



Monthly Newsletter

April 2026

Issue 2026 – 04

ICBDSR 2026 Annual Meeting – [REGISTRATION NOW OPEN!](#)



The 52nd Annual Meeting of the ICBDSR will be held **September 13-16th at the University Hospital in Magdeburg, Germany.**

Call for Abstracts – Don't Miss the Deadline!

We are pleased to announce that the call for abstracts for the ICBDSR 2026 is now open. Submission will be accepted online. Please visit the [ICBDSR 2026 Annual Meeting Website](#) for the abstract guidelines and the link to submit.

Abstract submissions are due by April 30, 2026.

The Abstract Themes for this year's meeting include:

- i. Birth defect surveillance and descriptive epidemiology (including surveillance methodology).
- ii. Risk factors for birth defects.
- iii. Rare diseases.
- iv. Molecular genetics / molecular epidemiology / epigenetics and birth defects.
- v. Prenatal screening, early pregnancy / birth/newborn screening and postnatal outcomes.
- vi. Psychosocial impacts of birth defects, quality of life, education, patient perspectives and related topics.
- vii. Birth defect surveillance and research in low- and middle-income countries.

Lisi-Hurtado Award Call for Nominations 2026

At this year's Annual Meeting in Germany, we will be awarding the Lisi-Hurtado Award. For information on the award and eligibility, please visit the [ICBDSR website](#).

Applications - The application package should be sent electronically to the ICBDSR Headquarters (centre@icbdsr.org), and should describe in detail the candidate's accomplishments, current research interests and projects, and why the nominator believes the candidate's career will promote knowledge of birth defects. Please read the full requirements and eligibility at the link above.

The winner will receive a plaque and a travel stipend to the Annual Meeting of the Clearinghouse.

Deadline for submitting applications is June 30, 2026.

Distinguished Service Award Call for Nominations 2026

At this year's Annual Meeting in Germany, we will be awarding the Distinguished Award. For information on the award and eligibility, please visit the [ICBDSR website](#).

Applications - The application package should be sent electronically to the ICBDSR Headquarters (centre@icbdsr.org), and should describe in detail the candidate's accomplishments, current research interests and projects, and why the nominator believes the candidate's career will promote knowledge of birth defects. Please read the full requirements and eligibility at the link above.

The winner will receive a plaque and a travel stipend to the Annual Meeting of the Clearinghouse.

Deadline for submitting applications is June 30, 2026.

Nominations Committee – Open position

We are also seeking expressions of interest for two members to join the ICBDSR Nominations Committee. Dr. Miriam Gatt, Malta and Dr. Jorge Lopez Camelo are finishing their terms with our sincere thanks.

The Nominating Committee, shall be composed of the ICBBD director, the Immediate Past Executive Chair and 2 Program Directors. The committee is responsible for proposing members for the Executive Committee. The term is 3 years. If you are interested in serving on the Nominating Committee, please send your name to centre@icbdsr.org for the consideration of the Executive Committee.

Nominations are due by June 30, 2026.

ICBDSR Birth Defect Report: Data Submission Schedule

Upcoming deadlines: May 31, 2026 | April 30, 2027 (and annually thereafter)

As program leaders and Clearinghouse members, accurate and timely data sharing is a shared responsibility. In response to member feedback, a clear two-year submission timeline has been established to support predictable planning. Timely submission allows for data cleaning, query resolution, and report preparation in advance of the annual meeting.

- For 2026, the list and definitions of anomalies remain unchanged from last year. Harmonization efforts will continue. The 2026 data submission form is available on the Clearinghouse website: [2026 Birth Defect Report Data Submission Form](#). Please review the "ReadMe" section carefully. A short video is also included. If you have questions, contact us early.
- Members with expertise in programming or web development are invited to contribute to the 2026 report preparation – please contact headquarters.
- For reference, the ICBDSR 2024 report is available [HERE](#). We are currently preparing the 2026 report; a draft will be circulated shortly for your review, and we will discuss it together at the annual meeting in Germany.

ICBD corner: birth defects and rare diseases, plus new skeletal dysplasia study proposal

Over the past three issues, the ICBBD Corner has built the case for integrating birth defect surveillance and rare disease tracking, making three connected points:

- birth defects and rare diseases affect the same children and belong under the same public health umbrella;
- integrated surveillance is feasible, with the "triple surveillance" model, national programs in Brazil and England, and newborn screening serving as working examples;
- the central bottleneck is infrastructure – without standardized registries, consistent coding, and population-based ascertainment, the evidence base remains too thin to drive policy or measure outcomes.

We invite members to share their own ideas for collaborative studies and projects that could advance this agenda

(centre@icbdsr.org).

Proposing a collaborative study on skeletal dysplasias: [SD interest and data availability – Fill out form](#)

With that framing in mind, we **propose a new international, multicenter epidemiologic study of skeletal dysplasias** (osteochondrodysplasias, OCDs) – a group of more than 450 heritable bone disorders now classified into 771 distinct entities associated with 552 genes (2023 ISDS Nosology revision). As a group, their birth prevalence is approximately 2.3–7.6 per 10,000, with under-ascertainment consistently noted; some forms are perinatally lethal (roughly 44% of detected cases die in the perinatal period), while non-lethal forms may go unrecognized in surveillance systems. Yet population-level data remain limited to single-country studies or specific conditions such as achondroplasia and osteogenesis imperfecta. No international multicenter study has systematically examined prevalence, perinatal outcomes, associated anomalies, and secular trends across the full spectrum, using harmonized surveillance data from multiple programs.

The proposed study would use existing ICBDSR member registry data to estimate birth prevalence by diagnostic group and region, characterize perinatal outcomes where possible, examine trends and risk factors (including paternal age and consanguinity), and assess coding comparability across programs using the 2023 ISDS Nosology as a shared reference. It is directly actionable with data already in hand, and would serve as a practical test case for the surveillance integration agenda this column has been building toward.

If your program is interested in learning more and possibly collaborating, please complete the **short survey form** (five questions total) – see URL above. We will then reach out to interested programs, and program leads to plan the next steps.

Literature corner: this month's selection from the ICB

In line with the proposed study in the ICB corner, this month's selection focuses on skeletal dysplasia. We list three epidemiologic studies that broadly looked at a range of skeletal dysplasia, one study (of several) that focused on achondroplasia, and the most recent skeletal dysplasia nosology.

1. Orioli IM, Castilla EE, Barbosa-Neto JG. The birth prevalence rates for the skeletal dysplasias. J Med Genet. 1986;23(4):328–332.

[PubMed](#)

A foundational population-based study using the ECLAMC database (349,470 births), reporting a crude birth prevalence of 2.3 per 10,000 with evidence of substantial under-registration. The most common recognizable types were achondroplasia, thanatophoric dysplasia/achondrogenesis, and osteogenesis imperfecta. The study established key methodological benchmarks for prevalence estimation in this group and remains widely cited as a baseline reference.

2. Orioli IM et al. Clinical epidemiology of skeletal dysplasias in South America. Am J Med Genet A. 2012;158A(5):1038–1045.

[PubMed](#)

An updated, larger population-based analysis from the same Latin American network, reporting an overall prevalence of 3.2 per 10,000 and perinatal lethality in 50% of cases. Prenatal ultrasound detected 73% of cases. Confirmed associations with higher paternal age and parental consanguinity. Applied ISDS classification across diagnostic groups, making it one of the most comprehensive registry-based baselines available for this condition category in any region.

3. Stevenson DA, Carey JC, Byrne JL, Srisukhumbowornchai S, Feldkamp ML. Analysis of skeletal dysplasias in the Utah population. Am J Med Genet A. 2012;158A(5):1046–1054.

[PubMed](#)

Using the Utah Birth Defect Network (UBDN), this study examined 153 skeletal dysplasia cases ascertained across 1999–2008, with all cases reviewed by three clinical geneticists and classified per the 2010 ISDS Nosology. Overall prevalence was 3.0 per 10,000 births and 20.0 per 10,000 stillbirths. The study is notable for its quality-controlled ascertainment

process and is a direct model for the type of registry-based, nosology-anchored approach we are proposing for an international collaboration.

4. Coi A et al. Epidemiology of achondroplasia: a population-based study in Europe. Am J Med Genet A. 2019;179(9):1791–1798.

[Full text](#)

The largest European population-level study of achondroplasia, drawing on 28 EUROCAT registries from 1991 to 2015 (434 cases; prevalence 3.72 per 100,000). Examined birth outcomes, prenatal diagnosis rates, associated anomalies, and the effect of parental age on de novo mutations. This study demonstrates the direct value of multicenter registry networks for rare skeletal disorders, and its design – harmonized data, multiple registries, long time window – is the template for what an ICBDSR collaboration could achieve across a broader set of OCD diagnostic groups.

5. Unger S et al. Nosology of genetic skeletal disorders: 2023 revision. Am J Med Genet A. 2023;191(5):1164–1209.

[Journal \(open access\)](#)

The 11th and current revision of the ISDS Nosology, now encompassing 771 entities across 41 groups associated with 552 genes. The most significant update is adoption of a dyadic naming system linking each phenotypic entity to its causative gene, improving precision and reducing coding errors. For surveillance purposes, this is the essential classification framework for any international epidemiologic study of skeletal dysplasias – aligning registry coding with this nosology is a prerequisite for meaningful cross-program comparability. Open access.

13th World Congress of Clinical Safety on June 13-14, 2026 – Kyoto, Japan

Hosted by International Association of Risk Management (IARMM). In-person and virtual registration options are available. For more information, please visit: <http://www.iarmm.org/13WCCS/>

17th International Skeletal Dysplasia Society Meeting

Toronto, Ontario, Canada | August 26–29, 2026

For more information, please visit: <https://www.isds.ch/index.php?page=isds-2026-meeting>

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

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