August 13, 2013

Big Kahuna Literature Review

Search strategy in title/abstract:

1. (Gene OR Synonyms) AND (Germline OR Germ-line OR Inherited OR Genetic Predisposition to Disease[MeSH Major Topic]) AND (Penetrance OR Allele Freq OR Incidence) AND (Any Diseases of Interest)
2. Segregation Analysis AND (Pancreas/Pancreatic or Prostate or Lung) AND (Cancer OR Adenomcarcinoma)
3. Gene-specific inclusions based upon reviews

Gene Synonyms based upon lit review and Cancer Gene Census for Germline Mutate cancer genes: <http://www.sanger.ac.uk/genetics/CGP/Census/germline_mutation.shtml>

Saved to chipmanj NCBI account

**Inclusion Criteria**

1. Reporting of penetrance, prevelance, allele frequency or risk factors affecting penetrance
2. If reporting on a gene variant, then include if:
   1. It is a common variant (such as a founder mutation variant)
   2. If a penetrance of variant is provided

**Exclusion Criteria**

1. Paper not published in English
2. Family studies of less than 5 families
3. The paper reports on a gene other than gene of interest (paper is captured b/c gene of interest is in abstract) \*
4. Full text of article could not be obtained (should be very rare).
5. The paper reports on a spectrum of gene variants but without any penetrance or prevalence information
6. The paper reports data that have subsequently been entirely included in larger studies
7. Paper reports study not conducted in humans

\* If the paper reports on MLH6, but the gene-specific lit review is for MLH1, make sure the MLH6 paper is among those obtained by the MLH6 search.

**BRCA1: 356 articles**

(672[Title/Abstract] OR BRCA1[Title/Abstract] OR BRCAI[Title/Abstract] OR BRCC1[Title/Abstract] OR ENSG00000012048[Title/Abstract] OR IRIS[Title/Abstract] OR PSCP[Title/Abstract] OR RNF53[Title/Abstract]) **AND**

(Germline[Title/Abstract] OR Germ-line[Title/Abstract] OR Inherited[Title/Abstract] OR "Genetic Predisposition to Disease"[MAJR]) **AND**

(Penetrance[Title/Abstract] OR Allele Frequency[Title/Abstract] OR Incidence[Title/Abstract]) **AND**

(Breast[Title/Abstract] OR Ovarian[Title/Abstract] OR Ovary[Title/Abstract] OR Pancreas[Title/Abstract] OR Pancreatic[Title/Abstract] OR Melanoma[Title/Abstract] OR Prostate[Title/Abstract] OR Lung[Title/Abstract] OR Colorectal[Title/Abstract] OR Colon[Title/Abstract] OR Endometrial[Title/Abstract] OR Endometrium[Title/Abstract] OR Sarcoma[Title/Abstract] OR Sarcomas[Title/Abstract])

**BRCA2: 287 articles**

(675[Title/Abstract] OR BRCA2[Title/Abstract] OR BRCC2[Title/Abstract] OR ENSG00000139618[Title/Abstract] OR FACD[Title/Abstract] OR FAD[Title/Abstract] OR FAD1[Title/Abstract] OR FANCB[Title/Abstract] OR FANCD[Title/Abstract] OR FANCD1[Title/Abstract] OR RP11-298P3\_4) **AND**

(Germline[Title/Abstract] OR Inherited[Title/Abstract] OR "Genetic Predisposition to Disease"[MAJR]) **AND**

(Penetrance[Title/Abstract] OR Allele Frequency[Title/Abstract] OR Incidence[Title/Abstract]) **AND**

AND (Breast[Title/Abstract] OR Ovarian[Title/Abstract] OR Ovary[Title/Abstract] OR Pancreas[Title/Abstract] OR Pancreatic[Title/Abstract] OR Melanoma[Title/Abstract] OR Prostate[Title/Abstract] OR Lung[Title/Abstract] OR Colorectal[Title/Abstract] OR Colon[Title/Abstract] OR Endometrial[Title/Abstract] OR Endometrium[Title/Abstract] OR Sarcoma[Title/Abstract] OR Sarcomas[Title/Abstract])

**CDK4: 30 articles**

1019[Title/Abstract] OR CCDS8953.1[Title/Abstract] OR CDK4[Title/Abstract] OR CMM3[Title/Abstract] OR ENSG00000135446[Title/Abstract] OR MGC14458[Title/Abstract] OR P11802[Title/Abstract] OR PSK-J3[Title/Abstract] **AND**

(Germline[Title/Abstract] OR Inherited[Title/Abstract] OR "Genetic Predisposition to Disease"[MAJR]) **AND**

(Penetrance[Title/Abstract] OR Allele Frequency[Title/Abstract] OR Incidence[Title/Abstract]) **AND**

(Breast[Title/Abstract] OR Ovarian[Title/Abstract] OR Ovary[Title/Abstract] OR Pancreas[Title/Abstract] OR Pancreatic[Title/Abstract] OR FAMM[Title/Abstract] OR Familial Atypical Multiple Mole Melanoma[Title/Abstract] OR Melanoma[Title/Abstract] OR Prostate[Title/Abstract] OR Lung[Title/Abstract] OR Colorectal[Title/Abstract] OR Colon[Title/Abstract] OR Endometrial[Title/Abstract] OR Endometrium[Title/Abstract] OR Sarcoma[Title/Abstract] OR Sarcomas[Title/Abstract])

**CDKN2A or P16: 35 articles**

(1029[Title/Abstract] OR ARF[Title/Abstract] OR CDK4I[Title/Abstract] OR CDKN2[Title/Abstract] OR CDKN2A[Title/Abstract] OR CMM2[Title/Abstract] OR ENSG00000147889[Title/Abstract] OR INK4[Title/Abstract] OR INK4a[Title/Abstract] OR MLM[Title/Abstract] OR MTS1[Title/Abstract] OR P42771[Title/Abstract] OR TP16[Title/Abstract] OR p14[Title/Abstract] OR p14ARF[Title/Abstract] p16[Title/Abstract] OR p16INK4[Title/Abstract] OR p16INK4a[Title/Abstract] OR p19[Title/Abstract]) **AND**

(Germline[Title/Abstract] OR Inherited[Title/Abstract] OR "Genetic Predisposition to Disease"[MAJR]) **AND**

(Penetrance[Title/Abstract] OR Allele Frequency[Title/Abstract] OR Incidence[Title/Abstract]) **AND**

(Breast[Title/Abstract] OR Ovarian[Title/Abstract] OR Ovary[Title/Abstract] OR Pancreas[Title/Abstract] OR Pancreatic[Title/Abstract] OR FAMM[Title/Abstract] OR Familial Atypical Multiple Mole Melanoma[Title/Abstract] OR Melanoma[Title/Abstract] OR Prostate[Title/Abstract] OR Lung[Title/Abstract] OR Colorectal[Title/Abstract] OR Colon[Title/Abstract] OR Endometrial[Title/Abstract] OR Endometrium[Title/Abstract] OR Sarcoma[Title/Abstract] OR Sarcomas[Title/Abstract])

**MLH1: 143 articles**

(4292[Title/Abstract] OR CCDS2663.1[Title/Abstract] OR COCA2[Title/Abstract] OR ENSG00000076242[Title/Abstract] OR

FCC2[Title/Abstract] OR HNPCC[Title/Abstract] OR HNPCC2[Title/Abstract] OR MGC5172[Title/Abstract] OR MLH1[Title/Abstract] OR P40692[Title/Abstract] OR Q0ZAJ3[Title/Abstract] OR Q0ZAJ6[Title/Abstract] OR Q53GX1[Title/Abstract] OR Q5GJ64[Title/Abstract] OR Q64FK0[Title/Abstract] OR Q6VBB8[Title/Abstract] OR hMLH1[Title/Abstract]) **AND**

(Germline[Title/Abstract] OR Inherited[Title/Abstract] OR "Genetic Predisposition to Disease"[MAJR]) **AND**

(Penetrance[Title/Abstract] OR Allele Frequency[Title/Abstract] OR Incidence[Title/Abstract]) **AND**

(Breast[Title/Abstract] OR Ovarian[Title/Abstract] OR Ovary[Title/Abstract] OR Pancreas[Title/Abstract] OR

Pancreatic[Title/Abstract] OR Melanoma[Title/Abstract] OR Prostate[Title/Abstract] OR Lung[Title/Abstract] OR

Colorectal[Title/Abstract] OR Colon[Title/Abstract] OR Endometrial[Title/Abstract] OR Endometrium[Title/Abstract] OR

Sarcoma[Title/Abstract] OR Sarcomas[Title/Abstract])

**MSH2: 130 articles**

(4436[Title/Abstract] OR CCDS1834.1[Title/Abstract] OR COCA1[Title/Abstract] OR ENSG00000095002[Title/Abstract] OR

FCC1[Title/Abstract] OR HNPCC[Title/Abstract] OR HNPCC1[Title/Abstract] OR MSH2[Title/Abstract] OR

P43246[Title/Abstract] OR Q0ZAI9[Title/Abstract] OR Q0ZAJ1[Title/Abstract] OR Q53FK0[Title/Abstract] OR

Q53GS1[Title/Abstract] OR Q53RU4[Title/Abstract] OR Q6VBB7[Title/Abstract]) **AND**

(Germline[Title/Abstract] OR Inherited[Title/Abstract] OR "Genetic Predisposition to Disease"[MAJR]) **AND**

(Penetrance[Title/Abstract] OR Allele Frequency[Title/Abstract] OR Incidence[Title/Abstract]) **AND**

(Breast[Title/Abstract] OR Ovarian[Title/Abstract] OR Ovary[Title/Abstract] OR Pancreas[Title/Abstract] OR

Pancreatic[Title/Abstract] OR Melanoma[Title/Abstract] OR Prostate[Title/Abstract] OR Lung[Title/Abstract] OR

Colorectal[Title/Abstract] OR Colon[Title/Abstract] OR Endometrial[Title/Abstract] OR Endometrium[Title/Abstract] OR

Sarcoma[Title/Abstract] OR Sarcomas[Title/Abstract])

**MSH6: 46 articles**

(2956[Title/Abstract] OR CCDS1836.1[Title/Abstract] OR ENSG00000116062[Title/Abstract] OR GTBP[Title/Abstract] OR HNPCC5[Title/Abstract] OR HSAP[Title/Abstract] OR MSH6[Title/Abstract] OR P52701[Title/Abstract] OR Q1L838[Title/Abstract] OR Q3SWU9[Title/Abstract])**AND**

(Germline[Title/Abstract] OR Inherited[Title/Abstract] OR "Genetic Predisposition to Disease"[MAJR]) **AND** (Penetrance[Title/Abstract] OR Allele Frequency[Title/Abstract] OR Incidence[Title/Abstract]) **AND** (Breast[Title/Abstract] OR Ovarian[Title/Abstract] OR Ovary[Title/Abstract] OR Pancreas[Title/Abstract] OR Pancreatic[Title/Abstract] OR Melanoma[Title/Abstract] OR Prostate[Title/Abstract] OR Lung[Title/Abstract] OR Colorectal[Title/Abstract] OR Colon[Title/Abstract] OR Endometrial[Title/Abstract] OR Endometrium[Title/Abstract] OR Sarcoma[Title/Abstract] OR Sarcomas[Title/Abstract])

**P53: 142 articles**

(7157[Title/Abstract] OR CCDS11118.1[Title/Abstract] OR ENSG00000141510[Title/Abstract] OR LFS1[Title/Abstract] OR P04637[Title/Abstract] OR P53[Title/Abstract] OR TP53[Title/Abstract] OR TRP53[Title/Abstract] OR p53[Title/Abstract] OR Li-Fraumeni[Title/Abstract]) **AND**

(Germline[Title/Abstract] OR Inherited[Title/Abstract] OR "Genetic Predisposition to Disease"[MAJR]) **AND**

(Penetrance[Title/Abstract] OR Allele Frequency[Title/Abstract] OR Incidence[Title/Abstract]) **AND**

(Breast[Title/Abstract] OR Ovarian[Title/Abstract] OR Ovary[Title/Abstract] OR Pancreas[Title/Abstract] OR Pancreatic[Title/Abstract] OR Melanoma[Title/Abstract] OR Prostate[Title/Abstract] OR Lung[Title/Abstract] OR Colorectal[Title/Abstract] OR Colon[Title/Abstract] OR Endometrial[Title/Abstract] OR Endometrium[Title/Abstract] OR Sarcoma[Title/Abstract] OR Sarcomas[Title/Abstract] OR "Adrenal Cortical"[Title/Abstract])

**PALB2: 26 articles**

(79728[Title/Abstract] OR ENSG00000083093[Title/Abstract] OR PALB2[Title/Abstract]) **AND**

(Germline[Title/Abstract] OR Inherited[Title/Abstract] OR "Genetic Predisposition to Disease"[MAJR]) **AND**

(Penetrance[Title/Abstract] OR Allele Frequency[Title/Abstract] OR Incidence[Title/Abstract]) **AND**

(Breast[Title/Abstract] OR Ovarian[Title/Abstract] OR Ovary[Title/Abstract] OR Pancreas[Title/Abstract] OR Pancreatic[Title/Abstract] OR Melanoma[Title/Abstract] OR Prostate[Title/Abstract] OR Lung[Title/Abstract] OR Colorectal[Title/Abstract] OR Colon[Title/Abstract] OR Endometrial[Title/Abstract] OR Endometrium[Title/Abstract] OR Sarcoma[Title/Abstract] OR Sarcomas[Title/Abstract])

**ATM: 42 articles**

(472[Title/Abstract] OR AT1[Title/Abstract] OR ATA[Title/Abstract] OR ATC[Title/Abstract] OR ATD[Title/Abstract] OR ATDC[Title/Abstract] OR ATE[Title/Abstract] OR ATM[Title/Abstract] OR DKFZp781A0353[Title/Abstract] OR ENSG00000149311[Title/Abstract] OR MGC74674[Title/Abstract] OR Q13315[Title/Abstract] OR TEL1[Title/Abstract] OR TELO1[Title/Abstract]) **AND**

(Germline[Title/Abstract] OR Germ-line[Title/Abstract] OR Inherited[Title/Abstract] OR "Genetic Predisposition to Disease"[MAJR]) **AND**

(Penetrance[Title/Abstract] OR Allele Frequency[Title/Abstract] OR Incidence[Title/Abstract]) **AND**

(Breast[Title/Abstract] OR Ovarian[Title/Abstract] OR Ovary[Title/Abstract] OR Pancreas[Title/Abstract] OR Pancreatic[Title/Abstract] OR Melanoma[Title/Abstract] OR Prostate[Title/Abstract] OR Lung[Title/Abstract] OR Colorectal[Title/Abstract] OR Colon[Title/Abstract] OR Endometrial[Title/Abstract] OR Endometrium[Title/Abstract] OR Sarcoma[Title/Abstract] OR Sarcomas[Title/Abstract])

**PANC, PROS, LUNG: 32 articles**

Segregation Analysis[Title/Abstract] **AND**

(Pancreas[Title/Abstract] OR Pancreatic[Title/Abstract] OR Prostate[Title/Abstract] OR Lung[Title/Abstract]) **AND**

(Cancer[Title/Abstract] OR Adenocarcinoma[Title/Abstract])

Sapna Suggestions:

PMS2, EPCAM: Lynch

MYH & APC - Polyposis ??, Mostly like FAP (recessive)

**Updates Log:**

**08/26/2013:**

Updated Lit Review Counts

Strategy:

* Added Germ-line to: (Germline[Title/Abstract] OR Germ-line[Title/Abstract]
* Ask GP about the CDKN2A search criteria. I have an AND P16 criteria. Was this a typo and should it be OR P16?
* Uploaded Literature Reviews into REDCap database to reduce and simplify number of review xls sheets to one per gene. Reviewers save their reviews by adding a suffix to the gene file (ex: 2013-08-07 PALB2 – LitReviewJC). The reviews will be uploaded into the database regularly, and once saved will be retained in a “Previous Reviews” folder in dropbox.
* All PDFs are saved centrally in an EndNote library: Kahuna Literature Review.enl

**08/13/2013:**

Added inclusion / exclusion criteria

**07/09/2013:**

Updated Lit Review Counts

Strategy, additions per meeting with Sapna Syngal:

* Added Major Topic MeSH term: "Genetic Predisposition to Disease" to: (Germline[Title/Abstract] OR Inherited[Title/Abstract] OR "Genetic Predisposition to Disease"[MAJR])
* Added Li-Fraumeni to listing of genes and “Adrenal Cortical” to listing of diseases in P53 search.
* Added synonyms to other genes
* Separated CDK4 from CDKN2A

**06/20/2013:**

Strategy:

* Per BM discussion and missing articles in CDKN2A, added Incidence to: (Penetrance[Title/Abstract] OR Allele Frequency[Title/Abstract] OR Incidence[Title/Abstract])

**04/05/2013:**

Updated Lit Review Counts

Strategy:

* Added “Gene-specific inclusions based upon reviews”

PANC, PROS, LUNG:

* Per AKlein’s suggestion, included criteria: AND (Cancer[Title/Abstract] OR Adenocarcinoma[Title/Abstract]).

CDKN2A:

* Fixed typo (CDKN2A was CDK2NA)
* Per BM meeting, added into criteria alternative gene nomenclatures: CDK4, CDKN2, MTSI, INK4, P16INK4
* Per BM meeting, added into criteria additional diseases: FAMM OR Familial Atypical Multiple Mole Melanoma

**03/29/2013:**

Updated Lit Review Counts

All non-segregation genes

* Per BM meeting, included criteria: AND (Germline[Title/Abstract] OR Inherited[Title/Abstract])