

遺伝性乳癌卵巣癌（HBOC）診療



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遺伝性乳癌卵巣癌 (HBOC) 診療ガイドライン

2024年版

Guidelines for Diagnosis and Treatment of Hereditary Breast and Ovarian Cancer 2024



日本遺伝性乳癌卵巣癌総合診療制度機構
Japanese Organization of Hereditary Breast and Ovarian Cancer

編

HBOCガイドラインが新しくなりました

どのような乳癌患者にBRCA遺伝学的検査を推奨するか？

ステートメント

以下の基準に合致する乳癌患者に対し、BRCA遺伝学的検査を推奨する。

- ①血縁者にすでにBRCA1/2 に病的バリエーション保持がわかっている
- ②既往歴・病理学的適応
 - 45歳以下の乳癌発症（全サブタイプ）
 - 60歳以下のトリプルネガティブ乳癌（TNBC）発症
 - 2個以上の原発性乳癌発症（両側，片側2箇所等）
 - 卵巣癌，卵管癌および腹膜癌を発症
 - 膵癌を発症
 - 男性乳癌を発症
- ③家族歴
 - 第三度近親者内*に乳癌または卵巣癌または膵臓癌発症者が1名以上いる
- ④治療適応
 - HER2陰性乳癌で化学療法歴がある手術不能や再発乳癌薬物療法，または再発高リスクの乳癌における術後薬物療法として，ポリ（ADP-リボース）ポリメラーゼ（PARP）阻害薬に対するコンパニオン診断の適格基準を満たす
- ⑤腫瘍組織プロファイリング検査やリキッドバイオプシーで，BRCA1または/かつBRCA2の生殖細胞系列の病的バリエーション保持が疑われる
- ⑥生殖細胞系列多遺伝子パネル検査（MGPT）で，BRCA1または/かつBRCA2の病的バリエーション保持が疑われる

* 第一度近親者：同胞，両親，子，第二度近親者：おじおば，祖父母，おいめい，第三度近親者：いとこ，孫，大おじ大おば

乳癌患者の検査基準に膵癌の病歴と家族歴が加えられた

乳癌

BQ

4

BRCA1/2病的バリエントを保持する乳癌患者に対し、乳房手術後放射線療法は推奨されるか？

ステートメント

乳房部分切除術が選択された場合には、術後放射線療法を行うことを推奨する。

乳房全切除術後に、再発リスクの高い患者では、BRCA1/2病的バリエントを保持していない乳癌患者と同様の臨床的適応に従って術後放射線療法を行うことを推奨する。

乳癌

BQ

4

BRCA1/2病的バリエントを保持する乳癌患者に対し，乳房手術後放射線療法は推

ステートメント

乳房部分切除術が選択された場合には，術後乳房全切除術後に，再発リスクの高い患者と同等の臨床的適応に従って術後

乳癌

CQ

3

BRCA1/2 病的バリエントを保持する乳癌患者に対し，乳房温存療法は推奨されるか？

推奨

BRCA1/2 病的バリエントを保持する乳癌患者において，温存乳房照射を必要とする乳房温存療法は条件付きで行わないことを推奨する。

推奨のタイプ：当該介入に反対する条件付きの推奨

エビデンスの確実性：中，合意率：100%（12/12名）

推奨の解説：本ガイドラインで実施したメタ解析の結果から，BRCA1/2 病的バリエントを保持する乳癌患者における乳房温存療法は，散发性乳癌患者に比べて温存乳房内再発率が高いことが示された。この傾向は観察期間が長くなるほど明確になることから，温存乳房内の新規乳癌の発症リスクは長期にわたって継続するものと推察された。ただし温存療法後の温存乳房内の新規乳癌発症のリスク，継続的な温存乳房のスクリーニングの必要性，また温存療法後の新規乳癌に対する乳房再建術の困難さ等を十分理解し，患者と医療従事者が十分なリスクコミュニケーションのうえshared decision making（SDM）を行い乳房温存療法を選択する場合には，これを否定しない。

乳癌

CQ

8

BRCA1/2 病的バリエントを保持する乳癌患者に対する周術期（術後）薬物療法に、PARP 阻害薬の投与は推奨されるか？

推奨

BRCA1/2 病的バリエントを保持する再発高リスク乳癌患者に対し周術期（術後）薬物療法に、PARP 阻害薬の投与を条件付きで推奨する。

推奨のタイプ：当該介入の条件付きの推奨

エビデンスの確実性：中、合意率：90%（9/10 名）

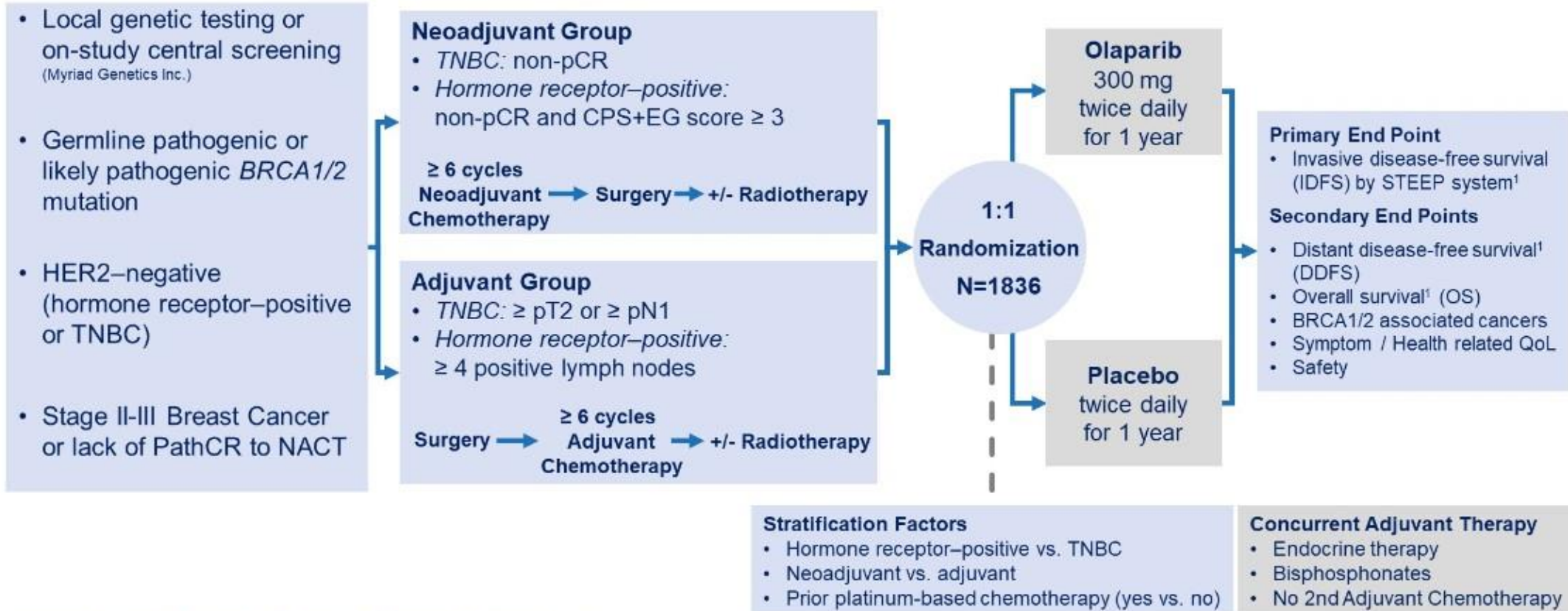
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ORIGINAL ARTICLE

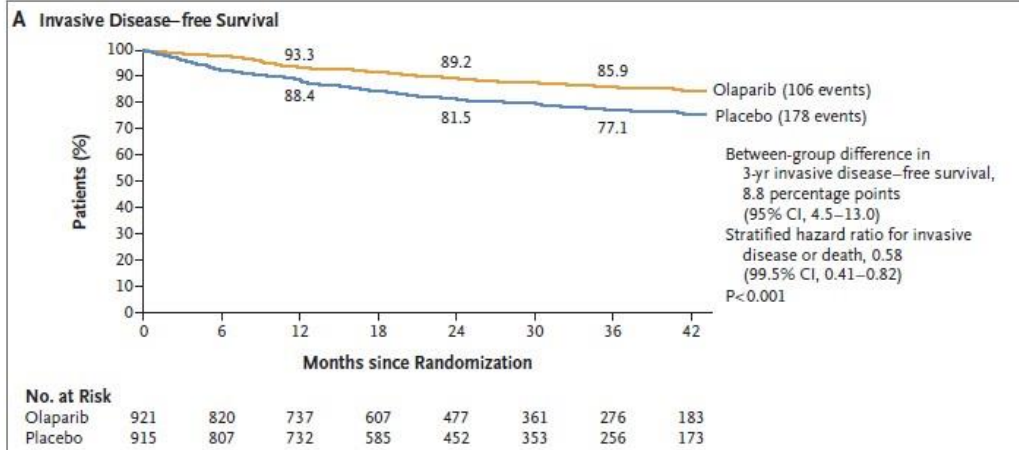
Adjuvant Olaparib for Patients with BRCA1- or BRCA2-Mutated Breast Cancer

A.N.J. Tutt, J.E. Garber, B. Kaufman, G. Viale, D. Fumagalli, P. Rastogi, R.D. Gelber, E. de Azambuja, A. Fielding, J. Balmaña, S.M. Domchek, K.A. Gelmon, S.J. Hollingsworth, L.A. Korde, B. Linderholm, H. Bandos, E. Senkus, J.M. Suga, Z. Shao, A.W. Pippas, Z. Nowecki, T. Huzarski, P.A. Ganz, P.C. Lucas, N. Baker, S. Loibl, R. McConnell, M. Piccart, R. Schmutzler, G.G. Steger, J.P. Costantino, A. Arahmani, N. Wolmark, E. McFadden, V. Karantza, S.R. Lakhani, G. Yothers, C. Campbell, and C.E. Geyer, Jr., for the OlympiA Clinical Trial Steering Committee and Investigators*

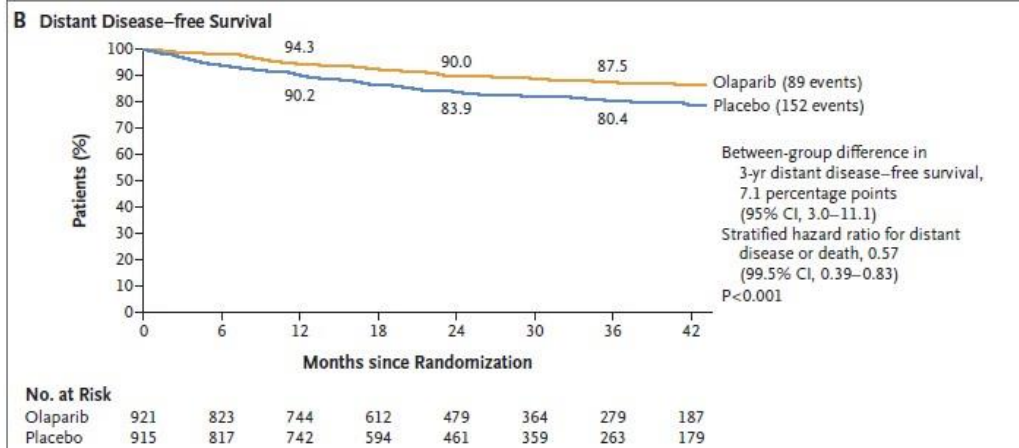
OlympiA: Trial schema



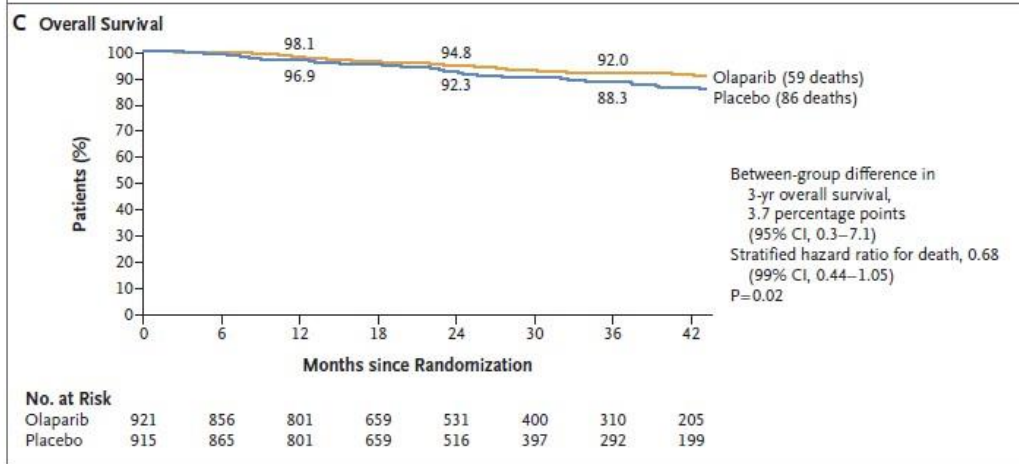
Hormone receptor +ve defined as ER and/or PgR positive (IHC staining $\geq 1\%$)
 Triple Negative defined as ER and PgR negative (IHC staining $< 1\%$)
¹Hudis CA, J Clin Oncol 2007



3年間の浸潤性無再発生存率はオラパリブ群85.9%、プラセボ群77.1%とオラパリブの投与によりIDFSの統計学的有意な3年IDFSの改善が認められた (p<0.001 ;HR, 0.58; 99.5% CI, 0.41-0.82)



3年間の遠隔無再発生存率はオラパリブ群87.5%、プラセボ群80.4%と遠隔無再発生存期間の延長が示された (P<0.001 ; HR, 0.57; 99.5% CI, 0.39-0.83)



生存率としてはオラパリブ群92%、プラセボ群88.3%となり有意差は認めなかった (P=0.02 ; HR0.68; 99% CI, 0.44 -1.05) 全生存期間 (Overall survival)への影響を確認するにはさらなる観察期間が必要と考えられた

BRCA1/2 病的バリエントを保持する乳癌患者で挙児希望がある場合に、生殖補助医療（ART）は推奨されるか？

ステートメント

BRCA1/2 病的バリエント保持乳癌患者における妊娠に関して、流産、早産や奇形の増加は認めない、また母体の予後を悪化させない等の、安全性に関するエビデンスが報告されている。

BRCA1/2 病的バリエント保持者において、特に乳癌等に罹患した場合には、将来妊娠・出産を希望し、原疾患の治療により卵巢機能の低下が予想される患者に対して、妊孕性温存療法として胚凍結（パートナーあり）、未受精卵子凍結（パートナーなし、若年者）、卵巢組織凍結を行うことが考慮される。しかしながら、それらの安全性ならびに有効性に関するエビデンスは限定的である。胚・未受精卵子凍結に関して、BRCA1/2 病的バリエント保持者では、卵巢予備能低下（卵胞数の減少）による採卵数ならびに凍結卵子数の低下に関する報告があるが、妊娠率や生児獲得率の低下につながる十分なエビデンスがあるとはいえない。

着床前遺伝学的検査（PGT-M）の選択肢がBRCA1/2 病的バリエント保持者にも生じ得るが、日本産科婦人科学会の見解における重篤性の定義からは、現時点でHBOCを理由とした申請がPGT-Mの対象として承認される可能性は低い。

Original Investigation

December 7, 2023

Pregnancy After Breast Cancer in Young *BRCA* Carriers

An International Hospital-Based Cohort Study

Matteo Lambertini, MD^{1,2}; Eva Blondeaux, MD³; Elisa Agostinetto, MD⁴; [et al](#)

» [Author Affiliations](#) | [Article Information](#)

JAMA. 2024;331(1):49-59. doi:10.1001/jama.2023.25463

FREE

2024 **ESMO BREAST CANCER**

Annual Congress

2660 – Safety of Assisted Reproductive Techniques in Young *BRCA* Carriers with a Pregnancy after Breast Cancer: Results from an International Cohort Study

Matteo Lambertini, Isotta Martha Magaton, Anne-Sophie Hamy, Sabine Linn, Rinat Bernstein-Molho, Fedro A. Peccatori, Alberta Ferrari, Estela Carrasco, Shani Paluch-Shimon, Elisa Agostinetto, Marta Venturelli, Ines Maria Vaz-Luis, Kenny A. Rodriguez-Wallberg, Hee Jeong Kim, Kimia Sorouri, Marco Bruzzone, Isabelle Demeestere, Hatem A. Azim Jr., Ann H. Partridge, Eva Blondeaux

Berlin (Germany)
May 16, 2024

  @matteolambe



ESMO daily news 2024より

No difference in risk recurrence or pregnancy outcomes was reported between young women with *BRCA1* or *BRCA2* mutations conceiving naturally and those using fertility treatments

For young women with a history of breast cancer harbouring *BRCA* mutations, undergoing assisted reproductive techniques (ART) did not appear to negatively influence maternal prognosis or pregnancy outcomes in a first analysis of a global study, as presented at the ESMO Breast Cancer 2024 (Berlin, 15–17 May) (Abstract 2660).

タラゾパリブ（製品名：ターゼナ）が承認されました



ターゼナ[®]カプセル製剤写真

がん化学療法歴のあるBRCA遺伝子変異陽性かつHER2陰性の手術不能又は再発乳癌：通常、成人にはタラゾパリブとして1日1回1mgを経口投与する

EMBRACA 試験



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ORIGINAL ARTICLE



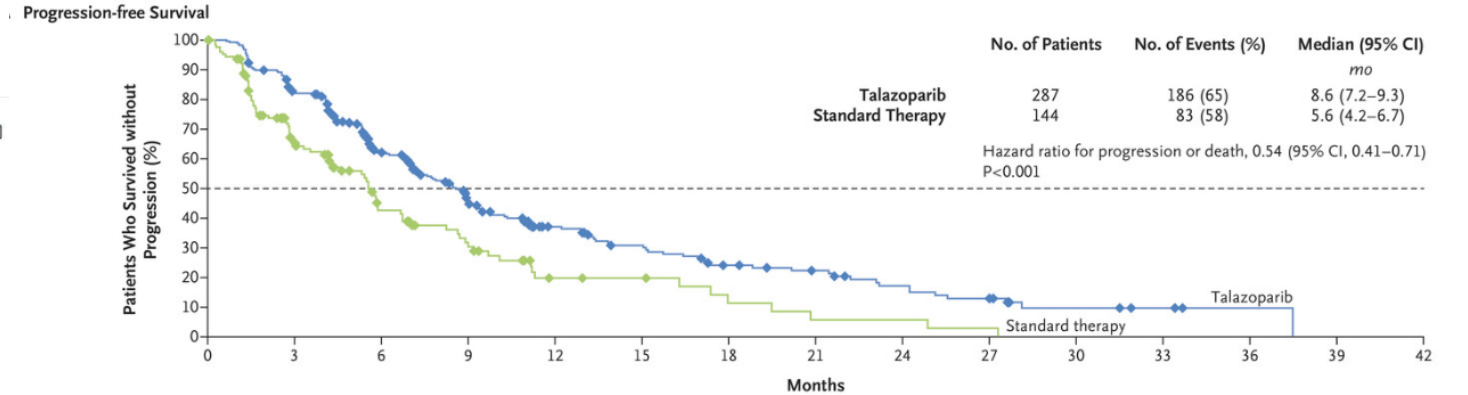
Talazoparib in Patients with Advanced Breast Cancer and a Germline BRCA Mutation

Authors: Jennifer K. Litton, M.D., Hope S. Rugo, M.D., Johannes Ettl, M.D., Sara A. Hurvitz, M.D., Anthony Gonçalves, M.D., Ph.D., Kyung-Hun Lee, M.D., Ph.D., Louis Fehrenbacher, M.D., [+10](#), and Joanne L. Blum, M.D., Ph.D. [Author Info & Affiliations](#)

Published August 15, 2018 | N Engl J Med 2018;379:753-763 | DOI: 10.1056/NEJMoa1802905 | VOL. 379 NO. 8

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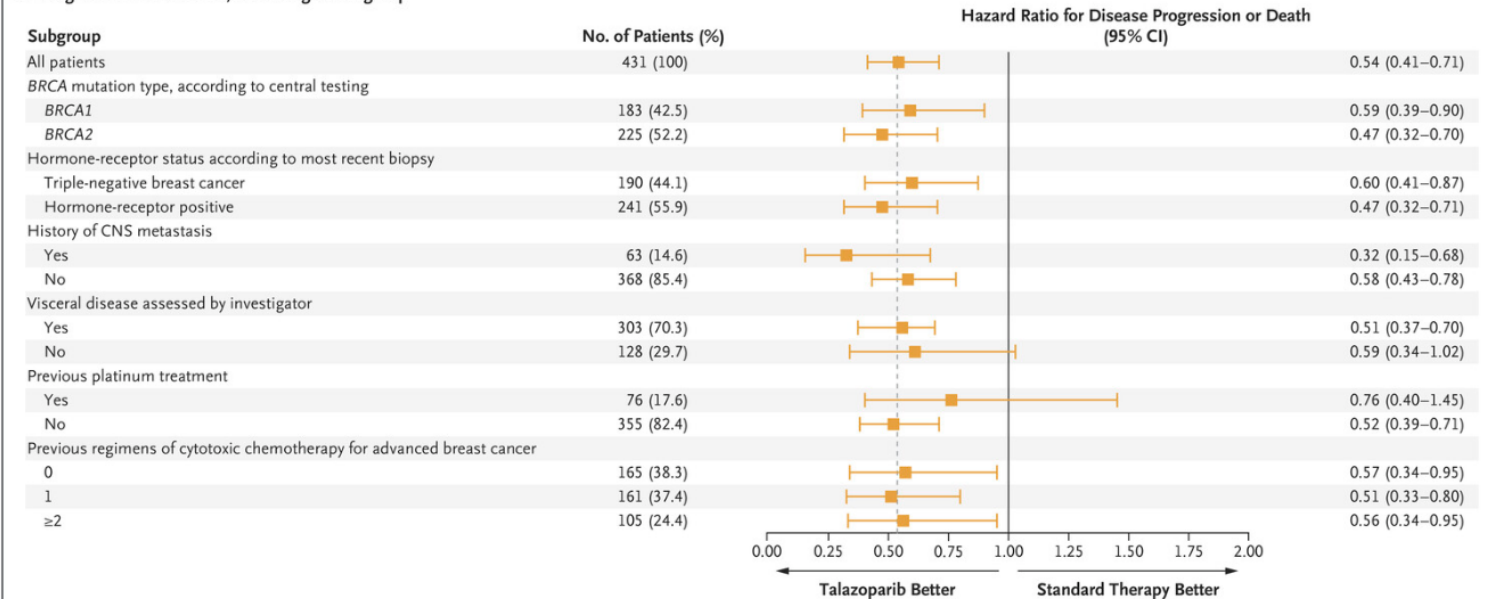
無増悪生存期間HR : 0.54
(95%CI : 0.41-0.71)



No. at Risk (events/cumulative events)

	0	3	6	9	12	15	18	21	24	27	30	33	36	39	42
Talazoparib	287 (0/0)	229 (50/50)	148 (53/103)	91 (34/137)	55 (17/154)	42 (9/163)	29 (9/172)	23 (2/174)	16 (5/179)	12 (4/183)	5 (2/185)	3 (0/185)	1 (0/185)	0 (1/186)	0 (0/186)
Standard therapy	144 (0/0)	68 (41/41)	34 (20/61)	22 (8/69)	9 (7/76)	8 (0/76)	4 (3/79)	2 (2/81)	2 (0/81)	1 (1/82)	0 (1/83)	0 (0/83)	0 (0/83)	0 (0/83)	0 (0/83)

B Progression-free Survival, According to Subgroup



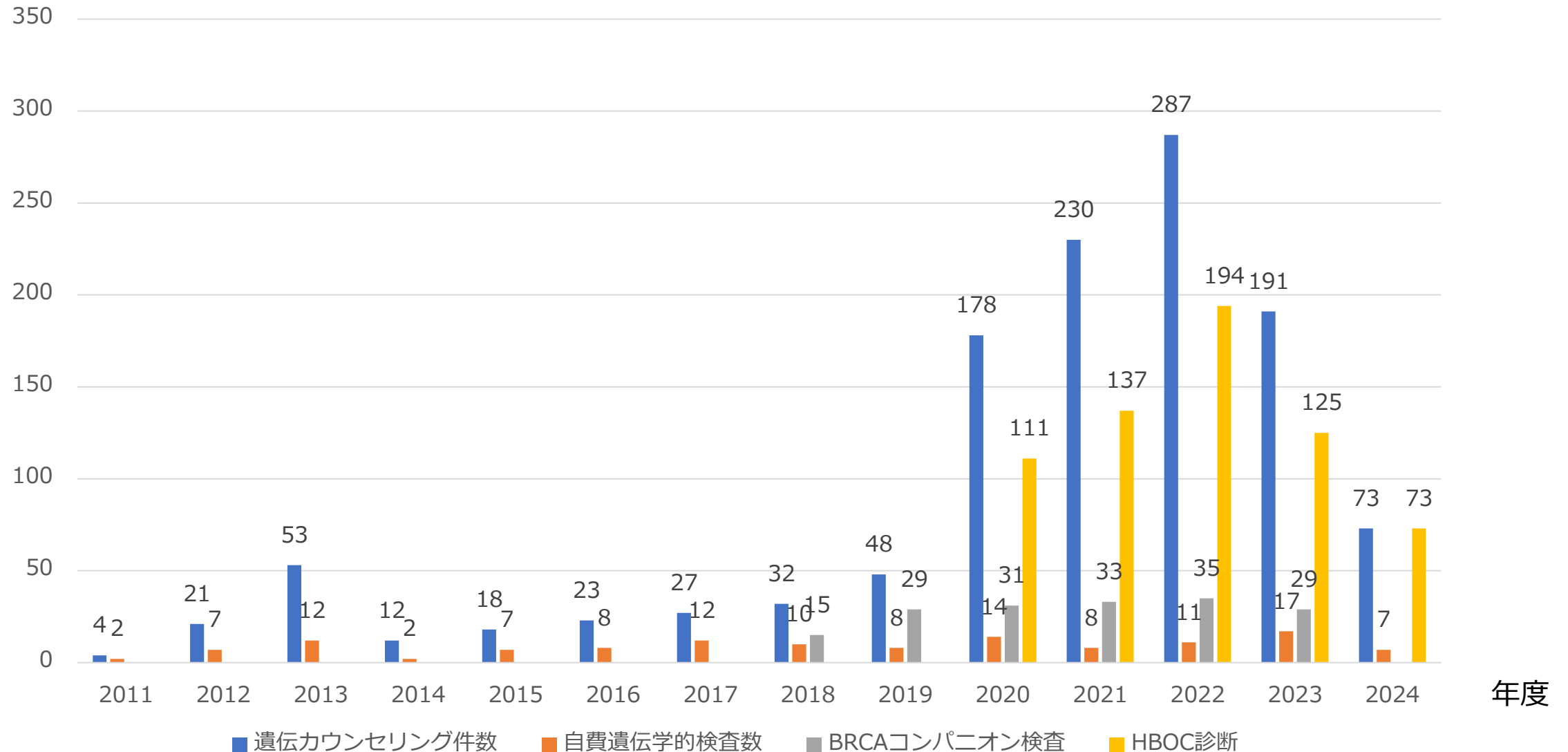
ターゼナの副作用

Table S3. Adverse Events

	Talazoparib Group (N=286)					Standard-Therapy Group (N=126)				
	Any Event	Grade 1 Event	Grade 2 Event	Grade 3 Event	Grade 4 Event	Any Event	Grade 1 Event	Grade 2 Event	Grade 3 Event	Grade 4 Event
	<i>Number of patients (percent)</i>									
Hematologic event*										
Patients with ≥1 hematologic adverse event, No. (%)	194 (67.8)	8 (2.8)	29 (10.1)	140 (49.0)	17 (5.9)	63 (50.0)	5 (4.0)	10 (7.9)	29 (23.0)	19 (15.1)
Anemia	151 (52.8)	10 (3.5)	29 (10.1)	110 (38.5)	2 (0.7)	23 (18.3)	8 (6.3)	9 (7.1)	5 (4.0)	1 (0.8)
Neutropenia	99 (34.6)	5 (1.7)	34 (11.9)	51 (17.8)	9 (3.1)	54 (42.9)	2 (1.6)	8 (6.3)	25 (19.8)	19 (15.1)
Thrombocytopenia	77 (26.9)	14 (4.9)	21 (7.3)	32 (11.2)	10 (3.5)	9 (7.1)	3 (2.4)	4 (3.2)	2 (1.6)	0
Leukopenia	49 (17.1)	6 (2.1)	24 (8.4)	18 (6.3)	1 (0.3)	17 (13.5)	1 (0.8)	5 (4.0)	8 (6.3)	3 (2.4)
Lymphopenia	21 (7.3)	4 (1.4)	8 (2.8)	9 (3.1)	0	4 (3.2)	1 (0.8)	2 (1.6)	0	1 (0.8)
Febrile neutropenia	1 (0.3)	0	0	0	1 (0.3)	1 (0.8)	0	0	0	1 (0.8)
Nonhematologic event†										
Patients with ≥1 nonhematologic adverse event, No. (%)	282 (98.6)	NR	NR	Grade 3-4 91 (31.8)		123 (97.6)	NR	NR	Grade 3-4 48 (38.1)	
Fatigue	144 (50.3)	84 (29.4)	55 (19.2)	5 (1.7)	0	54 (42.9)	33 (26.2)	17 (13.5)	4 (3.2)	0
Nausea	139 (48.6)	97 (33.9)	41 (14.3)	1 (0.3)	0	59 (46.8)	34 (27.0)	23 (18.3)	2 (1.6)	0
Headache	93 (32.5)	66 (23.1)	22 (7.7)	5 (1.7)	0	28 (22.2)	20 (15.9)	7 (5.6)	1 (0.8)	0
Alopecia	72 (25.2)	65 (22.7)	7 (2.4)	NA	NA	35 (27.8)	25 (19.8)	10 (7.9)	NA	NA
Vomiting	71 (24.8)	45 (15.7)	19 (6.6)	7 (2.4)	0	29 (23.0)	14 (11.1)	13 (10.3)	2 (1.6)	0
Diarrhea	63 (22.0)	50 (17.5)	11 (3.8)	2 (0.7)	0	33 (26.2)	14 (11.1)	12 (9.5)	7 (5.6)	0
Constipation	63 (22.0)	44 (15.4)	18 (6.3)	1 (0.3)	0	27 (21.4)	16 (12.7)	11 (8.7)	0	0
Decreased appetite	61 (21.3)	44 (15.4)	16 (5.6)	1 (0.3)	0	28 (22.2)	19 (15.1)	8 (6.3)	1 (0.8)	0
Back pain	60 (21.0)	36 (12.6)	17 (5.9)	7 (2.4)	0	20 (15.9)	12 (9.5)	6 (4.8)	2 (1.6)	0
Dyspnea	50 (17.5)	28 (9.8)	15 (5.2)	7 (2.4)	0	19 (15.1)	12 (9.5)	4 (3.2)	3 (2.4)	0
Palmar-plantar erythrodysesthesia syndrome	4 (1.4)	3 (1.0)	0	1 (0.3)	0	28 (22.2)	12 (9.5)	13 (10.3)	3 (2.4)	0
Pleural effusion	6 (2.1)	0	1 (0.3)	5 (1.7)	0	11 (8.7)	1 (0.8)	5 (4.0)	5 (4.0)	0

NA denotes not applicable; NR denotes not reported. * The thrombocytopenia category includes thrombocytopenia and decreased platelet count. The neutropenia category includes neutropenia, decreased neutrophil count, and neutropenic sepsis. The anemia category includes anemia and decreased hemoglobin level. No cases of acute myeloid leukemia or the

BRCA遺伝カウンセリングと検査数

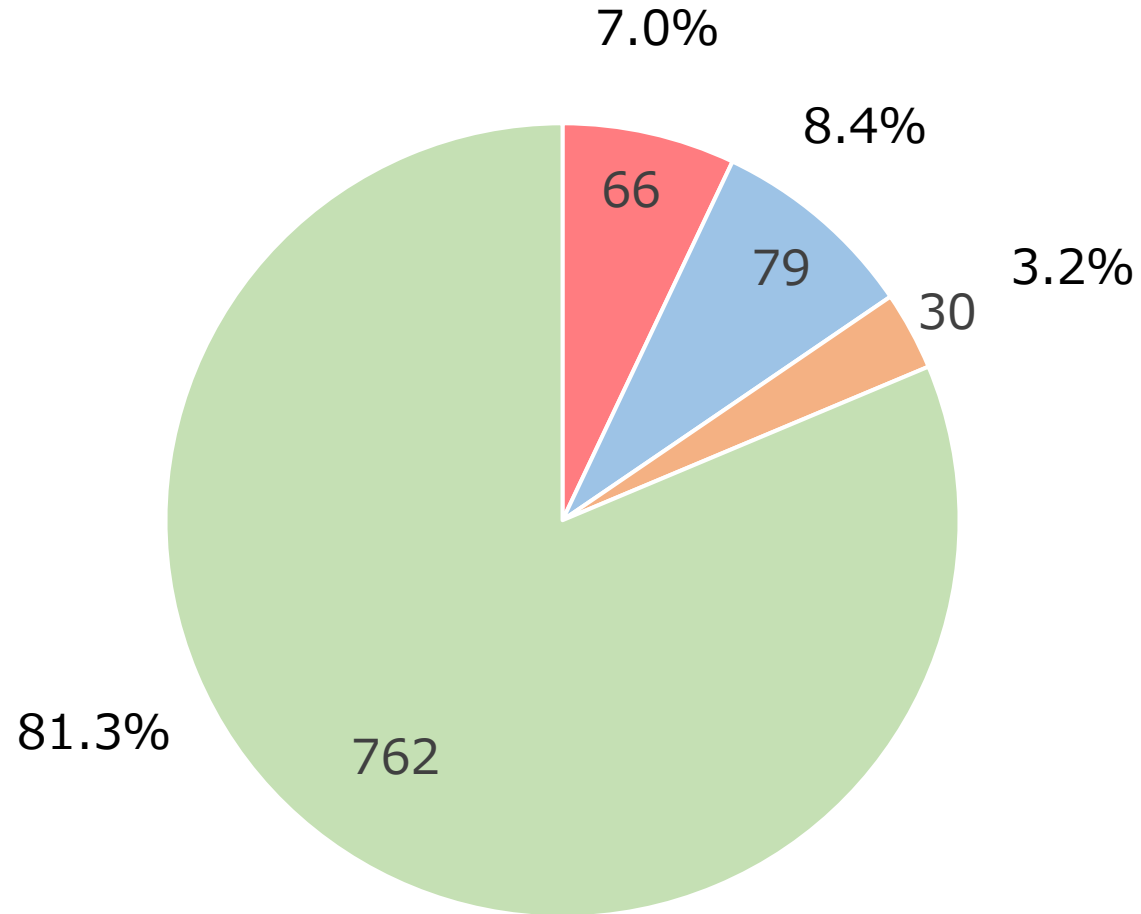


* 2024年度は4月～8月
** 遺伝カウンセリング件数は延べ数

2024年8月までの遺伝カウンセリングと検査数

遺伝カウンセリング (延べ数)	1197
自費検査 (自費検査の時期と血縁者診断を含む)	125
コンパニオン診断	172
HBOC診断	640
検査数 計	937

2024年8月までの陽性率



■ BRCA1 ■ BRCA2 ■ VUS ■ 通常のバリエーション

サーベイランス



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NCCN Guidelines Version 1.2025 **BRCA-Pathogenic/Likely Pathogenic Variant - Positive Management**

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[Discussion](#)

BRCA PATHOGENIC/LIKELY PATHOGENIC VARIANT-POSITIVE MANAGEMENT

Site	Screening/Surveillance Procedure and Interval
General	<ul style="list-style-type: none"> • Education regarding signs and symptoms of cancer(s), especially those associated with <i>BRCA</i> P/LP variants.
Breast cancer (female)	<ul style="list-style-type: none"> • Breast awareness^a starting at age 18 years. • Clinical breast exam, every 6–12 months,^b starting at age 25 years. • Breast screening^{c,d} <ul style="list-style-type: none"> ▶ Age 25–29 years, annual breast MRI^e screening with and without contrast^f (or mammogram, only if MRI is unavailable) or individualized based on family history if a breast cancer diagnosis before age 30 is present. ▶ Age 30–75 years, annual mammogram and breast MRI^e screening with and without contrast. ▶ Age >75 years, management should be considered on an individual basis. ▶ For individuals with a <i>BRCA</i> P/LP variant who are treated for breast cancer and have not had a bilateral mastectomy, screening with annual mammogram and breast MRI should continue as described above. ▶ Discuss option of RRM <ul style="list-style-type: none"> ◇ Counseling should include a discussion regarding degree of protection, reconstruction options, and risks. In addition, the family history and residual breast cancer risk with age and life expectancy should be considered during counseling. <ul style="list-style-type: none"> – Address psychosocial and quality-of-life aspects of undergoing RRM. • Consider risk reduction agents as options for breast cancer, including discussion of risks and benefits (see Discussion for details). (NCCN Guidelines for Breast Cancer Risk Reduction).
Breast cancer (male)	<ul style="list-style-type: none"> • Breast self-exam training and education starting at age 35 years. • Clinical breast exam, every 12 months, starting at age 35 years. • Consider annual mammogram, especially for those with <i>BRCA2</i> P/LP variants in whom the lifetime risk of breast cancer is up to 7%, starting at age 50 or 10 years before the earliest known male breast cancer in the family (whichever comes first).^{g,h}

サーベイランス

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NCCN Guidelines Version 1.2025 BRCA-Pathogenic/Likely Pathogenic Variant - Positive Management

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BRCA PATHOGENIC/LIKELY PATHOGENIC VARIANT-POSITIVE MANAGEMENT

Site		
Ovarian/ Fallopian Tube/ Peritoneal/ Uterine Cancers	<ul style="list-style-type: none"> • Counseling includes a discussion of reproductive options, extent of cancer risk balanced with cancer worry; degree of protection for breast, ovarian and uterine cancer; management of menopausal symptoms; hormone replacement therapy (HRT); and related medical or surgical history. • Considerations for salpingectomy with delayed oophorectomy, BSO, or non-surgical risk reduction strategies can apply to moderate-penetrance genes as well, with attention to age-related cancer risk of the known PV and family history. 	
	Reproductive considerations in premenopausal women	<ul style="list-style-type: none"> • If desired, refer to fertility specialists for discussion of age-related fertility considerations, options for in vitro fertilization, egg- and embryo-cryopreservation, and consideration of preimplantation genetic testing, gestational carrier, and adoption. • If eggs/embryos are cryopreserved, pregnancy may be achieved with uterus in place, with or without fallopian tubes or ovaries. • Individuals with P/LP <i>BRCA1</i> variant may have earlier menopause and oocyte aging.^{1,2}
	Non-surgical risk reduction	<ul style="list-style-type: none"> • Consultation with gynecologic oncologist or gynecologist with expertise/experience in genetic susceptibility to gynecologic cancer recommended. • Consideration of combination estrogen/progestin (E/P) contraception (such as oral contraceptive pills [OCP]) for ovulation suppression. Overall, studies in P/LP variant carriers support significant risk reduction benefits for ovarian cancer.^{3,4,5} See Discussion for risk/benefits of OCP. • Levonorgestrel intrauterine device (LNG-IUD) has been shown to reduce risk for ovarian cancer in the average-risk population.^{6,7}
	Surgical risk reduction with bilateral salpingo-oophorectomy	<ul style="list-style-type: none"> • Based on age-related risks of ovarian/fallopian tube cancer: <ul style="list-style-type: none"> ▶ <i>BRCA1</i>: Recommend RRSO between 35 and 40 years. ▶ <i>BRCA2</i>: Because ovarian cancer onset in patients with <i>BRCA2</i> P/LP variants is an average of 8–10 years later than in patients with <i>BRCA1</i> P/LP variants,⁸ it is reasonable to delay RRSO for management of ovarian cancer risk until age 40–45 years in patients with <i>BRCA2</i> P/LP variants unless age at diagnosis in the family warrants earlier age for consideration of prophylactic surgery. • CA-125 and pelvic ultrasound are recommended for preoperative planning. • See Risk-Reducing Salpingo-Oophorectomy (RRSO) Protocol in NCCN Guidelines for Ovarian Cancer - Principles of Surgery. Appropriate surgical and pathologic expertise is strongly recommended. SEE-FIM (Sectioning and Extensively Examining the Fimbriated End) protocol for pathologic assessment and pelvic washings should be performed. • If serous tubal intraepithelial carcinoma (STIC lesion) is found, further consultation with a gynecologist oncologist is recommended. • In addition, in premenopausal individuals, oophorectomy likely reduces the risk of developing breast cancer but the magnitude is uncertain and may be gene-specific. • Address bone health, cardiovascular health, psychosocial health, neurologic health, sexual health, and generalized quality-of-life aspects of undergoing RRSO. Consider preoperative menopause management consultation if patient is still premenopausal at time of RRSO.^{9,10} • HRT is generally not contraindicated and thus should be discussed with premenopausal patients who do not have a personal history of breast cancer.^{11,12}

サーベイランス

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NCCN Guidelines Version 1.2025 BRCA-Pathogenic/Likely Pathogenic Variant - Positive Management

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BRCA PATHOGENIC/LIKELY PATHOGENIC VARIANT-POSITIVE MANAGEMENT

Site	Screening/Surveillance Procedure and Interval
Pancreatic cancer	<ul style="list-style-type: none">For pancreatic cancer screening recommendations, see PANC-A.
Prostate cancer	<ul style="list-style-type: none">Starting at age 40 years: (Guidelines for Prostate Cancer Early Detection)<ul style="list-style-type: none">Recommend prostate cancer screening for <i>BRCA2</i> carriers.Consider prostate cancer screening for <i>BRCA1</i> carriers.
Melanoma	<ul style="list-style-type: none">No specific screening guidelines exist for melanoma, but general melanoma risk management is appropriate, such as annual full-body skin examination and minimizing ultraviolet (UV) exposure.
Risk to relatives	<ul style="list-style-type: none">Principles of Cancer Risk Assessment and Counseling (EVAL-A)
Reproductive options	<ul style="list-style-type: none">Principles of Cancer Risk Assessment and Counseling (EVAL-A)

Consider pancreatic cancer screening (preferably in the setting of a longitudinal study) for the following:

<ul style="list-style-type: none">Individuals with P/LP germline variants in <i>STK11</i>	<ul style="list-style-type: none">Beginning at age 30–35 years (or 10 years younger than the earliest exocrine pancreatic cancer diagnosis in the family, whichever is earlier).
<ul style="list-style-type: none">Individuals with P/LP germline variants in <i>CDKN2A</i>	<ul style="list-style-type: none">Beginning at age 40 years (or 10 years younger than the earliest exocrine pancreatic cancer diagnosis in the family, whichever is earlier).
<ul style="list-style-type: none">Individuals with P/LP germline variants in <i>ATM</i> or <i>BRCA2</i>	<ul style="list-style-type: none">Beginning at age 50 years (or 10 years younger than the earliest exocrine pancreatic cancer diagnosis in the family, whichever is earlier).
<ul style="list-style-type: none">Individuals with P/LP germline variants in one of the other pancreatic cancer susceptibility genes (<i>BRCA1</i>, <i>MLH1</i>, <i>MSH2</i>, <i>MSH6</i>, <i>EPCAM</i>, <i>PALB2</i>, <i>TP53</i>)	<ul style="list-style-type: none">GENE-A<ul style="list-style-type: none">Beginning at age 50 years (or 10 years younger than the earliest exocrine pancreatic cancer diagnosis in the family, whichever is earlier) for individuals with exocrine pancreatic cancer in ≥1 first- or second-degree relatives from the same side of (or presumed to be from the same side of) the family as the identified P/LP germline variant.^aThe Panel does not currently recommend pancreatic cancer screening for carriers of P/LP variants in genes other than <i>ATM</i>, <i>BRCA2</i>, <i>STK11</i>, and <i>CDKN2A</i> in the absence of a close family history of exocrine pancreatic cancer.



駒込病院ではHBOCに関わる検査、診療およびカウンセリングをおこなっています 他施設で検査を受けた方もご相談ください

- BRCA病的バリエーション保持者、VUSバリエーション保持者への遺伝カウンセリング
- BRCA病的バリエーション保持者のリスク低減手術
(乳癌および卵巣癌発症者は保険診療、未発症者は自費診療)
- BRCA病的バリエーション保持者へのサーベイランス
- 家族歴や病歴から何らかの遺伝性がんリスクを疑われる (BRCA病的バリエーションが判明しなかった など)

