Introduction/Background Ataxia-telangiectasia is an autosomal recessive neurodegenerative disorder with immunodeficiency and an increased risk of developing cancer, caused by mutations in the ataxia-telangiectasia mutated (ATM) gene. Female carriers have an increased risk (20–30%) of developing breast cancer.

It is known that heterozygosity for a pathogenic ATM variant is present in 1%–2% of the adult population.

Methodology Retrospective observational study. Review of patients followed in the inherited cancer unit at Gregorio Marañón University General Hospital between 1st January 2012 until 28th February 2020.

The statistical analysis was carried out using SPSS 26.0.

Results During the indicated period, we followed 333 patients with confirmed genetic mutations that predispose to developing gynaecological cancer. Of the total, 1.5% (5/333) were carriers of a pathogenic ATM mutation and 0.6% (2/333) of a variant of uncertain meaning ATM mutation.

Within the cohort of patients carrying a pathogenic ATM mutation, all of them had family history of breast cancer and one also of ovarian cancer. 3/5 of the patients were diagnosed with breast cancer at 38, 40 and 48 years old, respectively. One of them, had a second contralateral breast cancer at 58 years old.

The histology of the primary breast cancer was: ductal carcinoma in situ, invasive ductal carcinoma and invasive lobular carcinoma. Tumor stage was 0 in one case and stage I in two cases. Primary treatment was surgery in all patients: unilateral mastectomy with homolateral axillary lymphadenectomy in one patient and conservative surgery with selective sentinel lymph node biopsy in the other 2. Adjuvant treatment was only needed in 2 patients: radiation therapy and hormone therapy in one case and radiation therapy, chemotherapy and hormone therapy in the other.

Characteristics of these patients are summarized in table 1.

Abstract 194 Table 1

Table 1.

Ataxia-telangiectasia (n=7)	Frequency (%)
Mutation type	
Pathogenic	5/7 (71.4%)
Variant of uncertain meaning	2/7 (28.6%)
Family history of cancer (ATM pathogenic mutation)	
First degree	2/5 (40%)
Second degree	1/5 (20%)
Unknown	2/5 (40%)
Personal history of cancer (First tumour location)	
No cancer	2/5 (40%)
Breast	3/5 (60%)
Histology of the primary breast cancer	
Ductal carcinoma in situ	1/3 (33.3%)
Invasive ductal carcinoma	1/3 (33.3%)
Invasive lobular carcinoma	1/3 (33.3%)
Breast cancer stage	
Stage 0	1/3 (33.3%)
Stage I	2/3 (66.6%)
Primary breast cancer treatment	
Unilateral mastectomy with homolateral axillary lymphadenectomy	1/3 (33.3%)
Conservative surgery with selective sentinel lymph node biopsy	2/3 (66.6%)
Adjuvant treatment	
Not needed	1/3 (33.3%)
Radiation therapy and hormone therapy	1/3 (33.3%)
Radiation therapy, chemotherapy and hormone therapy	1/3 (33.3%)

Conclusion Patients carrying ATM mutations have a moderate-high risk of developing breast cancer and should be followed in specialized hereditary cancer units, in tertiary hospitals.

Disclosures No conflicts of interest to disclose.

229

BREAST CANCER PATIENTS AT INCREASED RISK OF DEVELOPING TYPE II ENDOMETRIAL CANCER; RELATIVE AND ABSOLUTE RISK ESTIMATION AND IMPLICATIONS FOR GENETIC COUNSELLING

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Introduction/Background Breast cancer (BC) is a recognised risk factor for endometrial cancer (EC). Emerging literature indicates it confers a higher risk of type II EC (T2EC) than type I EC (T1EC). Some surgeons offer a prophylactic hysterectomy to BC patients referred for risk-reducing bilateral salpingo-oophorectomy yet insufficient evidence prevents this from being standard practice. We aimed to quantify their relative and absolute risks of developing both endometrial cancer subtypes and identify a higher-risk group that should be considered for prophylactic hysterectomy.

Methodology This retrospective observational case-control study compared patients diagnosed with BC between 2008 and 2014, who subsequently developed EC within 10 years to those who did not. Absolute and relative risks were calculated using the numbers of regional BC and EC cases within this group, alongside 2009 UK female population and EC incidence statistics. Binary logistic regression generated adjusted odds ratios for patient- and disease-specific variables.

Results Forty-five BC patients developed endometrial cancer; twenty-four had T1EC and twenty-one had T2EC. Their relative risk of developing EC was greater than the general population's (RR 10.24, $p < 0.0001$). Notably, this was higher for T2EC (RR 32.37, $p < 0.001$) than T1EC (RR 6.12, $p < 0.0001$). Nonetheless, the absolute risk remained low. Tamoxifen exposure was significantly more prevalent amongst T2EC patients (adjusted odds ratio 79.61, $p = 0.003$). Increased age at BC diagnosis was associated with T1EC (adjusted odds ratio 1.10, $p = 0.043$) and T2EC (adjusted odds ratio 1.13, $p = 0.03$). Neither smoking status nor family history of BC was significantly associated with any outcome.

Conclusion Women with BC were more likely to develop T2EC than T1EC. The absolute risk was low, but the cumulative risk was substantial enough to consider prophylactic hysterectomy in the premenopausal group. Tamoxifen exposure was significantly predictive of EC, particularly T2EC, and might facilitate risk estimation. Older women at BC diagnosis who received tamoxifen treatment should be screened and closely monitored for EC. Clarification of the menopausal status will help make more meaningful recommendations.

Disclosures All authors state that they do not have any conflicts of interest.