Introduction

Breast cancer is still the most commonly diagnosed cancer (2.08 million new cases) and the leading cause of cancer death (0.66 million deaths) among females (1). Consistently, in China, breast cancer statistics is identical with that worldwide. Approximately 11% of worldwide breast cancer occurs in China and that the incidence has increased rapidly in recent decades (2). However, breast cancer in China is not comprehensively understood compared with
About 5–10% of breast cancer burden follows a Mendelian inheritance pattern and is characterized as hereditary (3). The breast cancer-associated genes BRCA1 on chromosome 17q and BRCA2 on chromosome 13q are the most well-known breast cancer susceptibility genes, indicating from high-risk breast cancer families and influencing both treatment options and clinical management (4,5). In United States, the average cumulative risks of breast cancer in BRCA1 and BRCA2 mutation carriers by age 70 years are in the ranges of 60–70% and 45–55% (6). However, multigene panel testing of breast cancer predisposition genes have been extensively conducted in Europe and America, which is relatively rare in Asia region. In China, rare large-scale research has been conducted to examine the BRCA1/2 mutations. In 2015, we participated into the multicenter study by Li et al. and assessed the frequency of germline mutations in 40 cancer predisposition genes (7). Finally, we acquired 159 patients with BRCA1/2 mutations among 937 Chinese breast cancer patients with high hereditary risk. However, the characteristics of BRCA1/2 mutation subtypes, as well as its association with clinicopathological features need further discussion.

In this study, we reported the screening results of BRCA1/2 mutation in our center and analyzed the association of BRCA1/2 mutation with clinicopathological characteristics. Based on the special genome mutation, we reviewed the literature and discussed their possible mechanism in breast cancer incidence and prognosis.

Methods

Participants

Between November 2015 and May 2016, breast cancer patients admitted to Xijing Hospital with high hereditary risk were recruited. Inclusion criteria: (I) onset age ≤35 years (early-onset breast cancer); (II) at least one first or second-degree relative with breast cancer, ovarian cancer (OC), primary peritoneal cancer, or fallopian tube carcinoma; (III) two primary breast cancer; (IV) male breast cancer; (V) breast cancer with OC, fallopian tube carcinoma or primary peritoneal cancer. This protocol was approved by the Ethics Committee of the Xijing Hospital of The Fourth Military Medical University (No. KY20150916-4) and undertaken in accordance with the Good Clinical Practice guidelines and the Declaration of Helsinki. All patients were fully consented and asked to provide written informed consent before enrollment.

BRCA1/2 screening

Participants were asked for 5 mL fresh peripheral venous blood, which was transferred into a coded Ethylenediaminetetraacetic Acid (EDTA) tube at 4 °C. Then, the collected blood samples were sent to Annoroad Gene Technology (Beijing, China) Co. Ltd. for gene testing on an Illumina HiSeq 2500 platform (Illumina, San Diego, CA, USA), as previous described (7). The transcripts of BRCA1 and BRCA2 were NM_007294 and NM_000059, including 49 coding regions, 160 thousand of nucleotide sites. The main endpoints were single base polymorphisms (SNPs) in gene coding region and the base sequence insertion or deletion. Bioinformatics analysis was used to obtain mutant sites. BRCA1/2 mutations were explained based on the references included in comprehensive database (ClinVar and BIC database), clinical practice guideline and the latest medical papers.

Statistical analysis

SPSS 22.0 for windows was used for statistical analysis. All participants were followed up by specified research nurses and there are two investigators independently extracted the data using a predesigned data extraction form. Normal descriptive data were represented by mean ± standard deviation. Student’s t-test was used to compare the mean after checking the homogeneity of variance. Enumeration data was compared using χ² test, and unadjusted odds ratio and adjusted odd ratio were estimated by logistic regression for each indictor. P<0.05 was considered statistically significant.

Results

Baseline characteristics

Between November 2015 and May 2016, 82 patients (77 females and 5 males) were assessed for eligibility. The basic features were presented in Table 1. The average age was 35.7±9.04 years old. For menopausal status, 76 (92.7%) patients were premenopausal, while 6 (7.3%) of them were postmenopausal. Besides, the expression of ER (estrogen receptor), PR (progesterone receptor), HER2 (human epidermal growth factor receptor type 2) and Ki67 were investigated. Enrolled patients were proved with high
hereditary risk. Family breast cancer history and other cancer history were found in 23 (28.0%) and 35 (42.7%) of them.

Results of BRCA1/2 mutation

Twenty participants (24.4%) were found with BRCA1/2 mutation among 82 patients with high risk (Table 2). Eight patients were detected with BRCA1 mutation, 13 patients were detected with BRCA2 mutation. There is only one case with both BRCA1 and BRCA2 mutation (BRCA2: c.2971A>G (A/G type) mutation indicating good prognosis was found in 8 participants. Mutations about drug-sensitivity, including BRCA1: c.4837A>G (G/G type) and BRCA1: c.2612C>T (T/T type), were explored in 5 patients. G/G genome type and T/T genome type seem to present together. Pathogenic variant (BRCA1 c.4485-2A>C, BRCA1 c.5470-5477del, BRCA1: c.190T>C or BRCA2: c.3109G>T) and potential pathogenic variant (BRCA2: c.31delT, BRCA2: c.6408delA, BRCA2: c.6705delG or BRCA2: c.677delC) were detected in 8 participants. Above all, we found that prognosis-related mutations were enriched in BRCA2 gene, while drug-sensitive related mutations were always observed in BRCA1 gene.

Description of the mutation types in ClinVar and BIC databases

Searching the mutant genomic types in ClinVar and BIC databases, we found some potential novel mutant types in BRCA2 gene, such as BRCA2 c.31delT, BRCA2 c.6408delA, BRCA2 c.6705delG and BRCA2 c.677delC genome types (Table 3).

Association of BRCA1/2 mutation with clinicopathological characteristics

To explore the relationship between BRCA1/2 mutation and clinical features, we classified participants into mutation group and wild group. As shown in Table 4, HER2 expression, family cancer history and family breast/OC history were significantly different between BRCA1/2 mutation group and BRCA1/2 wild group. Multiple logistic analysis showed that HER2 [odds ratio (OR) 4.58; 95% confidence interval (CI), 1.182–17.74; P=0.028] was independent factor for BRCA1/2 mutation (Table 5). Among 32 patients with positive HER2, only 9% (3/32) of them were detected with BRCA1 or BRCA2 mutation. But...
Table 2 List of patients with BRCA1/2 mutation

| No. | Sex   | BMI  | Age | Pathology | Family cancer history | Cancer type | Pathology | Mutation                                | Genome type | Effects |
|-----|-------|------|-----|-----------|-----------------------|-------------|----------|-----------------------------------------|-------------|---------|
| 1   | Female | 23.37| 41  | IBC       | Yes                   | OC          | BRCA1: c.4485-2A>C | A/C         | DM         |
| 2   | Female | 24.35| 30  | IBC + IDC | No                    | –           | BRCA1: c.5470-5477 del | TGGCGCAAT/- | DM         |
| 3   | Female | 27.39| 43  | IBC       | Yes                   | BC          | BRCA1: c.190T>C | T/C         | DM         |
| 4   | Female | 26.91| 35  | IBC       | Yes                   | GC          | BRCA1: c.4837A>G | G/G; T/T    | A; B       |
| 5   | Female | 30.12| 47  | IDC       | Yes                   | OC          | BRCA1: c.4837A>G | G/G; T/T    | A; B       |
| 6   | Female | 20.06| 62  | IDC       | Yes                   | OC          | BRCA1: c.4837A>G | G/G; T/T    | A; B       |
| 7   | Female | 20.94| 33  | IBC       | No                    | –           | BRCA1: c.4837A>G | G/G; T/T    | A; B       |
| 8   | Female | 21.08| 37  | IBC       | No                    | –           | BRCA2: c.2971A>G | A/G         | GP         |
| 9   | Female | 21.88| 34  | IBC       | No                    | –           | BRCA2: c.2971A>G | A/G         | GP         |
| 10  | Female | 20.69| 38  | IBC       | Yes                   | BC          | BRCA2: c.2971A>G | A/G         | GP         |
| 11  | Female | 24.24| 35  | IBC       | No                    | –           | BRCA2: c.2971A>G | A/G         | GP         |
| 12  | Female | 21.48| 39  | IBC       | Yes                   | BC          | BRCA2: c.2971A>G | A/G         | GP         |
| 13  | Female | 23.74| 31  | MBC       | No                    | –           | BRCA2: c.2971A>G | A/G         | GP         |
| 14  | Female | 22.10| 38  | IBC       | No                    | –           | BRCA2: c.2971A>G | A/G         | GP         |
| 15  | Female | 21.48| 28  | IBC       | Yes                   | BC          | BRCA2: c.2971A>G | A/G; G/G; T/T| GP; A; B    |
| 16  | Female | 19.71| 31  | IBC       | Yes                   | BC          | BRCA2: c.31delT  | T/-         | PDM        |
| 17  | Female | 22.43| 36  | IBC       | Yes                   | BC          | BRCA2: c.6408delA | A/-         | PDM        |
| 18  | Female | 27.34| 51  | IDC       | No                    | –           | BRCA2: c.6705delG | G/-         | PDM        |
| 19  | Female | 24.65| 46  | IBC       | Yes                   | BC, EC      | BRCA2: c.677delC | C/-         | PDM        |
| 20  | Female | 20.32| 59  | IBC       | Yes                   | BC*4        | BRCA2: c.3109C>T | C/T         | DM         |

IBC, invasive breast carcinoma; IDC, invasive ductal carcinoma; MBC, mucinous breast carcinoma; OC, ovarian cancer; BC, breast cancer; GC, gastric cancer; EC, esophagus cancer; DM, detrimental mutation; PDM, potential detrimental mutation; GP, good prognosis; A, sensitive to platinum drugs; B, prolonged survival for cisplatin + paclitaxel regimen.

for 50 cases of HER2 negative breast cancer patients, 34% (17/50) were found mutant BRCA1 or BRCA2 gene.

**Discussion**

BRCA1/BRCA2 mutations have been identified as main contributor of hereditary breast cancer, increasing the lifetime risk of breast cancer in women (8). However, this paradigm has not been studied extensively and accurately in China. In this study, we investigated the BRCA1/2 mutation rate and mutation features for breast cancer patients in northwest China. The mutation rate of BRCA1/2 in high hereditary risk breast cancer patients was 24.4%, which was similar to that in other regions. In 2015, Riahi et al. estimate 25% pathogenic mutations in BRCA1/2 genes in early-onset and familial breast/OC among Tunisian women (9). In 2016, Cao et al. observed a total mutation frequency of 23.3% in BRCA1 and BRCA2 genes among patients in eastern China (10). Differently, BRCA2 mutations seem to be a few more than BRCA1 mutations (11).
Table 3 Searching of ClinVar and BIC databases

| Gene  | Mutant     | Exon | Nucleotide | AA change  | BIC entries | Clinvar entries | Novel genome |
|-------|------------|------|------------|------------|-------------|----------------|--------------|
| BRCA1 | c.4485-2A>C | 14   | 4485-2     | Asn to His | Yes         | Yes            | No           |
|       | c.5470-5477del | 23   | 5470-5477  | Frameshift | 2           | 3              | No           |
|       | c.190T>C    | 64   | 190        | Cys to Arg | 14          | 4              | No           |
|       | c.4837A>G   | 15   | 4837       | Ser to Gly | None        | None           | No           |
|       | c.2612C>T   | 10   | 2612       | Pro to Leu | None        | None           | No           |
| BRCA2 | c.3109C>T   | 11   | 3109       | p.Q1037X   | 14          | 6              | No           |
|       | c.2971A>G   | 11   | 2971       | Asn to Asp | None        | None           | No           |
|       | c.31delT    | –    | 31         | Frameshift | None        | None           | Yes          |
|       | c.6408delA  | –    | 6408       | Frameshift | None        | None           | No           |
|       | c.6705delG  | –    | 6705       | Frameshift | None        | None           | Yes          |
|       | c.677delC   | –    | 677        | Frameshift | None        | None           | Yes          |

2013, Blay et al. found 59 (23%) families with pathogenic germ line mutation, 39 in BRCA1 and 20 in BRCA2, in hereditary breast and OC families from Asturias (Northern Spain) (12). However, this is a very selective cohort. More convincing data of BRCA1/2 mutations should be achieved in cohort with larger samples.

In this study, we found 8 pathogenic variants in BRCA1 and BRCA2 genes, some of which have been reported previously. In 2001, BRCA1: c.4485-2A>C (BRCA1 A/C genome type) alteration was found among OC families in Japan and was determined as harmful mutation (13). Mutation of BRCA1: c.5470-5477del (TGCCCAAT/-type) has been observed in several studies. In 2004, Choi et al. found BRCA1: c.5470-5477del mutation and described it as c.5589del8, in Korean breast cancer families (14). Similarly, BRCA1: c.5470-5477del mutation was detected in 2 out of 645 women from Shanghai, China (15). In 2007, BRCA1: c.5470-5477del mutations were observed in several cases of breast cancer patients and were regarded as the possible founder mutations for Chinese population (16). As founder mutations of Italian people, the detrimental function of BRCA1: c.190T>C (BRCA1 T/C genome type) mutation was verified in two studies from Italy (17,18). BRCA2: c.3109C>T (BRCA2 C/T genome type) mutation was explored and was regarded as the founder mutations of Southern Chinese people (19-21). Importantly, 4 potential harmful mutations, including BRCA2: c.31delT (T/-), BRCA2: c.6408delA (A/-), BRCA2: c.6705delG (G/-) and BRCA2: c.677delC (C/-) were detected in the study, which have never been reported before. In 2015, Rebbeck et al. exclaimed that c.31delT, c.6408delA, c.6705delG and c.677delC genome types located in BRC domain (c.3006-6255) and DNA binding domain (c.7437-8001) of BRCA2 gene (16). Frame-shifting mutation induced sequence changing of DNA binding domain and OB (oligonucleotide-binding) folds domain in BRCA2 gene was the possible mechanism. DNA binding domain and OB folds domain have been reported to participate into the repair of double-strand DNA breaks (DSBs) by homologous recombination (22).

BRCA1 gene, involving homologous recombination, nonhomologous end joining, and mismatch repair, plays crucial role in regulating DNA damage induced by DNA-damaging agents such as platinum (23). Patients with lower BRCA1 expression obtain better survival after platinum-based neoadjuvant chemotherapy (24). In 2010, Shim demonstrated significant prolongation of overall survival (OS) and progression-free survival (PFS) in advanced gastric cancer patients with BRCA1 T/T mutation, after treating with taxane and cisplatin regimen. In 2010, Shim demonstrated significant prolongation of overall survival (OS) and progression-free survival (PFS) in advanced gastric cancer patients with BRCA1 T/T mutation, after treating with taxane and cisplatin regimen. For non-small cell lung cancer patients treated with first-line paclitaxel-cisplatin chemotherapy, BRCA1 T/T mutation was proved as modest prognostic
### Table 4 Relationship between BRCA1/2 mutation and clinicopathological characteristics

| Item                              | Mutation group (N=20) | Wild group (N=62) | $\chi^2 / F$ | P    |
|-----------------------------------|-----------------------|-------------------|-------------|------|
| Sex                               |                       |                   | 1.718       | 0.19 |
| Female                            | 20                    | 57                |             |      |
| Male                              | 0                     | 5                 |             |      |
| Age                               | 39.7±9.04             | 34.2±8.61         | 0.813       | 0.37 |
| BMI                               | 23.22±2.80            | 22.6±3.41         | 0.826       | 0.366|
| Number of pregnancy               | 1.45 [0–4]            | 1.53±1.30         | 0.881       | 0.351|
| Lactation                         |                       |                   | 1.373       | 0.241|
| Yes                               | 16                    | 41                |             |      |
| No                                | 4                     | 21                |             |      |
| Menopausal status                 |                       |                   | 2.302       | 0.129|
| Premenopause                      | 17                    | 59                |             |      |
| Postmenopause                     | 3                     | 3                 |             |      |
| Menstruation                      |                       |                   |             |      |
| Age of menarche                   | 13.8±1.61             | 13.4±1.31         | 1.595       | 0.210|
| Menstrual cycle                   | 28.53±3.65            | 28.8±1.91         | 3.224       | 0.076|
| Estrogen receptor                 |                       |                   | 0.009       | 0.926|
| Positive                          | 13                    | 41                |             |      |
| Negative                          | 7                     | 21                |             |      |
| Progesterone receptor             |                       |                   | 0.011       | 0.918|
| Positive                          | 12                    | 38                |             |      |
| Negative                          | 8                     | 24                |             |      |
| HER2                              |                       |                   | 6.416       | 0.011|
| Positive                          | 3                     | 29                |             |      |
| Negative                          | 17                    | 33                |             |      |
| Ki67                               |                       |                   | 0.984       | 0.321|
| ≤20                               | 5                     | 23                |             |      |
| >20                               | 15                    | 39                |             |      |
| Pathology                         |                       |                   | 2.092       | 0.351|
| Carcinoma in situ                 | 0                     | 6                 |             |      |
| Invasive carcinoma                | 19                    | 53                |             |      |
| Other                             | 1                     | 3                 |             |      |
| Family cancer history             | 12                    | 21                | 4.293       | 0.038|
| Family breast/ovarian cancer history | 11                    | 16                | 5.836       | 0.016|
| Family other cancer history       | 2                     | 7                 | 0.026       | 0.872|
markers (27).

This study has several limitations. First, the sample size was relatively small. Second, there is no follow-up data because of the short follow-up period. With follow-up continued, the final prognosis and survival data will be gained. Third, only a limited number of SNPs in \textit{BRCA1} and \textit{BRCA2} genes were enrolled into detection, while the other polymorphisms in these genes may be important.

In conclusion, the \textit{BRCA1}/2 mutation features in our hospital and the total mutation rate of \textit{BRCA1}/2 in high hereditary risk breast cancer patients was similar to that in other regions. HER2 expression was independent factor for \textit{BRCA1} and \textit{BRCA2} mutation for breast cancer with high risk. Four novel genome types, including \textit{BRCA2}: c.31delT (T/-), \textit{BRCA2}: c.6408delA (A/-), \textit{BRCA2}: c.6705delG (G/-) and \textit{BRCA2}: c.677delC (C/-), might be potential harmful mutations, which needs further verification.

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### Footnote

**Conflicts of Interest:** All authors have completed the ICMJE uniform disclosure form (available at https://dx.doi.org/10.21037/tcr.2019.08.32). The authors have no conflicts of interest to declare.

**Ethical Statement:** The authors are accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. This protocol was approved by the Ethics Committee of the Xijing Hospital of The Fourth Military Medical University (No. KY20150916-4) and undertaken in accordance with the Good Clinical Practice guidelines and the Declaration of Helsinki (as revised in 2013). All patients were fully consented and asked to provide written informed consent before enrollment.

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