Disruptive Technology: Dr. Diana Bianchi Talks Prenatal DNA Testing

By Zelia Worman, PhD

As part of the summer lecture series from the NIH Office of Intramural Training and Education, the NICHD Director Dr. Diana W. Bianchi (pictured right) presented on the power of genomics in noninvasive screening for prenatal genetic conditions. Her talk, entitled “Noninvasive Prenatal Genomics: A Success Story in Genomic Medicine,” focused on how disruptive technologies have shaped the field of prenatal diagnostics. Dr. Bianchi is a pediatric medical geneticist, specialist in neonatal-perinatal medicine, and the principal investigator of a research group in the National Human Genome Research Institute. She has led research efforts on noninvasive prenatal detection of trisomy 13, 18, and 21 (commonly known as Down syndrome) and their treatment in utero.

Noninvasive prenatal testing uses a simple blood draw from a pregnant woman, followed by massively parallel sequencing to screen for genomic abnormalities in a fetus. In 1979, Dr. Bianchi and her mentor, Dr. Leonard “Len” Herzenberg, repurposed fluorescence-activated cell sorting (FACS) to detect the presence of cell-free fetal DNA from the mother’s blood, to identify aneuploidies (an abnormal number of chromosomes) in a noninvasive way. During her talk, Dr. Bianchi touched on part of the inspiration for her life’s work: Michael Herzenberg, her mentor’s son, who was born with Down syndrome in an era when neurological disorders were not understood and no prenatal screenings were available.

She describes how 10 percent of a pregnant woman’s blood sample is fetal, and through deep-sequencing and new bioinformatics algorithms, geneticists can estimate the abnormal amounts of sequence that align in chromosomes 13, 18, or 21. This test is so often correct, that it has been misinterpreted as a

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Letter from the Editor

I’m typing this letter while sitting in a car on the side of U.S. Highway 26 near a tiny rest stop in Wyoming. A line of cars, hundreds deep, stretches in front and behind me. As far as I can see, not much would awaken such a ruckus in this sleepy little pocket of the Cowboy State—other than a chance to witness the moon eclipse the sun, completely, for nearly two minutes! The Wyoming wind has whipped away any threat of cloud cover, and we all sit quietly in our vehicles, waiting for what many have described as a once-in-a-lifetime, mind-altering experience. A perfectly black moon surrounded by the enchantment of the sun’s corona.

I know what you’re thinking. What does this have to do with a fellow’s newsletter at the NICHD? Or maybe I’ve completely lost you to an Internet search for pictures of the eclipse. For those who are still reading, I’ll explain.

An eclipse offers a moment to be awe-struck, suspending belief that such an experience could never occur. Nothing quite prepares you for the first time you remove your protective glasses to witness what looks like a bottomless hole in the sky. It’s the same feeling I get when I read about a scientific breakthrough or a researcher’s attempt to do the unimaginable. That sense of wow, is this really happening?

An eclipse and groundbreaking science share a wow factor, but there is a major difference. The intensity of an eclipse lasts for but a few minutes—if you’re lucky. It’s fleeting. But the outcomes of research have an infinite impact. A single discovery has the potential to alter the course of humanity. Now that’s jaw dropping.

Check out this issue’s front-page article about NICHD Director Dr. Diana Bianchi’s research, and you’ll see what I mean.

Your Editor in Chief,
Shana R. Spindler, PhD

Questions, comments, or ideas? Send us an email at Shana.Spindler@gmail.com. We love to hear from you!
Dr. Diana Bianchi Talks Prenatal DNA Testing
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diagnostic. Dr. Bianchi emphasized that a negative result is 99 percent accurate. However, a positive result from this screening still needs to be confirmed through amniocentesis or chorionic villus sampling. Moreover, Dr. Bianchi added that false positive results have provided incidental findings that have led to discoveries in fetal-placental biology and even the detection of undiagnosed cancer in the mother.

Non-invasive prenatal screening is now offered as a routine test in high-risk pregnancies, and the goal is to make it as accessible as possible.

From the infancy of karyotyping to the current use of genomic sequencing, Dr. Bianchi has been at the edge of discovery, providing an exciting vision for the future. In fact, her current research goes one step further. Although the work is still preliminary, Dr. Bianchi and her team are addressing the possibility of early treatment for Down syndrome in the neuronal developmental stage of the fetus, *in utero*. She screened FDA-approved drugs that may have an impact in the pathways of neuronal development, are harmless for the mother and baby, and that can help restore some of the neuronal activity that is lost in Down syndrome patients. Her screening results are encouraging, and her group is advancing to behavioral testing of promising drugs in mice.

Overall, Dr. Bianchi’s talk generated enthusiasm about the bright future of medical genomics among our summer students. These new technologies can be “disruptive” and change how obstetricians are trained, while evolving prenatal testing to keep up with modern medical genetics. “Just like Google or Airbnb changed how research and booking a room works, noninvasive prenatal genomics is altering the obstetrician profession around the world,” said Dr. Bianchi.
College Teaching for the 21\textsuperscript{st} Century
Workshop Recap

Editor Q&A with class participant Copelan Gammon

Postbac fellow Copelan Gammon recounts key learning points from the annual “College Teaching for the 21\textsuperscript{st} Century” workshop. This year, Dr. Kate Monzo, an integrated life sciences teaching fellow at the University of Maryland and former postdoctoral fellow from the Weinstein lab, led the workshop.

\textit{For fellows who have never heard about this course before, can you give a brief overview on the subject matter and layout of the course?}

“College Teaching for the 21st Century,” taught by Dr. Kate Monzo, tells you what you need to know to successfully design and teach a college class that results in learning and isn’t given the nickname “Napping 101.” Kate gives a comprehensive introduction to “backward” course design and student-oriented teaching strategies.

Traditional course design typically involves identifying content first, preparing material, and creating an assessment last: a road trip with no clearly identified destination or map to help you get there. In contrast, backward design requires identifying the desired outcome first, developing an appropriate assessment for the learning outcome, and then planning learning experiences and instruction that will directly help students achieve that outcome.

The course is divided into six, two-hour sessions, each focusing on a different topic:

1. Backward design
2. Learning outcomes
3. Assessment
4. Active learning
5. Designing active learning activities
6. Writing a teaching philosophy

\textit{Since the title of the course is “College Teaching for the 21\textsuperscript{st} Century,” how do you think teaching differs in the 21\textsuperscript{st} century?}

Until recently, lecturing has been the undisputed standard in education. With more research in the field of education, science confirms what many students who don’t respond well to lectures already know; student-oriented, rather than teacher-oriented strategies, produce better learning outcomes. Lecturing, a teacher-oriented method, produces little student engagement and requires less complex cognition than teaching strategies that call for active student participation. Lecturing in a 21\textsuperscript{st} century classroom full of laptops also risks students who are more engaged in social media than cell biology.

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In contrast to traditional lecture, research shows that activities requiring complex levels of cognition promote deeper learning and are remembered longer. Bloom’s Taxonomy is a hierarchical model that allows educators to classify learning objectives by level of cognitive complexity. From least to most complex, Bloom identifies the abilities to remember, understand, apply, analyze, evaluate, and create. For example, rote memorization falls much lower on the hierarchy than writing an original research proposal or identifying the correct formula to apply to solve a problem.

How do you think the workshop’s course material applies to teaching at a smaller liberal arts college versus a large research university?

The principle of building a strong course through backward design is relevant for all schools and courses regardless of size. When discussing learning activities, we identified challenges to implementation based on factors such as class size, time allotted, layout of room, etc. Working with Dr. Monzo, we came up with creative solutions to make these strategies work with any class. For instance, in a large class, effectively utilizing your teaching assistants to turn one large group into several smaller groups can allow for more active student participation.

Part of the course includes reading current research related to college teaching and learning. What were a few of the main findings you covered? Were there any findings in the research that surprised you?

The literature highlighted the success of various teaching strategies—such as active learning—in achieving learning outcomes, and it also covered the importance of fostering student metacognition for academic growth. The finding that surprised me the most focused on the innocuous classroom clicker, which I had previously only used for taking attendance and brief multiple-choice quizzes. The report demonstrated that clickers could be effectively used for deep learning by providing a framework for more thoughtful discussion by using “best answer” questions, or initiating conversation about common misconceptions.

The class description mentions well-designed learning outcomes and effective assessment strategies. What’s the difference between these two concepts, and what’s an example from the course about how to make them well-designed and effective?

Learning outcomes set a clear goal for what the student will know or be able to do after a learning activity. Assessment measures how well the student achieved the learning outcome. Vague outcomes like, “Students will learn about zebrafish,” are not easily assessable and should be avoided. Learning outcomes and assessments are also more effective if they are on the same or similar level of cognitive complexity. If a learning outcome states, “Students will be able to differentiate between live monarch and viceroy butterflies,” an assessment asking the learner to list the key differences between the two
College Teaching for the 21st Century Workshop Recap

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varieties would not be an appropriate or effective assessment of that learning outcome. A better assessment might be to give the students specimen samples to sort.

I’ve heard the phrase “active learning” on several occasions. What is active learning, and can you give a few examples of active learning activities?

Active learning, as compared to passive learning, deeply engages the learner with the content, using the teacher as a facilitator to help learners be actively involved in their own learning. Listening to a lecture is a common form of passive learning. Active learning activities come in diverse forms. For example, the “Think, Pair, Share” technique asks learners to first think independently, then come together in pairs to discuss, and finally share their thoughts with the class. One of my personal favorites come from a psychology and law class I taught, where students participated in a mock forensic interview with a “psychopath.” I asked students to familiarize themselves with the criteria, ask relevant and revealing questions, and use the information they discovered to evaluate whether the actor met the criteria for psychopathy.

What is a teaching statement? Any tips on how to write a good one?

Your teaching philosophy statement is a brief document that clearly details your teaching approach, methods, and experience. Make it personal. Teaching philosophies are as diverse as teachers, and your statement should reflect who you are and what your goals are in the classroom.

How did this class change or validate your view on teaching?

As both a learner and occasional teacher, I found myself constantly reflecting on how I could have incorporated information and tools from this class to improve the ones I’ve taken or taught in the past. It clarified some of the frustrations I have previously encountered as a student in a massive lecture hall, and validated my love of active learning activities.

What was the most important take away from the class, for you?

Most students need to be taught how to learn. When I arrived at college I realized I was never taught how to learn or study properly, and was used to succeeding without a lot of hard work. Teaching how to learn at higher cognitive levels, along with self-reflection, would have been a useful tool, and it is a module I would add to the beginning of any course.

For fellows who are interested in taking the course, but aren’t sure if they want to dedicate the time to it, what would you say to them?

The time commitment is small in comparison to what you will gain. Even for those who aren’t planning to teach soon, the content of this course will make you a better learner as well. I highly recommend this course to anyone!
The Rep Report
By Suna Gulay, PhD

As the current NICHD Basic Sciences Representative, I represent NICHD postdoctoral fellows at the Fellows Committee (FelCom) meeting every month. Do you have a concern or question that you want brought up at the next meeting? Contact me at suna.gulay@nih.gov!

Are you interested in gaining leadership experience and soft skills? Serve on an NICHD committee (look for Dr. Yvette Pittman’s emails and articles) and/or FelCom! FelCom has many openings this month:

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<tr>
<th>POSITION</th>
<th>SEND CV AND SHORT STATEMENT OF INTEREST TO:</th>
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<tbody>
<tr>
<td>Animal research advisory committee liaison</td>
<td>Sarah Morgan, <a href="mailto:sarah.morgan2@nih.gov">sarah.morgan2@nih.gov</a></td>
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<tr>
<td>Basic sciences co-chair for Child Care Board</td>
<td>Sarah Morgan, <a href="mailto:sarah.morgan2@nih.gov">sarah.morgan2@nih.gov</a></td>
</tr>
<tr>
<td>Career development subcommittee membership</td>
<td>Qiong Fu, <a href="mailto:qiong.fu@nih.gov">qiong.fu@nih.gov</a></td>
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<tr>
<td>FARE subcommittee membership (Official announcement at FARE awards ceremony on September 14th)</td>
<td>John Gallagher, <a href="mailto:john.gallagher2@nih.gov">john.gallagher2@nih.gov</a> or Pushpanathan Muthuirulan, <a href="mailto:pushpanathan.muthuirulan@nih.gov">pushpanathan.muthuirulan@nih.gov</a></td>
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<tr>
<td>Mentoring subcommittee membership</td>
<td>Gloria Laryea, <a href="mailto:gloria.laryea@nih.gov">gloria.laryea@nih.gov</a> or Jennifer West, <a href="mailto:jennifer.west@nih.gov">jennifer.west@nih.gov</a></td>
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FelCom meets every month on the first Thursday at 4 p.m. in Building 1, Wilson Hall. The decisions for new members and liaisons are generally made at the next monthly FelCom or subcommittee meeting, so apply soon!
Congratulations to the 2018 NICHD FARE Recipients

Congratulations to all fellows and graduate students who received the annual Fellows Award for Research Excellence (FARE) during the 2018 competition. FARE is an NIH-wide competition that recognizes the important research of intramural fellows. Twenty-nine NICHD awardees—a 31 percent success rate—is a terrific accomplishment, and we had the second highest number of winners after NCI.

To learn more about FARE, please visit https://www.training.nih.gov/felcom/fare.

Below is the list of award recipients and their mentors. The FARE award ceremony will take place on September 14, 9:30 – 10 a.m., in Masur Auditorium, Building 10, during the annual NIH Research Festival (see September events).

Vasilisa Aksenova (Dasso)
Griffith Bell (Yeung)**
Dan Benjamini (Basser)
Razvan Chereji (Clark)
Junho Cho (Chou)
Eric Christenson (Banerjee)
Antony Cougnoux (Porter)
Hadis Dashtestani (Gandjbakhche)
Raffaella De Pace (Bonifacino)
Tyler Ekins (McBain)
Atena Farkhondeh Kalat (Bonifacino)
Jakob Gutzmann (Hoffman)
Eric Horstick (Burgess)
Joo Yun Jun (Yanovski)*
Hyun Min Jung (Weinstein)*

Chuljin Lee (Banerjee)
Pei-Chung Lee (Machner)*
Jeremy Luk (Simons-Morton)**
Chad McCormick (Zimmerberg)
Mayumi Miller (Weinstein)
Mona Orr (Storz)
Joshua Pemberton (Balla)
Karen Plevock Haase (Dasso)
Shristi Rawal (Zhang)* **
Amandine Rovini (Bezrukov)
Raffaello Verardi (Banerjee)
Jason Wester (McBain)*
Ashley Xiao (Loh)
Sara Young-Baird (Dever)

* Previous winners
** Member of the Division of Intramural Population Health Research
Meet Our New Fellows

We are happy to welcome new fellows to the NICHD family. If you recently arrived at the NICHD and would like us to introduce you in our quarterly “Meet Our New Fellows” column, please contact our editor, Dr. Shana Spindler, at Shana.Spindler@gmail.com.

ARIANNE FOSTER
Postbac Fellow
Home city: Galloway, New Jersey, USA
Degree institution: Rowan University, Bachelor’s degree
NICHD mentor: Dr. Juan Bonifacino
Area of research: I study protein trafficking in neurons. Neurons are polarized cells, and I look at how the lack of certain adaptor proteins affects the transport of various cargoes.

AKANSHA JAIN
Postbac Fellow
Home city: Oakton, Virginia, USA
Degree institution: University of Pennsylvania, Bachelor’s degree
NICHD mentor: Dr. Juan Bonifacino
Area of research: Intracellular protein trafficking and organelle positioning.

AUDREY LEE
Postbac Fellow
Home city: Upper Saddle River, New Jersey, USA
Degree institution: Brown University, Bachelor’s degree
NICHD mentor: Dr. Jeffrey Baron
Area of research: I study the molecular factors regulating skeletal growth and development at the growth plate.

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Meet Our New Fellows
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ANIKA PRABHU
Postdoctoral Fellow
Home city: Sydney, Australia
Degree institution: University of New South Wales, PhD
NICHD mentor: Dr. Forbes D. Porter
Area of research: I study the dysregulation of cholesterol homeostasis in lysosomal storage diseases.

CHRISTA VENTRESCA
Postbac Fellow
Home city: Guilford, Connecticut, USA
Degree institution: Vassar College, Bachelor’s degree
NICHD mentor: Dr. Mary Lilly
Area of research: I study cell development and metabolism within the context of fruit fly oogenesis.
September Announcements

THE 2017 FELLOWS RETREAT PLANNING: AN OPPORTUNITY AWAITS!
You are invited to serve on the Steering Committee for planning the 14th Annual Meeting for postdoctoral, clinical, and visiting fellows and graduate students. Please send a quick note to Dr. Yvette Pittman (yvette.pittman@nih.gov), NICHD Office of Education, to express your interest.

The group builds the program for the meeting, invites speakers, reviews abstracts, selects fellow/student presenters, and moderates some of the sessions, among other responsibilities. It’s a great opportunity to sharpen your soft skills while working in a team to plan this annual spring event!

We hope to start our monthly, one-hour meetings in October.

NICHD FELLOWS ADVISORY COMMITTEE: SEEKING NEW MEMBERS!
The Office of Education formed an advisory committee in 2016, and we are seeking several more dedicated members to help us develop and initiate academic support programs for the institute. Both domestic and visiting fellows are needed. We want to achieve a broad representation, culturally and academically, so we can address the needs of all our trainees at NICHD. The committee meets monthly to exchange ideas and informally discuss ways we can enhance and tailor the training experience within the NICHD intramural program.

Some potential topics for our committee are how to:
» Increase the participation for training activities
» Expose fellows to various careers in science
» Identify teaching opportunities
» Identify internal and external research funding mechanisms
» Establish a structure for sharing scientific and career resources within the institute

The committee meets once a month on Thursdays, from 3:30 to 4:30 p.m. Our Fall dates are listed below:
» September 14
» October 12
» November 9
» December 7

Don't miss this opportunity to serve your intramural NICHD community. Please contact Dr. Yvette Pittman at yvette.pittman@nih.gov if you are interested in joining the group.
September Events

WEDNESDAY – FRIDAY, SEPTEMBER 13 – 15, ALL DAY
NIH Research Festival 2017
Building 10

This annual event brings together researchers from across all NIH institutes for a multi-day opportunity to share research findings and form new collaborations. The general schedule of events and plenary session topics are available at https://researchfestival.nih.gov/2017.

THURSDAY, SEPTEMBER 14, 3:30 – 4:30 PM
NICHD Fellows Advisory Committee meeting

The committee meets monthly to exchange ideas and informally discuss ways we can enhance and tailor the training experience within the NICHD intramural program. The committee meets once a month on Thursdays, from 3:30 to 4:30 p.m. Please contact Dr. Yvette Pittman at yvette.pittman@nih.gov if you are interested in joining the group.

MONDAY, SEPTEMBER 25, 8:30 AM – 7 PM
NICHD DIR Scientific Retreat
Lipsett Auditorium and adjacent FAES Terrace

All intramural researchers—PIs and lab members—are encouraged to attend this single day, annual event. Apart from an exciting line-up of talks and image competition, every lab will have the chance to present at least one poster. Please plan to step away from your labs and join your colleagues for what is shaping up to be an exciting retreat!

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September Events
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WEDNESDAY, SEPTEMBER 27, 12:30 – 1:30 PM
NICHD Postbac Orientation Session & Pizza Lunch
Building 31, room 2A48

Our institute has approximately 50 postbacs conducting both clinical and basic science research. We would like to bring our postbacs together to meet each other and introduce the volunteer and training opportunities on campus. Learn about:
» ICU simulator rounds
» The annual postbac course
» Genetics clinic shadowing
» Children’s Inn volunteer opportunities
» And more!

The NICHD Office of Education aims to enrich fellows’ NIH experience with career development, outreach, and social activities. If you would like to attend this informational event, please contact Dr. Yvette Pittman at yvette.pittman@nih.gov.