HEARTLAND GENETICS SERVICES COLLABORATIVE

11TH ANNUAL CONFERENCE

APRIL 29 – MAY 1, 2015

KANSAS CITY, MO

WELCOME LETTER

ELEVENTH ANNUAL CONFERENCE

APRIL 29 – MAY 1, 2015

KANSAS CITY, MISSOURI

Dear Colleagues and Partners,

Welcome to the eleventh 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 exhibitors, poster presentations, to participate in the work group sessions 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
Dustin Foster

Conference Planning Committee:
Elizabeth Abbey
Jo Ann Bolick
Carol Johnson
Jeremy Penn
Mark Smith

Heartland Advisory Board:

Stan Berberich
Joni Bosch
Lucy Fossen
Cathy Harbison
David King
Julie Luedtke
Kim Piper
Laurie Smith
Larry Weatherford

JoAnn Bolick
Jeanne Egger
Esperanza Font-Montgomery
Jamey Kendall
Tiffany Lepard-Tassin
Jeremy Penn
Sharmini Rogers
Sharon Vaz
Jackie Whitfield

HEARTLAND GENETICS SERVICES COLLABORATIVE

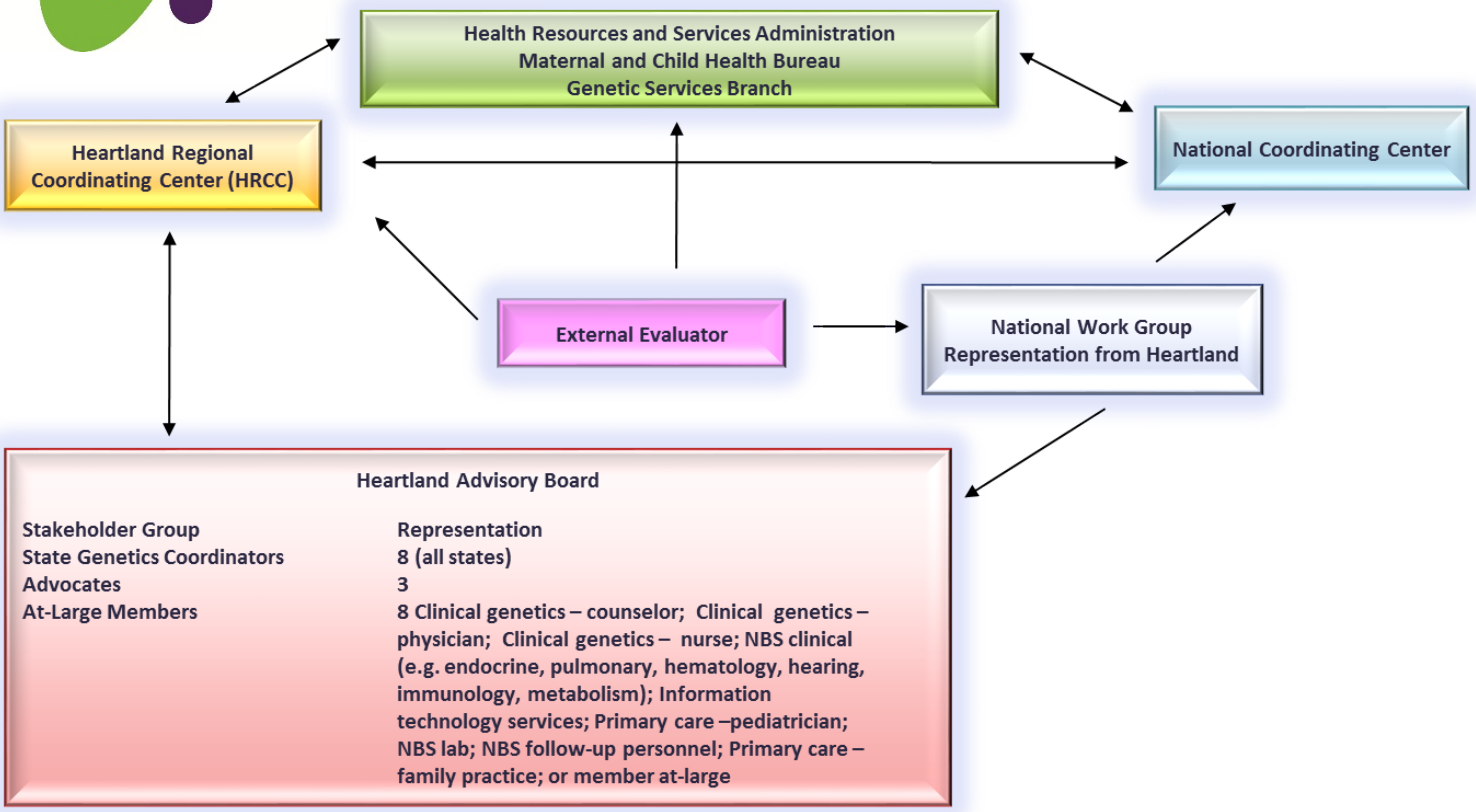
MISSION STATEMENT

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

ORGANIZATIONAL CHART



Newborn Screening Work Group
(15-20 core members)

- 3 Advocates
- 2 Metabolic clinicians
- 3 Long Term Follow-Up Coordinators
- 2 Newborn Screening Laboratorians
- 3 Newborn Screening Coordinators
- 4 State Genetics Coordinators

Clinical Services Work Group
(8-10 core members)

- 2 Advocates
- 4 Clinicians
- 2 Laboratorians
- 2 State Genetics Coordinators
- 1-2 Primary Care Providers

Advocacy Work Group
(16 core members)

- 2 Advocates per state

Ad Hoc Work Groups

- Early Hearing Detection and Intervention
- Hispanic Access Advisory Committee

APRIL 29 – MAY 1, 2015

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Baebies
615 Davis Drive, Suite 800
PO Box 14403
Durham, NC 27709
www.baebies.com

EXHIBITORS

National Coordinating Center (NCC)
7220 Wisconsin Avenue, Suite 300
Bethesda, MD 20814
www.nccrcg.org

&

National Genetics Education and Consumer Network (NGECN)
4301 Connecticut Ave NW, Suite 404
Washington DC, 20008
www.geneticalliance.org/programs/genesinlife/consumernetwork

This project is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) under H46MC24089 and Heartland Genetics Services Collaborative for $600,000 with no nongovernmental funding sources. This information or content and conclusions are those of the author and should not be construed as the official position or policy of, nor should any endorsements be inferred by HRSA, HHS or the U.S. Government.

HEARTLAND GENETICS SERVICES COLLABORATIVE

AGENDA

ANNUAL CONFERENCE AND ANCILLARY MEETINGS
APRIL 29 – MAY 1, 2015
AIRPORT MARRIOTT
KANSAS CITY, MISSOURI

Wednesday, April 29

TIME	EVENT	LOCATION
09:00 – 04:15	**EHDI Workshop** (designated invitees)	Salon B
02:00 – 04:00	**Advocate Work Group**	Liberty
04:00 – 07:30	**Annual Meeting Registration Open**	Ballroom Foyer
04:00 – 08:00	**Heartland Advisory Board Meeting** *Dinner provided for Advisory Board members and invited guests*	Salon C

Thursday, April 30

07:00 – 05:30	**Registration Open**	Ballroom Foyer
07:00	**Sponsors/Exhibitors Set Up**	Salon A
07:00 – 08:00	**BREAKFAST**	Salon B
08:00 – 08:10	**Welcome Remarks** *Brad Schaefer, MD* *Heartland Co-Director*	Salon C - E
08:10 – 08:30	**Perspectives from HRSA** *Joan Scott, MS, CGC* *Chief, Genetics Services Branch*	Salon C - E
08:30 – 08:50	**National Coordinating Center (NCC) Report** *David Flannery, MD* *NCC Medical Director*	Salon C - E
08:50 – 09:10	**State of the Heartland Region** *Klaas Wierenga, MD* *Heartland Co-Director*	Salon C - E
09:10 – 09:20	**BREAK**	Salon A
09:20 – 09:40	**Perspectives from EHDI** *Karl White, PhD* *Director, National Center for Hearing Assessment and Management*	Salon C - E
09:40 – 10:00	**NewSTEPs Updates** *Careema Yusuf, MPH* *Senior Specialist, NewSTEPs*	Salon C - E
10:00 – 10:20	**National Genetics Education and Consumer Network (NGECN)** *Sharon Romelczyk, MPA* *Program Manager, NGECN, Genetic Alliance*	Salon C - E

HEARTLAND GENETICS SERVICES COLLABORATIVE

AGENDA

10:20 – 10:30	**BREAK**	**Salon A**
	Pilot Projects Update	**Salon C - E**
10:30 – 11:00	▪ **Williams Syndrome Project** *Beth Kozel, MD, PhD* *Washington University School of Medicine*	
11:00 – 11:30	▪ **Review of Best Practices in Documenting Newborn Screening Refusals for States** *Jeremy Penn, PhD* *North Dakota State University*	**Salon C - E**
11:30 – 01:00	**LUNCH PROVIDED**	**Salon B**
	Priority Initiatives in the Heartland Region	**Salon C - E**
01:00 – 01:45	▪ **Hispanic Access Project** *Mary Ann Coffman, MS, CGC* *Project Coordinator, Heartland Regional Coordinating Center*	
01:45 – 02:15	▪ **Genetics Systems Assessment** *Ann Chou, PhD, MPH, MA* *University of Oklahoma Health Sciences Center*	**Salon C - E**
02:15 – 02:30	**BREAK**	**Salon A**
02:30 – 03:00	▪ **Role of the Geneticist in the Medical Home** *Brad Schaefer, MD* *and* *Christine Bruha, BS* *University of Arkansas for Medical Sciences*	**Salon C - E**
03:00 – 03:30	**Heartland Telemedicine Update** *Brad Schaefer, MD* *Heartland Co-Director* *and* *Lori Williamson Dean, MS, CGC* *Heartland Program Manager*	**Salon C - E**
03:30 – 05:00	**Poster Session with Authors** *Light refreshments and optional cash beverages*	**Salon A**

Dinner on your own — see meeting packet for shuttle services and restaurants

HEARTLAND GENETICS SERVICES COLLABORATIVE

AGENDA

Friday, May 1

07:00 – 08:00	**BREAKFAST**	**Salon B**
08:00 – 09:30	**"What if I say something wrong?" (Panel)** *Julie Kaylor, MS, CGC* *Wendy Parent-Johnson, PhD* *Val Renault, MA* *Mark Smith, MS*	**Salon C - E**
09:30 – 10:00	**Newborn Screening in the Genomics Era: Implications for State Programs** *Aaron Goldenberg, PhD* *Case Western Reserve University*	**Salon C - E**
10:00 – 10:20	**BREAK TO CHECKOUT**	**Hotel Lobby**
10:20 – 12:30	**BREAKOUT SESSIONS** ▪ **Clinical Services Work Group** ▪ **Newborn Screening Work Group**	**Salon C - E**
12:30 – 01:40	**LUNCH BUFFET**	**Salon B**
01:40 – 02:00	**Fruits of our Labors** *Barb Jackson, PhD*	**Salon C - E**
02:00 – 2:15	**Beginnings and Endings** *Barb Jackson, PhD* *Heartland Evaluator* *and* *Anayeli Herrera Morales, MPH* *Heartland Program Coordinator*	**Salon C - E**
02:15 – 02:30	**Closing Remarks** *Lori Williamson Dean, MS, CGC* *Heartland Program Manager*	**Salon C - E**
02:30	**Adjourn**	

NAME:
G. Bradley Schaefer, MD, FAAP, FACMG
Professor of Genetics and Pediatrics
Founding Director, Division of Medical Genetics
University of Arkansas for Medical Sciences
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: Welcome to the 11th Annual Heartland Genetics Services Collaborative Conference; Role of the Geneticist in the Medical Home; and Heartland Telemedicine Update

NOTES:

NAME:
Joan A. Scott, MS, CGC
Chief, Genetic Services Branch
Division of Services for Children with Special Health Needs
Maternal and Child Health Bureau
jscott2@hrsa.gov

BIO: Ms. Scott is Chief, Genetic Services Branch in the Division of Children with Special Health Needs (DSCSHN), Maternal and Child Health Bureau (MCHB) at Health Resources and Services Administration (HRSA). She is a certified genetic counselor with more than 35 years' experience in clinical genetics, genetics education, laboratory medicine, the biotechnology industry, and the ethical, legal, social, and policy implications of advances in genomics. Ms. Scott's career has focused on the application of genomic discoveries to health care. Prior to coming to HRSA, she was Executive Director of the National Coalition for Health Professional Education in Genetics (NCHPEG), where she led a national effort to promote health professional education and access to information about advances in human genetics, and a Research Scientist in the Berman Institute of Bioethics at The Johns Hopkins University where she studied public and stakeholder attitudes about genomics.

Prior to NCHPEG, Ms. Scott was Director of the Genetics and Public Policy Center at Johns Hopkins University which was established to fill an important niche in the science policy landscape. There she led the Center's efforts to address policy issues related to advances in genetics, genetic testing quality and oversight, and public engagement in genetic research. Prior to coming to the Center in 2002, Ms. Scott was a director in GeneLogic, Inc. overseeing the operations of a large biorespository for use in genomic discovery. She also has served as general manager and director of Genetic Services at the clinical diagnostic lab OncorMed from 1994-1998. Clinically, she has practiced in a variety of academic, outreach, and private practice settings, including pediatric, adult, and reproductive genetic clinics.

Ms. Scott is a past president of the National Society of Genetic Counselors and founding member of the American Board of Genetic Counseling. She has served on numerous national committees and work groups including the IOM Roundtable on Translation Genome-based Research into Health; the Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group; the Secretary's Advisory Committee on Genetics, Health, and Society Task Force on DTC Genetic Testing; the Maryland Insurance Administration Workgroup on Genetic Testing; the National Cancer Institute's CaHUB Advisory Committee; and the Genetic Alliance Biobank Advisory Board. Ms. Scott holds an M.S. (Human Genetics Program) from Sarah Lawrence College and a B.A. in Anthropology and Zoology from Kent State University. She has been certified by the American Board of Medical Genetics with a subspecialty in Genetic Counseling, and was recertified by the American Board of Genetic Counseling in 2006.

TOPIC: Perspectives from HRSA

NOTES:

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

BIO: Since 2008, Ms. Keehn has served as the Project Manager for 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). 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:

NAME:
Klaas J. Wierenga, MD, MSc, FACMG
Associate Professor, Department of Pediatrics,
Division of Genetics
Director of Medical Genetics Residency Program
klaas-wierenga@ouhsc.edu

BIO: Dr. Klaas J. Wierenga is a medical geneticist at the University of Oklahoma Health Sciences Center (OUHSC) in Oklahoma City, OK, with degrees in Biology and Medicine from the Groningen State University in the Netherlands. He is Board-certified in Clinical and Biochemical Genetics. His clinical focus is to provide care, both diagnostic and therapeutic, to children with genetic disorders or suspected of having genetic or metabolic disorders.

At OUHSC, he is Director of the Specialty Clinic, and Program Director of the Medical Genetics Residency Program. He is also director of the 1st year Medical School course 'Molecular and Cellular Systems' for OU Medicine. Together with Dr. G. Bradley Schaefer, he is co-Director of the HRSA-funded 'Heartland Regional Genetics Collaborative.' He also serves as Medical Director of the Master's Program for Genetic Counseling.

His research interest focuses on the diagnostic process for individuals with rare Mendelian disorders, using information technology and novel technical genetic breakthroughs to aid in this process; and to empower the geneticist involved in patient care to access and utilize complex data, by making analysis of these data more accessible and user-friendly.

TOPIC: State of the Heartland Region

NOTES:

NAME:
Karl R. White, PhD
Director, National Center for Hearing Assessment
and Management
Emma Eccles Jones Endowed Chair in Early Childhood Education
Professor of Psychology
Utah State University
Karl.White@usu.edu

BIO: Dr. White holds faculty appointments as a Professor in the Department of Psychology and the Department of Communicative Disorders and Deaf Education at Utah State University and is the founding Director of the National Center for Hearing Assessment and Management (NCHAM). He has conducted dozens of competitively funded research projects and has published extensively about the issues and evidence related to implementing and improving the efficacy of Early Hearing Detection and Intervention (EHDI) programs. Projects currently underway at NCHAM are focused on developing more effective hearing screening and intervention programs through research, improving public health information systems, training and technical assistance, and information dissemination. www.infanthearing.org.

Prior to his work at NCHAM, he was the Director of Research and Development at the National Technical Institute for the Deaf, the Director of Research and Evaluation at the Center for Persons with Disabilities, and the Director of the Early Intervention Research Institute at Utah State University. Dr. White is nationally and internationally recognized as one of the world's leading authorities on early identification and treatment of hearing loss. His work has been recognized with awards from such diverse organizations as the Deafness Research Foundation, the American Association for Speech Language and Hearing, The Swedish Society of Medicine, and the Alexander Graham Bell Association for the Deaf and Hard of Hearing. He has hundreds of publications and presentations at scholarly meetings and has been an invited speaker to more than 35 countries, where he has assisted in the implementation of newborn hearing screening and intervention programs. He also serves on many national and international advisory groups for organizations such as the United States Department of Health and Human Services, the World Health Organization, March of Dimes, the American College of Medical Genetics, and the American Academy of Pediatrics.

TOPIC: Perspectives from EHDI

NOTES:

NAME:
Careema Yusuf, MPH
Senior Specialist, NewSTEPs,
Newborn Screening and Genetics
careema.yusuf@aphl.org

BIO: Careema Yusuf is the Senior Specialist for NewSTEPs at the Association of Public Health Laboratories (APHL). Her primary responsibilities are related to the Newborn Screening and Technical assistance and Evaluation Program's (NewSTEPs) data repository system and she serves as the primary representative for NewSTEPs on issues related to data coordination and using data for quality improvement activities. This includes providing technical assistance and support to state newborn screening (NBS) programs, report writing, and data analysis. She also serves as staff liaison for the APHL NBS Health Information Technology Workgroup and the APHL Hemoglobinopathy Laboratory Workgroup. She has a BSc in Biology and an MPH with a concentration in Epidemiology.

TOPIC: NewSTEPs Updates

NOTES:

NAME:
Sharon Romelczyk, MPA
Program Manager
National Genetics Education
and Consumer Network
Genetic Alliance
sromelczyk@geneticalliance.org

BIO: Sharon manages the National Genetics Education and Consumer Network (NGECN), a program focused on creating new partnerships and resources for consumers (individuals with genetic conditions and their families) to access quality genetic services. By working with consumers and consumer groups involved with genetics programs in their states and regional areas, she hopes to understand what gaps in access and knowledge there are and how Genetic Alliance can help address those gaps. Her passion is in improving access to health and healthcare for individuals with disabilities and special healthcare needs and believes that doing so involves authentic engagement of individuals and communities.

Most recently, Sharon served as a Senior Project Specialist for the Association of University Centers on Disabilities (AUCD), working closely with state-based programs funded by the National Center on Birth Defects and Developmental Disabilities (NCBDDD) at the Centers for Disease Control and Prevention (CDC) with the goal of improving health and preventing chronic diseases for people with a broad range of disabilities. She has demonstrated experience in leading national efforts to create collaborative products with state-based public health partners, national nonprofits, and government agencies. Sharon has a BA in Applied Sociology from Rowan University and an MPA with a concentration in Disability and Health Policy from the University of Delaware.

TOPIC: National Genetics Education and Consumer Network (NGECN) Updates

NOTES:

NAME:
Beth Kozel, MD, PhD
Assistant Professor of Pediatrics
Division of Genetics and Genomic Medicine
Washington University School of Medicine
Kozel_B@kids.wustl.edu

BIO: Dr. Kozel received her undergraduate degree at Washington University in St. Louis and did her MD-PhD training through the MSTP program at Washington University School of Medicine. After medical school, Dr. Kozel completed residencies in pediatrics and clinical genetics at St. Louis Children's Hospital. In January of 2009, Dr. Kozel initiated a post-doctoral project under the guidance of Dr. Robert Mecham and Dr. Jim Cheverud.

Currently, Dr. Kozel is an Assistant Professor of Medicine in the Department of Pediatrics, Division of Genetics and Genomic Medicine at Washington University School of Medicine and is the director of the Williams Syndrome Center at St. Louis Children's Hospital. Her research combines studies in mouse models and humans to identify genetic and environmental factors that modify the severity of elastin mediated vascular disease. She has identified several pathways that exacerbate or reduce the severity of vascular disease in this population, offering the potential to impact risk assessment and treatment.

TOPIC: Williams Syndrome Project

NOTES:

NAME:
Jeremy Penn, PhD
Director, Student Affairs Assessment
North Dakota State University
jeremy.penn@ndsu.edu

BIO: Jeremy Penn, Ph.D., is the Director of Student Affairs Assessment at North Dakota State University. In his current position he supports the University's efforts to assess, evaluate, and improve student wellness, success, and development programs. He earned a Ph.D. in Qualitative, Quantitative, and Psychometric Methods from the University of Nebraska-Lincoln in 2008.

Jeremy became involved in newborn screening as a parent advocate for SCID newborn screening and currently sits on the Education and Training subcommittee of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children. He is a member of the North Dakota Newborn Screening Advisory Board. He is a student in North Dakota State University's M.P.H. program and is interested in newborn screening and evaluation of public health programs.

TOPIC: Review of Best Practices in Documenting Newborn Screening Refusals for States

NOTES:

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 working in the division of the Maternal and Child Health Service at the Oklahoma State Department of Health (OSDH). 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 currently coordinates the Family Health History project and participates as an interviewer and research team member for the Hispanic Access project.

TOPIC: Hispanic Access Project

NOTES:

NAME:
Ann F. Chou, PhD, MPH, MA
Associate Professor
Health Administration and Policy,
College of Public Health
Family and Preventive Medicine,
College of Medicine
University of Oklahoma
ann-chou@ouhsc.edu

BIO: Ann F. Chou serves as Associate Professor in the Department of Health Administration and Policy at the College of Public Health and the Department of Family and Preventive Medicine at the College of Medicine, University of Oklahoma Health Sciences Center (OUHSC). 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.

TOPIC: Genetics Systems Assessment

NOTES:

NAME:
Christine Bruha, BS
Genetic Counseling Student
University of Arkansas for Medical Sciences
cbruha@uams.edu

BIO: Christine Bruha is a second year genetic counseling student at the University of Arkansas for Medical Sciences. She earned her B.S. in Biology from Florida State University in 2013. Upon graduation, she will work as a pediatric genetic counselor at the University of South Florida in Tampa. Christine has had a wide variety of clinical experiences ranging from the Victorian Clinical Genetics Service in Melbourne, Australia to St. Jude Children's Research Hospital in Memphis, TN. Her primary areas of interest are pediatric and cancer genetics. Christine is originally from Pensacola and her other interests include baking, reading, and all things Southern.

TOPIC: Role of the Geneticist in the Medical Home

NOTES:

NAME:
Lori Williamson Dean, MS, CGC
Department Chair and
Assistant Professor,
Department of Genetic Counseling
University of Arkansas for Medical Sciences
Program Manager,
Heartland Genetics Services Collaborative
LWDean@uams.edu

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. Public health genetics, teaching, and supporting individuals and families affected by genetic conditions are the hallmarks of her career. Lori has spent her life and career in the Heartland region. She was born and raised in the Ozarks of southern Missouri and spends weekends on her farm outside of Willow Springs, Missouri.

TOPIC: Heartland Telemedicine Update and Closing Remarks

NOTES:

NAME:
Julie Kaylor, MS, CGC
Laboratory Genetic Counselor
Molecular Genetics Pathology Laboratory
Arkansas Children's Hospital
kaylorja@archildrens.org

BIO: Julie Kaylor is a Clinical and Laboratory Genetic Counselor at the Molecular Genetics Pathology laboratory at Arkansas Children's Hospital. She works to improve the utilization of genetic testing and educate clinicians and the public regarding the use and potential of genetic testing. She serves on the Community Advisory Committee for Partners for Inclusive Communities of Arkansas and she has been an advocate and volunteer of 15 years for the Conway Human Development Center.

TOPIC: "What if I say something wrong?"

NOTES:

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

BIO: Dr. Wendy Parent-Johnson is Professor, 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 took this position approximately two years ago and in that role is responsible for administration and oversight of multiple grants, research, teaching, and supervision of 35 staff.

Her educational background includes a Masters Degree in Rehabilitation Counseling and a Ph.D. in Education with an emphasis on Transition for Individuals with severe Disabilities, both from Virginia Commonwealth University. She has thirty years experience in the areas of supported and customized employment and transition from school to work for individuals with severe disabilities.

Dr. Parent-Johnson has conducted presentations and published numerous book chapters, peer-reviewed journal articles, curricula, and informal written and electronic products related to transition and supported/customized employment and has co-authored several books on supported employment with a current one under contract with Paul Brookes Publishing Co. Most recently, her research and teaching has focused on health care transition, health disparities, and interdisciplinary health science education.

TOPIC: "What if I say something wrong?"

NOTES:

NAME:
Valerie A. Renault, MA
Communications Coordinator
Research and Training Center on Independent Living
The University of Kansas
vrenault@ku.edu

BIO: Val Renault is Communications Coordinator at the University of Kansas Research and Training Center (RTC) on Independent Living, which works to enhance the independence of people with disabilities. She brings experience with health care, advocacy and education to her work on disability research, issues and rights. At the RTC, she helped implement a research project to improve health care access for people with disabilities and produced a new edition of the center's internationally known guide to preferred language about people with disabilities.

TOPIC: "What if I say something wrong?"

NOTES:

NAME:
Mark A. Smith, MS
Resource and Family Support Coordinator
Nebraska Center on Disabilities
Munroe Meyer Institute for Genetics and Rehabilitation
msmitha@unmc.edu

BIO: Mark Smith is the Coordinator of the Individual and Family Resource Program at the Munroe-Meyer Institute, a University Center for Excellence in Developmental Disabilities at the University of Nebraska Medical Center. In this role, he also serves as the Family Faculty to the Munroe-Meyer Institute LEND program. For over 35 years, Mr. Smith has developed and implemented efforts and projects promoting opportunities for individuals with disabilities and their families to fully participate in their communities. He has served in a variety of roles, from coordinating the return of institutionalized individuals to their communities in Nebraska to working at the local, state, and national levels on the implementation of Early Intervention services for infants and toddlers with disabilities. He continues his efforts in this area as a member of the Executive Board of the Division of Early Childhood of the Council for Exceptional Children. As well, as the parent and sibling of individuals with disabilities, he was a founding member of the Early Intervention Family Alliance, a parent-led national organization focused on the need to insure the family voice be included in Early Intervention policy discussions at all levels.

Mr. Smith's areas of work and experience related to disability policy and practice vary widely, including national policy work on Genetic Medicine through the Genetic Alliance and Heartland Regional Genetic Collaborative, Mental Health through the National Association for Dual Diagnosis, Health Policy through the Maternal and Child Health Bureau, Individual and Family Support through the Association of University Centers on Disabilities (Mark currently serves on the AUCD Board), and Special Education policy and practice. Mr. Smith holds degrees from the University of Nebraska at Omaha and the University of Nebraska in Psychology. In 2011, Mr. Smith was a recipient of the Silver U Award for his work from the University of Nebraska Medical Center.

TOPIC: "What if I say something wrong?"

NOTES:

NAME:
Aaron Goldenberg, PhD, MPH
Assistant Professor of Bioethics
Assistant Director, Center for Genetic Research Ethics and Law
Case Western Reserve University
aaron.goldenberg@case.edu

BIO: Dr. Aaron Goldenberg is an Assistant Professor of Bioethics at Case Western Reserve University and the Associate Director of the Center for Genetic Research Ethics and Law at Case Western. Dr. Goldenberg has a background in bioethics and public health genetics.

Much of his work over the last 10 years has focused on the ethical, legal, and social implications of newborn screening, including projects related to the expansion of screening panels, the use of genomic technology in NBS programs, the education of parents about newborn screening, and the implications of using residual bloodspots for research purposes. He is currently the Co-PI of a HRSA funded project to explore the ethical and programmatic challenges of integrating genomics into Newborn Screening Programs.

TOPIC: Newborn Screening in the Genomics Era: Implications for State Programs

NOTES:

NAME:
Barbara Jackson, PhD
Munroe-Meyer Institute,
University of NE Medical Center
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: Fruits of our Labors and Beginnings and Endings

NOTES:

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

BIO: Anayeli (Ana) received her undergraduate degree at The University of Oklahoma in Norman, OK, 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, Ana 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: Beginnings and Endings

NOTES:

ADVOCATE WORK GROUP

DESCRIPTION

Everything that the RC 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 the Heartland RC 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.

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

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HEARTLAND GENETICS SERVICES COLLABORATIVE

AGENDA

ADVOCATE WORK GROUP MEETING
APRIL 29, 2015
2:00 – 4:00 PM
ROOM: LIBERTY
AIRPORT MARRIOTT
KANSAS CITY, MISSOURI

Agenda Items		
2:00 – 2:15 PM	Introductions	All
2:15 – 2:50 PM	Visioning Session: Direction of the Grant Led by Sharon Romelczyk	All
2:50 – 3:00 PM	**BREAK**	
3:00 – 3:20 PM	Work Group Membership & Participation (Please review the document from last WG Call) Recruitment Letter	All Michelle Black
3:20 – 3:30 PM	Reimbursement Policy	All
3:30 – 3:40 PM	Leader Advocates Experience at ACMG	Teresa Nold
3:40 – 3:50 PM	Genetic Alliance Meeting in DC	Mark Smith
3:50 – 4:00 PM	Hispanic Access Recruitment Experience	Mary Ann Coffman & Anayeli Herrera Morales

Facilitator: Jeanne Egger, Co-Chair
Note Taker: Dustin Foster

HEARTLAND GENETICS SERVICES COLLABORATIVE

NOTES

CLINICAL SERVICES WORK GROUP

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.

****This Work Group currently meets on the second Monday of every month at 3:00 – 4:00 PM CST.****

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HEARTLAND GENETICS SERVICES COLLABORATIVE

AGENDA

CLINICAL SERVICES WORK GROUP MEETING
MAY 1, 2015
AIRPORT MARRIOTT
KANSAS CITY, MISSOURI

Agenda Items	
1. Framework for a regional model for delivering genetics services	All
2. Updates and discussion about work for the next two years	Lori/Dr. Schaefer/Mary Ann/Wendy Parent-Johnson
▪ Hispanic Access ▪ Individualized Health Plan ▪ Healthcare Transition ▪ Family Health History ▪ Telemedicine ▪ Medical Home ▪ Primary Care Provider Education & Support	
3. Role of this group	All
4. Other	All

Facilitator: Lori / Dr. Schaefer

Note Taker: Lori / Mary Ann

HEARTLAND GENETICS SERVICES COLLABORATIVE

NOTES

EHDI Work Group

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.

This Work Group currently meets on the fourth Monday of every other month at 3:00 – 4:00 PM CST.

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HEARTLAND GENETICS SERVICES COLLABORATIVE

AGENDA

EHDI WORK GROUP
MAY 1, 2015
AIRPORT MARRIOTT
KANSAS CITY, MISSOURI

Agenda Items

Please join either the Clinical Services Work Group or Newborn Screening Work Group breakout sessions.

HEARTLAND GENETICS SERVICES COLLABORATIVE

NOTES

NBS WORK GROUP

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.

This Work Group currently meets on the fourth Wednesday of every month at 2:00 – 3:00 PM CST.

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HEARTLAND GENETICS SERVICES COLLABORATIVE

NBS WORK GROUP

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HEARTLAND GENETICS SERVICES COLLABORATIVE

AGENDA

NEWBORN SCREENING WORK GROUP MEETING
MAY 1, 2015
AIRPORT MARRIOTT
KANSAS CITY, MISSOURI

Agenda Items		
10:20 – 11:15 AM	SCID Update	All
11:15 – 11:25 AM	LSD Screening	Patrick Hopkins & Andrea Atherton
11:25 – 11:35 AM	Emergency Preparedness Update	Patrick Hopkins
11:35 – 12:05 PM	COIIN Update	Kim Piper & Carol Johnson
12:05 – 12:20 PM	Galactosemia Survey Results	Dr. Kahler & Jo Ann Bolick
12:20 – 12:30 PM	NewSTEPs Activities	Marci Sontag

Facilitator: Karen Eveans
Note Taker: Jo Ann Bolick

HEARTLAND GENETICS SERVICES COLLABORATIVE

NOTES

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HEARTLAND GENETICS SERVICES COLLABORATIVE

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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 4 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 Credentialing pending for Garden City, KS site	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 #2 being recruited. On-site training to occur late summer/early fall 2015 in AR and AZ.	Continue the project; Revise, as needed, after Cohort #1 complete; recruit Cohort #2	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.	Build on the IHP efforts from the Kansas Integrated Community Systems (D-70) grant.	Pilot implementation of the model and develop resource template Care Coordination Facilitator training scheduled May 7-8, 2015	Continue the project; pilot implementation of new strategies; parent-professional partnership workshop; activity hub; IHP education	None	Consultants: Wendy Parent-Johnson, Heather Smith, Charlotte Burt HRCC: Lori Dean; Clinical Services WG
	Obj. 3: Increase Heartland pediatric genetics providers' knowledge of and practice behaviors in healthcare transition for youth with genetic conditions.	Learning collaborative	Draft Transition Engagement Tool to be reviewed by LC Pilot curriculum for peds and FP residents; and GC trainees	Continue the project; pilot implementation of new strategies; qualitative study of adult care providers; complete products and disseminate	Explore adult care providers needs and perceptions of the role of the geneticist in transition and the transition process	Consultant: Wendy Parent-Johnson, Richard Parent-Johnson HRCC: Lori Williamson Dean;

HRSA PRIORITY AREA	HEARTLAND OBJECTIVES	OVERALL STRATEGIES	STATUS	YEAR 4 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 interviews Address issues raised from the interviews around education (patients, families, interpreters, and other public health providers)		None	HRCC: Klaas Wierenga, Mary Ann Coffman, Gene Hallford 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 submitted to GIM	Qualitative study of PCP, geneticists, and patient/family perceptions of the role of the geneticist in the medical home		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: Heartland Coordinating Center will update Heartland Collaborative members about national efforts in developing templates for management of patients with genetic	Shared survey data with NCC and national GPCI project coordinator.	Will use data to inform a PCP project around FHH and decision support tools.	Combine the effort to provide and promote decision support tools with the FHH project.	HRCC: Mary Ann Coffman; Clinical Services Work Group

HRSA PRIORITY AREA	HEARTLAND OBJECTIVES	OVERALL STRATEGIES	STATUS	YEAR 4 STRATEGIES	PROPOSED REVISION	RESPONSIBLE PARTY for PROJECT COMPLETION
		conditions. We are aware of ongoing efforts, as well as published printed and online resources. These resources and updates will be provided to Heartland members through work group meetings, at the annual meeting, and made available on the Heartland website				
		Strategy 3: Complete the "Heartland Collaborative Partners Program"	Second cohort completing their projects		None	HRCC: Anayeli Herrera Morales; NBS Work Group
		Strategy 4: Family Health History implementation of NCHPEG tool into Heartland clinical practices	Educational webinar to be offered May 2015 Planning group formed		Redesign project to include aspects of the AAP's GPCI project and the Heartland's clinical decision support project	HRCC: Mary Ann Coffman; Clinical Services Work Group
	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.	All-region workshop scheduled for April 2015		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		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 SACHDNC recommendations related to newborn and child screening and genetic follow-up and treatment services.	Strategy 1: Develop an implementation toolkit for Heartland states	Toolkit components are established. SCID toolkit completed. Pompe drafted.	Add disorders, as needed	None	HRCC: Karen Eavens; NBS Work Group
		Strategy 2: Monitor local, regional and national activities related to new conditions and SACHDNC recommendations to identify opportunities for Heartland states to participate in pilots, clinical practice guidelines,	Ongoing	Continue to send 1 to 2 state representatives to national work group meetings.	None	HRCC; State Genetics Coordinators; NBS Program Coordinators

HRSA PRIORITY AREA	HEARTLAND OBJECTIVES	OVERALL STRATEGIES	STATUS	YEAR 4 STRATEGIES	PROPOSED REVISION	RESPONSIBLE PARTY for PROJECT COMPLETION
		short and long-term follow-up of newborn screen identified patients. Once a SACHDNC 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.				
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	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.	Manuscripts in progress.			Consultants: Ann Chou HRCC: Klaas Wierenga and Lori Williamson Dean
		Strategy 2: Sustain GSA in the Heartland		Implement in Heartland in 2015-2016		
		Strategy 3: Plan and execute national rollout of the GSA tool.	NYMAC and MTN States implementation	Implement in other states/regions.		Consultant: Ann Chou; HRCC Team
	Obj. 2: Pilot Project Program	Tool for soliciting novel strategies to address specific problems	As needed	No plans for RFP in Year 4		

HRSA PRIORITY AREA	HEARTLAND OBJECTIVES	OVERALL STRATEGIES	STATUS	YEAR 4 STRATEGIES	PROPOSED REVISION	RESPONSIBLE PARTY for PROJECT COMPLETION
	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 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.	Report of 2014/2015 drill to be reported at Heartland annual meeting (2015)			Consultants: Stan Berberich and Patrick Hopkins HRCC: Lori Williamson Dean
	Obj 4: Facilitate communication and linkages of Heartland providers through a variety of methods.	Strategy 1: Heartland Annual Meeting	Spring 2015	None		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		Conduct NEGC surveys adapted for Heartland region and adapt policy brief		HRCC: Anayeli Herrera Morales

HEARTLAND GENETICS SERVICES COLLABORATIVE

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HEARTLAND GENETICS SERVICES COLLABORATIVE

ACRONYMS

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

Judith Benkendorf, MS, CGC
June 2011

Genetics Organizations

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

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

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..

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

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

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

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.

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.

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ACRONYMS

Genetics Education

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.

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.

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

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

Organized Medicine

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.

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.

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.

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

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

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

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ACRONYMS

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).

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).

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.

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.

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.

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ACRONYMS

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 Pediatrics (AAP): The ACMG of the pediatricians.

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

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

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.

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.

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.

HEARTLAND GENETICS SERVICES COLLABORATIVE

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.

HEARTLAND GENETICS SERVICES COLLABORATIVE

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.

GLOSSARY

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.

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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.

GLOSSARY

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.

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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.

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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).

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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.

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