

BayesMendelKnowledgeBase: An R Package for ASK2ME website service

Jiabei Yang

Dana Farber Cancer Institute

August 9, 2016

Gene/Cancer up till now...

- ▶ Risk of Cancer given Gene mutation
 - ▶ ATM: Breast(Female), Colorectal, Gastric, Pancreatic, Prostate;
 - ▶ BRCA1: Breast, Ovarian, Pancreatic;
 - ▶ BRCA2: Breast, Ovarian, Pancreatic;
 - ▶ BRIP1: Ovarian;
 - ▶ CDH1: Breast(Female), Gastric;
 - ▶ CHEK2: Breast, Colorectal, Prostate;
 - ▶ MLH1: Colorectal, Endometrial, Ovarian;
 - ▶ MSH2: Colorectal, Endometrial, Ovarian;
 - ▶ MSH6: Colorectal, Endometrial, Ovarian;
 - ▶ P16: Melanoma;
 - ▶ PALB2: Breast, Pancreatic;
 - ▶ PMS2: Colorectal, Endometrial, Ovarian;
 - ▶ PTEN: Breast(Female), Colorectal, Endometrial, Kidney, Melanoma, Thyroid;
 - ▶ RAD51C: Ovarian;
- ▶ History of mastectomy, oophorectomy, hysterectomy added for risk reduction

Next steps in the future

- ▶ Prior cancers
- ▶ Risks of breast cancer for males (ATM, CDH1, PTEN)
- ▶ Messed up in the rating of papers
 - ▶ MLH1, MSH2, MSH6/Colorectal not the BayesMendel paper
 - ▶ BRCA1/Ovarian, BRCA2/Breast Mocci or BayesMendel?
 - ▶ MLH1, MSH2, MSH6, PMS2, PTEN/Endometrial, P16/Melanoma not in the Grid file
- ▶ Double check on the paragraphs for other cancers
 - ▶ Estimates of PALB2/Breast for male in database (Zhang 2013)
 - ▶ Rating different for the same paper (Tan 2012)
 - ▶ BARD1 not yet in the database