Monthly Newsletter

April 2024

Issue 2024 -04

2024 ICBDSR ANNUAL MEETING

The meeting will celebrate 50 years of ICBDSR (1974 – 2024) and 60 years of congenital anomaly registration in Czech Republic (1964 – 2024).

The **50th Annual Meeting of the ICBDSR** will take place in Prague, Czech Republic, from **September 1 to 4, 2024** at the Hermitage Hotel. Information on hotel reservation and registration: 50th ICBDSR Annual Meeting – ICBDSR. Please note the deadline of **June 30, 2024,** for hotel booking. Reservations can be made by the "Reservation form" available in the ICBDSR website.

General plan of the meeting:

Sun, Sept 1	Executive Committee (EC) Meeting *(restricted to EC members)* Pre-Annual Meeting Workshop - Enhancing Birth Defect Surveillance: Interactive Case Studies for Quality Improvement *Welcome Reception at Hermitage Hotel, offered by ICBDSR*
Mon, Sept 2	Scientific Sessions – Meeting Room Verdana *Dinner at the Hermitage Hotel, included in the registration fee of delegates staying at the hotel*
Tue, Sept 3	ICBDSR Business Meeting and Scientific Session – Meeting Room Verdana (morning) Scientific Session – Meeting Room Verdana*Tour - personal payment, optional (afternoon), more details to come soon*
Wed, Sept 4	Joint Workshop with Czech Society of Medical Genetics and Genomics *Departure (afternoon)*

Please note: we have added a Pre-Annual Meeting Workshop on Sunday, September 1st. See the ICBD Corner in the March newsletter for details. There will be no charge to attend the workshop. Registration will be required, and more information will be circulated in the coming months.

Abstracts Submission:

We invite you to submit abstracts for oral and/or poster presentation at the 50th ICBDSR Annual Meeting Scientific Symposium, this will be an in-person meeting.

The theme for the 50th Annual Meeting Scientific Symposium will be:

Primary Prevention, Surveillance and Management of Birth Defects

Abstract topics should fit one of the following thematic areas:

- Birth defect surveillance and descriptive epidemiology (including surveillance methodology)
- Risk factors and primary prevention for birth defects
- Rare diseases
- Covid-19 and impact on reproductive outcomes, birth defects surveillance, prevention, and care
- Molecular genetics / molecular epidemiology / epigenetics and birth defects
- Prenatal screening, early pregnancy / birth / postnatal outcomes

- Psychosocial impacts of birth defects, quality of life, education, and other related issues
- Management and care for birth defects
- Patient and caregiver experiences
- Historical perspective of surveillance of congenital anomalies

Authors of accepted **oral sessions** should aim at 10 minutes presentation with 3-5 minutes questions.

Abstract format
The abstract header should include the title, author(s), and affiliation(s). Acronyms and abbreviations should be spelled out on first use. The abstract text should be structured using the following sub-headings: Background/Objectives, Methods, Results, and Discussion.

Abstract submission information
The abstract is to be submitted using the electronic abstract submission form in the ICBDSR website.
Deadline for submission of abstracts is **May 1, 2024.**

Abstract review and notification
The Annual Meeting Committee will peer-review the abstracts and will make the final decisions regarding acceptance of abstracts and designated presentation format. You will receive an email of notification regarding acceptance of abstracts by **June 15, 2024.**

Submit your abstract HERE.

ICBD corner: 29 April, Undiagnosed Disease Day

April 29, Undiagnosed Diseases Day, is a day to celebrate, reflect, and recommit to action – a day meant to shine the light on the community with rare and undiagnosed diseases, a day to ensure this community is **seen and heard**.

Rare and ultrarare diseases are everywhere and affect 1 in 13 people – each condition may affect a few or even a handful of people, but collectively (7000+ of them identified so far), they change the lives at least **300 million people** worldwide.

Every rare disease starts as undiagnosed. Providing people with a rare disease diagnosis can be difficult – it requires specialized skills, tools, and teams - but it is critical to help those affected with better care and treatments.

Still, many families go through a **diagnostic odyssey** spanning years, with countless visits and tests, at significant personal and societal cost.

As we recognize Undiagnosed Diseases Day, consider a **local activty** (a talk, a social post) to spread awareness and celebrate the courage, persistence and resilience of those living with an undiagnosed disease along with their dedicated caretakers and supporters.

One such event on April 29 is organized by the Undiagnosed Diseases Network Foundation (UDNF) in collaboration with the Wilhelm Foundation and Undiagnosed Diseases Network International (UDNI), and it will be hosted at Harvard Medical School as a virtual event. The day includes live presentations from global experts on undiagnosed diseases. You can register and participate via this link: https://www.undiagnosed-day.org/

Collaborative ICBDSR projects: Gastroschisis paper just published

Feldkamp ML, Canfield MA, Krikov S, Prieto-Merino D, Šípek A, Lelong N, Amar E, Rissmann A, Csaky-Szunyogh M, Tagliabue G, Pierini A, Gatt M, Bergman JEH, Szabova E, Bermejo-Sánchez E, Tucker D, Dastgiri S, Bidondo MP, Canessa A, Zarante I, Hurtado-Villa P, Martinez L, Mutchinick OM, Camelo JL, Benavides-Lara A, Thomas MA, Liu S, Nembhard WN, Gray EB, Nance AE, Mastroiacovo P, Botto LD. Gastroschisis prevalence patterns in 27 surveillance programs from 24 countries, International Clearinghouse for Birth Defects Surveillance and Research, 1980-2017. Birth defects research 2024; 116(2): e2306. DOI: 10.1002/bdr2.2306

https://onlinelibrary.wiley.com/doi/10.1002/bdr2.2306

News from the ICBDSR and the World: a busy quarter of the year ahead

The second quarter of the year provides many opportunities to advocate for communities and families impacted by birth defects. Consider putting together local activities and joining our many partners: we are stronger together.

- **April 7** is **World Health Day –** It is celebrated annually, and each year draws attention to a specific health topic of concern to people all over the world. The date of 7 April marks the anniversary of the founding of WHO in 1948.
  Website: https://www.who.int/campaigns/world-health-day

- **May 13-15** is the **World European Congress on Pediatrics and Neonatal Conference –** It will be held in Rome, Italy. The program will grab on both international and regional speakers who are experts in neonatal and pediatric knowledge. Topics would be focused on common and updated scientific knowledge with the theme of "Transforming the Future of Pediatric and Neonatal Medicine: Advancements, Discoveries, and Innovations".
  Website: https://kindcongress.com/event/world-european-congress-on-pediatrics-and-neonatal-european-pediatrics-2024/
  Email: europeanpediatricsciencie@outlook.com

- **June 22-26** is the **Society for Birth Defects Research and Prevention's 64th Annual Meeting: Addressing Disparities in Birth Defects Research and Prevention.** The robust scientific program represents active, timely research areas of high relevance to public health and significant usefulness to researchers in the fields of birth defects research and surveillance and related fields.
  Meeting website: https://www.birthdefectsresearch.org/meetings/2024/

Recently published: recent papers selected at the ICBD

To find the paper online, use the DOI for a browser search. If interested but unable to get the full text of the papers, please email us and we will see what we can do to help.

Undiagnosed Diseases (special topic: 29 April is Undiagnosed Disease Day)

International Undiagnosed Diseases Programs (UDPs): components and outcomes. Curic E, Ewans L, Pysar R, Taylan F, Botto LD, Nordgren A, Gahl W, Palmer EE. Orphanet J Rare Dis. 2023 Nov 9;18(1):348. doi: 10.1186/s13023-023-02966-1. PMID: 37946247; PMCID: PMC10633944.
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC10633944/

Cluster analysis and visualisation of electronic health records data to identify undiagnosed patients with rare genetic diseases. Moynihan D, Monaco S, Ting TW, Narasimhalu K, Hsieh J, Kam S, Lim JY, Lim WK, Davila S, Bylstra Y, Balakrishnan ID, Heng M, Chia E, Yeo KK, Goh BK, Gupta R, Tan T, Baynam G, Jamuar SS. Sci Rep. 2024 Mar 1;14(1):5056. doi: 10.1038/s41598-024-55424-8.PMID: 38424111

Lived experiences of genetic diagnosis for rare disease patients: a qualitative interview study. Modelhart A, Sturz D, Kremslehner L, Prainsack B. Orphanet J Rare Dis. 2024 Feb 14;19(1):68. doi: 10.1186/s13023-024-03058-4.PMID: 38355619

A qualitative evaluation of patient and parent experiences with an undiagnosed diseases program. Siebold D, Denton J, Hurst ACE, Moss I, Korf B. Am J Med Genet A. 2024 Feb;194(2):131-140. doi: 10.1002/ajmg.a.63417. Epub 2023 Sep 26.PMID: 37750194

Risk factors: recent reviews

Risk of Major Congenital Malformations and Exposure to Antiseizure Medication Monotherapy. Battino D, Tomson T, Bonizzoni E, et al. *JAMA Neurol.* Published online March 18, 2024. doi:10.1001/jamaneurol.2024.0258.

First Trimester Use of Buprenorphine or Methadone and the Risk of Congenital Malformations. Suarez EA, Bateman BT, Straub L, et al. *JAMA Intern Med.* 2024;184(3):242–251. doi:10.1001/jamainternmed.2023.6986

Maternal Diabetes and Overweight and Congenital Heart Defects in Offspring. Turunen R, Pulakka A, Metsälä J, et al. *JAMA Netw Open.* 2024;7(1):e2350579. doi:10.1001/jamanetworkopen.2023.50579

Outcomes

Congenital anomalies and predisposition to severe COVID-19 among pediatric patients in the United States. Goodman, L.F., Yu, P.T., Guner, Y. *et al. Pediatr Res* (2024). https://doi.org/10.1038/s41390-024-03076-9

Bilateral Renal Agenesis—Interpreting the RAFT Trial. Gyamfi-Bannerman C, Marc-Aurele K, Mestan K. *JAMA.* 2023;330(21):2059–2060. doi:10.1001/jama.2023.22747.

Diagnosis

Molecular Diagnostic Yield of Exome Sequencing in Patients With Congenital Hydrocephalus: A Systematic Review and Meta-Analysis. Greenberg ABW, Mehta NH, Allington G, Jin SC, Moreno-De-Luca A, Kahle KT. *JAMA Netw Open.* 2023;6(11):e2343384. doi:10.1001/jamanetworkopen.2023.43384

Exome and genome sequencing in a heterogeneous population of patients with rare disease: Identifying predictors of a diagnosis. Pucel J, Briere LC, Reuter C, Gochyyev P; Undiagnosed Diseases Network; LeBlanc K.Genet Med. 2024 Mar 1:101115. doi: 10.1016/j.gim.2024.101115. Online ahead of print.PMID: 38436216

Meetings and Events: A list of future Meetings & Events is available at http://www.icbdsr.org/meetings/.

International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR)
Website: www.icbdsr.org
ICBDSR training platform: www.icbdsrtraining.org
Facebook: www.facebook.com/ICBDSR
Instagram: https://www.instagram.com/icbdsr/
Twitter:@icbdsr https://twitter.com/ICBDSR