HEARTLAND GENETICS SERVICES COLLABORATIVE

12TH ANNUAL CONFERENCE

OCTOBER 12 – 14, 2016

LITTLE ROCK, AR

HEARTLAND GENETICS SERVICES COLLABORATIVE

WELCOME LETTER

TWELFTH ANNUAL CONFERENCE

OCTOBER 12 – 14, 2016

LITTLE ROCK, ARKANSAS

Dear Colleagues and Partners,

Welcome to the twelfth regional conference for the Heartland Genetics Services Collaborative. During this meeting, you will learn about the exciting work occurring in the regional collaboratives and learn more about topics that will challenge and inspire. We invite you to visit our sponsors and exhibitors throughout the meeting, to participate in the prioritization exercise on Friday, and, of course, to network with your colleagues. We warmly welcome the many newcomers this year and hope that you become involved in the collaborative.

Thank you for your attendance, participation, and contribution to the Collaborative's mission.

Warmest Regards,

Heartland Regional Coordinating Center Team:
Brad Schaefer
Klaas Wierenga
Lori Williamson Dean
Anayeli Herrera Morales

Conference Planning Committee:
Elizabeth Abbey
Jamey Kendall
Kimberly Noble Piper
Kathy Northrop
Joyal Meyer
Andrea Taylor
Robin Troxell
Larry Weatherford

Heartland Advisory Board:

Stan Berberich
Joni Bosch
Lucy Fossen
Cathy Harbison
David King
Julie Luedtke
Jeremy Penn
Lois Starr
Jackie Whitfield
JoAnn Bolick
Jeanne Egger
Esperanza Font-Montgomery
Jamey Kendall
Tiffany Lepard-Tassin
Kim Noble Piper
Sharmini Rogers
Larry Weatherford

HEARTLAND GENETICS SERVICES COLLABORATIVE

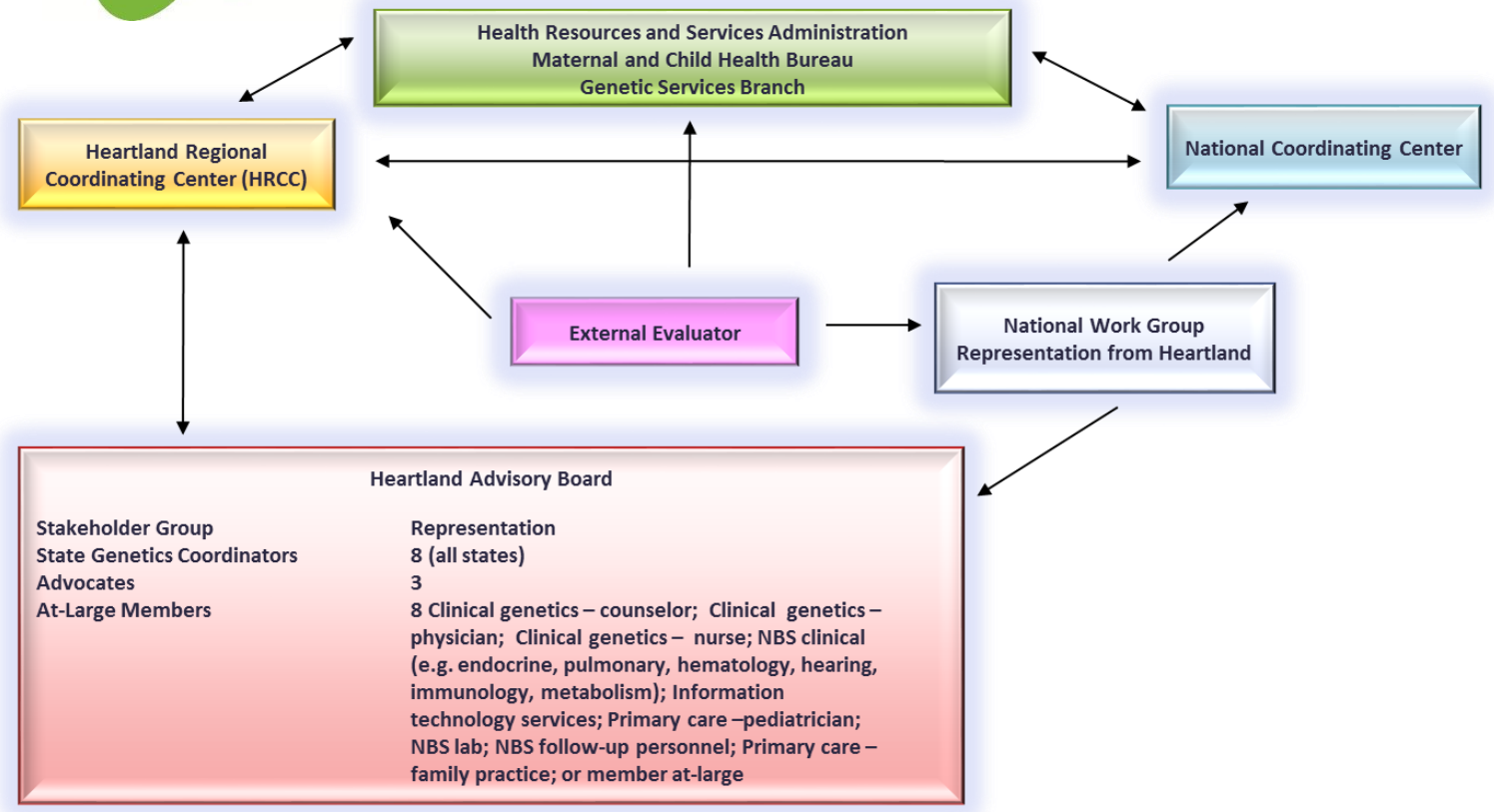
MISSION STATEMENT

February 2010

The Heartland Genetics Services Collaborative is focused on ensuring the best possible outcome for individuals with heritable disorders and optimizing the health of the population throughout the life cycle by improving understanding and awareness of genetics; expanding access to health care; and translating new findings to improve the quality of care within an eight-state region.

HEARTLAND GENETICS SERVICES COLLABORATIVE

ORGANIZATION CHART



Newborn Screening Work Group

(15-20 core members)

3 Advocates

2 Metabolic clinicians

3 Long Term Follow-Up Coordinators

2 Newborn Screening Laboratorians

Clinical Services Work Group

(8-10 core members)

2 Advocates

4 Clinicians

2 Laboratorians

2 State Genetics Coordinators

Early Hearing Detection and Intervention (EHDI)

EHDI coordinators, EHDI staff and Advocates

Advocacy Work Group

(16 core members)

2 Advocates per state

Hispanic Access Advisory Committee

Community members, Clinicians, Public Health

OCTOBER 12 – 14, 2016

HEARTLAND GENETICS SERVICES COLLABORATIVE

SPONSORS

PLATINUM

PerkinElmer

710 Bridgeport Ave
Shelton, CT 06484
www.perkinelmer.com

GOLD

GeneDx

207 Perry Parkway
Gaithersburg, MD 20877
www.genedx.com

Horizon Pharma

150 S Suanders Rd
Lake Forest, IL 60045
www.horizonpharma.com

Shire

300 Shire Way
Lexington, MA 02421
www.shire.com

SILVER

Ambry Genetics

15 Argonaut
Aliso Viejo, CA 92656
www.ambrygen.com

Recordati Rare Diseases

100 Corporate Drive, Ste 104
Lebanon, NJ 08833
www.recordatirarediseases.com

BRONZE

Fulgent Diagnostics

4978 Santa Anita Ave., Ste 205
Temple City, CA 91780
www.fulgentgenetics.com

PreventionGenetics

3800 South Business Park Ave
Marshfield, WI 54449
www.preventiongenetics.com

EXHIBITORS

Baby's First Test
4301 Connecticut Ave NW, Suite
404 Washington, DC 20008
www.babysfirsttest.org

National Coordinating Center (NCC) for the Regional Genetic Services Collaboratives
7101 Wisconsin Avenue, Suite 1101
Bethesda, MD 20814
www.nccrcg.org

University of Arkansas for Medical Sciences (UAMS) Sickle Cell Program
324 Jack Stephens
Little Rock, AR 72204
www.sicklecell.uams.edu

The Heartland Genetics and Newborn Screening Collaborative is supported by a grant from the Genetic Services Branch of the Maternal and Child Health Bureau (MCHB) of the Health Resources and Services Administration (HRSA). Grant Number: H46MC24089

HEARTLAND GENETICS SERVICES COLLABORATIVE

AGENDA

ANNUAL CONFERENCE AND ANCILLARY MEETINGS

OCTOBER 12 – 14, 2016

CAPITAL HOTEL

LITTLE ROCK, AR

WEDNESDAY, OCTOBER 12

TIME	EVENT	LOCATION
09:00 – 01:00	**Hispanic Access Advisory Committee Meeting (HAAC)** *Lunch provided for HAAC members and invited guests*	Quapaw Parlor
12:00 – 01:00	**Mentorship Networking Lunch** *Invited Mentors and Mentees*	Quapaw & Cherokee Foyer
01:00 – 03:00	**Advocate Work Group Meeting**	Ballroom
03:15 – 06:00	**Annual Conference Registration Open**	Outside Ballroom
03:30 – 07:30	**Heartland Advisory Board Meeting** *Dinner provided for Advisory Board members and invited guests*	Quapaw Parlor

~Dinner on your own~

THURSDAY, OCTOBER 13

TIME	EVENT	LOCATION
07:00 – 05:00	**Registration Open**	Outside Ballroom
07:00 – 08:00	**Breakfast**	Baxter Parlor
08:00 – 05:00	**Sponsors/Exhibitors Hall Open**	Cherokee & Quapaw
08:15 – 08:25	**Welcome Remarks** *Brad Schaefer, MD, FAAP, FACMG* *Heartland Co-Director* *and* *Lori Williamson Dean, MS, CGC* *Heartland Program Manager*	Ballroom
08:25 – 08:45	**Perspectives from the Health Resources and Services Administration (HRSA)** *Debi Sarkar, MPH* *Chief, Genetic Services Branch*	Ballroom
08:45 – 09:10	**National Coordinating Center (NCC) Report** *Alisha Keehn, MPA* *NCC Associate Program Director*	Ballroom

HEARTLAND GENETICS SERVICES COLLABORATIVE

AGENDA

THURSDAY, OCTOBER 13, CONT.

TIME	EVENT	LOCATION
09:10 – 09:30	**Perspectives from the National Center for Hearing Assessment and Management (NCHAM)** *Patti Martin, PhD* *Arkansas Children's Hospital*	Ballroom
09:30 – 09:50	**NewSTEPs Updates** *Sikha Singh, MHS, PMP* *NewSTEPs Manager*	Ballroom
09:50 – 10:00	**BREAK**	Cherokee & Quapaw Foyer
10:00 – 12:00	**Session A: Underserved Populations** *Moderated by Gene Hallford, PhD* *University of Oklahoma Health Sciences Center*	Ballroom
10:00 – 10:30	**Native Americans and Genomics: ELSI Project** *Paul Spicer, PhD* *University of Oklahoma*	Ballroom
10:30 – 11:00	**Pacific Islander Populations in the Midwest: Health and Culture** *Sheldon Riklon, MD* *University of Arkansas for Medical Sciences* *and* *Melisa Laelan* *University of Arkansas*	Ballroom
11:00 – 11:20	**Hispanic Access to Genetics Services: Recommendations from the Research** *Mary Ann Coffman, MS* *Heartland Hispanic Access Project Coordinator*	Ballroom
11:20 – 11:50	**Community Health Workers: Partners in the Care of Children** *Adriana Matiz, MD, FAAP* *Columbia University*	Ballroom
12:00 – 01:00	**LUNCH PROVIDED**	Baxter & Brooks Parlors
01:00 – 02:30	**Session B: Genetics across the Life Cycle** *Moderated by Kim Noble Piper, RN, BS, CPH,CPHG* *Iowa Department of Public Health*	Ballroom
01:00 – 02:00	**Newborn Screening (NBS) Timeliness** *Beth Tarini, MD, MS* *University of Iowa Hospitals and Clinics*	Ballroom

HEARTLAND GENETICS SERVICES COLLABORATIVE

AGENDA

THURSDAY, OCTOBER 13, CONT.

TIME	EVENT	LOCATION
01:00 – 02:00	**Newborn Screening (NBS) Timeliness, cont.** *Yvonne Kellar-Guenther, PhD* *Colorado School of Public Health* *and* *Stanton Berberich, PhD* *Iowa State Hygienic Laboratory*	Ballroom
02:00 – 02:25	**Long Term Follow Up: Longitudinal Pediatric Data Resource (LPDR)** *Sharmini Rogers, MBBS, MPH* *Missouri Department of Health and Senior Services*	Ballroom
02:25 – 02:45	**Beyond the Diagnosis: Providing Support to Children who are Deaf/Hard of Hearing and their Families** *Nannette Nicholson, PhD* *University of Arkansas for Medical Sciences* *and* *Patti Martin, PhD* *Arkansas Children's Hospital*	Ballroom
02:45 – 03:00	**BREAK**	Cherokee & Quapaw Foyer
03:00 – 03:25	**An Update on Current Practice and Challenges in Cancer Genetics** *Alex Obregon-Tito, PhD, MSc, MA, LCGC* *University of Arkansas for Medical Sciences*	Ballroom
03:25 – 03:50	**Public Health and Cancer Genetics** *Summer Cox, MPH* *Oregon Health Authority*	Ballroom
03:50 – 04:15	**Matched Tumor Normal Genetic Testing: The Role for Genetic Counselors** *Taylor Warner, MS* *University of Iowa Hospitals and Clinics*	Ballroom
04:15 – 04:45	**Direct to Consumer Testing** *Tiffany Lepard Tassin, MS, CGC* *University of Arkansas for Medical Sciences*	Ballroom
04:45 – 06:00	**HEARTLAND RECEPTION**	Baxter Parlor

~Dinner on your own~

HEARTLAND GENETICS SERVICES COLLABORATIVE

AGENDA

FRIDAY, OCTOBER 14

TIME	EVENT	LOCATION
07:30 – 08:30	**Breakfast**	Baxter Parlor
08:00 – 01:00	**Sponsors/Exhibitors Hall Open**	Cherokee & Quapaw
08:00 – 12:00	**Annual Conference Registration Open**	Outside Ballroom
08:30 – 10:30	**Session C: Models of Care** *Moderated by Shobana Kubendran, MS, CGC, MBBS* *University of Kansas Medical Center*	Ballroom
08:30 – 09:00	**Integrating Genomics into Primary Care** *Jen Leonhard, MS, LCGC* *Sanford Health System* *and* *Brad Schaefer, MD, FAAP, FACMG* *University of Arkansas for Medical Sciences*	Ballroom
09:00 – 09:10	**Heartland and Western States Telehealth Education Project** *Lori Williamson Dean, MS, CGC* *University of Arkansas for Medical Sciences*	Ballroom
09:10 – 09:30	**UAMS Sickle Cell Transition** *Leigh Ann Wilson, LCSW* *University of Arkansas for Medical Sciences*	Ballroom
09:30 – 09:50	**Improving Comprehensive Care and Time Efficiency at the Same Time – The HALI Project Parent Partner Model** *Brad Thompson, MA, LPC-S* *The HALI Project*	Ballroom
09:50 – 10:10	**Healthcare Transition for Patients with Genetic Conditions** *Wendy Parent-Johnson, PhD* *University of South Dakota*	Ballroom
10:10 – 10:30	**School Based Telehealth Centers** *Alan Faulkner* *University of Arkansas for Medical Sciences*	Ballroom
10:30 – 10:45	**BREAK: PLEASE CHECKOUT**	Ballroom
10:45 – 12:00	**Session D: Partnerships** *Moderated by Larry Weatherford, BA* *Heartland Advocate*	Ballroom
10:45 – 11:10	**Oklahoma Pediatric Research Network** *Ann Chou, PhD* *University of Oklahoma Health Sciences Center*	Ballroom

HEARTLAND GENETICS SERVICES COLLABORATIVE

AGENDA

FRIDAY, OCTOBER 14, CONT.

TIME	EVENT	LOCATION
11:10 – 12:00	**Cultivating Partnership through Heartland Activities: Collaborative Partners, Care Coordination, Mentoring, LEND, Title V, and GSA** *Barb Jackson, PhD* *Lori Williamson Dean, MS, CGC* *and* *Ann Chou, PhD*	Ballroom
12:00 – 01:00	**LUNCH BUFFET**	Baxter & Brooks Parlor
01:00 – 02:30	**Session E: Prioritization Exercise** *Facilitated by Barb Jackson, PhD*	Ballroom
02:30 – 02:40	**Evaluation and Reimbursement** *Anayeli Herrera Morales, MPH* *Heartland Assistant Program Manager*	Ballroom
02:40 – 02:50	**Closing Remarks** *Klaas Wierenga, MD* *Heartland Co-Director*	Ballroom
02:50	**Adjourn**	

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
G. Bradley Schaefer, MD, FAAP, FACMG
Professor of Genetics and Pediatrics
Founding Director, Division of Medical Genetics
University of Arkansas for Medical Sciences Committee for the Future Endowed Chair in Medical Genetics
Section Chief, Division of Genetics and Metabolism
Little Rock, AR
SchaeferGB@uams.edu

BIO: Dr. Schaefer moved from the University of Nebraska to the University of Arkansas for Medical Sciences in 2008. He is the Founding Director of the Division of Medical Genetics and the Chief of the Section of Genetics and Metabolism in the Department of Pediatrics. He holds the Committee-for-the-Future Endowed Chair in Medical Genetics.

He has authored 250 scientific articles, book chapters, and invited reviews. He is on the editorial board of the Journal of Child Neurology and the National Advisory Board for the Sotos Syndrome Support Association. His clinical practice focuses on the genetics of neurologic conditions, neurosensory abnormalities, and craniofacial malformations. His research is in clinical genetics with emphasis in neurogenetics and neurodevelopmental disabilities.

TOPIC: Integrating Genomics into Primary Care

NOTES:

NAME:
Lori Williamson Dean, MS, CGC
Assistant Professor and Program Director, Department of Genetic Counseling
University of Arkansas for Medical Sciences
Project Manager, Heartland Genetics Services Collaborative

BIO: Lori has worked as a prenatal and pediatric genetic counselor at Arkansas Children's Hospital ('92-'93); and at the University of Missouri Department of Pediatrics and the Southern Missouri Regional Genetics Program ('93 – '03). In 2004, she began teaching genetic counseling students at the University of Oklahoma Health Sciences Center and is now chairman of the Department of Genetic Counseling at the University of Arkansas for Medical Sciences.

Lori has served as Program Manager for the Heartland Genetics Collaborative since its inception in 2004. Increasing access to genetics services and genetics education are the themes of her career. Lori has spent her life and career in the Heartland region.

TOPICS: Heartland and Western States Telehealth Education Project
Cultivating Partnership through Heartland Activities: Collaborative Partners, Care Coordination, Mentoring, LEND, Title V, and GSA

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Debi Sarkar, MPH
Designated Federal Official, Secretary's Advisory Committee on Heritable Disorders in Newborns and Children

BIO: Debi Sarkar, MPH, is the Designated Federal Official (DFO) for the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children, a federal advisory committee created by Congress to advise the Secretary of Health and Human Services on the most appropriate application of universal newborn screening tests, technologies, policies, guidelines, and standards. As DFO, she oversees planning, policy, communications, operations, and logistical meeting activities to support the work of the committee. Ms. Sarkar has been at the Health Resources and Services Administration (HRSA) since 2006 and has worked as both a project officer and a team lead. Currently, she is the acting chief for the Genetic Services Branch in the Division of Children with Special Health Needs of the Maternal and Child Health Bureau (MCHB) at HRSA. Prior to joining MCHB, Ms. Sarkar was in HRSA's Office of Health Information Technology working on policy related to how technology can be used to efficiently collect data and how that data can be used effectively for quality improvement. Ms. Sarkar grew up in Alabama and received a Master of Public Health degree from the University of Alabama at Birmingham.

TOPIC: Perspectives from the Health Resources and Services Administration (HRSA)

NOTES:

NAME:
Alisha Keehn, MPA
Project Manager, NCC
American College of Medical Genetics and Genomics
akeehn@acmg.net

BIO: Ms. Keehn is the Associate Program Director of the National Coordinating Center for the Regional Genetic and Newborn Screening Service Collaboratives (NCC) housed at the American College of Medical Genetics and Genomics (ACMG). Ms. Keehn has been working with the NCC since joining ACMG in 2008. Prior to joining ACMG, Ms. Keehn was the Project Director of the Southeast Regional NBS and Genetics Collaborative. Ms. Keehn received her Master of Public Administration from the Georgia State University Andrew Young School of Policy Studies with a concentration in Policy Analysis and Evaluation.

TOPIC: National Coordinating Center (NCC) Report

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Patti Martin, PhD
Director of Audiology and Speech-Language Pathology, Arkansas Children's Hospital
Adjunct Assistant Professor of Audiology, University of Arkansas for Medical Sciences
martinpf@archildrens.org

BIO: Dr. Patti Martin is the Director of Audiology and Speech-Language Pathology at Arkansas Children's Hospital (ACH) and an Adjunct Assistant Professor of Audiology in the College of Health Professions at the University of Arkansas for Medical Sciences. Her areas of expertise include newborn screening, pediatric assessment, family support and program expansion. She was elected to two terms on the American Speech-Language-Hearing Association Special Interest Division 9/Childhood Hearing and Hearing Disorders steering committee and is currently the ASHA representative to the Joint Committee on Infant Hearing. In addition, she is a consultant for the Technical Assistance Network through the National Center on Hearing Assessment and Management. Dr. Martin provides oversight and direction to a staff of over 100 and is involved in Au.D. /ENT resident physician training and education through the University of Arkansas for Medical Sciences. Furthermore, she is involved in multiple grant-funded projects and research centering on improving access, independence and outcomes for children with hearing loss. Her doctoral program emphasis was in Family and Disability Policy, reflecting her passion about the many ways that families impact outcomes of children who are deaf/hard of hearing.

TOPIC: Perspectives from the National Center for Hearing Assessment and Management (NCHAM) Beyond the Diagnosis: Providing Support to Children who are Deaf/Hard of Hearing and their Families

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Sikha Singh, MHS, PMP
NewSTEPs Manager

BIO: Sikha Singh is the Manager for NewSTEPs at the Association of Public Health Laboratories (APHL). She joined APHL in 2009 and has significant experience in high-throughput genomic sequencing. Ms. Singh holds a Master of Health Science degree from the Johns Hopkins University and a Bachelor of Science degree from Temple University. She is a member of the Delta Omega Honorary Society as well as the recipient of a Project Management Professional (PMP) certificate.

TOPIC: NewSTEPs Updates

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Gene Hallford, PhD
Assistant Professor of Research
University of Oklahoma Health Sciences Center
gene@hallford.com

BIO: Dr. Hallford joined the OUHSC in 1989 and has been actively involved in research ever since. Over that time, he advanced from research assistant to faculty, moving from Psychiatry and Behavioral Sciences to Pediatrics, but has always been involved in what he most enjoys; research and education. As a faculty member at OU College of Medicine, he teaches Clinical Research Methods and Co-Directs the OCTSI's Summer Scholars' training program in Clinical and Translational Science. Dr. Hallford also provides educational seminars on clinical research and qualitative research methods and, through the Oklahoma Center for Neuroscience provide lectures on research-related health issues in the Oklahoma City area. Recently, he joined UAMS as Adjunct Faculty to provide research-related instruction and to help mentor MS students in research methodology. In 1997, he completed a Master's degree in Experimental Psychology, where he acquired advanced training in experimental design and quantitative data analysis and recently completed his PhD, which focused on quantitative methods and analysis as well as research in the areas of health and intercultural communication. Clearly, Dr. Hallford's academic studies follow his interests, in both programs, research and research methods courses were what he most enjoyed. This interest has continued in his unfolding academic career at the OUHSC. Currently, he serves on the OUHSC's Master of Science in Clinical and Translational Science, Executive Committee, the Oklahoma City Community College Associate in Applied Science, Clinical Research Coordinator program, Advisory Committee, and the OUHSC, Department of Psychiatry and Behavioral Sciences' Chemical Dependency Post-Doctoral fellowship Executive Committee. And as Navigator Program Director, he works both with trainees and new faculty to assist them with developing their research interests and with senior faculty and OCTSI Core Directors to promote the Research Navigator service and find mentors for Navigator clients. Together these two activities, research and education, are what encourage him to come to work each day.

TOPIC: Moderator, Session A: Underserved Populations

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Paul Spicer, PhD
Professor of Anthropology
University of Oklahoma
paul.spicer@ou.edu

BIO: Paul Spicer is Professor of Anthropology at the University of Oklahoma, where he teaches on native North America, religion, human development, and bioethics. He is an applied medical anthropologist who has been Principal Investigator on projects in substance abuse, child development, genetics, and cancer. This research has been funded by the Administration on Children and Families, numerous institutes at the National Institutes of Health, and the Robert Wood Johnson Foundation. His current research and scholarship focuses on ecology, ethics, and human development in American Indian and Alaska Native communities. Dr. Spicer is Principal Investigator of the Center on American Indian and Alaska Native Genomic Research, a National Human Genome Research Institute Center of Excellence in Ethical, Legal and Social Implications Research. He also serves as Editor in Chief of the Infant Mental Health Journal and Vice President of the Board of Zero To Three: The National Center for Infants, Toddlers, and Families.

TOPIC: Native Americans and Genomics: ELSI Project

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Sheldon Riklon, MD
Associate Professor in the Department of Family and Preventive Medicine
University of Arkansas for Medical Sciences

BIO: Sheldon Riklon, MD, is a Marshallese family physician, born and raised in the Marshall Islands. He is one of only two Marshallese in the world who has completed medical school and residency training at an accredited program in the United States. After high school graduation, he moved to the State of Hawaii where he received his B.A. degree in Biology from the University of Hawaii at Hilo in 1989. He completed the Imi Ho'ola Post Graduate Program at the University of Hawaii at Manoa and matriculated through the John A. Burns School of Medicine (JABSOM) where he received his MD in 1998. Upon completion of his family medicine residency training in 2001, he returned home to the Marshall Islands where, among other duties, he served as a primary care physician at Majuro Hospital, chaired the national medical referral committee, chaired Majuro Hospital's Pharmacy and Therapeutics committee, was a member of the institutional review committee, and directed the Special Medical Care Program that provided healthcare to the radiation affected population in the Marshall Islands. In 2009, he returned to Hawaii and was a faculty member with University of Hawaii's JABSOM's Department of Family Medicine and Community Health and served as the Family Medicine Clerkship Director. Dr. Riklon has been actively involved among the Marshallese and the other Micronesian populations in Hawaii as one of the founding members and recent past-chair of the Micronesian Health Advisory Coalition ("MHAC"). MHAC aims to educate Micronesians residing in the State of Hawai`i and the rest of the United States on health issues affecting them, to advocate for Micronesians on current health inequity issues and to collaborate with other Micronesian advocacy groups.

Dr. Riklon is currently an Associate Professor in the Department of Family and Preventive Medicine and a co-investigator in the Office of Community Health and Research at the University of Arkansas for Medical Sciences' Northwest Campus. He is the inaugural recipient of the Peter O. Kohler, M.D., Endowed Distinguished Professorship in Health Disparities at UAMS-NW Campus.
He serves as a primary care physician at Community Clinic in Springdale that serves a large patient population of Marshallese and underserved populations in NW Arkansas.

Dr. Riklon's research surrounds culturally competent health research, health care services, and palliative care for indigenous Pacific Islanders. His research has contributed to a deeper understanding about traditional Marshallese care practices and patient-centered care for Marshallese communities. Dr. Riklon has also worked toward creating health equity for Pacific Islanders living in the US. Ongoing research with UAMS will include increasing patient centered research with underrepresented Pacific Islander populations, a family model of diabetes self- management education to reduce disparities, and increasing the access the Marshallese community have to healthy food options and culturally competent health care services.

TOPIC: Pacific Islander Populations in the Midwest: Health and Culture

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Melisa Laelan
Founder/Director, Arkansas Coalition of Marshallese
Commissioner, Arkansas Minority Health Commission
mlaelan@email.uark.edu

BIO: Melisa Laelan is the nation's only certified court interpreter for Marshallese. She served 10 years in the U.S. Army and reached the rank of sergeant before she settled in Northwest Arkansas and helped found a nonprofit to support the Marshallese community. The nonprofit organization, Arkansas Coalition of Marshallese, helps military veterans seek citizenship, hosts a community garden to provide fresh produce to Marshallese churches, among other work that addresses community needs. Laelan also serves on the Arkansas Minority Health Commission. ("2014 Forty under 40" profiles. Northwest Arkansas Business Journal. Retrieved October 6, 2016, from http://www.nwabusinessjournal.com/best-in-class/profile/1188/melisa-laelan)

TOPIC: Pacific Islander Populations in the Midwest: Health and Culture

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Mary Ann Coffman MS, CGC
Project Coordinator
Heartland Genetic Services Collaborative
mcoffman5@cox.net

BIO: Mary Ann began her career as a pediatric genetic counselor in 1986 at the Children's Hospital of Oklahoma. She then worked in the division of the Maternal and Child Health Service at the Oklahoma State Department of Health (OSDH) in newborn screening and congenital disorders. From 1993 to 2000, she worked as a genetic service consultant for the Office of Women's and Children's Health, Arizona Department of Health Services and the OSDH.

Mary Ann has worked as a project coordinator since 2009 for the Heartland Genetics Services Collaborative. In the past, she has coordinated several projects including Transition, genetics nursing curriculum, and the Individualized Healthcare Plan (IHP). She is currently the project coordinator for the Hispanic Access Project.

TOPIC: Hispanic Access to Genetics Services: Recommendations from the Research

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Adriana Matiz, MD, FAAP
Associate Professor of Pediatrics
Columbia University
lam2048@columbia.edu

BIO: Dr Adriana Matiz is an Associate Professor of Pediatrics at Columbia University Medical Center. She is also the medical director of New York Presbyterian Hospital's Center for Community Health Navigation where she oversees the clinical integration and model development of the Patient Navigator and Community Health Worker programs across the healthcare system. Dr Matiz has served as the medical director of WIN for Asthma, a nationally recognized program to strengthen community-wide asthma management for children in N. Manhattan which later expanded the community health worker model to include adult patients with diabetes. She has created an asthma medical home model for children, pediatric care management model and is currently developing a model of care for children with special health care needs in primary care practices to improve care and reduce preventable healthcare utilization. Her passion is to decrease health disparities and to expand the delivery of culturally competent care.

Dr Matiz is a native of Colombia, raised in NYC and a graduate of NYU College of Arts and Science and the School of Medicine. She completed her pediatric training and subsequent Chief Residency at Montefiore Medical Center/Albert Einstein College of Medicine and resides with her family in the Bronx.

TOPIC: Community Health Workers: Partners in the Care of Children

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Kimberly Noble Piper, RN, BS, CPH, CPHG
Executive Officer, Center for Congenital and Inherited Disorders
State Genetics Coordinator
Iowa Department of Public Health
kimberly.piper@idph.iowa.gov

BIO: Kimberly Noble Piper is the state genetics coordinator for Iowa and the executive officer for the Center for Congenital and Inherited Disorders at the Iowa Department of Public Health (IDPH). She is certified in public health and public health genomics. Ms. Noble Piper started her nursing career as a maternity nurse and manager of maternity services in a large hospital in Des Moines. Ms. Noble Piper was the perinatal nurse consultant for the IDPH for six years prior to becoming the state genetics coordinator in 2004.

As the executive officer for the Center for Congenital and Inherited Disorders, Ms. Noble Piper is responsible for administrative oversight of the state's public health genomic programs. This includes newborn screening, maternal prenatal screening, regional genetics and neuromuscular clinics, Iowa Registry for Congenital and Inherited Disorders (Iowa's birth defects and stillbirth registry), family health history programming, and life course theory.

TOPIC: Moderator, Session B: Genetics Across the Life Cycle

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Beth Tarini, MD, MS
Fred G. Smith Chair in Academic Pediatrics
Division Director of Genetic Pediatrics and Adolescent Medicine
University of Iowa Hospitals and Clinics
beth-tarini@uiowa.edu

BIO: Beth Tarini is the Fred G. Smith Chair in Academic Pediatrics and Division Director of General Pediatrics & Adolescent Medicine. She is an associate professor in the Stead Family Department of Pediatrics. Her research focuses on optimizing the use of genetic services in pediatrics, specifically the organization and delivery of health care services through population-based screening programs such as newborn screening. She also conducts research on parental medical decision-making and parent-provider communication about genetic testing.

Dr. Tarini received a Bachelor of Arts in Biology from Harvard University and her medical degree from the Albert Einstein College of Medicine. She completed her pediatric residency training at the University of Washington and graduated from the Robert Wood Johnson Clinical Scholars Program at the University of Washington, where she received a Master of Science in Health Services.

TOPIC: Newborn Screening (NBS) Timeliness

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Yvonne Kellar-Guenther, PhD
Associate Professor
Colorado School of Public Health
yvonne.kellar-guenther@ucdenver.edu

BIO: Yvonne Kellar-Guenther is an Associate Professor at the Colorado School of Public Health (CSPH). She became involved in newborn screening when NewSTEPs (Newborn Screening Technical assistance and Evaluation Program) began. Dr. Kellar-Guenther is the Program Evaluator for NewSTEPs and the Associate Director for NewSTEPs 360, both HRSA funded projects. She also was the lead for the NewSTEPs Timeliness CoIIN (Collaborative Improvement and Innovation Network). In addition to her work on NewSTEPs, Dr. Kellar-Guenther is the program evaluator on several public health projects and teaches Program Evaluation at CSPH.

TOPIC: Newborn Screening Timeliness

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Sharmini V. Rogers, MBBS, MPH
Bureau Chief, Genetics and Healthy Children
Missouri Department of Health and Senior Services
sharmini.rogers@health.mo.gov

BIO: Sharmini Rogers received her medical degree from India and practiced medicine in Malaysia for seven years before she moved to the States where she received a Master's degree in Public Health from the University of Michigan, Ann Arbor. She has worked for the Missouri Department of Health and Senior Services since 1999. She is currently the Chief of the Bureau of Genetics and Healthy Childhood in the Missouri Department of Health and Senior Services. She has overall responsibility of the Missouri Newborn Screening programs, Genetic Programs such as Cystic Fibrosis, Hemophilia, Sickle Cell and Formula Program for individuals identified with a metabolic disorder. In addition, to the genetic programs, Dr. Rogers also has responsibility for federally funded home visiting programs, and other programs that promote healthy pregnancies, healthy babies and children. Dr. Rogers is a member of the Department's Institutional Review Board, Member of the Heartland Regional Genetics Collaborative Services Advisory Board, the Co-Chair of the Regional Collaborative Long Term Follow up Work group and a Steering Team member of NewSteps.

TOPIC: Long Term Follow Up: Longitudinal Pediatric Data Resources

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Nannette Nicholson, PhD
Director of Audiology
University of Arkansas at Little Rock
nn@uams.edu

BIO: Nannette Nicholson is a Professor and the Director of Audiology in the Department of Audiology and Speech Pathology at the University of Arkansas for Medical Sciences and the University of Arkansas at Little Rock. She serves on the state Infant Hearing Advisory Board and the Board of Directors for Arkansas Hands and Voices. Dr. Nicholson has a passion for pediatric audiology and topics related to early hearing detection and intervention (EHDI) services for families of children diagnosed with hearing loss. She and her colleagues have recently completed a systematic review on home visiting programs for families of children who are deaf or hard of hearing. Her most recent publication is on the combined use of a baby simulator and standardized parents to teach parent counseling techniques to students in the Doctor of Audiology (Au.D.) program.

TOPIC: Beyond the Diagnosis: Providing Support to Children who are Deaf/Hard of Hearing and their Families

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Alexandra J. Obregon-Tito, PhD, MSc, MA, LCGC
Licensed and Certified Genetic Counselor
Assistant Professor, College of Health Professions
Department of Genetic Counseling
University of Arkansas for Medical Sciences
AJObregontito@uams.edu

BIO: Alexandra Obregon-Tito is a certified genetic counselor dually trained in molecular genetics and social sciences, with expertise in genetics and genomics. Her training and experience includes molecular work in a wet lab as well as fieldwork in diverse settings, including work with indigenous groups in Latin America. She is interested in the dissemination and implementation of clinical genetics and genomics in underserved communities across the world.

TOPIC: An Update on Current Practice and Challenges in Cancer Genetics

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Summer Lee Cox, MPH
ScreenWise Genetics Coordinator
Oregon Health Authority
summer.l.cox@state.or.us

BIO: Summer Lee Cox, M.P.H., is the Coordinator of the ScreenWise Genetics Program, within the Public Health Division of the Oregon Health Authority. The ScreenWise Genetics Program works to improve the health and well-being of Oregonians through the up-to-date use of genomics and the public health practices of surveillance, education and policy/system change. Over the past six years, Ms. Cox has overseen two Centers for Disease Control and Prevention (CDC) Division of Cancer Prevention and Control cooperative agreements and a CDC Office of Public Health Genomics cooperative agreement with the Oregon Health Authority. As coordinator or the ScreenWise Genetics Program, Ms. Cox directs program activities that are largely related to hereditary cancer syndrome policy, education, and surveillance.

Ms. Cox earned a Master of Public Health, concentrating in public health policy and administration, from the College of Urban & Public Affairs: Hatfield School of Government at Portland State University. She also earned a B.A. in Biology, concentrating in genetics, from the Robert D. Clark Honors College at the University of Oregon. Previous to working in ScreenWise, Ms. Cox gained 12 years of research experience working at Oregon Health & Sciences University (OHSU), primarily in molecular medical genetics, non-human primate genomics, and colony genetics management. Ms. Cox has given over a dozen presentations at national events and has authored over ten peer-reviewed publications in the fields of molecular genetics and public health genomics.

TOPIC: Public Health and Cancer Genetics

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Taylor Warner, MS
Genetic Counselor
University of Iowa Hospitals and Clinics
taylor-c-warner@uiowa.edu

BIO: Taylor graduated from the University of Arkansas for Medical Sciences Genetic Counseling Graduate program in 2016. As part of her graduate program, she had the opportunity to train at hospitals in Little Rock, Kansas City, and Sioux Falls. In June 2016, Taylor began working at the University of Iowa Hospitals and Clinics as a pediatric genetic counselor. Taylor's clinic responsibilities include general genetics, connective tissue, and outreach clinics.

TOPIC: Matched Tumor Normal Genetic Testing: The Role for Genetic Counselors

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Tiffany Lepard Tassin, MS, CGC
Assistant Professor, Department of Genetic Counseling
University of Arkansas for Medical Sciences
tlepard@uams.edu

BIO: Tiffany is an Assistant Professor in the Department of Genetic Counseling at the University of Arkansas for Medical Sciences. In addition to teaching, she is the Genetic Counseling Discipline Coordinator for the Leadership Education in Neurodevelopmental Disabilities (LEND) program. She is a member of the Heartland Genetics Collaborative advisory board and is on the clinical services workgroup. Her area of expertise is in genetic counseling education, interdisciplinary training and service, and the utilization of social media for clinical research.

TOPIC: Direct to Consumer Testing

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Shobana Kubendran, MS, CGC, MBBS
Director of Genetics Division, Department of Pediatrics
University of Kansas Medical Center
shobana.kubendran@wesleymc.com

BIO: Shobana Kubendran is a board-certified genetic counselor and Director of genetics division at the Department of Pediatrics, Kansas University School of Medicine in Wichita, Kansas (KUSM-W). Her areas of practice are prenatal and pediatric genetic counseling. She developed a unique telegenetics service model that increases access to genetic services in Wichita and promotes integration of genetics into primary care. She enjoys teaching genetics in academic and clinical settings. Her passion is developing telegenetics services and increasing access to services in underserved regions.

TOPIC: Moderator, Session C: Models of Care

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Jennifer Leonhard, MS, LCGC
Genetic Counselor
Sanford Health
Jennifer.Leonhard@SanfordHealth.org

BIO: Jennifer Leonhard is an alumnus from the UAMS genetic counseling program. She completed her clinical training both in Little Rock, AR as well as in Sioux Falls, SD at Sanford Health. As the first genetic counselor at Sanford Health in Bemidji, MN, Jen serves all areas of medicine in both the hospital and clinics. Her position is part of the Sanford Imagenetics program, which intends to embed genetics practice and genomic medicine into primary care.

TOPIC: Integrating Genomics into Primary Care

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Leigh Ann Wilson, LCSW
University of Arkansas for Medical Sciences Cancer Institute, Adult Sickle Cell Program
lawilson2@uams.edu

BIO: Leigh Ann Wilson is a Licensed Certified Social Work in Arkansas. She received a Master's in Social Work from the University of Arkansas in Fayetteville. She has worked in outpatient dialysis, outpatient mental health, Geriatrics and is now at the UAMS Adult Sickle Cell Clinical Program where she facilitates the transition clinic.

TOPIC: UAMS Sickle Cell Transition

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Brad Thompson, MA, NCC, LPC-S
Director, The HALI Project
bthompson@southwest.org

BIO: Brad Thompson is the Director of The HALI Project; a non-profit organization that provides counseling and education services. That work includes working with families, the education system, and the medical system.

Brad has been married to Karen for 29 years and they have two adult children; Justin and Hali. Hali deals with the challenges of multiple special needs, both physical and cognitive, and is the inspiration for their work.

Brad has a Master's Degree in Counseling from West Texas A&M University and operates a private marriage & family therapy practice which supports his nonprofit habit. Brad and Karen have 27 years of experience working with families.

Brad has been blessed with many opportunities to serve in the primary care and special needs communities. Since 2008, he has worked with and in primary care practices to improve quality of care, provider efficiency, family health literacy, and patient/family satisfaction through the Hali Project's Parent Partner Program.

He served on the Medical Home Advisory Committee for the American Academy of Pediatrics from 2004-2010. Since 2011, he has served on an advisory committee for the Patient Centered Primary Care Collaborative. He currently serves on two committees for the Mountain States Regional Genetics Collaborative. He has recently been chosen to serve on the Transitions Advisory Committee for the Child Neurology Foundation. He also currently serves as a co-chair for the Patients, Families, and Consumers Leadership Center of the Patient Centered Primary Care Collaborative. Brad has been a keynote and break-out session speaker for numerous medical home and patient centered care conferences across the country.

While we would never have chosen for Hali the challenges that she faces every day, we feel incredibly blessed to share the lessons we have learned over 27 years with her to people across the country.

TOPIC: Improving Comprehensive Care and Time Efficiency at the Same Time - The HALI Project Parent Partner Model

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Wendy Parent-Johnson, PhD, CRC, CESP
Executive Director, Center for Disabilities
Professor, Department of Pediatrics
Editor, Journal of Rehabilitation
Sanford School of Medicine
University of South Dakota
wendy.parentjohnson@usd.edu

BIO: Dr. Wendy Parent-Johnson is a Professor with the Department of Pediatrics, and Executive Director of the Center for Disabilities, a Center for Excellence in Developmental Disabilities, at the Sanford School of Medicine, University of South Dakota (USD). She is responsible for administration and oversight of multiple grants, research, teaching, training, technical assistance, and supervision of approximately 35 staff. Dr. Parent-Johnson has more than thirty years of experience in the areas of supported and customized employment, transition from school to work, and Employment First policy and practice. More recently, her research and teaching has focused on health care transition, health disparities, interdisciplinary health science education, and the integration of health and employment. Dr. Parent-Johnson is the Editor of the Journal of Rehabilitation.

TOPIC: Healthcare Transition for Patients with Genetic Conditions

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Alan Faulker, BA
Program Manager, Center for Distance Health
University of Arkansas for Medical Sciences
akfaulkner@uams.edu

BIO: Alan has been employed at UAMS as a Program Manager in the Center for Distance Health since 2011. Alan worked on the deployment and development of the Arkansas e-Link Network and worked with individual sites on training and implementing new technology into its facilities.

Prior to joining the CDH team, Alan worked for in broadcast news in the Little Rock and Memphis television markets as an assistant news director. While working in news, Alan ran the day-to-day operations of the newsroom, worked on many investigative reports, breaking news and worked with new technologies and developed way of implementing them into broadcast news.

Alan graduated from Arkansas Tech University in 1994 with a BA in Liberal and Fine Arts

TOPIC: School Based Telehealth Center

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Larry Weatherford, BA
Heartland Advocate and Advisory Board member
larryweatherford@gmail.com

BIO: Larry Weatherford is an original member of both the Oklahoma Genetics Advisory Council and the Heartland Genetics Services Collaborative. Motivated by a desire to expand newborn screening in Oklahoma after his child was diagnosed with Cystic Fibrosis, he has been a parent advocate for the past 19 years. He helped found the Oklahoma City Chapter of the Cystic Fibrosis Foundation and was its first Executive Director.

Weatherford is the Veterans Business Development Officer and Public Affairs Specialist at the Oklahoma District Office of the Small Business Administration. In this role he works with aspiring small business owners, entrepreneurs and works to grow existing small businesses. In addition, Weatherford makes presentations and consults with a variety of organizations on crisis communications, media training, and social media using experience gleaned from his past work in the United States Air Force, during the Oklahoma City bombing, and as the lead public information officer for the Oklahoma State Department of Health on issues such as bioterrorism and pandemic influenza.

Since graduating from the University of Oklahoma with degrees in journalism and political science, Weatherford has been involved in public affairs in a number of different areas including active duty service in the United States Air Force as a public affairs specialist, numerous political campaigns including work on two presidential campaigns, and ownership of a public relations and advertising firm.

An Eagle Scout, he remains active in Boy Scouts and served for eight years as volunteer in Big Brothers/Big Sisters. He is a member of American Legion Post 88 and served as publicity chair for the Cleveland County Veterans Memorial. Weatherford is a member of First Presbyterian Church in Norman.

TOPIC: Moderator, Session D: Partnerships

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Ann F. Chou, PhD, MPH, MA
Associate Professor, Department of Family and Preventative Medicine
University of Oklahoma Health Sciences Center
ann-chou@ouhsc.edu

BIO: Ann has a background in organizational behavior, strategic management, and implementation science. Her research interests include provider-patient relationship, quality improvement and measurement, adoption and implementation of innovations and the delivery of best practices. Ann has been involved with the Heartland Collaborative, working on Genetic System Assessment project, for a number of years. The project has evolved from the development and uptake of a tool for state genetic programs to being adopted by a number of other regions as a tool to assess needs in certain areas of their respective programs.

TOPIC: Oklahoma Pediatric Research Network
Cultivating Partnerships through Heartland Activities: Collaborative Partners, Care Coordination, Mentoring, LEND, Title V, and GSA

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Barbara Jackson Ph. D.
Munroe-Meyer Institute,
University of Nebraska Medical Center
Omaha, NE
bjjackso@unmc.edu

BIO: Dr. Jackson is the Director of the Department of Education and Interdisciplinary Center of Program Evaluation (ICPE) and Professor at Munroe-Meyer Institute at the University of Nebraska Medical Center. Dr. Jackson is the program evaluator for multiple federal, regional and state funded projects, including such projects that focus on: national literacy and parent education, newborn screening, mental health consultation in child care settings, school based early childhood, and home visitation. In her role as an evaluation consultant she has experience with a variety of assessment measures that can effectively measure child, family, and early child teacher outcomes. The ICPE has established systematic quality assurance procedures and an effective process for continuous improvement as a key aspect of the evaluation process.

TOPIC: Cultivating Partnership through Heartland Activities: Collaborative Partners, Care Coordination, Mentoring, LEND, Title V, and GSA
Moderator, Session E: Prioritization Exercise

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Anayeli Herrera Morales, MPH
Assistant Program Manager
Heartland Genetics Services Collaborative
University of Arkansas for Medical Sciences
Anayeli@uams.edu

BIO: Anayeli received her undergraduate degree from The University of Oklahoma majoring in Spanish and minoring in Italian. She completed her Masters of Public Health with an emphasis in epidemiology at the University of Oklahoma Health Sciences Center in May 2013. After graduation, Anayeli worked as a project assistant/study coordinator at the Heart, Lung, and Vascular Clinic at OU Physicians and Heart Catheterization Laboratory at OU Medical Center.

Anayeli joined the Heartland Genetics Services Collaborative as the Program Coordinator in February 2014. She has worked with the Heartland Regional Coordinating Center leadership to plan, operate, and evaluate public health genetics projects that relate to access, education, resource management, and resource linkages for public health genetics stakeholders, primary care providers, and the public in the eight-state region. She also participates as both an interviewer and research team member for the Hispanic Access Project.

TOPIC: Evaluation and Reimbursement

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

NAME:
Klaas Wierenga, MD
Associate Professor of Pediatrics and Direct of Specialty Clinic
University of Oklahoma Health Sciences Center
Heartland Co-Director

BIO: Dr. Klaas Wierenga is an Associated Professor of Pediatrics and Director of Specialty Clinic at the University of Oklahoma Health Sciences Center (OUHSC) in Oklahoma City. He is a Board-Certified in clinical and medical biochemical genetics. Together with Dr. G. Bradley Schaefer, he is co-Director of the Heartland Regional Genetics Collaborative. Dr. Wierenga is director of the Medical Genetics Residency Program, and medical director of the Genetic Counseling Master's Program at OUHSC. His passion is providing medical care for individuals and families affected with heritable disorders. His research interest focuses on combining modern genetic tools with information technology in the diagnosis of rare genetic disorders.

TOPIC: Closing Remarks

NOTES:

HEARTLAND GENETICS SERVICES COLLABORATIVE

ADVISORY BOARD

DESCRIPTION

The Advisory Board is the region's advisory body to the Heartland Regional Coordinating Center and the collaborative as a whole. The Advisory Board meets monthly and makes decisions that require rapid response, approves documents and pilot project awards, advises on key issues and plans, and provides key leadership to address Heartland's priorities. The Advisory Board works by email outside of regularly scheduled monthly phone conferences.

In January 2011, the Advisory Board adopted bylaws to better define the role of the Board and its members.

MEMBERSHIP

Membership includes the eight State Genetic Coordinators, three Advocates, Newborn Screening representatives, Clinical Genetics representatives, and Primary Care Providers. The co-Program Directors, Program Manager, Program Coordinator, Program Evaluator, and Administrative Assistant serve as ex officio members.

The AB members currently meet on the second Wednesday of every month at 2–3 PM CT.

ADVOCATES

DESCRIPTION

Everything that the Heartland Genetics Services Collaborative does should ultimately improve the lives of those individuals and families impacted by genetic conditions. Consumers and advocates are involved in all of Heartland's work groups, projects and Advisory Board. Additionally, this stakeholder group convenes its own work group to address broad, psychosocial concerns affecting those with genetic conditions that Heartland may be able to impact.

MEMBERSHIP

As for all Heartland Work Groups, the Advocate Work Group is open to those interested in advocacy and leadership. The work group is comprised of parents of children (including adult children) with genetic / chromosomal conditions, individuals with a genetic condition, and representatives from advocacy organizations. Core members, however, agree to a two-year term that includes participation in monthly teleconferences, projects, e-mail communication, and occasional in-person meetings. Core members may also be asked to represent Heartland on national committees or work groups. Attendance at the annual in-person meeting will be funded by the Heartland Regional Coordinating Center for core members.

CO-CHAIRS: Michelle Black and Jeanne Egger

This Work Group currently meets on the third Friday of every month at 1:00 – 2:00 PM CT.

HEARTLAND GENETICS SERVICES COLLABORATIVE

AGENDA

ADVOCATE WORK GROUP MEETING
WEDNESDAY, OCTOBER 12, 2016
1:00 – 3:00 PM
ROOM: BALLROOM
CAPITAL HOTEL
LITTLE ROCK, AR

AGENDA ITEMS

TIME	EVENT	PRESENTER
12:00 – 01:00	**Mentorship Networking Lunch** Invited Mentors and Mentees Quapaw & Cherokee Foyer	
01:00 – 01:15	**Brief Introductions** *Name, State, Why are you attending the Heartland meeting*	All
01:15 – 01:30	**Mentorship Project Update**	Mark Smith
01:30 – 01:50	**Telehealth: The Families' Perspective**	Paula Connolly
01:50 – 02:00	**Break**	
02:00 – 02:20	**Telegenetics Services in the Heartland Region**	Lori Williamson Dean
02:20 – 02:40	**Project Carson** *Grantee: Family Voices of North Dakota*	Donene Feist
02:40 – 03:00	**Individualized Health Plan (IHP) Project** *Heartland Update*	Emily Meier
03:00	**Adjourn**	

NEXT MEETING: **Friday, November 18, 2016 at 1-2 PM CT**

HEARTLAND GENETICS SERVICES COLLABORATIVE

CLINICAL SERVICES

DESCRIPTION

Expanding access to genetics services and improving the quality of genetics services are integral to our mission. Members of the Heartland Clinical Services Work Group prioritize, promote, oversee and conduct activities that facilitate the medical home; improve health outcomes; expand genetics services; improve quality of services; and address gaps in services for individuals and families affected by genetic conditions across the life span.

MEMBERSHIP

As for all Heartland Work Groups, the Clinical Services Work Group is open to those interested in varying aspects of clinical genetics. The work group is comprised of advocates, medical geneticists, genetic counselors, genetic nurses, primary care providers, and public health workers. Core members, however, agree to a two-year term that includes participation in monthly teleconferences, projects, e-mail communication, and occasional in-person meetings. Core members may also be asked to represent Heartland on national committees or work groups. Attendance at the annual in-person meeting will be funded by the Heartland Regional Coordinating Center for core members.

CS members currently meet on the second Monday of every month at 3:00 – 4:00 PM CT.

HEARTLAND GENETICS SERVICES COLLABORATIVE

EHDI

DESCRIPTION

Enhancing the capacity and quality of all newborn screening programs, including newborn hearing screening, is integral to our mission. Members of the Heartland Early Hearing Detection and Intervention (EHDI) Work Group prioritize, promote, oversee and conduct activities that facilitate best practices in newborn hearing screening through resource and information sharing; continuing education; contributing to new knowledge discovery; and expanding NBS long-term follow up.

MEMBERSHIP

As for all Heartland Work Groups, the EHDI Work Group is open to those interested in varying aspects of newborn hearing screening (NBHS). The work group is comprised of EHDI coordinators, EHDI follow up staff, advocates, genetic counselors, and audiologists. Core members, however, agree to a two-year term that includes participation in monthly teleconferences, projects, e-mail communication, and occasional in-person meetings. Core members may also be asked to represent Heartland on national committees or work groups. Attendance at the annual in-person meeting will be funded by the Heartland Regional Coordinating Center for core members.

EHDI currently meets on the fourth Monday of every other month at 3:00 – 4:00 PM CT.

HEARTLAND GENETICS SERVICES COLLABORATIVE

NBS

DESCRIPTION

Enhancing the capacity and quality of newborn screening programs is integral to our mission. Members of the Heartland Newborn Screening Work Group prioritize, promote, oversee and conduct activities that facilitate best practices in newborn screening through resource and information sharing; continuing education; participation in national projects; contributing to new knowledge discovery; and expanding NBS long-term follow up.

MEMBERSHIP

As for all Heartland Work Groups, the Newborn Screening Work Group is open to those interested in varying aspects of newborn screening (NBS). The work group is comprised of NBS laboratorians, NBS follow up coordinators (both short term and long term), advocates, medical geneticists, genetic counselors, genetic nurses, and primary care providers. Core members, however, agree to a two-year term that includes participation in monthly teleconferences, projects, e-mail communication, and occasional in-person meetings. Core members may also be asked to represent Heartland on national committees or work groups. Attendance at the annual in-person meeting will be funded by the Heartland Regional Coordinating Center for core members.

NBS currently meets on the fourth Wednesday of every month at 2:00 – 3:00 PM CT.

BIBLIOGRAPHY (2006-2016)

PEER-REVIEWED JOURNAL ARTICLES

SCHAEFER GB, LARSON IA, BOLICK, JA, WILLIAMSON DEAN, L. What is the role of clinical genetics in the patient-centered medical home?: A commentary from the Medical Home Workgroup of the Heartland Regional Genetics and Newborn Screening Collaborative. Genetics in Medicine. 2015 vol 18:440-442.

DAACK-HIRSCH S, JACKSON B, BELCHEZ CA, ELDER B, HURLEY R, KERR P, NISSEN MK. Integrating genetics and genomics into nursing curricula: you can do it too!. Nurs Clin North Am. 2013 Dec; 48(4):661-9.

CARLETON SM, PECK DS, GRASELA J, DIETIKER KL, PHILLIPS CL. DNA carrier testing and newborn screening for maple syrup urine disease in Old Order Mennonite communities. Genet Test Mol Biomarkers. 2010 Apr;14(2):205-8.

CHOU AF, NORRIS AI, WILLIAMSON L, GARCIA K, BAYSINGER J, MULVIHILL JJ. Quality assurance in medical and public health genetics services: A systematic review. *Am J Med Genet.* 2009; Part C Semin Med Genet 151C:214–234.

FERGESON MA, MULVIHILL JJ, SCHAEFER GB, DEHAAI KA, PIATT J, COMBS K, BRIGHT BC, NEAS BR: Low adherence to national guidelines for thyroid screening in Down syndrome. *Genet Med.* 2009; 11(7):548-51.

HOLSEN LM, ZARCONE JR, CHAMBERS R, BUTLER MG, BITTEL DC, BROOKS WM, THOMPSON TI, SAVAGE CR: Genetic subtype differences in neural circuitry of food motivation in Prader-Willi syndrome. *Int J Obes.* 2008; 33: 273-283.

HOLSEN LM, ZARCONE JR, BROOKS WM, BUTLER MG, THOMPSON TI, AHLUWALIA JS, NOLLEN NG, SAVAGE CR: Neural mechanisms underlying hyperphagia in Prader-Willi syndrome. *Obesity.* 2006;14:1028-1037.

MULTIMEDIA

PARENT-JOHNSON W. Tools and strategies to facilitate health care transition. An invited webinar presentation for the Health Care Transition Research Consortium. 2016

PARENT-JOHNSON R. The Transitions Clinic. Center for Disabilities Outreach Newsletter and republished in Association of University Centers on Disability 260, Fall 2015. www.usd.edu/medicine/center-for-disabilities.

SCHAEFER GB, LARSON I, BOLICK J, AND WILLIAMSON DEAN L. Role of clinical genetics in the patient-centered medical home. http://www.heartlandcollaborative.org/work-groups/clinical-services/role-geneticist-medical-home/.

SCHAEFER, GB AND WILLIAMSON DEAN, L. Telemedicine in the Heartland. Webinar for the Mountain States Telemedicine Work Group. November 10, 2014.

PARENT-JOHNSON R. Optimizing health: bridging healthcare transitions for youth with special health care needs. Newsletter article for The Outreach Fall 2014 edition. Accessible at http://www.usd.edu/medicine/center-for-disabilities/the-outreach

BIBLIOGRAPHY (2006-2016)

COMEAU, MEG. The Affordable Care Act: Implications for Children with Genetic Disorders. Webinar. May 6, 2014. Accessible at https://www.youtube.com/watch?v=dnRiSypEqlA&feature=youtu.be

PARENT-JOHNSON R., PARENT-JOHNSON W. Social Capital: Making Transitions Possible. Webinar. January 10, 2014. Accessible at https://www.youtube.com/watch?v=9Br3uXL0g0o

PARENT-JOHNSON R., PARENT-JOHNSON W. Heartland Region Transition Projects: Overview and Findings. Webinar. November 25, 2013. Accessible at https://www.youtube.com/watch?v=G8Oa_36ImTg

SWEARINGEN C. Arkansas Newborn Screening Long term Follow up Database. Webinar. November 20, 2013. Accessible at https://www.youtube.com/watch?v=0ovRZ1mF-B4

PARENT-JOHNSON R., PARENT-JOHNSON W. Heartland Transition Learning Collaborative. Webinar. October 25, 2013. Accessible at https://www.youtube.com/watch?v=9Cl9HlMyJr8

WHITE M., BOWEN-EUBANKS M. HIPAA and FERPA Working Together While Keeping the Privacy of the Student in Mind. Webinar. August 22, 2013. Accessible at https://www.youtube.com/watch?v=nC7qO72o9Sw

BIGLER K. Comparing School-Based Plans for CYSHCN and/or Genetic Conditions. Webinar. July 9, 2013. Accessible at https://www.youtube.com/watch?v=oecIqrax1GU

KEMPER A. Weighing the Evidence for Newborn Screening. Webinar. April 24, 2013. Accessible https://www.youtube.com/watch?v=DQkCOz6B12I

KIMBERLING W., JOLMA C. Hearing Loss and Deafness. April 2013. Accessible at http://www.medicalhomeportal.org/

SMITH H., BAXTER M. IHP: What is your State Doing? Webinar. December 12, 2012. Accessible at www.heartlandcollaborative.org.

MARTIN G., BRADSHAW E. CCHD Screening: An Overview for Moving Forward. Webinar. May 14, 2012. Accessible at www.heartlandcollaborative.org.

WILLIAMSON L. Heartland Genetics and Newborn Screening Collaborative. Webinar Presentation for the Genetic Alliance Meet Your Neighbor Series; October 22, 2008.

SMITH L, WELSH H. Medium chain acyl-CoA dehydrogenase deficiency. Utah MedHome Portal: June 2008. http://www.medicalhomeportal.org/diagnoses-and-conditions/mcadd/description

BUTLER MG, WELCH J, ROPE A, BYERLY K. Prader-Willi syndrome. Utah MedHome Portal: March 2008; updated 2015. http://www.medicalhomeportal.org/diagnoses-and-conditions/prader-willi- syndrome/description

BIBLIOGRAPHY (2006-2016)

PRESENTATIONS

INVITED PLATFORM PRESENTATIONS

PARENT-JOHNSON W &PARENT-JOHNSON R. Transition from pediatric to adult medical care for youth with disabilities and/or chronic health conditions. Sanford School of Medicine Pediatric Grand Rounds. 2015; Sioux Falls, SD.

PARENT-JOHNSON W. What's health got to do with it? Integrating education and health care transitions to achieve mutual outcomes. Association of People Supporting Employment First National Conference, June 23, 2015; Philadelphia, PA.

KEEHN A, MAISE D, WILLIAMSON DEAN L, BROWER, A. Using MCC/RC Year-Two Evaluation Findings to Build Healthier Communities, Develop Quality Improvement, and Demonstrate the Value of the Regional Approach to Improving Access. AMCHP January 26, 2015; Washington, DC.

KUBENDRAN S, SARONWALA A, SCHAEFER. A novel approach in telegenetics services- geneticist, pediatrician and genetic counselor team. ACMG March 27, 2014; Nashville, TN.

PARENT-JOHNSON W., WILLIAMSON-DEAN L. Transitioning and Medical Home: Role of the Genetic Counselor. Pre-Conference Symposium, National Society of Genetic Counselors Annual Education Conference, October 2013; Anaheim, CA.

MROCH A. Sanford Children's Hearing Loss Clinic: Start-up, Successes, and Challenges. Early Hearing Detection and Intervention Annual Meeting; March 2012; St Louis, MO.

KUBENDRAN S. Telegenetics in the Heartland Regional Collaborative. American Telemedicine Association Conference; May 2011; Tampa, FL.

WILLIAMSON L. Access to Services is a Local Issue: NBS initiatives of the Regional Collaboratives. National Society of Genetic Counselors Annual Education Conference; October, 2010; Dallas, TX.

KUBENDRAN S. Telehealth: Heartland Regional Genetics & Newborn Screening Collaborative Telemedicine Services. Genetic Alliance Annual Conference; July, 2010; Bethesda, MD.

WILLIAMSON L. The Regional Newborn Screening and Genetics Services Collaboratives. Sarah Lawrence College Public Health Genomics Certificate Program; June 2010; Bronxville, NY.

WILLIAMSON L. The Genetic Counselor's Role in Newborn Screening. Sarah Lawrence College Public Health Genomics Certificate Program; June 2010; Bronxville, NY.

HOPKINS PV. The Heartland newborn screening back-up testing project: Quality assurance findings. Association of Public Health Laboratories Conference; November 2008; San Antonio, TX.

WILLIAMSON L. Role of genetic counselors in the newborn screening system: The Oklahoma experience: Association of Public Health Laboratories Conference; November 2008, San Antonio, TX.

BENDURE WB, SCHAFFER GB: Regional evaluation of telemedicine use and availability for clinical genetic service. American Telemedicine Association Conference; May 2007; Nashville, TN.

POSTER PRESENTATIONS

HERRERA MORALES A. *Collaborative partners project: a regional approach to dissemination of newborn screening information.* Association of Public Health Laboratories Genetic Testing and Newborn Screening Symposium; February 2016; St. Louis, MO.

HERRERA MORALES A & BOLICK JA. *Heartland Collaborative Partners Project: Dissemination of newborn screening information in Arkansas.* Arkansas American Public Health Association; May 2016; Hot Springs, AR.

BRUHA C, DEAN L, HALLFORD G, AND SCHAEFER GB. *The role of the geneticist in the patient-centered medical home.* American College of Medical Genetics; March 2015; Salt Lake City, UT.

MANN S, KEEHN A, DEAN L, ANDERSSON H, AND SCHAEFER B. *Increasing telegenetics providers in the United States.* American College of Medical Genetics; March 2015; Salt Lake City, UT.

PARENT-JOHNSON W AND PARENT-JOHNSON R. *Bridging systems: using social capital in health care transitions.* 15th Annual Chronic Illness and Disability Conference; MD Anderson Cancer Center; October 1, 2014; HOUSTON, TX.

BURT C AND SMITH H. *Heartland Individualized Healthcare Plan Learning Collaborative.* National Association of School Nurses' (NASN) 46th Annual Conference; June 28, 2014; San Antonio, TX.

PARENT-JOHNSON R. *Transitioning and Medical Home: The Role of the Geneticist.* Got Transition? and Health Care Transition: Research Symposium; October 2013; Houston, TX.

MULVIHILL J, HOPKINS P, BERBERICH S. *Interstate Compatibility and Emergency Preparedness of Newborn Screening Laboratories at the Interface of Clinical Medicine and Public Health.* American Society of Human Genetics Annual Meeting; November 2012; San Francisco, CA.

SCHULZE J, WILLIAMSON DEAN L. *Transition in Practice: A KAP Survey of Medical Genetics Health Professionals in the Heartland and Mountain States Regional Genetics and Newborn Screening Collaboratives*. American College of Medical Genetics Annual Meeting; March 2012; Charlotte, NC.

DAVIS, A, VAZ S. *Newborn Screening Education for Parents*. Association of Public Health Laboratories Newborn Screening and Genetic Testing Symposium. November 2011; San Diego, CA.

BIBLIOGRAPHY (2006-2016)

MULVIHILL J, WHITEHEAD S, CHOU A. Genetics Systems Assessment Working Group. *Weighted Metrics for Assessing Quality of Regional Public Health and Clinical Genetics Services.* 12th International Congress of Human Genetics/61st Annual Meeting of The American Society of Human Genetics; October, 2011; Montreal, Canada.

CREEL LM, KUBENDREN, S, LANDGREN, S, KAYE, CI, HOOKER, J, SCHAEFER, B. *Use of Telemedicine to Deliver Genetics Education in Two Regional Genetics Collaboratives*. National Coalition for Health Professional Education in Genetics, 14th Annual Conference; September 2011; Bethesda, MD.

CHOU AF, AU S, WILLIAMSON L, ET AL: *Developing a Comprehensive Quality Metrics Set for Genetics Service Delivery*: Academy Health; June 2011; Seattle, WA.

MCCALLISTER T, HIMSTEDT L, ROSS L, ET AL: *The Heartland good spot/bad spot project*: Association of Public Health Laboratories Newborn Screening and Genetic Testing Symposium; May 2010; Orlando, FL.

WILLIAMSON L, JACKSON B, MULVIHILL JJ, NBS Work Group of the Heartland Regional Genetics and NBS Collaborative**.** *A regional approach to enhancing quality and service capacity of NBS Programs***.** Association of Public Health Laboratories Newborn Screening and Genetic Testing Symposium; May 2010; Orlando, Fl.

ROGERS SV. *Knowledge and attitudes of the public on storage of dried blood spots*. Association of Public Health Laboratories Newborn Screening and Genetic Testing Symposium; May 2010; Orlando, FL.

ROGERS SV. Newborn screening – What providers and parents need to know. Association of Public Health Laboratories Newborn Screening and Genetic Testing Symposium; May 2010; Orlando, FL.

BERBERICH SL, VALBRACHT M, RAMIREZ M, ET AL. *Enlarging national newborn screening backup capacity by establishing a network of harmonized newborn screening programs*. Association of Public Health Laboratories Newborn Screening and Genetic Testing Symposium; May 2010; Orlando, FL.

NORRIS A, CHOU A, WILLIAMSON L, WHITEHEAD S, MULVIHILL JJ. *Metrics development for quality assessment of state genetics programs*. American College of Medical Genetics Annual Meeting; March 2010; Albuquerque, NM.

MILES J, NALE D, TROXELL R, ET AL. *Can teleHealth replace genetics outreach clinics in rural Missouri?* American Telemedicine Association Annual Conference; April 2009; Las Vegas, NV.

HATFIELD J, CHOU A, WILLIAMSON L, ET AL. *Developing of quality indicators to assess state genetics services and education: A Pilot Study*. American College of Medical Genetics Annual Meeting; March 2009; Tampa, FL.

CONOVER E, SCHAEFER GB. *Heartland Multi-State Teratogen Education Project*. American College of Medical Genetics Annual Meeting; March 2009; Tampa, FL.

PHILLIPS C, COPELAND S. *Genetic testing for maple syrup urine disease in Mennonite communities*. Association of Public Health Laboratories Newborn Screening and Genetic Testing Symposium; November 2008; San Antonio, TX.

BIBLIOGRAPHY (2006-2016)

KIM J, OLNEY R, YANG Q. *Trends in tuberous sclerosis deaths in the United States, 1983-1997*. American Public Health Association Annual Meeting; October 2008; San Diego, CA.

SAVAGE CR, HOLSEN L M, CHAMBERS RJ, ET AL. *Brain activation to food pictures in overweight and healthy weight children during high and low food motivation states*. The Obesity Society Annual Scientific Meeting; October 2007; New Orleans, LA.

SAVAGE CR, HOLSEN LM, CHAMBERS RJ, BROOKS WM, BUTLER MG, & ZARCONE JR. *Brain activation to food pictures in obese and healthy weight adults during high and low food motivation states*. The Obesity Society Annual Scientific Meeting Annual Scientific Meeting; October 2006; Boston, MA.

Heartland Goals
1. Continue to ensure that individuals with genetic disorders and their families have access to quality care and appropriate genetic expertise and other subspecialty expertise;
2. Apply the translation of genome-based knowledge, genomics best practices, and new technologies to education and training, services, and dissemination to improve population health; and
3. Quantitatively and qualitatively evaluate through program evaluation outcomes of projects undertaken to accomplish the goals.

HRSA PRIORITY AREA	HEARTLAND OBJECTIVES	OVERALL STRATEGIES	STATUS	YEAR 5 STRATEGIES	PROPOSED REVISION	RESPONSIBLE PARTY for PROJECT COMPLETION
Treat in the context of a medical home that provides accessible, family-centered continuous, comprehensive, coordinated, compassionate, and culturally effective care.	Obj. 1: Increase the number and diversity of the referral indications for telemedicine patient visits in the Heartland.	1: Prioritize pilot project funding for telemedicine clinic expansion.	Baseline data collection to be completed before May 31, 2016	Ongoing	None	HRCC Team; Clinical Services Work Group; Advisory Board
		2: Provide technical assistance to new users of telemedicine for the delivery of public health or clinical genetics services.		Ongoing	None	HRCC: Brad Schaefer
		3: Educate genetics residents, genetic counseling students, and those in practice less than five years in partnership with Western States region.	Cohort #3 being recruited. Training scheduled for August 1, 2016.	Continue the project; Revise, as needed.	None	HRCC: Lori Williamson Dean
	Obj. 2: Use the Individualized Health Plan to enhance coordinated, continuous and family-centered care for children with genetic conditions in the school setting.	Regional implementation of the model for IHP Offer care coordination training and materials in Spanish	Training for participating school nurses July/Aug 2016 Care Coordination trainings in Spanish	Continue the project; pilot implementation of new strategies; parent-professional partnership workshop in Spanish	None	Consultants: Wendy Parent-Johnson, Molly Satter HRCC: Lori Dean; Anayeli Herrera Morales; Advocate WG
	Obj. 3: Increase Heartland pediatric genetics providers' knowledge of and practice behaviors in healthcare transition for youth with genetic conditions.	Education, clinical practice	Pilot completed Reformatting for dissemination to other residents and learners Transition clinic data collection continues	Regional implementation of module Interviews with adult providers	None	Consultant: Wendy Parent-Johnson, Richard Parent-Johnson HRCC: Lori Williamson Dean;

HRSA PRIORITY AREA	HEARTLAND OBJECTIVES	OVERALL STRATEGIES	STATUS	YEAR 5 STRATEGIES	PROPOSED REVISION	RESPONSIBLE PARTY for PROJECT COMPLETION
						Clinical Services Work Group
	Obj 4: Increase access to genetics services for patients from Hispanic communities in the Heartland region.	Conduct interviews to learn about barriers, facilitators, and patient experiences.	Analysis of interview transcripts will be complete by 5-31-16 HA Advisory Committee to meet; review and prioritize recommendations	Branching research projects HA Adv Cmte guidance on activities	None	HRCC: Klaas Wierenga, Mary Ann Coffman, Gene Hallford, Alexandra Obregon Tito, and Anayeli Herrera Morales; Clinical Services Work Group; HA Advisory Group
	Obj 5: Explore role of geneticist in the medical home	Write a white paper delineating the issues	Commentary paper published in GIM Interviews with parents and providers to be completed by end of project year	Data analysis and recommendations for next steps	None	HRCC: Brad Schaefer, Gene Hallford; Lori Dean; Clinical Services Work Group
Expand the pool of the genetic service workforce by determining needs and gaps across sectors to provide education and training, with emphasis on allied health providers, other subspecialties and educators.	Obj. 1: Increase primary care providers' access to tools and resources for the initial evaluation, follow-up and monitoring of patients with genetic disorders	Strategy 1: Disseminate results, tools, and resources of the pilot project (A Novel Approach in Telegenetic Services: Pediatrician and Genetic Counselor Team) currently underway to train a community pediatrician in the preliminary evaluation of the genetics patient through a presentation at the 2012 Heartland annual meeting, to Heartland states' American Academy of Pediatrics (AAP) chapters, to the MedHome Portal (Mountain States region), and on the Heartland website.	Manuscript in progress	None	None	HRCC: Brad Schaefer
		Strategy 2:		No action planned		
		Strategy 3: Complete the "Heartland Collaborative	Third cohort completed Results of second	None	None	HRCC: Anayeli Herrera Morales;

HRSA PRIORITY AREA	HEARTLAND OBJECTIVES	OVERALL STRATEGIES	STATUS	YEAR 5 STRATEGIES	PROPOSED REVISION	RESPONSIBLE PARTY for PROJECT COMPLETION
		Partners Program"	cohort presented at APHL NBSGT 2016			NBS Work Group
		Strategy 4: Family Health History implementation of NCHPEG tool into Heartland clinical practices		No action planned		
	Obj 2: Enhance the Heartland's Early Hearing Detection and Intervention (EHDI) programs through resource sharing and education.	Strategy 1: Complete the EHDI exchange program.	Completed	Complete post-evaluation	None	HRCC: Anayeli Herrera Morales; EHDI Work Group
		Strategy 2: Support genetics education of EHDI coordinators, for audiologists, educators, speech/language pathologists, otolaryngologists, parents, patients, and advocates in the region.	Schedule webinars re: genetics evaluation for hearing loss and resources/genetic counseling	Schedule webinars, if the group wants to do that	None	HRCC: Anayeli Herrera Morales; EHDI Work Group
Build capacity in state public health departments to enhance and sustain the delivery of newborn and child screening and genetic follow-up and treatment services.	Obj 1: Heartland will initiate an information system to facilitate Heartland states' adoption of ACHDNC recommendations related to newborn and child screening and genetic follow-up and treatment services.	Strategy 1: Develop an implementation toolkit for Heartland states	Remove from website	None		
		Strategy 2: Monitor local, regional and national activities related to new conditions and ACHDNC recommendations to identify opportunities for Heartland states to participate in pilots, clinical practice guidelines, short and long-term follow-up of newborn screen identified patients. Once a ACHDNC recommendation is published, begin to work with clinicians to identify newborns who are clinically diagnosed prior to adoption of newborn screening to provide a cohort for comparison with newborn screen identified cases.	Ongoing	Participation in national efforts	None	HRCC; State Genetics Coordinators; NBS Program Coordinators

HRSA PRIORITY AREA	HEARTLAND OBJECTIVES	OVERALL STRATEGIES	STATUS	YEAR 5 STRATEGIES	PROPOSED REVISION	RESPONSIBLE PARTY for PROJECT COMPLETION
Expand state and regional collaborative systems of cohorts of patients for long-term monitoring and analysis of follow-up and treatment for provider and/or patient access.	Obj. 1: Improve state systems to reflect best practices in LTFU by increasing the specificity of newborn screening, establishing an approach to the long-term follow-up of patients, incorporating electronic health information, and expanding the number of conditions for which screening tests are available.	Strategy 1: Conduct regional NBS workshops for key stakeholders (public health, clinical services, patients and advocates).	Workshop planned for 2016	None		HRCC: Anayeli Herrera Morales and NBS Work Group
		Strategy 2: Facilitate states' adoption of case definitions for the recommended uniform screening panel.	Ongoing	Use time during NBS and Advisory Board monthly meetings		HRCC Team; NBS Work Group; Advisory Board
		Strategy 3: Link Heartland clinicians to clinical, LTFU databases, such as IBEM-IS	Support Iowa and Nebraska; South Dakota, University of Missouri, and Children's Mercy in Kansas City added	Ongoing		HRCC Team; NBS Work Group
Any other program priority that addresses the needs of the region and the program goals.	Obj. 1: Expand the implementation of the Genetic Systems Assessment	Strategy 1: Revise GSA tool based on Heartland implementation experience in 2012.	Complete	Develop version 3.0		Consultants: Ann Chou HRCC: Klaas Wierenga and Lori Williamson Dean
		Strategy 2: Sustain GSA in the Heartland	Complete	None		
		Strategy 3: Plan and execute national rollout of the GSA tool.	Implemented in 6 of 7 RCs	Invite NEGC to participate in version 3.0		Consultant: Ann Chou; HRCC Team
	Obj. 2: Pilot Project Program	Tool for soliciting novel strategies to address specific problems	5 projects funded for March - May	As needed		
	Obj. 3: Sustain Emergency Preparedness in the Heartland region (*HRSA priority 9*)	Strategy 1: The Heartland Region has demonstrated the ability to back up other states during events that would prevent a NBS laboratory from screening. During the next five years, it will be important to maintain this preparedness through drills		None		Consultants: Stan Berberich and Patrick Hopkins HRCC: Lori Williamson Dean

HRSA PRIORITY AREA	HEARTLAND OBJECTIVES	OVERALL STRATEGIES	STATUS	YEAR 5 STRATEGIES	PROPOSED REVISION	RESPONSIBLE PARTY for PROJECT COMPLETION
		conducted every other year (2014 and 2016). Repeating the drills will assure readiness and capacity as well as further harmonize systems and processes within the states.				
	Obj 4: Facilitate communication and linkages of Heartland providers through a variety of methods.	Strategy 1: Heartland Annual Meeting	Workshops held, as needed, in spring 2015 due to annual meeting not being held	Fall 2016		HRCC Team; Advisory Board
		Strategy 2: The Heartland website	Updated website and Facebook presence added	Ongoing		HRCC: Anayeli Herrera Morales and Illuminage (CareSource)
		Strategy 3: Participate in key national or other regional work groups or meetings that keep the Heartland linked to and represented.	Ongoing	Ongoing		HRCC Team and Heartland liaisons
	Obj. 5: Education and facilitate implementation of the Affordable Care Act	Strategy 1: Convene stakeholders	Survey completed; data analysis to commence; print policy briefs	Disseminate policy briefs		HRCC: Anayeli Herrera Morales

CONFERENCE ATTENDEES

NAME	ORGANIZATION	E-MAIL
MIKE **ABRAHAM**	Horizon Pharma	mabraham@horizonpharma.com
JANET **ALBRECHT**	PerkinElmer	janet.albrecht@perkinelmer.com
ANDREA **ATHERTON**	Shire Pharmaceuticals	aatherton0@shire.com
JORDAN **BARNES**	University of Arkansas for Medical Sciences	hbarnes@uams.edu
KRYSTAL **BAUMERT**	Nebraska Newborn Screening Program	Krystal.Baumert@nebraska.gov
ANDREW **BELL**	University of Arkansas for Medical Sciences	abell3@uams.edu
CAROLINA **BELTRAN-CARRANZA**	Kansas University School of Medicine-Wichita	caritobc@yahoo.com
KATIE **BENTZ**	North Dakota Department of Health	kbentz@nd.gov
STANTON **BERBERICH**	State Hygienic Laboratory at The University of Iowa	stanton-berberich@uiowa.edu
MICHELLE **BLACK**	Kansas Department of Health & Environment	mcblack@kdheks.gov
JO ANN **BOLICK**	University of Arkansas for Medical Sciences	JABolick@uams.edu
NATASHA **BONHOMME**	Genetic Alliance	nbonhomme@geneticalliance.org
TANJA **BROWN**	Arkansas Department Of Health	tanja.brown@arkansas.gov
VELVET **BROWN-WATTS**	Supporters of Families with Sickle Cell Disease, Inc	swithsicklecell@att.net
JONI **BRUCE**	Oklahoma Family Network	Jonib@ofn.mobi
AMBER **BRYANT**	Bureau of Genetics and Healthy Childhood	amber.bryant@health.mo.gov
JENA **CALLEN-SCHOLZ**	Shire Pharmaceuticals Rare Diseases	jcallenscholz0@shire.com
JENNIFER **CAMPBELL**	Oklahoma Newborn Screening Program	jennifer-d-campbell@ouhsc.edu
SAMANTHA **CASSISI**	University of Arkansas for Medical Sciences	scassisi@uams.edu
LISA **CATON**	Oklahoma State Department of Health	lisarc@health.ok.gov
ANN **CHOU**	University of Oklahoma Health Sciences Ctr	ann-chou@ouhsc.edu
MARY ANN **COFFMAN**	Heartland Genetics Services Collaborative	mcoffman5@cox.net
TIA **CORLEY**	Advocate Mentee	tiasephora@gmail.com
SUMMER **COX**	Oregon Health Authority, Public Health Division, Center for Protection & Health Promotion, ScreenWise Program	summer.l.cox@state.or.us
LAUREN **CSONKA**	University of Arkansas for Medical Sciences	lcsonka@uams.edu
KATIE **EASTON**	Ambry Genetics	keaston@ambrygen.com
JEANNE **EGGER**	Advocate	jeanne.egger@nebraska.gov
JARED **EIFERT**	Ambry Genetics	jeifert@ambrygen.com
DARLA **EIKEN**	Missouri State Laboratory - Newborn Screening Lab	darla.eiken@health.mo.gov
KAREN **EVEANS**	Nebraska Newborn Screening Program	karen.eveans@nebraska.gov
RODNEY **FARLEY**	Arkansas Department of Human Services MCHB Title V CSHCN	Rodney.farley@dhs.arkansas.gov

CONFERENCE ATTENDEES

NAME	ORGANIZATION	E-MAIL
ALAN **FAULKNER**	University of Arkansas for Medical Sciences	akfaulkner@uams.edu
DONENE **FEIST**	Family Voices of North Dakota	fvnd@drtel.net
JAMIE **FISHER**	University of Arkansas for Medical Sciences	jfisher5@uams.edu
LUCY **FOSSEN**	South Dakota Department of Health, Office of Child and Family Services	lucy.fossen@state.sd.us
MILAGROS **GARCIA**	Boys Town	mirakuru.garcia@gmail.com
ERICA **GARDNER**	University of Arkansas for Medical Sciences	egardner@uams.edu
SHAWN **GESSAY**	University of Arkansas for Medical Sciences	sgessay@uams.edu
ANNELISE **GILE**	Kansas Department of Health & Environment	agile@kdheks.gov
SARAH **GROENEVELD**	South Dakota Newborn Screening Program	Sarah.Groeneveld@SanfordHealth.org
CLAIRE **HACKING**	University of Arkansas for Medical Sciences	chacking@uams.edu
SUE **HADDAD**	Oklahoma State Dept. of Health NBS LAB	soheilah@health.ok.gov
DANA **HAGEMAN**	Twist of Fate - ATS	chdheartmom@gmail.com
GENE **HALLFORD**	University of Oklahoma Health Sciences Center	gene@hallford.com
BRITTANY **HENSON**	University of Arkansas for Medical Sciences	bhenson2@uams.edu
ERICA **HERRERA**	OKLAHOMA FAMILY NETWORK	Erica-Herrera@ouhsc.edu
ANAYELI **HERRERA MORALES**	Heartland Genetics Services Collaborative	anayeli@uams.edu
BERNARD **HERRMAN**	Ambry Genetics	bherrman@ambrygen.com
NICHOLE **HETZEL**	GeneDx	nhetzel@genedx.com
LESLIE **HIMSTEDT**	Arkansas Department of Health	Leslie.Himstedt@arkansas.gov
JEFF **HOFFMAN**	ECHO, NCHAM, Utah State University	jeffhoffman.ehdi@gmail.com
BARBARA **HOLCOMBE**	University of Arkansas for Medical Sciences Genetics Department, Newborn Screening Coordinator	blholcombe@uams.edu
HSIN-WEI **HUANG**	University of Arkansas for Medical Sciences	hhuang2@uams.edu
BARBARA **JACKSON**	Munroe-Meyer Institute	bjjackso@unmc.edu
TAMMIE **JOHNSON**	North Dakota Children's Special Health Services	tamjohnson@nd.gov
HOLLY **JOHNSON**	University of Arkansas for Medical Sciences	hajohnson2@uams.edu
ALISHA **KEEHN**	National Coordinating Center - ACMG	akeehn@acmg.net
YVONNE **KELLAR-GUENTHER**	NewSTEPs & CO School of Public Health	yvonne.kellar-guenther@ucdenver.edu
JAMEY **KENDALL**	Kansas Department of Health & Environment	jkendall@kdheks.gov
DANA **KENNICUTT**	Horizon Pharma	dkennicutt@horizonpharma.com
DAVID **KING**	Memorial Hospital of Texas County, Guymon, Oklahoma	kingdmk@aol.com
TRACY **KLUG**	Missouri State Laboratory - Newborn Screening	tracy.klug@health.mo.gov

CONFERENCE ATTENDEES

NAME	ORGANIZATION	E-MAIL
CAROLINE **KOCH**	University of Arkansas for Medical Sciences	ckoch@uams.edu
SHOBANA **KUBENDRAN**	Kansas University Medical Center-Wichita	shobana.kubendran@wesleymc.com
JENNIFER **LEONHARD**	Sanford Health	jennifer.leonhard@sanfordhealth.org
TIFFANY **LEPARD TASSIN**	University of Arkansas for Medical Sciences	tlepard@uams.edu
MEREDITH **LEVISEE**	University of Arkansas for Medical Sciences	mlevisee@uams.edu
EDWARD **LIETZ**	Advocate	funsized09@gmail.com
SHANNON **LOYD**	Arkansas EHDI Program	shannonloyd@arkansas.gov
JULIE **LUEDTKE**	Nebraska Department of Health and Human Services	julie.luedtke@nebraska.gov
SHALOME **LYNCH**	Iowa Department of Public Health	shalome.lynch@idph.iowa.gov
PATTI **MARTIN**	National Center for Hearing Assessment And Management	martinpf@archildrens.org
ADRIANA **MATIZ**	Columbia University	lam2048@columbia.edu
GLENDA **MATTHEWS**	Arkansas Newborn Screening Program	glenda.matthews@arkansas.gov
TIA **MCDONALD**	University of Arkansas for Medical Sciences	tmcdonald2@uams.edu
EMILY **MEIER**	University of South Dakota Center for Disabilities	emily.a.meier@usd.edu
CLAIRE **MENA**	Genetic Alliance	cmena@geneticalliance.org
ROBERT **MENENDEZ**	Fulgent Diagnostics	roramen1974@gmail.com
LINDSEY **METTE**	University of Texas Health Sciences Center at San Antonio	mette@uthscsa.edu
JOYAL **MEYER**	North Dakota Department of Health	jbmeyer@nd.gov
TIFFANY **MOORE**	University of Arkansas for Medical Sciences	tpmoore@uams.edu
RUBY **MOORE**	Arkansas Department of Health	ruby.moore@arkansas.gov
FRANCES **MORGAN**	Kansas Department of Health and Environment Laboratories	fmorgan@kdheks.gov
CHAD **MORRIS**	Recordati Rare Diseases	morris.c@recordati.com
NANNETTE **NICHOLSON**	University of Arkansas for Medical Sciences	nn@uams.edu
KIMBERLY **NOBLE PIPER**	Iowa Department of Public Health	kimberly.piper@idph.iowa.gov
ALEXANDRA **OBREGON-TITO**	University of Arkansas for Medical Sciences	ajobregontito@uams.edu
MIKE **O'SHEA**	PerkinElmer	mike.oshea@perkinelmer.com
WENDY **PARENT-JOHNSON**	University of South Dakota	wendy.parentjohnson@usd.edu
CATHERINE **POTENTE**	Ambry Genetics	cpotente@ambrygen.com
PAT **PURIFOY**	Arkansas Department of Health/Newborn Screening	patricia.purifoy@arkansas.gov
JULIE **RABURN-MILLER**	Missouri Department of Health and Senior Services	julie.raburn-miller@health.mo.gov
DONNA **RICHARDSON**	University of Arkansas for Medical Sciences-Adult Sickle Cell Clinical Program	djrichardson@uams.edu

CONFERENCE ATTENDEES

NAME	ORGANIZATION	E-MAIL
SHELDON **RIKLON**	University of Arkansas for Medical Sciences-Northwest Campus	sriklon@uams.edu
LORA **ROBERTS**	Oklahoma Family Network	lorar@ofn.mobi
JENNIFER **ROBERTS**	Children's Mercy Hospital	jlroberts2@cmh.edu
SHARMINI **ROGERS**	Department of Health and Senior Services	sharmini.rogers@health.mo.gov
WILLIAM **ROWE**	Shire Pharmaceuticals	wrowe@shire.com
MARGARITA **SAENZ**	University of Colorado	margarita.saenz@childrenscolorado.org
BRAD **SCHAEFER**	University of Arkansas for Medical Sciences	SchaeferGB@uams.edu
JOSEPH **SCHNEIDER**	Shire Pharmaceuticals	Joschneider@shire.com
GRACIELA **SHARIF**	PTI Nebraska	gsharif@pti-nebraska.org
SARAH **SHILLCUTT**	Advocate	sarahshillcutt@gmail.com
SIKHA **SINGH**	APHL/NewSTEPs	sikha.singh@aphl.org
MARK **SMITH**	Munroe-Meyer Institute	msmitha@unmc.edu
HOWARD **SMITH**	St. Louis Area First Steps	hsmith@benchmarkhs.com
PAUL **SPICER**	University of Oklahoma	paul.spicer@ou.edu
LOIS **STARR**	University of Nebraska Medical Center	lstarr@unmc.edu
RYAN **STITES**	GeneDx	rstites@genedx.com
GREG **STUDDARD**	Baebies, Inc.	GStuddard@Baebies.com
BETH **TARINI**	University of Iowa	beth-tarini@uiowa.edu
TAWNYA **TAYLOR**	Mentee	Funsized09@gmail.com
ANDREA **TAYLOR**	A Twist of Fate-ATS	Andrea.taylor01@yahoo.com
BRAD **THOMPSON**	The HALI Project	bthompson@southwest.org
ROBIN **TROXELL**	Mercy Health - Springfield	robin.troxell@mercy.net
VALERIE **VAN ZEE**	State Hygienic Laboratory at the University of Iowa	valerie-vanzee@uiowa.edu
BRYAN **VOSS**	Recordati Rare Diseases	voss.b@recordati.com
JOHN **WALL**	Shire Pharmaceuticals	jwall0@shire.com
TAYLOR **WARNER**	University of Iowa Hospitals and Clinics	taylor-c-warner@uiowa.edu
LARRY **WEATHERFORD**	Advocate	larryweatherford@gmail.com
BRENDA **WEBB**	Child & Adol Newborn Screening	brenda.webb@arkansas.gov
BETHANY **WETHINGTON**	Mentee	bethanyann13@hotmail.com
MERYL **WETTON**	University of Arkansas for Medical Sciences	mwetton@uams.edu
KLAAS **WIERENGA**	The University of Oklahoma Health Sciences Center	Klaas-Wierenga@ouhsc.edu
LORI **WILLIAMSON DEAN**	University of Arkansas for Medical Sciences	lwdean@uams.edu
LEIGH ANN **WILSON**	University of Arkansas for Medical Sciences	LAWilson2@uams.edu
BREANNE **WITHERSPOON**	University of Arkansas for Medical Sciences	bmwitherspoon@uams.edu

HEARTLAND GENETICS SERVICES COLLABORATIVE

NETWORKING

SUBMITTED BY: _____

Let's mingle and get to know each other a little bit more with a chance to win a few prizes. The winners will be those with the most entries under the "who am I?" column. The prizes are two $50 gift cards and t-shirts donated by the UAMS Sickle Cell Program. Please tear the following pages from your program book, give them to Ana, or leave them at the registration table. Have fun getting to know each other!

FUN FACT:	WHO AM I?
Awesomeness in the flesh!	*Anayeli Herrera Morales*
Advancing Newborn Screening and Pediatric testing solutions worldwide.	
AuD student! Want to learn more things.	
Before working in health care, I worked in journalism covering mostly breaking news and crime.	
But mom, I do live in a barn.	
Coaching Little League Wrestling has been a highlight for me giving back to the youth of Oklahoma.	
Colorado native.	
Coordinated the delivery of the world's first surviving septuplets.	
Cover North Texas, Oklahoma, and Arkansas for Shire Genetics Franchise.	
Every time I go on vacation, I try to do something I have never done before.	
Favorite hobbies are rubbing dog bellies and making kid crack-up.	
For four summers I worked in Alaska as a tour bus driver.	
Fun fact is that I just ran out of those and am looking for new ones.	
Fun in small doses!	
Halloween is my favorite holiday!	
I am a brand new follow-up nurse as of August 2016.	
I am a concert lover!	
I am a huge Razorback football fan…Woo Pig!	
I am a new and aspiring golfer.	
I am a quilter and have 3 cats. My goal is to be a quilting cat lady when I grow up.	
I am a second year Au.D. student in Arkansas.	
I am an avid Congenital Heart Defect Advocate. We have 4 children, 2 with Pulmonary Atresia w/IVS. I have a wide sarcastic streak and a love of strong coffee.	
I am currently training for my 7th marathon.	
I am a new to NW Arkansas.	
I am really passionate about finding cures for Inborn Errors of Metabolism and other rare diseases.	
I am the music leader at my church.	
I became a certified scuba diver at 10 years of age and have been scuba diving ever since.	
I became a grandpa for the first time on July 23rd.	

HEARTLAND GENETICS SERVICES COLLABORATIVE

NETWORKING

SUBMITTED BY: _____

FUN FACT:	WHO AM I?
I bicycle on average 3000 miles a year.	
I can play two musical instruments.	
I coach a nationally ranked cheer team.	
I drove Perot family during the 1992 Presidential Debates at Washington University in St. Louis but did not vote for Ross Perot.	
I earned the money as a teen for my first car by bottle feeding baby calves then selling them.	
I enjoy collecting art. My mother thinks I have way too much, as if there is such a thing as too much art! I have a current interest in Oklahoma Modernists.	
I enjoy kayaking.	
I enjoy knitting and baseball.	
I enjoy scrapbooking.	
I enjoy watching foreign movies.	
I enjoy watching my children play sports and binge watching Netflix!	
I got away from two speeding tickets! Once because I told the officer that I was listening to the bible and another because I told him that I was late to see my spiritual director!	
I grew up in a family of 8 kids!	
I have 3 dogs that I love very much.	
I have 3 small dogs that are all named after movie characters.	
I have 6 children (ages 7-26), now that's fun!	
I have a cat that was born with one eye!	
I have an aunt with sickle cell anemia.	
I have been involved in NBS activities since 1993.	
I have been mistaken for my 6 year old's grandma.	
I have four boys including a set of identical triplets.	
I have had one hole- in-one in golf.	
I have never lost at family game nights. We have a rule about the girl always wins, and I live with 4 boys.	
I have three grandsons who live in London, UK with their parents: Romeo, Prince, and King :-).	
I have two Newfoundlands that weigh 130 pounds apiece, and they are still puppies.	
I have worked for three different states' public health systems: Oklahoma, Idaho, and Kansas.	
I have worked in early intervention for just over 20 years and still know some of the families I met from long ago. I still enjoy meeting new families and being part of their journey. I also enjoy writing and recording my own music in my small, in-home studio. I've done this since I was around 13 years old and find it very therapeutic!	

HEARTLAND GENETICS SERVICES COLLABORATIVE

NETWORKING

SUBMITTED BY: _____

FUN FACT:	WHO AM I?
I hold 5 Ohio state records in powerlifting.	
I just moved to Austin, Texas.	
I knew I wanted to be a genetic counselor in 8th grade after reading a paragraph in my science textbook.	
I like listening to 80's pop rock music.	
I like sports. I like being outdoors. I enjoy watching my kids playing, drawing and dancing. I love meeting new people. I'm very international. I like learning about others culture and customs. I'm a people person. I care for others. I also love learning about new things and putting them in practice. Thanks.	
I like to read anything regarding History of Illnesses, States and People - Articles, Books, or Magazines. Also I like to climb Pinnacle Mountain with friends.	
I live closer to Canada than most of the continental U.S.	
I love being a dance mom.	
I love exotic food and am always ready to try new food and new drinks.	
I love going to the beach.	
I love Legos. And Disney. Yes, I have that set. It's a problem.	
I love teaching kids how to ride horses.	
I love to drive fast cars.	
I love to play volleyball.	
I love to read. I always carry a book with me. When the kids were younger and would argue while in the car, I'd pull off to the side of the road and start reading until they stopped. The first time I tried it, I got three chapters read.	
I love to ride my R1200R BMW motorcycle!	
I love to sing and read books.	
I love to travel, however a jar of peanut butter MUST always come with me.	
I love traveling!	
I met my nieces (3 and 4 months) over the summer when I went back home (Puerto Rico) over the summer.	
I once sunburned and blistered the soles of my feet while sleeping on a tropical beach.	
I once watched a lazy turtle contest.	
I play ice hockey!	
I played basketball in junior high and high school and was on the team.	
I practice yoga.	
I refuse! This is silly.	
I showed registered Polled Hereford cattle during my childhood years.	
I spent the summer interning at the Miami Zoo working with African carnivores and great apes.	
I studied abroad in Sweden.	

HEARTLAND GENETICS SERVICES COLLABORATIVE

NETWORKING

SUBMITTED BY: _____

FUN FACT:	WHO AM I?
I studied genetic counseling abroad in Australia for the summer!	
I survive in the National Guard.	
I take a road trip to the beach at least three times a year.	
I was a trivia question.	
I was able to visit the wonderful country of Costa Rica this last summer with my children.	
I was born on Super Bowl Sunday!	
I was struck by lightning this summer...	
If I was independently wealthy I would be an artist, painting and drawing most of the day. I would take breaks to go hiking :)	
I'm a foster parent.	
I'm a UAMS alum!	
I'm an avid hiker and nature photographer.	
I'm interested in humanitarian audiology.	
In a former life, I drove base taxi in Okinawa Japan	
I've been fortunate to work at Arkansas Children's Hospital for over 30 years and am still having FUN!	
John Wayne (real name Marion Morrison) is in my family tree	
Just started working in Kansas on October 10th!	
Love to skate.	
Loves To Ride Harley's / I have 7 motorcycles!	
My home was destroyed in a tornado, and my family only ended up with minor cuts.	
My husband and I receive a Christmas card from the President of the United States every year.	
My luck number is 13.	
My married name is the same as the street that I lived on in college.	
My secret TV pleasure is to watch Shark Tank.	
My son loves to tell people that I have my job because of him and his new born screening.	
My wife and I are having our first child this December.	
New Born Screening LTFU	
Negril Jamaica is my favorite vacation destination.	
Not only have I flown over the eye of a hurricane but I have been on land when one has come to shore.	
Our family likes to sail and recently acquired a 1973 22' O'Day sailboat called "Drowsy Maggie."	
Restores old British Motorcycles	

HEARTLAND GENETICS SERVICES COLLABORATIVE

NETWORKING

SUBMITTED BY: _____

FUN FACT:	WHO AM I?
Some of my loves: our 18 month old redhead, sand volleyball, red wine, and bitmojis!	
Speak English with a Dutch accent.	
Spends weekend riding bikes with friends.	
Spent 2 years working in ovarian cancer research.	
The Philadelphia Eagles are my favorite NFL team.	
This is my 25th year with Genetic Networks (GPGSN then Heartland). I started when I was 10.	
Took part in two Guinness World Records.	
Twenty years! I've been doing this for 20 years! I still love the work and the evolving nature of the field. I have a love/hate relationship with bureaucracy, but remain (perhaps naively) optimistic that we can and will get the resources we need to provide the best services we can! I've been blessed to work with and meet the best folks around, working in newborn screening and genetics!	
Visited over 15 countries and counting.	
Winning the game "Pay the Rent" on "The Price is Right" is on my bucket list.	
I love being a Mimi to all 8 of my grandbabies!	

HEARTLAND GENETICS SERVICES COLLABORATIVE

ACRONYMS

An Introduction to the Acronyms Used in the ACMG Office (adapted)
Genetics Alphabet Soup

Genetics Organizations

American Board of Genetic Counseling (ABGC): Office in Kansas City; Certifies masters-trained genetic counselors and accredits graduate programs.

American Board of Medical Genetics (ABMG): Certifies medical geneticists; Dr. Mimi Blitzer is Executive Director, Sharon Robinson is Administrator.

American College of Medical Genetics (ACMG): The American College of Medical Genetics and Genomics (ACMG) is an organization composed of biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors and other health care professionals committed to the practice of medical genetics.

American Society of Human Genetics (ASHG): Focus is research and K-undergraduate + public education in human genetics (~8000 members); Annual Meeting is in the fall and ACMG's BOD and Committees meet during these days. Dr. Joann Boughman is EVP.

Genetic Alliance: Umbrella organization for over 600 professional and disease-specific organizations; these are the consumer advocates and they are involved in education and public policy. Office is in DC; Sharon Terry is Executive Director.

International Society of Nurses in Genetics (ISONG): Professional organization for nurses in genetics (~200-300 members).

National Society of Genetic Counselors (NSGC): Office in Chicago; main professional organization primarily for genetic counselors (~2500 members); Annual Meeting is in the fall; Meghan Carey is Executive Director.

National Organization of Rare Disorders (NORD): An advocacy and information organization for individuals with rare disorders (many are genetic) and for the orphan drugs often used to treat these conditions.

Newborn Screening and Genetics Resource Center (NNSGRC): Based at the University of Texas, this is a clearinghouse for information on NBS; Dr. Brad Therrell is the Director.

Society for Metabolic Disorders (SIMD): Small organization that is home to metabolic/biochemical geneticists, nutritionists and those interested in the conditions identified by newborn screening (NBS); focus is research and treatment. They have no Executive Office; in the "odd numbered" years, our Annual Meetings overlap.

HEARTLAND GENETICS SERVICES COLLABORATIVE

ACRONYMS

Genetics Education

Accreditation Council for Graduate Medical Education (ACGME): Oversees residency training in the US.

Association of Professors of Human and Medical Genetics (APHMG): Medical school genetics course directors, medical genetics residency and fellowship program directors. This group uses ASHG as its executive office. It meets once a year.

Genetics Residency Review Committee (RRC): Responsible for the accreditation of medical genetics residency programs.

National Coalition for health Professional Education in Genetics (NCHPEG—pronounced NITCH peg): An organization made up of representatives of >100 medical, nursing and health professional associations focused on integrating genetics into all health professional education. Office in Baltimore; Joan Scott, MS, CGC is the Executive Director.

Organized Medicine

American Board of Medical Specialties (ABMS): The Board of Boards. ABMG is a member; we were the 24th and last medical specialty board to be accepted ca. 1992.

American Medical Association (AMA): Represents organized medicine in the largest sense. Based in Chicago; Dr. Ray Lewandowski (Corpus Christi) is the ACMG's representative to the AMA House of Delegates (HOD). ACMG MD Fellows are encouraged to belong to the AMA in order for us to maintain our seat in the AMA HOD.

Association of American Medical Colleges (AAMC): The organization that represents the medical schools in the US; based in DC.

Council of Medical Specialty Societies (CMSS): Organization of the medical organizations. ACMG is a member. Dr. Mira Irons is the ACMG representative to CMSS.

National Board of Medical Examiners (NBME): Based in Philadelphia, they administer the medical licensing examinations for physicians in the USA.

Department of Health and Human Services (DHHS): Agencies/offices we deal with

Agency for Healthcare Quality and Research (AHRQ)

American Health Information Communities (AHIC): A public-private partnership based in the Office of HHS Secretary Leavitt to move American to electronic health records by 2014. Personalized medicine and genomics workgroups involve ACMG.

HEARTLAND GENETICS SERVICES COLLABORATIVE

ACRONYMS

Health Resources and Services Administration (HRSA): Deals with public health and patient care services. Maternal Child Health Bureau (MSHB), Genetics Services Branch (GSB) is the office we work with most frequently (GSB/MCHB/HRSA). Joan Scott, MS, CGC is the Branch Chief. GSB has funded the

National Coordinating Center (NCC) for the Regional Genetics and Newborn Screening Collaborative (RC) Groups, based at ACMG. The US is divided into 7 RCs (see www.nccrcg.org).

Health Resources and Services Administration (HRSA): an agency of the U.S. Department of Health and Human Services, primary Federal agency for improving health and achieving health equity through access to quality services, a skilled health workforce and innovative programs; provides health care to people who are geographically isolated, economically or medically vulnerable.

Centers for Disease Control and Prevention (CDC): Based in Atlanta this is the nation's public health and epidemiology agency. It has a number of Centers and Office that deal with genetics and NBS issues. The Branch we interact the most with is:

- Newborn Screening and Molecular Biology Branch (NSMBB). Chief: Carla Cuthbert, Ph.D. FACMG, FCCMG
    - Newborn Screening Quality Assurance Program (NSQAP). Joanne Mei, PhD is Team Lead.
    - Newborn Screening Translation Research Initiative (NSTRI). Robert Vogt, PhD is Team Lead.
    - Biochemical Mass Spectrometry Laboratory (BMSL). Victor de Jesus, PhD is Team Lead.
    - Molecular Quality Improvement Program (MQIP). Suzanne Cordovado, PhD is Team Lead.

National Institutes of Health (NIH): Institutes and Offices we work with include (Francis Collins, MD, PhD NIH Director is a medical geneticist and ACMG Founding Fellow)

- Office of Rare Diseases (ORD): Coordinates research and information on rare diseases; Dr. Steven Groft is Director

- National Cancer Institute (NCI)

- National Human Genome Research Institute (NHGRI): Dr. Eric Green is Director

- *The Eunice Kennedy Shriver* National Institute for Child Health and Human Development (NICHD): This is where NBS research has its home. Newborn Screening Translational Research Network (NBSTRN) funding comes to ACMG from NICHD. Alan Guttmacher, MD is the Director of NICHD. Geneticist Dr. Jim Hansen of NICHD works closely with ACMC.

- National Library of Medicine (NLM): Works with partners to standardize terminology used in medicine and genetics. Most contact is with Lister Hill National Center for Biomedical Communications and the National Center for Biotechnology Information (NCBI).

HEARTLAND GENETICS SERVICES COLLABORATIVE

ACRONYMS

US Food and Drug Administration (FDA): Oversees the safety of medications, medical devices including some genetics tests and food. Has 5 Centers, all are local.

Department of Health and Human Services (DHHS): Advisory Committees

Secretary's Advisory Committee on Genetics Health and Society (SACGHS): Run out of NIH, this committee met 3 times a year until its charter ended in October 2010. It was involved in making recommendations about the oversight of genetic testing, as well as gene patents and a number of other issues of deep interest to ACMG. Several ACMG members were appointed to this committee. Websites with all documents still exists.

Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC): Run out of GSB/MSHB/HRSA, this committee deals with issues related to NBS.

Other Medical/Scientific Organizations

American Academy of Family Physicians (AAFP): The ACMG of family physicians.

American Academy of Pediatrics (AAP): The ACMG of the pediatricians.

American College of Obstetrics and Gynecology (ACOG): the ACMG for Ob/Gyn physicians.

American Public Health Association (APHA): The professional organization for public health professionals.

Association of Public Health Laboratories (APHL): National organization that includes the state NBS labs; based in Sliver Spring, MD. Jelili Ojodu is ACMG's main contact with APHL.

Ethical, Legal, and Social Implications (ELSI) program: integral part of the Human Genome Project, to address and identify issues raised by genomic research.

Genetics Society of America (GSA): Sponsors a number of research conferences, supports education of students, and publishes the journal "Genetics."

HALI Project: a project directed by Brad Thompson, inspired by his daughter Hali, with the aim to "bridge the gap" amongst services for children of special needs by providing training, education, and counseling to families.

Hispanic Access Advisory Committee (HAAC)

Institute of Medicine (IOM): The medical/health care policy arm of the National Academy of Sciences. They hold panels, roundtables and advisory committees on which Mike Watson often participates.

HEARTLAND GENETICS SERVICES COLLABORATIVE

ACRONYMS

Leadership Education in Neuro-developmental Disabilities (LEND): LEND programs provide long-term, graduate level interdisciplinary training as well as interdisciplinary services and care. The purpose of the LEND training program is to improve the health of infants, children, and adolescents with disabilities. They accomplish this by preparing trainees from diverse professional disciplines to assume leadership roles in their respective fields and by ensuring high levels of interdisciplinary clinical competence.

Longitudinal Pediatric Data Resource (LPDR)

National Center for Hearing Assessment and Management (NCHAM)

Newborn Screening (NBS)

Newborn Screening Technical assistance and Evaluation (NewSTEPS) Program: national newborn screening program designed to provide data, technical assistance, and training to newborn screening programs across the country and to assist states with quality improvement initiatives. NewSTEPs is a comprehensive resource center for state newborn screening programs and stakeholders.

Title V: The Title V Program is located in the Division of Developmental Disabilities Services (DDS) Children's Services unit and is a federal program that serves children with chronic medical conditions.

HEARTLAND GENETICS SERVICES COLLABORATIVE

GLOSSARY

SOURCE: HTTP://WWW.GENESINLIFE.ORG/GLOSSARY

A

abnormal result: a possible result of a screening test. An abnormal result does not determine a diagnosis, and means additional testing is needed to see if the individual has a condition. Also referred to as positive result.

acquired mutations: a change within a sequence of DNA caused by environment factors (sun, radiation, or chemicals), aging, or chance.

acute: describes an illness that only affects an individual for a short period of time.

ADA: the Americans with Disabilities Act of 1990 gives civil rights protections to individuals with disabilities and guarantees equal opportunity for individuals with disabilities in public accommodations, employment, transportation, state and local government services, and telecommunications.

adenine: one of four chemical bases in DNA, denoted (A), with the other three being cytosine (C), guanine (G), and thymine (T).

advocacy group: a group of people who work together to support a cause.

alkaptonuria: a rare genetic disorder in which a person's urine turns a dark brownish-black color when exposed to air.

allele: one of two or more versions of a gene. An individual inherits two alleles for each gene, one from each parent.

amino acids: amino acids are a set of 20 different molecules used to build proteins.

annotation: the process of identifying the locations of genes in a genome and determining the role of those genes. Annotation is a process that often follows gene sequencing.

autosomal chromosomes: all of the human chromosomes except for the X and Y chromosomes.

autosomal dominant: a pattern of inheritance where having only copy of the gene that does not work correctly results in the condition, and the condition affects males and females equally.

autosomal recessive: a pattern of inheritance where copies of the gene that do not work properly are needed in order to have the condition, and the condition affects males and females equally.

B

Biobank: a collection of human biological samples (such as blood and tissue) and medical information about the people who gave their samples for research studies.

GLOSSARY

biomedicine: when the principles of natural sciences are used to evaluate and treat medical conditions.

biorepository: a collection of human biological samples (such as blood and tissue) and medical information about the people who gave their samples for research studies.

blinding: in a scientific experiment, a blind is where some of the people involved are prevented from knowing certain information that might lead to conscious or subconscious bias on their part, making the results not completely accurate.

blood sample: when blood is drawn from the human body in order to be tested for medical purposes.

BRCA 1 and 2: the first two genes found to be associated with inherited forms of breast cancer.

C

cancer: a group of diseases characterized by uncontrolled cell growth. Cancer begins when a single cell mutates, resulting in a breakdown of the normal regulatory controls that keep cell division in check.

carrier: a person who has a change in only one gene of a pair and the other gene of the pair is working normally. Carriers typically do not display the symptoms of the condition, but can pass on the mutation to offspring.

carrier screening: a type of genetic testing to determine if an individual is a carrier for a genetic disease.

cell: the basic building blocks of all living things.

chromosome: an organized structure of DNA containing many genes that is wrapped around proteins found in cells. Humans typically have 23 pairs of chromosomes or 46 total.

chronic: describes an illness that affects an individual for a long period of time, possibly their entire life.

chronic disease: a long-lasting health condition such as cancer, coronary heart disease, and diabetes.

CLIA: Clinical Laboratory Improvement Amendments are regulations created in 1988 by the Center for Medicare and Medicaid Services to ensure quality laboratory testing on humans.

clinical geneticist: a physician with training in genetics who meets with patients to evaluate, diagnose, and manage genetic disorders.

clinical testing: testing that is done to confirm if a person has a condition.

cloning: creating an organism that has the same genes as the original.

GLOSSARY

confirmatory test: confirm or rule out a medical condition in an individual with concerning symptoms or an out-of-range screening result.

congenital: a condition that is present from birth.

copy number variation: when the number of copies of a particular gene varies from one individual to the next.

cytogenetics: the branch of genetics that studies the number and structure of human chromosomes.

cytosine: one of four chemical bases in DNA, denoted (C), with the other three being adenine (A), guanine (G), and thymine (T).

D

deoxyribonucleic acid (DNA): a molecule found in chromosomes that carries genetic information. DNA is composed of four units (called bases) that are designated A, T, G, and C.

diagnostic genetic testing: genetic testing used to identify if an individual has a condition associated with symptoms they are showing.

direct-to-consumer genetic testing: a type of genetic testing that is available directly to the consumer without having to go through a healthcare professional.

disorder: a disturbance in physical or mental health functions.

DNA replication: the process by which a molecule of DNA is duplicated.

DNA sequence: the sequence of the bases of DNA spell out instructions for making all of the proteins needed by an organism.

Dolly: the first mammal ever cloned (a sheep).

dominant: individuals receive one version of a gene from each parent. Sometimes a version of a gene is dominant. Dominant genes have a more powerful effect than recessive genes and are thus more likely to be expressed or have a visible effect on the body. If a dominant gene and a recessive gene are inherited, the effects of the dominant gene will mask those of the recessive gene.

double helix: the twisted-ladder shape that two strands of DNA form.

Down syndrome: also called trisomy 21. Down syndrome is a genetic disease in which a person inherits an extra copy of chromosome 21. The extra chromosome causes problems with the way the body and brain develop.

GLOSSARY

dried-blood-spot testing: testing the small amount of dried blood on the filter paper cards used in newborn screening.

DTC: Direct-To-Consumer is a type of genetic test that is available directly to the consumer without having to go through a health care professional.

E

emergency preparedness: the act of being prepared with your medical information in case an emergency event ever occurs.

endocrinologist: a doctor that specializes in disorders of the glands.

environmental factors: chemicals, sun, and radiation that can cause mutations in DNA and can result in a disease that was acquired and not inherited.

enzyme: a protein that helps with chemical reactions in the body.

epigenetic markings: changes in how the expression of a gene is regulated that is not caused by a change in the gene sequence.

epigenetics: an emerging field of science that studies heritable changes caused by the activation and deactivation of genes without any change in the underlying DNA sequence of the organism.

epigenome: it consists of chemical compounds that modify, or mark, the genome in a way that tells it what to do, where to do it, and when to do it.

eukaryote: A type of cell that has a nucleus and membrane-bound organelles.

exon: a section of DNA that serves as the set of instructions for constructing a protein.

expression: the process by which the information from a gene is translated and used to make a functional product, either a protein or a strand of RNA. When this occurs, the gene is said to have been expressed.

F

false negative result: the result of a diagnostic test came back as normal when the disease is actually present. Tests are designed to make sure this type of mistake happens as little as possible.

false positive result: the result of a diagnostic test came back positive or abnormal when the disease is not actually present. Tests are designed to make sure this type of mistake happens as little as possible.

family health history: a record of medical information about an individual and their family members, as well as information about the eating habits, activities, and environments the family shares.

family tree: a record of members of a family and their relationships.

fatal: Causing death.

FDA: the Food and Drug Administration is the governmental organization responsible for protecting the public by assuring the safety, efficacy, and security of drugs, biological products, medical devices, food, and cosmetics.

first degree relative: a family member who shares about 50 percent of their genes with a particular individual in a family. First degree relatives include parents, offspring, and siblings.

follow-up testing: testing procedure that takes place after a positive or abnormal test result. Follow-up testing is designed to limit false positive results.

fragile X syndrome: a genetic disorder caused by mutations in a gene on the X chromosome. Fragile X syndrome affects mostly males. It is the most common form of inherited intellectual disability (mental retardation). Other symptoms include distinctive facial features and poor muscle tone.

fraternal twins: results from the fertilization of two separate eggs during the same pregnancy. Fraternal twins may be of the same or different sexes and they share half of their genes just like any other siblings.

frequency: the number of times something happens in a specific group.

G

gene: a sequence of DNA that carries the instructions for making a sequence of RNA, which in turn instructs and assists in the creation of proteins.

gene regulation: the process of turning genes on and off, which ensures that the appropriate genes are expressed at the proper times.

gene therapy: an experimental technique for treating disease that works by introducing a healthy copy of a non-functioning gene into the patient's cells.

genetic code: the instructions in a gene that tell the cell how to make a specific protein. A, C, G, and T are the letters of the DNA code.

genetic counselor: a healthcare provider who has special training in genetic conditions. Genetic counselors help families understand genetic disorders and counsel families in making decisions about the testing or management of a genetic disorder.

HEARTLAND GENETICS SERVICES COLLABORATIVE

GLOSSARY

genetic disease: a condition that is caused by changes in genes or chromosomes.

genetic disorder: a disease that is caused by an abnormality in an individual's DNA.

genetic map: shows where genes are located relative to each other on chromosomes.

genetic marker: a DNA sequence with a known physical location on a chromosome. Genetic markers can help link an inherited disease with the responsible gene.

genetic testing: a laboratory test to look for a change in a gene of an individual. The results of a genetic test can be used to confirm or rule out a diagnosis of a genetic disease.

geneticist: a doctor or scientist who studies heredity and how genes work and contribute to disease.

genetics: the scientific study of how particular qualities or traits are passed down from parents to child.

genome: the complete DNA sequence in the chromosomes of an individual.

genomics: the study of the entire genome of an organism whereas genetics refers to the study of a particular gene.

genotype: a trait or gene that an individual inherits.

GINA: the Federal legislation passed in 2008 that makes it unlawful to discriminate against individuals based on their genes for health insurance or employment purposes.

guanine: one of four chemical bases in DNA, denoted (G), with the other three being adenine (A), cytosine (C), and thymine (T).

gynecologist: a doctor who specializes in the health care of women.

H

healthcare provider: a doctor, nurse, physician's assistant, or genetic counselor.

hereditary mutations: a change within a gene that can be passed to offspring.

heredity: the passing of traits from parents to offspring.

heterozygous: refers to having inherited different forms of a particular gene from each parent.

histone: proteins that DNA wraps around as it coils into chromosomes. These proteins keep the DNA from becoming tangled and damaged.

homozygous: a genetic condition where an individual inherits the same alleles for a particular gene from both parents.

Human Genome Project: an international project that mapped and sequenced the entire human.

human subjects protections: the government has policies to protect people that participate in genetics research.

Huntington's disease: a genetic disorder that affects muscle coordination and brain function. Nerve cells in certain parts of the brain waste away, or degenerate, so it is called a neurodegenerative disorder. It is inherited through an autosomal dominant pattern.

I

identical twins: result from the fertilization of a single egg that splits in two. Identical twins share all of their genes and are always of the same sex.

immunity: an inherited, acquired, or produced resistance to infection by a specific pathogen.

immunization: the process of producing immunity to an infectious organism or agent in an individual or animal through vaccination.

immunologist: a doctor that specializes in conditions of the immune system.

in-range screening result: the clinical test did not show any signs of conditions.

informed consent: permission given by an individual to proceed with a specific test, procedure, or research study with an understanding of the risks and benefits of the activity.

inheritance: passing of genes and traits from parents to child.

Institutional Review Board (IRB): makes sure that risks to people are as low as possible in a research study.

isolate DNA: to separate DNA from the other cell components.

K

karyotype: refers to an individual's full set of chromosomes. May also refer to a photographic representation of an individual's chromosomes with all 23 pairs positioned next to one another.

knockout: refers to an organism that has been genetically engineered such that one or more specific genes are inactivated or do not work properly. Scientists create knockouts (often in mice) so that they

can study the impact of these genes when they do not function and learn something about the genes' function.

L

linkage: the close association of genes or other DNA sequences on the same chromosome. The closer two genes are to each other on the chromosome, the greater the probability that they will be inherited together.

M

medical geneticist: a doctor who specializes in genetics and genetic disorders.

medical home: the facility or physician that coordinates the care of an individual with a complex medical condition.

Mendelian inheritance: refers to patterns of inheritance that are characteristic of organisms that reproduce sexually.

metabolic disorder: a disorder or defect in the way the body breaks down food or other products (metabolism).

methylation: when a base on the DNA strand is altered by the addition of a methyl group. This change causes that section of DNA to coil more tightly, preventing the genes around if from being used or expressed. This process is important as embryos develop and new cells take on specific roles in the body, but errors in DNA methylation have been linked to many human diseases.

mitochondria: membrane-bound cell organelles that generate most of the chemical energy needed to power the cell's biochemical reactions.

mitochondrial inheritance: the mitochondrion, an organelle in the cell, contains its own genome. Mutations in these genes are responsible for several known genetic diseases. Individuals only inherit mitochondrial DNA from their mothers.

mitosis: the process occurring in cells where all the chromosomes are replicated and the cell contents are equally divided into two daughter cells.

model organisms: organisms used in medical research to mimic a disease found in humans and to study its prevention, diagnosis, and treatment.

mutation: any change that occurs in a gene. These may occur because of errors in the replication process or directly from the environment. Most mutations do not have any effect, some may have positive effects, and many have harmful effects.

N

natural selection: the evolutionary process where the organism best adapted to it's environment survives.

negative test result: a possible result of a screening or diagnostic test. If the result came back from a genetic screening test, it means that the test did not find any evidence of the genetic condition for which it was testing. If the result came back from a genetic diagnostic test, then the test did not find any evidence that the person has the genetic condition for which it was testing.

neonatal: during the first month of life.

newborn screening: a process of testing newborn babies for some serious, but treatable, conditions.

non-invasive: a medical test or procedure that does not require a doctor to insert any device through the skin or into a body opening.

nucleus: a membrane-bounded region inside each cell that provides a sanctuary for genetic information, including the long strands of DNA that encode this genetic information.

O

oncogene: a mutated gene that contributes to the development of a cancer.

opt-out: a patient's right to refuse screening tests.

organ: a collection of tissues that structurally form a functional unit specialized to perform a particular function. Your heart, kidneys, and lungs are examples of organs.

organelle: a subcellular structure that has one or more specific jobs to perform in the cell, much like an organ does in the body.

out-of-range result: this result means that the screening test did show signs that the individual may be at higher risk of having one or more conditions.

P

patient confidentiality: the right of an individual patient to have personal, identifiable medical information kept private.

pediatrician: a primary care physician who specializes in the medical care of infants, children, and adolescents.

GLOSSARY

pedigree: a genetic representation of a family tree that diagrams the inheritance of a trait or disease though several generations.

pharmacogenetics: the study of how genetics determine drug behavior and why some drugs work differently between individuals.

phenotype: an individual's observable characteristics or traits.

physician: a person licensed to practice medicine, also known as a medical doctor.

positive screen (positive test result): a possible result of a screening or diagnostic test. If the result came back from a genetic screening test, further testing must be done to determine if the person has the condition which was being tested for. If the result came back from a genetic diagnostic test, then the person has the condition and can pursue treatment options for that condition.

predictive genetic test: a genetic test for individuals not yet showing symptoms of a genetic disorder but have a family history of the condition or an increased risk of developing the condition.

predispositional: symptoms are likely, but not certain to develop if testing suggests you have disease gene.

prenatal: anytime before the birth of the baby.

prenatal care providers: healthcare professionals who aid a woman throughout her pregnancy.

presymptomatic: you will eventually develop symptoms if testing suggests you have the disease gene.

primary care provider: a doctor trained to treat a wide variety of health-related problems.

privacy protections: ensure that blood spots cannot be accessed by a third party, including insurers and law enforcement.

Progeria: a rare disease characterized by accelerated aging.

prokaryote: a type of cell that does not have a nucleus or membrane-bound organelles.

prostate cancer: a disease characterized by uncontrolled cell growth in the prostate gland, which is part of the male reproductive system.

proteins: make up many parts of every cell in the body. Proteins are made up of amino acids. The order of these amino acids determines what form and job a protein has.

protein sequencing: the process of determining the order of amino acids (the molecules that make up proteins) of a particular protein.

public health: the science and practice of protecting and improving the health of a community.

pulmonologist: a doctor that specializes in lung conditions and diseases.

Q

quality assurance: process of defining the quality of performance required for each step in the testing process.

quality control: monitoring the degree of adherence to defined criteria, taking corrective action when the system fails and documenting all of these events to convey the total quality of performance.

R

rare health conditions: an uncommon disorder that affects the ability of the human body to function normally.

recessive: a quality found in the relationship between two versions of a gene. Individuals receive one version of a gene, called an allele, from each parent. If the alleles are different, the dominant allele will be expressed, while the effect of the other allele, called recessive, is masked. In the case of a recessive genetic disorder, an individual must inherit two copies of the mutated allele in order for the disease to be present.

referral: individuals receive one version of a gene from each parent. Sometimes a version of a gene is recessive. Recessive genes have a less powerful effect than dominant genes. If a dominant gene and a recessive gene are inherited, the effects of the recessive gene are not visible or are masked by the more powerful dominant gene's effects. If two recessive genes are inherited, then the effects of the recessive genes are visible.

registry: a collection of medical information, clinical data, and demographics (age, male or female, etc.) about people with a specific disease or condition.

research geneticist: geneticists who focus on research and study the origin, treatment, and prevention of genetic conditions.

retesting: when a test needs to be repeated in order to clarify, confirm or reject the results of the initial test.

ribonucleic acid (RNA): a molecule similar to DNA. Unlike DNA, RNA is single-stranded. An RNA strand has a backbone made of alternating sugar (ribose) and phosphate groups. Attached to each sugar is one of four bases--adenine (A), uracil (U), cytosine (C), or guanine (G).

S

SCID: Severe Combined Immunodeficiency is an inherited condition affecting the immune system causing individuals to be more susceptible to infectious diseases.

screening tests: tests that analyze DNA samples to detect the presence of a gene or genes associated with an inherited disorder.

sequencing: DNA sequencing is a detailed description of the order of the chemical buildings blocks, or bases, in a given stretch of DNA. The sequence of bases tells scientists the type of genetic information that is carried in a particular segment of DNA.

sex chromosome: a sex chromosome is a type of chromosome that participates in sex determination. Humans have two sex chromosomes, the X and the Y. Females have two X chromosomes in their cells, while males have both X and a Y chromosomes in their cells.

sex linked: a trait in which a gene is located on a sex chromosome.

sickle cell anemia: a disorder that is passed down through families and causes red blood cells to form an abnormal crescent, or sickle, shape. These sickled red blood cells cannot carry enough oxygen to the body. It is inherited in the autosomal recessive pattern.

single nucleotide polymorphisms (SNPs): a type of polymorphism involving variation of a single base pair. Scientists are studying how single nucleotide polymorphisms, or SNPs (pronounced snips), in the human genome correlate with disease, drug response, and other phenotypes.

social worker: a trained professional who provides social services to those in need.

somatic cells: any cell in the body except for sperm and egg cells.

specialist: a healthcare provider who has special knowledge about a condition or a specific part of a condition.

standard medical procedure: surgery or practice that is a common and well accepted as the best course of treatment.

state assistance: payment given to individuals by government agencies on the basis of need.

stem cell: a cell with the potential to form many of the different cell types found in the body.

support group: a group of people who are all impacted by the same condition and come together to share experiences and help one another.

symptom: evidence of a disorder or disease that directly affects and is noticed by the patient, such as a rash, pain, nausea, or a runny nose.

T

transcription: the process within the cell that uses DNA instructions to create pieces of RNA, which can then be used to make proteins or perform other tasks throughout the body.

testing outcomes: the possible results you can receive after participating in a test such as positive, negative, or inconclusive.

thymine: One of four chemical bases in DNA, denoted (T), with the other three being adenine (A), cytosine (C), and guanine (G).

trait: a specific characteristic of an individual.

transcription factor: proteins that bind to specific sections of DNA and control transcription or the process of using DNA instructions to create new strands of RNA.

transition process: the time when an individual with a genetic condition or special healthcare needs must change his or her system of care to reflect his or her age. After reaching adolescence or adulthood an individual will likely need to change health care providers, most likely from a pediatrician to an adult physician.

treatable condition: a condition with a known treatment that can improve the survival and/or quality of life of an individual.

true positive result: a small percentage of individuals with out-of-range results do have the condition and must pursue treatment options.

U

uracil: one of four chemical bases that are part of RNA, denoted (U). The other three bases are adenine (A), cytosine (C), and guanine (G).

V

virus: an infectious agent that occupies a place near the boundary between the living and the nonliving.Viruses enter host cells and hijack the enzymes and materials of the host cells to make more copies of themselves.

W

whole genome sequencing: whole genome sequencing is the mapping out of a person's unique DNA. Your genome is the unique blueprint for your body.

working copy: a gene that functions the way it is intended to.

X

X chromosome: one of two sex chromosomes. Humans have two sex chromosomes, the X and the Y. Females have two X chromosomes in their cells. Males have X and Y chromosomes in their cells.

X-linked dominant: an inheritance pattern where X-linked means that the disease gene is located on the X sex chromosome and dominant means that having only one copy of the gene that does not work properly causes the condition. Affects more females than males.

X-linked recessive: an inheritance pattern where X-linked means that the disease gene is located on the X sex chromosome and recessive means that two copies of the gene that does not work properly are needed to have the condition. Affects more males than females.

Y

Y chromosome: one of two sex chromosomes. Humans have two sex chromosomes, the X and the Y. Males have X and Y chromosomes in their cells.

HEARTLAND GENETICS SERVICES COLLABORATIVE

NOTES

HEARTLAND GENETICS SERVICES COLLABORATIVE

NOTES

HEARTLAND GENETICS SERVICES COLLABORATIVE

NOTES

HEARTLAND GENETICS SERVICES COLLABORATIVE

NOTES

HEARTLAND GENETICS SERVICES COLLABORATIVE

NOTES

HEARTLAND GENETICS SERVICES COLLABORATIVE

NOTES