1. Discuss the indications for genetic counseling in women with a personal or family history of cancer.
2. Describe the physical, emotional, and social impact of genetic conditions that increase cancer risk.
3. Identify options for screening and cancer prevention for women at high risk of breast cancer.
4. Discuss how health care professionals can encourage discussion amongst family members regarding genetic testing.
The Family Tree – the Why

- Ruth Pedersen (died of ovarian cancer)
  - Louis Pedersen, Jr (+)
    - Gwen Blum (+)
      - Hannah Johnk (+)
      - Haley Blum (+)
      - Lucas Blum (-)
    - Five daughters (-)
    - One daughter (unknown)
  - Ruth’s sister (died of breast cancer)
    - Louis’s 1st cousin A (unknown)
      - Daughter (+) prophylactic surgery
      - Daughter (-)
    - Louis’s 1st cousin B (unknown)
      - Daughter (+) prophylactic surgery
      - Son (unknown)

What is BRCA1?

- BRCA1 stands for breast cancer gene. We all have this.
- Human gene that produces tumor suppressor proteins
- It was first identified in 1990 at University of California-Berkeley
- A positive test result means that you have inherited a mutation in the breast cancer gene, and therefore have a much higher risk of developing breast cancer and ovarian cancer

Medical Advice

- Lifetime risk for those with the BRCA1 mutation goes from 12% to 87% for breast cancer, and 6% to 44% for ovarian cancer.
- OPTIONS
  - Breast cancer
    - Prophylactic double mastectomy with reconstruction
    - More frequent screening
    - Alternating mammograms and breast MRI every 6 months
  - Ovarian cancer
    - Remove the ovaries and fallopian tubes, or not
    - No effective screening for ovarian cancer
Removal of ovaries was an easy decision
› Done having children
› Post-menopausal
› No definitive screening options. It is a silent killer.

Removal of breasts was much tougher
› Physicians and counselors input
› Discussion with friends and family
› BRCA Sisterhood on facebook
› Discussion of reconstruction options
› Family friend going through breast cancer at the time (eventually fatal)

Greater impact for pre-menopausal women
Still have side effects for post-menopausal women

Prophylactic Double Mastectomy and Reconstruction

Considerations:
› Single surgery?
› Skin/nipple sparing?
› Implants or DIEP flap?
› Perioperative risks
› Size
Hannah’s perspective

- Pregnant at the time of diagnosis
- Screening recommended every 6 months at age 30
- Unable to have MRI or mammo when pregnant or nursing
- Not done having children
- Testing not recommended until age 18