Scientific Programme Committee

Guillermo Lay-Son, MD, MSc, *Chair*

Clinical Assistant Professor, Pontificia Universidad Catolica de Chile
Head, Genetics Unit, Complejo Asistencial Dr. Sotero del Rio

Dr. Guillermo Lay-Son is a clinician and academic in Medical Genetics at the Faculty of Medicine of the Pontificia Universidad Católica de Chile and Head of the Genetics Unit of the Complejo Asistencial Dr. Sótero del Río, Santiago, Chile. Currently, Dr. Lay-Son is involved in undergraduate teaching as Head of the course of Genetics for Medicine and is a teacher in other courses and internships. In the postgraduate area, Dr. Lay-Son is a teaching tutor for residents of Clinical Genetics program, as well as tutor in charge for residents of other specialties and subspecialties. As a clinical geneticist, Dr. Lay-Son has a special interest in dysmorphology and skeletal dysplasias and works in the Group for the Study of Genetic Abnormalities of the Skeleton (GEAGE). In addition to his work at the University, Dr. Lay-Son works in the care of patients with rare diseases of genetic origin with scarce economic resources in the public health system. He has collaborated on several research projects focused on the study of the molecular basis of genetic diseases, especially in the context of congenital anomalies, osteochondrodysplasias and other genetic disorders such as hereditary breast cancer, cystic fibrosis, 22q11.2 microdeletion and Marfan syndrome. As principal investigator, Dr. Lay-Son contributes to the implementation and evaluation of new technologies such as chromosomal microarrays, next generation sequencing as panels and exome in low-income patients.

Cara Mai, DrPH, *Co-Chair*

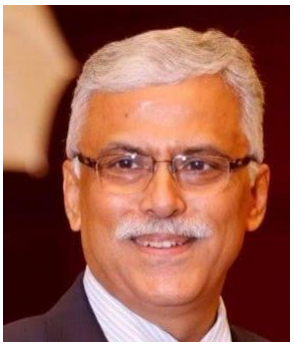
Health Scientist, National Center on Birth Defects and Development
U.S. Centers for Disease Control and Prevention

Dr. Cara Mai is a health scientist with the National Center on Birth Defects and Developmental Disabilities at the U.S. Centers for Disease Control and Prevention (CDC) in Atlanta, Georgia. Dr. Mai has worked at CDC for over two decades and has provided technical and scientific direction to programs working to develop or enhance their birth defects systems and to use the data for public health action. Her areas of interest include surveillance methodology, prevention strategies, data utilization, dissemination and evaluation.

Hannah Blencowe, MD, *Steering Committee*

Assistant Professor
London School if Hygiene and Tropical Medicine

Dr. Blencowe's research is focused on maternal, perinatal and child health: contributing to global estimates of preterm birth, stillbirths and low birthweight. She has also coordinated novel estimates on congenital disorders and disability after neonatal complications. In addition to improving data, she is passionate to explore how data can be used in policy and programmes towards ending preventable deaths. Dr. Blencowe contributes to ongoing UN-led work to improve perinatal data through providing country technical support and supporting further research as part of the UN-IGME Core Stillbirth Estimation Group, the Every Newborn Action Plan metrics group and the Vulnerable Newborns working group.



Rajesh Mehta, MD, *Steering Committee*

Expert Advisor (formerly WHO SEARO)

Dr. Rajesh Mehta is a pediatrician by training. He has been a teacher and trainer in medical college and worked with the World Health Organization for about 16 years. He was the Regional Adviser for Newborn, Child and Adolescent in the South-East Asia Regional Office, New Delhi. He provided strategic advice and support to the eleven countries of the South-East Asia Region to strengthen newborn, child and adolescent health, including birth defects, early childhood development and improving quality of healthcare. Dr. Mehta was a Core Group Member of the global Folate Task Team and has been a strong advocate for food fortification strategies. He has numerous publications including papers in medical journals, chapters in several textbooks and has prepared several national and regional strategies and training packages. He is a recipient of Fellow of Indian Academy of Pediatrics and a Salzburg Global Fellow on Early Child Development..

Salimah Walani, PhD, MPH, MSN, RN, *Steering Committee*

Expert Advisor

Dr. Salimah Walani is a known leader in maternal and newborn health. She is currently the Global Policy & Advocacy Advisor with MiracleFeet and an independent global health consultant, working with WHO, PAHO and March of Dimes. Before joining MiracleFeet, Salimah Walani was Vice President for Global Programs at March of Dimes where she worked with partners to improve birth outcomes in low- and middle-income countries. Salimah led the organization of the last three International Conferences on Birth Defects and Disabilities in the Developing World –Tanzania (2015), Colombia (2017), and Sri Lanka (2020). Dr. Walani has served on numerous technical advisory groups in maternal and newborn health and has been involved in surveillance, prevention, care and advocacy for birth defects. Dr. Walani holds a PhD from New York University, an MPH from Harvard University, and an MSN from Simmons College in Boston.

Jennifer Williams, PhD, MSN, MPH, FNP-BC, *Steering Committee*

Captain, U.S. Public Health Service

Team Lead, Neural Tube Defects Surveillance and Prevention Team

U.S. Centers for Disease Control and Prevention National Center on Birth Defects and Developmental Disabilities, Division of Birth Defects and Infant Disorders

Captain Jenny Williams is a nurse epidemiologist in the National Center on Birth Defects and Developmental Disabilities at the U.S. Centers for Disease Control and Prevention (CDC) in Atlanta, Georgia. She is a certified family nurse practitioner and has a PhD in nursing research and dual master degrees in nursing and in global public health. Captain Williams started at CDC in 2001 as an Epidemic Intelligence Service officer. Her work focuses on birth defects surveillance and birth defects prevention programs, maternal conditions that affect child health and emergency response. Currently, she is the team lead of the Neural Tube Defect Surveillance and Prevention Team at CDC.

Mariana Aracena Alvarez, MD

Clinical Assistant Professor
Pontificia Universidad Catolica de Chile

Dr. Aracena is a Chilean-certified pediatrician and clinical geneticist. She received her Medical degree from the Universidad Chile (1979) and her Medical Doctor Specialist in Pediatrics with a mention in Genetics from the Universidad de Concepción-Chile (1987); and a Master in Medical Sciences, mention in Genetics, from the Universidad de Chile (1995). Dr. Aracena also received her Diploma in Medical Education, School of Medicine, Pontificia Universidad Católica de Chile in 2017 and is an Associate Assistant Professor at Sección de Genética y Enfermedades Metabólicas, División de Pediatría, Pontificia Universidad Católica de Chile. Dr. Aracena's areas of interest are dysmorphology, natural history of genetic or teratogenic syndromes and clinical cytogenetics. Dr. Aracena has developed her professional career in both healthcare and academics and has participated in various scientific publications.

Dr. Eva Bermejo

Director, Institute of Rare Diseases Research (IIER)
ECEMC's Scientific Coordinator, Epidemiology & Clinical Genetics
Research Unit on Congenital Anomalies, Instituto de Salud Carlos III (ISCIII)

Dr. Bermejo is the Director of the Institute for Rare Diseases Research (IIER), Institute of Health Carlos III (ISCIII). She is a tenured Scientific Researcher on Congenital Malformations at IIER-ISCIII; and Scientific Coordinator and Principal Investigator of ECEMC (Spanish Collaborative Study of Congenital Malformations) and its Clinical Network and is responsible for the Epidemiology and Clinical Genetics Section. Dr. Bermejo served at the Executive Committee of ICBDSR for 6 years (Chair in 2015-2017). She was Scientific Director of the Spanish National Biobank of ISCIII. She is accredited in Human Genetics by the Spanish Association of Human Genetics (AEGH). Dr. Bermejo has a Master's Degree in Science (Genetics), and PhD in Science (Genetics), Summa Cum Laude.

Carolina Goic Boroevic, BSW, Mecon

Executive Director, Foro Nacional de Cáncer Fundation
Assistant Professor, *Pontificia Universidad Catolica de Chile*

Carolina Goic has a MS in applied economics and social work, is an assistant professor at the Medicine Faculty of the Pontificia Universidad Católica de Chile and an associate researcher in the Public Policy Center of Universidad Católica. For sixteen years she was a very active politician recognized for her contributions to health issues. As a former senator and congresswoman, she presided and integrated the health and science committees. Today, she is the Executive Director of the Foro Nacional de Cáncer Foundation and a member of the Advisory Council of the Chilesincáncer Foundation. She is also the Founder of "TODAS" foundation that seeks to strengthen female leadership.

Ayesha De Costa, MD

Scientist, Department of Maternal, Newborn, Child and Adolescent Health,
World Health Organization, Geneva

Dr. Ayesha De Costa is a medical doctor and serves as scientist at the Maternal Newborn Child and Adolescent Health and Aging Department at the World Health Organization. She supports research, country programs and guideline development in the area of newborn and child health, including birth defects in low income settings.

A. Pablo Duran MD, MPH, PhD,

B. Regional Advisor in Perinatal Health

Pan American Health Organization, World Health Organization

Dr. Pablo Duran is a Medical Doctor and has a PhD from University of Buenos Aires; he received a MPH from University of El Salvador (Buenos Aires, Argentina) and specialized in Pediatrics at the Children's Hospital Pedro de Elizalde. Dr. Duran is a Fellow in International Nutrition and Nutritional Epidemiology (Cornell University, Ithaca, NY, USA) with more than 20 years of experience. He is an Associate Researcher, Council of Research in Health at the Ministry of Health, Buenos Aires, Argentina. He is the Regional Advisor on Perinatal Health at the Latin American Center for Perinatology (CLAP/SMR), Pan American Health Organization/World Health Organization. Dr. Duran leads activities in the perinatal area and newborns in coordination with other specific areas of Family and Community Health and develops technical cooperation activities in related topics to the health of the newborn and its influence on the course of life.

Gloria Duran, MD

Clinical Associate Professor, Congenital Errors of Metabolism

Pontificia Universidad Catolica de Chile

Dr. Gloria Duran is a Clinical Geneticist and has worked in inherited metabolic disorders for thirty years. She is a staff member in the Genetic and Inborn Errors of Metabolisms Section of Pediatrics Division, Pontificia Universidad Catolica de Chile and Associated Clinic Professor of the Pediatrics Division at the School of Medicine, Pontificia Universidad Católica de Chile.

Fernando Carvajal Encina, MD

Vice President of Neonatology

Chilean Pediatric Society

Dr. Fernando Carvajal is a Neonatologist and Master in Clinical Epidemiology. He is the Vice President of the Neonatology Branch for the Chilean Pediatric Society and Head of the Neonatal Intensive Care Unit, La Serena Hospital, Chile. Dr. Carvajal is also a member of the Neonatology Advisory Group, Healthcare Network Management Division, Ministry of Health. Additionally, Dr. Carvajal is a member of the Editorial Committee of the Evidence in Pediatrics journal of the Spanish Association of Pediatrics. He is also Assistant Professor Faculty of Medicine Universidad Católica del Norte (UCN) and a Member of the academic faculty of the Magister in Public Health UCN.

Boris Groisman, MD

Chair, ICBDSR Executive Committee

Red Nacional de Anomaías Congénitas de Argentina, Administracion Nacional de Laboratorios e Institutos de Salud

Boris Groisman is a medical geneticist and epidemiologist from Argentina. He´s a member of the coordination of the National Network of Congenital Anomalies of Argentina (RENAC), a birth defects surveillance program covering around 300,000 births per year in the country. His areas of expertise are epidemiological analysis, information and communications technology, public health surveillance, e-learning and medical genetics.

Shuchita Gupta, MBBS, MD, MPH, PhD

Medical Officer (Newborn Health), World Health Organization

Dr. Shuchita Gupta is a pediatrician specializing in research and public health. She is currently working as a Medical Officer (Newborn Health) with the Department of Maternal Newborn, Child and Adolescent Health and Aging at the World Health Organization, based in the South East Asia Regional Office in India. Her work focuses on coordinating multi-country research on newborn health, guideline development and programmatic support to the Member States in the Region.

Dr. Kapila Jayaratne, MBBS, MSc, DCH, MD

National Programme Manager, Maternal & Child Morbidity and Mortality, Family Health Bureau, Ministry of Health

Dr. Kapila Jayaratne is a Consultant Community Physician working as the National Programme Manager for Child Morbidity & Mortality at the Family Health Bureau, Ministry of Health, Sri Lanka. Dr. Jayaratne graduated and obtained his master degree and doctorate in Community Medicine from the University of Colombo. He had his post-doctoral training at the University of Melbourne. He pioneered the establishment of a national birth defects surveillance system and reshaped surveillance of stillbirths, infant deaths and child deaths in Sri Lanka. He restructured conducting pathological post-mortems on feto-infant deaths. Dr. Jayaratne, as the conference chair, successfully organized ICBD2020 in Sri Lanka.

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Madhulika Kabra, MBBS, MD

Professor & Officer In-Charge, Genetics Division, Department of Pediatrics, All India Institute of Medical Sciences

Chair, World Health Organization Collaborating Centre

Dr. Madhulika Kabra is a Professor & Officer In-Charge, Division of Genetics, Department of Pediatrics, All India Institute of Medical Sciences, New Delhi, India. She is Chair of the WHO Collaborating Centre for training in Clinical and Laboratory Genetics in Developing Countries. WHO Collaborating Centre is the Quality Assurance Center for Birth Defects Surveillance in the Southeast Asia region. Dr. Kabra is also the Chairperson of the Center of Excellence for Rare Diseases Committee (AIIMS, New Delhi designated as one of the 8 COE for Rare Diseases) and is a member of the panel of experts of the WHO Collaborative Network for Rare Diseases. Her particular areas of interest include intellectual disability, storage disorders, newborn screening and birth defects.

Marcela Lagos, MD

Molecular Genetics Laboratory Director

Pontificia Universidad Catolica de Chile

Dr. Marcela Lagos earned her medical degree from the University of Chile's Medical School. She did her Clinical Pathology Residency at Pontificia Universidad Católica de Chile's Medical School. She then worked in a private clinic laboratory. Dr. Lagos returned to work at the Pontificia Universidad Católica de Chile and completed her Clinical Molecular Genetics fellowship at Harvard University, Boston, MA, USA. Dr. Lagos is currently the Director of the Molecular Genetics Laboratory Red Salud UCCHRISTUS and a Clinical Associate Professor at Pontificia Universidad Católica de Chile.

Mariela Larrandaburu, PhD

President Latin American Network of Human Genetics (RELAGH) Ministry of Public Health of Uruguay, Medicine School Catholic University Uruguay

Dr. Mariela Larrandaburu is a Medical Geneticist, President of Latin American Network of Human Genetics-RELAGH. Her expertise is in clinical genetics, genetic epidemiology and public health. She has a Diploma in Human Genetics in Children and Adults, University of the Republic of Uruguay, MSc and PhD in Genetics and Molecular Biology, Federal University of Rio Grande do Sul, Brazil. She is a Technical Advisor for the Disability and Rehabilitation Program of the Ministry of Public Health of Uruguay, and a Professor at the Catholic University of Uruguay in Medicine School. She was selected to join WHO Technical Working Group on Burden of Birth Defect-TWGBBD.

Dr. Jorge López-Camelo, PhD

Research Director, Center for Medical Education and Clinical Research (CEMIC-CONICET), Buenos Aires, Argentina; Coordinator ECLAMC , Buenos Aires

Dr. Jorge Lopez-Camelo has a PhD in Science, is a specialist in Genetics at the Federal University of Rio de Janeiro, Brazil since 1994. He is a Senior Researcher with CONICET (National Scientific and Technical Research Council of Argentina) and the Director of CEMIC (the Spanish acronym for Center for Medical Education and Clinical Research), a research institute located in CEMIC, supported by CONICET.

Cecilia Mellado, MD

Clinical Associate Professor; Head, Genetics and Congenital Errors of Metabolism Section Pontificia Universidad Catolica de Chile

Dr. Mellado is a Clinical Geneticist, Head of the Genetics and Inborn Disorders of Metabolism Section, Associate Clinical Professor and the Medical Genetics Residency Program Director in the Division of Pediatrics at the Pontificia Universidad Católica de Chile. She is also an attending physician in Clinical Genetics at the UC-CHRISTUS Healthcare Network and at the public hospital Complejo Asistencial Dr. Sótero del Río. She also serves as associate director of the National Congenital Anomalies Registry of Chile (RENACH). Dr. Mellado received her MD from the Universidad de Concepción and completed her residency in Medical Genetics at the Universidad de Chile.

Rosa Pardo, MD

Associate Professor (D), Head, Genetics Section, Department of Medicine Hospital Clínico Universidad de Chile

Dr. Rosa Pardo is the Head, Genetics Section- Department of Medicine, and Pediatrician of Neonatology Unit-Department of Gynecology & Obstetrics, Hospital Clínico Universidad de Chile. Dr. Pardo is a Medical Doctor (1995 National University, Colombia), Pediatrician (1999, Pontifical Xaverian University, Colombia) and Clinical Geneticist (2003, University of Chile). She is associate Professor and Head of the Postgraduate Program on Clinical Genetics at the University of Chile. Dr. Pardo is also the Co-Director of the National Registry of Congenital Anomalies of Chile (RENACH). In this capacity she is working to improve the quality of life for people with rare diseases and disabilities via expanded access to health services.

Jose Poblete, MD

Associate Professor and Chief of the Obstetrics and Gynecology Division
Chilean Society of Obstetrics and Gynecology

Dr. Jose Poblete is an Obstetrician and Gynecologist, MMF specialist. He is an Associate Professor and Chief of the Obstetrics and Gynecology Division at the Pontificia Universidad Catolica de Chile. Dr. Poblete is a past-President of the Chilean Society of Obstetrics and Gynecology.

Neena Raina , PhD

Director, Department of UHC/ Family Health (FGL)

Dr. Neena Raina, Ph.D, is working as Director, Department of UHC/ Family Health (FGL), Health through the Life Course in WHO, Regional Office for South-East Asia Region. She has worked as senior advisor, Maternal Child and Adolescent Health and development in WHO. She has been working in WHO for the past 24 years. Dr. Raina has worked in the areas of reproductive, maternal, newborn, child and adolescent health for the past 42 years. She has published about 200 articles/chapters in journals/books.

Maria Ines Romero, MD, MPH

Programa Nacional de Salud de la Infancia
Ministry of Health, Chile

Dr. María Inés Romero is a Specialist in Pediatrics and Master of Public Health (Universidad de Chile & Harvard University). Her experience includes clinical practice, teaching and research (full professor) at the Universidad de Chile, Universidad Catolica de Chile and Universidad San Sebastian. As an editor and author of books and articles in Chilean and international publications, she has focused on Child and Adolescent development, Health Public Systems and Policies and Curriculum Development in Medical Education. She has also contributed as an international consultant (PAHO/WHO). Currently Dr. Romero serves at the Ministry of Health in the Disease Prevention and Control Division.

Dr. Ignacio Zarante, MD, MSc, PhD

Full Professor, Institute of Human Genetics, Pontificia Universidad Javeriana, Bogotá

Dr. Zarante is a Medical geneticist, master's in Biology and Doctorate in Biological Sciences at the Javeriana University. He has 20 years of experience in Clinical Genetics, 10 years in Forensic Genetics and 22 years in clinical and basic research, as well as in undergraduate and postgraduate teaching. Professor at the Institute of Human Genetics of the Faculty of Medicine of the Pontificia Universidad Javeriana. Dr. Zarante is the President of the Colombian Association of Medical Geneticists; Advisor to the Surveillance and Monitoring Program for children with congenital defects of the Bogotá Health Secretariat; and, Coordinator of the orphan disease care center of the San Ignacio University Hospital.