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The state of public health genetics in Rhode Island. H.F.L. Mark[1,2,3,4] R.Caldarone[1], A. Zimmerman[1], S.Viner-Brown[1], P. Simon[1], A. Colt[3], W. Hollinshead[1] and P. Nolan[1]. [1]Rhode Island Department of Health, [2]KRAM Corporation, [3]Rhode Island Public Health Foundation, [4]Brown University School of Medicine, all in Providence, Rhode Island.

Rhode Island is a small state, with a population of approximately one million; 11% live below the poverty line; 10% are uninsured; 10% are minorities; and 13% have no regular source of medical care. There are approximately 77,000 children under the age of 6 years; 20% live in poverty. As reported by vital records, the state has approximately 13,000 births per year; 3.2% have congenital anomalies; less than 1% have inborn errors of metabolism or hemoglobinopathies; but 41% are deemed at developmental risk. There are a total of 1,900 children in the RI Department of Health's early intervention program. One of the departmental goals is to ensure that all children with special health care needs receive diagnostic, therapeutic, and habilitative services on a timely basis. Towards this end, the department has established a new initiative to integrate genetics in public health, utilizing both existing and new sources of funding (from the Maternal and Child Health Bureau of Healthcare Resources and Services Administration). A novel information infrastructure will be implemented to identify children with such needs; identify health resources and services in use; generate aggregate data for surveillance, needs assessment, planning, and evaluation; generate profiles for tracking and followup services; and provide patient information to pediatric and specialty physicians. All of the above will be achieved through a universal, longitudinal, child health information database (KIDSNET), which is linked to 18% of pediatric medical-care sites in the state, and serves more than half of the children in the system. A revised state genetics plan is currently being written to incorporate all relevant elements, with special emphasis on genetics education for consumers, nongeneticist healthcare professionals and members of the legislature.

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An assessment of genetic knowledge and utilization among mental health care providers and consumers. Petty, EM,[1,2] Madeo, AC,[1,2] Smith, LB,[1,2] Milner, KK.[3] Depts. of Internal Medicine,[1] Human Genetics,[2] and Psychiatry,[3] Univ. of Michigan, Ann Arbor, Michigan.

Advances in genetics are ushering in a new era of molecular medicine that will impact all fields of medicine including psychiatry. Identification of genetic alterations associated with mental illness should facilitate development of innovative strategies for improved clinical care of affected and at-risk individuals through use of molecular diagnostics, genetic risk assessments, and pharmacogenetic agents. While these applications may ultimately improve disease-related morbidity, they could have negative consequences if they are inappropriately applied or interpreted. The risks of potential negative consequences will increase if mental health care providers and consumers are not well informed about the appropriate uses, benefits, risks, and limitations of emerging genetic information and technology. A recent survey of psychiatrists and Alliance of the Mentally Ill members regarding prenatal genetic testing revealed that both groups supported the development of prenatal testing for diseases with psychiatric manifestations (Milner et al, 1999). Surprisingly, their support for prenatal testing of adult-onset disorders, including Huntington disease, was in contrast to current practices by most clinical geneticists. We subsequently hypothesized that mental health professionals and consumers may have misconceptions about genetics due to a lack of exposure to up-to-date and accurate genetic information, and, that such misconceptions may negatively impact upon the availability of appropriate genetic counseling for individuals with, or at risk for, psychiatric disorders. To begin addressing these hypotheses we developed a survey to assess genetic knowledge, awareness of genetic services, and utilization of genetic resources. It was sent to over 900 mental health care providers. Analysis of 200 completed surveys reveals that most respondents understand some basic genetic principles. However, diverse opinions regarding genetic contributions to mental illness were noted. Few providers routinely refer clients with family histories of mental illness for genetic counseling. A similar survey for consumers is currently being administered. Results from provider and consumer surveys will be further analyzed to help determine what educational strategies and clinical services will be most helpful in providing genetic resources to mental health professionals and clients.

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MCHB *SPRANS* Projects: Collaborative efforts to guide primary care providers to access and use medical genetic information electronically. V. Proud[1], K. Silvey[2], C. Barash[3], B. Pletcher[4] and N. Hanson[5]. [1]Children's Hospital Eastern Va Med S Norfolk VA, [2]Ore. Health Sci. Univ. Eugene OR, [3]Boston MA, [4]Med. College NJ Newark, and [5]Children's Hosp. Med. Cen., Seattle WA.

Information from the Human Genome Project is changing the way primary care providers (PCPs) think about and use genetics information in clinical practice. The Maternal and Child Health Bureau Genetic Services Branch made the education of PCPs a priority in order to improve effective utilization of genetic services. Projects identified needs and developed products to accurately present the changing practice of medical genetics in a way that is useful to physicians and nurses. Surveys, focus groups, and hundreds of interviews during these projects identified significant gaps in knowledge about basic concepts necessary to make appropriate diagnosis and management decisions. PCPs often hesitate to use genetics information because of the belief that having a genetic diagnosis is "bad" and should be avoided at all costs, the perception of limited treatment options, and prejudices complicated by the media hype about inappropriate use of genetic information.

Educational programs for PCPs evolved from simple one-on-one presentations to the creation of comprehensive information resources for use in daily practice. Electronic products developed through these projects include software, Internet sites and a WebManager. The internationally known Internet based GeneTests[TM] and GeneClinics[TM] present information on Molecular laboratories doing testing and peer reviewed disease specific profiles. Genes in Your Practice© is a CD-ROM information package that presents the basics of Human and Medical Genetics. GenesAtWork© is an Internet site that highlights information for the primary care provider including frequent newsletters on practical genetic issues. INFOGENETICS© includes a CD-ROM WebManager and Internet site that focus the clinician's search for a diagnosis on OMIM© and the Birth Defects Encyclopedia© to generate a genetics differential about a specific patient problem. Genetics professionals and PCP's together with managed care administrators and a strong consumer community must continue to work together to develop tools to make the best possible information available to all electronically.

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Ethical Issues Encountered in Establishment of the Texas Birth Defect Research Center (TBDRC) A Scheuerle and D Wright, Texas Birth Defects Research Center, Dallas, Texas.

The Texas Birth Defect Research Center (TBDRC) is funded by the Centers for Disease Control and Prevention and administered by the Birth Defects Monitoring Division of the Texas Department of Health. Texas is one of eight states in the National Birth Defects Prevention Study. As part of the Study, Texas contributes cases and controls to a composite database, performs interviews with mothers, and collects biological specimens. The cases are identified from the existing Texas Birth Defects Registry (TBDR) and use of these data in an interstate study required review by Institutional Review Boards at the state and federal level, as well as coordination with the IRBs of other states.

The TBDR is a legislated surveillance system and it is not intrinsically a research entity. Because the mission of the TBDR includes social services referral and birth defects prevention, the information collected is not anonymous. Confidentiality protocols have been established for use of the Registry data by researchers, but organization of the NBDPS required a more formal review of human subjects protections. The ethical questions addressed by Texas in coordinating with the NBDPS fall into 10 categories:

1) Informed consent; 2) Sensitivity to ethnic/cultural minorities; 3) Participation of minors in research; 4) Patient/parent confidentiality; 5) Patient/parent right to refuse; 6) Patient/parent right to withdraw; 7) Patient/parent privacy right vs public health need for data; 8) Compensation for participation; 9) Use of electronically transmitted data, and; 10) What to do when Texas state law differs from that of other states.

In organization of the TBDRC, the two largest issues for Texas have been participation of minor parents who are not emancipated and translation of the questionnaire into a culturally sensitive version of spanish. As the study has progressed, the most difficulty has been balancing the need for subject participation with a non-coercive, non-harassing contact process.