ISHGG FOURTH ANNUAL OUTCOME (October 2020- September 2021)

(To avoid a lengthy report, we have tried to mention a project under one category only eg. a publication can also be a grant, a collaboration, and an on-going project; however, it may not appear in the other three categories). The Covid-19 pandemic affected the work of some of our Summiteers.

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

2019 Batch


52. Monye HI, Olawoye O, Ugalahi MO, Onebunne EO, Oluleye TS. Genetic eye diseases and services - awareness, knowledge and perceptions of adult patients of the University college Hospital, Ibadan. 2021. (In preparation).


**2018 Batch**


182. Wangi KYW. Health Professions in the era of precision medicine. Online newspaper: Kompasiana; May 2021.


2017 Batch


199. Cordeiro BQS, Carvalho WR, Fonseca EM, Martins CCP, Moreno MD, Quinelato V, Casado PL. Vertical discrepancy in height of Morse Cone pillars submitted to different torque forces. Materials. 2021. (Accepted).


2016 Batch


256. Borda V, Alvim I, Mendes M, Silva-Carvalho C, Soares-Souza GB, Leal TP, Furlan V, Scliar MO, Guio H. The genetic structure and adaptation of Andean highlanders and Amazonians are influenced by the interplay between geography and culture. Proc Natl Acad Sci USA. 2020; 117(51):32557-32565


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

2019 Batch

1. Al Hajjar N (PI), Berindan Neagoe I, Pîrlorg R. Increasing the capacity, competitiveness and visibility, research of UMF Iuliu Hațieganu by forming and supporting multidisciplinary teams in research excellence. 2021. (Iuliu Hatieganu University of Medicine and Pharmacy, Received).


3. Berindan Neagoe I (PI), Pîrlorg R (Co-PI). Development and support of the medical entrepreneurial culture among UMF Cluj-Napoca students through dissemination, training and mentoring. 2021. (Iuliu Hatieganu University of Medicine and Pharmacy, Received).

4. Chianumba R (PI), Alli LA. Patterns of liver function parameters among patients living with sickle cell disease on hydroxyurea therapy. 2021. (Institutional Research Fund grant; Submitted).

5. Gyawali P (PI), Quinonez S. Medical genetics need assessment in Nepal using a mobile health application. 2021. (Global Genomic Medicine Collaborative Seed Award; In preparation).

6. Hussein N (PI), Lee YK, Ng CJ, Ramli R, John P, Jafry N. Developing research capacity to improve the implementation of thalassaemia screening in Sabah, Malaysia using the Stanford design thinking framework. 2021. (University Malaya Specialist Centre (UMSC) Care Fund; Submitted).

7. Hussein N (PI), Lee YK, Ng CJ, Ramli R, John P, Jafry N. Developing research capacity to improve the implementation of thalassaemia screening in Sabah, Malaysia using the Stanford design thinking framework. 2021. (University Malaya Specialist Centre (UMSC) Care Fund; Submitted).


9. Iqbal M (PI). Genetic analysis of retinitis pigmentosa in consanguineous families from southern Punjab, Pakistan. 2021. (Research Grant, ORIC, The Islamia University of Bahawalpur Pakistan; Received).

11. Nyamjav D, Osgonbaatar T, Tumenbayar A, Baatar N, Delgerduuren M, Alimaa A. Primary care approach in Adolescents. 2021. (Health Promotion Fund, MOH, Mongolia; Received).

12. Oo MTZ (PI), Khant AK. Fractional excretion of magnesium in children with nephrotic syndrome. 2020. (Department of Medical Research, Lower Myanmar Yangon; Received).


14. Adewole OO (PI). Finding people with tuberculosis through strategic deployment of a non-sputum based (Sweat TB Test) in Nigeria. Grant challenges Africa Phase II. 2021. (African Academy of Science; Received).

15. Adewole OO (PI), Arun V (Co-PI). Statins as treatment adjunct in active pulmonary tuberculosis. 2020. (Cure Within Reach; Received).

16. Afolabi BB (PI), Galadanci H (Co-PI), Balogun M (Co-PI), Adeyemo TA (Co-PI), Sam-Agudu N (Co-PI). Intravenous versus oral iron for iron deficiency anemia in pregnant Nigerian women (IVON): an open label, randomized controlled trial. Clinical Research Protocol. 2020-2023. (Bill and Melinda Gates Foundation; Received).

17. Alatise O (PI), Kingham P (Co-PI), Abdulkareem F (Co-I). Nigerian cancer research training programme. 2021. (OAK Foundation, Switzerland; Received).


22. Karina Milla-Neyra et al. Cornejo-Olivas M (co-investigator). Redefining the genetics of spinocerebellar ataxia type 10 in admixed and Amerindian population from Peru. 2020. (FONDECYT/CONCYTEC Peru; Received).

23. Kingham P (PI), Alatise O (Co-PI) Abdulkareem FB (Co-I). The efficacy and feasibility of fecal immunochemistry for colorectal cancer screening in Nigeria. 2020. (Prevent Cancer Foundation; Received).

24. Kingham P (PI), Olusegun a (Co-PI), Abdulkareem FB (Site-PI). Expanding cancer research capacity in Nigeria with Team Science. 2021. (NIH-D43; Submitted).

25. Kingham P(PI), Alatise O (Co-PI), Du (Co-PI), Abdulkareem FB (Co-I). Determining the risk factor profile and biology of colorectal cancer in Nigeria. 2020. (NIH-R01; Received).


32. Olopade OI (PI), Odunsi AO (Co-PI), Ojengbede O, Adejumo PO, Abdulkareem FB (Member-TAC. Global Scholars in Oncology Associated Research (Global-SOAR) training program. 2021. (NIH-D43; Submitted)

33. Olopade OI(PI), Ntekim A, Popoola AO, Arowolo OA, Sowunmi A, Olopade CS, Ibraheem A, Abdulkareem FB (Project Site Director), Daramola OA, et al. Assessing REsponse to neoadjuvant Taxotere and TrAstuzumab in Nigerian women with HER2-positive breast cancer (ARETTA), a multicenter study. 2019. (Universities of Lagos, Nigeria; University of Ibadan, Nigeria; Obafemi Awolowo University, Nigeria; Ile-Ife and Lagos State University, Nigeria; University of Chicago, USA; Continued).


35. Wei JJ, Abdulkareem FB (Co-I), Silas Olugbenga (Co-I). Develop a telepathology program in promoting cervical cancer research and training in Nigeria. 2020. (Global Health Institute, Northwestern University; Continued).

2017 Batch


38. Calalo CM, Cases RKC, Asor BBN, Abad PJB, Asuncion PC. A content analysis of Facebook groups on congenital adrenal hyperplasia. 2021. (Funded by the Newborn Screening Reference Center – University of the Philippines Manila; Under ethics review).

39. Guntekin Ergun S, Yaylacioglu Tuncay F, Talim B, Dincer P. Examination of ocular developmental pathologies by creating an Lrp5 mutant zebrafish model using CRISPR/Cas9. 2021. (TUBITAK 1002 Grant; Received).

41. Mochly-Rosen D (PI), Manasa J (PI), Dhoro M (Co-PI), Masimirembwa C (Co-PI). Improving translational research in Zimbabwe with a focus on infectious diseases. 2021 (National Institutes of Health Global Infectious Disease Research Training Program; In preparation).

42. (Stanford University USA, University of Zimbabwe, African Institute of Biomedical Science & Technology, Zimbabwe; In preparation).

43. Owusu M (PI), Sambian D, Godfred A. Enhancing global health security: Expanding efforts and strategies to protect, diagnose and improve public health in Ghana. 2020. (CDC Fund through the Centre for Health System Strengthening; Accepted).


45. Roy S (PI), Nuruddin M, Osmani M, Gregor K. Screening of common blinding and ocular tumor among under six-year children in Chittagong district of Bangladesh. 2019. (International Rotary Club, Global Grant; Received - extension applied for due to Covid).


2016 Batch

47. Bozaykut Eker P (PI), Park T (PI), Deniz E (Consultant). Cardiac analysis on the superior hypoxia-tolerance through in vivo and in vitro approaches. 2021. (Joint grant from TUBITAK, Turkey and NFS, USA; Received).

48. Toklu Keskin N (PI), Deniz E (Consultant). Comparison of hot spot mutant p53 proteins’ effects on glioblastoma. 2021. (TUBITAK, Turkey; Received).


50. Butali A (PI), Adeyemo WL (Co-investigator), Eshete MA. Refining the genetic and genomic architecture of non-syndromic orofacial clefts. 2020-2025. (NIH/NIDCR; Received).


52. Hou, L, Sagay A, Ogunsola FT, Akanmu AS, Murphy M (PI), Adeyemo WL (Co-I), Abdulkareem FB. Nigeria research training grant for HIV and malignancies. 2019-2024. (D43TW009575-06, Northwestern University, USA; Continued).

53. Butali A (PI) Adeyemo WL (Co-I). Orofacial clefts, whole genome sequencing and incidental findings: Ethical considerations. 2020-2025. (NIH/NIDCR; Received).


56. Ntakirutimana G (PI), Masaisa F (Co-PI), Mukandekezi A (Co-PI). Uwizeyimana G (Co-PI). Rugwizangoga B (Co-PI) and Wineza A (Co-PI). Assessment of molecular diagnosis and response of Chronic Myeloid Leukaemia (CML) using reverse transcriptase- polymerase chain reaction at CHUK. 2021. (University Teaching Hospital of Kigali (CHUK), Directorate of Research; Received).


61. Ariani Y (PI), Yuridian R (Co-PI). The role of variant of COL1A1 gene in 7 major ethnic groups in Indonesia towards wrinkle appearance. 2021. (Paragon Technology and Innovation Corp. Indonesia; Accepted).

62. Ariani Y (PI), Bawolaksono (Co-PI). Melasma and it’s correlation to variant of MC1R gene in Indonesian women population. 2021. (Paragon Technology and Innovation corp. Indonesia; Accepted).

C. Collaborations (NIH and Other Institutions):

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

2019 Batch

1. LLamos-Paneque A, Gahl WA, Adams D, (Undiagnosed Diseases Network International (UDNI). Data Sharing Project. (Specialty Hospital No.1 FF. AA, Quito- Medical Genetics Branch; NHGRI/NIH).

2. LLamos-Paneque A, Stephen G, Tifft C. ICORD’s new initiative: Rare diseases in the Caribbean and Latin America (ERCAL). (Specialty Hospital No.1 FF. AA, Ecuador- ORDR; NHGRI/NIH).


2018 Batch

4. Pasechnikova N, Zborovska O, Savina O. Molecular and clinical research in Ukrainian families with the inherited eye diseases. (V. Filatov Institute of Eye Diseases and Tissue Therapy; NEI/NIH).

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


7. Riazuddin S, Naeem MA, Hejtmancik JF, Gottsch JD, Riazuddin SA, Khan SY, Ali M, Qazi ZA, Butt NH, et al. Hereditary vision impairment in Pakistan (CEMB, University of the Punjab, Lahore-Pakistan; NEI/NIH; Wilmer Eye Institute, Johns Hopkins University, USA; Layton Rehmatulla Benevolent Trust Eye Hospital, Lahore; Allama Iqbal Medical College, University of Health Sciences, Lahore, Pakistan).

2017 Batch

2016 Batch
9. Ekure E, Kruszka P, Adeyemo A. Atlas of human malformation syndromes in diverse populations. (University of Lagos/Lagos University Teaching Hospital, Nigeria; NHGRI/NIH).
10. Ekure E, Adeyemo A. Genetics of congenital heart diseases in Africa. (University of Lagos/Lagos University Teaching Hospital, Nigeria; NHGRI/NIH).

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

2019 Batch
3. Akay G, Posey JE, Llamos-Paneque A. Genome sequencing to elucidate causes and mechanisms of mendelian genetic disorders. (Baylor College of Medicine, USA; Genetics Service of Hospital No.1 FF. AA. Quito-Ecuador).
4. Bocoum A, Koita A, Rizig M, Maiga Y, Guinto CO. Parkinson’s disease in Africa: Developing a clinically characterized DNA; resource for Genome-Wide Association (GWAS) and other collaborative genetic studies new research (Centre Hospitalier Universitaire du Point “G”, Bamako, Mali; Centre Hospitalier Universitaire Gabriel Touré, Bamako, Mali; UCL Queen Square Institute of Neurology, London, UK).
5. Chowdhury EH, Amarakoon GGTT, Mosema KBA, Khant AK. Associations/complications with Trisomy 21in children. (Samtse Genral Hospital, Bhutan; Faculty of Medicine and Allied Sciences, Rajarata University of Sri Lanka, Sri Lanka; Instut National de Recherche Biomedical, Biamamarie Mutombo Hospital, Kinshasa, DR Congo; Yangon Children Hospital, Myanmar).
6. **Cornejo-Olivas M, Llamos-Paneque, A.** Collaboration in technical support regarding molecular diagnosis of neurogenetics diseases like spinocerebellar ataxias, Huntington disease, steinert Disease. (Neurogenetics Research Center in Perú; Genetics Service of Hospital No.1 FF. AA. Quito, Ecuador).


8. **Hussein N**, Quinonez SC. Introducing MiGene Family History App in Malaysia. (Family History Working Group, University of Malaya, Malaysia; Global Genomic Medicine Collaborative).

9. **Iqbal M**, Ahmed ZM. Analyses of DNA variants causing otitis media in Pakistani population. (Department of Biotechnology, The Islamia University of Bahawalpur, Pakistan; University of Maryland, School of Medicine, USA).

10. **Iqbal M**, Ansar M. Mutational analysis of DNA from consanguineous Pakistani families having ophthalmological disorders through whole exome sequencing (Department of Biotechnology, The Islamia University of Bahawalpur, Pakistan; Institute of Molecular and Clinical Ophthalmology Basel (IOB) Basel, Switzerland).

11. **Iqbal M, Naeem MA.** Identification, clinical evaluation, and genetic analysis of RP families (Department of Biotechnology, The Islamia University of Bahawalpur, Pakistan; National Centre of Excellence in Molecular Biology, University of the Punjab, Lahore, Pakistan).

12. **López-Star B, Llamos Paneque A.** Consultation on ophthalmogenetic case. (Mexican Society of Ophthalmology; Mexican Institute of Ophthalmology; Specialty Hospital No.1 FF. AA, Quito, Ecuador).


14. Olorunniji F (PI), **Ali LA.** Adeoye R. Development of a platform for rapid on-site testing of novel viral infections in Nigeria based on CRISPR, isothermal PCR amplification, and split G-quadruplex DNAzyme technologies. (GCRF, Liverpool John Moores University, UK; University of Abuja, Nigeria; University of Ilorin, Nigeria).

15. Santra MK, Rapole S, Tripathi V, Shridhar PR, Shanamugham D, Tilak TVSVGK, **Mutreja D**, Deshmukh C, Tamhankar AS, Chatterjee S. Mechanism & therapeutic application of neuroimmune communication in colorectal cancer (Dept. of Biotechnology, Ministry of Science & Technology, India; NCCS, Pune; NCL Pune; DMH Pune; School of Chemistry, University of Hyderabad).


**2018 Batch**

17. 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).

18. **Cornejo-Olivas M, Dutta AK, Dueñas M, Dwivedi A, Mistri M.** Chapter on ‘Nutrigenomics’, Springer Nature book on Biochemical aspects of nutrients (micro and macro) and nutricles and...
therapeutic potential of nutrients along with nutrigenomics. (National Institute of Neurological Sciences, Peru; All India Institute of Medical Sciences Kalyani, India; Hospital Edgardo Rebagliati Martins, Peru, Army Hospital Research and Referral (AHRR), India, FRIGE's Institute of Human Genetics, India).

19. Düttke AK, Lemke L, Paul L, Riggs ER. ClinGen GRIN Variant Curation Expert Panel (VCEP). (AIIMS Kalyani, India; Institute of Human Genetics, Leipzig University Hospital, Germany; Geisinger Health System, Pennsylvania, USA).

20. Dwivedi A, Kumar R, Singh AK, Dey M. To study the clinical utility and diagnostic yield of exome sequencing in prenatal diagnosis of congenital anomalies (AHRR, Delhi & Base Hospital Delhi Cantt, India).

21. Gitaka J, Clark T. Molecular epidemiology of Plasmodium falciparum. (London School of Hygiene and Tropical Medicine, UK; Mount Kenya University, University of Nairobi).

22. Jaja C, Edem-Hotah J. The insights into microbiome and environmental contributions to sickle cell disease and leg ulcers (The INSIGHTS Study). (University of Sierra Leone College of Medicine and Allied Health Sciences, Sierra Leone; University of South Carolina, USA).


25. Luquetti D, Dueñas-Roque M. Phenotypic and genomic characterization of microtia in the Andean Population. (Washington University, USA; Seattle Children’s Hospital, USA; Hospital Nacional Edgardo Rebagliati Martins, Perú).


27. Messaoud O, Düttke AK, Cornejo-Olivas MR, Bhuiyan ZA, Banerjee K. Editing a special issue ‘Monogenic vs Oligogenic Reclassification’ https://www.frontiersin.org/research-topics/13904/monogenic-vs-oligogenic-reclassification in Frontiers in Genetics (https://www.frontiersin.org/journals/genetics) (Institut Pasteur de Tunis, Tunisia; All India Institute of Medical Sciences, Kalyani, India; National Institute of Neurological Sciences, Lima, Peru; Centre Hospitalier Universitaire Vaudois, Lausanne, Switzerland; Institute White Plains, United States).

28. Messaoud O, Banerjee K, Cornejo-Olivas M, Düttke AK, Bhuiyan ZA. Special article series on ‘Monogenic vs Oligogenic Reclassification” in Frontiers in Genetics/ Frontiers in Pediatrics (https://www.frontiersin.org/research-topics/13904/monogenic-vs-oligogenic-reclassification). (Institut Pasteur de Tunis, Tunisia; Weill Cornell Medicine, USA; National Institute of Neurological Sciences, Peru; All India Institute of Medical Sciences Kalyani, India; Centre Hospitalier Universitaire Vaudois (CHUV) Lausanne, Switzerland).

29. Messaoud O, Haddad S, Charfi-Kaddour S, Gargouri M, Drissi LB, Hammami A, Bardaoui A. The use of advanced materials for molecular biology applications. (Institut Pasteur de Tunis, Tunisia; Faculty of Sciences of Tunis, Tunisia; Faculty of Sciences of Bizerte, Tunisia; Borj Cedria technopole, Tunisia; Faculty of Sciences, Rabat, Morocco).
30. **Messaoud O**, Khalil A, Nasr M, Azzab S, Sunoqrot S, Amro A. Screening of nanoparticles, biomaterials, natural extracts and disinfectants on coronavirus strain using several antiviral tests. (Institut Pasteur de Tunis, Tunisia; Egypt Japan University of Science and Technology (EJUST), Egypt; The Public Authority Ain Shams University, Egypt; Al-Zaytoonah University of Jordan, Jordan; Al-Quds University, Palestine)

31. **Naeem MA**, Ali M, Butt H, Qazi ZA, Mehmood A, Shahid AA, Riazuddin S. Clinical study on mesenchymal stem cell modality for management of Retinitis Pigmentosa in patients. (LRBT Hospital; Stem Cell Labs at Jinnah Burn & Reconstructive Surgery Center, Jinnah Hospital Complex, Lahore; National Centre of Excellence in Molecular Biology, University of the Punjab Lahore, Pakistan).

32. **Nakousi-Capurro N**, Cavalcanti DP. Description of genotype-phenotype relations in patients with osteogenesis imperfecta in a Brazilian Cohort. (Clinical Genetics Service, Carlos Van Buren of Valparaíso, Chile; Department of Clinical Genetics, Faculty of Medical Sciences at University of Campinas, Sao Paulo, Brazil).

33. Omisore AD, Sutton EJ, Omidiji O, **Abdulkareem FB**, Daramola AO. Tablet-based mobile health ultrasound for point-of-care breast cancer diagnosis in Nigeria. (Obafemi Awolowo University, Nigeria; University of Lagos, Nigeria; Memorial Sloan Kettering Cancer Center, USA).


35. Pasechnikova N, Zborovska O, **Savina O**, Kremenska U. Genetic diagnostics; medical and genetic consultation for families with retinoblastoma in an ISIDA-IVF genetical laboratory with a view to implementing pre-implantation diagnostics during the period of planning for the next pregnancy in the said families. (V. Filatov Institute of eye diseases and tissue therapy; ISIDA-IVF clinic, Ukraine).

36. **Savina O**, Rykov O, Kremenska U. Genetic testing; medical and genetic consultation for families with Aniridia and WAGR syndrome - implement pre-implantation diagnostics during family planning. (Department of Ophthalmology Shupyk’s National Medical Academy of Postgraduate Education; ISIDA-IVF clinic; NGO - Aniridia. WAGR, Kyiv ophthalmological clinic, Ukraine; In discussion).


**2017 Batch**


40. **Casado PL**, Patrick Shmidtlin. Frontier in Dental Medicine: Biomarkers in periodontology and peri-implant diseases. (Fluminense Federal University, Brazil; University of Zurich, Switzerland).
41. Finger PT, Tomar A, Kivela T, Krema H, Yousef YA. Treatment for radiation retinopathy, for patients with choroidal melanoma treated by radioactive plaque therapy. (King Hussein Cancer Center, Jordan; International multicenter study with New York Eye Cancer Center, USA).


43. Petlichkovski A, Sukarova-Angelovska E, Brusco A, Rubeis SD. Whole-exome sequencing of proband families with neurodevelopmental disorders. (Institute for Immunobiology and Human Genetics, Medical Faculty Skopje, Macedonia; Pediatric University Clinic, Medical Faculty Skopje, Macedonia, University of Turin, Italy; Icahn School of Medicine at Mount Sinai, New York, USA).

44. Petlichkovski A, Sukarova-Angelovska E. Analysis of genes involved in folate metabolism, in children with congenital anomalies associated with palate cleft (Institute for Immunobiology and Human Genetics, Neurology University Clinic, Medical Faculty Skopje, Pediatric University Clinic, Medical Faculty Skopje, Macedonia.)

45. Roy S, Osmani M, Geary A, Dean W. Fundamental virtual reality simulation for manual small incision cataract surgery validity: Evaluation, efficacy and acceptability study in Bangladesh, China, Ethiopia, India, Mongolia, Togo, United Kingdom and United States of America. (Chittagong Eye Infirmary, Bangladesh; London School of Hygiene, UK; 13 centers from 8 countries).


47. Thakur N, Mayo C, Matt BJ. WHO Collective Global Network for Rare Disease (WHO CGN4RD).

2016 Batch

48. Adeoye AM, Bongani M, Connolly SJ. INVestigation of rheumatiC AFTreatment Using vitamin K antagonists, rivaroxaban or aspirin Studies (INVITUS trial). (University of Ibadan, Nigeria; University of Cape Town, S. Africa; Population Health Research Institute, Canada).

49. Adeoye AM, Karaye KK, Loeb M, Yusuf S. A randomized controlled trial of Influenza Vaccine to prevent adverse Vascular Events (IVVETrial). (University of Ibadan, Nigeria; Bayero University, Kano, Nigeria; McMaster University, Canada).

50. Ariani Y, Beben B, Khaireunisa S. Genomic variations of hair aging in 7 major ethnic groups in Indonesia. (Indonesian Medical Education and Research Institute (IMERI) - University of Indonesia; Biostatistics Department - University of South Australia; Paragon Technology and Innovation Corp. Indonesia).

51. Gui H, Bogi E. To develop personalized medicine in Latino America (INBIOMEDIC, Peru; Copenhagen Institute for future studies, Denmark).


53. Okafor FU. Introduction of genetics and genomics in the nursing curriculum (The University of Benin; Benson Idahoasa University, Nigeria; In discussion).
D. New Research Projects: (Total- 46; 2019- 17; 2018- 8; 2017- 10; 2016- 11)

2019 Batch

1. Amarakoon GG, Wijayarathne HK. Awareness on existing neonatal screening among mothers in a tertiary care center-Sri Lanka.
3. Chaudhry A, Bocoum A, Rizig M. Identifying gaps in knowledge about Parkinson’s disease amongst medical professionals in Africa.
7. Hoe HK, Sherina NS, Hussein N, Malik TF. Exploring the views and experiences of parents on school-based thalassemia screening in Klang district, Malaysia.
8. LLamos-Paneque A. Andean Duchenne Muscular Dystrophy Registry Initiative, coordinated by PTC Therapeutics.
12. Monye HI, Kyari F, Ugalahi MO. Ophthalmologists’ knowledge, perceptions, and practice patterns of ophthalmic genetics services in Nigeria (Submitted for ethical review).
13. Ng WL (PI), Hussein N (Co-PI), Lee YK, Ng CJ, Kee BP, Malik TFA, Qureshi N, Then SM, Kwan Z. HLA-b*58:01 Allele testing in primary care to prevent allopurinol induced-severe cutaneous adverse reaction (SCAR).
17. Win KPP, Khant AK. Thyroid hormone status and growth parameter in children with different stages of chronic kidney disease.

2018 Batch
18. Anorue EI, Gbotolorun MO, Oluwarotimi AC, Arotiba GT. Health related quality of life of patients presenting with orofacial Hard tissue injuries at Lagos University Teaching Hospital, Idi-araba Lagos. A prospective longitudinal study.


20. Cornejo-Olivas M. Closing the diagnostic gap in inherited neuropathies and related disorders.


25. Utumativishima JN, Nkeshimana M, Mutimura E. Associations of SARS-CoV-2 with CVD risks in Rwandan adults admitted at Tertiary Hospital, Kigali.

**2017 Batch**


27. Fatima SS (supervisor). Methylation status of promoter region of FTO, POMC and IGF genes in familial Type 2 Diabetes.


32. Roy S, Hoque F. Cosmetic outcome of congenital ptosis treated with prang suture and without prang suture.

33. Roy S, Hoque F. Demographic profile, presentation and treatment outcome of ocular surface neoplasia treated in a tertiary eye hospital in Bangladesh.


35. Yüksel M, Kayhan G, Yaylacioglu Tuncay F, Ergun MA, Ozdek Ş. Genotype and phenotype characteristics of Stargardt's Disease in Turkish population.

**2016 Batch**


37. Gio H, Jaramillo L. Metagenomic analysis and antimicrobial resistance in urban and hospital’s wastewater of Huánuco.

41. Okafor FU. Translate knowledge in genetics and genomes nursing to clinical settings, community, and primary health facility.
42. Sjarif DR, Ariani Y, Dewi BE. Development of iduronate 2-sulfatase enzyme using mRNA method as a treatment for mucopolysaccharidosis type II.
43. Sjarif DR, Ariani Y, Fadilah. Developing pipeline for detection of genes causing rare diseases using whole exome sequencing data.
44. Subhedar K, Tibrewal S, Sen P. Maternal vitamin A deficiency and MAC (Microphthalmos, Anophthalmos and Coloboma) disorders.
45. Tibrewal S, Ratna Ria, Das S. Genotype-phenotype correlation of retinoblastoma in north India.
46. Uwineza A. Qualitative assessment of faculties’ perspectives toward precision medicine-Education innovation project: Integration of Precision Medicine in medical curriculum at the University of Rwanda.

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

2019 Batch
2. Alcausin MML, Tumulak MJR, Silao CL, Fabella TD. Clinical characterization and identification of CFTR gene mutations in newborns with positive screen for cystic fibrosis in the expanded newborn screening program. (Summit lectures on proposal/grant writing helped with the proposal of this project).
4. Badoe E, Ameyaw EK, Wiafe SA, Manu E, Asafo-Agyei SB, Thomford NE, Ashong J. Molecular diagnosis of MPS patients in Ghana: The RDGI-BioMarin Project. (Knowledge from Summit helped design project).
5. Chowdhury EH, Roy S, Hoque F. Epidemiological study of Retinoblastoma in a Tertiary Eye Care Center of Bangladesh. (Knowledge gained at Summit improved research quality).
7. Hassib N, Sayed I. An attempt to diagnose typical and atypical cases by detection genes causing tooth structure anomalies. (Summit lectures improved skills in diagnosis of rare genetic disorders).
8. Hussein N, Lee YK, Qureshi N, John P, Azmi A, Ng CJ. Identifying gaps in thalassaemia screening: Exploring the views, experiences, and beliefs among thalassaemia carriers in Sabah,
Malaysia. (Awareness on challenges of underprivileged population gained at the Summit helped with better communication; improved health service).

9. **Kars ME, Ozcelik T.** Prevalence of actionable secondary findings in 3,599 whole exomes and genomes from Turkey. (Knowledge gained from Summit lectures, personalized training and field trips helped with the interpretation of genetic variants and sequencing data analysis).

10. **Llamos-Paneque A, Hernández-Iñiguez P, Rivas-Iglesias C, Sanchez-Salazar J, Onofre Perez EJ.** Clinical characterization of pediatric patients with Steinert's myotonic dystrophy diagnosed at the Hospital of Specialties of the Armed Forces No. 1. (Knowledge from Summit lectures in neurogenetics emphasized on triplet expansion disorders; helped in execution of the study.)

11. **Llamos-Paneque A, Lamar-Segura, E, Nacato-Pachacama K, Guzmán A.** Cytogenetic variants in patients with Turner syndrome diagnosed at the Hospital of Specialties of the Armed Forces No. 1 of Quito, Ecuador. (Summit helped reaffirm usefulness of registries for epidemiological studies, clinical characterization of genetic disorders).

12. **López-Star B, Pérez- Serrano R, Ochoa L.** Identification of individuals carrying the rs7677751 polymorphism of the PDGFRA gene in patients with Keratoconus. (Summit lectures on eye disorders provided knowledge for execution of study).

13. **López-Star B.** Pérez- Serrano R. Identification of rs7677751 polymorphism in PDGFRA gene, in patients with corneal astigmatism. (Dr. Hufnagel’s team (NEI), helped understand the best choice of genes associated with astigmatism in Mexican population).

14. **LoTempio J, Bramble MS, Mosema KBA, Kamangu EN, Mumba-Ngoyi D, Tshala-Katumbay D, Vilain E.** Building high-quality genome-enabled reference sets to address genetic diversity in Congolese ethnic groups in the Democratic Republic of Congo. (Summit lectures on Bioethics and Variant Interpretation helped develop human subject consent protocols).

15. **Luong LH, Hoang LT, Le NT, Tran TN.** Study of genetics component in cardiovascular malformation and rare vascular disorder. (Summit encouraged discussions, collaborations and initiation of this project).

16. **Luong LH, Hoang LT, Le NT, Tran TN.** Study of rare immunodeficiency disorder. (Knowledge and credentials from the Summit initiated discussions and project).

17. **Mhandire K, Dandara C.** Pharmacogenomic determinants of resistant hypertension in Africans: Towards a gene-based treatment outcome prediction tool. (Dr. Marino L, a Summit contact, shared online resources that helped me pursue training in programming and bioinformatics; helped in setting-up this machine learning based project; helped transition to dry lab).

18. **Monye HI, Olusanya BA, Tongo K.** Normative values for ocular biometric parameters in preterm babies and associated socio-demographic and clinical factors at the University College Hospital, Ibadan. (Summit lectures and field trips stimulated interest in newborn screening-ophtalmic context, study will provide baseline data for monitoring preterm babies).

19. **Mosema KBA, Spencer D, Bramble MS, Likuba EB, Mumba-Ngoyi D, Tshala-Katumbay D, Vilain E, Délot E, Vilain E.** Disorders/differences of sex development in Central Africa: Genetics, psychosocial adaptation and Perceptions. (New skill acquired at the Summit and lectures on genome sequencing and variants interpretation helped write research proposal, and conduct research for a PhD thesis).

20. **Mutreja D, Komala SB, Singh V, Sreenivasa S.** A longitudinal prospective study on role of serum hepcidin as a biomarker for iron status in chronic kidney disease (CKD). (Summit reiterated personalized approach for diagnosis of CKD related anemia utilizing hepcidin).

21. **Mutreja D, Venkatesan S, Sharma S, Tilak TVSGK, Boruah D.** A pilot study to evaluate the role of vascular endothelial growth factor and its comparison with microvessel density in
angiogenesis of hematological malignancies. (Summit emphasized importance of gene disease association in hematologic malignancies).

22. Nnodu O (PI), Isa H, Tanko Y, Otu T, Alli LA, Chianumba R, Agumadu UN, Ekong J, Elmi Z, Rooks H, Menzel S, Ekong R. A pilot study on sickle cell disease to identify an unusual haplotype at 1 the BCL11A locus in Nigeria. (Summit lecture on Genetics of sickle cell disease enhanced my contribution; improved research quality).


24. Sayed I, Abdel-Kader M. Clinical and Molecular characterization of syndromic and non-syndromic tooth agenesis with suggested management in cases with severely atrophied alveolar ridges. (Summit lectures improved skills in diagnosis of rare genetic disorders).


27. Wiafe SA, Anyane-Yeboa K, Brew YA, Badoe E, Ameyaw EK, Thomford NE, Ashong J. Clinical and genetic evaluation of undiagnosed genetic and rare disease patients through the IHOPE Program in Ghana. (Knowledge from Summit improved design, developed protocol).


2018 Batch

29. Acharya M, Dutta AK, Mukherjee S. Understanding genetic architecture underlying hereditary non- syndromic hearing loss. (Summit lectures, NCBI resources on genetics of hearing loss and variant prioritization were critical).

30. Adeyemo TA, Bolarinwa AB, Olatinwo AT, Adiat AL, Akanmu AS. Clinical and laboratory phenotypes of persons living with haemophilia in Southwest Nigeria. (Knowledge from Summit improved case reporting).

31. Adeyemo TA. Akinsete AM, Ojewunmi OO, Akinsete A. Sickle cell disease registry of Nigeria (SCDRN). Targets 5000 SCD for accurate clinical and patient-reported data to support clinical research, interventions, management, and quality of care of patients. (Summit helped initiate registry).

32. Afolabi BB (PI), Babah O, Adeyemo TA. Low dose aspirin for preventing intrauterine growth restriction and preeclampsia in sickle cell pregnancy (PIPSICKLE): a randomised controlled trial. (Knowledge from Summit supported study design, implementation).

33. Ajoloko EA, Oluwarotimi AC, Adeyemi MO. Efficacy of sodium bicarbonate buffered lidocaine hydrochloride in reduction of injection, onset of action and depth of anaesthesia intra-
alveolar extraction of mandibular first and second molars: A randomized controlled double-blind study. (Knowledge from Summit helped design study).

34. Cornejo-Olivas M, Cosentino C. Study for research on genetics of Parkinson disease in Peru, a LARGE PD study. (Summit lectures in Bioethics and Scientific Writing helped improve informed consent form and proposal).

35. Cornejo-Olivas M, Cubas-Montecino D, Bazalar-Montoya J. DNA biobank implementation in Neurogenetics Division, Instituto Nacional de Ciencias Neurologicas. (Summit lectures on broad informed consent, biobanking and ethics supported proposal and design of standardized procedures of our first biobank).


37. Dueñas-Roque M, Mayorga G, Prötzel A, Gamarra N, Ota A. Birth defects registry in the Health Social Security of Peru. A pilot study at a reference hospital. (Summit introduced me to researchers in congenital malformations, motivated me to continue this work).

38. Dueñas-Roque M, Segura P, Soria M, Calderón M, Rondón P. Epidemiology of lysosomal storage disorders in the Health Social Security of Peru. (Summit encouraged me to do more research in Peru).


41. Lertwilaiwittaya P, Pho-iam T, Soontrapa P, Boonyapisit K, Limwongse C. Prevalence of C9orf72 hexanucleotide repeat expansion in different neurodegenerative diseases in Thailand. (Summit connected me to Dr. Bryan Traynor who helped obtain a positive control sample to establish local test in Thailand).

42. Lertwilaiwittaya P, Roothumnong E, Thongnoppakhun W, Limwongse C, Pithukpakorn M. Next generation sequencing in breast-ovarian cancer in Thailand. (Summit lectures on writing manuscripts helped write the proposal and manuscript draft).

43. Mazzetti-Soler P, Cornejo-Olivas M, Cosentino C, Torres L. Illanes-Manrique M. Enroll-HD: A world observational study for Huntington’s disease families; A CHDI foundation project. (Dr. K. Fischbeck’s (NINDS) Neurogenetics talk at the Summit, suggestions and comments helped improve recruitment strategy and MTA agreement).

44. Menzel Stephan, Adeyemo TA, Nnodu O. Identification of novel mechanisms of fetal-haemoglobin induction by common genetic variation in patients with sickle cell disease. (Summit encouraged research collaborations).

45. Messaoud O. Genetic investigation of Xeroderma pigmentosum (Knowledge gained from Summit personalized trainings helped interpret next generation sequencing data).


2017 Batch
48. Aglan MS, Otaify GA. Targeted next-generation sequencing in the diagnosis of osteogenesis imperfecta and Bruck syndrome. (Knowledge gained from Summit helped improve reporting of results).
49. Avogbe PH, Brun LVC, Sanni A. Comparison of HPV detection in urine and cervical samples collected from Beninese women. (Resources from the Summit helped study design).
51. Besil-Suárez B, Benítez Cordero Y, Roblejo-Balbuena H. Clinical-epidemiological surveillance of microcephaly and other congenital defects after infection by Zika in pregnant women, products of conception and Cuban newborns. (Summit provided tools for research design in complex diseases, analysis and interpretation of NGS data).
53. Casado PL, Aguiar T, Jordao M, Quinelato V. Relationship of geographic tongue before and during COVID-19 pandemic in patients under treatment. (Summit helped understand research on multifactorial diseases).
56. Casado PL, Luiza Motta L, Aguiar T, Quinelato V. Analysis of alcohol consumption during the covid-19 pandemic in Brazilian students. (Knowledge from Summit provided research basis for associated risk in diseases).
57. Casado PL, Quinelato V, Aguiar T. Analysis of 7 different techniques to isolate microRNA. (Summit workshop on Bioinformatics helped understand the relationship of miRNA with DNA and messenger RNA; helped improve execution of project).
58. Casado PL, Quinelato V, Aguiar T. Genetic profile of patients underwent peri-implant maintenance therapy: relationship with peri-implant disease incidence. (Summit lectures helped understand characteristics to be considered when studying multifactorial diseases, such as peri-implant diseases; improved the project, sample collection, medical history analysis, selection of main related genes).
59. Casado PL, Santos M, Telma Aguiar T, Valquiria Quinelato V. Analysis of the development of bruxism during the covid-19 pandemic in Brazilian students. (Knowledge from Summit provided research basis for associated risk in diseases).
60. James O, Adekunle AA. Wound healing following palatoplasty: A randomized study using honey or warm saline mouth bath. (Summit gave insight in exploring better care options for subjects born with orofacial clefts).
61. Lloyd W, Mgasa A, Makani J. Blood transfusion for sickle cell patients attending Temeke Regional Referral Hospital in Tanzania. (Summit enhanced knowledge, focus on genetics of SCD).

62. Lotz-Esquivel S, Alvarado-Aguilar M. Acute intermittent porphyria in Costa Rica. (Knowledge from Summit provided tools for soliciting specialized treatment and laboratory tests for these patients).

63. Mahfoudh W, Faleh R. Circulating tumor DNA as biomarkers for early diagnosis, treatment response and prognosis of breast cancer. (Project was inspired by Summit lectures and tailored training).

64. Marcheco Teruel B, Benítez Cordero Y, Suarez Besil B, Rojas Betancourt IA, Lantigua Cruz PA. Evaluation of the impact of the national program for diagnosis, handling and prevention of genetic diseases and congenital defects. (Summit reaffirmed usefulness of data for personalized medicine, clinical validity of gene-disease associations, registries for genetic epidemiological studies).

65. Nandal R, Biswas S, Laller KS. A study of serum vitamin-d levels in young adults with ST Elevation Myocardial Infarction (STEMI). (Summit enhanced research methodology, protocol preparation and grant writing).


67. Roy S. Effectivity of interferon alfa 2a in treatment of ocular surface neoplasia. (Knowledge from Summit inspired exploration of alternative treatments).

68. Roy S. Presentation of genetic diseases in a Tertiary Eye Care Center of Bangladesh. (Summit inspired the diagnosis of other genetic diseases presented with retinoblastoma, the first of its kind in Bangladesh).

69. Torres-Mejía G (PI), Fejerman L, Ángeles-Llerenas A, Gómez Flores-Ramos L. LAGENO-BC & CONFLUENCE research proposal. (Knowledge from Summit helped write manuscript).


2016 Batch


73. Bode-Thomas F, Yilgwan CS, Amusa GA, Isa MS, Uche U, Ugwu C, Udo P, Sani MU, … Ekure EN, et al. Nigerian National Registry for rheumatic heart disease. (Summit emphasized on relevance of birth defects registry including congenital heart disease, which was translated to another cardiac disease of public health importance in our setting).

74. Butali A, Adeyemo WL. Refining the genetic and genomic architecture of non-syndromic orofacial clefts. (Knowledge from Summit of immense value in advancing this project).
75. Dasanayaka NN, Sirisena ND, Samaranayake N. Telomere length, gene expression profile and DNA methylation status of selected genes in a Sri Lankan group of practitioners of meditation and mindfulness techniques (Knowledge from Summit supported study design and implementation).

76. Deniz E. Genome-wide screening with CRISPR/Cas9 and modelling of resistance mechanisms developed against cytotoxic drugs in cancer treatment. (Knowledge from Summit in human genomic variations helped analyzing bioinformatics data).

77. Ekure EN, Adeyemo AA, Sokunbi OJ, Muenke M, Kruszka P. Case reports on genetic diseases associated with congenital heart defects -Emery-Dreifuss muscular dystrophy syndrome, neurofibromatosis and Hajdu Cheney syndrome. (Summit’s introduction to the electronic atlas and importance of highlighting genetic syndromes in diverse populations for diagnosis, led to the necessity of publishing findings related to children of sub-Saharan African descent).


79. Krawitz P, Ekure EN, Opoola ZA, Lesi FEA. Computer-assisted image analysis and exome sequencing for the diagnosis of genetic disorders. (Summit introduced utilization of the Face2Gene software; resulted in collaboration with German PI).

80. Lallar M, Kaur L, Singh UP. Attitude, expectation and perception towards genetic counselling in parents of children with genetic disease. (Genetic counselling sessions at the Summit helped modulate sessions for patients).

81. Lallar M, Kaur L. Diagnostic yield of fetal and perinatal autopsy. (Knowledge from Summit on molecular testing and web resources helped in analysis of genetic diseases).

82. Mekkawy MK, Hussen DF. Molecular cytogenetic characterization and breakpoint mapping of chromosomal rearrangements towards better management of patients with genetic disorders and carriers (Molecular cytogenetic techniques learned at Summit helped in accurate diagnosis).

83. Okafor FU. Analysis of data for ‘Reproductive health genetics and genomics education among faculty and students in basic nursing programmes in default Old Western Region, Nigeria’ (PhD Thesis). (Knowledge from Summit will guide action for inclusion of genetics and genomics in nursing).

84. Roblejo-Balbuena H, Marcheco-Teruel B, Benitez Cordero Y, Monzón Benítez G. Genetic factors associated with the clinical severity of COVID 19 in Cuban patients. (Summit provided tools for research design in complex diseases; analysis and interpretation of NGS data).

85. Sjarif DR, Ariani Y, Priambodo R, Prakoso NM, Hafifah CN, Yuliarti K. Lysosomal storage disease study: developing screening and diagnostic methods for MPS II, MPS IVA, Gaucher disease, Pompe disease and NCL (Neuronal Ceroid Lipofuscinosis) type 2. (Project was inspired by Summit lectures on Inborn Error of Metabolism, Tools for molecular genetics, Diagnostic odyssey, Variant interpretation, Variant nomenclature.)

86. Tibrewal S, Ratna R, Dubey S. Childhood glaucoma – inheritance pattern and role of consanguinity. (Summit gave confidence to embark on this project).

87. Tibrewal S, Ratna R, Ganesh S. Familial inheritance pattern in childhood cataract in north India. (Summit gave confidence to embark on this project).

F. Other Accomplishments/Initiatives:
1. Abubakar S. Guest Researcher at Social Network Methods Section, NHGRI, Visiting Program, DHHS (2020); Lectured on ethical, legal & social implication (ELSI) of Genetics and Genomics in the Medical Ethics & Law Course for Masters programs (MPH; Global Health), at College of Health Sciences and the World Bank Funded Africa Center of Excellence for Population Health and Policy, Bayero University, Kano, Nigeria; Appointed as Reviewer, BMC Health Services Research, Springer Nature (Global Publishers, 2021); Supervised undergraduate medical student on knowledge of Family History Taking Tools among Health Professionals in Nigeria.

2. Akinmola OO. Advanced in dialogue with management for starting a genetic counselling unit in the Tertiary Hospital, Lagos University Teaching Hospital; Lectured on Management of Neonatal Diabetes, Genetics as a tool for precision medicine, to optimize outcome for patients, Newborn screening: Neonatal hypothyroidism, at Paediatric Endocrinology Training Centre For West Africa; Lectured on Human genetics to Chemical Pathologist Society of Nigeria; Member, G2MC Rare Disease Working Group for pilot study at different sites; Attended the virtual International conference on COVID-19, International Federation of Clinical Chemistry (Feb. 2021); Identified a rare disease of Kallmann syndrome, counselled and referred appropriately for treatment.

3. Alamin TA. Joined 2-year Master of Medical Education program (2021); Introduced genomics in undergraduate medical laboratory sciences curriculum; Participated in initiation of the Sudanese Human Genetic Association.

4. Alli LA. Appointed External examiner, first Professional Examination in Medical Biochemistry and Molecular Biology, Kaduna State University (Dec. 2020); Appointed, Visiting Lecturer, Medical Biochemistry and Molecular Biology for Preclinical Medical students, Nile University, Abuja Nigeria (Jun. 2021); Used materials from Summit to lecture on Basic molecular biology, Genetic disorders, and Inborn errors of metabolism; Strengthened the Biochemistry curriculum on genetics and genomics for preclinical medical trainees; Reviewed research articles for Pharmaceutical Biology and British Journal of Medicine and Medical Research; Supervised, mentored postgraduate research students (5 MSc, 1 PhD) on projects based on human genetics, molecular biology and ethnopharmacology management of SCA.


7. Bocoum A. Joined International Parkinson’s disease Genomic Consortium; Member, African Section Educational Committee; Awarded, GP2MSc scholarship; Established Malian National Registry of Parkinson’s disease.
8. **Chowdhury EH.** Summit certificate helped secure position of a specialist in the Department of Pediatrics, Samtse General Hospital (SGH), Royal Government of Bhutan (2020); Previously was Associate Consultant, at Department of Pediatrics, Chittagong Eye Infirmary and Training Complex, Chittagong, Bangladesh; Initiated registry for congenital abnormalities in Department of Pediatrics, SGH; Emphasized importance of genetic counseling based on Summit learnings; Trained colleagues to establish research/clinical facilities in SGH; Emphasized on personalized care; Presented on ‘Recent management protocol of common genetic diseases in children’, CME program in SGH; Identified and managed pediatric genetic disorders (19) in Department of Pediatrics, SGH; Diagnosed cases Down’s Syndrome (7), Hemophilia (3), Congenital Hypothyroidism (3), Achondroplasia (2), Neurofibromatosis (1), Turner’s Syndrome (1), Duchenne Muscular Dystrophy (1), Marfan’s Syndrome (1); Referred cases for cardiac surgery (4), Ophthalmic surgery (1), physiotherapy (2); Diagnosis improved quality of life of affected individuals and their families due to known prognosis and access to appropriate support.

9. **Guven Y.** Presented poster on ‘Three Nance Horan syndrome families from Turkey: Three different approaches for molecular diagnosis’, at ESHG Conference (virtual, Aug. 2021); Presented poster on ‘Gorlin-Goltz Syndrome with a de novo variant in PTCH1: A case report’, at 28th Congress of the IAPD (virtual, Jun. 2021); Diagnosed, referred patients (12) to genetics department for molecular confirmation and medical referral- Ectodermal dysplasia (4), Nance Horan syndrome (1), Primary Failure of eruption (1), Oligodontia cases (8); Early diagnosis, surgical interventions, dental prosthodontic and restorative therapies, preventive and follow-up protocols, contributed to socio-psychological well-being, improved quality of life of children.

10. **Gyawali P.** Young Investigator, Global Genomic Medicine Collaborative (G2MC); Member, Family Health History Flagship Project under G2MC; Corresponding Member, International Federation of Clinical Chemistry, Task Force in Global Newborn Screening; Master’s thesis supervisor, Global Health, University of Copenhagen.

11. **Hussein N.** Coordinator, Postgraduate Family Medicine Specialty, University of Malaya, Malaysia; Audited postgraduate curriculum to increase awareness, strengthen Family Medicine in genetics and genomics; Received scholarship to attend RARD (S. Africa, Jun. 2020; postponed due to COVID-19 pandemic); Invited speaker, by Tumulak MJR (Summiteer) for an online session, on ‘Cancer genetics in primary care’ and ‘Communicating genetic risk’, University Malaya (Jun. 2021); Planned topics and workshops on ‘Use of technology in patient care’, ‘Patient education and counselling’, ‘Chronic kidney disease’ and ‘Family planning’; Mentored Family Medicine trainees on research projects in ‘Challenges among primary care doctors’ on prenatal genetic counselling and testing’, ‘Parental consent in school-based thalassemia screening’; Developed guidelines, supervised trainees in tele-consultation of patients during COVID-19 pandemic; Identified genetic disorder- alpha thalassemia (1).

12. **Iqbal M.** Supervised graduate research students (4 PhD, 7 MS) on molecular and genetic basis of different ophthalmological disorders; Developed curriculum, delivered lectures on Advanced Molecular Genetics to graduate level students; Appointed as External Examiner, reviewed theses (1PhD, 7 MS/MPhil); Reviewed research articles (3) for Journal of Animal and Plant Sciences (JAPS) and Book Chapter for Frontiers in Molecular Pharming.

13. **Kars ME.** Presented seminar on ‘Characterization of the fine-scale genetic structure of the Turkish Population’, at Molecular Biology and Genetics Department, Bilkent University (Mar. 2021); Used materials from Summit for Teaching Assistantship on Human Genetics and Introduction to Human Biology (2020-2021); Shared knowledge with fellows and undergraduate...
students (7), during their rotations in the genetics laboratory; Used knowledge and resources gained from the Summit for data analysis and presentations to the Thesis committee (2020-2021).

14. **Khant AK.** Speaker on ‘Acute Kidney Injury in Children’, for MS students in Paediatric, organized by Myanmar Paediatric Society (Virtual, Oct. 2020); Examined, diagnosed, treated children (~200) with kidney disease- acute kidney injury due to viper bite/G6PD deficiency/drug overdose/sepsis, prolong shock (30), chronic kidney disease (20), recurrent urinary tract infection (10), nephrotic syndrome (40), steroid resistant nephrotic syndrome (30), rapidly progressive glomerulonephritis (10), systemic lupus erythromatosus (40), vasculitis (15), IgA nephropathy (10); Counselling patients (40) with hereditary renal disease- congenital nephrotic syndrome (5), congenital abnormalities of kidney urinary tract (11), meningomyelocele with neurogenic bladder (7), syndromic child with renal abnormalities (5), Lowe syndrome with renal tubular acidosis (2), focal segmental glomerulosclerosis (8) and several others (5).

15. **LLamos-Paneque A.** Professor, Human Genetics (undergraduate level), School of Dentistry, International University of Ecuador; Updated topics in human genetics; Lectured on clinical genetics to medical students, professionals at Equinoctial Technological University of Ecuador and Army Hospital of Quito; Selected Member, Editorial Board of Molecular Cytogenetics Journal; Summit provided access to research groups to drive medical genetics in low-income countries; Established contact with Ibero-American Network of Neurocutaneous Diseases (Drs. F. Ramos, MT. Acosta, NIH)- presented and attended discussions on neurocutaneous cases and therapeutic advances; Summit improved genetic counseling skills; Implemented genetic counseling service at Maternal Fetal Unit (50 cases/yr) for prenatal patients that tested positive for genetics conditions; Identified cases (2) with structural chromosomes anomalies including supermarket chromosome, detected through molecular cytogenetics studies with German collaborator (Dr. T. Liehr), as service is not available in Ecuador; Evaluated genetic cases (450) through molecular tests - mono gene etiology (100), chromosome (150) through cytogenetic studies, or multifactorial (200); Registered new cases (250) in the Registry of Genetic Diseases, Specialty Hospital No.1 FF. AA, Quito, Ecuador.

16. **López-Star B.** Speaker on ‘Experience as NEI/NIH Fellow’, at Mexico-United States Symposium on Vision Health: Priorities and Opportunities for Bilateral and Regional Engagement in Scientific and Public Health Research (Jan. 2021); Trained junior staff in ophthalmogenetics; Established system to record, monitor patients (63) with inherited retinal Disease; Evaluated, diagnosed, treated ophthalmogenetic cases (8)- Marfan syndrome (4), pigmenyary retinosis (4); Referred patients (3) for medical management to Children’s and Women’s Hospital, Mexico; Diagnosis improved quality of life for patients and their families due to known prognosis and access to appropriate support; None were candidates for treatment, but couples made informed decisions in family planning; Collaborated with the Asociación Para Evitar la Ceguera, Mexico to refer patients with congenital amaurosis of Leber (1).

17. **Luong LH.** Affiliated with National E Hospital, Vietnam (2020); Helped establish lab with focus on genetics of immune disorders- immunodeficiency disorders, auto-inflammatory diseases, and autoimmune disorders.

18. **Mhandire K.** Awarded, Postdoctoral fellowship at Roswell Park Cancer Institute, USA (Mar. 2021); Completed 54 hours of ‘Lockdown Learning Bioinformatics’ (https://www.youtube.com/channel/UC7aizSyonJqZI3O3U4SDTiA).

19. **Monye HI.** Completed online courses on ‘The genomic era: The future of genetics in healthcare’, ‘Genomic technology in clinical diagnostics: Next generation sequencing’, offered by St George’s, University of London (Jan-May 2021), ‘Genomic scenarios in primary care’, offered
by Wellcome Connecting Science (Jan. 2021); These positively influenced patient care; Awarded, Developing Country Eye Researcher Travel Fellowship (2021), by Association for Research in Vision and Ophthalmology; Awarded, Best abstract for ‘The impact of the COVID 19 pandemic on the ophthalmology residency training programme in Nigeria’, at the 52nd Annual Scientific Conference of the Association of Resident Doctors, University College Hospital, Ibadan; Member, Global Genomic Medicine Collaborative (G2MC) Family Health History and Evidence Workgroups, paired-reviewer for a systematic review on evidence of the clinical utility of genomic medicine in patient outcomes.

20. **Mosema KBA**. Lectured in genetics and pediatrics to medical students at Université Protestante du Congo, Kinshasa and Université Uele, Isiro, Haut-Uele Province; Continued establishment of Unit of Sickle Cell Disease (screening, treatment and follow-up), at Biamba Marie Mutombo Hospital, Kinshasa City; Created a WhatsApp group for clinical training in genetics and genomics for medical students of several universities in the DR Congo (UPC, UU, UK etc); Diagnosed cases- holoprosencephaly (1), achondroplasia (1) trisomy 13 (1).

21. **Mushi TL**. Founder, Executive Director of–INNOVATION FOR HEALTH DEVELOPMENT (iHD Foundation), an NGO, to promote community development through evidence-based research, advocacy and lifesaving initiatives that include advancements in genetics and genomics, precision medicine, empowerment of health workforce to reduce maternal-child mortality rates, respecting human rights, in Tanzania.

22. **Mutreja D**. Delivered online lectures on Genomics to faculty and colleagues, lectured post-graduate residents and medical students; Shared resource materials from Summit with medical students, postgraduate residents, colleagues from other specialties; Section Editor (Pathology), Indian Journal of Medical Paediatric Oncology (2020-21); Identified cases (18) by histopathologic /microscopic diagnosis- Lafora disease (1), solitary rectal ulcer syndrome(3), thalassemia (4), sickle cell disease (2), Gaucher disease (1); Improved quality of life by prompt diagnosis and medical management.

23. **Nair L**. Completed Medical Genetics Residency (Oct. 2021); Mentored Genetic Counselling students in basic genetics; Conducted webinars for Pediatricians, Pediatric Neurologists, Developmental Pediatricians, Genetic Counselling students on ‘Genetic disorders’, ‘Genetic tests and their clinical implications’; Conducted OPD, evaluated, diagnosed patients with various genetic disorders, provided genetic testing and genetic counselling; Identified cases- Down syndrome (6), unexplained intellectual disability/ developmental delay (32), recurrent pregnancy loss/ infertility (10); Evaluated and diagnosed rare genetic disorders like CLCN1 gene associated myotonic dystrophy (1), VPS51 gene associated Pontocerebellar hypoplasia type 13 (1), SCN3A gene associated neurodevelopmental disorder (1), pyruvate carboxylase deficiency (1), neurofibromatosis type 1 (1); Started Down syndrome registry at NIMS Medicity, Trivandrum, Kerala.

24. **Okunola O**. Attended Training Fellowship in Stigma research, FIC/NIH (virtual, Jun. 2021), Summit influenced selection; Used experience and materials from Summit to train, mentor medical students, resident doctors, colleagues in medical genetics and genomics, research grant applications; Liaison, working group with paediatric nephrologists to establish congenital kidney disease registry with follow up; Diagnosed patients (20) with inherited kidney diseases (all autosomal dominant polycystic kidney disease), screened, followed up on siblings; Patients and family reported improvement in quality of life due to accurate diagnosis and pathway to care.

26. **Paredes-Moscosso SR.** Submitted abstracts on ‘Abundance of Variants of Uncertain Significance (VUS) found using multi-gene panels in Peruvian patients with hereditary breast cancer’, ‘Modelling with CRISPR/Cas9 the BRCA1 gene ‘Variants of Uncertain Significance’ identified in Peruvian Patients’, and ‘Identification of a *TREM2* mutation (W44X) in a familial case of Alzheimer’s Disease from Lima (Peru) by Whole-Exome Sequencing’, to Asociación Latinoamericana de Genética (2021, Accepted); Selected, mentored a training program, ‘Introduction to Mentoring in Higher Education’, sponsored by CONCYTEC and The British Council (Nov. 2020); Invited reviewer by Pîrlog, R (Summiteer), for ‘Innovation in Science’ call by Asaiah, a Romanian Entrepreneurship Association connecting students from Iuliu Hatleganu University of Medicine and Pharmacy (Romania) with local and international stakeholders (Nov. 2020); Adviser, Multisectoral Working Group for National Policy for the Development of Science, Technology and Technological Innovation (Feb. 2021); Awarded, scholarship for Diploma in Science Communication, funded by CONCYTEC and organized by Universidad Peruana Cayetano Heredia (Mar. 2021); Awarded, training grants for ‘Innovation and Science Diplomacy’, sponsored and organized by The Sao Paulo Innovation and Science Diplomacy School (InnSciD SP) and The World Academy of Sciences (Jul. 2021) and AAAS (Aug. 2021).

27. **Pirlog R.** Awarded, accepted Fulbright scholarship at MD Anderson Cancer Center, Tx, USA (Aug. 2021) for research on role of tumor microenvironment in early development of lung cancer; Pursued an internship in molecular pathology at the University Hospital of Rouen, France (Nov. 2020- May 2021); Started a molecular pathology laboratory; Helped secure funds to sustain a local biobank and its infrastructure for alignment with international consortiums.

28. **Sayed I.** Promoted to Associate Professor of Oro-dental Genetics (Oct. 2020); Identified cases (61) - neurogenetic manifestation (32), skeletal manifestation (18), ectodermal dysplasia patients (8); Referred patients (3) for prosthetic management of ectodermal dysplasia.

29. **Tiong SY.** Integrated and implemented genetic counselling into mainstream prenatal service as part of antenatal and post-natal care (Jun. 2021); Increased awareness among expectant mothers on newborn screening, inherited metabolic disorders (IEM), at Loh Guan Lye Specialists Centre, Penang; Offered IEM genetic tests with an average of 7.5% uptake; Involved in integrating genetic counselling at Breast Cancer Centre, Loh Guan Lye Specialists Centre, Penang (Dec. 2020); Prepared bilingual (English, Chinese) flyers to increase awareness in newborn screening, metabolic disorders, hereditary breast and ovarian cancers; Committee member, Genetic Counselling Society Malaysia (2020-2021); Referred 74 new cases in cancer and paediatrics for genetic counselling.

30. **Tumulak MJR.** Thesis panel member for genetic counseling students (13 students); Genetic counselor for studies on ‘Determination of the prevalence of genetic mutation in high risk Filipino hereditary breast cancer patients’, ‘Genotypic and phenotypic variations of inherited retinal degenerations among Filipino patients in a tertiary hospital’; Invited, resource speaker on ‘The role of genetic counselors in newborn screening programs’ by various forums- Rare Disease Ghana Initiative (virtual, Oct. 2020), 18th National Newborn Screening Convention (virtual, Oct. 2020); 6th International Conference, Board of Genetic Counseling, India (virtual, Jul. 2021); Organized training on genetic counseling for hemoglobinopathies and fatty acid oxidation disorders, Food and Nutrition Research Institute InGest Study (Jul. 2021); Provided pre-test and post-test genetic counseling to patients – breast cancer patients (100), cystic fibrosis patients (50), alpha and beta thalassemia patients (600), and ophthalmology patients (3).

32. **Wiafe SA.** Co-Chair, Developing Nations Working Group- Undiagnosed Disease Network International (2020); Vice Chair, Patient Advocacy Constituent Committee, International Research Consortium for Rare Diseases (Jul. 2021); Panel member, WHO Collaborative Global Network for Rare Diseases; Organized, 3rd Symposium on Birth Defects and Rare Disease (virtual, Mar. 2021); Attended 9th Conference on Rare and Undiagnosed Diseases and Mayo Clinic Science Session (virtual, April, 2021); Speaker, on ‘Improving the standards of care for rare disease patients in Ghana’, IEEPO Changemaker (Jun. 2021); Panelist on ‘Unlocking the power of diversity in genomic data diversity’, African Health Online (virtual, July 2021); Enrolled patients (30) with undiagnosed rare genetic disease in IHOPE Program, Ghana; Enrolled patients (3) with MPS in RDI-Bio Marin Project.

33. **Yadav S.** Fellow in Obstetrics and Gynecology (Advanced ultrasound course), at Mediscan, Chennai, India (Jan. 2021); Delivered lecture on ‘Role of prenatal screening’, for Genetic counsellors organized by Future Medicine Academy (Feb. 2021); Attended, ‘Expert fetal ultrasound- one day extempro course’ (virtual, Jul. 2021), organized by Ukranian Evidence Based Medicine Association; Shared knowledge from Summit in genomics with students from different specialties; Perfored antenatal scans and genetic sonograms (100); Identified fetuses with congenital malformations- congenital diaphragmatic hernia (1), multicystic dysplastic kidneys (2), congenital talipus (1); Provided preconception, antenatal counseling to patients (50).


2018 Batch

35. **Abdulkareem F.** Obtained Doctor of Medicine (MD), from National Postgraduate Medical College of Nigeria (Jan. 2021); Obtained a certificate in Genomic Technologies in Clinical Diagnostics-Molecular Techniques, from St George's, University of London (Dec. 2020); Member, of Accreditation Panels at Eko Medical University of Medicine & Health Sciences and University of Nigeria Teaching Hospital, Enugu (Feb. 2021); Appointed, Curriculum review committee for Biorisk management in Nigerian universities, participated in workshops organized by Sandia National Laboratories, USA, in Abuja (Mar. 2021) and by Faculty of Pathology National Postgraduate Medical College (May 2021); Participated in Laboratory Assessors’ Training Workshop Phase 3 on ISO/IEC for ISO 15189 organized by NINAS Abuja (May 2021); Attended Topics in Human Genetics Summer Course, University of Iowa (Jun. 2021); Did professional assessments for promotions at University of Nigeria Nznukka, Delta State University and University of Sokoto, Nigeria; Attended 15th Annual Scientific Conference of College of Nigerian Pathologists, (Nov. 2020); Attended, Genomic Frontiers Conference (virtual, Jan. 2021); Invited speaker on ‘-Unknown Case’, at Pathology Symposium of African Research for Oncology (virtual, Feb. 2021); Accepted, oral presentation on ‘Risk factors for abnormal cervical cytology in women living with HIV in Nigeria’, virtual AORTIC (Nov. 2021); My NGO, FAJIM Medicare Foundation (https: fajimmf.org.ng) commemorated World Diabetes Day (Nov. 2020), World Cancer Day (Feb. 2021), Hypertension Day (May 2021); Raised awareness and medical outreach for free screening and treatment in colorectal and breast cancer through radio jingles,
television interviews, webinars; Due to COVID no cases of congenital abnormalities were recorded for this period.

36. **Adewole OO.** Appointed, Country Ambassador, Member of Trustee Royal Society of Tropical Medicine and Hygiene (2021); Lectured, trained residents, medical students in genomics and its implications, roles in clinical practice; Shared knowledge from the Summit.

37. **Adeyemo TA.** Mentored new junior faculty (3), College of Medicine University of Lagos and Lagos University Teaching Hospital; Expanded World Bleeding Disorder Registry; Identified, patients (80) with inherited bleeding disorders; Counselling referred patients for care.

38. **Cornejo-Olivas M.** Completed 2-year Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Principal investigator, Coordinator of the First DNA Bank in the public system, Peru; Appointed member, Council for Rare and Orphan Disorders by Peruvian Ministry of Health; Nominated as Executive Committee member for the Pan American Section of the International Parkinson and Movement Disorder Society (MDS); As MDSgene Taskforce member completed systematic review on SCA17; Invited speaker, ERCAL Latin American initiative (Feb. 2021), and Telehealth program, by Ministry of Health (Jun. 2021); Faculty, XXVII Peruvian Congress in Neurology (Jun. 2021); Mentored trainees (3) from Northern Pacific Global Health Research Fellows Training Consortium (FIC/NIH), Global Brain Health Institute, University of California, and Young Members MDS Mentoring Program.

39. **Daich Varela M.** Started, PhD at University College of London, UK (Feb. 2021); Coordinator of Ophthalmic genetics module for post-graduate degree courses in Ophthalmology, University of Buenos Aires, Argentina; Presented talks on ‘Inherited retinal disorders’ at Buenos Aires (Nov. 2020) and in London (Feb. 2021); Continued working on gene therapy clinical trials; Evaluated ophthalmic genetics patients; Identified, diagnosed and managed cases (70- CSNB (10), rod-cone dystrophy (20), cone-rod dystrophy (20), coloboma (5), albinism (5), other rarer syndromic diseases (10); Referred all for counseling and medical management.

40. **Das S.** Completed fellowship in Clinical Genetics, Dept. of Medical Genetics, Christian Medical College Hospital, Vellore, India (2021); Trained medical and nursing students in basic clinical genetics; Volunteered with Organization for Rare Diseases India; Involved in neuromuscular and perinatal clinic for patient management; Identified, diagnosed and counselled patients (~350) with various genetic disorders- juvenile amyotrophic lateral sclerosis 2 (1), autosomal recessive pontocerebellar hypoplasia with microcephaly (1), arginemia (1), ataxia telangiectasia (3), benign familial infantile seizures (2), familial infantile convulsions with paroxysmal choreoathetosis (1), 46XX,t(7;15)(q35;q26.2) (1), biotinidase deficiency (1), Charcot-Marie-Tooth disease, axonal, type 2A2A (3), FSHD 1 (1), DMD (17), achondroplasia (2), cystinuria (1), homocystinuria (2), Glass syndrome (1), Coach syndrome (1), Coffin Lowry syndrome (1), Cohen syndrome (1) amongst others; Referred patients to specialty departments for interdisciplinary management.

41. **Dueñas-Roque MM.** Medical Geneticist, at Genetic Department, Hospital Nacional Edgardo Rebagliati Martin; Professor, Medical Genetics for undergraduate students, Medical School, Universidad San Ignacio de Loyola, Perú; Successfully completed the Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Shared knowledge from Summit with students; Submitted abstracts ‘Deployment of clinical whole genome sequencing in support of more than 1,000 resource-limited patients: Four years of the iHope Program’ to ASHG (2021) and ‘Variable phenotype within dopa responsive dystonia, experience from two Peruvian cases’ to Movement Disorders Meeting (2021); Participated in ICORD/ERCAL group, an international initiative to bridge the gap in rare diseases in Latin America (virtual, May 2021); Enrolled as
young investigator, participated in Evidence Working Group and flagship projects in Global Genomic Medicine Collaborative (G2MC); Summit influenced, helped establish an Undiagnosed Disease Program (UDP) at Hospital Edgardo Rebagliati Martins, EsSalud; Enrolled cases (65 trios) in iHope, Illumina Project for identification of genetic causes of rare diseases in Peru.

42. **Dutta AK.** Established, Paediatric Genetics OPD, at AIIMS Kalyani (India); Summit inspired initiation of a quasi-experimental project for increasing medical graduates’ interest in, Inborn Errors of Metabolism and as partial fulfilment for the Foundation for Advancement of International Medical Education and Research (FAIMER) Fellowship (May 2021); Faculty for the Society of Fetal Medicine Postgraduate Teaching Program, presented on ‘What are aneuploidies, chromosomal abnormalities and non-disjunction’ (Nov. 2020); Resource person for webinar series ‘Genomics in clinical practice’, organized by Bangladesh Physicians (Mar. 2021), presented on ‘Multifactorial vs polygenic & monogenic disorders’, ‘Basic genetic testing and ethical issues’, ‘Genetic diagnosis-database and search engines’, ‘Inborn errors of metabolism case scenarios’, ‘Basic genetic counselling’; Summit curriculum, resource materials helped prepare for these talks; Joined the Education Working Group in Global Genomic Medicine Collaborative (2021).

43. **Dwivedi A.** Created, designed various platforms to spread awareness on genetic disorders among armed forces personnel, families; Conducted webinars on cancer genetics, genetics of renal disorders & lysosomal storage disorders etc.; Used materials from Summit to educate fellow residents, researchers; Invited Speaker on World Rare Disease Day by Takeda, delivered lecture on ‘Care for rare diseases’ (virtual, Feb. 2021); Panelist on ‘Lysosomal storage disorders’, at 6th South Asian symposia (virtual, Apr. 2021); Presented poster on ‘A cross sectional survey to assess the knowledge, attitude and practices towards genetic disorders and testing among non-geneticist clinicians of Indian Armed Forces’, 6th Annual International Conference of the Board of Genetic Counseling India (virtual, Jul. 2021); Member of Global Genomic Medicine Collaboration; Initiated molecular testing including MLPA, Sanger sequencing for common genetic disorders (Thalassemia, Achondroplasia etc.), at AHRR, Delhi; Diagnosed patients (680) with various genetic disorders (Chromosomal, Mendelian and disorders of Genomic imprinting); Knowledge from Summit helped manage patients.

44. **Edem-Hotah J.** Qualified as Foundation Fellow of the College of Nursing and Midwifery, Sierra Leone Postgraduate College of Nursing and Midwifery (May 2021); Invited by the Dean, Faculty of Basic and Health Sciences, Ernest Bai Koroma University of Science and Technology to revise Public Health curriculum (BSc Hons) for inclusion of genetics and genomics pedagogy, research and management of sickle cell disease (Mar-Jun. 2021)- used materials from the Summit; Observed increased awareness for genetics and genomics amongst COMAHS faculty and students, as well as political and technical leaderships of the Ministry of Health and Sanitation and related agencies; Initiated training of nurses and midwives and counselling at the Postgraduate College of Nursing and Midwifery.

45. **Eshete MA.** Work impacted by COVID-19.

46. **Gitaka J.** Established Sickle cell surveillance at Mary Help of the Sick Mission Hospital in Thika, Kenya; Developed, strengthened genomics content in Clinical Medicine Course curriculum, at Mount Kenya University; Referred patients (10) for genetic counselling and medical management; Helped allay anxiety due to known pathway to care.

47. **Kemevor-Asima D.** Expanded, National Rare Disease Registry andCongenital Disease Registry, in collaboration with Mr. S. Wiafe (Summiteer) of Rare Disease Ghana Initiative; Increased
awareness to newborn screening and metabolic diseases, sensitized clinicians and organizers of clinical workshops.

48. **Lertwilaiwittaya P.** Started 4-year Combined Internal Medicine and Medical Genetics Residency Program at University of Alabama at Birmingham, USA (Jun. 2021); Prior physician, at Division of Medical Genetics, Department of Medicine and Siriraj Genomics Laboratory, and Department of R&D, Faculty of Medicine Siriraj Hospital, Mahidol University, Thailand (May 2021); Completed 2-year Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Presented posters on ‘Uptake and utilization of germline BRCA1/2 testing for Thai woman with breast cancer’ and ‘Unfavorable 5-year outcomes in the first familial amyloidosis polyneuropathy case that underwent liver transplantation in Thailand’, at American College of Medical Genetics (2021); Participated in Genomics Thailand (Whole Genome for 50,000 Thais) recruitment process; Co-founded Monday Oncogenetics Clinic, Siriraj Hospital; Increased the cohort of cancer patients (1940) for NGS testing.


50. **Messaoud O.** Completed 2-year Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Used resources from Summit for lecture on Human Genetics, at Faculty of Medicine of Tunis; Young investigator, Global Genomic Medicine Collaborative ([https://g2mc.org](https://g2mc.org)); Reviewer, Medical Hypotheses journal and Topic Editor for the journal Frontiers in Genetics; Organized, participated, session moderator for an international event- Innovation-H Competition ‘Increasing mobility for people with handicaps’ (virtual, Oct. 2020); Participated in virtual trainings and workshops on ‘Facilitating online meetings and workshops’, ‘Integrating digital learning tools in higher education’, and ‘Leadership skills in academia’ (virtual, Nov. 2020); Participated in RDWG-ClinGen workshop, organised by H3Africa and ClinGen (virtual, Feb. 2021);

51. **Mistri M.** Received, Best Young Researcher Award, from International Academic and Research Excellence Committee for exceptional caliber, outstanding performance as academician, researcher, mentor, advisor and leader (2020); Published first largest series of Indian molecular data for various lysosomal storage disorders that include Niemann-Pick disease; As Member, Global Genomic Medicine Collaboration, implemented Undiagnosed and Rare Disease Flagship Project (March 2021).

52. **Muttamba W.** Summit introduced concept, importance of biobanking and sequencing; Involved in multi country initiative to increase biobanking and sequencing capacity in low- and middle-income countries as part of the COVID-19 pandemic response; Identified a case of sickle cell disease, referred for further management.

53. **Naeem MA.** Member, Entry Test Examination Committee for MPhil/PhD at CEMB (Dec. 2020); Supervised PhD students (2), who successfully graduated (2020); Trained MS/MPhil research internees (3) from University of Education (UE), University of the Central Punjab (UCP) and Kinnard College for Women (KCW), at Vision Impairment Lab of CEMB, Lahore, Pakistan.

54. **Nakousi-Capurro N.** Clinical Geneticist at Hospital Carlos Van Buren of Valparaíso, Chile (2020); Faculty at Universidad de Valparaíso as Associate Professor (May 2021); Designed courses, lectured medical students in genetics and clinical genetics; Provided on-site clinical genetics training to neuropsychiatric residents and medical students; Assisted in hosting 3 online courses (a) Skeletal dysplasias (first Latin American course), organized by Chilean Clinical Geneticists, (b) Genomic medicine, organized by the Society of Brazilian Geneticists, GeneOne and DASA educa., (c) Human and mammalian genetics and genomics: The McKusick Short
Course 202, organized by Johns Hopkins University and the Jackson Lab, USA (Apr.-Jul. 2021); Drafted proposal for acquisition of NGS technology for Carlos Van Buren Hospital; Presented online talks on ‘Prenatal testing techniques for chromosomal disorders’, for Regional and in-hospital gynecologists; Established protocols with breast cancer team for prioritizing patients for genetic testing; Collaborated with a telemedicine platform to deliver clinical care to patients and education/information to colleagues in genetics.

55. **Oluwarotimi AC.** Currently the sub-Dean, College of Medicine, University of Lagos; Chairman, Medical and Dental Consultant Association of Nigeria, Lagos University Teaching Hospital Chapter; Member, Dental Therapist Registration Board of Nigeria; National Liaison Officer, Nigerian Dental Association. (2019 –till date). Chairman, Constitution Amendment Committee, Nigerian Dental Association (2019/2020); Coordinator, Brush Twice Daily Programme organized by Unilever Global/ Nigeria. (2008- Till Date).


57. **Savina O.** Presented on ‘Labyrinths of ophthalmogenetics’, at International Applied Science Conference-You need to see your childhood (Ukraine, Jul. 2021); Participated in the TV show Doctor Knows on ‘Heterochromia’ (Ukraine, Jul. 2021); Introduced ophthalmogenetics consultation at Dobrobut Medical Center; Identified ophthalmic genetic cases (18)- aniridia (6), retinitis pigmentosa (4), Stargardt macular dystrophy (3), macular dystrophy with central cone involvement (1), autosomal dominant occult macular dystrophy (1), Leber congenital amaurosis, type 4 (1), Leber hereditary optic neuropathy (1), X-linked retinitis pigmentosa (1); Performed genetic testing, identified mutations, provided genetic counseling.


59. **Utumatwishima JN.**

60. **Wangi KYW.** Enrolled for Ph.D. at Penn State University, USA; Applied for a fellowship at Trinity International University, Illinois, USA; Promotor, National virtual seminar on Development of Genetic Nursing in Indonesia, presented on ‘The Ethical challenges in genetics and genomics nursing in Indonesia’ (Indonesia, Oct. 2021); Awarded Nursing bursary in Genomics and Healthcare, by Wellcome Genome Campus, UK (virtual, Jul. 2021); Developed undergraduate nursing course (3 credits) ‘Trend & current issues in nursing’ includes-Introduction to genetics & genomics medicine, newborn genetic screening & prenatal genetic screening; and nursing role in genetic counseling (Jan - May 2021).

*2017 Batch*
61. **Abad PJ.** Member, Scientific Advisory Board on ‘Interventions using genomics-based strategies towards enhanced nutrition recommendations’, Department of Science and Technology, Food and Nutrition Research Institution (Mar. 2021-present); Served on Global Panel: Genomic practice and research during a pandemic, World Congress of the International Society of Nurses in Genetics (Nov. 2020); Served as abstract reviewer for Congress of the International Society of Nurses in Genetics (Nov. 2021); Served as external reviewer on ‘Investigation of dilated cardiomyopathy in Vietnamese patients using Next-generation sequencing’, Center for Genetics and Reproductive Health, Ho Ch Minh City, Viet Nam; Co-organized, moderated three webinars on telegenetics cancer counseling – focused on genetic counselors making a difference (Nov. 2020), peer supervision in genetic counseling (Dec. 2020), and genetics education for health professionals (Feb. 2021).

62. **Avogbe PH.** Used resources from Summit to design a new course on genetics and genomics databases (Masters’ program), Polytechnic School at the University of Abomey-Calavi, Benin; Delivered new courses in biochemistry and molecular biology; Lectured on Genetic basis of disease, Next-generation sequencing methods, Cancer genetics and genomics to Graduate level students, University of Abomey-Calavi, Benin.

63. **Benítez-Cordero Y.** Delivered lectures in Genetic counseling to graduate level (MS) students, at National Center of Medical Genetics, Venezuela and at ECLAMC Congenital Malformations Conference on Most frequent congenital defects in pregnant adolescents: 10-year study in Cuba; Member of International Clearinghouse for Birth Defects Prevention & Research; Mentored residents (5) in clinical genetics; Evaluated genetic cases (20) of monogenic, chromosomal or multifactorial etiology; Codified cases (2105) for the Registry; Referred cases (5) for genetics analysis and medical management- congenital myopathy (3), spinal muscular atrophy (2).

64. **Casado PL.** Presented talk on ‘Periodontal disease: a risk factor for Sars-Cov-2 infection’, for community engagement in dental practice (virtual, 2021); Trained and supervised graduate students (2 PhD) for research in molecular biology and implant dentistry, at Fluminense Federal University, Brazil; Invited, Editor for Brazilian Dental Journal.

65. **Dhoro M.** Completed 2-year Certificate program in Medical Genetics and Genomics (NHGRI, Jun. 2021); Lectured on genetic basis of disease and gene therapy, diagnosis of genetic disorders, genetic counselling, genomic medicine and pharmacogenetics to Clinical Pharmacologists (medical doctors, postgraduates); Mentored graduate level students (MPhil, DPhil, Clinical pharmacology) on research projects and proposals related to HIV genetic diversity and drug resistance in TB and HIV coinfections; Appointed on Panel, for review of applications for Africa Rapid Grant Fund, Covid-19 Programme Call (2020); Reviewed, edited abstracts for University of Zimbabwe Annual Medical Research Day.

66. **Fatima SS.** Work impacted by COVID-19.


68. **James O.**

69. **Lotz-Esquível S.** Completed 2-year Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Member of the Medical Genetics and Metabolism Department, National Children's Hospital; Provided care for pediatric patients with inborn errors of metabolism and
genetic conditions (4 patients/week); Member, Rare and Orphan Disease Multidisciplinary Clinic, Hospital San Juan de Dios (8 patients/week); Aided in transition of adolescents to adults’ hospital and with transfer of knowledge between hospitals; Expanded prospective patients database for ongoing and future publications (~275 patients); Requested, obtained specialized treatment and laboratory tests for these patients, previously not available in National Health System (San José, Costa Rica); Knowledge acquired through Summit provided opportunity to work at National Referral Centers; Improved quality of life for patients and families by providing optimized genetic services using multidisciplinary approaches.


71. **Mehrez MI.** Promoted to Assistant Professor (Feb. 2021); Presented on ‘3D imaging modalities and craniofacial anomalies’, National Research Centre (Cairo, Feb. 2021); Participated in an online Master’s course ‘Logical Reasoning in Human Genetics’ (Jan. 2021 - present), offered by Columbia Global Centers, Tunisia and Institut Pasteur de Tunis; Reviewed a paper ‘Immunofluorescence studies to dissect the impact of Cockayne syndrome A alterations on the protein interaction and cellular localization’ for Journal of Genetic Engineering and Biotechnology (May 2021); Identified cases (7) clinically- GAPO syndrome (2), Silver Russell (1), Sotos (1), Rubinstein-Taybi (2), Richieri-Costa-Periera (1); Referred cases (4) for oral rehabilitation- GAPO (2), Mandibulacral dysplasia (1), Silver-Russel (1); Dental management of conditions resulted in improved quality of life and psychological well-being of patients.


73. **Nandal R.** Appointed, Teaching faculty for DNB-SS Cardiology course at Pandit Bhagwat Dayal Sharma University of Health Sciences, Rohtak, lectured MBBS students and MDs; Conducted weekly congenital heart diseases clinics; Used facial dysmorphism recognition tools in dysmorphic patients with congenital heart diseases; Established three generation pedigree history as integral tool for patients with cardiomyopathy, sudden death suspected channelopathy, hyperlipidemia and congenital heart disease patients; Identified syndromic children (>50), of thesee-Noonan syndrome (5), Marfan syndrome (1), William syndrome (1), Di Georgi syndrome (1); Referred all identified and suspect syndromic and dysmorphic patients (20) for genetic counselling; Updated congenital heart disease registry.

74. **Otaify GA.** Completed the 2-year Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Introduced, lectured on clinical genetics to undergraduate students, NewGiza University, Egypt; Summit helped in preparation of lectures; Submitted abstract on ‘Five novel mutations in FKBP10 and PLOD2 expanding the molecular and phenotypic spectrum in 10 new patients with rare Bruck syndrome’, to ASHG (2021); Conducted outpatient consultations, management and counseling of patients with genetic diseases, including hereditary bone disorders (8-15 new cases/week), with follow-up for treatment or continued investigation till diagnosis (30); Summit helped in better approach for diagnosis of genetic cases, management plans for patients with metabolic disorders; Lectured on ‘Clinical interpretation of genes and variants for disease causality in WES data’ at Generation Clinics and Labs to expand genetics education and training in the private sector.
75. **Owusu M.** Lead, for diagnostic testing for COVID-19, at the Kumasi Centre for Collaborative Research in Tropical Medicine; Engaged in media interactions on COVID-19.

76. **Petlichovski A.** Completed 2-year Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Included Summit lectures and data in medical school and specialization curricula; Implemented, new Genetic Counseling Service Center at the Institute for Immunobiology and Human Genetics (Macedonia); Performed genetic testing, provided genetic counseling for patients (>20) with rare genetic disorders- Noonan syndrome (3), Rett Syndrome (2), phenylketonuria (1), congenital glaucoma (1), neonatal severe encephalopathy (1), ichthyosis vulgaris (1), SRTD-6 (1), familial Mediterranean fever (4), Gaucher disease (1), Marfan Syndrome (3), Stickler Syndrome (1), Ehlers – Danlos Syndrome (1), hereditary angio edema (3) and for couples (200) with recurrent spontaneous abortion.

77. **Roy S.** Passed, Advanced exam in International Council of Ophthalmology (Mar. 2021); Member, Royal College of Surgeon of Edinburgh and Fellow of International Council of Ophthalmology (2021); Received Fazlul Hoque Memorial Award from Ophthalmological Society of Bangladesh for research paper on ‘Outcome of periocular capillary hemangioma treated with oral propranolol’ (Dec. 2020); Speaker on ‘Retinoblastoma – our endless journey’ and ‘Retinoblastoma today’, at international webinars, organized by Young ophthalmological Society of Bangladesh (Jan. 2021) and L.V. Prasad Eye Institute (May 2021); Speaker on ‘Childhood ocular cancer’ an awareness TV program (Mar. 2021); Arranged CME program on ‘Black fungus infection of eye and recent management protocol’ at the Hospital; Mentored graduate level research students (BSc in Optometry), Institute of Community Ophthalmology, Chittagong University; Identified genetic disorder cases (73)- Retinoblastoma (40), Neurofibromatosis (5), Blepharophimosis syndrome (3), Thyroid ophthalmopathy (25) etc.; Treated patients (55), some were referred for medical management; Patients (42) were satisfied with their outcome.

78. **Sanhueza Díaz C.** Assistant Professor, for MS course ‘Techniques and Methodologies in Genetics’, at Faculty of Medicine, University of Chile; Delivered lectures on Chromosomopathies at University of Chile.

79. **Thakur N.** Developed next generation sequencing for prenatal testing using amniotic fluid for the first time in Nepal; Knowledge gained from Summit was key in validation and analysis; Optimized, performed preimplantation genetic screening (9), clinical exome testing (23) helped diagnose new rare genetic disorders (13); BRCA1/BRCA2 tests were conducted (15) at Nova International Diagnostics Nepal; Conducted Symposium on ‘Rare diseases’ on the occasion of ‘World Rare Disease Day-2021’, featured on [https://rarediseaseday.org/friend/2506](https://rarediseaseday.org/friend/2506) and national newspapers; Conducted symposium, presented on ‘The current state of Down Syndrome, its challenges and future perspectives’, on World Down Syndrome Day (Mar. 2021), Co-organizer for International Brain Research Organization-Asia Pacific Regional Committee Nepal School in association with Neuroscience Society of Nepal, on ‘Understanding Neuroscience and the Spectrum of Neurogenetic Disorders’ (Aug. 2021); Lab coordinator for COVID-19 RT-PCR tests (45,000), trained laboratory scientists (40) on RT-PCR techniques at Bir Hospital; Provided genetic counselling, diagnostic services to patients (>200) with genetic disorders- recurrent abortion (56), Down syndrome (22), Turner syndrome (8), chromosomal anomaly (34), rare genetic diseases (14), beta thalassemia (12), sickle cell anemia (28) and congenital birth defects-Duchenne muscular dystrophy (17), spinal muscular atrophy (7), metachromatic leukodystrophy (4), hemophilia A (6), familial mental retardation syndrome (5), Crouzon syndrome (3), disorder of sexual dysfunction (8), cleft lip and palate (8), at Bir Hospital; Referred patients and their families for appropriate supportive therapies for improvement in quality of life.
80. **Torres-Mejía G.** Presented two talks on ‘History of breast cancer policy in Mexico’ to strengthen advocacy for ‘united’ civil society organizations, organized by the Salvati Foundation, A.C. and ‘Breast cancer: Risk factors and early detection’, to Chronic Diseases INSP staff (Oct. 2020); Mentored graduate students (2 MS) on gene-environment interactions in breast cancer.

81. **Tuncay Yaylacioglu F.** Accepted to the ICO-NEI Ocular Genetics Fellowship Program (August 2021-2022); Completed 2-year Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Presented talk on ‘Challenges in mimicking missense variants’ at a Zebrafish Workshop on creating rare disease specific CRISPR-Cas9 platforms in zebrafish and ensuring their sustainability at Hacettepe University Zebrafish Unit (Mar. 2021).

82. **Vishnoposka SA.** Taught graduate courses ‘Human Genetics’, ‘School of Clinical Genomics’, ‘Human Physiology’ to undergraduates; Presented abstract on ‘Novel pathogenic variants in LHX3, LHX4 and GLI2 identified in pediatric patients with congenital hypopituitarism: From variant calling to variant testing’, at ENDO (virtual, Mar. 2021).

83. **Yousef YA.** Identified, diagnosed, and treated patients with retinoblastoma (22); Referred genetically tested (14) with familial and non-familial retinoblastoma for counseling and medical management; Informed patients negative for germline disease about extremely low risk of transmission to offsprings; Improved quality of life by alleviating stress. (2016 Batch)

84. **Adeoye AM.** Joint appointment with Institute of Cardiovascular Diseases, & College of Medicine, University of Ibadan, Nigeria- Head Molecular Cardiology and Preventive Cardiology unit; Currently Chairman, Medical Advisory Committee /Director of Clinical Services, Research, and Training at the University College Hospital, Ibadan, Nigeria, and the first Teaching Hospital in Nigeria.


86. **Adeyemo WL.** Awarded Fellowship of the Academy of Medicine Specialties of Nigeria (FAcadMedS).

87. **Ariani Y.** Appointed Lecturer, in Pediatrics program for dysmorphology and genetic risk assessment; Supervised postgraduate students (3) on genetics research; Speaker on ‘Host genetic susceptibility towards COVID-19’, at 6th Annual International Conference and Exhibition, Indonesian Medical Education and Research Institute (Nov. 2020); Speaker on ‘Dysmorphology in Inborn error of metabolism’, at 4th Indonesian Pediatric Nutrition and Metabolic Update (Indonesia, Apr. 2021); Presented 4 papers on LSD research at Science and Mathematics International Conference (Indonesia, Oct. 2020), published in American Institute of Physics Conference Proceedings (Apr. 2021); Developed prenatal genetic tests for alpha and beta thalassemia, and spinal muscular atrophy at Morula International IVF Center, Jakarta; Managed cases (~200 post-natal, prenatal and preimplantation) in collaboration with other pediatric subspecialists and OB/GYN Department.

88. **Deniz E.** Secured a postdoctoral fellowship at the Dept. of Oncology, Georgetown University, Washington DC (Jun. 2021); Supervised graduate students (2 MS) in genomics and genomic editing projects (Turkey, 2021); Speaker on ‘Identifying novel gene products conferring drug resistance in cancer chemotherapy’, at 8th Multidisciplinary Cancer Research Congress (Turkey, Jan. 2021).
89. **Ekure EN.** Completed 2-year Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Speaker on ‘Rheumatic heart disease in Nigeria: Insights from a community based survey in Lagos, Nigeria’, at Nigerian Cardiac Society Quarterly Seminar (Oct. 2020); Identified, diagnosed, treated pediatric congenital heart defect cases (149), at Lagos University Teaching Hospital, Nigeria; Counselling patients (131) that needed surgical management; All received medical treatment but only few (42) were surgical managed.

90. **Guio H.** INBIOMEDIC Research Center was certified by the Ministry of Health to test COVID-19 patients. 2021; Completed the Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Started a master course in Precision Oncology (Spain, 2021).

91. **Hussen DF.** Promoted to Associate Professor (Jan. 2021); Collaboration with NIH on manuscript on Syndromes (Williams syndrome, Turner syndrome and Rubinstein Taybi syndrome in diverse populations) aided the promotion; Obtained Diploma in Psychiatric Medicine (May 2021), to assist in psychiatric evaluation of patients with genetic disorders.

92. **Lallar M.** Speaker on ‘Basics of genetics: Non-Mendelian inheritance’ and ‘Genetic techniques: Cytogenetic and molecular’, a class series in association with Bangladesh Society of Medical Genetics (Bangladesh, Oct. 2020); Faculty in workshop on ‘Decision making in Genetics and PGT’, at 16th annual Fertivision Conference, Indian Fertility Society (India, Dec. 2020); Delivered talk on ‘Genetic testing in infertile couples’, FOGSI Webinar (India, Jan. 2021); Faculty in workshop on ‘Panel discussion on aneuploidy screening’ in North Zone YUVA FOGSI (India, Apr. 2021); Appointed faculty for Certificate Programme in Genetic Counselling, Future Medical Academy and Sri Ramachandra Institute of Higher Education and Research, Chennai; Trained students (40) over one year; Presented cases at Society of Foetal Medicine, India; Provided genetic counselling to antenatal patients (>500)- aneuploidy screen risk (200), fetal malformations (150), teratogenic exposure (10), family history /previous child with genetic disease (140); Provided prenatal diagnosis to patients (>50) with various single gene and chromosomal disorders-Thalassemia (25), balanced translocation carriers (5), lysosomal storage disorders (10), spinocerebellar ataxia (2), IEM (8); Continued to maintain birth defect registry for the institution; Mobilized parents and social workers to form support groups for pediatric patients with genetic diseases.

93. **Malasa L.** Completed, Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Appointed, Assistant Lecturer, Department of Paediatrics and Child Health, Hubert Kairuki Memorial University, Dar es Salaam, Tanzania (Feb. 2021); Attended training on ‘Teaching methodology and instructional assessment skills’ conducted by the Centre for Educational Development in Health, Arusha, Tanzania (Oct. 2020); Attended the Massive Open Online Course on ‘Implementation research with a focus on infectious diseases of poverty’, organized jointly by the University of Pennsylvania School of Nursing, the University of Pennsylvania Center for Global Health, USA, and the WHO Special Programme for Research and Training in Tropical Diseases (Oct. 2020).

94. **Okafor FU.** Completed promotion formalities for Professorship in Nursing Science- awaiting external assessment; Supervised nursing student projects.

95. **Roblejo Balbuena H.** Obtained PhD (Apr. 2021); Lectured on Genetic counseling at graduate level (MS), at National Center of Medical Genetics, Venezuela; Mentored residents (2) in clinical genetics. Evaluated genetic cases (19) of monogenic, chromosomal or multifactorial etiology; Registered new cases (4) in the Genetic Diseases' Register of the Pediatric Hospital Center, Havana.
96. **Seven M.** Lectured on ‘Human development and theoretical basis of nursing practice’ in undergraduate nursing program, University of Massachusetts Amherst, College of Nursing (2020-2021); Dissertation advisor for PhD student on ‘The experiences and needs of individuals with a variant of uncertain significance (VUS) on genetic tests for hereditary cancer syndromes: A grounded theory study’, University of Massachusetts Amherst, College of Nursing (2021); Presented seminar on ‘Social network characteristics to improve cancer related health behaviors’, University of Massachusetts, Institute of Diversity Sciences Health Group Seminar (Nov. 2020). Organizing Committee Member, Virtual Congress Wellcome Genome Campus and presented on ‘Genetic knowledge and practice among Turkish nurses: A literature review, nursing, genomics and healthcare’ (virtual, Jul. 2021).

97. **Sirisena ND.** Awarded the Senate Award for Research Excellence at the Inauguration Ceremony of University Annual Research Symposium (Jan. 2021); Awarded first runner-up poster for ‘Design and implementation of a novel pharmacogenetic assay for the identification on CYP2D6*10 genetic variant in a cohort of oestrogen receptor positive breast cancer patients’, presented at Annual Research Symposium of the Faculty of Medicine, University of Colombo (Dec. 2020); Introduced Continuing Professional Development course titled ‘Training Program in Clinical Genetics’ for medical professionals, Faculty of Medicine, Colombo (Jan./Jul. 2021); Diagnosed patients (>250) with various genetic disorders- hereditary cancer (33), dysmorphism/developmental delay (48), thrombophilia (72), haematono-cological diseases (47), thalassemia (10), and congenital birth defects- Down syndrome (35), Turner syndrome (10), ambiguous genitalia (22); Offered genetic counseling, diagnostic services at the Human Genetics Unit, and referred patients for appropriate supportive therapies for improvement in quality of life.

98. **Tibrewal S.** As Head, expanded the genetic clinic at Dr Shroff’s Charity Eye Hospital (SCEH); Conducted regular educational classes (18) for the faculty and trainees at SCEH; Initiated monthly case presentations of clinico-genetic correlation scenarios (Nov. 2020); Prepared one-page ready reckoner for different genetic disorders (I-GENES, Snapshots in Ophthalmology); Received and extended 2021 ARVO Developing Country Eye Researcher Travel Fellowship; Speaker on ‘The role of genetics in pediatric eye disorders’, at Delhi Ophthalmological Society Conference (Mar. 2021); Panelist for ‘Mainstreaming genetic counseling’, at Annual meeting of Board of Genetic Counsellors in India (Jul. 2021); Examined pediatric cases (3364); Counselling patients (328 ) with help of the genetic counselor during COVID pandemic; Performed genetic testing in patients (34) and provided definitive genetic diagnosis- retinoblastoma (15,) retinitis pigmentosa (4), Leber’s congenital amaurosis (4), Leber’s hereditary optic neuropathy (2), Down’s syndrome (4), Aniridia with lens subluxation (1), dominant optic atrophy (1), isolated foveal hypoplasia (1), megalocornea microspherophakia (1), Stargardt’s disease (1).

99. **Uwineza A.** Completed Medical Genetics and Genomics Certificate Program (NHGRI, Jun. 2021); Joined the G2MC Young Investigator Committee (Sept 2020); Participated in Rare Disease Group-ClinGen Workshop, H3Africa (Feb 2021); Participated Pre-Congress Symposium (Mar. 2021), for International Congress of Human Genetics to be held in 2022; Examined suspect cases of genetic disorders (672) referred from other departments at CHUK; Identified genetic disorders (495)- sex development disorder (27), chromosomal abnormalities (433), monogenic syndrome (35); Offered pre and post- test counselling to patients that received genetic tests; These measures alleviated stress, helped improve quality of life of patients and their families.