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BRCA1 testing

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Mutations in *BRCA1* confer a high lifetime risk for both breast and ovarian cancer. Many different *BRCA1* mutations have been described in families with early-onset breast and ovarian cancer [1, 2]. The presence of recurrent mutations in *BRCA1* suggests the presence of founder effects. *BRCA1* "de novo" mutations are very rare. 66 families were studied in Szczecin with strong aggregations of breast/ovarian cancers. Mutations were found in 35 (53%) of the 66 families. Three *BRCA1* abnormalities–5382insC, C61G, and 4153delA–accounted for 51, 20, and 11% of the identified mutations, respectively [3]. Similar results were reported in Gliwice, Gdańsk and Poznań centres [4-6]. Additional evaluation of 200 families originating from all regions of Poland (with at least 3 breast/ovarian cancers) revealed that constitutional *BRCA1* mutations can be detected in 64% (128/200) of these families, and 90% of all of them carried one of the 3 common founder mutations (5382insC, C61G and 4153delA) [7]. Genetic testing for these *BRCA1* mutations is inexpensive and relatively simple in Poland (~100 Euro including genetic counselling). It is based on the "multiplex PCR" method with almost 100% specificity in detection of three common Polish *BRCA1* mutations. In 2006, 3500 unselected incident cases of early-onset breast cancers were screened for presence of the three common Polish *BRCA1* mutations. The proportion of cases with an identified mutation was 5.7% [8]. In a similar study of 500 unselected ovarian cancers, common *BRCA1* mutations were detected in 13% of cases [9, 10]. It is possible to increase women's awareness about hereditary cancer through the popular press. Genetic testing was offered to 5000 Polish women through an announcement placed in a popular women's magazine „Twój Styl". A total of 5024 women who qualified received a free genetic test for three mutations in *BRCA1* which are common in Poland. Out of these, 198 women (3.9%) were found to carry a *BRCA1* mutation. Genetic testing for *BRCA1* mutations in Poland should be recommended for adult females who:

- fulfil clinical criteria of hereditary breast/ovarian cancer,
- are affected with breast or ovarian cancer,
- are healthy but have at least one relative with breast/ovarian cancer (breast cancer diagnosed under age 50).

Genetic testing for presence of *BRCA1* mutations should be preceded and followed by genetic counselling. So far nearly 4500 *BRCA1* mutation carriers have been detected at the International Hereditary Cancer Centre in Szczecin (the world's largest registry of females diagnosed and under surveillance by a single centre).

We believe that the "Polish model" of solving the problem of *BRCA1/BRCA2* testing can be valuable for many populations with a relatively high level of genetic homogeneity.

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