2017 | ACMG Annual Clinical Genetics Meeting

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

Whorls and Swirls: The Skin as Nature's Window to Mosaicism

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

These series of talks will highlight the growing understanding of cutaneous mosaicism and its pathogenetic mechanisms. From clinic to bench, we will discuss practical clinical tips and cutting edge science in the context of nature's human canvas, the skin. We will review disorders with visible evidence of mosaicism. Although the association with underlying abnormality is well-known, the guidelines for patient assessment remain controversial. This session will highlight the current guidelines and recommendations for patient care. The session will also discuss recently developed genetic testing approaches that have translated into improved diagnosis and genetic counseling for individuals with mosaic/segmental presentation.

Dr. Jennifer Hand (A Coffee Break: Café-au-Lait and Pigmentary Mosaicism), Dr. Ludwine Messiaen (On Gonadal, Gonosomal and Somatic Mosaicism in NF1/SPRED1), and *Dr.* Katherine Rauen *(*The New Road Map(K): RASopathies) declined to release their slides but you will be able to listen to their presentations.

LEARNING OBJECTIVES

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

- Review recognized patterns and pathogenetic mechanisms of cutaneous mosaicism
- Describe pitfalls and benefits in testing for cutaneous mosaicism
- Illustrate challenges in diagnosis, management and genetic counseling of mosaic disorders
- Describe a practical approach for cutaneous examination for the practicing geneticist

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

- A Coffee Break: Café-au-Lait and Pigmentary Mosaicism- Jennifer Hand, MD, FAAD, FACMG
- The New Road Map(K): RASopathies - Katherine Rauen, MD, PhD
- On Gonadal, Gonosomal and Somatic Mosaicism in NF1/SPRED1 – Tackling the Challenges Through Cell-of-Origin Specific Approaches- Ludwine Messiaen, PhD
- Coffee with a Shot of PIK3CA- Dusica Babovic-Vuksanovic, MD, FACMG
- Mosaicism in TSC: Genetic and Clinical Implications- David Kwiatkowski, MD, PhD, ABIM

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

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Karen W. Gripp, MD, FACMG – Wiley Publishing Inc., 3; FDNA, 4; Novartis, 4 (PC)
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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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SPEAKERS AND MODERATORS

Faculty Disclosures:

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Moderator: Erin Conboy, MD
Mayo Clinic
No financial relationships to disclose.

Dr. Erin Conboy, M.D. is currently a second year medical genetics resident at the Mayo Clinic in Rochester, MN. She also completed her pediatrics residency at Mayo Clinic and is a board certified in Pediatrics. She is especially interested in pediatric genetics, and in particular inborn errors of metabolism. Her research interests are varied. She have published a case series on Noonan Syndrome with Multiple Lentigines, currently has a grant to look at the causes of hyperammonemia after Roux-en-Y gastric bypass surgery, and recently presented an interesting case at ASHG of liver failure in a patient with sitosterolemia.

Speaker: Dianalee McKnight, PhD, FACMG
GeneDx
Financial relationships to disclose. (Self): Employee of GeneDx

Dianalee McKnight is an ABMG-certified Clinical Molecular Geneticist and Associate Director of the Neurogenetics program at GeneDx. She received her B.S. in Biology from the University of Maryland and her Ph.D. in Physiology at The Pennsylvania State University. Dianalee then worked as a post-doctoral fellow at the National Institutes of Health in the National Institute of Dental and Craniofacial Research for 5 years. While at the NIH, Dianalee completed her medical training in the Metropolitan Washington D.C. program at the National Human Genome Research Institute. Dianalee joined GeneDx in 2010 and her interests include neurology, endocrinology and craniofacial disorders.

Speaker: Jennifer Hand, MD, FAAD, FACMG
Associate Professor of Dermatology and Pediatrics; Assistant Professor of Clinical Genomics, Mayo Clinic, Mayo Clinic
No financial relationships to disclose.

Jennifer Hand is an Associate Professor of Dermatology and Pediatrics, and an Assistant Professor of Clinical Genomics at the Mayo Clinic in Rochester, Minnesota. Her passion for genetics dates back to college where she completed her B.S. Degree at the University of California, Berkeley. As a board certified Dermatologist, Pediatric Dermatologist and Clinical Geneticist, she is proud to serve on the Advisory Board for FIRST, the National support organization for patients with Ichthyosis. She also serves as UpToDate Section Editor for Genodermatoses and is a Consulting Editor for GeneReviews.

Speaker: Katherine Rauen, MD, PhD
Professor, Department of Pediatrics; Chief, Division of Genomic Medicine; Director, NF/Ras Pathway Clinic, University of California Davis, MIND Institute
No financial relationships to disclose.

Katherine (Kate) Rauen, MD, PhD is a Professor in the Department of Pediatrics, Division of Genomic Medicine at UC Davis where she currently serves as the Chief of Genomic Medicine and holds the Albert Holmes Rowe Chair in Human Genetics. She received a MS in Human Physiology and a PhD in Genetics from UC Davis and obtained her MD at UC Irvine. Dr. Rauen did her residency training in Pediatrics and fellowship in Medical Genetics at UC San Francisco. Dr. Rauen is internationally known for her pioneering work in the early application of arrayCGH technology in clinical genetics and as a leader and major contributor to the understanding of the RASopathies. Her research program involves the clinical and basic science study of cancer syndromes with effort to identify underlying genetic abnormalities affecting common developmental and cancer pathways. Dr. Rauen led the research team that discovered the genetic cause of CFC syndrome and independently identified the genetic cause of Costello syndrome. Dr. Rauen is committed to academic medicine, medical education, and advancing best practices for patients with RASopathies. She has successfully obtained both intramural and extramural funding for her research activities, and currently holds a 5-year NIH grant studying skeletal myogenesis in Costello syndrome and CFC. She is the innovator of the world-renowned NF/Ras Pathway Clinic which she initiated in 2007 and this clinic has now been emulated around the globe. She serves on the medical advisory board of CFC International, is a Co-Director for the Costello Syndrome Family Network, and serves on the advisory broads for RASopathies Network USA and Global Genes. Dr. Rauen was recently awarded the Presidential Early Career Award for Scientists and Engineers (PECASE) on her work for CFC and Costello syndrome. This award is the highest honor bestowed by the United States Government on science and engineering professionals in their early research careers.

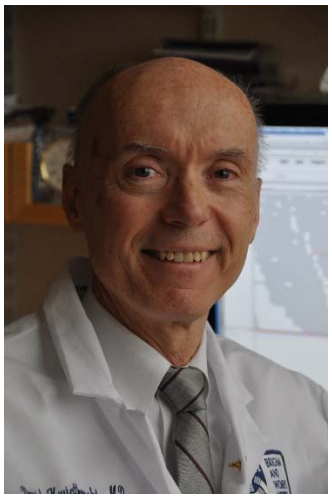
Speaker: Ludwine Messiaen, PhD
Lab Director, Department of Genetics, University of Alabama at Birmingham, University of Alabama at Birmingham
No financial relationships to disclose.

My research is focused on the development and provision of comprehensive genetic tests for a number of rare and common genetic disorders. My laboratory currently offers molecular diagnostic testing for all forms of neurofibromatoses (NF1, NF2, schwannomatosis, spinal NF, NF-Noonan, segmental or mosaic NF), Legius syndrome, Tuberous Sclerosis, the ras-o-pathies, Von Hippel-Lindau disorder, PTEN—disorder, autosomal recessive polycystic kidney disease, and others. My research has a special focus on Neurofibromatosis type1 and phenotypically

overlapping disorders including Legius syndrome. We are interested to fully explore the diverse spectrum of NF1 mutations. We explore NF1 genotype-phenotype correlations using several complementary approaches, including comparison of discrete phenotypic signs in unrelated patients carrying an identical NF1 mutation, characterization of cells/tissues affected in patients with mosaic or segmental NF, definition of the mutational spectrum in patients with variant forms of NF (spinal NF, Watson syndrome, NF-Noonan).

Speaker: Dusica Babovic-Vuksanovic, MD, FACMG
Clinical Geneticist, Mayo Clinic
No financial relationships to disclose.

Speaker: David Kwiatkowski, MD, PhD, ABIM
Professor of Medicine, Harvard Medical School, Senior Physician, Brigham and Women's Hospital, Dana Farber Cancer Institute, Brigham and Women's Hospital
No financial relationships to disclose.



Dr. David Kwiatkowski is best known for his groundbreaking identification of the TSC1 gene, and a long series of genetic analyses in TSC that have elucidated the genetics of TSC and the mechanisms of tumor development in TSC. In addition, his laboratory has generated numerous mouse models of TSC, in which preclinical studies have shown major benefit to treatment with rapalog drugs (rapamycin and everolimus), enabling translation of these drugs to patient care, and eventual demonstration of their benefit in treatment of several kinds of TSC tumors. Dr. Kwiatkowski can perhaps best be described as a mathematician-geneticist-oncologist. He is a Professor of Medicine at Harvard Medical School, an Associate Member of The Broad Institute of MIT and Harvard, and a Senior Physician and practicing thoracic oncologist at the Dana-Farber Cancer Institute/BWH. He is a member of the American Society of Clinical Investigation and the Association of American Physicians, and Program Leader, Dana Farber Harvard Cancer Center Cancer Genetics Program. He is an author of over 280 original research publications; has served on grant review panels for over 35 different organizations, including several branches of the NIH, and 8 originating in Europe; is a member of several external or scientific advisory boards for research programs; and is Senior Editor for Cancer Genetics for PLOS Genetics. For the past several years he has been actively engaged in the NCI TCGA (The Cancer Genome Atlas) project, participating in numerous analysis working groups (AWGs), and co-

leading the bladder cancer and PI3kinase-mTOR AWGs. He has taught and mentored numerous students and post-doctoral fellows.

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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For Technical Support call	1-800-504-5379

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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
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7101 Wisconsin Avenue, Suite 1101 | Bethesda, MD 20814
Telephone: 301-718-9603 | Fax: 301-718-9604 | E-mail: education@acmg.net | Website: www.acmg.net/education