Literature Review

Option 1

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 an 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.

Option 2

Use NCBI online resources to obtain all relevant papers denoting gene penetrance, prevalence, and important gene modifiers.

If those papers also cited a key paper not already identified from the online resource, we added that to the list of relevant papers.

**Do we need to keep allele freq? Is penetrance/incidence enough?**

Select one paper per gene/cancer combination

1. Save list of papers from option 2/download all abstracts.
2. After obtaining list from option 2, if expert available, consult with an expert (Sapna, Kevin) on which paper to select.
3. If no expert, select paper based on:
   1. Meta analysis/review.
   2. Sample size.
   3. Number of citations.
   4. Should we look at the quantity they estimate? (penetrance vs. hazard ratio)