№2024-018 変更
2024年3月

検査内容変更のお知らせ

謹啓　時下ますますご清栄のこととお慶び申し上げます。
平素は格別のご高配を賜り厚くお礼申し上げます。
この度、下記検査項目におきまして、検査内容の変更をご案内いたします。
健康と医療の未来に貢献すべく、より良い検査サービスのご提供に努めてまいります。
謹白

記

■ 変更日　　2024 年 3 月 15 日（金）ご報告分より

（報告書の変更は、3月16日早朝以降に医療機関へメール送信した分が対象です）

■ 対象項目

項目コード	検　査　項　目
2690 5	BRCA1/2 遺伝子検査（乳癌）
R638 8	BRCA1/2 遺伝子検査（HBOC）

■ 変更内容

本検査のコンパニオン診断の適応薬剤としてオラパリブに次いで、乳癌のみタラゾパリブトシル酸塩が承認されました。

これに伴い総合検査案内欄外および検査報告書を一部変更いたします。

1.　総合検査案内：欄外の変更

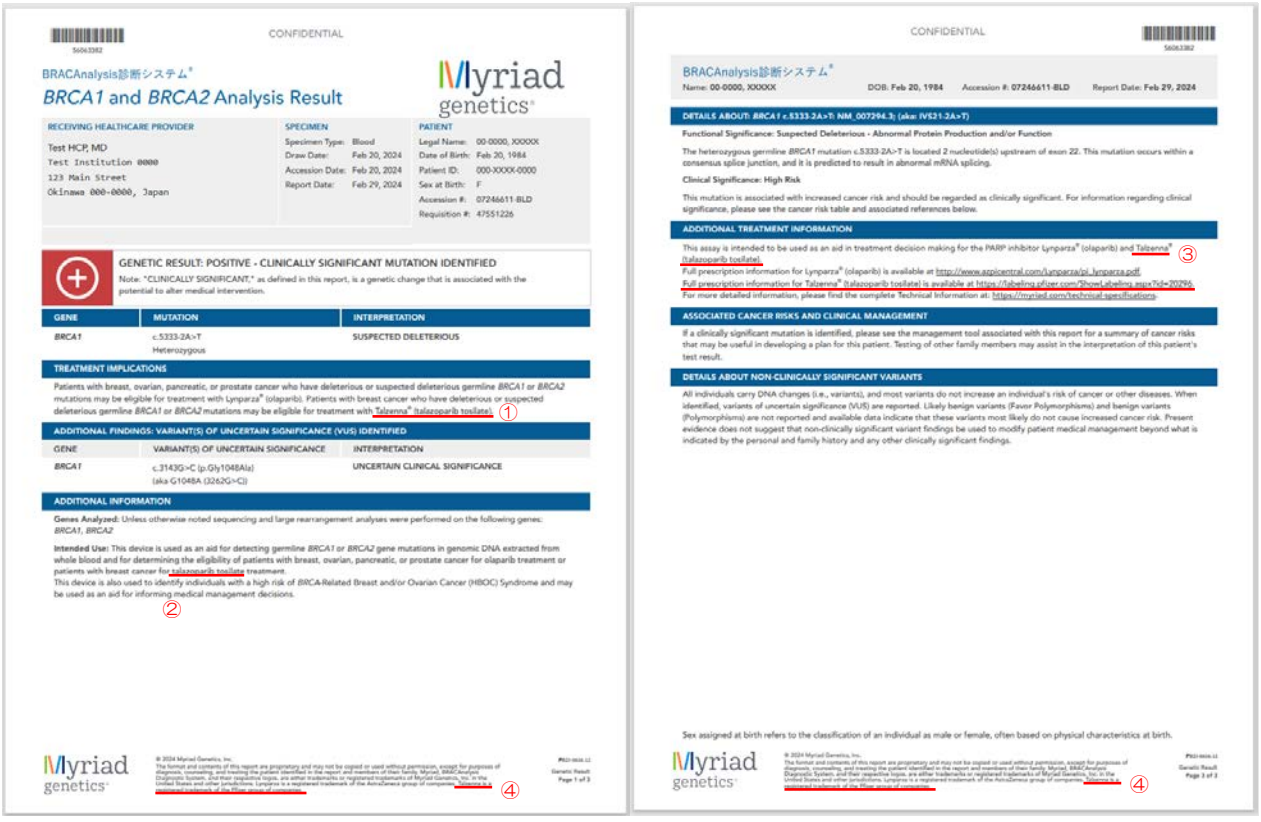
変更内容：欄外にタラゾパリブトシル酸塩に関する記載（赤字）を追加

新	現
患者の臨床的に意義のあるバリアント分類を医療従事者に提供し、オラパリブへの適応を判定するための補助に用いられるコンパニオン診断です。 **＊乳癌のみオラパリブ及びタラゾパリブトシル酸塩の適応を判定するための補助に用いられるコンパニオン診断です。**	患者の臨床的に意義のあるバリアント分類を医療従事者に提供し、オラパリブへの適応を判定するための補助に用いられるコンパニオン診断です。

2.　検査報告書：4か所にタラゾパリブトシル酸塩に関する記載を追加

変更内容：①TREATMENT IMPLICATIONS、②ADDITIONAL INFORMATION、③ADDITIONAL TREATMENT INFORMATIONに、「乳癌の場合はタラゾパリブトシル酸塩も適応であること」、及び④フッター部にタラゾパリブトシル酸塩に関する記載を追加（下図赤線）

新報告書見本（一部抜粋）



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BRACAnalysis診断システム®

BRCA1 and *BRCA2* Analysis Result

Myriad genetics®

RECEIVING HEALTHCARE PROVIDER

Test HCP, MD
Test Institution 0000
123 Main Street
Okinawa 000-0000, Japan

SPECIMEN

Specimen Type: Blood
Draw Date: Feb 20, 2024
Accession Date: Feb 20, 2024
Report Date: Feb 29, 2024

PATIENT

Legal Name: 00-0000, XXXXX
Date of Birth: Feb 20, 1984
Patient ID: 000-XXXX-0000
Sex at Birth: F
Accession #: 07246611-BLD
Requisition #: 47551226

GENETIC RESULT: POSITIVE - CLINICALLY SIGNIFICANT MUTATION IDENTIFIED

Note: "CLINICALLY SIGNIFICANT," as defined in this report, is a genetic change that is associated with the potential to alter medical intervention.

GENE	MUTATION	INTERPRETATION
BRCA1	c.5333-2A>T Heterozygous	SUSPECTED DELETERIOUS

TREATMENT IMPLICATIONS

Patients with breast, ovarian, pancreatic, or prostate cancer who have deleterious or suspected deleterious germline *BRCA1* or *BRCA2* mutations may be eligible for treatment with Lynparza® (olaparib). Patients with breast cancer who have deleterious or suspected deleterious germline *BRCA1* or *BRCA2* mutations may be eligible for treatment with Talzenna® (talazoparib tosilate). ①

ADDITIONAL FINDINGS: VARIANT(S) OF UNCERTAIN SIGNIFICANCE (VUS) IDENTIFIED

GENE	VARIANT(S) OF UNCERTAIN SIGNIFICANCE	INTERPRETATION
BRCA1	c.3143G>C (p.Gly1048Ala) (aka G1048A (3262G>C))	UNCERTAIN CLINICAL SIGNIFICANCE

ADDITIONAL INFORMATION

Genes Analyzed: Unless otherwise noted sequencing and large rearrangement analyses were performed on the following genes: *BRCA1, BRCA2*

Intended Use: This device is used as an aid for detecting germline *BRCA1* or *BRCA2* gene mutations in genomic DNA extracted from whole blood and for determining the eligibility of patients with breast, ovarian, pancreatic, or prostate cancer for olaparib treatment or patients with breast cancer for talazoparib tosilate treatment.
This device is also used to identify individuals with a high risk of *BRCA*-Related Breast and/or Ovarian Cancer (HBOC) Syndrome and may be used as an aid for informing medical management decisions.
②

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BRACAnalysis診断システム®

Name: 00-0000, XXXXX　DOB: Feb 20, 1984　Accession #: 07246611-BLD　Report Date: Feb 29, 2024

DETAILS ABOUT *BRCA1* c.5333-2A>T: NM_007294.3; (aka: IVS21-2A>T)

Functional Significance: Suspected Deleterious - Abnormal Protein Production and/or Function

The heterozygous germline *BRCA1* mutation c.5333-2A>T is located 2 nucleotide(s) upstream of exon 22. This mutation occurs within a consensus splice junction, and it is predicted to result in abnormal mRNA splicing.

Clinical Significance: High Risk

This mutation is associated with increased cancer risk and should be regarded as clinically significant. For information regarding clinical significance, please see the cancer risk table and associated references below.

ADDITIONAL TREATMENT INFORMATION

This assay is intended to be used as an aid in treatment decision making for the PARP inhibitor Lynparza® (olaparib) and Talzenna® (talazoparib tosilate). ③
Full prescription information for Lynparza® (olaparib) is available at http://www.azpicentral.com/lynparza/pi_lynparza.pdf.
Full prescription information for Talzenna® (talazoparib tosilate) is available at https://labeling.pfizer.com/ShowLabeling.aspx?id=20296.
For more detailed information, please find the complete Technical Information at: https://myriad.com/technical-specifications.

ASSOCIATED CANCER RISKS AND CLINICAL MANAGEMENT

If a clinically significant mutation is identified, please see the management tool associated with this report for a summary of cancer risks that may be useful in developing a plan for this patient. Testing of other family members may assist in the interpretation of this patient's test result.

DETAILS ABOUT NON-CLINICALLY SIGNIFICANT VARIANTS

All individuals carry DNA changes (i.e., variants), and most variants do not increase an individual's risk of cancer or other diseases. When identified, variants of uncertain significance (VUS) are reported. Likely benign variants (Favor Polymorphisms) and benign variants (Polymorphisms) are not reported and available data indicate that these variants most likely do not cause increased cancer risk. Present evidence does not suggest that non-clinically significant variant findings be used to modify patient medical management beyond what is indicated by the personal and family history and any other clinically significant findings.

Sex assigned at birth refers to the classification of an individual as male or female, often based on physical characteristics at birth.

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報告書の変更内容は、下記項目で共通です。

- 2690 5 BRCA1/2 遺伝子検査（乳癌）
- R221 7 BRCA1/2 遺伝子検査（卵巣癌）
- U052 0 BRCA1/2 遺伝子検査（膵癌）
- R629 0 BRCA1/2 遺伝子検査（前立腺癌）
- R638 8 BRCA1/2 遺伝子検査（HBOC）