GeneCancer

Hereditary Cancer Risk Analysis · 108+ Genes · 15+ Cancer Types · NCCN-Aligned Screening Plan

PATIENT **Sarah Mitchell**	DATE OF BIRTH **1986-05-14 (Age 38)**	SAMPLE TYPE **Buccal (Cheek) Swab**	SAMPLE ID **GM-CA-2024-01187**
REPORT ID **GCA-2024-66130**	COLLECTION DATE **2024-02-12**	REPORT DATE **2024-02-19**	LABORATORY **Gene Matrix CLIA Lab, Chicago, IL**
CLIA / NPI **14D2276402 / 1225729312**	ORDERING PROVIDER **Self-Ordered (DTC)**	METHOD **MassArray / NGS + Del-Dup Analysis**	

108+ GENES ANALYZED	**15+** CANCER TYPES SCREENED	**1** PATHOGENIC VARIANT	**99.9%** ANALYTICAL ACCURACY

/ Executive Summary

Analysis of your 108-gene hereditary cancer panel identified one pathogenic variant: BRCA1. Your genes carry a significant increase in breast and ovarian cancer risk — your lifetime risk of breast cancer is up to 72% (vs. 12.5% for the average person), and ovarian cancer risk is up to 44%. No pathogenic variants were detected in BRCA2, PALB2, the Lynch syndrome genes, TP53, or the remaining panel genes. Early detection with enhanced screening is proven to save lives — a personalized, NCCN-aligned screening plan follows.

Family impact: Other family members likely carry this same change. Each first-degree relative (parents, siblings, children) has a 50% chance of carrying the BRCA1 variant. Cascade testing is recommended — share this report with your family in a supportive, informative manner.

/ Genetic Findings

GENE	VARIANT	GENOTYPE	IMPACT	RECOMMENDATION
BRCA1	`c.5266dupC (p.Gln1756Profs)`	Heterozygous	PATHOGENIC Your body's "tumor off switch" is partially missing. Lifetime breast cancer risk up to 72%, ovarian up to 44%. Cancer can develop at a much younger age than average.	Enhanced screening: annual breast MRI plus mammogram starting now; ovarian surveillance (CA-125 + transvaginal ultrasound). Discuss risk-reducing options with specialists. Genetic counseling arranged.
BRCA2	`No pathogenic variant detected`	—	NEGATIVE Full sequencing and deletion/duplication analysis negative.	Population-risk screening for BRCA2-associated cancers.

PALB2	No pathogenic variant detected	—	NEGATIVE No elevated PALB2-associated breast or pancreatic cancer risk identified.	Routine screening per BRCA1-driven plan.
MLH1 / MSH2 / MSH6 / PMS2 / EPCAM	No pathogenic variants detected	—	NEGATIVE Lynch syndrome not indicated by this panel.	Colonoscopy every 5–10 years per general-population guidelines unless family history dictates otherwise.
TP53	No pathogenic variant detected	—	NEGATIVE Li-Fraumeni syndrome not indicated.	No additional surveillance required from this gene.
CHEK2 / ATM / CDKN2A / APC	No pathogenic variants detected	—	NEGATIVE Moderate-penetrance breast, melanoma, and colorectal genes negative.	Continue the enhanced screening schedule outlined below.

/ Risk Summary by Cancer Type

Your assessed risk category across the six major hereditary cancer groups covered by this panel, integrating genetic findings with clinical guidelines.

CANCER GROUP	RESULT	CLINICAL IMPLICATION
Breast & Ovarian — BRCA1, BRCA2, PALB2	HIGH	Pathogenic BRCA1 variant confirmed. Enhanced MRI/mammogram screening and specialist referral indicated.
Colorectal — MLH1, MSH2, MSH6, PMS2, EPCAM, APC, MUTYH	MODERATE	No pathogenic variant; family-history-based surveillance still advised.
Prostate — HOXB13, BRCA2, ATM, CHEK2	MODERATE	Applies to male relatives who carry the familial BRCA1 variant — PSA screening discussion recommended.
Lynch Syndrome — mismatch repair genes	MODERATE	Panel negative; standard colonoscopy schedule.
Skin (Melanoma) — CDKN2A, CDK4, BAP1, MC1R	MODERATE	Panel negative; annual dermatology exam advised.
Pancreatic & Gastric — BRCA2, ATM, PALB2, CDKN2A, CDH1, STK11	MODERATE	BRCA1 carriers may qualify for pancreatic surveillance programs — discuss with your specialist.

/ Personalized Recommendations

Enhanced Screening Schedule (NCCN-Aligned)

- Breast MRI every 12 months, alternating with mammography every 12 months (imaging every 6 months in practice).
- Ovarian surveillance: transvaginal ultrasound and CA-125 every 6–12 months as directed by your gynecologic oncologist.
- Report any new symptoms promptly to your healthcare team; keep detailed personal health records and family history.

Prevention & Lifestyle

- Maintain healthy body weight (BMI 18.5–24.9) and exercise 150+ minutes of moderate activity weekly.
- Avoid tobacco and limit alcohol consumption; ensure all vaccinations are current (HPV vaccine if eligible).
- Discuss chemoprevention options: Tamoxifen/Raloxifene for breast, Aspirin for colorectal, with your physician.
- Consider risk-reducing surgeries only after thorough discussion with specialists.

Family & Support

- Share findings with family in a supportive, informative manner — first-degree relatives have a 50% chance of carrying this variant.
- Consider joining hereditary cancer support groups; seek mental health counseling if experiencing anxiety about cancer risk.
- A board-certified genetic counselor session is included with this result — schedule within 30 days.

Laboratory: Gene Matrix LLC · 1375 W Fulton St STE 545, Chicago, IL 60607 · CLIA ID: 14D2276402 · NPI: 1225729312 · (847) 302-9668 · support@genematrix.io · Mon–Fri 8AM–5PM CT. Testing performed in a CLIA-certified, CAP-accredited laboratory using next-generation sequencing. Analytical accuracy 99.9%. All findings reviewed by board-certified geneticists.
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