Conclusions: Using a condensed genetic counseling video and providing an option for genetic testing during a patient’s initial appointment significantly increased the frequency of genetic testing used by patients with ovarian, fallopian, or peritoneal cancer. Current technology can be used to provide immediate and interactive methods of counseling and may dramatically increase the utilization of genetic testing in patients with ovarian, fallopian, or peritoneal cancer.

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22 - Scientific Plenary
Bone density testing underutilized in BRCA population following risk-reducing salpingo-oophorectomy
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Objectives: Characterize bone health surveillance patterns, bone mineral density (BMD) outcomes, and fracture risk after risk-reducing salpingo-oophorectomy (RRSO) in patients with BRCA mutations.

Methods: An institutional review board–approved, retrospective review was performed of health surveillance among BRCA1/2 mutation carriers after RRSO from the years 2000 to 2013. Women with occult carcinoma at RRSO were excluded from analysis. The primary outcome was the number of women who had a dual-energy X-ray absorptiometry (DEXA) scan after RRSO. Secondary outcomes included new diagnoses and time to diagnosis of osteopenia and osteoporosis. Incidence of fracture was also included. Information regarding hormone replacement therapy (HRT) was also recorded. Use of aromatase inhibitors, chemotherapy, and osteoporotic agents was not recorded.

Results: A total of 192 BRCA mutation carriers underwent RRSO. Median age at the time of RRSO was 48 years. Approximately, 65% of the cohort was premenopausal at the time of surgery. Median follow-up was 6.5 years from date of surgery. DEXA scanning was performed in 97 (51%) women after RRSO, of which 48 patients had 1 or more test. Age, preoperative menopausal status, use of HRT, and length of follow-up were comparable between BRCA mutation carriers who had DEXA surveillance and those who did not. Seventy-six (78%) women had abnormal findings. Fifty-eight (60%) had osteopenia and 19 (20%) had osteoporosis. Median time to abnormal bone density was 24 months (range, 1–151). Fracture was seen in 10 patients (5%). In women younger than 50 years, the frequencies of osteopenia and osteoporosis were 66% and 11% compared with 50% and 31% in postmenopausal women (P = .08). Thirty-five women (46%) who had DEXA surveillance used HRT. Women who used HRT had lower frequencies of osteopenia and osteoporosis than women who did not use HRT, 74% and 0.06% vs 83% and 22%, respectively (P = .09; OR 0.26, CI 0.06–1.22).

Conclusions: Significant bone loss is common and develops rapidly in women after RRSO. Women with BRCA mutations who undergo RRSO are underscreened for BMD. HRT is associated with a lower risk of significant bone loss osteoporosis. Guidelines for screening in these individuals should be firmly established to reduce osteoporotic-related fracture risk in this population.

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23 - Scientific Plenary
Higher rates of clinically actionable multigene panel results in Ashkenazi Jewish patients
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Objective: To investigate whether higher rates of clinically actionable multigene panel results in Ashkenazi Jewish patients result in clinically actionable and nonactionable mutations that may confer a higher risk of breast, ovarian, fallopian, and peritoneal cancer.

Methods: Four hundred and forty-four patients underwent multigene panel testing. The median patient age was 54 years (range, 25–91 years) and 435 (96%) were women. One hundred and thirty-three patients (28%) were Ashkenazi Jewish (AJ) and 93 (21%) were not Caucasian. Three hundred and forty-four patients (78%) had a personal history of cancer. Forty-nine patients had ovarian cancer, 26 endometrial cancer, and 251 breast cancer. We identified 62 pathogenic mutations in 56 patients (12%) and 291 variants of uncertain significance in 196 patients (43%). Overall, 41 pathogenic mutations identified (66%) were actionable. Having a personal or family history of cancer or a specific diagnosis of ovarian, endometrial, or breast cancer did not affect the likelihood of identifying a clinically actionable mutation. Twenty pathogenic mutations were identified in 19 AJ patients, 18 of which were in genes other than BRCA1/2. Among those with pathogenic mutations, AJ patients were significantly more likely than non-AJ patients to harbor an actionable mutation (17 [85%] vs 24 [57%]; P = .04). (See Fig. 1.)

Conclusions: With the rapid acceptance of multigene panels, there is a pressing need to understand how this testing will affect patient management. We found that screening and prevention recommendations existed for 66% of the pathogenic mutations identified. In the AJ population, 85% of identified mutations were actionable, only 2 of which were in the BRCA1/2 genes. Our findings suggest that panel testing may be especially useful in the AJ population.

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24 - Scientific Plenary
Hereditary cancer panel testing in an unscreened endometrial carcinoma cohort
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Objectives: To characterize bone health surveillance patterns, bone mineral density (BMD) outcomes, and fracture risk after risk-reducing salpingo-oophorectomy in patients with ovarian, fallopian, or peritoneal cancer.

Methods: We reviewed the medical records of all patients who underwent multigene panel testing at a single institution between May 2012 and December 2014.

Results: Four hundred and fifty-four patients underwent multigene panel testing. The median patient age was 54 years (range, 25–91 years) and 435 (96%) were women. One hundred and thirty-three patients (28%) were Ashkenazi Jewish (AJ) and 93 (21%) were not Caucasian. Three hundred and forty-four patients (78%) had a personal history of cancer. Forty-nine patients had ovarian cancer, 26 endometrial cancer, and 251 breast cancer. We identified 62 pathogenic mutations in 56 patients (12%) and 291 variants of uncertain significance in 196 patients (43%). Overall, 41 pathogenic mutations identified (66%) were actionable. Having a personal or family history of cancer or a specific diagnosis of ovarian, endometrial, or breast cancer did not affect the likelihood of identifying a clinically actionable mutation. Twenty pathogenic mutations were identified in 19 AJ patients, 18 of which were in genes other than BRCA1/2. Among those with pathogenic mutations, AJ patients were significantly more likely than non-AJ patients to harbor an actionable mutation (17 [85%] vs 24 [57%]; P = .04). (See Fig. 1.)

Conclusions: With the rapid acceptance of multigene panels, there is a pressing need to understand how this testing will affect patient management. We found that screening and prevention recommendations existed for 66% of the pathogenic mutations identified. In the AJ population, 85% of identified mutations were actionable, only 2 of which were in the BRCA1/2 genes. Our findings suggest that panel testing may be especially useful in the AJ population.

Fig. 1. Mutations in Ashkenazi Jewish and non-Ashkenazi Jewish Patients.

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