2017 | ACMG Annual Clinical Genetics Meeting

ACMG American College of Medical Genetics and Genomics
Translating Genes Into Health®

Cardinal Signs of Six Selected Syndromes

Held in Phoenix, Arizona, March 21-25, 2017

Date of Release: April 3, 2017
Expiration Date: April 3, 2020 (CME, NSGC, P.A.C.E.®)
Estimate Time of Completion: 2 hours
Course must be completed by the expiration dates

COURSE DESCRIPTION

This series will include succinct descriptions of six syndromes. Each description will include the "cardinal signs" (key manifestations which should trigger a clinical diagnosis), additional key components of the syndrome, cause (if known), natural history, and differential diagnosis. At the end of the session, attendees should be more knowledgeable about each of the discussed entities.
Dr. Joseph Hersh (Primrose Syndrome) declined to release his slides but you will be able to listen to his presentation.

LEARNING OBJECTIVES

At the conclusion of this course, participants should be able to:

- Describe the cardinal signs of each condition
- Explain the mode of inheritance and cause of each disorder
- Recognize the natural history of each condition
- Describe a reasonable differential diagnosis for each disorder

TARGET AUDIENCE

All healthcare professionals interested in the diagnosis, management, treatment and prevention of genetic conditions and increasing their understanding of the genetic basis of common, chronic health problems affecting both children and adults will find the programming applicable to their practice. These select sessions from the ACMG Annual Meeting are targeted for the following professionals:

- Medical and clinical geneticists
- Physicians of all specialties with an interest in genetics, genomics and the genetic basis of disease
- Genetic counselors
- Laboratory geneticists, directors, technicians and technologists

- Researchers
- Pathologists
- Educators
- Nurses
- Dietitians
- Physician assistants
- Biotechnology and pharmaceutical development professionals
- Fellows, Trainees and Students
- Public health professionals
- Genetic/consumer advocates
- Others with an interest in the science and art of medical genetics and genomics

SESSIONS

- Update on De Barsy Syndrome-Mary Willis, MD, PhD, FACMG
- X-linked Hypophosphaemic Rickets: More Than Just Bowed Legs-Gary S. Gottesman, MD,
- Cardinal Signs of Ellis-van Creveld Syndrome "Strangers on a Train"- Caleb Bupp, MD, FACMG
- Bohring-Opitz: Not a Subtype of C Syndrome Anymore!- Bianca Russell, MD
- Review of Wiedemann-Rautenstrauch Syndrome-Allison Jay, MD, FACMG
- Primrose Syndrome-Joseph MD, FACMG

Accreditation:

The American College of Medical Genetics and Genomics is accredited by the Accreditation Council for Continuing Medical Education (ACCME) to provide continuing medical education for physicians.

Credit Designation:
The American College of Medical Genetics and Genomics designates this activity for a maximum of 2 *AMA PRA Category 1 Credit*s™. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

Genetic Counselor Credit

The National Society of Genetic Counselors (NSGC) has authorized American College of Medical Genetics and Genomics to offer up to 2 Category 1 contact hours for this OnDemand course. The American Board of Genetic Counseling (ABGC) will accept CEUs earned for this course for the purposes of genetic counselor certification and recertification. Reporting of credits is sent to NSGC quarterly. Additional fee (~$25) applies for NSGC credit that is billed by NSGC.

P.A.C.E. CEUs – Laboratory Directors and Laboratory Personnel

ACMG is approved as a provider of continuing education programs in the clinical laboratory sciences by the American Society for Clinical Laboratory Science (ASCLS) Professional Acknowledgment for Continuing Education (P.A.C.E.®) Program. The American College of Medical Genetics and Genomics designates this course for a maximum of 2 contact hours. ACMG is approved by the Florida Board of Clinical Laboratory Personnel as CE Provider. ACMG is approved by the California Department of Health Services through the ASCLS P.A.C.E.® Program as CE Provider #275.

HIPAA Compliance

The ACMG supports medical information privacy. While the ACMG is not a "covered entity" under HIPAA 1996 and therefore is not required to meet these standards, ACMG wishes to take reasonable steps to ensure that the presentation of individually identifiable health information at ACMG-sponsored events has been properly authorized. All presenters have completed a form indicating whether they intend to present any form of individually

identifiable healthcare information. If so, they were asked either to attest that a HIPAA-compliant consent form is on file at their institution, or to send ACMG a copy of the ACMG HIPAA compliance form. This information is on record at the ACMG Administrative Office and will be made available on request.

Content Validation

ACMG follows the ACCME policy on Content Validation for CME activities, which requires:

Content Validation and Fair Balance

1. ACMG follows the ACCME policy on Content Validation for CME activities, which requires:
   a) All recommendations involving clinical medicine must be based on evidence that is accepted within the profession of medicine as adequate justification for their indications and contraindications in the care of patients.
   b) All scientific research referred to, reported or used in CME in support or justification of patient care recommendations must conform to the generally accepted standards of experimental design, data collection and analysis.

2. Activities that fall outside the definition of CME/CE; "Educational activities that serve to maintain, develop, or increase the knowledge, skills, and professional performance and relationships that a physician uses to provide services for patients, the public, or the profession" (source: ACCME and AMA) will not be certified for credit. CME activities that promote recommendations, treatment, or manners of practicing medicine or pharmacy that are not within the definition of CME/CE or, are known to have risks or dangers that outweigh the benefits or, are known to be ineffective in the treatment of patients.
3. Presentations and CME/CE activity materials must give a balanced view of therapeutic options; use of generic names will contribute to this impartiality. If the CME/CE educational materials or content includes trade names, where available, trade names from several companies must be used.

Off-label Uses of Products

When an off-label use of a product, or an investigational use not yet approved for any purpose, is discussed during an educational activity, the accredited sponsor shall require the speaker to disclose that the product is not labeled for the use under discussion, or that the product is still investigational. Discussions of such uses shall focus on those uses that have been subject of objective investigation.

Disclaimer: *ACMG educational programs are designed primarily as an educational tool for health care providers who wish to increase their understanding of the application of genomic technologies to patient care. The ACMG does not endorse, or recommend the use of this educational program to make patient diagnoses, particular by individuals not trained in medical genetics. Adherence to the information provided in these programs does not necessarily ensure a successful diagnostic outcome. The program should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed at obtaining the same results. In determining the propriety of any specific procedure or test, a healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen.*

2017 ACMG Program and Education Committee Members Disclosures

Members of the ACMG Staff, Education and Program Committees involved in planning the 2017 ACMG Annual Clinical Genetics Meeting are required to disclose relevant relationships which could be perceived by some as a real or apparent conflict of interest in planning. All disclosures have been reviewed and conflicts of interest resolved by the Education

Committee COI sub-committee or the Executive Director and CME Associate Director and conflicts of interest are disclosed. In the cases where a conflict existed then the committee member refrained from the discussion.

Following is a list of program and education committee members who have disclosed one or more such relationships and names of companies with which those relationships exist:

EC = Education Committee; PC = Program Committee; S = ACMG Staff

1. Major stockholder/ownership interest 2. Grant/Research Support (External) 3. Salary/Employment/Royalty(ies)/Honoraria 4. Consultant/consulting fees/other remuneration 5. Speakers' bureau	6. Non-remunerative positions of influence such as officer, board member, trustee, or public spokesperson (All Committee Members Below are on ACMG Committees –Members with other affiliations are listed) 7. Receipt of intellectual property 8. Other

Georgianne L. Arnold, MD, FACMG - Horizon, 2; Recordati, 2; Biomarin, 2; Actelion, 2; SIMD, 6; ACGME, 6; AAP, 6; ASHG, 6 (PC)
Karen W. Gripp, MD, FACMG – Wiley Publishing Inc., 3; FDNA, 4; Novartis, 4 (PC)
Fuki M. Hisama, MD, FACMG – Horizon Pharmaceuticals, 4; ABMGG, 6 (PC)
Christine A. Curtis, PhD, FACMG - CSI Laboratories, 3 (EC)
Thomas E. Mullen, PhD, MS, FACMG - Good Start Genetics Inc., 3 (EC)
Christian P. Schaaf, MD, PhD, FACMG - Springer Publishing Company, 3 (PC), (EC)
Katrina M. Dipple, MD, PhD, FACMG – ACGME, 6; AAP, 6; ASHG, 6; LA BioMed DSMB, 4; SIMD 6 (PC)

Following is a list of committee, education members and staff who have no relationships to disclose:

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Penelope Freire, CMP (PC), (S)

SPEAKERS AND MODERATORS

Faculty Disclosures:

As a sponsor accredited by the ACCME, the American College of Medical Genetics and Genomics must ensure balance, independence, objectivity and scientific rigor in all its sponsored educational activities. All faculty participating in a CME-certified activity are expected to disclose to the audience any relevant financial interest(s) or other relationship(s) with the manufacturer(s) of any commercial product(s), provider(s) of commercial services or any commercial supporters, including diagnostic laboratories, of the activity discussed in an educational presentation. Relevant financial interest(s) or other relationship(s) can include such things as grants or research support, consultancy, major stock holder, etc. The intent of this disclosure is not to prevent a planner or speaker with a relevant financial or other relationship from course planning or making a presentation, but rather to provide learners with information on which they can make their own judgments. It remains for the audience to determine

whether the speaker's interests or relationships may influence the presentation with regard to exposition or conclusion. All conflicts of interests have been reviewed and resolved by the education and CME subcommittee.

Moderator: Leah Burke, MD, FACMG, FAAP
Professor, Departments of Pediatrics and Medicine, UVM Larner College of Medicine, The University of Vermont Medical Center
No financial relationships to disclose.

Dr. Burke is board certified in both Clinical Genetics and Pediatrics. She is the Director of the UVM Medical Center Clinical Genetics Program and a Professor at the UVM Larner College of Medicine. She serves on the Council on Genetics of the American Academy of Pediatrics and is on the Pediatric Genetic Testing Expert Review Panel for the American Society of Human Genetics. In the ACMG, she has served as a moderator in numerous sessions and organized the Diagnostic Dilemmas Session from 2004- 2013. She is active in the New England Genetics Collaborative as a work group leader and member of the Coordinating Council.

Speaker: Mary Willis, MD, PhD, FACMG
Naval Medical Center San Diego
No financial relationships to disclose.

Dr. Mary J Willis MD PhD is the clinical geneticist and a medical educator at Naval Medical Center San Diego. NMCSD is the largest military medical facility in the western United States and one of the largest centers for military medical education, home to 24 accredited training programs. Dr. Willis is passionate about her work, and is honored to have served active and retired military personnel and their families for the last 9 years. In addition to a busy clinical practice she serves as subject matter expert in genetics and genomics for the Navy and also for the Department of Defense. She served 9 years as Navy representative on the Military Newborn screening work group and 4 years as ex-officio representative for the DOD on the U.S. Department of Health and Human Services Secretary's Advisory Committee on Heritable Disorders in Newborns and Children, SACHDNC. She is also a member of the Western States Genetic Services Collective. She is board certified by American Board of Medical Genetics and Genomics. Dr. Willis received her Ph.D. from University of Colorado, Boulder and her M.D. from University of Colorado School of Medicine. She completed residencies in Pediatrics and Medical Genetics at University of California San Diego. She is Assistant Professor of Pediatrics for Uniformed Service University of Health Sciences as well as clinical faculty and site director for UCSD medical genetics fellowship NMCSD training site.

Speaker: Gary Gottesman, MD, FAAP, FACMG
Medical Geneticist, Shriners Hospital for Children - Saint Louis
Financial relationships to disclose. (Self): Consultant fees for Ultragenyx Pharmaceutical Inc.

Gary S. Gottesman, MD, is the medical geneticist in the Center for Metabolic Bone Disease and Molecular Research at the Shriners Hospital for Children in St. Louis. As a Fellow of the American College of Medical Genetics and Genomics he is a faculty member for the Genetics and Genomics Review Course and previously served on the Social, Ethical and Legal Issues Committee. After graduating from Harvard College he attended the University of Michigan Medical School and completed his pediatrics and medical genetics training at the St. Louis Children's Hospital and Washington University School of Medicine. He was in the first class of the Howard Hughes Medical Institute - National Institutes of Health Research Scholars Program in 1985-1986. His medical genetics fellowship was funded by the Pediatric Scientist Development Program. He is board certified in Pediatrics and Medical Genetics. After almost two years on the faculty at Washington University School of Medicine, jointly working at the Shriners Hospitals for Children-Saint Louis, Dr. Gottesman moved to the Saint Louis University (SLU) School of Medicine for several years. He left for a position at the National Human Genome Research Institute and subsequently moved to the National Institute on Aging. Dr. Gottesman returned to SLU as Director of the Division of Medical Genetics in 2003. He served on the Cardinal Glennon Children's Medical Center Ethics Committee for seven years. In 2008, he became Academic Chair of the Department of Physician Assistant Education and Program Director of the Physician Assistant Program at SLU, before returning to the Shriners Hospitals for Children-St. Louis in early 2011. Dr. Gottesman's areas of interest in medical genetics include: metabolic bone diseases, heritable disorders of connective tissue, skeletal dysplasias, neurofibromatosis, neurogenetics, clinical dysmorphology, inborn errors of metabolism, medical ethics and education.

Speaker: Caleb Bupp, MD, FACMG
Clinical Geneticist, Spectrum Health, Spectrum Health
No financial relationships to disclose.

Dr. Caleb Bupp is a clinical geneticist with Spectrum Health in Grand Rapids, Michigan. He completed medical school at the University of Toledo in 2009 and then pediatrics residency through the University of Louisville at Kosair Children's

Hospital in 2012. Genetics training was done at the Greenwood Genetic Center. His clinical interests include dysmorphology, genetic causes of intellectual disability and autism, and genetics education.

Speaker: Bianca Russell, MD
Cincinnati Children's Hospital
No financial relationships to disclose.

Bianca Russell is a 4th year combined pediatrics and human genetics resident at Cincinnati Children's Hospital. She received her MD from the University of California, Irvine in 2013 and her bachelor's degree in 2008 from Connecticut College. She is interested in clinical genetics and the natural history of genetic disorders. She manages a rare disease registry for Bohring-Opitz Syndrome and serves on the medical advisory board for the Bohring-Opitz Syndrome Foundation.

Speaker: Allison Jay, MD, FACMG, ABMGG certified in Clinical Genetics, Medical Biochemical Genetics
Director of Cancer Genetics Program, St. John Hospital
No financial relationships to disclose.

Dr. Allison Jay is the Director of the St. John Cancer Genetics Program. She completed a residency in Medical Genetics at Wayne/State Detroit Medical Center from 2010-2012, and then completed Medical Biochemical Genetics fellowship there from 2012-2013. She subsequently completed a Clinical Biochemical Genetics fellowship at the Biochemical Genetics laboratory at the Mayo Clinic from September 2013-September 2014. She is board certified in Clinical Genetics, and Medical Biochemical Genetics. Her present interests include caring for patients affected with hereditary cancer, pediatric and adult patients with metabolic disease, and contributing to the care of newborns through consultations.

Speaker: Joseph Hersh, MD, FACMG
Professor, Department of Pediatrics, University of Louisville
No financial relationships to disclose.

University of Missouri School of Medicine for pediatrics residency Cincinnati children's Hospital University of Connecticut for genetics and development Fellowships University of Louisville pediatrics faculty and Director, Weisskopf Center

Participation Instructions

1. Participant logs into ondemand.acmg.net
2. Once logged in the participant will access the session they would like to view. They will be asked if they would like to claim credit for the meeting, or if they will not claim credit for the meeting. Then, this information (Course description) will appear, and participant will have to attest that they have read the information. They will then click Continue.
3. After that, the participant will be able to select the credit types to claim.
4. For each session with a post-test, the participant will need to mark and complete the matching pre-test.
5. Then the participant will watch the session presentations.
6. Participant will complete viewing all session content. "Check marks" indicate which presentations have been viewed.
7. After viewing all presentations within a session, participant will click the "Claim Credit for Session" button under the CME dropdown at the top of the page.
8. Participant should take and then successfully pass the post-test. If they do not pass with a score of 80% or higher, they will have unlimited tries to pass the post-test.
9. Participant will continue the steps above to earn credits for additional sessions.
10. If a session does not have a test attached, the participant will not need to take a pre- or posttest, but will have to complete a Concurrent or Plenary session-specific evaluation to claim credit.
11. To print their certificate, the participant will click the "Print Certificate" button under the CME dropdown at the top of the page. Participant must complete the meeting evaluation (one time only) before they can access their certificate. Participant will then choose their certificate(s). The certificate(s) will be automatically updated as they earn new credits.

Stream Requirements	
Network	For best results, use a hardwired network connection instead of wireless

Full Screen Viewing	If you would like to view the webcast full screen, display the tool bar at the bottom and click the double arrow in the far right corner. The screen will enlarge to the full screen of you system. To restore the size, press the "ESC" key
Refresh Browser Window	If the webcast freezes and does not recover in 3-4 seconds, refresh browser window
Freezing or Stuttering Issues	Adjust the amount of bandwidth needed by putting your mouse anywhere over the video window. A tool bar will appear at the bottom. On the right side you will see a "HD" button, click on that button and you will see a list of options. The top is "auto", with decreasing numbers below. Select a lower bandwidth (such as 360p) to see if your webcast improves
For Technical Support call	1-800-504-5379

Mobile Viewing Requirements

Android Devices	Android 2.3+ with Adobe Flash Player 10.2 or better installed Install Flash Player
Apple Devices	iOS 4+

Online Viewing Requirements

Bandwidth	512kbps
Required Hardware and Software	Screen resolution of 1024X768 or larger Sound card and speakers/headphones

Browser	Microsoft Internet Explorer 7.0 or better Mozilla Firefox 4 or better Safari 5 or better
Windows	Operating System: Windows 8 desktop mode, Windows 7; Windows Vista; Windows XP Service Pack 2 or 3 x86 or x64 (Browsers must be in 32-bit mode) 1.6-gigahertz (GHz) or higher processor 512MB of RAM
Mac OS	Operating System: Apple Mac OS X 10.4.8 or above Intel Core™ Duo 1.83GHz or faster processor 512MB of RAM

Registration and Fees

ondemand.acmg.net
ACMG Members and ACMG Trainees: ($50)
Non-members ($60)
Additional fee (~$25) applies for NSGC credit that is billed by NSGC.

Questions regarding CE credit should be directed to education@acmg.net.

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