

Where can
you see us
next?

2013 Joint Meeting of
the Newborn
Screening and Genetic
Testing Symposium
and International
Society for Neonatal
Screening
May 5-6, 2013
Atlanta, GA

MARK YOUR CALENDARS!

NBSTRN Clinical
Centers Executive
Committee Meeting
April 4-5, 2013
Washington, DC

NBSTRN Network
Meeting
April 8-9, 2013
Washington, DC

NBSTRN Standing
Committee Meeting
April 7th & 9th, 2013
Washington, DC

NBSTRN NEWSLETTER



N B S T R N
NEWBORN
SCREENING
TRANSLATIONAL
RESEARCH
NETWORK

April 2013

Return of Results (ROR)

The NBSTRN website has a new resource! Developed by the NBSTRN Bioethics and Legal Issues Workgroup, "Points to Consider for Research Conducted Using NBSTRN Resources" is intended to address research conducted using the Virtual Repository of Dried Blood Spots (VRDBS), Longitudinal Pediatric Data Resource (LPDR), or other NBSTRN resources. Prior to beginning a research project, researchers should consider what to do if their work finds information of possible clinical utility to an individual or population in the study. When applicable, researchers should have an IRB approved plan to manage ROR.

Frequently, research in this area may be conducted with anonymous or aggregate de-identified data without specific informed consent. Thus, in many cases, it may not be possible for the researcher to return individual results to research participants. Even in research using anonymous or de-identified data; however, there may be rare cases where results have clear clinical significance for individuals or a population, and where return of results (ROR) would be beneficial for the individual or the population. If de-identified data have been used, and if the state has retained individual identifiers, the state may be able to act as an "Honest Broker," to re-identify individuals. Alternatively, when anonymous data have been used, the State may be able to identify a specific population that may benefit from knowledge of the research results. Researchers should have a plan, in cooperation with the state as Honest Broker and the appropriate IRB, to consider whether, when, and how results ought to be returned. If return of results is part of the plan; the rules, roles, and responsibilities of all parties involved should be clearly defined in the research protocol.

Whether and how results *should* be returned to participants varies greatly depending on the research methods (e.g. population data), the information obtained (e.g. how likely to impact an individual's health), whether the samples are identifiable, and the agreements made in any consent for a specific protocol. The website has a table which provides a framework for approaching ROR in different types of research.

Go to NBSTRN.org to read more!!!!

Spotlight on NBS Researcher

This month's researcher of the month is a pediatrician by training, a professor of Laboratory Medicine since 1998 and the T. Denny Sanford Professor of Pediatrics at the Mayo Clinic College of Medicine in Rochester, MN. He currently serves as co-director of the Biochemical Genetics Laboratory and as vice-chair of Information Management in the Department of Laboratory Medicine & Pathology. He holds joint appointments in the Department of Pediatrics & Adolescent Medicine and in the Department of Medical Genetics. His clinical interests include inborn errors of metabolism, particularly mitochondrial fatty acid oxidation disorders, newborn screening, and metabolic disorders misdiagnosed either as child abuse or sudden and unexpected death. This researcher has focused his past research on clinical and biochemical characterization of newly discovered metabolic disorders, as well as clinical applications of tandem mass spectrometry. His current research interest is the clinical implementation of multivariate pattern recognition software that improves the interpretation of complex profiles of laboratory results. The goal of this work is to integrate all clinically significant results available to diagnose a particular condition in a single score in a manner that is objective, evidence-based, and open to worldwide collaboration and data sharing.

Don't know who we are talking about? Visit <https://www.nbstrn.org/about/spotlight> for the full article!

Experiencing Genetic Counseling

Are you looking for an innovative, experiential approach to teaching medical genetics concepts? Order the NEWLY updated interactive resource available from ACMG!

Experience the Future: Discover the Power of Genetics is a dynamic interactive tool originally developed for The Endocrine Society's 2002 Annual Meeting. Revised in 2012, this program has proven to be a successful way to introduce health professionals and students to the experience of discussing genetic information and receiving genetic test results through live, simulated counseling encounters with experienced genetic counselors.

Each of the seven disease-specific modules (colon cancer, cystic fibrosis, fragile X syndrome, hereditary breast-ovarian cancer, hemochromatosis, multiple endocrine neoplasia type II and sickle cell disease) provides both faculty and participants with all the necessary program materials. Participants are assigned a specific case scenario, followed by a personal genetic counseling experience. Each module includes a curriculum for the delivery and discussion of genetic testing results in a face-to-face setting with a trained genetic counselor, working with a medical geneticist. This program offers participants the opportunity to personally experience the complexity and nuances of interactions with families and patients receiving genetic testing information. Save trees, all program materials come on an easy to use flash-drive!



If you have topics for inclusion in future newsletters, please let us know by emailing ismotrich@acmg.net



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