordinator, External Assessor, Chinese University of Hong Kong, for Master of Family Medicine Programme, Universiti Malaya (Jan. 2024); Project Reviewer for - Consensus on the National Standards for Palliative Care: A Delphi Study (May 2024) and Joint development of eLearning training modules on evidence-based medicine (JET EBM) for primary care physicians in Singapore and Malaysia (SingHealth Polyclinics, Singapore; University of Malaya, Malaysia, Apr. 2024); Participant, Postgraduate Curriculum Workshop and e-portfolio (Mar. 2024); Author, 'Malaysia Guidelines for Early Detection and Management of Students with Asthma Symptoms in Schools' (2023-2024); On Committee for translation of book on 'Testing Treatments – Better Research for Better Healthcare' (English to Malay), a Cochrane Group initiative (2024-2025); Presented on 'Developing and Evaluating a Mobile Phone-based Early Alert System Using High-Resolution Air Quality Forecast To Improve Asthma Control In Malaysia' and 'Optimising The Preparation Of A Site Visit For A Multi-Country Research Study: A Strategic Approach', at the 12th International Primary Care Respiratory Group World Conference (May 2024); Speaker on 'Why Family Matters – Family History Revisit' at Faculty of Medicine UMHealth Webinar (Apr. 2024); Speaker, facilitator for workshops on 'National Policies in Malaysia Air Quality' (May 2024), 'Introduction To Research In Clinical Practice' (Apr. 2024), 'Genetic Screening In Primary Care' and 'Psychological Issues In Genetic Testing' (Jan. 2024), 'Developing An Asthma Registry' (Jan. 2024), 'Exercise In Elderly' (Dec. 2023), Practice Diary Calibration Workshop for Postgraduate Examiners (Nov. 2023); Grant Reviewer for 'Development And Feasibility Of An Asthma Web-based Training Program For Malaysian Educators', Universiti Malaya Research Excellence Grant (UMREG) (Dec. 2023); Mentored PhD candidate on Air Quality and Respiratory Health; Supervised Family Medicine trainees in developing a research proposal 'Motivational Interviewing On Obesity Among Doctors In Primary Care'; Audited postgraduate curriculum, increased awareness, strengthened Family Medicine training through emphasis on family history, instilled skills in community genetics, promoted genetic screening in primary care; Conceptualized, initiated discussion with State Health Department Thalassemia Committee on developing research capacity to improve

implementation of cascade screening in primary care settings in Sabah, Malaysia, using the Stanford design thinking framework.

12. **Iqbal M**. Delivered lectures on 'Advanced Molecular Genetics And Classical Genetics' to graduate & undergraduate students; Submitted abstract on 'Biallelic variants in CAPN10 cause microcephaly, intellectual disability and developmental delay' to ASHG (USA, 2024); Reviewed Dossiers for promotion to Associate Professor for University of Hafr Al Batin, KSA and IUB, Pakistan; Participated on Board of Studies at different universities in Pakistan; Appointed, External Examiner, reviewed PhD (3) and MS (10) theses; Supervised graduate research students (5 PhD, 7 MS) on molecular and genetic basis of different hereditary disorders.
13. **Kars ME**. Postdoctoral Fellow speaker on 'Investigating IBD-PD Comorbidity, IBD Therapeutic Targets And Monogenic IBD Genes Through EHR And Sequencing Data Of Diverse Populations', at the IBD Conference (New York, Jun. 2024); Presented posters on 'Therapeutic targets for inflammatory bowel disease: A PheWAS approach' and 'Investigating the genetic determinants of the comorbidity between inflammatory bowel disease and Parkinson's disease', at the Digestive Disease Week (Washington DC, May 2024); Presented poster on 'Application of network-based heterogeneity clustering for investigation of genotype-phenotype correlations in BioMe BioBank', at ASHG (Washington DC, Nov. 2023); Speaker on 'Investigating The Genetic Underpinnings Of Cardiometabolic Traits Through Phenome-Wide Association Studies', at the 2nd Annual Leducq Meeting (NY, Oct. 2023); Journal Reviewer - Frontiers in Genetics.
14. **Khant AK**. Senior Consultant Pediatrician, Pediatric Nephrology Dept., Yangon Children Hospital; Attended, Joint 5th Primer in Paediatric Nephrology for Asia and 5th IPNA-AsPNA Master for Junior Classes CME Interactive Course, Singapore (Aug. 2024); Lectured undergraduate medical students, post graduate pediatric students, at University of Medicine 1, 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; Identified, diagnosed, treated pediatric cases (450) in the emergency department - acute diarrhea (120), acute viral infection (170), dengue hemorrhagic fever (60), acute respiratory tract infection (65), pyrexia of unknown origin (25), syndromes (10); renal replacement therapy for acute kidney injury (25), chronic kidney disease children (5); Counselled patients with hereditary renal disease (10), congenital nephrotic syndrome (1), congenital abnormalities of kidney urinary tract (10), meningomyelocele with neurogenic bladder (4), syndromic child with renal abnormalities (3), Lowe syndrome with renal tubular acidosis (1), Prune Belly syndrome (1), Caroli syndrome (1), focal segmental glomerulosclerosis (7) and several others.
15. **LLamos-Paneque A**. Professor, Biology & Human Genetics (undergraduate level), School of Dentistry, International University of Ecuador and Technion: Israel Institute of Technology; Updated topics in rare diseases in Human Genetics such as TTr-Amyloidosis, Epigenetics; 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; Initiated work with Dr. Lina Ghaloul Gonzalez, Division of Genetic and Genomic Medicine, UPMC Children's Hospital of Pittsburgh, for diet and crisis management of glutaric aciduria and methylmalonic acidemia patients (5 cases), diagnosed in Ecuador, resulting in integration of nutritionists and pediatricians for training in inborn errors of metabolism; Attended Online courses on 'Cytogenomics Today: Link Between Diagnosis and Research', by Mexican Society of Human Genetics (Jun. 2024); 'Oncogenetics' and 'Precision

Medicine' sponsored by Mexican Society of Human Genetics (virtual, Jan. 2024); Attended events to improve skills in genomic field, particularly in rare diseases- II Rare Disease Expert Forum, sponsored by Stendal, S.A. (Mexico, May 2024), 'Grand Launch of the Treatment for Patients with hATTR', at Cartagena de Indias, by PTC Therapeutics S.A. (Mar. 2024); Summit improved genetic counseling skills, implemented this at a private clinic's Maternal Fetal Unit (~70 cases/yr); Evaluated cases (400)- mono gene aetiology (45) confirmed by molecular genetics, chromosome (58) confirmed by cytogenetics or multifactorial (190); Registered new cases (293) in the Registry of Genetic Diseases, Specialty Hospital No.1 FF. AA, Quito, Ecuador; Continued register cases with the Ecuadorian Duchenne Muscular Dystrophy Registry.

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 (~2 patients/ month); Hired two ophthalmologists- ophthalmo-pediatrician (1) and ophthalmo-oncologist; Emphasized on genetic testing; Member, Pan American Inherited Retinal Diseases Group (since Feb. 2020); Trained junior staff in ophthalmo-genetics; Continued to record, monitor cases (141) with inherited retinal disorders; Evaluated, diagnosed, treated ophthalmo-genetic cases (23)- congenital cataract (6), congenital glaucoma (6), retinitis pigmentosa (11); 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 on family planning.
17. **Luong LH**. Affiliated with National E Hospital, Vietnam (2022); PhD candidate in the International Postgraduate Program- iPS Cell Regenerative Medicine, at Kansai Medical University, Japan (since Sep. 2022).
18. **Mhandire K**. Appointed, Genetic Variant Scientist- Review Analyst, GeneDx, MD, US (since Feb. 2022); Led a mechanism for disease curation- curated 14 loss of function, 5 gain of function genetic disease mechanisms for diagnostic use; Used knowledge and experience gained from the Summit, including ACMG variant interpretation, NGS applications, whole exome/genome sequencing, and networking for lectures and training; Lectured NIH Laboratory Genetics and Genomics Fellowship trainees on 'Introduction to ACMG Variant Interpretation and HGVS Nomenclature', 'Targeted Familial Testing, and 'Use of Population Databases in Variant Interpretation'; Attended the UK Biobank Scientific Conference (virtual, Dec. 2023) and IDWeek Conference (MA, Oct. 2023); Trained Scientists (3) in neurodevelopmental disorders and whole exome sequence variant interpretation; Analyzed and reported cases in whole exome sequencing (>1500), NGS panel sequencing (>800), targeted familial (>300).
19. **Monye HI**. Appointed to a post-doctoral role by the Eyes of Africa – Genetics of Blindness Study Team (2024); Attended, digital course on 'Basics of Clinical Trial Management', by BIO Ventures for Global Health (virtual, Jul. 2024 - ongoing); Continued sensitization of ophthalmologists, trainees on evaluation of genetic eye diseases with knowledge, skills and resources gained at Summit; Initiated, developed a database for patients with retinitis pigmentosa at the Tulsi Chanrai Foundation Eye Hospital, Abuja, Nigeria; Enrolled 23 patients.
20. **Mosema KBA**. Lectured on 'Genetics' and 'Pediatrics' to medical students at Université de l'Uele, Isiro, Haut-Uele Province; Attended workshop on 'Data Science for Diverse Scholars in Down Syndrome' (DS3), organized by INCLUDE DCC, University of Colorado, Boulder (USA, Jul. 2024); Presented talk on 'Systematic Screening Of Sickle Cell Disease In Children And Family Centered Care In The Biamba Marie Mutombo Hospital', at the 1st Meeting of the

Congolese Sickle Cell Society, Kinshasa, (DRC, Jun. 2024); Presented posters on 'Diagnosing rare conditions in low-resource settings: The experience of the differences of sex development clinic in the Democratic Republic of the Congo', 'Feasibility of low-cost genetic newborn screening with AI-based tool in a limited resource setting', at ASHG Annual Meeting (Nov. 2023); 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 cases (2) and advised care for (3) Down's Syndrome patients.

21. **Mushi TL**. Completed Masters in Medicine in Pediatrics and Child Health (Sep. 2023); Trained colleagues in newborn screening and role of genetics and genomics in precision medicine; Initiated a Pulmonary Hypertension Registry at the Jakaya Kikwete Cardiac Institute, Tanzania- diagnosis based on clinical features, physical examination and echocardiography; Established, continued to enroll in a Birth Defect Registry at Bugando Medical Centre, Tanzania; About 1731 were born with congenital heart disease(s), ventricular septal defect (463, 35.5%), atrial septal defect (162, 12.45%), tetralogy Fallot (232, 17.8%), coartation of aorta, hypoplastic left heart syndrome (4, 0.3%), truncus arteriosus (27, 2.8%), transposition of great vessels (54, 4.01%), and others (13, 0.75%); Down syndrome (76, 5.85%), Edward' syndrome (1, 0.078%), Turner syndrome (1, 0.07%) and others (3, 0.17%), acquired heart disease (432, 25.2%), rheumatic heart disease (152, 35.1%), cardiomyopathies (59, 13.6%), myocarditis (101, 5.8%), Kawasaki disease (2, 0.46%) and others (82, 18.9%); Offered counselling as part of routine management due to unavailability of a geneticist or genetics counselor; Medically managed congenital heart disorders - hemodynamic instability with diuretics, furosemide, and spironolactone, ventricular septal and atrial septal closure defects, tetralogy of Fallot with surgical interventions.
22. **Mutreja D**. Submitted report as PI of Armed Forces Medical Research Committee on projects titled 'A Pilot Study to Evaluate the Role of Vascular Endothelial Growth Factor and its Comparison with Microvessel Density in Angiogenesis of Hematological Malignancies' and 'Flow Cytometric Detection of Minimal Residual Disease in Cases of B-Acute Lymphoblastic Leukemia'; Delivered lectures on genomics to faculty/colleagues, post graduate residents at Command Hospital Air Force Bangalore, India; Shared resource materials from Summit with postgraduates from other specialties; Lectured on 'Gene Editing & CRISPR Cas 9 Technology' at AFCON-PATH, Army Hospital Research & Referral New Delhi (India, Nov. 2023); Presented talk on 'Clinicopathologic Correlation' as pathology discussant at Medical Education & Research Trust, Association of Physicians of India CME, API Bhavana, Bangalore (India, Oct.-Nov. 2023); Section Editor (Pathology) for Indian Journal of Medical Paediatric Oncology (2021-23); Identified cases (6) by cytogenetics and histopathologic /microscopic diagnosis- thalassemia (4), sickle cell disease (2).
23. **Nair L**. Conducted weekly webinars for pediatricians, including developmental and neurological pediatricians, and counseling students on 'Genetic Disorders', 'Genetic Tests and Their Clinical Implications'; Mentored, lectured Genetic Counsellor students on 'Basic Genetics'; Conducted OPD, evaluated, diagnosed patients with various genetic disorders, provided genetic testing and counselling; Identified cases- Down syndrome (3), unexplained intellectual disability/ developmental delay (27), recurrent pregnancy loss/ infertility (18); Conducted prenatal molecular genetic evaluation of 7 pregnant women, identified pathogenic variant 22q11.1 tetrasomy in one fetus; Evaluated, diagnosed cases with rare genetic disorders -15q11.2q13.3 tetrasomy (1), Xp22.2 monosomy (1), 22q11.21 trisomy (1), blended phenotype of 4p16.3 deletion (Wolf Hirschhorn syndrome) and 8p23.3 trisomy (1).

24. **Okunola OO**. Used experience and materials from Summit to train, mentor medical students, Residents, colleagues in medical genetics and genomics, research grant applications; Lectured post graduate and undergraduate medical students on hypertensive renal disease, genetics of ADPKD and APOLI gene in Blacks; Liaison on working group with pediatric nephrologists, established congenital kidney disease registry with follow up; Developed innovative model for diabetic renal clinics to limit morbidity and retard progression of disease by addressing co-morbidities (ophthalmological, cardiovascular, chiropody, limb salvage) in patients and their relatives; Diagnosed patients (27) with inherited kidney diseases (all autosomal dominant polycystic kidney disease, ADPKD), others (22) with congenital anomalies of the kidney and urinary tract (CAKUT)- ureteropelvic junction obstruction (12), renal agenesis (5) and horse shoe kidneys (5); 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, passed the preliminary qualifying examination (May 2024).
26. **Paredes-Moscosso, SR**. Accepted, speaker on 'Bridging Science To The World: Science Communication Principles And Methodologies For Impactful And Well Structured Messages' at CILAC 2024 (Colombia, Dec. 2024); Awardee for course on 'Organoids: Advances And Applications', by Wellcome Trust, UK (Online, Sep. 2024); Speaker on 'In Vitro Modeling By CRISPR/Cas9 Of A VUS Mutation Found In Peruvian Patients With Hereditary Breast Cancer', at Sinapsis 2024, by Heinrich Heine Universität Düsseldorf (Germany, Jul. 2024) and Universidad Católica San Pablo (Perú, Jul. 2024); Peer reviewer, BMC Cancer (Jun. 2024) and AAAS-TWAS Science Diplomacy Center (Feb. 2024).
27. **Pirlog R**. Started Fellowship in Molecular Pathology at CHU de Nice, Nice, France, supported by a Giordano Scholarship awarded by the European Society of Pathology; Elected member, Steering Committee of the Young Cancer Professionals Group, European Cancer Organization.
28. **Sayed I**. Associate Professor of Oro-dental Genetics; Presented talk on 'ED Experience - Clinical, Molecular And Dental Care', and poster on 'Clinical overlap between *NECTIN1* and *NECTIN4* Related Ectodermal Dysplasia', at the 1st MENA Region and African International EB Congress (Egypt, Apr. 2024); Identified cases (32)- neurogenetic manifestation (9), skeletal manifestation (15), ectodermal dysplasia patients (8); Referred patients (6) for prosthetic management of ectodermal dysplasia; Supervised, mentored a PhD student.
29. **Tiong SY**. Elected Committee Member, Genetic Counselling Society Malaysia (GCSM, 2023-2024); Lectured on 'Inherited Metabolic Disorders (IMD) Newborn Screening' for an Antenatal Class and clinic, to increase awareness among parents with follow up at delivery (May 2024); Continued to implement mandatory newborn screening for 33 types of inherited metabolic disorders at Loh Guan Lye Specialists Centre, along with genetic counselling service; Uptake of IMD new born screening increased to 98% (Oct. 2023- Jun. 2024) from 56% (Oct. 2022- Jun. 2023); Received 37 new referrals for genetic counselling (Oct. 2023-Jun. 2024).
30. **Tumulak MJR**. Promoted to Research Assistant Professor I (2023); Speaker on 'A Content Analysis Of Facebook Groups On Congenital Adrenal Hyperplasia' at the 21st Newborn Screening Convention in Manila, Philippines (Oct. 2023) and presented poster at the Human Genetics in Asia Conference in Tokyo, Japan (Oct. 2023); Organized workshop on Genetic Counseling on Hemoglobin Disorders to healthcare providers in Manila (Philippines, Apr., Jun. 2024); Thesis panel member for genetic counseling students (10 students); Provided clinical interpretations on alpha and beta thalassemia mutation results (500); Provided pre- and post-test

genetic counseling to patients – breast cancer patients (10), cystic fibrosis patients (10), alpha and beta thalassemia patients (600), fatty acid disorders patients (70), and IRD study participants (30).

31. **Wiafe SA**. Appointed as a member of the Global Commission to End Diagnostic Odyssey in Children Living with a Rare Disease (Apr. 2024); Speaker on 'Challenges and Lessons Learned In Implementing Undiagnosed Disease Program in Ghana' (S. Korea, Sept. 2024); Attended the 2nd Undiagnosed Disease Hackathon (Netherlands, Jun. 2024); Submitted abstract on 'Challenges in the diagnosis and management of children with spinal muscular atrophy in Ghana: A five-year retrospective review', to the 18th International Child Neurology Conference (S. Africa, May 2024); Attended the Sanford/PPALS PACT Course (USA, May 2024); Attended the Global Skin African Regional Meeting (Kenya, Apr. 2024); Attended the Global SMAdvocacy (Belguim, Mar. 2024); Attended RareX2024 (S. Africa, Feb. 2024); Organized a workshop for pediatricians on genetics and rare diseases at the Annual General Meeting of the Pediatric Society of Ghana (Ghana, Feb. 2024); Attended the World Orphan Drug Congress Europe 2023 and the RDI Membership meeting (Spain, Oct. 2023); Speaker on 'Report On The Developing Nations Working Group' and 'Champions Initiative' at the 12th UDNI Conference, (Georgia, Oct. 2023); Speaker on 'Report From The Taskforce On Impact' at the Consortium Assembly meeting and the IRDIRC Conference (Canada, Oct. 2023); Enrolled 220 patients unto the Congenital Anomaly and Rare Disease Registry; Enrolled 115 cases unto the Undiagnosed Disease Program; Diagnosed 20 cases through the Undiagnosed Disease Program - Sandhoff disease (2), and 1 each of Pitt-Hopkins syndrome, mucopolysaccharidosis type II, mucopolysaccharidosis type VI , familial isolated hypoparathyroidism, Noonan syndrome, POLR3-related leukodystrophy, ataxia-telangiectasia, glycogen storage disease type III, GRIN1-related neurodevelopmental disorder, megalencephalic leukoencephalopathy with subcortical cysts, nemaline myopathy, spinal muscular atrophy, Gaucher disease, Wilson disease, 8q23.3-Q24.22 deletion, GM1 gangliosidosis, spinocerebellar ataxia Type 7, FOXP1 syndrome.
32. **Yadav S**. Clinical Geneticist at Faith Diagnostic and Fetal Centre (FDFC), Mohali, India; Started Fetal Medicine Fellowship program at FDFC; Completed Observership (30 days) in Oncogenetics at Tata Cancer Institute (ACTREC), Mumbai (Jan. 2024); Started new Genetics Clinic at FDFC, emphasis on comprehensive genetic testing, counseling, family screening for high risk cancer patients; Guest lecturer on 'Genetic Tests – When, Where and Why?', at FOGSI-FDMSEC National Conference (India, Jun. 2024); Presented poster on 'Molecular alterations at 11p15.5 locus in cases of isolated hemihyperplasia – Series of 32 cases', at the National Conference of Medical Genetics (India, Dec. 2023); Debated on 'Utility of Universal Extended Carrier Screening In India', at the 2nd Annual Symposium on Genetic Disease, New Delhi (India, Nov. 2023); Moderator, 3rd South Asia Gaucher Summit on 'Recent Updates in Gaucher's Disease', discussed recent advances in neurological manifestations specifically in skeletal phenotypes (virtual, Oct. 2023); Used materials and knowledge from Summit to create awareness on genetic disorders among clinicians; Trained Fetal Medicine and Genetics Fellows (4); Performed antenatal invasive fetal procedures – amniocentesis (10) , CVS (2); Involved in preconception and antenatal genetic counselling of patients - carrier of beta thalassemia (8 couples), NIPS screen positive (7), previous baby with cystic fibrosis (1), screen positive on antenatal screening (18), couple carrier screening for previous neonatal deaths, babies with neonatal death (3), spinal muscle atrophy (2), Kabuki syndrome (1); Multiple congenital malformations (12) were diagnosed using antenatal advanced 2D/3D ultrasonography and fetal

ECHO, including jugular lymph sacs (1), cystic hygroma(1) Binder's phenotype (2), CAKUT (2), club feet (2), skeletal dysplasia (2), cardiac defects like DORV (1), rhabdomyoma (1).

33. **Yesilcinar I**. Secured a new position as Associate Professor, at Community Department, College of Nursing & Health Sciences, UMass Dartmouth- experiences and collaboration opportunities from the Summit in prenatal health helped secure this position (Aug. 2024).