ANNUAL REPORT
2006-2007

RESOURCE FOR
GENETIC & EPIDEMIOLOGIC RESEARCH
(RGE)

Jahn Barlow
Director

July 1, 2006 - June 30, 2007

EXECUTIVE SUMMARY

2006-2007 was a year of noteworthy change and growth for RGE. After nearly two decades of service as the RGE Director, Ms. Jean Wylie resigned from her position at the University of Utah in January 2007. Mr. Jahn Barlow was hired as the new Director in February. In May, Dr. Geri Mineau, Research Professor in the Department of Oncological Sciences, and Dr. Barry Nangle, Director of the Utah Center for Health Data in the Utah Department of Health, were elected Chair and Vice Chair respectively of the RGE Review Committee. In June 2007, the RGE Review Committee was expanded with the addition of three new faculty members from the School of Medicine.

The Utah Population Database (UPDB) continued to grow and expand as a research resource during 2006-2007. Two new agreements were completed in 2007 that will significantly expand the UPDB infrastructure. In April 2007, the Master Linkage File Research Agreement was finalized between the University and Intermountain Healthcare, which authorizes the linking of Intermountain's Enterprise Data Warehouse to the UPDB to facilitate joint research projects between the two institutions. As part of the agreement, Intermountain becomes a data contributor to RGE and will designate a representative to the RGE Review Committee. In May 2007, the University and the Utah Department of Health (UDOH) signed a three year RGE agreement to incorporate the UDOH inpatient hospital discharge, ambulatory surgery, and emergency department encounter data sets into the UPDB.

The net number of projects using UPDB increased from 64 to 77, including an increase in the net number of projects using the UUHSC Enterprise Data Warehouse and UPDB from 17 to 23.

Organizational Activity and Support

Vice President for Research, University of Utah

RGE continues to report to Dr. Raymond Gesteland, Vice President for Research, through Dr. Jeffrey Botkin, Associate Vice President for Research Integrity. Additionally, Dr. Gesteland's office provides partial support of the Pedigree and Population Resource at Huntsman Cancer Institute for the maintenance and operation of the UPDB.

Senior Vice President for Health Sciences, University of Utah

The Senior Vice President for Health Sciences, Dr. Loris Betz, continues to provide substantial support for the operating costs of RGE through the Dean's Office of the School of Medicine. Dr. Betz' office also contributes to the annual maintenance and operational support of the UPDB. Additionally, in February 2007, Dr. Betz provided funding to the Information Technology Services Data Resource Center and the Pedigree and Population Resource at Huntsman Cancer Institute to update the links between the UUHSC Enterprise Data Warehouse and the UPDB as infrastructure support for a growing number of research projects using combined data from the two resources.

Huntsman Cancer Institute at the University of Utah

Pedigree and Population Resource

Huntsman Cancer Institute (HCI) continues to provide significant financial support for the Pedigree and Population Resource (PPR) as a core resource of HCI. Under the leadership of Dr. Geri Mineau, PPR manages the data within the UPDB, provides consultation to present and future users of the data, and adds to and improves the data. Additionally, HCI underwrites the computer hardware, security, backup and database management costs of the UPDB through the Computing and Technology Group, as well as software development and maintenance through the Informatics Core Resource.

High Risk Cancer Clinics

RGE continues to provide external review, through the RGE Review Committee, of all proposals to use data resources from the HCI High Risk Cancer Clinics.

RGE Data Contributors

Cancer Data Registry of Idaho (CDRI)
www.idcancer.org

A data contributor since 1999, the CDRI provides cancer diagnosis data for the State of Idaho from 1969 through 2004. While only 10% of individuals statewide link to UPDB, records from the counties in southeastern Idaho link at rates of 20% or more.

Ms. Stacey Carson, Vice President of Operations and Registry Services, serves as the CDRI contact for RGE. All requests to use CDRI data are reviewed by the Cancer Analysis Working Group prior to access.

Data Resource Center, University of Utah Health Sciences Center (DRC)
http://uuhsc.utah.edu/drc

Beginning in 2003, demographic records stored in the UUHSC Enterprise Data Warehouse (EDW) for more than 1.7 million patients at the University Hospitals and Clinics have been linked to records in the UPDB. No patient information or medical records are incorporated into the UPDB; only a link between the records is maintained. The data are combined only for limited, project specific access to both sets of data when a project is approved by RGE.

Ms. Cheri Hunter is the representative to the RGE Review Committee from the UUHSC Data Resource Center.

Driver License Division, Utah Department of Public Safety (DLD)
http://driverlicense.utah.gov/

A data contributor since 1999, the Driver License Division provides records that facilitate follow-up information for individuals living in Utah, as well as height and weight data, residential histories, and migration data. Recent projects have also used the data to identify and recruit population-based controls for research. Nearly 6 million current and previous address records from the DLD have been assigned a geo-code by the University DIGIT Lab using the Universal Transverse Mercator system. Approximately 70% of the individuals on DLD records link to one or more other records in the UPDB.

Mr. Randy Campbell, Records Officer, serves as the reviewer for requests to access DLD data from the UPDB.

The Church of Jesus Christ of Latter-day Saints
www.familysearch.org

In the 1970s, more than 185,000 families were identified from Family Group Sheets in the archives of the Utah Genealogical Society. These records were computerized to create electronic pedigree structures that were linked across generations producing the large multigenerational families available in the UDPB. More than 1.6 million individuals born between the late 1700s and the mid 1900s are included in these families descending from the Utah pioneers. Currently, the Ancestral File is available to update or add new genealogy information to the UPDB as needed.

Mr. Glen Buckner serves as the reviewer for requests to access the data from the genealogy records in UPDB.

Utah Cancer Registry (UCR)
http://ucr.utah.edu

A data contributor since the 1970s, UCR is one of the original members of National Cancer Institute Surveillance, Epidemiology, and End Results (SEER) program. UCR provides statewide cancer records for Utah from 1966 through 2004 through annual updates to the UPDB.

Dr. Antoinette (Nan) Stroup, Deputy Director, is the UCR representative to RGE.

Utah Department of Health (UDOH)
http://health.utah.gov

A data contributor since 1977, the Department of Health continues to contribute valuable data sets to the UPDB, including annual updates of birth certificates, death certificates, fetal death records, marriage and divorce records. These records link to other records in UPDB at very high rates. They provide valuable demographic and epidemiologic data, such as fertility, longevity, residential histories, birth complications, and causes of death. Family information from these records is also linked to the existing genealogy records expanding the pedigrees of the Utah pioneer families into current generations. Birth and marriage records also help to "reconstitute" smaller 3 generation families which are not connected to the existing genealogy records.

Dr. Barry Nangle, Director of the Utah Center for Health Data, which includes the Office of Vital Records and Statistics, serves as the UDOH representative to RGE.

UPDB: DEVELOPMENT, MANAGEMENT, AND IMPROVEMENT

UPDB continues to receive annual updates from our data contributors for births, marriages, divorces, deaths, cancer records, and driver licenses in Utah. A copy of the Ancestral File from the Church of Jesus Christ of Latter-day Saints is available to UPDB, and genealogies can be added or updated as needed. There are more than 9 million records in the UPDB. Multiple records for an individual are matched to create a "person-oriented record." From more than 15 million individuals named in these records, more than 6.5 million unique individuals have been created.

Record Type	New Records	Total Records
Genealogy	1,281	1,602,035
Birth Certificates (1915-21, 1947-2005)	79,650	2,159,379
Marriage Certificates (1978-2005)	24,749	569,556
Divorce Records (1978-2005)	10,003	249,189
Death Certificates (1904-2005)	13,750	708,118
Fetal Deaths	1,713	7,508
Social Security Death Index	173	479,437
Utah Cancer Records	9,747	226,014
Idaho Cancer Records	6,687	123,737
Driver License Division	88,012	2,863,257
1880 Territorial Census	-	142,711
Project specific records, incorporated into UPDB	10,589	71,085
Obituary records (began fall 2006)	1,442	1,442
Total Records	**247,796**	**9,203,468**

These longitudinal records capture numerous data points from many of the key events associated with an individual over a lifespan, facilitating a wide range of topics for genetic and epidemiologic research. Efforts continue to incorporate new data sets into the UPDB, as well as linking to external data sets that can be combined with UPDB data for specific research projects approved by RGE. Progress in 2007 includes new agreements with the UDOH and Intermountain Healthcare and improvements in the existing links with the UUHSC Data Resource Center.

Utah Department of Health Hospital Data

In April 2007, researchers from Huntsman Cancer Institute (HCI) and the Utah Department of Health (UDOH) were awarded a grant (R01 RR02746) by the NIH National Center for Research Resources to integrate two extremely large data repositories. In May 2007, the University and UDOH signed an amendment to the existing RGE agreement authorizing the incorporation of the UDOH health facility administrative data into the UPDB. Under this joint project, *Sharing Statewide Health Data for Genetic Research,* claims data (ICD9 codes) from statewide administrative databases will be merged and linked into UPDB. The claims data covers a period from 1996 to present and includes more than 2 million records from Hospital Inpatient Discharge, approximately 2 million records from Ambulatory Surgery, and more than 5 million encounters with the Emergency Department.

Huntsman –Intermountain Cancer Care Program

The Huntsman-Intermountain Cancer Care Program (HICCP) Database Access and Research Subcommittee continues its work on a project to link the UPDB and the Intermountain Enterprise Data Warehouse (IEDW). Linkage of IEDW and UPDB will expand data capture to statewide levels and allow a vast new store of medical information to be linked to genealogy. Investigators will be able to query diagnostic information and identify disorders that cluster in families and potential subjects for research projects. The Master Linkage File Research Agreement was finalized between the University and Intermountain Healthcare in April, 2007. Approximately 5.3 million individuals in the IEDW will be linked to "person" records in UPDB; an anonymous Master Linkage File (MLF) will be created.

Data Resource Center, University of Utah Health Sciences Center

With funding provided by Dr. Betz, Senior Vice President for Health Sciences, PPR and the UUHSC Data Resource Center (DRC) updated the links between the UPDB and the Enterprise Data Warehouse (EDW) in April 2007. The Pedigree and Population Resource (PPR) at Huntsman Cancer Institute received over 1.8 million demographic records for patients in the EDW from the DRC, including 223,084 new patient records. PPR linked the new patients and attempted to increase the linkage of previous patients. This resulted in 74.3% of the patients linking to one or more UPDB records. Because many researchers are interested in conducting family studies, PPR analyzed the pedigree quality of the linked records and find that:

25.7% are not linked
16.4% have no familial relationships
6.5% have parent-child only relationships
11.5% have two generation families
39.9% have multi-generational pedigrees with three or more generations.

When **restricted to Utah residents**, PPR links 83% of residents to other UPDB record(s):

17.0% do not link
17.3% have no familial relationships
6.8% have parent-child only relationships, 1 or 2 parents with a single child
12.8% have two generation families with 4 or more family members
46.1% have multi-generational pedigrees with three or more generations, some in families with as many as 11 generations. There are 613,595 in this group and 569,337 (92.8%) are living.

The use of these links by research projects continues to expand, with 23 projects (nearly 1/3 of UPDB projects) requesting access to these data during 2007.

UPDB Operations and Management Group

This group meets weekly and is responsible for implementing the operation of UPDB, including computer equipment, security, backups, database design and data management. It includes computer professionals and programmers from Huntsman Cancer Institute's Pedigree and Population Resource (PPR), Computer and Technology Group (CATG), and Informatics Core Resource as well as the PPR and RGE Directors. The UPDB uses a relational database management system, Microsoft SQL Server 2005. This group provides the administration of UPDB, including ongoing database performance and tuning, index maintenance, management of security environment which involves software updates, user access, and monitoring network access. When new data are added to UPDB, this group modifies the database design, develops applications, writes triggers and loads the data.

Pedigree and Population Resource
Geri Mineau, Director
Alison Fraser, Database Manager
Carole Schaefer, Software Engineer
Richard Pimentel, Software Engineer
Andrew Hammer, Programmer/Developer

Computer and Technology Group
Dinny Berry, Database Administrator

Informatics Core Resource
Samir Courdy, Director
Cindy Spigle, Sr. Software Engineer

RGE Review Committee

In August 2006, Dr. Randall Burt replaced Dr. Mary Beckerle as the HCI representative to the RGE Review Committee. Dr. Christopher Sans, Business Development Manager, joined the RGE Review Committee in November 2006 as an ex-officio member representing LineaGen Research Corporation located in the University Technology Commercialization Office. Mr. Jahn Barlow replaced Ms. Jean Wylie as the RGE Director and an ex-officio member of the Committee in February 2007. Dr. Geri Mineau and Dr. Barry Nangle were elected Chair and Vice Chair respectively of the RGE Review Committee in May 2007. Three new faculty members from the School of Medicine were nominated and approved to serve on the Committee in May 2007: Dr. Stephen Guthery, Dr. Sampath Prahalad and Dr. Allen Sawitzke.

Committee Members as of June 30, 2007

Randall Burt, M.D.
Senior Director of Prevention and Outreach
Huntsman Cancer Institute

Stephen Guthery, M.D., M.Sc.
Department of Pediatrics

Cheri Hunter
Data Resource Center
UUHSC Information Technology Service

Lynn Jorde, Ph.D.
Eccles Institute of Human Genetics

Geri Mineau, Ph.D., Chair
Director, Pedigree and Population Resource
Huntsman Cancer Institute

Barry Nangle , Ph.D., Vice Chair
Director, Utah Health Data Center
Utah Department of Health

Sampath Prahalad, M.D., M.Sc.
Department of Pediatrics

Allen Sawitzke, M.D.
Department of Internal Medicine

Antoinette (Nan) Stroup, Ph.D.
Deputy Director, Utah Cancer Registry

George L. White, Ph.D., M.S.P.H.
Department of Family and Preventive Medicine

By mail
Glen Buckner
The Church of Jesus Christ of Latter-day Saints

Randy Campbell
Records Officer, Driver License Division
Utah Department of Public Safety

Stacey Carson, RHIT, CTR
Vice President, Operations and Registry Services
Cancer Data Registry of Idaho

Ex officio
Jahn Barlow, M.P.A.
Director, RGE

Christopher Sans, Ph.D.
Business Development Manager
LineaGen Research Corporation
Technology Commercialization Office

RESEARCH PROJECTS APPROVED TO USE UPDB

Continuing Projects:

1. An Influence Diagram for the Management of Community-acquired Pneumonia

   Principal Investigator: Dominik Aronsky, M.D., Ph.D.
   Vanderbilt University
   Department of Biomedical Informatics
   Data used: death certificates

2. A Study of Cancer Rates and TCE Ground Water Contamination

   Principal Investigator: Wayne Ball, Ph.D.
   Utah Department of Health
   Data used: genealogy, UCR, DLD

3. Molecular Analysis of FFPE Cancer Tissue

   Principal Investigator: Philip Bernard, M.D.
   Department of Pathology
   Data used: genealogy, death certificates, UCR, DRC

4. Development of a Registry for Familial Pancreatic Cancer

   Principal Investigator: Randall Burt, M.D.
   Department of Internal Medicine, Gastroenterology Division
   Data used: genealogy, death certificates, UCR, CDRI, DLD

5. High Risk Familial Colon Cancer: Genotype and Phenotype

   Principal Investigator: Randall Burt, M.D.
   Department of Internal Medicine, Gastroenterology Division
   Data used: genealogy, death certificates, UCR, CDRI, DRC, DLD

6. High Risk Breast Cancer Clinic

   Principal Investigator: Saundra Buys, M.D.
   Department of Internal Medicine, Oncology Division
   Data used: genealogy, death certificates, UCR, CDRI, DLD

7. Genetic Analysis Techniques for Common Cancers

Principal Investigator: Nicola Camp, Ph.D.
Department of Biomedical Informatics, Division of Genetic Epidemiology
Data used: genealogy, death certificates, UCR, DLD

8. Analysis of Familiality by Phenotype

Principal Investigator: Lisa Cannon-Albright, Ph.D.
Department of Biomedical Informatics, Division of Genetic Epidemiology
Data used: genealogy, death and birth certificates, UCR, CDRI, DRC, DLD

9. Familial Analysis of Pelvic Organ Prolapse (POP) and Urinary Incontinence

Principal Investigator: Lisa Cannon-Albright, Ph.D.
Department of Biomedical Informatics, Division of Genetic Epidemiology
Data used: genealogy, birth certificates, death certificates, UCR, DRC, DLD

10. Familiality Investigation of Chronic Pain and Treatment Responses

Principal Investigator: Lisa Cannon-Albright, Ph.D.
Department of Biomedical Informatics, Division of Genetic Epidemiology
Data used: genealogy, DRC, DLD

11. Familiality Investigation of Clinical Characteristics of Prostate Cancer

Principal Investigator: Lisa Cannon-Albright, Ph.D.
Department of Biomedical Informatics, Division of Genetic Epidemiology
Data used: genealogy

12. Familiality Student Activities

Principal Investigator: Lisa Cannon-Albright, Ph.D.
Department of Biomedical Informatics, Division of Genetic Epidemiology
Data used: genealogy, death certificates, UCR, DLD

13. Fine Mapping and Isolation of the Familial Wilm's Tumor Gene FWT1

Principal Investigator: Lisa Cannon-Albright, Ph.D.
Department of Biomedical Informatics, Division of Genetic Epidemiology
Data used: genealogy, death certificates, UCR, DLD

14. Genetic Analysis of Colorectal Cancer

Principal Investigator: Lisa Cannon-Albright, Ph.D.
Department of Biomedical Informatics, Division of Genetic Epidemiology
Data used: genealogy, death certificates, UCR, DLD

15. Genetic Epidemiology of Prostate Cancer

Principal Investigator: Lisa Cannon-Albright, Ph.D.
Department of Biomedical Informatics, Division of Genetic Epidemiology
Data used: genealogy, death certificates, UCR, DLD

16. Genetics of Breast Cancer

Principal Investigator: Lisa Cannon-Albright, Ph.D.
Department of Biomedical Informatics, Division of Genetic Epidemiology
Data used: genealogy, death certificates, UCR, DLD

17. Genetics of Familial Aneurysms

Principal Investigator: Lisa Cannon-Albright, Ph.D.
Department of Biomedical Informatics, Division of Genetic Epidemiology
Data used: genealogy, death certificates, DRC, DLD

18. Identification of Chronic Lymphocytic Leukemia (CLL) Predisposition Loci

Principal Investigator: Lisa Cannon-Albright, Ph.D.
Department of Biomedical Informatics, Division of Genetic Epidemiology
Data used: genealogy, death certificates, UCR, DLD

19. Identification of Melanoma Predisposition Loci

Principal Investigator: Lisa Cannon-Albright, Ph.D.
Department of Biomedical Informatics, Division of Genetic Epidemiology
Data used: genealogy, death certificates, UCR, DLD

20. Inherited Predisposition to Diabetes

Principal Investigator: Lisa Cannon-Albright, Ph.D.
Department of Biomedical Informatics, Division of Genetic Epidemiology
Data used: genealogy, DLD, UCR

21. Using Record Matching Techniques to Identify Duplicate Person Records

Principal Investigator: Scott DuVall
Department of Biomedical Informatics
Data used: all

22. Linkage of Utah Birth Defects Network surveillance data with UPDB

Principal Investigator: Marcia Feldkamp, M.S.P.H.
Utah Department of Health
Data used: genealogy, birth and death certificates, fetal death, UCR, DRC, DLD

23. Co-Ancestry in Primary Pulmonary Hypertension

Principal Investigator: C. Gregory Elliott, M.D.
Intermountain Healthcare
Department of Internal Medicine, Division of Pulmonary Medicine
Data used: genealogy, death certificates

24. Spatial Variation in Body Mass Index

Principal Investigator: Jessie Fan, Ph.D.
Department of Family and Consumer Studies
Data used: genealogy, birth certificates, DLD

25. Role of Clonal Lymphocytosis of Unknown Significance (CLUS) in the Multi-Step Pathogenesis of Familial Chronic Lymphocytic Leukemia (CLL)

Principal Investigator: Martha Glenn, M.D.
Department of Internal Medicine, Oncology Division
Data used: genealogy, UCR, CDRI

26. Role of Family History in Cardiovascular Mortality of Hemodialysis Patients

Principal Investigator: Alexander Goldfarb-Rumyantzev, M.D., Ph.D.
Department of Internal Medicine, Nephrology Division
Data used: genealogy, death certificates, DLD

27. Chronic Respiratory Failure in Children

Principal Investigator: Melissa Gowans, M.D.
Department of Pediatrics
Data used: death certificates

28. Inflammatory bowel disease in Utah: Using the Utah Population Database to assess familial risk

Principal Investigator: Stephen Guthery, M.D., M.Sc.
Department of Pediatrics, Division of Pediatric Gastroenterology
Data used: genealogy, birth and death certificates, DRC

29. Familial kinship analysis in patients with Mullerian anomalies

Principal Investigator: Ahmad Hammoud, M.D.
Department of Obstetrics and Gynecology
Data used: genealogy, birth certificates

30. Origins of Higher Order Multiple Pregnancies

Principal Investigator: Ivan Huang, M.D.
Department of Obstetrics and Gynecology
Data used: birth certificates

31. Effectiveness of Utah's Shaken Baby Prevention Program

Principal Investigator: Heather Keenan, M.D.
Department of Pediatrics, Critical Care
Data used: birth certificates

32. Familial Aggregation and Coaggregation of Cancer

Principal Investigator: Richard Kerber, Ph.D.
Department of Oncological Sciences/Huntsman Cancer Institute
Data used: genealogy, birth and death certificates, UCR, DLD

33. The Cache County Family-Based Cohort Study on Aging

Principal Investigator: Richard Kerber, Ph.D.
Department of Oncological Sciences/Huntsman Cancer Institute
Data used: genealogy, birth and death certificates, DLD

34. Genetic Characterization of Cancer Risk in Families

Principal Investigator: Richard Kerber, Ph.D.
Department of Oncological Sciences/Huntsman Cancer Institute
Data used: genealogy, birth and death certificates, UCR, DLD

35. Promoting Colonoscopy in Rural High-Risk Families via Telegenetics

Principal Investigator: Anita Kinney, Ph.D.
Department of Internal Medicine, Clinical Epidemiology Division
Data used: genealogy, UCR

36. Rocky Mountain Cancer Genetics Coalition

Principal Investigator: Anita Kinney, Ph.D.
Department of Internal Medicine, Clinical Epidemiology Division
Data used: genealogy, death certificates, UCR, CDRI, DLD

37. Familiality of Psoriasis in Utah

Principal Investigator: Gerald Krueger, M.D.
Department of Dermatology
Data used: genealogy, DRC

38. Genetic Characterization of Novel Subtypes of Inherited Neuropathies

Principal Investigator: Victoria Lawson, M.D.
Department of Neurology
Data used: genealogy, DRC

39. The Tom C. Mathews Familial Melanoma Research Clinic

Principal Investigator: Sancy Leachman, M.D., Ph.D.
Department of Dermatology
Data used: genealogy, death certificates, UCR, CDRI, DLD

40. Genetic Study of Extended Pedigrees with Autism

Principal Investigator: William McMahon, M.D.
Department of Psychiatry
Data used: genealogy, birth and death certificates, DLD

41. Pregnancy Outcomes in Childhood Cancer Survivors

Principal Investigator: Geri Mineau, Ph.D.
Department of Oncological Sciences/Huntsman Cancer Institute
Data used: genealogy, birth and death certificates, fetal death certificates, UCR, DLD

42. Retrospective Chart Review for Veteran Suicide Identification

Principal Investigator: Michelle Moskos, Ph.D., M.P.H.
Department of Pediatrics
Data used: death certificates

43. Intermountain Heart Collaborative Study

Principal Investigator: J. Brent Muhlestein, M.D.
Intermountain Healthcare
Cardiovascular Department
Data used: DLD

44. Genetic Epidemiology of Breast Cancer

Principal Investigator: Susan Neuhausen, Ph.D.
University of California, Irvine
Department of Medicine, Division of Epidemiology
Data used: genealogy, death certificates, CDRI, UCR

45. W. M. Keck Foundation, University of Utah Genetic Reference Project

Principal Investigator: Andy Peiffer, M.D., Ph.D.
Department of Human Genetics
Data used: genealogy, birth and death certificates

46. Utah Infertility Registry-Polycystic Ovary Syndrome (UIR-PCOS)

Principal Investigator: C. Matthew Peterson, M.D.
Department of Obstetrics and Gynecology
Data used: genealogy, DRC

47. The Intermountain States Registry of Childhood Rheumatic Disease

Principal Investigator: Sampath Prahalad, M.D.
Department of Pediatrics, Immunology and Rheumatology
Data used: genealogy, birth certificates

48. Intermountain Multiple Sclerosis Project

Principal Investigator: John Rose, M.D.
Department of Neurology
Data used: genealogy, death certificates, DRC

49. Utah Rheumatoid Arthritis Project

Principal Investigator: Allen Sawitzke, M.D.
Department of Internal Medicine, Rheumatology Division
Data used: genealogy, DRC, DLD

50. Genetic and Molecular Characterization of Interstitial Lung Disease

Principal Investigator: Mary Beth Scholand, M.D.
Department of Internal Medicine, Pulmonary Division
Data used: genealogy, death certificates, DRC

51. Genetic Influences in Autism in Tuberous Sclerosis

Principal Investigator: Susan L. Smalley, Ph.D.
University of California, Los Angeles
Department of Psychiatry
Data used: genealogy

52. Kinship and Socio-Demographic Determinants of Mortality

Principal Investigator: Ken Smith, Ph.D.
Department of Family and Consumer Studies
Data used: genealogy, death certificates

53. Effects of Cancer Risk on Fertility in a Large Kindred with Hereditary Breast and Ovarian Cancer

Principal Investigator: Ken Smith, Ph.D.
Department of Family and Consumer Studies
Data used: birth certificates

54. The Utah Study of Fertility, Longevity and Aging

Principal Investigator: Ken Smith, Ph.D.
Department of Family and Consumer Studies
Data used: genealogy, birth and death certificates, UCR, CDRI, DLD

55. Study of Time to Pregnancy in Normal Fertility

Principal Investigator: Joseph Stanford, M.D.
Department of Family and Preventive Medicine
Data used: birth certificates

56. Validating prediction models of kidney transplant outcome using local data

Principal Investigator: Hongying Tang
Department of Biomedical Informatics
Data used: death certificates, DRC

57. Intergenerational Predisposition to Obstetric Complications

Principal Investigator: Michael Varner, M.D.
Department of Obstetrics/Gynecology
Data used: genealogy, birth and death certificates, fetal deaths, UCR, CDRI, DLD, DRC

58. Thyroid Disease in Persons Exposed to Radioactive Fallout from Atomic Weapons Testing at the Nevada Test Site: Phase III MORTALITY STUDY COMPONENT

Principal Investigator: George White, Ph.D., M.S.P.H.
Department of Family and Preventive Medicines
Data used: birth and death certificates

59. Cancer risks in mothers of twins and their offspring

Principal Investigator: David Whiteman, Ph.D.
Queensland Institute of Medical Research, Australia
Department of Epidemiology
Data used (under a CDITA): genealogy, birth and death certificates, UCR

60. Genetics and Molecular Studies of Eye Disease

Principal Investigator: Kang Zhang, M.D.
Department of Ophthalmology
Data used: genealogy, DRC, DLD

New Projects:

1. An Asthma Susceptibility Gene in Utah

Principal Investigator: Lisa Cannon-Albright, Ph.D.
Department of Biomedical Informatics, Division of Genetic Epidemiology
Data used: genealogy, death certificates, UCR, DRC, DLD

2. Analysis of the Familial Component to Disease in a Biomedical Resource with Linked Genealogy

   Principal Investigator: Lisa Cannon-Albright, Ph.D.
   Department of Biomedical Informatics, Division of Genetic Epidemiology
   Data used: genealogy, death certificates, UCR, DRC, DLD

3. Familiality of Pituitary Tumors using UPDB

   Principal Investigator: Lisa Cannon-Albright, Ph.D.
   Department of Biomedical Informatics, Division of Genetic Epidemiology
   Data used: genealogy, death certificates, UCR, DRC, DLD

4. Probabilistic Linkage Methodological Research: Vaccine Registry Linkage

   Principal Investigator: J. Michael Dean, M.D., M.B.A.
   Department of Pediatrics, Critical Care
   Data used: birth certificates

5. Genetics of Preterm Birth

   Principal Investigator: M. Sean Esplin, M.D.
   Department of Obstetrics and Gynecology
   Data used: genealogy, birth and death certificates, fetal death data, DRC, DLD

6. Genetic Factors in Solid Organ Transplant Outcome

   Principal Investigator: Alexander Goldfarb-Rumyantzev, M.D., Ph.D.
   Department of Internal Medicine, Nephrology Division
   Data used: genealogy, death certificates, UCR, DRC

7. Facility-based Health Care Utilization for Children Who Experience Abuse

   Principal Investigator: Elizabeth Guenther, M.D., M.P.H.
   Department of Pediatrics, Division of Pediatric Emergency Medicine
   Data used: birth certificates, DLD

8. Undocumented Status and Health Indicators in Utah

   Principal Investigator: Ken Jameson, Ph.D.
   Department of Economics
   Data used: DLD

9. Bridging Geographic Barriers: Remote Cancer Genetic Counseling for Rural Women

Principal Investigator: Anita Kinney, Ph.D.
Department of Internal Medicine, Clinical Epidemiology Division
Data used: genealogy, UCR, CDRI

10. Life Course and Health Trajectories of Adolescent Parent - Phase I

Principal Investigator: Kim Korinek, Ph.D.
Department of Sociology
Data used: birth certificates, DLD

11. Neurogenetics of Auditory Processing Disorder in Utah Families

Principal Investigator: Thomas N. Parks, Ph.D.
Department of Neurobiology and Anatomy
Data used: genealogy, DRC

12. Endometriosis: Natural History, Diagnosis and Outcomes

Principal Investigator: C. Matthew Peterson, M.D.
Department of Obstetrics and Gynecology
Data used: genealogy, birth certificates, DLD

13. Predictors of Disease and/or Therapeutic Response in the Treatment of Osteoarthritis

Principal Investigator: Allen Sawitzke, M.D.
Department of Internal Medicine, Rheumatology Division
Data used: genealogy, DRC

14. Graduate course in quantitative historical demography

Principal Investigator: Ken Smith, Ph.D.
Department of Family and Consumer Studies
Data used: genealogy, death certificates

15. The Genetic Basis for Developmental Dysplasia of the Hip

Principal Investigator: David Stevenson, M.D.
Department of Pediatrics, Division of Medical Genetics
Data used: DRC

16. Body Weight from Birth to Adolescence

Principal Investigator: Rebecca Utz, Ph.D.
Department of Sociology
Data used: birth certificates, DLD

17. Historical Social Mobility Analysis

Principal Investigator: Marco van Leeuwen
International Institute of Social History
Department of Sociology, Utrecht University
Data used: genealogy, birth and death certificates

Single search or pilot projects

1. Sally Glaser, Director, Greater Bay Area Cancer Registry, requested preliminary counts on familiality of Hodgkin lymphoma for a grant submission.

2. David Stevenson, Department of Pediatrics, requested a count of the number of patients with developmental dysplasia of the hip in the DRC and the number that linked to UPDB.

3. Maria Norton, Utah State University, requested some frequency statistics on the Cache County cohort for an NIH grant application.

4. Ken Jameson, Department of Economics, requested counts and cross tabs on health information for immigrants for an NSF grant.

5. Lisa Cannon-Albright and Nephi Walton, Department of Biomedical Informatics, requested counts for individuals with known imprinting disorders for autoimmune disorders from the EDW who link to 3 or more generations of genealogy in the UPDB.

6. David Virshup and Vickie Venne, Huntsman Cancer Institute, requested a search of UPDB to determine if two families diagnosed with Li-Fraumeni Syndrome and no identifiable *p53* mutation shared a common ancestor.

RESEARCH PROJECTS APPROVED TO USE HCI RESOURCES

On-going projects using High Risk Breast Cancer Clinic (HRBCC) resources:

1. Genetic and Environmental Modifiers of Penetrance in BRCA1 and BRCA2 Mutation Carriers

   Principal Investigator: Saundra Buys, M.D.[1]
   Department of Internal Medicine, Oncology Division

2. Ovarian Cancer Screening Study

   Principal Investigator: Saundra Buys, M.D.
   Department of Internal Medicine, Oncology Division

3. Psychological Assessment of HRCC Breast Cancer Participants Following Genetic Testing

   Principal Investigator: Anita Kinney, Ph.D.
   Department of Internal Medicine, Clinical Epidemiology Division

4. Genetic Epidemiology of Breast Cancer: BRCA1 and BRCA2

   Principal Investigator: Susan Neuhausen, Ph.D.
   University of California, Irvine

5. Cancer Susceptibility Genetic Testing and Adult Sibling Relationships

   Principal Investigator: Ken Smith, Ph.D.
   Department of Family and Consumer Studies

Continuing Projects Using Familial Colon Cancer Clinic (FCCC) and Registry resources:

1. Behavioral and Psychosocial Issues Related to Screening and Genetic Testing for Familial Adenomatous Polyposis

   Principal Investigator: Anita Kinney, Ph.D.
   Department of Internal Medicine, Clinical Epidemiology Division

1 Dr. Buys is the Rocky Mountain Cancer Genetics Coalition Co-Principal Investigator for this multi-site project accessing Registry information through the Cancer Genetics Network

PUBLICATION REVIEWS - UPDB

PAPERS

1. Harrell, C, Smith, K and Mineau, G. Are Girls Good and Boys Bad? The Effects of Sex Composition of Offspring on Parental Mortality Past Age 50.

2. Penn, D and Smith, K. Differential fitness costs of reproduction between the sexes.

3. Tsai, I-C, Woolf, M, Neklason, D, Branford, W, Yost, H, Burt, R and Virshup, D. A Disease-Associated Casein Kinase I δ Mutation May Promote Adenomatous Polyps Formation via a Wnt/β-catenin Independent Mechanism.

4. Camp, N, Farnham, J and Cannon-Albright, L. Localization of a Prostate Cancer Predisposition Gener to an 880 kilobase Region on Chromosome 22q12.3 in Utah High-Risk Pedigrees.

5. Thomas, A, Camp, N, Farnham, J, Allen-Brady, K and Cannon-Albright, L. Shared genomic segment analysis. A novel approach to mapping disease predisposition genes in extended pedigrees using SNP genotype assays.

6. Maul, J, Burt, R and Cannon-Albright, L. A familial component to rectal cancer, independent of colon cancer risk.

7. Neklason, D, Stevens, J, Boucher, K, Matsunami, N, Barlow, J, Mineau, G, Leppert, M, and Burt, R. American Founder Mutation for Attenuated Familial Adenomatous Polyps.

8. Moskos, M. VA linkage project.

9. Camp, N, Cannon-Albright, L, Farnham, J, et. al. Compelling evidence for a prostate cancer gene at 22q12.3 by the International Consortium for Prostate Cancer Genetics.

10. Camp, N, Farnham, J, Allen-Brady, K and Cannon-Albright, L. Narrowing the 8q24 prostate cancer locus to 2.0 megabases using Utah high-risk extended pedigrees.

ABSTRACTS

1. Christensen, GB, Farnham, J, Camp, N, and Cannon-Albright, L. Survey of Excess Familiality in Prostate Cancer.

2. Christensen, GB, Farnham, J, Camp, N, and Cannon-Albright, L. Genome-wide linkage analysis for aggressive prostate cancer in Utah high-risk pedigrees.

3. Christensen, GB, Farnham, J, Camp, N, and Cannon-Albright, L. Genetic susceptibility of prostate cancer: genome-wide screen of men with non-metastatic disease.

4. Matthews, RD, Neumayer, LA and Cannon-Albright, L. Risk of cancer of other sites among relatives of non-medullary thyroid cancer patients.

5. Camp, NJ, Werner, TL and Cannon-Albright, L. The Familiality of Multiple Myeloma in the Utah Population Database.

6. Teerlink, C and Cannon-Albright, L. A genealogical assessment of heritable predisposition to asthma mortality.

7. Randall, L, Lessnick, S, Kerber, R, Coffin, C, Scaife, C, Andtbacka, R and Cannon-Albright, L. A heritable contribution to malignant fibrous lesions.

8. Cannon-Albright, L, Nygaard, I and Norton, P. The Heritable Contributions to Lower Urinary Tract Symptoms.

9. Matthews, R, Neumayer, L and Cannon-Albright, L. Non-Medullary Thyroid Cancer and Relative Risk of Other Malignancy: An Analysis of the Utah Population Database and Utah Cancer Registry.

THESES

1. Galazka, M. Familiality of Diabetes Mellitus in Utah.