Myriad Genetic Laboratories, Inc.

Mutation Prevalence Tables



The below tables represent observations of deleterious mutations by Myriad Genetic Laboratories through its clinical testing service, reflective of our current test offering. Data obtained through testing performed under specific research protocols is not included here. The information included in these tables was obtained from a routine laboratory requisition form and has not been independently verified by Myriad Genetic Laboratories. Patients for whom relevant information was not provided were not included in this tabulation. Table 2 includes patients tested only for three prevalent founder mutations as well as patients tested by full sequence analysis. The method used to develop the prevalence tables has been published in, Frank TS et al: Clinical Characteristics of Individuals With Germline Mutations in BRCA1 and BRCA2: Analysis of 10,000 Individuals. Journal of Clinical Oncology. 20:1480-1490, 2002. Please call 1-800-469-7423 with any questions or comments.

1. The Prevalence of Deleterious Mutations in BRCA1 and BRCA2 (Excludes Individuals of Ashkenazi Ancestry)

	Family History (Includes at least one first- or second-degree relative)					
Patient's History	No breast cancer <50, or ovarian cancer in any relative†	Breast cancer <50 in one relative; no ovarian cancer in any relative	Breast cancer <50 in more than one relative; no ovarian cancer in any relative	Ovarian cancer at any age in one relative; no breast cancer <50 in any relative	Ovarian cancer in more than one relative; no breast cancer <50 in any relative	Breast cancer <50 and ovarian cancer at any age ^{††}
No breast cancer or ovarian cancer at any age	1.5%	2.6%	5.6%	3.0%	5.3%	7.2%
Breast cancer ≥50	2.2%	3.8%	8.0%	4.9%	9.5%	10.6%
Breast cancer <50	4.7%	10.4%	21.2%	10.3%	21.9%	26.6%
Male breast cancer	6.9%	17.4%	36.6%	15.9%	*33.3%	28.3%
Ovarian cancer at any age, no breast cancer	7.7%	14.3%	27.4%	14.7%	22.7%	34.4%
Breast cancer ≥50 and ovarian cancer at any age	12.1%	23.6%	50.0%	23.6%	44.2%	39.4%
Breast cancer <50 and ovarian cancer at any age	26.3%	40.0%	64.5%	41.2%	45.5%	57.4%

[†]May include families with breast cancer ≥50 (in women or men)

Number of observations in Table 1 is 162,914

2. The Prevalence of Deleterious Mutations in BRCA1 and BRCA2 in Individuals of Ashkenazi Ancestry

Family History (Includes at least one first- or second-degree relative) Ovarian cancer Ovarian cancer No breast Breast cancer Breast cancer <50 <50 in more than cancer <50, at any age in one in more than one or ovarian in one relative; no one relative; no relative; no breast relative; no breast Breast cancer <50 cancer <50 in cancer <50 in cancer, in ovarian cancer in ovarian cancer in and ovarian cancer Patient's History any relative† any relative any relative any relative any relative at any age^{††} No breast cancer 8.2% 13.0% 16.4% 12.7% 22.3% 22.9% or ovarian cancer at any age Breast cancer ≥50 3.3% 7.1% 10.8% 13.2% 13.6% 16.7% Breast cancer <50 7.9% 17.5% 26.9% 18.1% 20.0% 33.0% *46.2% Male breast cancer 13.5% 26.8% *21.1% *66.7% *55.6% Ovarian cancer at 16.2% 26.4% 47.4% 26.2% any age, no breast 57.1% 57.8% cancer Breast cancer ≥50 20.5% 18.2% *30.0% *31.3% *100.0% *55.6% and ovarian cancer at any age Breast cancer <50 and ovarian cancer 42.1% *63.2% *85.7% *62.5% *100.0% *36.4% at any age

In Situ data is now included in the "affected" categories

Number of observations in Table 2 is 26,015

^{††}Includes family members with either or both diagnoses

^{*}N<20

In Situ data is now included in the "affected" categories

[†]May include families with breast cancer ≥50 (in women or men)

^{††}Includes family members with either or both diagnoses