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FULL-TEXT ARTICLE

Genetic epidemiology of BRCA1 mutations in Norway.

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Abstract

Familial breast-ovarian cancer has been demonstrated to be frequent but unevenly distributed in Norway. This was assumed to be caused by the reduced population size created by the medieval Bubonic plagues 25 generations ago, and by the following rapid expansion. We have previously reported that four mutations account for 68% of the BRCA1 mutation carriers. Subsequent analysis has resulted in a total of 100 separate families carrying one of these founder mutations. The four mutations occurred on one specific BRCA1 haplotype each. The 1675delA, 816delGT and 3347delAG families originated from the South-West coast of Norway with a few families in the north, while the traceable ancestors of the 1135insA families clustered along the historical inland road from the South-East to mid-Norway. The carriers of each of the four mutations today are descendants of one or a few individuals surviving the plagues. We may identify the majority of BRCA1 mutation carriers in Norway by screening for local founder mutations.

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Publication type, MeSH terms

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