A HYBRID APPROACH TO EVALUATION OF BRCA MUTATIONS PREVALENCE FOR ASHKENAZI JEWISH WOMEN

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Many studies have estimated the prevalence of BRCA1/2 mutations in Ashkenazi Jewish women conditional on the individual's personal and family history of cancer. In an attempt to summarize all relevant data we extracted the estimates of prevalence reported for various subpopulations from 52 papers. Each subpopulation was defined by a 'profile' i.e. by a specific combination of personal and family history characteristics. The main difficulty in this process arose from the great variation across studies in the specifications of these profiles. To overcome the problem we used a 'hybrid' approach, combining data from the literature with the data on our own sample of 989 Israeli women. We first developed a standardized description of personal and family history, in such a way that any specified profile could be considered as a subset of cells in a multi-dimensional table. Using our sample we estimated the odds ratio of BRCA1/2 mutations and the frequency for each cell in the table. We then estimated the prevalence of BRCA 1/2 mutations in each cell by 'distributing' the estimated prevalence of each specified profile across its constituent cells using the odds ratios and frequencies from the sample.
In the presentation we will discuss the assumptions, details of the calculations and the results obtained.