2017 | ACMG Annual Clinical Genetics Meeting

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

Genetic Challenges and Controversies in Suspected Child Abuse Cases: Distinguishing Fracture Facts from Fracture Friction

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

Date of Release: April 3, 2017s
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

The clinical geneticist can be called upon to play a role in the multidisciplinary evaluation of children with clinical findings concerning for child abuse. In considering child abuse from a clinical genetics perspective, presentations can be categorized into fractures, skin lesions, hemorrhage, growth disturbances, and concern for caregiver-fabricated illness (previously known as Munchausen syndrome by proxy).This session focuses on fractures and current diagnostic challenges and public misperceptions and controversies. The first diagnostic challenge is that in suspected child abuse cases, the clinician must decide whether and when to pursue testing for osteogenesis imperfecta. A second diagnostic challenge is that more rare disorders (beyond osteogenesis imperfecta) that predispose to fracture, often easily diagnosable by clinical history and radiographs, could be missed due to lack of training. An area of current public controversy is whether non-mobile infants with multiple unexplained fractures could be diagnosed with Ehlers-Danlos syndrome as a feasible explanation. For example, the media has featured sympathetic stories of parents who claim they are victims of false child abuse allegations (after charges have been dismissed due to medical experts who gave testimony for the defense). This session addresses these challenges by discussing current clinical, radiological, and molecular approaches for diagnosing osteogenesis imperfecta and other bone fragility disorders that predispose to fractures. Next, this session considers the evidence in the literature (or lack thereof) suggesting that Ehlers-Danlos syndrome predisposes to bone fragility in infants. Finally, this session considers practice changes in order to both diagnose and reduce the likelihood of abuse in children with disabilities, a particularly vulnerable population. The goal is to empower clinical geneticists to utilize a unified and up-to-date approach to help distinguish the rare causes from the real cases of child abuse, and those critical distinctions and correct diagnoses may be life-saving for some infants and children.

LEARNING OBJECTIVES

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

- Consider Current Approaches to Clinical, Radiological, and Molecular Work-up for Osteogenesis Imperfecta in Suspected Child Abuse Cases
- Recognize that Bone Fragility Disorders Beyond Osteogenesis Imperfecta Include Disorders with Decreased Bone Density, Increased Bone Density, and Rarely Normal Bone Density
- Discuss whether Ehlers-Danlos Syndrome predisposes to bone fragility in infants and children
- Consider Red Flags that May Indicate Abuse in Children with Disabilities

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

- From the Genetics Trenches: Distinguishing Fracture Facts from Fracture Fiction in Two Clinical Cases- Natasha Shur, MD, FACMG
- What Every Clinical Geneticist Should Know About Testing for Osteogenesis Imperfecta in Suspected Child Abuse Cases- Peter Byers, MD
- Beyond Osteogenesis Imperfecta: Rare Disorders Associated with Bone Fragility- Michael Bober, MD, PhD
- Does Ehlers-Danlos Syndrome Cause Bone Fragility: Public Perception versus Clinical Evidence.- Brad Tinkle, MD PhD, FACMG, FAAP
- Recognition and Prevention of Child Abuse in the Child with Disability- Catherine Nowak, MD, FAAP

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.

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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

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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)

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SPEAKERS AND MODERATORS

Faculty Disclosures:

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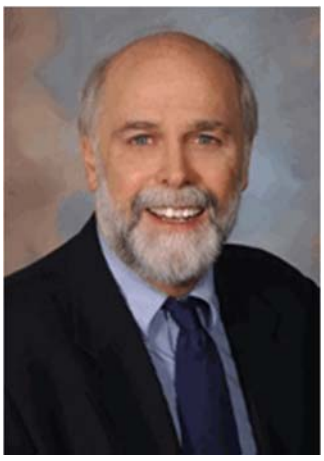
Moderator: John Carey, MD, MPH
Professor, Department of Pediatrics, Division of Medical Genetics, University of Utah, University of Utah School of Medicine
No financial relationships to disclose.

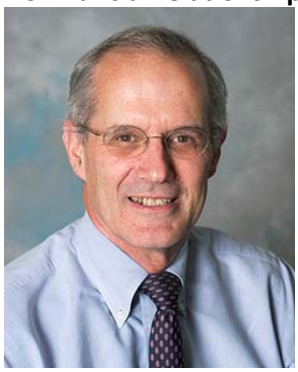


John C. Carey, MD, MPH, is Professor and formerly Vice Chair of Academic Affairs, Department of Pediatrics, at the University of Utah. Throughout his career, Dr. Carey has been interested in birth defect syndromes and the care of children with these conditions. Dr. Carey graduated from Villanova University in 1968 with an A.B. and obtained his M.D. from Georgetown University School of Medicine in 1972. He trained in pediatrics, genetics and dysmorphology as a resident and fellow at the University of California San Francisco, 1972-1979. Dr. Carey obtained an M.P.H. from the University of California at Berkeley in 1976 in between his residency and fellowship years. Dr. Carey joined the faculty at University of Utah Health Sciences Center in 1979. He became Chief of the Division of Medical Genetics in 1985 and remained in that leadership position until 1999 when he stepped down to assume the role as Editor-in-Chief of the American Journal of Medical Genetics. He held that editorial position from 2001 through 2016. Dr. Carey established the Medical Genetics Fellowship Program at the University of Utah in 1985 and was the Program Director until he stepped down in 2014 but continues as a mentor and teacher in the Program. Dr. Carey's research focus has been in congenital malformations, neurofibromatosis, and syndrome delineation. He has authored or co-authored over 300 papers, chapters, invited articles, and editorials for scientific journals. He also co-authored the textbook, "Medical Genetics," by Jorde, Carey, & Bamshad, now in its 5th edition. The book is a widely used text in schools of medicine throughout North America, South America, and Europe. Dr. Carey has served as medical adviser and "founding professional" for the Support Organization for Trisomy 18, 13 and Related Disorders (SOFT) since 1980. The medical and ethical aspects of care of infants and children with these conditions are currently his major academic interest.

Moderator Speaker: Natasha Shur, MD, FACMG
Division Head, Genetics, Albany Medical Center
No financial relationships to disclose.

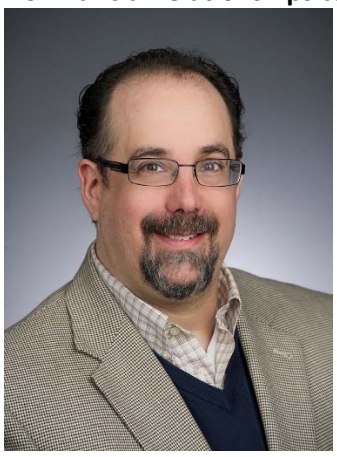
Dr. Shur is currently Division Head of Genetics in the Department of Pediatrics at Albany Medical Center in Albany, New York. She completed her genetics residency and pediatrics residency at the Children's Hospital at Montefiore Medical Center and earned her medical degree at the Albert Einstein College of Medicine of Yeshiva University in the Bronx.

Speaker: Peter Byers, MD
Professor, Departments of Pathology and Medicine (Medical Genetics), University of Washington
No financial relationships to disclose.



Peter H. Byers, MD, Professor of Pathology and Professor of Medicine (Medical Genetics), is a graduate of Reed College, in Portland, and Case Western Reserve University School of Medicine. He completed two years of internal medical residency at UCSF, then was a Research Fellow at the National Institutes of Health where he studied the way that collagen molecules were made, and then came to the University of Washington as a Fellow in Medical Genetics and joined the faculty in 1977 and became Professor in 1986. He has continued to study the way molecules that are important parts of structures in the body, like bone, tendon, and blood vessel walls and to understand how changes in these proteins and the changes in the genes that provide the information to cells to make them determine the clinical presentations. His group has been one of the dominant international laboratories to study inherited forms of brittle bone disease and conditions that give rise to inherited aneurysm disorders. Studies include gene discovery, identification of mosaicism to account for recurrence of dominant condition in children of unaffected parents, the molecular mechanisms of disorders, and their natural history. These studies paved the way to understand how genetic testing can help to identify children with genetic disorders among those thought to have been abused. He created and runs a diagnostic laboratory that was among the first in the nation to bring diagnostic studies for these disorders to clinical practice and in 2014 was named as the inaugural Director of the new UW MedicineCenter for Precision Diagnostics. His role in understanding genetic disorders was recognized by Antoine Marfan Award by the Marfan Foundation and Colonel Sanders Lifetime Achievement Award by the March of Dimes.

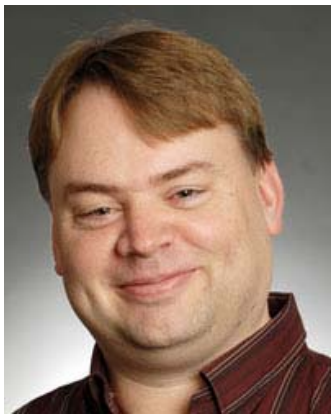
Speaker: Michael Bober, MD, PhD
A.I. duPont Hospital for Children
No financial relationships to disclose.



Dr. Michael B. Bober is the Director of the Skeletal Dysplasia Program at the Alfred I. duPont Hospital for Children in Wilmington, DE and a Professor of Pediatrics at Thomas Jefferson University's Stanley Kimmel Medical College. He completed a combined M.D./Ph.D. program in Biomedical Engineering at Tulane University. His dissertation research focused on the genetic response of bone to mechanical stimulation. He then went on to complete a Pediatrics Residency at Tulane University and a Medical Genetics Residency and Fellowship at Johns Hopkins University. He is a

board certified in Pediatrics, Clinical Genetics and Molecular Genetics. His clinical practice is focused on the diagnosis and management of children with skeletal dysplasia.

Speaker: Brad Tinkle, MD PhD, FACMG, FAAP
Advocate Children's Hospital
Financial relationships to disclose. (Self): Speakers bureau for Alexion Pharmaceuticals Conflict companies: Alexion Pharmaceuticals; Dr. Brad R Tinkle Consulting Corp; Tinkle Family Racing.; IMEDECS; Best Doctors; Ehlers-Danlos Society; Hypermobility Syndromes Association; EDS Support UK; Left Paw Press.



Dr. Tinkle is a clinical and clinical molecular geneticist and Division Head of Clinical Genetics at Advocate Children's Hospital. Dr. Tinkle is a recognized international expert in connective tissue disorders serving on various medical advisory groups and speaks internationally on the subject. Dr. Tinkle started at Purdue University finishing with a BSE in genetic engineering. He went on to George Washington University creating transgenic mouse models for human disease, earning his PhD in human genetics. He graduated from the Indiana University School of Medicine wanting to pursue genetic medicine. He completed a combined pediatrics and clinical genetics residency from Cincinnati Children's Hospital. He then completed a fellowship in clinical molecular genetics before becoming a staff physician. While in Cincinnati, Dr. Tinkle specialized in disorders of connective tissue running various multidisciplinary clinics for connective tissue disorders (especially Ehlers-Danlos syndrome), Marfan and Marfan-related disorders, as well as skeletal dysplasias. He authored two books on joint hypermobility/EDS geared towards the general public in addition to his more than 30 publications. In 2012, Dr. Tinkle assumed the role of medical director of clinical genetics for Advocate Children's Hospital in the greater Chicago area.

Speaker: Catherine Nowak, MD, FAAP
Boston Children's Hospital
No financial relationships to disclose.

Dr. Nowak is the Clinical Director of The Feingold Center at Boston Children's Hospital - Waltham, MA. Dr. Nowak received her medical degree from McGill University and is Board Certified in both Pediatrics and Clinical Genetics. Following her pediatric internship and residency at the University of Massachusetts Medical Center, she completed a

fellowship in Clinical Genetics and Birth Defects with Dr. Murray Feingold at the National Birth Defects Center. She served as Chief of the Genetics Division at UMASS Memorial Medical Center in Worcester, MA from 2002-2006. In 2006, she returned to the NBDC, renamed the Feingold Center for Children which is now part of the division of Genetics at Boston Children's Hospital.

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.
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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.
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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
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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

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