Rebekah S. Zimmerman, Ph.D., FACMG

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EDUCATION

2010: Clinical Molecular Genetics Fellowship, Harvard Medical School, Genetics Training Program, Boston, Massachusetts

2008: Ph.D., Genetics, Tufts University, Sackler School of Graduate Biomedical Sciences, Boston, Massachusetts

2002: B.S., Biochemistry, Syracuse University, Syracuse, New York

CERTIFICATION/LICENSURE

2015: New Jersey Board of Medical Examiners: Licensed Bioanalytical Lab Director

2012: New York State Department of Health: Certificate of Qualification as Laboratory Director in Genetic Testing (Limited to Molecular)

2011: American Board of Medical Genetics: Certification in Clinical Molecular Genetics

PROFESSIONAL EXPERIENCE

July 2014- present: Laboratory Director & Director of Clinical Genetics: Foundation for Embryonic Competence (FEC), Basking Ridge, New Jersey

- Directs the preimplantation genetic diagnosis (PGD) laboratory, including laboratory operations and personnel, validations, quality control, department budgeting, etc.
- Reviews and releases final interpretations and reports on comprehensive chromosome screening and single gene disorder cases
- Was instrumental in the validation and implementation of a next generation sequencing-based comprehensive chromosomes screen for aneuploidy (NexCCS)
- Responsible for the inspection preparation for regulatory agencies (CLIA, NJ DOH, NY DOH)
- Prepares validations for submission to appropriate regulatory agencies (NJ DOH, NY DOH)
- Provides continuing education for Fellows and other medical staff

March 2011-July 2014: Director, Reproductive Genetics: GenPath/BioReference Laboratories, Inc. Elmwood Park, New Jersey

- Designed, validated and implemented testing for several high volume/high throughput genotyping based laboratory developed tests (LDTs)
- Oversaw CAP, CLIA and NYS regulatory requirements for the department
- Obtained New Jersey and New York State DOH approval for numerous LDTs
- Reviewed and released results for molecular genetic tests from several molecular techniques: Illumina GoldenGate/BeadXpress, Taqman real time PCR for copy number detection, fragment analysis by capillary electrophoresis, invader assays, Luminex xTAG, allele specific primer extension, Sanger sequencing
- Implemented the use of Ion Torrent next generation sequencing for high throughput genotyping
- Provided scientific expertise when evaluating new platforms to bring into the company for diagnostic test development

April 2013 – January 2014: Director, Inherited Cancer Confirmation Lab: GenPath/BioReference Laboratories, Inc. Elmwood Park, New Jersey

- Implemented an automated Sanger sequencing confirmation lab for the Inherited Cancer program
- Obtained New Jersey and New York State DOH approval for all appropriate testing

June 2010-March 2011: Associate Laboratory Director: Correlagen Diagnostics, Inc., Waltham, Massachusetts

- Reviewed, revised and released patient reports for Sanger-based sequencing of over 100 genes
- Communicated with physicians to provide further interpretation of results or counsel on performing family studies
- Assisted the Laboratory Director in overseeing the CLIA and CAP regulatory requirements
- Contributed to validation studies of next generation sequencing platform for transition into clinical testing
- Provided expertise to Variant Sciences group for interpretation of complicated results

OTHER MOLECULAR DIAGNOSTICS EXPERIENCE

2008-2009: Fellow: Laboratory for Molecular Medicine, Partners HealthCare Center for Personalized Genetic Medicine, Cambridge, Massachusetts

- Performed the validation of a microarray-based resequencing test for Dilated Cardiomyopathy using the Affymetrix Genechip platform that makes it possible to screen a larger number of genes simultaneously, significantly cutting the cost of the test and turn-around time while increasing the clinical sensitivity s
- Improved Sanger-based sequencing test for Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) by developing assays for a new gene to add to the previous testing panel
- Performed thorough assessment of novel variants found during testing; including database and literature searches, species conservation, familial segregation and *in silico* analysis
- Performed interpretation of results and drafted reports for sign-out

April – May 2009: Fellow (rotation): Children's Hospital Boston DNA Diagnostic Laboratory, Boston, Massachusetts

- Performed primary review of chromosomal microarray data; assessed the pathogenicity of copy number changes
- Reviewed data obtained through Southern blotting/methylation analysis, MLPA, capillary-based sequencing

February – April 2009: Fellow (rotation): Genzyme Genetics, Westborough, Massachusetts

- Rotated through testing groups and was exposed to several platforms (ASO hybridization, Southern blotting, real time PCR by Taqman, Allele-specific PCR, etc.) for a variety of diseases
- Reviewed data closely with Directors and observed case sign-out

RESEARCH EXPERIENCE

Postdoctoral Research: Children's Hospital Boston, Department of Cardiovascular Research, 2009-2010 (Advisors: Drs. Amy Roberts and David Clapham)

- Determined that RAF1, a gene implicated in Noonan syndrome, may also be involved in rare cases of isolated, pediatric-onset hypertrophic cardiomyopathy
- Explored new genes involved in congenital heart disease, focusing on causes of left-sided heart defects including hypoplastic left heart syndrome, bicuspid aortic valve, coarctation of the aorta, etc.

Doctoral Research: Program in Genetics, Sackler School of Graduate Biomedical Sciences, Tufts University, 2002-2008 (thesis advisor: Dr. Naomi Rosenberg)

- Investigated the role of the tumor suppressor p19Arf in preleukemic and transformed cells using the Abelson Murine Leukemia Virus (Ab-MLV) transformation model
- Showed that the p19Arf protein expression is necessary, but not sufficient for the induction of the apoptotic response that follows oncogene expression
- Determined that Abelson disease is accelerated in mice lacking either the Ink4a/Arf locus or the Arf gene alone
- Found that changes in the localization of p19Arf in vitro coincided with the onset of the apoptotic crisis phase, suggesting a role for post-translational regulation of p19Arf

TEACHING EXPERIENCE

Laboratory for Molecular Medicine; Cambridge, Massachusetts, 2008-2009

- Trained technicians and new Fellows in sequence review, assessment of novel variants, set up of assays for Sanger sequencing and Affymetrix GeneChip resequencing arrays

Tufts University, Sackler School of Graduate Biomedical Sciences; Boston, Massachusetts
Instructor: Taught Biology at after-school workshop for middle school children, 2002-2003
Rotation Mentor: Supervised projects of four different first-year graduate students rotating in the lab, 2003-2006

Syracuse University, Syracuse, New York
Teaching Assistant: Calculus I and II, 1999-2002
Tutor: Calculus I, II & III and Differential Equations, Department of Engineering and Computer Science, 2001-2002

PROFESSIONAL SOCIETIES

American Society of Human Genetics, 2015-present
American Society of Reproductive Medicine, 2015-present
American College of Medical Genetics and Genomics, 2008-present
New England Regional Genetics Group, 2009-2011
National Postdoctoral Association, 2008-2010
National Society of Collegiate Scholars, Syracuse University Chapter, 1998-2002

AWARDS AND HONORS

Reviewer, Journal of Assisted Reproduction and Genetics, 2014-present
Richard King Trainee Award for Best Publication in Genetics in Medicine, 2011
Reviewer, American Journal of Medical Genetics, 2010
Partners in Excellence Award, For Outstanding Performance and Commitment to Excellence, 2009
Sackler Biomedical Travel Fellowship, Tufts University, 2007
Undergraduate Honors for Research and Scholarship, Dept. of Biology, Syracuse University, 2002
Syracuse University Chancellor's Scholar, 1998-2002

PUBLICATIONS

Morin SJ, Eccles J, Iturriaga A, Zimmerman RS. Translocations, inversions and other chromosome rearrangements. Fertility and Sterility. In press.

Goodrich D, Tao X, Bohrer C, Lonczak A, Xing T, Zimmerman R, Zhan Y, Scott RT Jr, Treff NR. A randomized and blinded comparison of qPCR and NGS-based detection of aneuploidy in a cell line mixture model of blastocyst biopsy mosaicism. J Assist Reprod Genet. 2016 Aug 6

Franasiak JM, Olcha M, Bergh PA, Hong KH, Werner MD, Forman EJ, Zimmerman RS, Scott RT Jr. Expanded carrier screening in an infertile population: how often is clinical decision making affected? Genet Med. 2016 Mar 3.

Zimmerman, R.S., Jalas, C, Tao X, Fedick, A.M., Kim, J., Pepe, R.J., Northrop, L.E., Scott, R.T., Treff, N. R. Development and validation of concurrent preimplantation genetic diagnosis for single gene disorders and comprehensive chromosomal aneuploidy screening without whole-genome amplification. Fertil Steril. 206 Feb;05(2):286-94.

Zimmerman R.S., Cox S, Lakdawala NK, Cirino A, Mancini-DiNardo D, Clark E, Leon A, Duffy E, White E, Baxter S, Alaamery M, Farwell L, Weiss S, Seidman CE, Seidman JG, Ho CY, Rehm HL, Funke BH. *A Novel Custom Resequencing Array for Dilated Cardiomyopathy (DCM).* Genet Med. 200 May;2(5):268-78; PMID: 20474083

Lakdawala N., Dellefave L., Redwood. C.S., Sparks E., Cirino A., Depalma S., Colan, S., Funke B, Zimmerman R.S., et al. *Familial Dilated Cardiomyopathy caused by an Alpha-Tropomyosin Mutation: The Distinctive Natural History of Sarcomeric DCM*, J Am Coll Cardiol, 200; 55:320-329, PMID: 207437

Zimmerman R.S. and Rosenberg N. *Changes in p9Arf Localization Accompany Apoptotic Crisis During Pre-B Cell Transformation by Abelson Murine Leukemia Virus*, J Virol. 2008 Sep;82(7):8383-9, PMID: 857962

PRESENTATIONS

Zimmerman RS, Treff N, Zhan Y, Tao X, Scott III RT, Scott K, Scott Jr RT. Prospective Randomized and Blinded Comparison of NGS CCS Platforms. Poster Presentation. 2016 ASRM Scientific Congress & Expo, Salt Lake City, Utah.

Juneau CR, Scott K, Neal S, Morin SJ, Zhan Y, Zimmerman RS, Treff N, Franasiak JM, Scott Jr RT. Reliable Detection of Segmental Aneuploidy Identified by Next Generation Sequencing (NGS). Oral Presentation. 2016 ASRM Scientific Congress & Expo, Salt Lake City, Utah.

Eccles JM, Iturriaga A, Jalas C, Behrens A, Kleinman E, Scott Jr RT, Treff N, Zimmerman R. Experiences in Single Gene Disorder (SGD) Preimplantation Genetic Diagnosis (PGD): A Focus on Indication for Testing, Family Member Availability and its Influence on Test Design Paradigms. Oral Presentation. 2016 ASRM Scientific Congress & Expo, Salt Lake City, Utah.

Zimmerman RS. Thorough Validation and Implementation of Preimplantation Genetic Screening for Aneuploidy by NGS. Invited Presentation. 8th Annual Next Generation Dx Meeting, August 23, 2016, Washington, DC

Zimmerman RS, Jalas C, Kleinman E, Eccles J, Iturriaga A, Treff N, Scott Jr RT. Expanded PGD Options for Couples with Unavailable Familial Samples, Germline Mosaicism, or *De Novo* Mutations. Poster Presentation. Preimplantation Genetic Diagnosis International Society (PGDIS) Congress, May 2016, Bologna, Italy.

Zimmerman RS. The Impact of Expanded Carrier Screening on PGD. Invited Presentation. The Human Genetics Association of New Jersey's New Horizons in Genetics Throughout the Lifetime, May 3, 2016, Morristown, New Jersey

Zimmerman, R.S., Jalas, C., Tao, X., Fedick, A., Scott, R. T., Treff, N. R., Combined PGD and CCS from the Same Biopsy: Comprehensive Analysis While Bypassing the Intrinsic Limits of WGA; Presented at the 7st American Society for Reproductive Medicine Annual Meeting, Baltimore, Maryland, 205; Abstract #2459

Jalas, C, Tao, X., Gabriele, D., Scott, R.T., Zimmerman, R., Treff, Nathan; Validation of a Workflow for Rapid Concurrent Preimplantation Genetic Diagnosis of 24-Chromosomes Aneuplody, Single Gene Disorders, And Micro-Deletion and Duplications; Presented at American College of Medical Genetics and Genomics, Salt Lake City, Utah, 205; Abstract #4

Klein RT, Dugan EK, Weisberger J, Zimmerman R. 203 Poster Presentation: Expanded Carrier Screening for Consanguineous Couples. National Society of Genetic Counselors 32nd Annual Education Conference, Anaheim, California.

Zimmerman R, Klein RT, Feier A, Weisberger J, Chung W. 203, Poster Presentation: Supporting Data for a Twenty Five Autosomal Recessive Disease Panel for Ashkenazi Jewish Carrier Screening. The American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting, Phoenix, Arizona.

Klein RT, Gomez A, Shulman L, Feier A, Weisberger J, Zimmerman R. 203, Poster Presentation: Utilization of Expanded Carrier Screening for African American and Hispanic Individuals. The American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting, Phoenix, Arizona.

Klein RT, Kramer E., Feier, A., Zimmerman R., 202, Poster Presentation: A Model for Providing Patient Education and Care When Offering a Comprehensive Pan-Ethnic Carrier Screen. National Society of Genetic Counselors 3st Annual Education Conference, Boston, Massachusetts.

"Pan-Ethnic Carrier Screening", Holy Name Medical Center, OB Grand Rounds, May, 202

"InheriGen: Pan-Ethnic Carrier Screening", UMDNJ-Robert Wood Johnson Medical School, Division of Maternal Fetal Medicine, June, 202.

Zimmerman R.S. et al., 200. Poster Presentation: Evidence for *RAF* in causing non-syndromic, pediatric hypertrophic cardiomyopathy. Dr. M. Judah Folkman Research Day, Children's Hospital Boston, Boston, Massachusetts.

Zimmerman R.S. et al., 200. Poster Presentation: Evidence for *RAF* in causing non-syndromic, pediatric hypertrophic cardiomyopathy. The American College of Medical Genetics 7th Annual Clinical Genetics Meeting, Albequerque, New Mexico.

Zimmerman R.S. et al., 2009. Poster Presentation: *A Novel Custom Resequencing Array for Dilated Cardiomyopathy (DCM).* The American College of Medical Genetics 6th Annual Clinical Genetics Meeting, Tampa, Florida.

Stackpole R., and Rosenberg N. 2007. Poster Presentation: *Nucleolar Localization of p9Arf is Important for Tumor Suppressor Function During Transformation by the Abl Oncogene.* The American Society of Human Genetics 57th Annual Meeting, San Diego, California.

Ng P., Stackpole R., et al., 2002. Poster Presentation: *Malignant Melanoma in Xiphophorus (Platy X Swordtail) Hybrids as a Potential Non-Mammalian In Vivo Model for Discovery and Evaluation of Anti-Neoplastic and Anti-Metastatic Agents*, Undergraduate Research Conference, Syracuse University, Syracuse, New York.

Ng P., Stackpole R., et al., 200. Poster Presentation: *Histological Determination of Metastases in Malignant Melanoma in Xiphophorus (Platy X Swordtail) Hybrids*, Undergraduate Research Conference, Syracuse University, Syracuse, New York.