# University of California, San Francisco CURRICULUM VITAE

Name: Aleksandar Rajkovic, MD, PhD

**Position:** Professor In Residence

Pathology

School of Medicine

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#### **EDUCATION**

1981 - 1985	Johns Hopkins University, Baltimore, Maryland	BS	Chemistry	
1985 - 1992	Case Western Reserve University, Cleveland, Ohio	MD/PHD	Medicine/Molecular Biology	Fritz Rottman
1992 - 1993	Metrohealth Medical Center, Cleveland, Ohio	Internship, General Medicine	Internal Medicine	
1993 - 1997	Metrohealth Medical Center, Cleveland, Ohio	Residency	Obstetrics and Gynecology	
1997 - 1997	Metrohealth Medical Center, Cleveland, Ohio	Fellowship	Maternal Fetal Medicine	
1997 - 1999	Baylor College of Medicine	Medical Genetic Residency	Medical Genetics	

# LICENSES, CERTIFICATION

1993	Medical License, State of OHIO
1997	Medical License, State of TEXAS
2009	Medical License, State of PENNSYLVANIA
2018	Medical License, State of CALIFORNIA

#### PRINCIPAL POSITIONS HELD

1997 - 2001 Baylor College of Medicine Clinical Instructor Department of Obstetrics and

Gynecology

2001 - 2006	Baylor College of Medicine	Assistant Professor	Department of Obstetrics and Gynecology
2006 - 2009	Baylor College of Medicine	Associate Professor	Department of Obstetrics and Gynecology
2009 - 2013	University of Pittsburgh School of Medicine	Associate Professor	Department of Obstetrics and Gynecology
2011 - 2018	University of Pittsburgh School of Medicine	Marcus Allen Hogge Chair i Reproductive Sciences	Department of Obstetrics, Gynecology & Reproductive Sciences
2013 - 2018	University of Pittsburgh School of Medicine	Professor	Department of Obstetrics, Gynecology & Reproductive Sciences (Primary Appointment)
2013 - 2018	University of Pittsburgh, Graduate Schof Public Health	nool Professor	Department of Human Genetics
2013 - 2018	University of Pittsburgh School of Medicine	Professor	Department of Pathology
2018 - present	University of California, San Francisco	Distinguished Professor	Department of Pathology
2018 - present	University of California, San Francisco	o Professor	Department of Obstetrics and Gynecology
2018 - present	University of California, San Francisco	Professor	Institute of Human Genetics
2018 - present	Chief Genomic Officer	Professor	UCSF Health
OTHER POSIT	TIONS HELD CONCURRENTLY		
2006 - 2009	Baylor College of Medicine, Houston, TX	Research Director, Womens Reproductive Health Research	Department of Obstetrics and Gynecology

2007 - 2009	Baylor College of Medicine, Houston, TX	Director of Research	Department of Obstetrics and Gynecology		
2009 - 2018	Magee-Womens Hospital of UPMC	Director of Reproductive Genetics	Department of Obstetrics, Gynecology & Reproductive Sciences		
2009 - 2018	Magee-Womens Hospital of UPMC	Co-Director, Fetal Diagnostic Center			
2012 - 2018	University of Pittsburgh School of Medicine	Program Director	Residency/Fellowship in Medical Genetics		
2013 - 2018	Magee-Womens Hospital of UPMC	Medical Director, Pittsburgh Genomics Laboratory	Department of Obstetrics, Gynecology & Reproductive Sciences		
2012 - 2018	Magee-Womens Hospital of UPMC	Medical Director, Pregnancy Screening Lab	Department of Obstetrics, Gynecology & Reproductive Sciences		
2018 - present	University of California, San Francisco	Distinguished Professor	Department of Pathology		
2018 - present	University of California, San Francisco	Professor	Department of Obstetrics and Gynecology		
2018 - present	University of California, San Francisco	Professor	Institute of Human Genetics		
2018 - present	University of California, San Francisco	Chief Genomic Officer	UCSF Health		
HONORS AND	HONORS AND AWARDS				

1985	Phi Beta Kappa Society	Honor Society
1985	American Chemical Society Award for Outstanding Achievement in Chemistry	•

1997	First Place Award, Research Day, Cleveland Society of Obstetrics and Gynecology	Cleveland Society of Obstetrics and Gynecology
1997	Peter Adam Research Award	Metrohealth Medical Center, Cleveland, Ohio
2013	Marcus Allen Hogge Chair in Reproductive Sciences, University of Pittsburgh	Endowed Chair
2013	Elected to American Society for Clinical Investigation	American Society for Clinical Investigation
2016	Keynote Speaker, International Federation of Fertility Societies.	International Federation of Fertility Societies.
2018	Elected to Association of American Physicians	Association of American Physicians
2019	Stuart Lindsay Distinguished Professor in Experimental Pathology I	University of California, San Francisco
2020	Elected to American Gynecological and Obstetrical Society	American Gynecological and Obstetrical Society

#### **KEYWORDS/AREAS OF INTEREST**

Reproductive Genomics Oogenesis Ovarian Development Germline differentiation Uterine leiomyomas

### **CLINICAL ACTIVITIES**

#### **CLINICAL ACTIVITIES SUMMARY**

I am board-certified in clinical genetics and genomics, as well as obstetrics and gynecology. My clinical activities include medical director of cytogenomic and genomic laboratories, prenatal genetic diagnosis and counseling, as well as adult/cancer genetics. I previously acted as a medical director for the clinical genomics laboratories at the Magee-Womens Hospital, and served as the medical director for the overall genetic counseling/diagnostic services at Magee. Currently, I serve as the Chief Genomic Officer, overseeing clinical genetics and genomics at the UCSF Health.

#### **CLINICAL SERVICES**

1999 - 2009	Prenatal Diagnosis, Genetics and Ultrasound	Two days a week
1999 - 2009	Labor and Delivery Coverage	One day a week
1999 - 2009	Gynecologic Surgery	One day a week
2009 - present	Prenatal Diagnosis, Genetics and Ultrasound	Half a day a week

2009 - present Adult Genetics

Half a day a week

# PROFESSIONAL ACTIVITIES

#### **MEMBERSHIPS**

1999 - present American Society of Human Genetics
1999 - present American College of Medical Genetics
2001 - present International Society for Prenatal Diagnosis
2002 - present American College of Obstetricians and Gynecologists
2003 - present Society for the Study of Reproduction
2000 - present Society for Reproductive Investigation
2007 - 2009 Perinatal Research Society
2013 - present American Society for Clinical Investigation (ASCI)
2018 - present Association of American Physicians (AAP)

#### SERVICE TO PROFESSIONAL ORGANIZATIONS

2008 - 2010	American College of Obstetricians and Gynecologists	ACOG Committee on Genetics, Member
2016 - present	American College of Obstetricians and Gynecologists	ACOG Committee on Genetics, ASRM Liaison
2006 - 2007	Society for the Study of Reproduction	Program Committee
2008 - 2008	Society for the Study of Reproduction	Clinical Outreach
2014 - 2014	Society for the Study of Reproduction	Program Committee Translational Research
2020 - present	ACMG Foundation Board of Directors	Member

#### SERVICE TO PROFESSIONAL PUBLICATIONS

2007 - 2010 Associate Editor: Molecular and Human Reproduction Associate
 2009 - present Associate Editor: Journal of Assisted Reproduction and Genetics
 2009 - 2011 Associate Editor: Endocrine Reviews

# **INVITED PRESENTATIONS - INTERNATIONAL**

2002	Maribor Congress on Infertility: "Genetic Regulation of Mammalian Oogenesis;" Maribor, Slovenia	Invited Speaker
2005	Society for the Study of Reproduction, Annual Meeting: "Nobox deficiency and Gene Expression in the Newborn Ovary," Quebec City	Invited Speaker
2005	Mammalian Oogenesis and Epigenetic Modification Workshop: "Transcriptional regulation of early Folliculogenesis," Tokyo, Japan	Invited Speaker
2006	ESHRE Course on Ovarian Development, ESHRE Annual Meeting, "Autosomal gene mutations impairing female reproduction, implications from animal models, Lisbon, Portugal.	Invited Speaker
2008	ESHRE 2008 Conference: "X chromosome genes with a role in gametogenesis," Barcelona.	Invited Speaker
2010	3rd International Symposium on Reproductive Endocrinology and Genetics: "Genetics of Premature Ovarian Failure," Hangzhou, Zhejiang Province, China	Invited Speaker
2010	Sixth World Congress on Ovulation Induction: "Genetics of Folliculogenesis," Naples, Italy.	Invited Speaker
2011	1st Congress of TARTEN (Trans-Atlantic Reproductive Technologies Network), "Molecular mechanisms behind premature ovarian aging," Istanbul, Turkey.	Invited Speaker
2011	Invited Speaker at the CHA Medical University International Reproductive Symposium, "Genomic Approaches to Reproductive Pathology," Seoul, South Korea.	Invited Speaker
2015	Canadian Fertility and Andrology Society (CFAS) Annual Meeting: "Depletion of the Follicle Reserve" Nova Scotia, Canada	Invited Speaker
2015	RIMED Symposium: "Reproductive Genomics: "New Revelations and Implications for Personalized Medicine" Rome, Italy.	Invited Speaker
2015	60th Annual Scientific Meeting of Argentine Society for Clinical Investigation (SAIC): "Recent Advances of Genomic Testing in Perinatal Medicine" Mar del Plata, Argentina.	Invited Speaker
2015	IFFS/JSRM International Meeting 2015: "What have we learned from animal models?" Yokohama, Japan.	Invited Speaker

2016	Society for Reproductive Investigation Ovarian Biology Meeting: Ovarian Ageing: Epidemiologic, Genetics and Metabolic Determinants: "Genetics of Ovarian Failure" Montreal, Canada.	Invited Speaker
2016	Society for Reproductive Investigation Annual Meeting: "Premature Ovarian Failure: Whole Exome Sequencing Approaches to Identify Genetic Causes", Montreal, Canada.	Invited Speaker
2016	Invited Speaker at Shangdong University; Conference on Reproductive Medicine: "Genesis of Leiomyomas" Shanghai, China.	Invited Speaker
2016	22nd IFFS World Congress: "Diagnostic Advances in Reproductive Medicine through Genomics" India Expo Mart in New Delhi, India.	Invited Speaker
2017	4th World Congress of Reproductive Biology 2017. "Oocyte Differentiation During Embryogenesis is Independent of Meiosis and Driven by Interplay of Multiple Transcriptional Regulators"; Okinawa Convention Center, Okinawa, Japan	Invited Speaker
2018	ICE-SEMDSA December 2018, Cape Town, South Africa, "Animal Models of Premature Ovarian Failure".	Invited Speaker
2019	International IVIRMA Congress	Invited Speaker
2020	Chinese Association of Reproductive Medicine/International Federation of Fertility Societies	Invited Speaker
INVITED PRE	SENTATIONS - NATIONAL	
2000	AAAS 2000 Annual Meeting: "Homocysteine and Reproductive Pathology" as part of the Symposia on "New Knowledge on Homocyst(e)ine," Washington, D.C.	Invited Speaker
2002	Invited Speaker, Grand Rounds, Dept of Obstetrics and Gynecology, BCM: "Stem cells and implications for therapy. Houston, TX.	Invited Speaker
2002	Invited Speaker at the Texas Forum for Female Reproduction, 8th Annual Meeting, Houston, TX, "Identification of oocyte-specific transcription factors by in silico subtraction"	Invited Speaker
2002	Invited Speaker at the Postgraduate Courses, American Society for Reproductive Medicine Annual Meeting, Seattle, WA. Talks presented were entitled: "Functional Genomics and Reproductive Pathology", "Genes Leading to Ovarian Dysfunction in Women and Mice", "Genes Causing Testicular Failure and Azoospermia/Oligospermia."	Invited Speaker

2002	Invited Speaker "Lack of Nobox, an oocyte-specific homeobox gene, disrupts early Folliculogenesis," Gordon Research Conference, Mammalian Gametogenesis & Embryogenesis, June 30 - July 5, 2002, Connecticut College, New London, CT.	Invited Speaker
2002	Invited Speaker, Department of Obstetrics and Gynecology, Baylor College of Medicine: "Human Genome Project and Reproductive Pathology," June 2003.	Invited Speaker
2003	Invited Speaker to Reproductive Health Sciences Branch, NIH; Bethesda, MD: "Identification and functional characterization of oocyte-specific transcription factors".	Invited Speaker
2003	Plenary Session Speaker at the 50th Society for Gynecologic Investigation Meeting: "Transcriptional regulation of early Folliculogenesis," Washington, D.C.	Invited Speaker
2003	Speaker at the Womens Health in The 21st Century Symposium: "Meeting Primary Care Needs Through Increasing Practice Skills," talk entitled, "The Future of Gene Therapy," Houston, TX.	Invited Speaker
2003	Invited Speaker at Metrohealth Medical Center, Case Western Reserve University: Obstetrics and Gynecology Grand Rounds, Cleveland, Ohio: "Functional Genomics and Womens Health"	Invited Speaker
2003	Invited Speaker at Metrohealth Medical Center, Case Western Reserve University, Obstetrics and Gynecology Research Seminar, Cleveland, Ohio: "Genetics of Early Folliculogenesis"	Invited Speaker
2004	Grand Rounds, Dept of Obstetrics and Gynecology, BCM, "Markers of Ovarian Cancer"	Invited Speaker
2004	Invited Speaker at Northwestern Medical School, Department of Obstetrics and Gynecology, Grand Rounds, Chicago, Illinois: "Transcriptional Control of Early Folliculogenesis.	Invited Speaker
2004	Society for Gynecologic Investigation, 51st Meeting; Houston, TX: "Nohlh is a helix-loop-helix, oocyte specific transcription factor, critical in mammalian gametogenesis"	Invited Speaker
2004	Invited Speaker at the Gordon Research Conference on Reproductive Tract Biology: "Oocyte-specific transcription factors and regulation of Folliculogenesis"	Invited Speaker
2005	Invited Speaker at the Reproductive Scientist Development Program Meeting: "Masters and Slaves in Oogenesis", Los Angeles.	Invited Speaker

2005	Grand Rounds, Dept of Obstetrics and Gynecology, BCM: "Stem Cells in Reproductive Biology"	Invited Speaker
2006	Organizer and Invited Speaker at the Reproductive Scientist Developmental Program Meeting, Santa Fe, New Mexico; "Ovarian Biology"	Invited Speaker
2006	Invited Speaker, Mary C. Martin Lecture, University of California, San Francisco: "Regulators in oogenesis and spermatogenesis".	Invited Speaker
2006	Invited Speaker at the Gene Therapy Seminar Series, Baylor College of Medicine, Houston, TX. "Gene Therapy and Womens Health, is there anything on the horizon".	Invited Speaker
2006	Invited Speaker at the NIH Reproductive Biology Seminar, University of Texas, Houston. "The role of germ cell specific regulators in folliculogenesis". November 10, 2006.	Invited Speaker
2007	Invited Speaker at the NIH Reproductive Biology Seminar, Bethesda, Maryland, "Regulators of Female gametogenesis".	Invited Speaker
2007	Invited Speaker at the Society for the Study of Reproduction 40th Annual Meeting: "From Oocyte to Embryo," San Antonio, TX.	Invited Speaker
2008	Invited Speaker at the Society for Gynecological Investigation: "Frontiers in oocyte and early embryo development," San Diego, 2008.	Invited Speaker
2008	Invited Speaker at the PREGER Course, "The role of Gametes in Preimplantation Embryo Biology," Philadelphia, 2008.	Invited Speaker
2009	Invited Speaker at the Human Genetics Seminar, Emory University, Dept of Human Genetics, Atlanta, GA. "Oocyte regulators in development and disease".	Invited Speaker
2009	Invited Speaker at the Endocrinology National Meeting: "Novel Directions in Premature Ovarian Failure," Washington, DC.	Invited Speaker
2009	Invited Speaker at the Reproductive Biology Seminar Series, UT Southwestern, Dallas: "Oocyte regulators in development and disease".	Invited Speaker
2010	Grand Rounds Department of Obstetrics and Gynecology, Magee-Womens Hospital, UPMC: "Array CCGH in Obstetrics and Gynecology".	Invited Speaker
2010	Invited Speaker at the D.C. Johnson Seminar Series, Center for Reproductive Sciences, "Genetics of Premature Ovarian Failure," KU Medical Center, Kansas City, KS.	Invited Speaker

2011	Invited Speaker at the Society for Maternal-Fetal Medicine and American Association of Obstetricians and Gynecologist Foundation Annual Scholars' Retreat, "Oogenesis and Ovarian Failure," Magee-Womens Research Institute and Foundation, Pittsburgh, PA	Invited Speaker
2011	Invited Speaker at the RSDP conference, "Career Paths for Physician Scientists", Boulder, Colorado.	Invited Speaker
2011	Invited Speaker at the 9th Annual Women and Infants Health Care Conference: "Current State of the Art in Genetic Consultation and Diagnosis from Pre-Conception to Birth," Magee-Womens Hospital of UPMC, Pittsburgh, PA.	Invited Speaker
2012	Invited Speaker at the 45th Annual Meeting of the Society for the Study of Reproduction: "Molecular Mechanisms and Consequences of Premature Ovarian Aging," The Penn State University, State College, PA	Invited Speaker
2012	Invited Speaker at the Magee-Womens Hospital of UPMC Board of Directors and V.P. Meeting: "The Future of Genetic Testing," Magee-Womens Hospital of UPMC, Pittsburgh, PA	Invited Speaker
2012	Invited Speaker at the NIH/NICHD & American Society for Reproductive Medicine Conference on Ovarian Reserve: Regulation and Implications for Womens Health: "The State-of-the Science in the Identification of Genetic Markers for Premature Ovarian Insufficiency." San Diego, CA.	Invited Speaker
2012	Invited Speaker at the Center for Reproductive Biology and Health: "Molecular Determinants of Reproductive Life Span", Pennsylvania State University, State College, PA.	Invited Speaker
2013	Invited Speaker at Lectures in Reproductive Science (LRS) Seminar Series: "Primordial Follicle Activation," Northwestern University, Evanston, IL.	Invited Speaker
2013	Provost's Inaugural Lecture, Marcus Allen Hogge Chair in Reproductive Sciences, University of Pittsburgh: "Ex Ovo Omnia", Pittsburgh, PA.	Invited Speaker
2014	Molecular Medicine Research Seminar, Children's Hospital of Pittsburgh: "Molecular Mechanisms Behind Premature Ovarian Aging." Lawrenceville, PA	Invited Speaker
2014	Invited Speaker at Oregon Health & Science University, Oregon National Primate Research Center, Division of Reproductive and Developmental Seminar Series: "Molecular Mechanisms Behind Premature Ovarian Aging," Portland, Oregon	Invited Speaker

2014	Invited Speaker at the University of Pittsburgh Annual Celebration of Science, Science 2014 - Sustain It!: "Stem Cells Remixed," Pittsburgh, PA; October 3, 2014	Invited Speaker
2014	Invited Speaker at American Society for Reproductive Medicine, "Gamete Reserve II - Ovary", Honolulu, HI	Invited Speaker
2014	Invited Speaker at OvaScience Symposium "Ovarian Follicles: From Basic Science to Clinical Application", Palo Alto, CA	Invited Speaker
2014	Invited Speaker at the Center for Research on Reproduction and Womens Health, "Molecular Mechanisms Behind Premature Ovarian Aging", Perelman School of Medicine at the University of Pennsylvania, Philadelphia, PA	Invited Speaker
2014	Invited Speaker at Grand Rounds, Department of Obstetrics and Gynecology, Magee-Womens Hospital, UPMC: "Update on Prenatal Aneuploidy Detection," Pittsburgh, PA.	Invited Speaker
2015	Invited Speaker at the Center for Fertility and Reproductive Endocrinology at Magee-Womens Hospital, UPMC, IVF Retreat: Preimplantation Genetic Screening," Pittsburgh, PA; January 12, 2015	Invited Speaker
2015	Invited Speaker at the Premalignant Working Group Research Symposium at Magee-Womens Hospital, UPMC: "Med 12 Associated Reproductive Pathologies" Pittsburgh, PA.	Invited Speaker
2015	Invited Speaker at the Florida International University, Herbert Wertheim College of Medicine Research Seminar Series: "Molecular Mechanism Behind Premature Ovarian Aging" Miami, FL	Invited Speaker
2015	Invited Speaker at the Society for Reproductive Investigation 62nd Annual Scientific Meeting: "Exomes, Chromosomes and Ovarian Failure" San Francisco, CA.	Invited Speaker
2015	Invited Speaker at Grand Rounds, the Center for Human Reproduction: "Genomic Markers of Ovarian Reserve" New York, New York; May 12, 2015.	Invited Speaker
2015	Invited Speaker at the MCCBP Work In Progress Seminar Fall 2015 "Exome Sequencing, Gonadal Failure and Genomic Instability" University of Pittsburgh Cancer Institute, Pittsburgh, PA	Invited Speaker
2015	Invited Speaker at the RSDP 2015 Scholar Retreat: "Mediator Complex Subunit 12 (Med12) Exon 2 Missense Mutation Causes Uterine Leiomyomas and Genomic Instability" Boulder, Colorado; September 12, 2015.	Invited Speaker

2015	Invited Speaker at the P50 Outreach Seminar: "Genomic Approaches to Gonadal Failure" Weill Cornell Medical College, New York, New York.	Invited Speaker
2015	Invited Speaker at the RIMED Symposium: "Reproductive Genomics: "New Revelations and Implications for Personalized Medicine" Rome, Italy.	Invited Speaker
2016	Invited Speaker at Grand Rounds, Department of Obstetrics Gynecology, UPMC Mercy Hospital: "Preimplantation Genetic Testing: Are We Ready For Whole Genome Sequencing," Pittsburgh, PA.	Invited Speaker
2016	Speaker for the 2016 P01 Seminar Series at Northwestern University Department of Obstetrics and Gynecology, "MED12 Functions in Reproductive Pathology" Chicago, IL.	Invited Speaker
2016	Invited Speaker at UPMC Hamot Research Days 2016 Genetics and Genomics in Medicine: "Controversies Surrounding Prenatal Genetic Screening" Erie, PA	Invited Speaker
2016	Invited Speaker at the 2016 Annual Meeting of the Society for the Study of Reproduction: "Med12 and Reproductive Pathology" San Diego, California.	Invited Speaker
2016	Invited Speaker at the 2016 Gordon Research Conference Mammalian Reproduction: "Reproductive Aging and Overall Health: Is There a Link?" Waterville, New Hampshire; august 21-26, 2016.	Invited Speaker
2016	Invited Speaker at the 5th Annual Sanford Imagenetics Genomic Medicine Symposium: "Genomic Approaches to Reproductive Medicine" Sanford Center - Sioux Falls, SD	Invited Speaker
2016	22nd IFFS World Congress: "Diagnostic Advances in Reproductive Medicine through Genomics" India Expo Mart in New Delhi, India.	Invited Speaker
2016	Magee-Womens Research Institute 2016 Annual Retreat: "Genetics of Early Folliculogenesis"; Pittsburgh, PA	Invited Speaker
2016	Translational Reproductive Biology and Clinical Reproductive Endocrinology Conference: "Genetic Control of Follicle Recruitment"; New York, New York; November 19, 2016.	Invited Speaker
2016	Womens Cancer Research Center Work-In-Progress Seminar: "Genetic Underpinnings of Leiomyoma"; Pittsburgh, PA	Invited Speaker

2017	Magee-Womens Research Institute Work-in-Progress Conference and Research Seminar Series. "Reproductive Aging and Overall Health"; Magee-Womens Research Institute, Pittsburgh, PA.	Invited Speaker
2018	Augusta University Grand Rounds: Diagnostic Advances in Reproductive Medicine through Genomics	Invited Speaker
2018	Diagnostic Advances in Reproductive Medicine Through Genomics Baylor Health Care Systems, Dallas TX - September 2018.	Invited Speaker
2018	Committee on Genetics ACOG, Washington DC October 2018.	Invited Speaker
2018	Magee Women's Research Summit October 2018.	Invited Speaker
2018	NICHD Strategic Planning, October 2018.	Invited Speaker
2018	ASHG San Diego, CA October 2018	Invited Speaker
2018	Grand Rounds and Reproductive Seminar, University of Colorado, Denver, CO,	Invited Speaker
2018	University of Nevada, Reno, NV, Reproductive Aging	Invited Speaker
2019	Precision Medicine World Conference, January 2019	Invited Speaker
2019	Buck Institute, Novato California, Developmental Origins of Reproductive Aging	Invited Speaker
2019	Illumina Symposium on Complex and Mendelian Disease Research Symposium	Invited Speaker
2019	ASRM Genetics of Premature Ovarian Aging	Invited Speaker
2019	AMA, Chicago. Precision Medicine Roundtable	Invited Speaker
2019	ASRM Preimplantation Genetic Screening for aneuploidy and beyond: FISH, microarrays, qRT-PCR, next gene sequencing, and SOMA	Invited Speaker
2019	ASHG Genetics of Premature Ovarian Insufficiency	Invited Speaker
2019	UC Santa Cruz Seminar Series: Clinical Genetics and Genomics at UCSF	Invited Speaker
2020	ENDO, San Francisco, Developmental Origins of Reproductive Aging	Invited Speaker
2020	SSR Ovarian Workshop, Ottawa, Canada	Invited Speaker
2020	Precision Medicine World Congress, Women's Health	Chair
2020	Precision Medicine World Congress, Unique Populations Study Collaborations	Chair

# **INVITED PRESENTATIONS - REGIONAL AND OTHER INVITED PRESENTATIONS**

2010	Grand Rounds Department of Obstetrics and Gynecology, Magee-Womens Hospital, UPMC: "Array CCGH in Obstetrics and Gynecology".	Invited Speaker
2011	Invited Speaker at the Society for Maternal-Fetal Medicine and American Association of Obstetricians and Gynecologist Foundation Annual Scholars' Retreat, "Oogenesis and Ovarian Failure," Magee-Womens Research Institute and Foundation, Pittsburgh, PA	Invited Speaker
2011	Invited Speaker at the 9th Annual Women and Infants Health Care Conference: "Current State of the Art in Genetic Consultation and Diagnosis from Pre-Conception to Birth," Magee-Womens Hospital of UPMC, Pittsburgh, PA.	Invited Speaker
2012	Invited Speaker at the Magee-Womens Hospital of UPMC Board of Directors and V.P. Meeting: "The Future of Genetic Testing," Magee-Womens Hospital of UPMC, Pittsburgh, PA	Invited Speaker
2013	Provost's Inaugural Lecture, Marcus Allen Hogge Chair in Reproductive Sciences, University of Pittsburgh: "Ex Ovo Omnia", Pittsburgh, PA.	Invited Speaker
2014	Invited Speaker at Grand Rounds, Department of Obstetrics and Gynecology, Magee-Womens Hospital, UPMC: "Update on Prenatal Aneuploidy Detection," Pittsburgh, PA.	Invited Speaker
2015	Invited Speaker at the Center for Fertility and Reproductive Endocrinology at Magee-Womens Hospital, UPMC, IVF Retreat: Preimplantation Genetic Screening," Pittsburgh, PA; January 12, 2015	Invited Speaker
2015	Invited Speaker at the Premalignant Working Group Research Symposium at Magee-Womens Hospital, UPMC: "Med 12 Associated Reproductive Pathologies" Pittsburgh, PA.	Invited Speaker
2015	Invited Speaker at the MCCBP Work In Progress Seminar Fall 2015 "Exome Sequencing, Gonadal Failure and Genomic Instability" University of Pittsburgh Cancer Institute, Pittsburgh, PA	Invited Speaker
2016	Invited Speaker at Grand Rounds, Department of Obstetrics Gynecology, UPMC Mercy Hospital: "Preimplantation Genetic Testing: Are We Ready For Whole Genome Sequencing," Pittsburgh, PA.	Invited Speaker

2016	Invited Speaker at Grand Rounds, Department of Obstetrics Gynecology, UPMC Mercy Hospital: "Preimplantation Genetic Testing: Are We Ready For Whole Genome Sequencing," Pittsburgh, PA.	Invited Speaker
2016	Invited Speaker at the Magee-Womens Research Institute 2016 Annual Retreat: "Genetics of Early Folliculogenesis"; Pittsburgh, PA	Invited Speaker
2016	Invited Speaker at the Womens Cancer Research Center Work-In-Progress Seminar: "Genetic Underpinnings of Leiomyoma"; Pittsburgh, PA	Invited Speaker
2017	Invited Speaker at the Magee-Womens Research Institute Work-in-Progress Conference and Research Seminar Series. "Reproductive Aging and Overall Health"; Magee-Womens Research Institute, Pittsburgh, PA.	Invited Speaker

# **CONTINUING EDUCATION AND PROFESSIONAL DEVELOPMENT ACTIVITIES**

2011 Leadership Development for Physicians in Academic Health Centers, Harvard School of Public Health

# **GOVERNMENT AND OTHER PROFESSIONAL SERVICE**

2008 - 2012	NIH CMIR Study Section, Bethesda, MD	Regular Member
2009 - present	NIH Special Emphasis Panels	Ad hoc Member
2011 - 2011	NIH/NICHD VISION Workshop on Pregnancy and Pregnancy Outcomes, Bethesda, MD.	Panel Member
2011 - 2011	NIH/NICHD Vision Workshop on Reproduction. Bethesda, MD	Panel Member
2014 - 2014	NIH Fragile-X Associated Primary Ovarian Insufficiency, The Eunice Kennedy Shriver National Institute of Child Health and Human, Rockville, MD	Panel Member
2017 - 2017	NIH Panel IRAP Review	Panel Member
2018 - 2018	NIH Panel Genetics Health and Disease Panel.	Panel Member
2018 - 2018	NIH Fragile-X Associated Primary Ovarian Insufficiency Meeting, Bethesda, MD	Co-chair
2018 - present	BIOS Advisory Committee	Member
2018 - present	Institute of Human Genetics Task Force	Member
2019 - present	Reproductive Genetics Technology Consortium	Member

# UNIVERSITY AND PUBLIC SERVICE

#### **SERVICE ACTIVITIES SUMMARY**

I have served on numerous committees at the Departmental and University level. At the level of the University, I serve on the Precision Medicine Committee, and have served on the search committee for the Ob/Gyn chair in 2015, as well as the current search committee for the Division Director of Reproductive endocrinology. I also served on various committees at the Magee Womens Research Institute, as detailed below. I also served on research and training committees, such as WRHR advisory committee (I am the research director) and T32 training grant (advisory committee).

# UNIVERSITY SERVICE UC SYSTEM AND MULTI-CAMPUS SERVICE

2019 - present	UC Clinical Genomics Group	Member
2019 - present	UC Genomic Data Sharing Group	Member
2019 - present	UC Lab Council	Member

#### **UCSF CAMPUSWIDE**

2018 - present	Precision Medicine Committee	Member
2018 - present	Center for Maternal-Fetal Precision Medicine	Advisory Board Member
2018 - present	Institute of Human Genetics Education Committee	Member
2019 - present	Research Data Science Council	Member

#### **SCHOOL OF MEDICINE**

2018 - present	Institute of Human Genetics, Steering Committee	Member
2019 - present	CRS Search Committee	Member
2019 - present	Institute of Human Genetics Search Committee	Member
2018 - 2019	HGTF Task Force	Chair, Clinical Genomics

# **SERVICE AT OTHER UNIVERSITIES**

2002 - 2009	Baylor College of Medicine	Obstetrics and Gynecology Resident Admissions Committee
2004 - 2008	Baylor College of Medicine	Medical School Admissions Committee

2008 - 2009	Baylor College of Medicine	Grand Rounds Committee
2007 - 2009	Baylor College of Medicine	Resident Research Committee
2008 - 2009	Baylor College of Medicine	Residency Education Committee
2005 - 2009	Baylor College of Medicine	Medical Scientist Training Program (MSTP) Faculty Operating Committee
2006 - 2009	Baylor College of Medicine	Scientific Integrity Committee
2009 - 2018	University of Pittsburgh	Obstetrics and Gynecology Departmental Steering Committee
2011 - 2018	University of Pittsburgh	Magee-Womens Research Institute Postdoctoral Selection Committee
2011 - 2018	University of Pittsburgh	Personalized Medicine Task Force
2011 - 2018	University of Pittsburgh	Magee-Womens Research Institute Steering Committee
2012 - 2018	University of Pittsburgh	UPMC Graduate Medical Education Committee
2013 - 2018	University of Pittsburgh	Medical Scientist Training Program
2013 - 2018	University of Pittsburgh	Molecular Genetics and Developmental Biology Graduate Program Faculty Member

2013 - 2018	University of Pittsburgh	Magee-Womens Basic and Translational Reproductive Health Training Program, Advisory Committee
2015 - 2018	University of Pittsburgh	Magee-Womens Basic and Translational Reproductive Health Training Program, Research Director
2017 - 2018	University of Pittsburgh	T32 Training grant co-Director: Reproductive Development from Gonad to Fetus

#### COMMUNITY AND PUBLIC SERVICE

2012 - 2017	March of Dimes Basil O'Connor Starter Scholar Resear Award Advisory Committee	ch Committee Member
2013 - 2006	March of Dimes Trans-Disciplinary Centers (TDC) Advisory CommitteeFounding Member: Doctors for Change, Houston, Texas	Committee MemberFounding Member
2006 - 2009	Doctors for Change, Houston, Texas. Dedicated to improving access to healthcare for all.	Founding Member

#### CONTRIBUTIONS TO DIVERSITY

#### **CONTRIBUTIONS TO DIVERSITY**

Initiated population based genomic sequencing of UCSF patients (https://www.3dhealthstudy.org/) with emphasis on increasing diversity among participants and collaborating with Nguyen Tung and Special Populations for Health Equity in Research and Education (SPHERE) Committee, which consists of diverse community leaders and a UCSF team with experience with diverse research participants.

#### **TEACHING AND MENTORING**

#### **TEACHING SUMMARY**

Over the past decade, I have participated as a guest lecturer to several courses dealing with genetics, from animal models, to practical aspects of prenatal, adult/cancer genetics, as well as ethics of clinical and research genetics. Moreover, I participate monthly in didactic lectures of ob/gyn and pathology residents during their rotations in the genetics department. These lectures focus on reproductive and cancer genomics. I have also participated in 1-2 per year for 10-hour total involvement in small group sessions of third year medical students that rotate through the department of obstetrics and gynecology. I served as the Program Director for the

Clinical Genetics and Genomics training program at University of Pittsburgh that encompassed: 1). Clinical Genetics and Genomics Residency (2 per year), 2). Laboratory Genetics and Genomics fellowship (2 per year), and 3) Clinical Biochemical Genetics. I was involved with hands on, laboratory and patient-based teaching in addition to overall evaluation in resident and fellowship competencies.

#### RESEARCH AND CREATIVE ACTIVITIES

#### **RESEARCH AWARDS - CURRENT**

leiomvomas form

1. P50 HD098580 PI 20% % effort Rajkovic (PI) NICHD 04/01/2019 03/31/2024

THE ORIGIN AND CELLULAR HETEROGENEITY \$ 352,000 direct/yr \$ 1,760,000 total

OF UTERINE LEIOMYOMAS

Proposal focuses on understanding the origins of leiomyomas using cell lineage tracing techniques as well as single cell sequencing

I managed the project, performed experiments, supervised postdocs and technicians, and participated in drafting manuscripts.

2. R01 HD088629-01 Principal Investigator 20% % effort Rajkovic (PI) NICHD 07/01/2017 06/30/2022

Med12 Mechanisms of Uterine Leiomyoma Formation \$ 277,221 direct/yr \$ 1,386,105 total

Proposal focuses on the new model of Med12 tumorigenesis and mechanisms of how uterine

I managed the project, performed experiments, supervised postdocs and technicians, and participated in drafting manuscripts.

3. 2016229-142062	PI	5% % effort	Rajkovic (PI)
UCSF School of Medicin	ie	06/01/2019	05/31/2022
PILOT WHOLE GENOM	E SEQUENCING AND	\$ 1,252,374	\$ 1,252,374 total
RETURN OF RESULTS	AT UCSF	direct/yr 1	

The purpose of this project is to ultimately determine best consenting and recruitment practices for UCSF patients, optimal return of results to individuals and their families, bioethical considerations, novel predictive algorithms for disease, novel diagnostics and OMICS guided interventions. Such knowledge will be critical to routine responsible use of genomic testing in populations and continuously engage UCSF consumers/patients with their own genomics/OMICS and clinical health, create trust and collaboration between consumer and clinician, and improve personal health maintenance and outcomes

I am the PI and oversee implementation and execution of this project.

4. PI 5% % effort Rajkovic (PI)

ITGSC - UCSF IT Information Access and Innovation 08/01/2020 07/31/2022

Genomic Data Repository IT Infrastructure to Improve \$ 253,000 direct/yr \$ 253,000 total Information Access and Innovation 1

USCF does not have a centralized genetic repository readily accessible to clinicians and researchers. With exponential rise in genetic testing across many services, there is a need by many stakeholders to centralize these data sets. Our proposed solution is a Genomic Information Commons (GIMS) database system specialized for the storage and access of genetic and phenotypic data that will help clinicians and researchers determine disease pathogenicity and research targets. In summary, the GIMS will be a research tool for all providers at UCSF to understand underlying genetic mechanisms for patients with complex disorders and identify targets for future therapeutics. We propose to leverage existing software platforms created by researchers at Children's Hospital of Philadelphia as part of the NIH funded Gabriella Miller Kids Pediatric Research Program. We would like to collaborate with them and create an integrated resource for UCSF researchers and clinicians to provide a centralized access with harmonized genomic and phenotypic data for research and future clinical purposes.

I am the overall PI and will oversee efforts to implement Genomic Data repository and to interact with BCHSI

#### **RESEARCH AWARDS - SUBMITTED**

1. R01 HD105256	Multi-PI	10% % effort	Rajkovic (PI)

NHGRI 4/2021 3/2026

' Trio analysis of Recurrent pregnancy loss Integrated \$1,000,000 \$5,000,000 total bioinformatics genOmics Study (TRIOS)' direct/yr 1

Identify genomics of early pregnancy losses

This is a collaboration with Stanford and I will be the UCSF PI of this project.

2. U24 HD104584	PI	10% % effort	Rajkovic (PI)
NICHD		04/01/2021	03/31/2024
Reproduction and Infer	tility Expert Gene Curation Panel	·	\$ 750,000 total
		direct/vr 1	

Panel to curate genes for little understood area of infertility

I will oversee the project, reviewers and coordinate all the interactions to curate genomic variants.

#### **RESEARCH AWARDS - PAST**

1.	R01 HD088629	co-PI	5% % effort	Rajkovic (PI)
	NICHD		09/01/2018	08/31/2019
	Small molecule GPR10 ar	ntagonists for the	\$ 300,000 direct/yr	\$ 300,000 total
	treatment of uterine leiom	yoma	1	

Proposal focuses on discovering small molecules to target leiomyomas

I managed the project, performed experiments, supervised postdocs and technicians, and participated in drafting manuscripts.

2.	R01 HD044858-09 NICHD	Principal Investigator	30% % effort 9/25/2014	Rajkovic (PI) 6/30/2019
	Transcriptional Regulation	n of Early Folliculogenesis	\$ 271,984 direct/yr 1	\$ 1,359,920 total
	-	<ul><li>ovarian folliculogenesis and primary follicle formation</li></ul>		8 transcriptional
	I managed the project, pe participated in drafting ma	rformed experiments, superanuscripts.	ervised postdocs and	I technicians, and
3.	R01 HD044858 NICHD	Principal Investigator	30% % effort 04/01/2004	Rajkovic (PI) 03/31/2009
	Transcriptional Regulation	n of Early Folliculogenesis	\$ 175,000 direct/yr	\$ 875,000 total
	In this proposal we studie oogenesis and primordial	d the role of newly discove follicle formation	ered homeobox, Nobe	ox, in driving early
	I managed the project, pe participated in drafting ma	rformed experiments, superanuscripts.	ervised postdocs and	I technicians, and
١.	K12 HD060128	Research Director	5% % effort	Dale (PI)
	NICHD		04/27/2005	03/31/2010
	Womens Reproductive He Program, Baylor College	•	\$ 596,570 direct/yr	\$ 1,789,710 tota
	This is a K12 training grad careers, at Baylor College	nt for obstetricians and gyn e of Medicine	ecologists interested	l in research
	•	vise and help scholars ach sory committee and individu	•	
j.	R01 HD070647-04	Principal Investigator	25% % effort	Rajkovic (PI)
	NICHD		04/1/2012	1/31/2018
	Genomic Basis of Premat	ture Ovarian Insufficiency	\$ 463,794 direct/yr 1	\$ 2,153,105 tota
	Whole exome sequencing	g of individuals with POI to	determine their gene	etic etiology
	I managed the project, pe participated in drafting ma	rformed experiments, superanuscripts.	ervised postdocs and	I technicians, and
3.	R21 HD074278	Principal Investigator	10% % effort	Rajkovic (PI)
	NICHD		05/01/2014	04/30/2016
	Genomic integrity of the > Specific Autosomal Gene	Chromosome and Ovary-	\$ 168,372 direct/yr 1	\$ 275,000 total
	We constructed high reso individuals with ovarian in	lution X chromosome to de sufficiency	etermine copy numbe	er variants in
	I managed the project, pe participated in drafting ma	rformed experiments, supe anuscripts.	ervised postdocs and	I technicians, and
7.	R01 HD074278	Co-investigator	5% % effort	Peters (PI)
	NICHD		05/15/2011	05/10/2016

Non-Invasive Detection of Fetal Aneuploidy by Next- \$ 466,647 direct/yr \$ 2,333,236 total Generation DNA Sequencing 1

Research on cell free DNA and its utility in aneuploidy detection

I served as a co-investigator on the project, facilitated sample collection from our prenatal diagnosis clinic, and participated in drafting manuscripts.

8. R01 HD56351 Principal Investigator 20% % effort Rajkovic (PI)
NICHD 09/30/2009 08/31/2012

LIM Homeodomain Regulated Genetic Pathways in \$261,229 direct/yr \$783,687 total Oogenesis and Ovarian Failure 1

The project focused on downstream pathways of LHX8 in oocyte differentiation and folliculogenesis

I managed the project, performed experiments, supervised postdocs and technicians, and participated in drafting manuscripts

9. R03 HD054829 Principal Investigator 10% % effort Rajkovic (PI)

NICHD 09/04/2009 07/31/2011

The Role of Hormad1 in Germ Cell Development \$50,000 direct/yr \$100,000 total and Meiosis

The project characterized a novel meiotic factor we discovered during a screen for oogenesis specific genes

I managed the project, performed experiments, supervised postdocs and technicians, and participated in drafting manuscripts.

10. R21 HD058125 Principal Investigator 5% % effort Rajkovic (PI)

NICHD 04/01/2008 03/31/2011

Genetics of Human Ovarian Failure \$ 150,000 direct/yr \$ 275,0000 total

Candidate gene approaches to human ovarian failure

I managed the project, performed experiments, supervised postdocs and technicians, and participated in drafting manuscripts.

11. R21 HD058125 Principal Investigator 10% % effort Rajkovic (PI)

NICHD 04/01/08 03/31/11

Genetics of Human Ovarian Failure \$ 150000 direct/yr \$ 275000 total

Candidate gene approaches to human ovarian failure

I managed the project, performed experiments, supervised postdocs and technicians, and participated in drafting manuscripts.

12. N01 WH4-2110	Principal Investigator	5% % effort	Rajkovic (PI)
NHLBI		6/01/2005	7/01/2009
Womens Health Initiative	, Baylor Field Office	\$ 500,000 direct/yr	\$ 2,000,000 total

Baylor Field Office maintained records of participants and follow ups, as well as participated in analysis and manuscript drafting.

I was Principal Investigator for the Baylor College of Medicine Field Office for the Womens Health Initiative (WHI) study; I oversaw a staff of three individuals and participated in data analyses and manuscript drafting.

13. March of Dimes, #6- FY05-70	Principal Investigator	5% % effort	Rajkovic (PI)
March of Dimes		06/01/2005	05/31/2008
Genetic Pathways in Earl	y Folliculogenesis	\$ 100,000 direct/yr 1	\$ 300,000 total

Studies focused on mechanisms behind germ cell specific regulation of oogenesis I managed the project, performed experiments, supervised postdocs and technicians, and participated in drafting manuscripts.

	14. R03 HD47514	Principal Investigator	10% % effort	Rajkovic (PI)	
	NICHD		08/01/2004	07/31/2006	
The Role of Obox in Mammalian Oogenesis" Studies on the New Family of Homeobox Genes,		\$ 50,000 direct/yr	\$ 100,000 total		
	Ohox	lly of Homeobox Genes,	ı		

We discovered a whole new family of oocyte specific homeobox genes, Obox, and further characterized its function

I managed the project, performed experiments, supervised postdocs and technicians, and participated in drafting manuscripts.

<ol><li>15. Basil O'Connor Award</li></ol>	Principal Investigator	5% % effort	Rajkovic (PI)	
March of Dimes		2/01/2003	1/31/2005	
Genetics of Mammalian	Oogenesis	\$ 50,000 direct/yr	\$ 100,000 total	
		1		

My project focused on discovering novel genes in oogenesis, using a combination of in silico analysis and knockout animal models

I managed the project, performed experiments, supervised postdocs and technicians, and participated in drafting manuscripts.

16. K12 HD063087-07	Research Director	10% % effort	Rajkovic (PI)
NICHD		09/01/2015	06/30/2020
Magee-Womens Basic a	and Translational	\$ 332,783 direct/y	r \$ 1,663,915 total
Reproductive Health Tra	nining Program	1	

This is a K12 training grant for obstetricians and gynecologists interested in research careers

Research director who recruits, evaluates and oversees training of qualified OB/GYN scholars.

# PEER REVIEWED PUBLICATIONS

Fishel BR, Ragheb JA, Rajkovic A, Haribabu B, Schweinfest CW, Dottin RP. (1985)
 Molecular cloning of a cDNA complementary to a UDP-glucose pyrophosphorylase mRNA
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## **BOOKS AND CHAPTERS**

- 1. Rajkovic A, Pangas S, Matzuk M. (2005) "Follicular Development: Mouse, Sheep and Human Models." In: Neill JD, editor. Knobil and Neill's Physiology of Reproduction, Volume 2, Third Edition. San Diego, CA: Elsevier Academic Press; 2005. p. 383-410.
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- 3. Hogge WA and Rajkovic A. (2014) Practical Genetics for the Ob-Gyn. McGraw-Hill Professional. Publication Date: October 21, 2014.
- Pangas S and Rajkovic A. (2014) Follicular Development: Mouse, Sheep and Human Models. In: Plant TM, Zeleznik AJ, editors. Knobil & Neill's Physiology of Reproduction, Fourth Edition. India: Elsevier; 2014. Book Chapter. Publication Date: December 19, 2014

# SIGNIFICANT PUBLICATIONS

 Shin YH, Ren Y, Suzuki H, Golnoski KJ, Ahn HW, Mico V, Rajkovic A. (2017) Transcription Factors SOHLH2 Coordinate Differentiation without Affecting Meiosis I. J Clin Invest. 2017 May 15. pii:90281:10. 1172/JCI90281. PMID: 28504655

Previous studies have suggested that oocyte differentiation proceeds independent of meiosis. Although this has been suggested, it has never been shown how it happens and factors that drive it. In this manuscript, our group shows for the first time that Sohlh1 and Sohlh2 are the universal drivers of gamete differentiation independent of meiosis, and that these events are sexually dimorphic, as downstream pathways are different in female and male gonads, yet Sohlh1 and Sohlh2 are universal drivers. These findings are important considering efforts to differentiate oocytes from iPS cells and modify germline. Role: senior author, designed the research question, directed and interacted with postdocs and collaborators, co-wrote the manuscript.

- Mittal P, Shin YH, Yatsenko SA, Castro CA, Surti U, Rajkovic A. (2015) Med12 gain-offunction mutation causes leiomyomas and genomic instability. J Clin Invest. 2015 Jul 20. pii: 81534. doi:10.1172/JCI81534. PubMed PMID: 26193636
  - We (PMID 22428002) and others have shown that 70% of uterine leiomyomas carry a variant in the exon 2 of Med12 gene. This association was intriguing but no experiments were conclusive that it is causative. Here we showed for the first time, that this variant causes uterine hyperplasia and leiomyomas in the mouse model, and that Med12 variant effects are gain of function and not due to loss of function. This publication has significant impact for other tumors, where variants in Med12 have recently been discovered, including breast phyllodes tumors and prostate cancer. Role: senior author, designed the research question, directed and interacted with postdocs and collaborators, co-wrote the manuscript.
- 3. AlAsiri S, Basit S, Wood-Trageser MA, Yatsenko SA, Jeffries EP, Surti U, Ketterer DM, Afzal S, Ramzan K, Faiyaz-Ul Haque M, Jiang H, Trakselis MA, Rajkovic A. (2014) Exome sequencing reveals MCM8 mutation underlies ovarian failure and chromosomal instability. J Clin Invest. Dec 1. pii: 78473. doi: 10.1172/JCI78473. PMID: 25437880

This paper shows for the first time that MCM8 mutations inherited in autosomal recessive fashion can cause gonadal dysgenesis and genomic instability. Previous menopause GWAS studies showed that SNPs in the MCM8 gene show some of the strongest association with menopause, however, whether this association was a true cause and effect with MCM8 or some surrounding genetic element was unknown. Our family based whole exome sequencing study as well as a follow up study (PMID:25480036) on its partner, MCM9, showed that MCM8 and MCM9 play a direct role in the spectrum of disorders from gonadal dysgenesis to premature menopause. Role: senior author, designed the research question, directed and interacted with postdocs and collaborators, co-wrote the manuscript.

- 4. Wood-Trageser MA, Gurbuz F, Yatsenko SA, Jeffries EP, Kotan LD, Surti U, Ketterer DM, Matic J, J, Jiang H, Trakselis MA, Topaloglu AK, Rajkovic A. (2014) MCM9 mutations are associated with ovarian failure, short stature, and chromosomal instability. Am J Hum Genet Dec 4;95(6):754-62. doi: 10.1016/j.ajhg.2014.11.002. PubMed PMID: 25480036; PubMed Central PMCID: PMC425997
  - We spent significant effort determining genetic causes of human gonadal failure, and this manuscript discovers MCM9 as a new gene involved in gonadal dysgenesis. Moreover, this gene causes chromosomal instability, just like MCM8, and has led to a hypothesis that a subset of women with infertility may have adverse medical events due to genetic variation at DNA damage response genes. These and other studies have opened investigation into the concept of fertility and overall health. Role: senior author, designed the research question, directed and interacted with postdocs and collaborators, co-wrote the manuscript.
- 5. Suzuki H, Ahn HW, Chu T, Bowden W, Gassei K, Orwig K, Rajkovic A. (2012) SOHLH1 and SOHLH2 coordinate spermatogonial differentiation. Dev Biol. 2012 Jan; 361(2):301-12. PubMed PMID: 22056784; PubMed Central PMCID: PMC3249242.
  - We previously discovered that Sohlh1 and Sohlh2 encode spermatogonia and oocyte specific helix-loop-helix transcriptional regulators, and in this manuscript, we showed that these two unique regulators physically interact and regulate spermatogonial differentiation as heterodimers. We also showed that SOHLH1 and SOHLH2 suppress genes involved in SSC maintenance, and induce genes important for spermatogonial differentiation. Role: senior author, designed the research question, directed and interacted with postdocs and collaborators, co-wrote the manuscript.
- Shin Y, Choi Y, Erdin SU, Yatsenko SA, Kloc M, Yang F, Wang PJ, Meistrich ML, Rajkovic, A (2010) HORMAD1 mutation disrupts synaptonemal complex formation, recombination, and chromosome segregation in Mammalian meiosis. PLoS Genet. 2010 Nov: 6(11):e1001190. PubMed PMID: 21079677; PubMed Central PMCID: PMC2973818
  - Little has been known regarding germ cell specific checkpoints of meiosis. In this manuscript as well as subsequent manuscript (PMID 23759310), we describe functional studies of our previous discovery of a highly expressed and novel germ cell specific gene, Nohma (PMID 15567723) later renamed Hormad1. In above manuscript, we showed that Hormad1 gene encodes a first known germ cell specific checkpoint protein that modulates DNA double-strand break repair during female meiosis, and the first to regulate meiotic sex chromosome inactivation. HORMAD1 does checkpoint surveillance in part via DSB-independent pathways, because Hormad1 deficiency can rescue Spo11 mutants. Moreover, Hormad1 deficiency promotes DMC1-independent DSB repairs, which in turn helps asynaptic Hormad1 deficient oocytes resist perinatal loss.

7. 24. Pangas S, Choi Y, Ballow D, Zhao Y, Westphal H, Matzuk M, Rajkovic A. (2006) Oogenesis requires germ cell-specific transcriptional regulators SOHLH1 and LHX8. Proc Natl Acad Sci U.S.A. 2006 May; 103(21): 8090-5. PubMed Central PMCID: PMC1472434.

We discovered a transcriptional cascade that regulates oogenesis and involves Sohlh1 and downstream factor Lhx8. Sohlh1 and Lhx8 have become markers for oocyte differentiation and have been used by many for studies involving this important event. Moreover, these genes regulate oogenesis, as well as early embryo development, by impacting accumulation of maternal effect genes. This study added to the growing number of germ cell specific transcriptional regulators like Nobox, that are involved in oocyte differentiation and growth. Our and other laboratories have shown that many of these transcriptional regulators are involved in the causes of human gonadal dysgenesis.

- 8. Chlebowski RT, Kuller LH, Prentice RL, Stefanick ML, Manson JE, Gass M, Aragaki AK, Ockene JK, Lane DS, Sarto GE, Rajkovic A, Schenken R, Hendrix SL, Ravdin PM, Rohan TE, Yasmeen S, Anderson G; WHI Investigators. (2009) Breast cancer after use of estrogen plus progestin in postmenopausal women. N Engl J Med. 2009 Feb; 360(6):573-87. PubMed PMID: 19196674; PubMed Central PMCID: PMC3963492.
  - I was the principal investigator of the Baylor College of Medicine Women's Health Initiative field office, that recruited and collected data, as part of the WHI study. Although not a first or senior author on this publication, I contributed significantly to numerous manuscripts that were generated from the WHI, including this one. In this manuscript, we showed that hormone replacement therapy was associated with a significant rise in breast cancer, and the risk declined markedly after discontinuation of combined hormone therapy. This study sparked much debate over timing of hormone replacement therapy, which continues.
- 9. Rajkovic A, Pangas SA, Ballow D, Suzumori N, Matzuk MM. (2004) NOBOX deficiency disrupts early folliculogenesis and oocyte-specific gene expression. Science. 2004 Aug; 305(5687):1157-9. PubMed PMID: 15326356.
  - Transcriptional cascade of early oogenesis was poorly understood, prior to this manuscript. I discovered the first homeobox gene (Nobox) that is oocyte-specific and regulates post-meiotic oocyte differentiation. These studies paved way for discovery of multiple other transcriptional regulators and showed that transcriptional regulation in early folliculogenesis is essential not only for the synthesis of genes important in oocyte growth and differentiation, but also for the synthesis of maternal effect genes that are critical in early post-fertilization embryo development.
- 10. Ballow D, Meistrich M, Matzuk M, Rajkovic A, (2006) SOHLH1 is essential in spermatogonial differentiation. Dev Biol. 2006 Jun; 294(1):161-7. PubMed PMID: 16564520.
  - Spermatogonial differentiation is a unique process that precedes meiosis I in males. I discovered Sohlh1 and Sohlh2 (PMID 16765102), two germ cell specific transcriptional regulators. We previously showed that Sohlh1 drives oocyte differentiation and in this manuscript, we showed that Sohlh1 is the first spermatogonial specific gene that specifically affects differentiation without affecting spermatogonial proliferation and maintenance. These and other studies have led to further investigation into the differentiation cascades of Sohlh1 and Sohlh2, and provided markers for differentiation that are indispensable in the burgeoning field of gamete differentiation and modification in vitro.

11. X-linked ADGRG2 mutation and obstructive azoospermia in a large Pakistani family. Khan MJ, Pollock N, Jiang H, Castro C, Nazli R, Ahmed J, Basit S, Rajkovic A, Yatsenko AN. Sci Rep. 2018 Nov 2;8(1):16280. doi: 10.1038/s41598-018-34262-5. PMID: 30389958

### PATENTS ISSUED OR PENDING

- 1. "Novel germ cell-specific contraceptive target", USPTO Application #: 20030144205
- 2. "Tex14 compositions and modulators thereof to alter intercellular bridge development and function", USPTO Application #: 20060223099
- 3. "Ovarian-derived Hydrogels for Biomedical and Biotechnology Applications", Application #. 62/303,993
- 4. PCT Patent Application No. PCT/US2018/043948, entitled: "Gene Therapy for Treatment Of Infertility".

#### **CONFERENCE ABSTRACTS**

- Abstract presentation at the 48th Annual Meeting of the Society for Gynecologic Investigation, March 14-17, 2001. "Discovery of oocyte-specific transcripts by expressed sequence tag database analysis." Rajkovic A, Yan C, Klysik M, Matzuk M.
- 2. Abstract presentation at the ESHRE, Lisbon, Portugal, April 6-7, 2001. "OBOX, a new family of homeobox genes preferentially expressed in oocytes." Rajkovic A, Yan C, Yan W, Klysik M, Matzuk M.
- 3. Abstract presentation at the Texas Forum for Female Reproduction, 8th Annual Meeting, Houston, TX, May 2-3, 2002. "Identification of oocyte-specific transcription factors by in silico subtraction."
- 4. Abstract presentation at the Connecticut College, Gordon Research Conference, Mammalian Gametogenesis and Embryogenesis, New London, CT, June 30 - July 5, 2002. "Lack of NOBOX, an oocyte-specific homeobox gene, disrupts early folliculogenesis." Rajkovic A.
- 5. Abstract presentation at the American Society for Human Genetics 52nd Annual Meeting, Baltimore, MD, October 15-19, 2002. "Recurrent hydatidiform mole: Further mapping and candidate gene analysis on chromosome 19q13.4." Panichkul P, Rajkovic A, Kashork CD, Shaffer LG, Van den Veyver IB. Baylor College of Medicine, Houston, TX.
- 6. Abstract presentation at the 50th Annual Meeting of the Society for Gynecologic Investigation, Washington, D.C., March 28, 2003, "Transcriptional regulation of early folliculogenesis." Rajkovic A, Suzumori N, Yan C.
- Abstract presentation at the 50th Annual Meeting of the Society for Gynecologic Investigation, Washington, D.C., March 28, 2003, "Identification of expressed sequence tags preferentially expressed in human placentas by in silico subtraction." Miner D., Rajkovic A.
- 8. Abstract presentation at the 51st Annual Meeting of the Society for Gynecologic Investigation, Houston, TX, March 24-27, 2004. "NOHLH is a helix-loop-helix, oocyte specific transcription factor, critical in mammalian gametogenesis." Rajkovic A, Norwood H, Ballow D, Lerman B.

- 9. Abstract presentation at the 52nd Annual Meeting of the Society for Gynecologic Investigation, Los Angeles, CA, March 25, 2005. "NOBOX regulates multiple novel genes potentially critical in oocyte survival." Ballow B, Rajkovic A.
- Abstract Presentation at the Society for the Study of Reproduction Meeting, Quebec City, Canada, July 25, 2005. "NOBOX deficiency and gene expression in the newborn ovaries." Choi Y, Rajkovic A.
- 11. Abstract presentation at the Society for the Study of Reproduction Meeting, Quebec City, Canada, July 27, 2005. "NOHLH, a novel helix loop helix transcription factor, is critical in mammalian gametogenesis." Rajkovic A, Pangas S, Matzuk M, Ballow D.
- 12. Abstract presentation at the Society for the Study of Reproduction Meeting, Omaha, Nebraska, July 31, 2006. "Lhx8 is essential in mammalian folliculogenesis." Choi Y, Rajkovic A.
- 13. Abstract presentation at the Society for the Study of Reproduction Meeting, Omaha, Nebraska, July 30, 2006. "Sohlh1 is essential for spermatogonial differentiation." Ballow D, Meistrich ML, Matzuk M, Rajkovic A.
- 14. Abstract presentation at the Keystone Symposium, February, 21, 2007. "Reproduction: Advances and Challenges, Homeobox transcription factor, NOBOX interacts with the peptidylarginine deiminase 6 promoter." Choi Y., Rajkovic A.
- 15. Abstract presentation at the Society for Gynecological Investigation, Reno, Nevada, March 14-17, 2007. "The role of GDF9 in Premature Ovarian Failure." Zhao H, Qin Y, Kovanci E, Simpson JL, Chen ZJ, Rajkovic A.
- 16. Abstract presentation at the Society for Gynecological Investigation, Reno, Nevada, March 14-17, 2007. "Mutation analysis of NANOS3 in 80 Chinese and 88 Caucasian women with premature ovarian failure." Qin Y, Zhao H, Kovanci E, Simpson JL, Chen ZJ, Rajkovic A.
- 17. Abstract presentation at the Texas Forum for Reproductive Sciences, April 19, 2007. "LHX8 regulates oogenesis upstream of NOBOX." Choi Y., Rajkovic A.
- 18. Abstract presentation at the Society for the Study of Reproduction, July 22, 2007. "Oocyte regulators in ovarian development and failure." Rajkovic A.
- 19. Abstract presentation at the Society for the Study of Reproduction, July 22, 2007. "LHX8 deficiency disrupts early folliculogenesis and oocyte-specific gene expression in the mouse ovary." Choi Y, Ballow D, Zhao Y, Westphal H and Rajkovic A.
- 20. Abstract presentation at the American Society for Human Genetics, October 23-27, 2007. "FIGLA mutations in women with premature ovarian failure." Zhao H, Chen Z-J, Qin Y, Wang S, Simpson JL, Rajkovic A.
- 21. Abstract presentation at the Society for the Study of Reproduction, May 25-30, 2008. "Gene expression of newborn mouse ovary lacking a LIM-homeobox gene, LHX8." Choi Y, Ballow D, Xin Y, Rajkovic A.
- 22. Abstract presentation at the 55th Annual Meeting of the Society for Gynecological Investigation, March 26-29, 2008. "Animal models in the study of Oogenesis."
- 23. Abstract presentation at the 56th Annual Meeting of the Society for Gynecological Investigation, March 17-21, 2009. "Detection of novel CNVs in uterine Leiomyomas using high resolution SNP arrays." Bowden W, Skorupski J, Kovanci E, Rajkovic A.

24. Abstract presentation at the Society for the Study of Reproduction, July 20, 2009. 
"Massive Parallel Sequencing of small RNAs from newborn mouse ovaries identifies novel miRNAs preferentially expressed in the ovaries." Ahn HW, Zhao H, Harris R, Coarfa C, Milosavljevic A, Morin RD, Marra MA, Rajkovic A.

- 25. Abstract presentation at the Society for the Study of Reproduction, July 22, 2009. "HORMAD1 Is a germ cell-specific HORMA containing protein required for synaptonemal complex formation in male meiosis." Shin Y., Raikovic A.
- 26. Abstract presentation at the American Society for Human Genetics Annual Meeting, Honolulu, HI, October 20-24, 2009. "Evaluation of 9p24 risk locus and colorectal adenoma and cancer: a pooled analysis of four studies". Hutter CM, Slattery ML, Berndt SI, Hsu L, Duggan DJ, Muehling J, Caan BJ, Beresford SA, Rajkovic A, Sarto GE, Marshall JR, Hammad N, Wallace RB, Makar KW, Prentice RL, Potter JD, Hayes RB, Peters U
- 27. Abstract presentation at the Proceedings of the 42th Annual Meeting of the Society of Gynecologic Oncologists (SGO), Orlando, FL, March 2010. "HORMA Domain Containing Protein-1 (HORMAD1) and Outcomes in Ovarian Cancer Patients." Shahzad M, Matsuo K, Shin Y, Stone R, Bottsford-Miller J, Lu C, Han H, Rajkovic A, Sood A.
- 28. Abstract presentation at the Association of Genetic Technologists, Phoenix, AZ, June 3-6, 2010. "Prenatal diagnosis of a complex unbalanced rearrangement involving chromosomes 2, 5, and 7 along with multiple interstitial deletions." Kochmar S, Thorson H, Rajkovic A, Sathanoori M, Clemens M, Madan-Khetarpal S, Davis S, Hoover J, Surti U; University of Pittsburgh Medical Center, Pittsburgh, PA.
- 29. Abstract presentation at the 43rd Annual Meeting of the Society for the Study of Reproduction (SSR), July 30 August 3, 2010, Milwaukee, WI. "SOHLH1 and SOHLH2 double knockouts reveal cooperative function in the male germ cell differentiation pathways." Suzuki H, Ahn HW, Bowden W, Rajkovic R.
- 30. Abstract presentation at the 43rd Annual Meeting of the Society for the Study of Reproduction (SSR), July 30 August 3, 2010, Milwaukee, WI. "HORMAD1 Mutation Disrupts Chromosomal Segregation in Oogenesis." Shin YH, Choi YS, Uckac S, Yatsenko SA, Yang F, Wang J, Rajkovic A.
- 31. Abstract presentation at the American Society for Reproductive Medicine, October 2010. "Complex X chromosome rearrangement delineated by array CGH in a woman with primary ovarian insufficiency." Ochalski M, Engle N, Wakim A, Rajkovic A, Surti U.
- 32. Abstract presentation at the 2011 American College of Medical Genetics Annual Clinical Genetics Meeting (ACMG 2011), March 16-20, 2011, Vancouver, British Columbia, Canada. "Congenital diaphragmatic hernia in a prenatally diagnosed 17q12 microdeletion syndrome." Hendrix N, Surti U, Clemens M, Canavan T, Rajkovic A. Magee-Womens Hospital of UPMC, Pittsburgh, PA USA.
- 33. Abstract presentation at the 2011 American College of Medical Genetics Annual Clinical Genetics Meeting (ACMG 2011), March 16-20, 2011, Vancouver, British Columbia, Canada. "The Clinical Utility of Array Comparative Genomic Hybridization in the Prenatal Testing of Fetuses with Multiple Anomalies." Davis S, Surti U, Clemens M, Rajkovic A. Magee-Womens Hospital of UPMC, Pittsburgh, PA.
- 34. Abstract poster presentation at the Society for the Study of Reproduction Annual Meeting Annual Meeting. July 29 August 4, 2011, Oregon Convention Center, Portland, OR. August 2, 2011 Poster Session B Meiosis and Fertilization: "HORMAD1 modulates double

stranded break repair during female meiosis." Shin YH, McGuire M, and Rajkovic A. Magee-Womens Research Institute, Department of Obstetrics and Gynecology and Reproductive Sciences, University of Pittsburgh, Pittsburgh, PA 15213.

- 35. Abstract presentation at the Society for the Study of Reproduction Annual Meeting, July 29 August 4, 2011, Oregon Convention Center, Portland, OR. "LHX8 in oocytes is required for survival of primordial follicle pool and follicular development." Krishna Jagarlamudi and Rajkovic A. Magee-Womens Research Institute. University of Pittsburgh, Pittsburgh, PA.
- 36. Abstract presentation at the 12th International Congress of Human Genetics (ICHG/ASHG), October 11-15, 2011, Montreal Quebec, Canada. "Development and validation of a CGH microarray for clinical diagnosis of hematological malignancies." Yatsenko SA, Gollin SM, Hu J, Sathanoori M, Surti U, Rajkovic A. Ob/GYN & Reproductive Sci, Univ. of Pittsburgh, Magee-Womens Hospital of the UPMC, Pittsburgh, PA USA.
- 37. Abstract presentation at the National Society of Genetic Counselors 30th Annual Education Conference (NSGC), October 27-30, 2011, San Diego Marriott, San Diego, California. "An unusual presentation of Fanconi anemia." Knickelbein K, Dudley B, Thull D, Peffer A, Rajkovic A, Puhalla S. University of Pittsburgh Medical Center, Pittsburgh, PA USA.
- 38. Abstract presentation at the 59th Annual Meeting of the Society for Gynecologic Investigation March 21-24, 2012 Manchester Grand Hyatt, San Diego, CA, USA. "Granulocyte Colony Stimulating Factor Maintains Ovarian Follicle Numbers in Mice Treated with High-Dose Chemotherapy by Decreasing Chemotherapy-Related Ischemia." Skaznik-Wikiel ME, McGuire M, Sukhwani M, Krivak TC, Rajkovic A, and Orwig K. Magee-Womens Research Institute, University of Pittsburgh, Pittsburgh, PA.
- 39. Abstract presentation at the 2012 American Society of Clinical Oncology (ASCO) Annual Meeting, June 1-5, 2012, McCormick Place, Chicago, IL. "Estrogen plus progestin (E+P) and breast cancer incidence and mortality." Chlebowski RT, Anderson GL, Kuller LH, Aragaki, AK, Manson, JE, Stefanick, ML, Johnson K, Gass, M, Lane DS, Ocken J, Sarto G, Wactawski-Wende J, Rajkovic A, Prentice RL. Womens Health Initiative Group.
- 40. Abstract presentation at the 62nd Annual Meeting of the American Society of Human Genetics, November 6-10, 2012, Moscone Center, San Francisco, CA. "Genetic variation and age at natural menarche and menopause in African American women from the Population Architecture using Genomics and Epidemiology (PAGE) Study." Malinowski J, Spencer KL, Carty CL, Franceschini N, Fernandez-Rhodes L, Young A, Cheng I, Ritchie MD, Haiman CA, Wilkens L, Wu C, Matise TC, Carlson CS, Brennan K, Park A, Rajkovic A, Hindorff LA, Buyske S, and Crawford DC.
- 41. Abstract presentation at the 62nd Annual Meeting of the American Society of Human Genetics, November 6-10, 2012, Moscone Center, San Francisco, CA. "Mediator complex subunit 12 (MED12) mutations in uterine leiomyosarcomas." McGuire MM, Jones M, Trucco G, Surti U, Edwards RP, Rajkovic A. Department of Obstetrics, Gynecology and Reproductive Sciences, Magee-Womens Research Institute, Pittsburgh, PA; Department of Pathology, Magee-Womens Hospital of UPMC, Pittsburgh, PA.
- 42. Abstract presentation at the 62nd Annual Meeting of the American Society of Human Genetics, November 6-10, 2012, Moscone Center, San Francisco, CA. "Computational methods for detecting whole-genome triploid samples with Agilent CGH+SNP arrays."

- Adapalli A, Ashutosh A, Peter B, Ghosh J, Curry B, Rajkovic A, Surti U, Yatsenko S. Agilent Genomics, Agilent Labs, Santa Clara, CA; Department of Obstetrics, Gynecology and Reproductive Sciences, University of Pittsburgh, PA.
- 43. Abstract presentation at the 62nd Annual Meeting of the American Society of Human Genetics, November 6-10, 2012, Moscone Center, San Francisco, CA. "Association of A Deletion Downstream of NKX2-1 With Benign Hereditary Chorea: Transfer of Benign CNV to Likely Pathogenic Category." Surti U, Liao J, Coffman K, Hu J, Sathanori M, Yatsenko SA, Madan-Khetarpal S, McGuire M, Rajkovic A. University of Pittsburgh Medical Center, Pittsburgh, PA.
- 44. Abstract presentation at the 62nd Annual Meeting of the American Society of Human Genetics, November 6-10, 2012, Moscone Center, San Francisco, CA. "High resolution copy number profiling of the X chromosome in clinical diagnosis." Yatsenko SA, Madan-Khetarpal S, Rajkovic A, Surti U. Department of Obstetrics, Gynecology and Reproductive Sciences, Department of Pathology, University of Pittsburgh School of Medicine; and the Department of Medical Genetics, Children's Hospital of Pittsburgh of UPMC, Pittsburgh, PA.
- 45. Abstract presentation at Cold Spring Harbor Germ Cell Meeting, October 2-6, 2012. LIM-homeobox protein 8 inhibits primordial oocyte activation that is independent of the KIT signaling pathways. Suzuki H, Jagarlamudi K and Rajkovic A, University of Pittsburgh Department of Obstetrics, Gynecology and Reproductive Sciences, Pittsburgh, PA.
- 46. Abstract presentation at the Society for the Study of Reproduction Annual Meeting, August 13-16, 2012. "Molecular Mechanisms and Consequences of Premature Ovarian Aging". Rajkovic A, University of Pittsburgh Department of Obstetrics, Gynecology and Reproductive Sciences, Pittsburgh, PA.
- 47. Abstract presentation at American Urology Association Annual Meeting (AUA2013) May 4-8, 2013, in San Diego, CA. "High resolution copy number profiling of the X chromosome in clinical diagnosis in sex-reversal DSD patients." Schneck FX, Yatsenko SA, Fox J, Rajkovic A, Witchel SF. Department of Urology, Division of Pediatric Endocrinology and Department of Pediatrics, Children's Hospital of Pittsburgh of UPMC, Department of Obstetrics, Gynecology and Reproductive Sciences, Magee-Womens Hospital of UPMC, University of Pittsburgh, Pittsburgh, PA.
- 48. Abstract presentation at American College of Medical Genetics and Genomics Annual Meeting, March 19-23, 2013, Phoenix, AZ. "Clinical utility of limited chromosome analysis as an adjunct to first-tier cytogenomic microarray testing." Sathanoori M, Surti U, Hu J, Yatsenko S, Rajkovic A. University of Pittsburgh School of Medicine, Magee-Womens Hospital of University of Pittsburgh Medical Center, Pittsburgh, PA.
- 49. Abstract presentation at American College of Medical Genetics and Genomics Annual Meeting, March 19-23, 2013, Phoenix, AZ, "Improved interpretation of chorionic villus sampling results using microarray in conjunction with traditional cytogenetics". Steele D, Rajkovic A, Yatsenko S, Sathanoori M, Hu J, Surti U. Center for Medical Genetics and Genomics, Magee-Womens Hospital of UPMC, Pittsburgh, PA.
- 50. Abstract presentation at American College of Medical Genetics and Genomics Annual Meeting, March 19-23, 2013, Phoenix, AZ, "10p11.23-p12.1 microdeletion: Mohawk (MKX) gene haploinsufficiency affects testis migration". Mroczkowski HJ, Yatsenko SA, Arnold G, Schneck F, Rajkovic A. Departments of Obstetrics, Gynecology and

Reproductive Sciences, Pathology, Pediatrics, Urology, Human Genetics, Magee-Womens Research Institute, University of Pittsburgh School of Medicine, Pittsburgh, PA.

- 51. Abstract presentation at American College of Medical Genetics and Genomics Annual Meeting, March 19-23, 2013, Phoenix, AZ. "Prenatal diagnosis of trisomy 6q25.3-qter by array-CGH in a fetus with ultrasound abnormalities and an apparently normal karyotype". Thakur P, Rajkovic A, Yatsenko S. Division of Medical Genetics, Department of Obstetrics, Gynecology and Reproductive Sciences, Magee-Womens Hospital of University of Pittsburgh Medical Center, Pittsburgh, PA.
- 52. Abstract presentation at the 46th Annual Meeting of the Society for the Study of Reproduction, July 22-26, 2013, Montreal, Canada." The role of mediator complex subunit 12 (Med12) in reproductive tract biology". Mittal P, Shin YH, Rajkovic A. Departments of Human Genetics, Obstetrics, Gynecology and Reproductive Sciences, Magee-Womens Research Institute, Department of Pathology, University of Pittsburgh, Pittsburgh, PA.
- 53. Abstract presentation at the Endocrine Society's 95th Annual Meeting & Expo, June 15-18, 2013, San Francisco, CA. "Hypergonadotropic hypogonadism associated with triplication of the short arm of chromosome 11." Gurtunca N, Yatsenko SA, Rajkovic A, Witchel SF. Children's Hospital of Pittsburgh of UPMC, Magee-Womens Hospital of UPMC, University of Pittsburgh School of Medicine, Pittsburgh, PA.
- 54. Abstract presentation at the 63rd Annual Meeting of the American Society of Human Genetics, October 22-26, 2013, Boston, MA. "High resolution copy number analysis of genes involved in gonadal differentiation in patients with disorder of sexual development". Yatsenko SA, Schneck FX, Fox J, Madan-Khetarpal S, Witchel SF, Surti U, Rajkovic A. Departments of Obstetrics, Gynecology and Reproductive Sciences, Pathology, Urology, Pediatrics and Medical Genetics, Magee-Womens Hospital of UPMC, Children's Hospital of Pittsburgh of UPMC, University of Pittsburgh School of Medicine, Pittsburgh, PA.
- 55. Abstract presentation at the 63rd Annual Meeting of the American Society of Human Genetics, October 22-26, 2013, Boston, MA. "Overlapping phenotype of Silver-Russell-like and 14q32 microdeletion syndromes in a child with submicroscopic 11p15.5 duplication and 14q32 deletion". Mroczkowski HK, Lowenstein DB, Abdel-Humid H, Saller DN, Rajkovic A, Yatsenko SA. Department of Obstetrics, Gynecology and Reproductive Sciences, Magee-Womens Research Institute, and Departments of Pediatric Neurology, Pathology, Human Genetics, University of Pittsburgh School of Medicine, Pittsburgh, PA.
- 56. Abstract presentation at the Annual American College of Medical Genetics Clinical Genetics Meeting, March 25-29, 2014, Nashville, TN. "Impact of early speech intervention for a child with a FOXP1 gene microdeletion." Mroczkowski HJ, Azage M, Canale EM, Cummings D, Finegold D, Rajkovic A, Yatsenko S. University of Pittsburgh School of Medicine, UPMC Children's Hospital of Pittsburgh, Magee-Womens Hospital of UPMC, Pittsburgh, PA.
- 57. Abstract in presentation at the ASA 38th Annual Meeting, Testis Workshop, Hyatt Regency, San Antonio, Texas (April 2013). "Intact full length RNAs are well-preserved in undamaged sperm irrespective of clinical parameter" Georgiadis A, Jaffe T, Sanfillippo J, Orwig K, Volk E, Rajkovic A, Yatsenko AN. Department of Obstetrics, Gynecology and Reproductive Sciences, Magee-Womens Research Institute, and Departments of Pediatric Neurology, Pathology, Human Genetics, University of Pittsburgh School of Medicine, Pittsburgh, PA

58. Abstract in presentation at the 29th Annual Meeting of the European Society of Human Reproduction and Embryology, London, England (2013). "Multiple mutations discovered in a familial case of azoospermia using whole exome sequencing". Yatsenko, AN, Georgiadis AP, McGuire MM, Zorrilla M, Bunce KD, Peters D. Rajkovic A. Olszewska M, Kurpisz M. Department of Obstetrics, Gynecology and Reproductive Sciences, Magee-Womens Research Institute, and Departments of Pediatric Neurology, Pathology, Human Genetics, University of Pittsburgh School of Medicine, Pittsburgh, PA.

- 59. Abstract presentation at the Human Genome Meeting 2014, Human Genome Organisation, April 27-30, 2014, Geneva, Switzerland. "Linkage analysis coupled with exome sequencing identified defects in the gene MCM8 causing premature ovarian insufficiency. Basit S, AlAsiri S, Wood M, Rajkovic A. Center for Genetics and Inherited Diseases, Taibah University Madinah, Madinah Al-Munawarah, Assisted Reproductive Technologies Unit, King Khalid University Hospital, Riyadh, Saudi Arabia, Magee-Womens Research Institute, Department of Obstetrics and Gynecology and Reproductive Sciences, University of Pittsburgh, Pittsburgh, PA.
- 60. Abstract presentation at the 2014 Society for the Study of Reproduction Annual Meeting, July 19-23, 2014 Grand Rapids, MI. "Mutant MCM8 induces human premature ovarian insufficiency and chromosomal instability", Wood MA, Yatsenko SA, AlAsiri S, Jeffries E, Afzal S, Ramzan K, Faiyaz M, Haque UI, Jiang H, Trakselis M, Basit S, Rajkovic A. Department of Obstetrics, Gynecology and Reproductive Sciences, Magee-Womens Research Institute and Department of Chemistry, Chevron Science Center, University of Pittsburgh, IVF & ART Unit, IVF & ART Unit, Department of Obstetrics and Gynecology, Prince Naif Centre for Immunology Research, College of Medicine, King Saud University, Riyadh, Saudi Arabia and Center for Genetics and Inherited Diseases, Taibah University Madinah Al-Munawarah, Saudi Arabia.
- 61. Poster presentation at the 2014 Society for the Study of Reproduction Annual Meeting, July 19-23, 2014 Grand Rapids, MI. "Lhx8 suppresses primordial follicle activation through LIN28A-AKT dependent pathways" (Poster). Ren Y, Suzuki H, Jagarlamudi K, Shin Y, Jiang, H, Golnoski K, Rajkovic A. Magee-Womens Research Institute, Department of Obstetrics and Gynecology and Reproductive Sciences, University of Pittsburgh, Pittsburgh, PA.
- 62. Poster presentation at the 2014 Society for the Study of Reproduction Annual Meeting, July 19-23, 2014 Grand Rapids, MI. "Embryonic nucleocytoplasmic shuttling of SOHLH2 associates with oocyte differentiation (poster)". Shin Y, Suzuki H, McGuire M, Rajkovic A. Magee-Womens Research Institute, Department of Obstetrics and Gynecology and Reproductive Sciences, University of Pittsburgh, Pittsburgh, PA.
- 63. Platform Presentation at the Society for the Study of Reproduction Annual Meeting July 19-23, 2014. Grand Rapids, Michigan. "Mediator complex subunit 12 (Med12) missense mutation causes uterine leiomyomas in mice". Mittal P, Shin Y, McGuire M, Rajkovic A. Magee-Womens Research Institute, Department of Obstetrics and Gynecology and Reproductive Sciences, University of Pittsburgh, Pittsburgh, PA
- 64. Abstract presentation at the Association of Genetic Technologists 40th Annual Meeting, June 4-6, 2015, Savannah, Georgia. "Molecular analysis and clinical consequences of an Xp-Yq unbalanced translocation in two families". Ketterer DM, Kochmar SJ, Madan-Khetarpal S, Hu J, Surti U, Rajkovic A, Yatsenko SA. Department of Obstetrics, Gynecology and Reproductive Sciences, Department of Pathology, University of

- Pittsburgh School of Medicine; and the Department of Medical Genetics, Children's Hospital of Pittsburgh of UPMC, Pittsburgh, PA
- 65. Poster presentation at the 2015 Society for the Study of Reproduction annual Meeting, June 18-22, San Juan Puerto Rico. "MCM9 mutations are associated with ovarian failure, short stature and chromosomal instability". Rajkovic A, Wood-Trageser MA, Gurbuz F, Yatsenko SA, Jeffries EP, Kotan LD, Surti U, Ketterer DM, Matic J, Chipkin J, Jiang H, Trakselis MA, Topaloglu AK. Magee-Womens Research Institute, Department of Obstetrics and Gynecology and Reproductive Sciences, Department of Human Genetics, University of Pittsburgh, Pittsburgh, PA.
- 66. Poster presentation at the 2015 Society for the Study of Reproduction Annual Meeting June 18-22, 2014, San Juan Puerto Rico. "DCAF17 homozygous mutations cause hypergonadotropic hypogonadism in humans and mice" F. Diao, M.A. Wood-Trageser, Y. Shin, Y. Sheng, F. Gurbuz, L.D. Kotan, H. Jiang, A.K. Topaloglu, A. Rajkovic. Magee-Womens Research Institute, Department of Obstetrics and Gynecology and Reproductive Sciences, University of Pittsburgh, Pittsburgh, PA.
- 67. Poster presentation at the 2015 Society for the Study of Reproduction Annual Meeting June 18-22, 2014, San Juan Puerto Rico "SOHLH1 and SOHLH2 interaction is essential for their cellular localization and oocyte differentiation" Shin Y, Golnoski K, Suzuki H, Ahn H, Rajkovic A. Magee-Womens Research Institute, Department of Obstetrics and Gynecology and Reproductive Sciences, University of Pittsburgh, Pittsburgh, PA.
- 68. Platform Presentation (Trainee Oral Talk Finalist) at the Society for the Study of Reproduction Annual Meeting: San Juan, Puerto Rico. June 18-22, 2015, "Med12 Exon 2 Mutation Causes Genomic Instability" Mittal P, Shin Y, Yatsenko SA, Castro C, Rajkovic, A. Magee-Womens Research Institute, Department of Obstetrics and Gynecology and Reproductive Sciences, University of Pittsburgh, Pittsburgh, PA.
- 69. Platform presentation at the American College of Medical Genetics Annual Meeting, March 8-12, 2016, Tampa, Florida. "Clinical Significance of Single Chromosome Regions Of Homozygosity Detected by Microarray: Uniparental Disomy, Genomic Imprinting, Autosomal Recessive Mutations, Mitotic Recombination, Chromosomal Aberrations and Mosaicism" Yatsenko SA, Ou Z, Sebastian J, Arnold GL, Friehling E, Madan-Khetarpal S, El-Gharbawy A, Hu J, Surti U, Bellissimo D, Rajkovic A. "Departments of Pediatrics, Human Genetics, Obstetrics, Gynecology and Reproductive Sciences, Children's Hospital of Pittsburgh of UPMC, Magee-Womens Research Institute, Department of Pathology, University of Pittsburgh, Pittsburgh, PA.
- 70. Top poster presentation at the American College of Medical Genetics Annual Meeting, March 8-12, 2016, Tampa, Florida "The Clinical Utility and Diagnostic Yield of Chromosomal Microarray in the Neonatal Population" Sklirou E, Sebastian J, Telesco RR, T Yanowitz T, Arnold GL, Hu J, Surti U, Emery SP, Saller DN Jr., Madan-Khetarpal S, Brozanski B, Rajkovic A, Yatsenko SA. Departments of Pediatrics, Human Genetics, Obstetrics, Gynecology and Reproductive Sciences, Children's Hospital of Pittsburgh of UPMC, Magee-Womens Research Institute, Department of Pathology, University of Pittsburgh, Pittsburgh, PA.
- 71. Poster presentation at the American College of Medical Genetics Annual Meeting, March 8-12, 2016, Tampa, Florida. "Prenatal Diagnosis of a Submicroscopic Complex 14q32 Rearrangement in a Fetus with Multiple Cardiac Anomalies" Marinescu PS, Saller DN Jr., Yatsenko SA, Rajkovic A. Magee-Womens Research Institute, Department of Obstetrics

and Gynecology and Reproductive Sciences, Human Genetics, and Pathology, University of Pittsburgh, Pittsburgh, PA

- 72. Platform presentation at the American College of Medical Genetics Annual Meeting, March 8-12, 2016, Tampa, Fl. "Expansion of Phenotype and Genotypic Data in CRB2-Related Syndrome" Lamont R, TanW-H, Innes M, Parboosingh J, Schneidman-Duhovny D, Saller DN. Jr, Yatsenko SA, Rajkovic A, Krall M, Gray K, Mehta L, Pappas J, Rodan L, Steele D, Vanderwall R, Bernier F, Slavotinek A. Magee-Womens Research Institute, Department of Obstetrics and Gynecology and Reproductive Sciences, Human Genetics, and Pathology, University of Pittsburgh, Pittsburgh, PA
- 73. Abstract presentation at 2016 World Biomaterials Congress, May 18-22, 2016, Montreal, Canada "The Effect of ECM Stiffness on Ovarian Follicle Development" Buckenmeyer MJ, Shin Y, Rajkovic A, Brown, B. Magee-Womens Research Institute, Department of Obstetrics and Gynecology and Reproductive Sciences, University of Pittsburgh, Pittsburgh, PA
- 74. Oral presentation at 64th SRI Annual Scientific Meeting, March 15-18, 2017, Orlando, Florida " Med12 is Critical in Reproductive Tract Development" Wang X, Mittal P, Rajkovic A. Magee-Womens Research Institute, Pittsburgh, PA
- 75. Poster presentation at ENDO 2017, April 1-4, 2017, Orlando, Florida "Brain Stem Hypoplasia and Hypogonadotropic Hypogonadism in a Patient with Charge Syndrome Due to a Novel De Novo Mutation in Chromodomain Helicase DNA-Binding Protein 7 (CHD7)" Morsi A, Katari S, Rajkovic A, Gurtunca NChildrens Hospital of Pittsburgh of UPMC, Pittsburgh, PA, University of Pittsburgh, PH, Medical Director of Pittsburgh Cytogenetics Laboratory, Magee Womens Research Institute and Foundation, Department of Obstetrics and Gynecology and Reproductive Sciences University of Pittsburgh, Children's Hospital of Pittsburgh of UPMC, Pittsburgh, PA
- 76. Abstract presentation at the 42nd Annual Meeting of the American Society of Andrology, April 19 April 22, 2017, Miami, Florida "Prenatal and Postnatal Genetic Diagnosis of 45X/46,XY Mosaicism and its Clinical Implications: A 20-Year Study" Aarabi M, Surti U, Witchel S, Schneck F, Rajkovic A, Yatsenko S. Medical Genetics & Genomics Laboratories, Magee-Womens Hospital of UPMC, Department of Obstetrics, Gynecology and Reproductive Sciences, University of Pittsburgh School of Medicine, Department of Pathology, University of Pittsburgh School of Medicine, Department of Human Genetics, Graduate School of Public Health, University of Pittsburgh, Pediatric Endocrinology, Children's Hospital of Pittsburgh of UPMC, Pediatric Urology, Children's Hospital of Pittsburgh of UPMC, Magee-Womens Research Institute, Pittsburgh, PA
- 77. Oral presentation at 50th Annual Meeting Society for the Study of Reproduction (SSR). "SOHLH1 and SOHLH2 Coordinate Oocyte Differentiation without Affecting Meiosis I" Ren, Y, Rajkovic A. Magee-Womens Research Institute, Pittsburgh, PA
- 78. Abstract presentation at the 10th International Meeting of Pediatric Endocrinology, September 14 17, 2017, Washington, D.C. "Molecular and Cellular Characterization of Patients with 46,XX Testicular and Ovotesticular Disorders of Sex Development" Touzon MS, Berensztein E, Ramirez P, Garrido NA, Marino R, Galluzzo L, Aliberti P, Costanzo M, Guercio G, Vaiani E, Ciaccio M, Rivarola MA, Belgorosky A, Hospital de Pediatria Prof. Dr. Juan P. Garrahan, Buenos Aires, Argentina; Yatsenko SA, Rajkovic A, Magee-Womens Research Institute, Pittsburgh, PA

79. Poster presentation at ENDO 2018 Meeting, March 17-20, 2018, Chicago, Illinois. "Characterization of a Large Cohort of Subjects with Disorders of Sex Development Admitted to a Single Pediatric Hospital"

- 80. Poster presentation at the 2018 ACMG Annual Clinical Genetics Meeting, April 11-14, 2018, Charlotte, North Carolina. "Copy Number Variations in Patients with Oral Clefts" Ou Z, Hu J, Madan-Khetarpal S, Ortiz D, El Gharbawy A, Sebastian J, Saller D, Bellissimo D, Rajkovic A, Yatsenko SA, Pittsburgh Cytogenetics Laboratory, Center for Medical Genetics and Genomics, Magee Womens Hospital of UPMC, Department of Obstetrics, Gynecology & Reproductive Sciences, University of Pittsburgh School of Medicine, Children's Hospital of Pittsburgh of UPMC, Department of Pathology, University of Pittsburgh School of Medicine, Pittsburgh, PA
- 81. Abstract presentation at the 2018 ACMG Annual Clinical Genetics Meeting, April 11-13, 2018, Charlotte, North Carolina. "Incidental Finding of Dystrophinopathy in Patients with Abnormal Neurodevelopmental Presentations and Normal Muscle Tone", Aarabi M, Abdel-Hamid H, Sebastian J, Ortiz D, Madan-Khetarpal S, Rajkovic A, Bellissimo D, Yatsenko SA, Medical Genetics & Genomics Laboratories, Magee Womens Hospital of UPMC, Department of Obstretrics, Gynecology and Reproductive Sciences, University of Pittsburgh School of Medicine, Pediatric Neurology, Children's Hospital of Pittsburgh of UPMC, Department of Pathology, University of Pittsburgh School of Medicine, Department of Human Genetics, Graduate School of Public Health, University of Pittsburgh, Magee Womens Research Institute, Pittsburgh, PA