**Collaborative Cancer Epidemiology: Cancer Consortia – Supplemental Materials**

**Supplemental Methods & Results**

*Exclusion Criteria for Scoring by Scientific Area and Translational Stage -* Papers were excluded from scoring by scientific area if they were book chapters, news articles, editorials, or commentaries (N=101); meeting summaries that did not include a synthesis of evidence and guidelines or recommendations were also excluded (N=5). Papers were not coded if their abstracts and texts were not found or were unavailable (N=30), or if they did not fit into any of the defined scientific areas (N=11). Papers were excluded from coding for translational stage if they were not coded for scientific area (N=147, see above), if they were review articles (N=309), or if they were descriptions of resources or infrastructures that presented no empirical data (N=57).

*Comparison of Translational Stage of CEC versus non-CEC associated cancer genetics papers -* A literature analysis by Schully *et al.*(1) estimated that only 0.64% of cancer genetics papers published in 2007 could be considered T2 and beyond. They conducted a PubMed (2) search for cancer related papers, involving human subjects, published in 2007. They identified 20,266 papers containing at least one of the following key words in their titles or abstracts: biomarker, gene, genetic, genome, genomic, epigenetic, epigenomic, gene mapping, genetic testing, and personalized health. After reviewing 660 abstracts and extrapolating those results back to 20,266 articles, they estimated that 130 (0.64%) were T2 or above. (See supplemental table 3 for T-score definitions and examples.) We selected papers within our dataset that would have been retrieved using the following PubMed search: *human AND cancer AND ("biomarker"[TIAB] OR "biomarkers"[TIAB] OR "gene"[TIAB] OR "genes"[TIAB] OR "genetic"[TIAB] OR "genetics"[TIAB] OR "genome"[TIAB] OR "genomes"[TIAB] OR "genomic"[TIAB] OR "genomics"[TIAB] OR "epigenetic"[TIAB] OR "epigenetics"[TIAB] OR "epigenomic"[TIAB] OR "epigenomics"[TIAB] OR "gene mapping"[TIAB] OR "genetic testing"[TIAB] OR "personalized health"[TIAB])* and identified 171 of that were published in 2007. Six (3.5%) of the CEC cancer genetics papers were scored as T2 or above. Assuming that the 171 EGRP CEC cancer genetics papers , published in 2007, would have been included Schully *et al.’s* search results 3.5% of EGRP CEC cancer genetic papers were T2 and above while 0.62% of non-EGRP CEC cancer genetics papers were T2 and above. This difference was found to be statistically significant (P-value 2.4x10-6) using the Pearson's chi-squared test.

*Acknowledgement of CEC contributions in associated papers –* Publications were collected from the websites and grant applications of four, established epidemiology consortia: Gene, Environment Association Studies Consortium (GENEVA); International Consortium for Prostate Cancer Genetics Consortium (ICPCG); Breast Cancer Family Registry (BCFR); and Genetic Associations and Mechanisms in Oncology (GAME-ON). A total of 472 publications were reviewed to determine how the contributing consortium was acknowledged. The type of acknowledgment for each paper was coded into one of 7 hierarchical categories: 1) by CEC name or abbreviation in the title, 2) by CEC name or abbreviation as a group author, 3) by CEC name in the acknowledgements, 4) by associated grant number in the acknowledgments or funding section, 5) by CEC name in the methods or other article text, 6) by CEC name in the supplemental materials only, or 7) the CEC was not acknowledged. The number and percentage of papers acknowledging consortium contributions by category is presented in Supplemental Table 2

**Table S1.** Scientific areas, their definitions, and examples from the literature analysis.

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| **Notation (abbreviation)** | **Definition** | **Examples from Literature Review** |
| Gene Discovery – Linkage (LK) | Scans for association using microsatellite markers including replication studies, fine mapping of gene regions, and segregation analyses. | Family-based association analysis of 42 hereditary prostate cancer families identifies the Apolipoprotein L3 region on chromosome 22q12 as a risk locus (3) |
| Gene Discovery – Sequencing (NGS) | Next generation sequencing studies including whole genome and whole exome sequencing. | Mutations in 12 genes for inherited ovarian, fallopian tube, and peritoneal carcinoma identified by massively parallel sequencing (4) |
| Gene Discovery – Loss of Heterozygosity (LOH) | Studies seeking to identify cancer genes by comparing the presence of heterozygosity at genetic loci in germline and tumor tissue DNA. | Genome-wide allelotypes of familial pancreatic adenocarcinomas and familial and sporadic intraductal papillary mucinous neoplasms (5) |
| Gene Discovery – Genome-wide Association (GWA) | Genome-wide association studies using high density SNP arrays including replication studies, and fine mapping of GWAS hits and meta and pooled analyses of GWAS data. | A genome-wide association study of upper aerodigestive tract cancers conducted within the INHANCE consortium (6)  Associations of common variants at 1p11.2 and 14q24.1 (RAD51L1) with breast cancer risk and heterogeneity by tumor subtype: findings from the Breast Cancer Association Consortium (7) |
| Candidate Gene (CG) | Genetic association studies of variants based on *a priori* knowledge of biological function; includes replication of observed associations from other CG studies. | Nonsynonymous polymorphisms in genes in the one-carbon metabolism pathway and associations with colorectal cancer (8)  7q21-rs6964587 and breast cancer risk: an extended case-control study by the Breast Cancer Association Consortium (9) |
| Gene Characterization (GC) | Studies of allele frequencies, genotype-phenotype correlations, penetrance estimates, gene-gene interactions (including modifier genes), and germline-somatic genotype interactions. | Lifetime risk of melanoma in CDKN2A mutation carriers in a population-based sample (10)  Multiple pigmentation gene polymorphisms account for a substantial proportion of risk of cutaneous malignant melanoma (11) |
| Gene-Environment (GE) | Studies of differential genotypic effects by environmental and lifestyle factors, not including family history or genetically determined phenotypes. | Prospective study of N-acetyltransferase-2 genotypes, meat intake, smoking and risk of colorectal cancer (12) |
| Environment, Lifestyle, and Descriptive Epidemiology (EL) | Studies that examine, describe, or quantify association of risk factors with disease, including co-morbidities. | Passive smoking and the use of noncigarette tobacco products in association with risk for pancreatic cancer: a case-control study (13)  Autoimmune disorders and risk of non-Hodgkin lymphoma subtypes: a pooled analysis within the InterLymph Consortium (14) |
| Clinical and Translational (CT) | Studies of clinical factors and prognosis and survival; clinical trials; assessments of validity and sensitivity of clinical and genetic screening; and case reports. | BRCAPRO validation, sensitivity of genetic testing of BRCA1/BRCA2, and prevalence of other breast cancer susceptibility genes (15)  Distinctive clinical course and pattern of relapse in adolescents with medulloblastoma (16) |
| Resources (RS) | Descriptive papers of consortia, biorepositories, databases, or other research tools; consensus reports and guidelines on research practices. | GLIOGENE an International Consortium to Understand Familial Glioma (17)  Assessing pathogenicity: overview of results from the IARC Unclassified Genetic Variants Working Group (18) |
| Methods and Technologies (MT) | Development of new assays, analytic approaches, methodologies, questionnaires or surveys and other research tools. | MicroRNA expression profiling using microarrays (19)  Leveraging genetic variability across populations for the identification of causal variants (20) |
| Biology (BIO) | Functional studies of associated genes, investigations of molecular pathways *in vitro* or in animal models, gene expression analysis in tumor or normal tissue. | Secondary mutations as a mechanism of cisplatin resistance in BRCA2-mutated cancers(21)  Overexpression of HER-2 in ovarian carcinomas (22) |
| Behavioral (BR) | Studies on behavioral determinants of screening, risk affecting behaviors, and lifestyles. Includes results of focus groups and surveys of provider and patient attitudes and opinions. | Attitudes, knowledge, and risk perceptions of women with breast and/or ovarian cancer considering testing for BRCA1 and BRCA2 (23)  The impact of direct-to-consumer marketing of cancer genetic testing on women according to their genetic risk (24) |
| Molecular Epidemiology (ME) | Association studies of epigenetic factors on outcome, risk, and disease status; discovery and validation of disease biomarkers; association molecular factors, including tumor subtypes, with disease status, survival, and other outcomes. | Short telomere length and breast cancer risk: a study in sister sets (25)  CCL3 (MIP-1alpha) plasma levels and the risk for disease progression in chronic lymphocytic leukemia (26)  CpG island methylator phenotype underlies sporadic microsatellite instability and is tightly associated with BRAF mutation in colorectal cancer (27) |

**Table S2.** Acknowledgment CEC contributions in 472 papers, associated with 4 established CEC, by acknowledgement type.

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| --- | --- | --- | --- | --- | --- |
|  | **GENEVA** | **ICPCG** | **BCFR** | **GAME-ON** | **All Consortia** |
| **Acknowledgement Type** | **Manuscripts** | **Manuscripts** | **Manuscripts** | **Manuscripts** |
|  | **N (%)** | **N (%)** | **N (%)** | **N (%)** | **N (%)** |
| Title | 2 (4%) | 8 (10%) | 22 (10%) | 0 (0%) | 32 (7%) |
| Author line | 9 (18%) | 1 (1%) | 14 (6%) | 2 (2%) | 26 (5%) |
| Acknowledgment section | 18 (35%) | 5 (6%) | 50 (22%) | 10 (8%) | 83 (18%) |
| Associated grant number in acknowledgment or funding section only | 13 (25%) | 63 (81%) | 89 (40%) | 47 (40%) | 212 (45%) |
| Methods or other article text only | 3 (6%) | 0 | 3 (1%) | 1 (1%) | 7 (1%) |
| Supplemental materials only | 4 (8%) | 1 (1%) | 3 (1%) | 1 (1%) | 9 (2%) |
| Not acknowledged | 2 (4%) | 0 | 44 (20%) | 57 (48%) | 103 (22%) |
| **Total Manuscripts** | **51** | **78** | **225** | **118** | **472** |

**Supplemental References**

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