International Clearinghouse for Birth Defects Surveillance and Research Monthly Newsletter

July 2022 ***Issue 2022-07***

Remembering Paula Margarita Hurtado Villa (1978-2022)

July 13 of this year marked a significant loss for all of us who work in birth defects surveillance and clinical genetics. On that day we lost our dear friend Paula Margarita Hurtado Villa. She was a Medical Geneticist and Bioethicist, and professor in the Department of Basic Health Sciences at the Javeriana University in Cali, Colombia.

Paula at the 2016 ICBDSR Annual Meeting in Bratislava

Paula was the first director of that department and helped develop one of the most important health faculties in Colombia. Paula created the Birth Defects Surveillance Program in Cali in 2010, which later became the Program for the Prevention and Surveillance of Orphan Diseases and Birth Defects (PREVERDEC). The program was a member of ECLAMC and, since 2015, a full member of the International Clearinghouse for Birth Defects Surveillance and Research. She authored more than 20 scientific articles and 3 chapters of academic books.

Since her graduation, Paula practiced medical genetics at the Imbanaco Clinic in Cali, serving hundreds of patients from all socio-economic levels. In her work as an epidemiologist, Paula strongly supported the concept of triple surveillance and broadening the monitoring of birth defects to other rare diseases.

She was a wonderful friend, travel companion, and a kind soul; a dedicated physician with an extraordinary sense of humanity, a keen mind that developed creative and innovative solutions, a trustworthy and hard worker; and a beloved daughter, sister, aunt, loving wife.

All who knew her remember how her smile lit the room.She did so much in so little time. She taught us great things – things that we can now apply in her honor. She will be very much missed.

ICBDSR ANNUAL MEETING

The **48th ICBDSR Annual Meeting** will be heldfrom **September 18thto 21st, 2022** in Bologna, Italy, at Hotel Relais Bellaria. It will bean in-person meeting with the possibility to join the scientific session by live stream. Please, fill in the application form to attend the Scientific Session by leave stream.

presentation format, either in person or virtually (pre-recorded). The finalagenda of the meeting is under preparation.

Information on hotel reservation and registration: http://www.icbdsr.org/48th-icbdsr-annual-meeting/
Please note: deadline for hotel reservation expired on June 20, 2022. Late reservations are still accepted upon availability of rooms.

Venues of future Annual Meetings:
2023: Malta
2024: Prague (Czech Republic)

ICBD corner: News and comments from the International Centre on Birth Defects (ICBD)

Birth Defect Training Case Studies - Updated. Thank you to those who have expressed an interest in trying out the case studies. Based on feedback we received, we at ICBD we have updated the three extended case studies on birth defect identification, description, reporting, and coding.

We are preparing to send these out to the programs who volunteered to pilot test them – send us an email if you want to be added to the list. What we ask in return is a timely review with feedback and evaluation.

The case studies expose the trainees to concepts such as birth defect sequence (e.g. spina bifida sequence) and non-random associations (VACTERL association), and to basic information about some common risk factors, genetic (e.g., trisomy 18, deletion 22q11) and otherwise (maternal diabetes, folic acid insufficiency).

The target audience is clinical providers at the point of care (e.g., nurses, junior doctors, medical students) as well as abstractors and a range of surveillance staff.

Methodologically, the case studies highlight the value of checklists and clinical clues for high quality activities, and are aligned with the content of the Quick Reference Handbook and the Manual for Program Managers (second edition), recently issues by WHO/CDC/ICBDSR.

For the ICBD,
Lorenzo Botto, ICBD Director

What's Going on

International Conference on Birth Defects and Disabilities in the Developing World – ICBD 2023

"Accelerating action for birth defects and disabilities: surveillance, prevention, management and family-centered care" – March 1-4, 2023, Santiago, Chile
The abstracts submission is now open. Full information at https://www.marchofdimes.org/icbd.aspx

2022 Clubfoot Africa Conference

Registration is now open for the 2022 Clubfoot Africa Conference 'Moulding the Future'.
The three day event organised by Steps Charity NPC in partnership with the South African Paediatric Orthopaedic Society will take place in Cape Town, South Africa and virtually on 21 – 23 November.

International Federation for Spina Bifida and Hydrocephalus (IFSBH)

Grand Rounds for Clinicians working with SpinaBifida and Hydrocephalus- August 12, 2022
This session will be on #FetalSurgery with Dr. Agnieszka Pastuszka (Urologist at Medical University of Warsaw, Poland).
Registrations are now open here

A selection from the recent literature, and recommendations from readers

All readers are kindly invited to contribute to this section of the Newsletter, sending their suggestions to centre@icbdsr.org by the 1st Friday of the month

Outcomes and Longitudinal Studies

Baldacci S, Santoro M, Pierini A, Mezzasalma L, Gorini F, Coi A. Healthcare Burden of Rare Diseases: A Population-Based Study in Tuscany (Italy). Int J Environ Res Public Health. 2022 Jun 21;19(13):7553. doi: 10.3390/ijerph19137553. PMID: 35805212; PMCID: PMC9265803.

Urhoj SK, Tan J, Morris JK, Given J, Astolfi G, Baldacci S, Barisic I, Brigden J, Cavero-Carbonell C, Evans H, Gissler M, Heino A, Jordan S, Lutke R, Odak L, Puccini A, Santoro M, Scanlon I, de Walle HEK, Wellesley D, Zurriaga Ó, Loane M, Garne E. Hospital length of stay among children with and without congenital anomalies across 11 European regions-A population-based data linkage study. PLoS One. 2022 Jul 22;17(7):e0269874. doi: 10.1371/journal.pone.0269874. PMID: 35867669; PMCID: PMC9307180.

Prevention / Risk Factors

Kancherla V, Roos N, Walani SR. Relationship between achieving Sustainable Development Goals and promoting optimal care and prevention of birth defects globally. Birth Defects Res. 2022 Jul 1. doi: 10.1002/bdr2.2055. Epub ahead of print. PMID: 35776686.

Begashaw B, Tariku Z, Berhane A. Preconception of folic acid supplementation knowledge among Ethiopian women reproductive age group in areas with high burden of neural tube defects: a community based cross-sectional study. J Nutr Sci. 2022 Jun 20;11:e48. doi: 10.1017/jns.2022.32. PMID: 35836692; PMCID: PMC9241059.

Anele CR, Goldani MZ, Schüler-Faccini L, da Silva CH. Prevalence of Congenital Anomaly and Its Relationship with Maternal Education and Age According to Local Development in the Extreme South of Brazil. Int J Environ Res Public Health. 2022 Jul 1;19(13):8079. doi: 10.3390/ijerph19138079. PMID: 35805738; PMCID: PMC9265685

Zhang W, Yang Y, Liu Y, Zhou L, Yang Y, Pan L, Ba Y, Wang R, Huo Y, Ren X, Bai Y, Cheng N. Associations between congenital heart disease and air pollutants at different gestational weeks: a time-series analysis. Environ Geochem Health. 2022 Jul 23. doi: 10.1007/s10653-022-01315-8. Epub ahead of print. PMID: 35869374.

Finn J, Suhl J, Kancherla V, Conway KM, Oleson J, Sidhu A, Nestoridi E, Fisher SC, Rasmussen SA, Yang W,

and congenital diaphragmatic hernia. Birth Defects Res. 2022 Jun 27. doi: 10.1002/bdr2.2059. Epub ahead of print. PMID: 35757961.

Wang Y, Wang L, Yang Z, Chen F, Liu Z, Tang Z. Association between perinatal factors and hypospadias in newborns: a retrospective case-control study of 42,244 male infants. BMC Pregnancy Childbirth. 2022 Jul 20;22(1):579. doi: 10.1186/s12884-022-04906-6. PMID: 35858860; PMCID: PMC9301865.

Genetics and Genomics

Awotoye W, Mossey PA, Hetmanski JB, Gowans LJJ, Eshete MA, Adeyemo WL, Alade A, Zeng E, Adamson O, Naicker T, Anand D, Adeleke C, Busch T, Li M, Petrin A, Aregbesola BS, Braimah RO, Oginni FO, Oladele AO, Oladayo A, Kayali S, Olotu J, Hassan M, Pape J, Donkor P, Arthur FKN, Obiri-Yeboah S, Sabbah DK, Agbenorku P, Plange-Rhule G, Oti AA, Gogal RA, Beaty TH, Taub M, Marazita ML, Schnieders MJ, Lachke SA, Adeyemo AA, Murray JC, Butali A. Whole-genome sequencing reveals de-novo mutations associated with nonsyndromic cleft lip/palate. Sci Rep. 2022 Jul 11;12(1):11743. doi: 10.1038/s41598-022-15885-1. PMID: 35817949; PMCID: PMC9273634.

Li J, Yang W, Wang YJ, Ma C, Curry CJ, McGoldrick D, Nickerson DA, Chong JX, Blue EE, Mullikin JC, Reefhuis J, Nembhard WN, Romitti PA, Werler MM, Browne ML, Olshan AF, Finnell RH, Feldkamp ML, Pangilinan F, Almli LM, Bamshad MJ, Brody LC, Jenkins MM, Shaw GM; NISC Comparative Sequencing Program; University of Washington Center for Mendelian Genomics; National Birth Defects Prevention Study. Exome sequencing identifies genetic variants in anophthalmia and microphthalmia. Am J Med Genet A. 2022 Aug;188(8):2376-2388. doi: 10.1002/ajmg.a.62874. Epub 2022 Jun 18. PMID: 35716026; PMCID: PMC9283271.

Infections, including COVID-19

Hromić-Jahjefendić A, Barh D, Ramalho Pinto CH, Gabriel Rodrigues Gomes L, Picanço Machado JL, Afolabi OO, Tiwari S, Aljabali AAA, Tambuwala MM, Serrano-Aroca Á, Redwan EM, Uversky VN, Lundstrom K. Associations and Disease-Disease Interactions of COVID-19 with Congenital and Genetic Disorders: A Comprehensive Review. Viruses. 2022 Apr 27;14(5):910. doi: 10.3390/v14050910. PMID: 35632654; PMCID: PMC9146233.

Latos-Bieleńska A, Marcus E, Jamry-Dziurla A, Rankin J, Barisic I, Cavero- Carbonell C, Den Hond E, Garne E, Genard L, João Santos A, Lutke LR, Dias CM, Neergaard Pedersen C, Neville A, Niemann A, Odak L, Páramo-Rodríguez L, Pierini A, Rissmann A, Morris JK. COVID-19 and children with congenital anomalies: a European survey of parents' experiences of healthcare services. BMJ Open 2022;12:e061428. doi:10.1136/bmjopen-2022-061428

Recommendations from readers

No recommendations this month

News from ICBDSR Executive Committee and ICBDSR Members

Publication of REMERA data on malformations in 2020

The Register of malformations in Rhône-Alpes (Remera) has published on its website its **updated data** for the period **2006-2020** for its surveillance territory (**Ain, Isère, Loire, Rhône**) .

Online Self-Paced Course on Birth Defect Surveillance and Prevention

The online course remains available for interested professionals. The course has been developed by the International Centre on Birth Defects (ICBD Centre) and supported in part by funding from the National Center on Birth Defects and Developmental Disabilities, US Centers for Disease Control and Prevention, through Agreement with the Task Force for Global Health. The course is designed for clinicians, epidemiologists, public health professionals, and anyone interested in understanding birth defects and improving their prevention and care. The course includes videos, quizzes, a discussion forum, and publications/resources. It is available in

English and Spanish. Additional information is available at http://www.icbdsr.org/online-self-paced-course-on-birth-defect-surveillance-and-prevention/. Please, register through the online registration form.

Meetings and Events

A list of future Meetings & Events is available at http://www.icbdsr.org/meetings/.
All readers are kindly invited to contribute to this section of the Newsletter, sending information about future meetings/events at *centre@icbdsr.org*

World Birth Defects Day – WBDD, March 3rd

Continue to raise awareness on birth defects throughout the year!
Join the WBDD partners at https://www.worldbirthdefectsday.org/application-form/

Email info@worldbirthdefectsday.org
Follow @worldbdday on Twitter https://twitter.com/WorldBDDayand
Instagram worldbddayhttps://www.instagram.com/worldbdday/.
Join WBDD page on Facebook https://www.facebook.com/WBDDay/.

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