

# Deciphering Beckwith-Wiedemann Spectrum



**Friday, Saturday and Sunday  
July 21, 22 and 23, 2023**

## COURSE OVERVIEW

Deciphering Beckwith-Wiedemann Spectrum (BWSp) will provide current diagnostic and clinical management information to clinicians and caregivers with patients affected by BWSp. Conference attendees will be educated on the vast spectrum of presenting clinical features, current clinical management practices, and common medical procedures. Understanding this information is crucial in providing optimal care to patients with BWSp. The audience will be encouraged to further educate other clinicians and caregivers at their home institutions.

## TARGET AUDIENCE

This course is intended for families of children with BWSp and practicing clinicians, including, but not limited to pediatricians, family practitioners, general practitioners, genetic counselors, nurse practitioners, nurses, physician assistants, physicians-in-training, other allied healthcare professionals and families. Other specialists are welcome to attend.

## COURSE OBJECTIVES

Upon completion of this course, participants should be able to:

- More accurately diagnose patients with BWSp
- Improve their management of individuals with BWSp
- Share educational materials with other providers at their home institution

## GENERAL INFORMATION AND COURSE REGISTRATION FEES

### In-Person Registration Fees

Physician .....	\$75
Other Healthcare Providers .....	\$50
Family .....	.\$10/adult

*Attending the conference in person includes continental breakfast, refreshments, lunch, and course materials.*

### Virtual Registration Fees

Healthcare Providers .....	\$40
Family .....	.\$20/adult

*Attending the conference virtually includes livestream access to the conference and access to view online course materials.*

**REGISTER ONLINE: [CHOP.CLOUD-CME.COM](https://chop.cloud-cme.com)**

## HOTEL ACCOMMODATIONS

A block of rooms has been reserved at reduced rates at the Sheraton University City, located just a few blocks from Children's Hospital. The group overnight rate is \$199 single and double occupancy plus a 16.37 percent room tax. Participants are required to make their own hotel reservations by calling the Sheraton directly at 215-387-8700. Please mention the name of the conference when making your reservation. The special room rate will be available until Monday, May 15, 2023. After that date, reservations will be accepted on a space and rate availability basis.



## ACCME ACCREDITATION STATEMENT

Children's Hospital of Philadelphia is accredited by the Accreditation Council for Continuing Medical Education (ACCME) to provide continuing education for physicians.

## AMA CREDIT DESIGNATION STATEMENT

Children's Hospital of Philadelphia designates this live activity for a maximum of 7.25 *AMA PRA Category 1 Credits*<sup>™</sup>. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

## NURSING CONTINUING PROFESSIONAL DEVELOPMENT APPROVED PROVIDER STATEMENT

Nurses will receive a CME certificate of completion. Nurses may use CME category 1 credits for re-licensure in many states, as well as most re-certifications. Please check your state board and certifying body for re-licensure and re-certification requirements.

## CONTINUING GENETIC COUNSELOR CREDITS

Genetic counselors may apply directly for Category 2 CEUs after they have attended the CHOP BWSp Conference. This event meets the criteria to be applicable for Category 2 CEUs. For more information on how to apply: <https://www.nsgc.org/portals/0/docs/education/Who,%20When,%20How%20Much%20at%202022.pdf>

## REFUND AND CANCELLATION POLICY

Children's Hospital of Philadelphia reserves the right to cancel or postpone any course due to any unforeseen circumstances. In the event of cancellation or postponement, Children's Hospital of Philadelphia will refund any registration fees. In order to process refunds for course withdrawals, cancellations must be received in writing by Friday, July 7, 2023. No refunds will be issued thereafter.

## PROGRAM

### FRIDAY, JULY 21, 2023

- 5 p.m. Registration
- 6 p.m. Opening Reception and Welcome Remarks  
*Jennifer Kalish, MD, PhD*
- 6:30 p.m. Dinner
- 8 p.m. Parent Social Night

### SATURDAY, JULY 22, 2023

- 8 a.m. Registration and Breakfast
- BWSp Basics**
- 9 a.m. Introduction to BWSp and Clinical Features  
*Jennifer Kalish, MD, PhD*
- 9:15 a.m. (Epi)Genetics of BWSp  
*Evan R. Hathaway, MS, LCGC*
- 9:30 a.m. Molecular Aspects of BWSp  
*Arupa Ganguly, PhD*
- 9:45 a.m. Management of Hypoglycemia and Hyperinsulinism in BWSp  
*Diva D. De León-Crutchlow, MD, MSCE*
- 10 a.m. Q&A
- 10:15 a.m. Break
- Macroglossia Implications and Management**
- 10:45 a.m. Understanding Macroglossia in BWSp  
*Eric C. Liao, MD, PhD*
- 11 a.m. BWSp and Sleep  
*Christopher M. Cielo, DO*
- 11:15 a.m. Orthodontics and Jaw Development in BWSp  
*Hyun-Duck Nah-Cederquist, DMD, MSD, PhD*
- 11:30 a.m. Speech Development in BWSp  
*Cynthia Solot, MA*
- 11:45 a.m. Nutrition and Feeding in BWSp  
*Sherri S. Cohen, MD, MPH*

- Noon Q&A
- 12:15 p.m. Lunch
- 1 p.m. **Various Aspects of BWSp Medical Management**  
**The Diagnostic Odyssey of BWSp**  
*Mallory Byrne, MSN, APRN, CPNP-AC, CPN*
- 1:15 p.m. **BWSp and Lateralized Overgrowth**  
*Richard S. Davidson, MD*
- 1:30 p.m. **Tumor Screening in BWSp**  
*Suzanne MacFarland, MD*  
*Kristin Zelley, MS, LCGC*
- 1:45 p.m. **Radiology Findings in BWSp**  
*Lisa S. States, MD*
- 2 p.m. **BWSp and Development**  
*Emily Shabason, MD*
- 2:15 p.m. Q&A
- 2:30 p.m. Break
- 2:45 p.m. **Research Updates and Ongoing Projects**  
**Blood Tests for Wilms' Tumor Surveillance in Children with BWSp**  
*Jack Brzezinski, MD*
- 3 p.m. **Adrenal Masses in BWSp**  
*Diana Carli, MD*
- 3:15 p.m. **AIBWS Assessment of Adults with Beckwith-Wiedemann Syndrome**  
*Andrea Gazzin, MD*
- 3:30 p.m. **Adult Experiences with BWS: A Discovery Cohort**  
*William Drust, MS*  
*Jennifer M. Kalish, MD, PhD*
- 3:45 p.m. **The Phenotypic Spectrum of Twins in BWSp**  
*Jennifer L. Cohen, MD*
- 4 p.m. Q&A
- 4:15 p.m. **Research Updates and Ongoing Projects**  
**Impact of Assisted Reproductive Technologies on Genomic Imprinting in a Mouse Model**  
*Marisa Bartolomei, PhD*

- 4:25 p.m. **CDKN1C Mutations Revisited in BWS**  
*Steve D. Klein, MD, PhD*
- 4:35 p.m. **Utilizing Novel Technologies in BWS Research**  
*Snehal Nirgude, PhD*
- 4:45 p.m. **Lateralized Growth in Beckwith-Wiedemann Syndrome**  
*Alessandro Mussa, MD, PhD*
- 4:55 p.m. **Epiphyseodesis for Leg Length Discrepancy in BWS**  
*Maurizio De Pellegrin, MD*  
*Lorenzo Marcucci, MD*
- 5:05 p.m. **Miultilocus Imprinting Disorder (MLID) in a cohort of patients with BWSp**  
*Silvia Russo, PhD*
- 5:15 p.m. **Q&A**
- 5:30 p.m. **Announcements and Adjourn for the Day**  
*Jennifer M. Kalish, MD, PhD*
- 5:45 p.m. **Dinner**
- 8 p.m. **Parent Social Night**

## **SUNDAY, JULY 23, 2023**

- 8 a.m. **Breakfast**
- 9 a.m. **BWSp Through the Years: Parent Perspectives**
- 9:40 a.m. **Q&A**
- 10 a.m. **BWSp Through the Years: Family Perspectives**
- 10:40 a.m. **Q&A**
- 11 a.m. **Break**
- 11:30 a.m. **BWSp Through the Years: Adult and Adolescent Perspectives**
- 12:10 p.m. **Q&A**
- 12:20 p.m. **The Transition from Pediatric to Adult Care**  
*Natalie Stollon, MSW, MPH*
- 12:30 p.m. **Resources for Families**  
*Addie Fishstein, LSW*
- 12:45 p.m. **Updates on International Collaboration**  
*Monica Bertolotti*
- 1 p.m. **Next Steps in BWS Research**  
*Jennifer M. Kalish, MD, PhD*
- 1:15 p.m. **Adjourn — Boxed Lunch**

Unless otherwise noted, faculty is from Children's Hospital of Philadelphia and/or the Perelman School of Medicine at the University of Pennsylvania.

## COURSE DIRECTOR

**Jennifer M. Kalish, MD, PhD**  
Director, Beckwith-Wiedemann  
Syndrome Program of Excellence  
Attending Physician, Human Genetics  
Assistant Professor of Pediatrics

## CONTINUING GENETIC COUNSELOR EDUCATION PLANNERS

**Evan R. Hathaway, MS, LCGC**  
Genetic Counselor, BWS Clinic

## COURSE FACULTY

**Marisa S. Bartolomei, PhD**  
Co-director, Epigenetics Program  
Professor of Cell and Developmental  
Biology

**Monica Bertoletti**  
AIBWS ODV  
Italy

**Jack Brzezinski, MD**  
Paediatric Oncologist, Division of  
Haematology/Oncology  
The Hospital for Sick Children  
Canada

**Mallory Byrne, MSN, APRN,  
CPNP-AC, CPN**  
Nurse Practitioner  
Department of Pediatrics

**Diana Carli, MD**  
Attending Physician, Department of  
Public Health and Pediatrics  
University of Torino, Italy

**Christopher M. Cielo, DO**  
Attending Physician, Pulmonary  
Medicine  
Assistant Professor of Pediatrics

**Jennifer L. Cohen, MD**  
Attending Physician  
Assistant Professor of Pediatrics  
Division of Medical Genetics  
Duke University (Durham, North  
Carolina) Medical Center

**Sherri S. Cohen, MD, MPH**  
Attending Physician, Gastroenterology,  
Hepatology and Nutrition  
Medical Director, Pediatric Feeding  
and Swallowing Center  
Program Director, Nutrition  
Fellowship Program  
Clinical Associate Professor

**Richard S. Davidson, MD**  
Attending Surgeon, Orthopaedic  
Surgery  
Professor of Orthopaedic Surgery

**Diva D. De León-Crutchlow, MD,  
MSCE**  
Attending Physician, Endocrinology  
Chief, Endocrinology and Diabetes  
Director, Congenital  
Hyperinsulinism Center

**Maurizio De Pellegrin, MD**  
Pediatric Orthopedic Unit  
Piccole Figlie Hospital,  
Parma, Italy  
Department of Orthopedic, ASST  
Ospedale Papa Giovanni XXIII,  
Bergamo, Italy

**William Drust, MS**  
IRB Analyst I, Research  
Administration

**Addie Fishstein, LSW**  
Social Worker, Patient and Family  
Services

**Arupa Ganguly, PhD**  
Director, Genetic Diagnostic  
Laboratory  
Professor of Genetics

**Andrea Gazzin, MD**  
Department of Pediatric and Public  
Health Sciences  
University of Turin  
Torino, Italy

**Steve D. Klein, MD, PhD**  
Fellow, Human Genetics

**Eric C. Liao, MD, PhD**  
Director, Center for Craniofacial  
Innovation  
Surgery Vice Chair of Academic Affairs  
Professor of Surgery

**Suzanne MacFarland, MD**  
Attending Physician  
Division of Oncology

**Lorenzo Marcucci, MD**  
Orthopedic Residency Program  
University of Verona  
Verona, Italy

**Alessandro Mussa, MD, PhD**  
Attending Physician, Department  
of Public Health and Pediatrics  
University of Torino, Italy  
Scientific Committee of the Italian  
Beckwith-Wiedemann Syndrome  
Association

**Hyun-Duck Nah-Cederquist,  
DMD, MSD, PhD**  
Orthodontist  
Director, Craniofacial Research  
Laboratory  
Clinical Associate Professor,  
University of Pennsylvania School  
of Dental Medicine

**Snehal Nirgude, PhD**  
Fellow, Human Genetics

**Silvia Russo, PhD**  
Director of the Laboratory for  
Experimental Research of Medical  
Cytogenetics and Molecular Genetics  
Italian Auxological Institute

**Emily Shabason, MD**  
Attending Physician, Division of  
Developmental and Behavioral  
Pediatrics

**Cynthia B. Solot, MA, CCC-SLP**  
Senior Speech-Language Pathologist  
Co-director, Velopharyngeal  
Dysfunction Program

**Lisa J. States, MD**  
Director, Outpatient Satellite Imaging  
Principal Investigator, 18F-DOPA PET  
Research Studies  
Attending Physician, Radiology  
Associate Professor of Clinical  
Radiology

**Natalie Stollon, MSW, MPH**  
Community Education Program Mgr

**Kristin Zelly, MS, LCGC**  
Senior Genetic Counselor, Cancer  
Predisposition Program