

Germline loss-of-function *BRCA1* and *BRCA2* mutations and risk of *de novo* hematopoietic malignancies

Ryan J. Stubbins,^{1,2,3*} Anase S. Asom,^{1*} Peng Wang,⁴ Angela M. Lager,⁴ Alexander Gary⁴ and Lucy A. Godley^{1,5}

¹Section of Hematology/Oncology, Department of Medicine, The University of Chicago, Chicago, IL, USA; ²Division of Hematology, Department of Medicine, University of British Columbia, Vancouver, British Columbia, Canada; ³Leukemia/BMT Program of BC, BC Cancer, Vancouver, British Columbia, Canada; ⁴Department of Pathology, The University of Chicago, Chicago, IL, USA; and ⁵Department of Human Genetics, The University of Chicago, Chicago, IL, USA

**RJS and ASA contributed equally as first authors.*

Correspondence:

L.A. GODLEY - lucy.godley@northwestern.edu

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Supplemental Material for :

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Supplemental Table 1. Germline and somatic *BRCA1/2* mutations detected in the University of Chicago patient cohort.

Supplemental Table 1 outlines the 35 patients comprising the University of Chicago patient cohort; their hematopoietic malignancies; germline *BRCA* mutations; and results of somatic mutation testing.

Abbreviations used: NA, not applicable.

Supplemental Table 1. Germline and somatic BRCA1/2 mutations detected in the University of Chicago patient cohort.

ID	Hematologic Dx #1	Hematologic Dx #2	Gene	cDNA	pChange	Transcript	Chr	GRCh38 Start	GRCh38 End	Variation ID	ClinVar or LoF Annotation	Somatic Testing from Home Mailig?	Additional cytogenetic abnormality or somatic BRCA mutation	Evaluable for loss of heterozygosity	Evidence of possible loss of heterozygosity
1	Myeloid	NA	BRCA1	Deletion exons 13-15	NA	NM_007294.3	NA	NA	NA	NA	Loss of function (LOF) - Large deletion	Not performed	Normal karyotype	Yes	No
2	Myeloid	NA	BRCA1	c.68_69del	p.Glu23fs	NM_007294.4	17	43124028	43124029	17662	Pathogenic	Not performed	Not performed	No	NA
3	Myeloid	NA	BRCA1	c.68_69del	p.Glu23fs	NM_007294.4	17	43124028	43124029	17662	Pathogenic	No coverage	Normal karyotype	Yes	No
4	Myeloid	NA	BRCA1	c.2989_2990dup	p.Asn97fs	NM_007294.4	17	43092540	43092541	54738	Pathogenic	Not performed	Normal karyotype	Yes	No
5	Lymphoid	Myeloid	BRCA1	c.5266_5267dup	p.Gln1756fs	NM_007294.4	17	43057061	43057062	988271	Pathogenic	Not performed	del(20q)	Yes	No
6	Myeloid	NA	BRCA1	c.68_69del	p.Glu23fs	NM_007294.4	17	43124028	43124029	17662	Pathogenic	Not performed	Not performed	No	NA
8	Myeloid	NA	BRCA1	c.3048_3052dup	p.Asn1018fs	NM_007294.4	17	43092478	43092479	54763	Pathogenic	No coverage	Normal karyotype	Yes	No
9	Lymphoid	NA	BRCA1	c.181T>G	p.Cys61Gly	NM_007294.4	17	43106487	43106487	17661	Pathogenic	Not performed	Not performed	No	NA
10	Myeloid	NA	BRCA1	Deletion exon 17	NA	NM_007294.3	NA	NA	NA	NA	Loss of function (LOF) - Large deletion	Yes	der(5;17)(p10;q10) and -17	Yes	Yes
12	Myeloid	NA	BRCA1	c.5266_5267dup	p.Gln1756fs	NM_007294.4	17	43057061	43057062	988271	Pathogenic	Not performed	Normal karyotype	Yes	No
13	Lymphoid	Myeloid	BRCA1	c.5503_11416del	NA	NM_007294.3	NA	NA	NA	NA	Loss of function (LOF) - Large deletion	Yes	Complex karyotype, no obvious loss of chr17	Yes	No
16	Myeloid	NA	BRCA1	Deletion exon 12	NA	NM_007294.3	NA	NA	NA	NA	Loss of function (LOF) - Large deletion	Not performed	Not performed	No	NA
17	Lymphoid	NA	BRCA1	c.427G>T	p.Glu143Ter	NM_007294.4	17	43104136	43104136	37581	Pathogenic	Not performed	Multiple clones but no loss of chr17	Yes	No
18	Lymphoid	NA	BRCA1	c.697_698del	p.Val233fs	NM_007294.4	17	43094833	43094834	37695	Pathogenic	Not performed	Not performed	No	NA
19	Myeloma	Lymphoid	BRCA2	c.5828del	p.Ser1943fs	NM_000059.3	13	32340183	32340183	37999	Pathogenic	No coverage	Not performed	No	NA
20	Myeloid	NA	BRCA2	c.7977-1G>C	NA	NM_000059.4	13	32363178	32363178	38132	Pathogenic	No coverage	No chr13 abnormality	Yes	No
22	Myeloma	NA	BRCA2	c.6535_6536ins8	NA	NM_000059.3	13	32340890	32340891	126108	Pathogenic	Not performed	Clone with +13	Yes	unknown
23	Myeloid	NA	BRCA2	c.1753A>T	p.Lys585Ter	NM_000059.4	13	32333231	32333231	487414	Pathogenic/Likely Pathogenic	Not performed	Not performed	No	NA
25	Myeloid	NA	BRCA2	c.5073dup	p.Trp1692fs	NM_000059.4	13	32339421	32339422	37943	Pathogenic	Yes	Not performed	Yes	No
28	Myeloid	NA	BRCA2	c.5946del	p.Ser1982fs	NM_000059.4	13	32340301	32340301	9325	Pathogenic	Not performed	Not performed	No	NA
29	Myeloid	NA	BRCA2	c.8673_8674del	p.Arg2892fs	NM_000059.4	13	32376710	32376711	52656	Pathogenic	Yes	Normal karyotype, also BRCA2 p.D1420Y (Benign)	Yes	No
32	Myeloid	NA	BRCA2	c.4638del	p.Phe1546fs	NM_000059.4	13	32338989	32338989	37915	Pathogenic	Yes	No chr13 abnormality	Yes	No
33	Myeloma	NA	BRCA2	c.8243G>A	p.Gly2748Asp	NM_000059.4	13	32363445	32363445	52535	Pathogenic	Not performed	Clone with -13	Yes	Yes
34	Lymphoid	NA	BRCA2	c.6468_6469del	p.Gln2157fs	NM_000059.3	13	32340817	32340818	38047	Pathogenic	Not performed	Normal karyotype	Yes	No
35	Myeloma	NA	BRCA2	c.8817_8820del	p.Lys2939fs	NM_000059.4	13	32379377	32379380	52684	Pathogenic	Yes	BRCA2 c.8814_8822del (Pathogenic) at VAF 27%	Yes	Yes