

Article

Spectrum of Germline Pathogenic Variants in BRCA1/2 Genes in the Apulian Southern Italy Population: Geographic Distribution and Evidence for Targeted Genetic Testing

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Supplementary materials:

Table S1. List of VUS identified in the analyzed cohort.

| BRCA1 | BRCA2 |
|---------------------|----------------|
| p.Pro1776Ser | p.Asp2566Glu |
| p.Arg1347Gly | p.Glu1811Ala |
| p.Arg1528Thr | p.Ile1298Thr |
| p.Asn1354Ser | p.Alala1170Val |
| p.Asp1152Asn | p.Alala1797Gly |
| p.Asp1435Gly | p.Alala1991Val |
| p.Asp67Tyr | p.Alala2351Thr |
| p.Asp96Gly | p.Alala3205Leu |
| p.Cys636Tyr | p.Arg155Thr |
| p.Gln1090Arg | p.Arg174His |
| p.Gln284Arg | p.Arg2108Cys |
| p.Gln284Arg | p.Arg2520Pro |
| p.Glu445Gln | p.Arg2787His |
| p.Glu761Gln | p.Arg3385His |
| p.Gly1201Cys | p.Asn1836Lys |
| p.His888Tyr | p.Asn1906ser |
| p.Ile1275Val | p.Asp125Glu |
| p.Lys970Asn | p.Asp191Asp |
| p.Pro1771Arg | p.Asp3112Asn |
| p.Pro568Leu | p.Asp3112Asn |
| p.Pro659Ala | p.Cys148Arg |
| p.Ser632Asn | p.Glu1110Gly |
| p.Ser784Leu | p.Glu1901Val |
| p.Thr333Ala | p.Glu2292Gly |
| p.Thr779Ala | p.Glu2961Ser |
| p.Val1378Ile | p.Glu2981Lys |
| p.Val1653Met | p.Glu3002asp |
| c.5468-10C>A | p.Gly2044Ala |
| c.67+8C>A | p.Gly267Glu |
| duplicazioneesone 3 | p.His1966Arg |
| c.135-3T>C | p.Ile1237Met; |
| | p.Ile1244Thr |
| | p.Ile1298Met |
| | p.Ile982Leu |

p.Leu914Phe
 p.Lys2013Glu
 p.Phe376Cys
 p.Phe590Cys
 p.Pro2589His
 p.Pro2796Ser
 p.Pro3194Gln
 p.Ser1328Thr
 p.Ser1468Thr
 p.Ser1750Phe
 p.Ser286Pro
 p.Ser3366Asnfs*4
 p.Thr1887Met
 p.Thr200Ile
 p.Thr2097Met
 p.Thr2222Cys
 p.Thr3288Ile
 p.Thr64Ile
 p.Tyr2624Cys
 p.Val1810Ile
 c.7435+6G>A
 c.7805+6C>T
 c.9502-12T>G
 c.4211G>C
 p.Ala1615Thr
 p.Ala1615Thr
 p.Arg278His

Table S2. Non coding PVs and Large Genomic Rearrangements (LGR).

| BRCA1 non-coding PVs and LGRs | <i>n</i> |
|--------------------------------------|-----------------|
| c.212+2T>A | 2 |
| c.547+2T>A | 1 |
| c.5153-1G>A | 4 |
| c.5278-2delA | 2 |
| exons 1-2 deletion | 3 |
| exons 1-12 deletion | 2 |
| exons 1-13 deletion | 1 |
| exons 1-20 deletion | 1 |
| promoter and exons 1-11 deletion | 1 |
| exon 7 deletion | 1 |
| exon 8 deletion | 2 |
| exon 19 deletion | 1 |
| exons 5-8 deletion | 1 |
| BRCA2 non-coding PVs and LGRs | <i>n</i> |
| c.67+1G>A | 2 |
| c.7976+1 G>A | 1 |
| c.8755-1G>A | 12 |
| exons 1-3 deletion | 1 |
| exon 14 deletion | 1 |