49th Annual Meeting and Scientific Session of the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR)

Primary Prevention, Surveillance and Management of Birth Defects

November 5th- 8th 2023

Malta

Cavalieri Art Hotel, St Julians

Organized by
ICBDSR
In collaboration with the Malta Congenital Anomalies Registry (MCAR),
Directorate for Health Information and Research, Malta

International Clearinghouse for Birth Defects
Surveillance and Research

DHIR
DIRECTORATE FOR HEALTH INFORMATION AND RESEARCH

VisitMalta
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PROGRAM

	Sunday, November 5th, 2023
14:00-16:00	**Executive Committee Meeting** *(Executive Committee members and ICBD staff only)*
18:00-19:30	**Registration** ICBDSR Annual Meeting
19:30	**Welcome Reception** *at Cavalieri Art Hotel, offered by ICBDSR with the support of Visit Malta Incentives & Meetings*

	Monday, November 6th, 2023 **Meeting Room Babylon**
8:30-9:00	**Registration**
9:00-9:15	**Welcome – Walter Busuttil, Chief Medical Officer, Malta and Boris Groisman, Chair ICBDSR**
9:15-9:30	**ICBDSR Executive Committee Talk** *Boris Groisman, ICBDSR Chair and Lorenzo D Botto, ICBD Director*
9:30-10:10	**J. David Erickson Lecture** – *Pablo Duran, Pan American Health Organization (PAHO)*
10:10-10:30	**Keynote Lecture** Surveillance research and NTDs prevalence estimates around the world: the importance of the WHA67.19 Resolution *Sylvia Roozen, Secretary General, International Federation for Spina Bifida and Hydrocephalus*
10:30-10:45	**EUROCAT Updates** *David Tucker, President EUROCAT Association, CARIS, Swansea University, Wales, UK*
10:45-11:00	**NBDPN Updates**–*Wendy A Nembhard, Chair of NBDPN, University of Arkansas for Medical Sciences, USA*
11:00-11:15	*Coffee break*
11:15-11:30	**From ConcePTION project to sustainable solution for medicines safety assessment during pregnancy and breastfeeding in Europe and beyond** *Miriam Sturkenboom, Department of Data Science & Biostastistics, University Medical Center Utrecht, Netherlands*
11:30-13:00	**Session I – Oral Presentations** **Birth defects surveillance – Methodology I** Global burden of birth defects estimation: methods, progress and selected results Kathleen L Strong Estimating the contribution of different disease groups to the global rare diseases burden using data from the Veneto region RD registry, 2002 through 2019 Monica Mazzucato MALFOVAR Registry / Repository Update Tidhar Steiner Using population-level, real world data to establish cytogenetic testing patterns and evaluate genotype-phenotype correlations in congenital anomalies in England Corinne R Mallinson Current activities of the JRC-EUROCAT Central Registry Agniezska Kinsner-Ovaskainen Validating routinely collected data to improve ascertainment of congenital anomalies to support national registration in England Jennifer M Broughan
13:00-14:15	*Lunch*

14:15-15:45	**Session II – Oral Presentations** **Birth defects surveillance - Descriptive epidemiology** Prenatal findings in craniofacial microsomia using the Alberta Congenital Anomalies Surveillance System, 1997-2019 Mary Ann Thomas Prevalence of skeletal dysplasia in Emilia Romagna, Italy Elisa Ballardini Monitoring an emerging maternal and infant health threat: expanding a birth defects surveillance program to include infants with congenital cytomegalovirus infections Paul A Romitti Diaphragmatic hernia in the Czech Republic. Incidence, prenatal diagnosis and sex ratio Antonin Šípek, Jr Population-based study on epidemiology and outcomes of Congenital Tracheo-Oesophageal Anomalies in South Wales over a 21-year period Margery Morgan Years of potential life lost for children and adults with Congenital Heart Defects, United States, 2007-2017 Wendy N Nembhard
15:45-16:00	*Coffee break*
16:00-16:45	**Poster session** *(meeting room Olympia)*
16:45–18:15	**Session III – Oral Presentations** **Risk factors and health outcomes of birth defects** A brief overview of the current state of knowledge: pregnancy outcome after first trimester exposure to statins Katarina Dathe Analysis of medication consumption during pregnancy in newborns with birth defects in Bogotá, and Cali, 2002-2019 MC León-Sanabria Profile of live births with birth defects in Brazil, their risk factors and associated infant mortality, 2011 to 2020 Julia A Gomes Health outcomes of children born with genetic syndromes in Europe: an undergoing EUROlinkCAT study Alessio Coi Factors associated with the survival of isolated cardiovascular and digestive congenital defects, in the first year of life, in Cuba 2011-2020 Yudelkis Benitez Cordero Radionuclides – Neural Tube Defects – Developmental anomalies – Chornobyl Wladimir Wertelecki
19:30	*Dinner at Cavalieri Art Hotel (included in the registration fee of delegates staying at the hotel)*

	Tuesday, November 7th, 2023 **Scientific session: Surveillance, management, care and transition to adulthood** **Meeting room Babylon**
8:00-8:30	**Registration**
8.30–10:30	**Session I - Surveillance and management** **Moderators:** Neville Calleja, Directorate for Health Information & Research, Ministry for Health, Malta; Charmaine Cordina, Directorate for Health Information & Research, Ministry for Health, Malta Surveillance of Congenital Anomalies in Malta - Setting the scene Miriam Gatt Neonatal mortality due to congenital anomalies in Malta and beyond Merle Wilhelm Evolution of Screening in Malta Mark Cordina The accuracy of antenatal ultrasound screening in Malta: a population-based study Jeremy Borg Myatt Fetal Cardiac Screening in Malta Sarah Darmanin Prenatal Orofacial screening in Malta Maria Christina Tabone Orofacial Clefts in Malta: Prevalence, Impact & Public Health Implications – A 20-Year Analysis Terence Micallef The epidemiology of congenital heart disease in Malta Victor Grech Maternal and Gestational Risk Factors of Congenital Diaphragmatic Hernia Sophie Hackenbruch
10:30–11:00	*Coffee break*
11:00–13:15	**Session II - Care, transition to adulthood and personal experiences** **Moderators:** Victor Grech, Paediatric Department, Mater Dei Hospital, Tal-Qroqq, Malta Miriam Gatt, Directorate for Health Information & Research, Ministry for Health, Malta Breaking the news Ryan Farrugia Several decades later - a look at long-term outcomes of congenital heart defects in Malta Maryanne Caruana The European Reference Networks - Raising the bar for patients with rare diseases and their healthcare providers across Europe James Clark Rare Diseases in Paediatrics : Outsourcing the specialised paediatric services into the Community Resource Centres Chris Sciberras Adult Down Syndrome Clinic Maria Callus Patient / Carer experiences **Closing** – *Yves Muscat Baron – Clinical Chairperson, Department of Obstetrics and Gynaecology*
13:15-14:15	*Lunch*
Afternoon	*Tour-personal payment, optional (afternoon)* *Social dinner offered to delegates by Ministry for Health (accompanying persons personal payment)*

	Wednesday, November 8th, 2023 **Meeting Room Babylon**
9:00-11:30	**ICBDSR Annual Business Meeting** *(ICBDSR members only)*
11:30-11:45	*Coffee break*
11:45 – 12.45	**Session IV– Oral Presentations** **Birth defects surveillance – Methodology II** The JRC-EUROCAT Information System Agniezska Kinsner-Ovaskainen A patient-initiated rare disease registry for South Africa Helen L Malherbe National Congenital Anomaly and Rare Disease Registration Service: how the Congenital Anomaly Official Statistics Report can influence and improve care Kathryn Johnson Semi-automated identification of ventricular septal defects from a clinical data warehouseJ Julie Thomas-Chabaneix
12:45 – 13:30	**Session on ICBDSR Report and collaborative projects**

Posters

(Alphabetical order by corresponding author's last name)

The posters will be shown on Monday 6th November

Meeting room "Olympia"

Using national congenital anomaly registration to evaluate the roll out of the non-invasive prenatal testing (NIPT) in England
Jennifer M Broughan

Infants with Congenital Anomalies of the Kidneys and Urinary Tract (CAKUT) in Malta
Gabrielle de Gray

Congenital anomalies as rare diseases: the importance of genomic linkage for congenital anomaly registries
Kathryn Johnson

The unexplored effects of social and health inequality on adverse outcomes to children born with Spina Bifida: 2010-2020
Kathryn Johnson

Sibling birth defects recurrence in Florida, 2000 to 2019
Russell S Kirby

Prenatal detection of birth defects: pathway of care in obstetric ultrasound services in Bogotá, Colombia
Maria Camila León-Sanabria

Echocardiography evaluation of congenital heart disease with its outcome in a Medical University Hospital of Dhaka, Bangladesh
M Abdul Mannan

Prevention of neural tube defects in Chile: more than two decades of folic acid fortification
Cecilia Mellado

WHO's Resolution calls for micronutrient fortification to prevent spina bifida and other Neural Tube Defects
Rosa Pardo

Epidemiology and risk ractors concerning transposition of the great vessels: A population-based case-control study in Bogotá and Cali, Colombia
Pablo Pineda-Sanabria

Characterization of risk factors for Clubfoot in Bogotá and Cali, between 2002-2020
Esteban Portilla

Polydactyly risk factors in Bogotá and Cali, Colombia between 2002-2020: A case-control study
Lina María Ramírez

Severe congenital protein C deficiency: epidemiology, diagnosis and long-term management
Csaba Siffel

Sex ratio and congenital anomalies in children born after assisted reproduction.
Antonin Šípek Sr

Application of Photon counting CT in the diagnostics of Post mortem fetal malformations for the assessment of skeletal , external skin surface and vasculature anomalies
Tidhar Steiner

Characterization of environmental exposures in the French ATENA registry
Julire Thomas-Chabaneix

Multidisciplinary consultation in fetuses with malformations: prenatal exome experience in a tertiary public hospital
Eduardo Tizzano

Prevalence of Congenital Heart Defects Among Boys with Hypospadias in England
Lee T Watson