Event Program

Share. Learn. Partner. Plan.

2018 Bloom's Syndrome Conference

We would like to thank our sponsors for their generous contributions and for their hard work in making this meeting possible.

And many donations made by BSA members and their families and friends!

Organizing Committee

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Nathan Ellis

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2018 Bloom's Syndrome Conference

Welcome to the 2018 Bloom's Syndrome Conference. We are excited to have you attend, and we hope that you will be informed and energized by the program we have put together. The Bloom's syndrome community is relatively small, but thanks to each of you, we are a cohesive and active support community focused on members' health and well-being. We hope that this conference will bring a new level of understanding and direction for people with Bloom's syndrome and their families, friends and health care teams. The conference is designed to address the following goals.

Share

This represents an extraordinary opportunity for us to share with one another our experiences. Although we are all unique individuals, we share common concerns and common values. To address our common concerns, we can work together with the Bloom's Syndrome Association and the Bloom's Syndrome Registry to collect information about our medical histories and ultimately to identify medical and other interventions that have been used successfully in persons with Bloom's syndrome.

Learn

Experts in the areas of basic and clinical research around Bloom's syndrome, endocrinology, cancer surveillance and treatment, and immunology will speak to the attendees about the latest advances in these areas. Our goal is for our participants to leave with an informed position about their condition and to be prepared to advocate for the most effective and appropriate evaluations and treatments available.

Partner

By co-hosting the event, we are closely tying together the needs of the Bloom's Syndrome Association member community and the mission of the Bloom's Syndrome Registry. Our goal is for our attendees to understand the importance of participating in the registry and to fully update their information. We will also be hearing about ways to partner with other members of the rare disease community.

Plan

A subset of the Bloom's Syndrome community recently authored a paper outlining the needs of the broader community, and we will have multiple sessions that address our plans for advocacy and action in the next year. We will create specific goals and enlist your help to make these goals a reality.

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Schedule & Agenda

Thursday, 8/9/18	**Unless otherwise noted, all sessions will be in the Salon ABC Meeting Room**
5:00pm – 8:00pm	Conference Check-In Location: Atrium Grove
5:30pm – 7:30pm	Welcome Reception Location: Atrium Grove

Friday, 8/10/18

7:00am – 9:00am	Conference Check-In Location: Atrium Grove
8:30am – 9:30am	Opening Keynote Session: Where are we? Chris Cunniff, MD
9:30am – 10:30am	Cancer Diagnosis and Treatment Carolyn Fein Levy, MD
10:30am – 10:45am	Refreshment Break
10:45am – 12:00pm	Cancer Surveillance Sharon Plon, MD, PhD
12:00pm – 1:00pm	Buffet Lunch Location: Atrium Grove
1:00pm – 2:00pm	Endocrinology Evaluation and Treatment Maria Vogiatzi, MD
2:00pm – 3:00pm	Immunology Overview Edith Schussler, MD
3:00pm – 3:15pm	Refreshment Break
3:15pm – 5:00pm	Bloom's Syndrome Perspectives Sheryl Grossman, MSW, and Panelists
5:00pm – 6:30pm	Free time
6:30pm – 8:30pm	Group Dinner Location: Hofbrauhaus, 5500 Park Place, Rosemont Within walking distance of the hotel

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Saturday, 8/11/18	**Unless otherwise noted, all sessions will be in Salon ABC Meeting Room**
8:30am – 9:30am	Building Community - Advocacy and Action Nicole Boice – CEO, Global Genes
9:30am – 10:30am	Building a Research Agenda Nathan Ellis, PhD Joanna Groden, PhD
10:30am – 10:45am	Refreshment Break
10:45am – 11:55am	Moderated Discussion: Taking the Next Steps
11:55am – 12:00pm	Closing Remarks Chris Cunniff, MD
12:00pm – 1:00pm	Lunch (box lunches) Location: Atrium Grove

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Session Descriptions

Below you will find short descriptions of the topics that each of the speakers will address. We will have electronic links to PDF's of the speakers' Powerpoint presentations available during and after the meeting. Please contact one of the Organizing Committee members if you would like for us to email the PDF's to you as an attachment or send it to you by mail in print form. We have included 3 blank pages at the end of the program for you to use for taking notes during the meeting.

Where are we?
Chris Cunniff, MD

Dr. Cunniff will provide an overview of the Bloom's Syndrome Registry, including the number of registrants, the kind of information that is contained in the Registry, and the samples that have been collected. He will provide an overview of the Health Supervision Recommendations for people with Bloom syndrome that are soon to be published in the American Journal of Medical Genetics and will update attendees on activities that are currently underway at the Registry and those that are planned for the near future.

Cancer Diagnosis and Treatment
Carolyn Fein Levy, MD

Dr. Levy will discuss common ways that cancers are treated in the general population and how these treatment strategies have been modified for people with Bloom's syndrome. She will provide information on which types of chemotherapy are believed to be well tolerated by people with Bloom syndrome and those that may be associated with problem side effects or long-term risks. She will also discuss new treatment approaches that are being developed and the ways these new treatments may be useful for people with Bloom syndrome.

Cancer Surveillance
Sharon Plon, MD, PhD

Dr. Plon will use her extensive background in cancer surveillance to discuss how we can best detect cancer at its earliest stages, when treatment is most successful. She will review information on how this is being done in conditions similar to Bloom's syndrome, where a genetic change causes significantly increased risks, compared to the general population. She will also provide input on how best to study the current approaches to cancer surveillance in people with Bloom's syndrome and to measure their effectiveness.

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Endocrinology Evaluation and Treatment
Maria Vogiatzi, MD

Dr. Vogiatzi will review her experience in screening, diagnosis and treatment of endocrine abnormalities in people with Bloom syndrome. These issues will include growth problems, thyroid abnormalities, type 2 diabetes, cholesterol and triglyceride abnormalities, puberty and fertility. She will provide information on treatment of these endocrine issues, including use of growth hormone.

Immunology Overview
Edith Schussler, MD

Dr. Schussler will provide background information on the immune system and how it does its job. She will discuss the basic parts of the immune system, including immune cells such as T and B-cells, as well as immunoglobulins and how they protect us from infection. She will describe what is currently known about the range of immune abnormalities that have been reported in people with Bloom syndrome. She will also give information about what kind of immune testing may be most helpful, when referral should be made to an immunologist, and how best to treat recurrent infections, including when immunoglobulin therapy is indicated.

Bloom's Syndrome Perspectives
Sheryl Grossman, MSW, and Panelists

Ms. Grossman has recorded a video presentation that provides her perspective as a person with Bloom's syndrome and as the facilitator for Bloom's Connect. Panelists will respond to the issues raised and will provide their own unique perspective as a person with Bloom's syndrome or a family member of someone with Bloom's syndrome

Building Community – Advocacy and Action
Nicole Boice, CEO, Global Genes

As CEO of Global Genes, Ms. Boice works to "build awareness, educate the global community, and provide critical connections and resources that equip advocates to become activists for their disease." She will share her experience in founding Global Genes and will discuss successful strategies for engagement of patients, patient support organizations, and their partners.

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Building a Research Agenda
Nathan Ellis, PhD, and Joanna Groden, PhD

Dr. Ellis and Dr. Groden have been at the forefront of Bloom's syndrome research and have been responsible for much of what is known about the fundamental functions of the *BLM* gene and how *BLM* gene abnormalities cause medical problems. Building upon their own success in genetic investigation, they will provide their recommendations for future research that will have the highest impact.

Taking the Next Steps
Moderated Panel Discussion

Representatives from the Bloom's Syndrome Association, Bloom's Syndrome Registry, the Bloom's Syndrome Nanocourse group, and a member of the Association from the Netherlands will reflect on the themes of the meeting and discuss ways to improve our understanding of Bloom's syndrome and to apply this knowledge for better health and social outcomes.

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Conference Presenters

Chris Cunniff, MD

Dr. Cunniff is a board-certified Pediatrician and Medical Geneticist and is the Chief of the Division of Medical Genetics at Weill Cornell Medical College in New York. He has been a practicing medical geneticist for over 25 years and is currently Professor of Pediatrics and Director of the Bloom's Syndrome Registry. He has served in leadership roles in medical genetics as Chair of the Committee on Genetics of the American Academy of Pediatrics, as a member of the Board of Directors of the American College of Medical Genetics, and as President of the American Board of Medical Genetics. In 2015 he assumed leadership of the Bloom's Syndrome Registry, which maintains medical information on over 275 persons with Bloom's Syndrome, as well as samples from over half of registrants. Working with other experts on Bloom's Syndrome, he has authored clinical care recommendations for people with Bloom's Syndrome, which are to appear soon in the American Journal of Medical Genetics. He is actively engaged in additional Bloom's Syndrome investigations on feeding and nutrition concerns, cancer onset and treatment, sensitivity to ultraviolet and ionizing radiation, and the genetic changes seen in cancer in people with Bloom's Syndrome. He is very pleased to be working together with participants and speakers at this conference to understand the medical and social needs of people with Bloom's Syndrome, to increase and share knowledge, and to plan for the future.

Carolyn Fein Levy, MD

Carolyn Fein Levy, MD is a Pediatric Hematologist/Oncologist at the Cohen Children's Medical Center (CCMC) of NY. She is an Assistant Professor of Pediatrics at Zucker School of Medicine at Hofstra/ Northwell and the Section Head of the Pediatric Oncology Rare Tumor and Sarcoma (PORTS) Program at CCMC. Dr. Fein Levy contributes to the field of pediatric oncology as the local principal investigator on several national Children's Oncology Group (COG) clinical trials. Working with Dr. Ken Onel, Head of the Genetics/Genomic program for Northwell Health, they have created the Center for Cancer Prevention and Wellness, a family-centered cancer risk assessment program. Dr Fein Levy cares for a number of patients with Bloom syndrome and has a specific interest in developing better chemotherapy treatment options for pediatric and adult patients with Bloom syndrome who develop cancer. Dr. Fein Levy volunteers as the camp doctor for Chai Lifeline's Camp Simcha since 1998, and since 2009 she has served as the associate medical director of the Camp Simcha Girls Medical Staff. As a cancer survivor, she brings a unique combination of medical expertise and personal experience and serves as a role model and living illustration of all the possibilities of life after cancer. She has been invited to share her story and clinical expertise as an inspirational speaker revealing her life as a cancer patient, survivor, wife, mother, and physician throughout the New York area.

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Conference Presenters

Sharon Plon, MD, PhD

Dr. Sharon Plon is a board-certified medical geneticist having completed her genetics training at the University of Washington. Dr. Plon has been at Baylor College of Medicine for 25 years and is now professor in the Departments of Pediatrics/Hematology-Oncology, Molecular and Human Genetics and Human Genome Sequencing Center at Baylor College of Medicine and Texas Children's Hospital. Dr. Plon serves on the National Advisory Council for Human Genome Research of the NIH and serves on the Board of Directors of the American Society of Human Genetics. Dr. Plon has dedicated her career to improving the understanding of genetic susceptibility to cancer including leading the NHGRI/NCI-funded BASIC3 clinical trial of whole exome sequencing for newly diagnosed childhood cancer patients now being expanded into several different centers in Texas (KidsCanSeq trial). Dr. Plon has also worked with the American Association of Cancer Research to develop an online resource for physicians with up to date recommendations for cancer surveillance in those children at genetic risk of cancer.

Maria Vogiatzi, MD

Maria G. Vogiatzi, MD. is a Pediatric Endocrinologist at the Children's Hospital of Philadelphia (CHOP). She is an Associate Professor of Pediatrics and the Director of Adrenal and Puberty Center at CHOP. A native of Greece, Dr. Vogiatzi obtained her medical degree at the Aristotelian University in Thessaloniki, Greece. She then came to US, where she completed a residency in pediatrics at SUNY, Downstate in NYC. She pursued further training in Pediatric Endocrinology at the New York Presbyterian/ Weill Cornell Medical Center (NYP/ WCMC) and at Texas Children's/ Baylor College of Medicine in Houston TX. After her training, Dr Vogiatzi joined the faculty of pediatrics at NYP/ WCMC, where she practiced for close to 18 years. During her tenure, she became the chief of Pediatric Endocrinology and the Program Director of the fellowship program at NYC/WCMC, positions that she held for about 10 years. She relocated to Philadelphia in 2015 and joined the faculty at CHOP. At CHOP, she leads the Adrenal and Puberty Center, a sub-specialty center that focuses on helping children with disorders that affect growth, development and adrenal function. Dr. Vogiatzi has conducted clinical and lab research, and her work has been supported by NIH and foundation awards. Specifically to Bloom syndrome, Dr Vogiatzi and her collaborators at the NYP/WCMC examined the endocrine abnormalities of children and adults affected with the syndrome and who were also part of Bloom's Syndrome Registry.

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Conference Presenters

Edith Schussler, MD

Dr. Edith Schussler, MD is a board certified Allergist and Immunologist and an Assistant Professor of Pediatrics at Weill Cornell Medical College. A native New Yorker, Dr. Schussler received her undergraduate degree at the Univeristy of California Berkeley, before completing premedical studies at Harvard University. She returned to New York to attend the Icahn School of Medicine, where she also completed her residency in pediatrics and fellowship in adult and pediatric Allergy and Immunology. Over the past year, she has presented her immunology research at national meetings in the U.S. and Europe. She has published on immune dysregulation in Gaucher disease, allergy to Botulinum antitoxin, food allergy and genetic immune deficiency. She is an active member of the education committee for the New York Asthma Allergy Society, the Early Career Immunologist Committee of the Clinical Immunology Society, and the Anaphylaxis Committee of the American Academy of Allergy Asthma & Immunology.

Sheryl Grossman

Ms. Grossman graduated from Washington University in St. Louis with a Bachelor's degree in psychology and a minor in Jewish and Near Eastern Studies and a special minor in Disability Studies. She received her Master's degree from Washington University in Social Work with a concentration in Disability Issues and Advocacy. She was certified by the American Association of Suicidology in 2000 for five years as a Certified Crisis Worker. Sheryl has worked in the Disability Rights field for more than 20 years on issues ranging from equal access to care and informed consent to education, employment, and assistive technology. She has spent the last 15 years specializing in multiply marginalized groups and rare conditions. As an adult with Bloom's Syndrome, Sheryl was driven to find others like herself and to establish a global, psychosocial support group in 1996: Bloom's Connect, which she still serves as facilitator for today.

Nicole Boice

Ms. Boice founded Global Genes in 2008, after being personally touched by rare disease through friends, who struggled for 2 ½ years to find a diagnosis for their son. Once diagnosed, they learned that there were neither treatments nor cures for their son's condition. She understands the importance of finding a diagnosis and has built an organization to help address this problem. Global Genes aims to help families affected by rare disease connect with tools, resources, and much needed support, helping eliminate the challenges of rare disease. Since its inception, Global Genes has impacted hundreds of thousands of patients worldwide. Ms. Boice has held numerous consulting, sales, and marketing executive roles in her 25 years of experience. She has worked with organizations in media, pharmaceutical and high-tech sectors – Schering Plough, CMP Media, United Business Media, and Burrill & Company. A graduate of the University of California San Diego, Nicole is a proud wife, mother of two children, and adopted mother of two dogs.

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Conference Presenters

Nathan Ames Ellis, PhD

Dr. Ellis is a cancer geneticist in the Department of Cellular and Molecular Medicine at the University of Arizona. He serves as Program Director of the Cancer Biology Program in the University of Arizona Cancer Center and Director of the Genetics graduate program. Dr. Ellis was born and raised in New York City, beginning his scientific career in somatic cell genetics and studying gene regulation, sex determination, and mammalian development. In 1990, he joined the New York Blood Center as an Assistant Member, where in collaboration with James German and Joanna Groden he characterized the molecular genetics of the gene mutated in Bloom's syndrome BLM. In 1997, Dr. Ellis took a position in the Department of Human Genetics at Memorial Sloan-Kettering Cancer Center, continuing studies of molecular mechanisms underlying genomic instability and the cancer genetic epidemiology and population genetics of colorectal cancer. He has since held positions at the University of Chicago and the University of Illinois at Chicago studying cancer health disparities and the relationship between genetic risk factors and environmental factors in colorectal carcinogenesis. He continues to study molecular mechanisms underlying genomic instability in Bloom's syndrome with the aspiration to develop better treatment options for persons with the syndrome who develop cancer.

Joanna Groden, PhD

Dr. Groden is a Professor and Vice Chair in the Department of Cancer Biology and Genetics at The Ohio State University. She serves as Program Director for the Pelotonia Fellowship Program in the OSU Comprehensive Cancer Center-Arthur G. James Cancer Hospital and Richard J. Solove Research Institute, and Co-Director of the Biomedical Sciences Graduate Program in the OSU College of Medicine. Dr. Groden began her research career in human genetics with James German at the New York Blood Center, and completed a PhD with his mentorship from Cornell University Graduate School of Medical Sciences studying somatic recombination in Bloom's syndrome. She then completed a postdoctoral fellowship at the University of Utah in the Department of Human Genetics where in collaboration with others, she identified and characterized the gene for an inherited form of colon cancer. In 1995, she continued her collaboration with James German and Nathan Ellis in the discovery of the BLM gene, mutated in Bloom's syndrome. She has held faculty and research leadership positions at the University of Cincinnati and OSU, studying both colorectal cancer genetics and DNA repair mechanisms, using genetically engineered mouse models of cancer in combination with biochemical and molecular biology approaches. She continues her work in Bloom's syndrome with the goal of learning how to translate scientific discoveries into the clinic.