You are currently completing this survey for ${m://ExternalDataReference}.

Survey Cover Page

Dear Healthcare Provider:
You are invited to participate in a research study exploring how recommendations that healthcare providers make to their patients may change based on the results of genetic testing.

This study is being conducted by Jill Dolinsky, MS, CGC at Ambry Genetics. Healthcare providers (e.g. physicians, genetic counselors, PhDs and nurses) who practice in the USA and are involved in genetic testing are welcome to participate. This invitation is related to your patient:

Patient Name: ${m://ExternalDataReference}

- This study will consist of two surveys regarding the clinical management of your patient. **This survey should be completed prior to the receipt of genetic testing results.** If this survey is completed, you will receive an invitation for the second survey after the receipt of genetic testing results.
- Each survey should take approximately 5-10 minutes to complete.
- Participation is voluntary and you may decide to leave the survey at any time.
- De-identified clinical and test result information about your patient may be included in the analysis of your responses. In no way will your responses or your patient’s information be identifiable to the investigators.
- After completing each survey you will have the option of entering a raffle. After completing this survey, you will be able to enter a raffle for a $50 Amazon gift card. After completing the second survey (administered after test results are reported), you will be able to enter a raffle for a $100 Amazon gift card. Drawings will be held four times a year on a quarterly basis. If you wish to enter the drawing you will be prompted to enter your email address, which will be stored separately from your survey responses.
- Should you have any questions regarding this research study you may contact the study team at ambrystudies@ambrygen.com
By continuing with the survey, you acknowledge that you have read the information above and consent to participate in this survey. Thank you in advance for your time and consideration.

Sincerely,

Jill Dolinsky
Director, Clinical Affairs
jdolinsky@ambrygen.com

Starting Block

Genetic testing category for this patient:

- Oncology
- Cardiology
- Neurology/General Genetics

Is your patient living?

- Yes
- No

Alive Oncology Block

Does your patient meet criteria for genetic testing?

- Yes
- No
- I don't know/I'm not sure
Please specify criteria used to determine eligibility for genetic testing (select all that apply):

- NCCN Testing Criteria
- Syndrome Specific Criteria (e.g. Amsterdam, Chompret, etc.)
- Prior probability >10% on BRCAPro, BOADICEA, IBIS, etc.
- Patient Insurance Company Criteria
- Other (specify):

Cancer types related to your patient's **current or past cancer screenings/interventions** (select all that apply):

- None
- Brain
- Breast
- Colorectal
- Leukemia/Lymphoma
- Melanoma
- Ovarian
- Pancreatic
- Prostate
- Uterine
- GI polyps
- Other type (specify):

Currently recommended or previously completed **brain cancer** screenings/interventions (select all that apply):

- Biopsy
- Imaging
- Surgery
- Radiation
- Systemic Therapy/Chemotherapy
- Chemopreventive Medications
Currently recommended or previously completed breast cancer screenings/interventions (select all that apply):

- Referral to Specialist
- Recommendations for changes in lifestyle/health habits
- Patient education in symptom awareness
- Other (specify):
- None

Currently recommended or previously completed colorectal cancer screenings/interventions (select all that apply):

- Self-Breast Exam
- Breast MRI
- Digital/Analog Mammogram (MMG)
- Tomosynthesis/3D Mammogram
- Ultrasound
- Bilateral Mastectomy
- Risk-Reducing Mastectomy (RRM)
- Risk-Reducing Salpingo Oophorectomy (RRSO)
- Chemopreventive Medications
- Radiation
- Systemic Therapy/Chemotherapy
- Referral to a Specialist
- Recommendations for changes in lifestyle/health habits
- Other (specify):
- None

- Colonoscopy
- Sigmoidoscopy
- Upper Endoscopy
- Video Capsule Endoscopy
- High-sensitivity Fecal Occult Blood Tests (FOBT)
- Stool DNA Test (FIT-DNA)
Currently recommended or previously completed melanoma screenings/interventions (select all that apply):

- Biopsy
- Skin Cancer Screening
- Eye Exam/Ocular Melanoma Screening
- Sun Screen Agents
- Medications
- Radiation
- Systemic Therapy/Chemotherapy
- Referral to Specialist
- Recommendations for changes in lifestyle/health habits
- Patient education in symptom awareness
- Other (specify):
- None

Currently recommended or previously completed ovarian cancer screenings/interventions (select all that apply):

- CA-125
- Transvaginal Ultrasound
- Salpingo-Oophorectomy
- Bilateral Salpingo-Oophorectomy
- None
Currently recommended or previously completed **pancreatic cancer** screenings/interventions (select all that apply):

- Abdominal Ultrasound
- Angiography
- Biopsy
- CA 19-9
- Carcinoembryonic Antigen (CEA)
- CT Scan
- Endoscopic Ultrasound
- Endoscopic Retrograde Cholangiopancreatography (ERCP)
- Liver Function Tests
- MR Angiography (MRA)
- MR Cholangiopancreatography (MRCP)
- Percutaneous Transhepatic Cholangiography (PTC)
- Positron Emission Tomography (PET)
- Somatostatin Receptor Scintigraphy (SCS)
- Chemopreventive Medications
- Radiation
- Systemic Therapy/Chemotherapy
- Surgical Resection
- Referral to Specialist

Currently recommended or previously completed **pancreatic cancer** screenings/interventions (select all that apply):

- Total Abdominal or Laparoscopic Hysterectomy and Bilateral Salpingo-Oophorectomy (TAH-BSO)
- Chemopreventive Medications
- Oral Contraceptives (OCP)
- Radiation
- Systemic Therapy/Chemotherapy
- Referral to Specialist
- Recommendations for changes in lifestyle/health habits
- Patient education on symptom awareness
- Other (specify):
- None
Currently recommended or previously completed prostate cancer screenings/interventions (select all that apply):

- Biopsy
- Prostate Specific Antigen (PSA)
- Prostate MRI
- Hormone Therapy
- Radiotherapy
- Prostatectomy
- Referral to Specialist
- Chemopreventive Medications
- Radiation
- Systemic Therapy/Chemotherapy
- Clinical Trial
- Recommendations for changes in lifestyle/health habits
- Patient education in symptom awareness
- Other (specify):
- None

Currently recommended or previously completed uterine cancer screenings/interventions (select all that apply):

- Biopsy
- CA-125
- CT Scan
- MRI
- Cystoscopy and Proctoscopy
- Hysteroscopy
Currently recommended or previously completed **leukemia/lymphoma** screenings/interventions (select all that apply):

- [ ] Biopsy
- [ ] Bone Marrow Aspiration and Biopsy
- [ ] Lumbar Puncture
- [ ] Complete Blood Count (CBC)
- [ ] Chest X-Ray or Chest CT Scan
- [ ] Blood Transfusion
- [ ] Stem Cell Transplant
- [ ] Radiotherapy
- [ ] Chemopreventive Medications
- [ ] Radiation
- [ ] Systemic Therapy/Chemotherapy
- [ ] Referral to Specialist
- [ ] Recommendations for changes in lifestyle/health habits
- [ ] Patient education in symptom awareness

**Other Blood Tests (specify):**

[ ]

**Other (specify):**

[ ]

[ ] None
Currently recommended or previously completed **Other Cancer** screenings/interventions (select all that apply):

- [ ] Biopsy
- [ ] Blood Testing
- [ ] CT scan
- [ ] MRI
- [ ] Ultrasound
- [ ] Medications
- [ ] Radiation
- [ ] Systemic Therapy/Chemotherapy
- [ ] Referral to Specialist
- [ ] Recommendations for changes in lifestyle/health habits
- [ ] Patient education in symptom awareness
- [ ] Other Imaging (specify):
- [ ] Other (specify):
- [ ] None

Currently recommended or past **GI polyps** monitoring/screening (select all that apply):

- [ ] Biopsy
- [ ] Blood Testing
- [ ] CT scan
- [ ] MRI
- [ ] Ultrasound
- [ ] Medications
- [ ] Referral to Specialist
- [ ] Recommendations for changes in lifestyle/health habits
- [ ] Patient education in symptom awareness
- [ ] Other Imaging (specify):
- [ ] Other (specify):
- [ ] None
Is there a known familial pathogenic/likely pathogenic variant in the family?

- Yes
- No

Please provide details, including the gene/syndrome and closest degree of relation that tested positive:

Has your patient been tested for the familial variant?

- Yes
- No

Has your patient had previous genetic testing?

- Yes
- No

Type of previous genetic testing (select all that apply):

- [ ] Clinical Testing
- [ ] Research Study Testing
- [ ] Healthy Screening Panel (e.g. 23andMe, Ancestry, etc.)

Previous Testing History (select all that apply):

- [ ] Single Site Analysis
- [ ] BRCA1/2
Lynch Syndrome Panel
Other Single Gene
Breast Cancer Panel
Ovarian Cancer Panel
Colorectal Cancer Panel
Comprehensive Cancer Panel
Other Gene Panel
Healthy Screening Panel (e.g. 23andMe, Ancestry, etc.)

Single Site Analysis Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Single Site Analysis Results - Specify gene(s) and variant(s), if known

BRCA1/2 Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

BRCA1/2 Results - Specify gene(s) and variant(s), if known

Lynch Syndrome Panel Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Lynch Syndrome Panel Results - Specify gene(s) and variant(s), if known
Other Single Gene Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Other Single Gene Results - Specify gene(s) and variant(s), if known

Breast Cancer Panel Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Breast Cancer Panel Results - Specify gene(s) and variant(s), if known

Ovarian Cancer Panel Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Ovarian Cancer Panel Results - Specify gene(s) and variant(s), if known

Colorectal Cancer Panel Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance
Colorectal Cancer Panel Results - Specify gene(s) and variant(s), if known

Comprehensive Cancer Panel Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Comprehensive Cancer Panel Results - Specify gene(s) and variant(s), if known

Other Gene Panel Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Other Gene Panel Results - Specify gene(s) and variant(s), if known

Healthy Screening Panel Results - Specify gene(s)/variant(s) and classification (if available)

Are there any additional details you would like to provide regarding the patient's treatment or management (optional)?
Deceased Oncology Block

Does your patient have a known pathogenic/likely pathogenic variant in their family?

- Yes
- No

Please provide details, including the gene/syndrome and closest degree of relation that tested positive:

Has your patient been tested for the familial variant?

- Yes
- No

Did your patient have previous genetic testing?

- Yes
- No

Type of previous genetic testing (select all that apply):

- Clinical Testing
- Research Study
- Healthy Screening Panel (e.g. 23andMe, Ancestry, etc.)

Previous Testing History (select all that apply):
Single Site Analysis Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Single Site Analysis Results - Specify gene(s) and variant(s), if known

BRCA1/2 Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

BRCA1/2 Results - Specify gene(s) and variant(s), if known

Lynch Syndrome Panel Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance
Lynch Syndrome Panel Results - Specify gene(s) and variant(s), if known

Other Single Gene Results
- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Other Single Gene Results - Specify gene(s) and variant(s), if known

Breast Cancer Panel Results
- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Breast Cancer Panel Results - Specify gene(s) and variant(s), if known

Ovarian Cancer Panel Results
- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Ovarian Cancer Panel Results - Specify gene(s) and variant(s), if known

Colorectal Cancer Panel Results
- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance
Colorectal Cancer Panel Results - Specify gene(s) and variant(s), if known

Comprehensive Cancer Panel Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Comprehensive Cancer Panel Results - Specify gene(s) and variant(s), if known

Other Gene Panel Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Other Gene Panel Results - Specify gene(s) and variant(s), if known

Healthy Screening Panel Results - Specify gene(s)/variant(s) and classification (if available)

Have you discussed or do you plan to discuss any of the following genetic counseling implications with the deceased patient's family members prior to receiving the results of the ordered genetic test?

- Reproductive testing options
- Cancer recurrence risk
- Identification/Testing of at-risk family members
- Other (specify):
Have you discussed or do you plan to discuss any of the following with the deceased patient's **family members** prior to receiving the results of the ordered genetic test?

- ☐ Referral to support organizations
- ☐ Changes in financial planning
- ☐ Discrimination in employment and/or in obtaining insurance
- ☐ Detection of mis-attributed paternity or adoption
- Legal implications (specify):
- Other (specify):
- ☐ None

Are there any additional details you would like to provide regarding the patient's **family members'** treatment or management (optional)?

Alive Cardiology Block

Did your patient meet criteria for genetic testing?

- ☐ Yes
- ☐ No
- ☐ I don't know/I'm not sure
Please specify criteria used to determine eligibility for genetic testing (select all that apply):

- [ ] Syndrome Specific Criteria (e.g. Marfan, Noonan, etc.)
- [ ] Patient's Insurance Company Criteria
- [ ] Other (specify):

Has your patient had a cardiac transplant?

- [ ] Yes
- [ ] No

Currently recommended screening/monitoring (select all that apply):

- [ ] Blood pressure monitoring
- [ ] Cardiac MRI
- [ ] Chest X-Ray/Angiography
- [ ] CT Scan
- [ ] Echocardiogram
- [ ] Electrophysiology study (EPS)
- [ ] Exercise stress test
- [ ] 24-hour ECG monitoring (Holter)
- [ ] Pulse monitoring
- [ ] Other (specify):
- [ ] None

Currently prescribed medications (select all that apply):

- [ ] Anticoagulants
- [ ] Antiplatelet agents and Dual Antiplatelet Therapy (DAPT)
- [ ] Angiotensin-converting enzyme (ACE) Inhibitors
- [ ] Angiotensin II receptor blockers (ARBs)
- [ ] Alpha-adrenoreceptor antagonists
Currently recommended interventions (select all that apply):

- Beta-adrenergic blocking agents
- Combined alpha- and beta-blocking agents
- Cholesterol lowering medications
- Calcium channel antagonists
- Digitalis preparations
- Diuretics
- Pain Medications
- Sodium channel antagonists
- Supplements
- Vasodilators
- Other (specify):
- None

Is there a known familial pathogenic/likely pathogenic variant in the family?

- Yes
- No

Please provide details, including the gene/syndrome and closest degree of relation that tested positive:
Has your patient been tested for the familial variant?

- [ ] Yes
- [ ] No

Has your patient had previous genetic testing?

- [ ] Yes
- [ ] No

Type of previous genetic testing (select all that apply):

- [ ] Clinical Testing
- [ ] Research Study Testing
- [ ] Healthy Screening Panel (e.g. 23andMe, Ancestry, etc.)

Previous Testing History (select all that apply):

- [ ] Chromosomal Microarray (CMA)
- [ ] Karyotype
- [ ] Single Site Analysis
- [ ] Single Gene
- [ ] Cardiology Gene Panel
- [ ] Non-Cardiology Gene Panel
- [ ] Healthy Screening Panel (e.g. 23andMe, Ancestry, etc.)

Chromosomal Microarray (CMA) Results
Chromosomal Microarray (CMA) Results - Specify CNV

Karyotype Results

- Abnormal
- Normal and/or Variant of Unknown Significance

Karyotype Results - Specify

Single Site Analysis Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Single Site Analysis Results - Specify gene(s) and variant(s), if known

Single Gene Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Single Gene Results - Specify gene(s) and variant(s), if known
Cardiology Gene Panel Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Cardiology Gene Panel Results - Specify gene(s) and variant(s), if known

Non-Cardiology Gene Panel

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Non-Cardiology Gene Panel - Specify gene(s) and variant(s), if known

Healthy Screening Panel Results - Specify gene(s)/variant(s) and classification (if available)

Are there any additional details you would like to provide regarding the patient's treatment or management (optional)?

Deceased Cardiology Block
Is there a known pathogenic/likely pathogenic variant in the family?

- Yes
- No

Please provide details, including the gene/syndrome and closest degree of relation that tested positive:

Has your patient been tested for the familial variant?

- Yes
- No

Did your patient have previous genetic testing?

- Yes
- No

Type of previous genetic testing (select all that apply):

- Clinical Testing
- Research Study Testing
- Healthy Screening Panel (e.g. 23andMe, Ancestry, etc.)

Previous Testing History (select all that apply):

- Chromosomal Microarray (CMA)
- Karyotype
Healthy Screening Panel Results - Specify gene(s)/variant(s) and classification (if available)

Chromosomal Microarray (CMA) Results
- Pathogenic/Likely Pathogenic Copy Number Variant (CNV)
- Non-Pathogenic/Likely Benign Copy Number Variant (CNV)/Variant of Unknown Significance

Chromosomal Microarray (CMA) Results - Specify CNV

Karyotype Results
- Abnormal
- Normal and/or Variant of Unknown Significance

Karyotype Results - Specify

Single Site Analysis Results
- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance
Single Site Analysis Results - Specify gene(s) and variant(s), if known

Single Gene Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Cardiology Gene Panel Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Cardiology Gene Panel Results - Specify gene(s) and variant(s), if known

Non-Cardiology Gene Panel

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Non-Cardiology Gene Panel - Specify gene(s) and variant(s), if known

Have you discussed or do you plan to discuss any of the following genetic counseling implications with the deceased patient's family members prior to receiving the results of the ordered genetic test?
Have you discussed or do you plan to discuss any of the following with the deceased patient's family members prior to receiving the results of the ordered genetic test?

- Referral to support organizations
- Changes in financial planning
- Discrimination in employment and/or in obtaining insurance
- Detection of misattributed paternity or adoption

Legal implications (specify):

Other (specify):

- None

Are there any additional details you would like to provide regarding the patient's family members' treatment or management (optional)?

Alive General Block

Did your patient meet criteria for genetic testing?
Please specify criteria used to determine eligibility for genetic testing (select all that apply):

☐ Syndrome Specific Criteria (e.g. Marfan, Noonan, etc.)
☐ Patient's Insurance Company Criteria
☐ Other (specify):

Is there a known pathogenic/likely pathogenic variant in the family?

☐ Yes
☐ No

Please provide details, including the gene/syndrome and closest degree of relation that tested positive:







Has your patient been tested for the familial variant?

☐ Yes
☐ No

Has your patient had previous genetic testing?

☐ Yes
☐ No
Type of previous genetic testing (select all that apply):

- Clinical Testing
- Research Study Testing
- Healthy Screening Panel (e.g. 23andMe, Ancestry, etc.)

Previous Genetic Testing (select all that apply):

- Chromosomal Microarray (CMA)
- Karyotype
- Single Site Analysis
- Single Gene
- Gene Panel
- Whole Exome Sequencing/Whole Genome Sequencing
- Healthy Screening Panel (e.g. 23andMe, Ancestry, etc.)
- Other (specify):

Healthy Screening Panel Results - Specify gene(s)/variant(s) and classification (if available)

Chromosomal Microarray (CMA) Results

- Pathogenic/Likely Pathogenic Copy Number Variant (CNV)
- Non-Pathogenic/Likely Benign Copy Number Variant (CNV)

Chromosomal Microarray (CMA) Results - Specify CNV

Karyotype Results
Karyotype Results - Specify

- Abnormal
- Normal and/or Variant of Unknown Significance

Single Site Analysis Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Single Site Analysis Results - Specify gene(s) and variant(s), if known

Single Gene Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Single Gene Results - Specify gene(s) and variant(s), if known

Gene Panel Type (e.g. Noonan Syndrome Panel, Neurodevelopmental Panel, etc.)

Gene Panel Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance
Gene Panel Results - Specify gene(s) and variant(s), if known

Whole Exome Sequencing/Whole Genome Sequencing Results
- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Whole Exome Sequencing/Whole Genome Sequencing Results - Specify gene(s) and variant(s), if known

Other Testing Type (e.g. Chromosomal breakage analysis, methylation studies, mitochondrial sequencing, etc.)

Other Testing Results - Specify

Current monitoring/screening (select all that apply):
- Biochemical testing
- Bone Age
- Echocardiogram
- EEG
- EMG
- X-Ray/Skeletal Survey
Current interventions (select all that apply):

- [ ] Dietary Intervention
- [ ] Medications
- [ ] Supplements
- [ ] Physical Therapy
- [ ] Speech Therapy
- [ ] Occupational Therapy
- [ ] Other (specify):
- [ ] None

Dietary Intervention - Specify



Medications - Specify



Supplements - Specify



Specialties currently following the patient (select all that apply):

- [ ] CT Scan (specify location)
- [ ] MRI (specify location)
- [ ] Ultrasound (specify location)
- [ ] Other imaging (specify type and location):
- [ ] Other (specify):
- [ ] None
Are there any additional details you would like to provide regarding the patient's treatment or management (optional)?

Deceased General Block

Is there a known pathogenic/likely pathogenic variant in the family?

- Yes
- No

Please provide details, including the gene/syndrome and closest degree of relation that tested positive:
Was your patient tested for the familial variant?

- Yes
- No

Did your patient have previous genetic testing?

- Yes
- No

Type of previous genetic testing (select all that apply):

- Clinical Testing
- Research Study Testing
- Healthy Screening Panel (e.g. 23andMe, Ancestry, etc.)

Previous Genetic Testing (select all that apply):

- Chromosomal Microarray (CMA)
- Karyotype
- Single Site Analysis
- Single Gene
- Gene Panel
- Whole Exome Sequencing/Whole Genome Sequencing
- Healthy Screening Panel (e.g. 23andMe, Ancestry, etc.)
- Other (specify):
Healthy Screening Panel Results - Specify gene(s)/variant(s) and classification (if available)

Chromosomal Microarray (CMA) Results

- Pathogenic/Likely Pathogenic Copy Number Variant (CNV)
- Non-Pathogenic/Likely Benign Copy Number Variant (CNV)

Chromosomal Microarray (CMA) Results - Specify CNV

Karyotype Results

- Abnormal
- Normal and/or Variant of Unknown Significance

Karyotype Results - Specify

Single Site Analysis Results

- Pathogenic/Likely Pathogenic
- Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Single Site Analysis Results - Specify gene(s) and variant(s), if known

Single Gene Results

- Pathogenic/Likely Pathogenic
Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Single Gene Results - Specify gene(s) and variant(s), if known

Gene Panel Type (e.g. Noonan Syndrome Panel, Neurodevelopmental Panel, etc.)

Gene Panel Results

Pathogenic/Likely Pathogenic
Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Gene Panel Results - Specify gene(s) and variant(s), if known

Whole Exome Sequencing/Whole Genome Sequencing Results

Pathogenic/Likely Pathogenic
Non-Pathogenic/Likely Benign/Variant of Unknown Significance

Whole Exome Sequencing/Whole Genome Sequencing Results - Specify gene(s) and variant(s), if known

Other Testing Type (e.g. Chromosomal breakage analysis, methylation studies, mitochondrial sequencing, etc.)
Other Testing Results - Specify

Have you discussed or do you plan to discuss any of the following **genetic counseling implications** with the deceased patient's **family members** prior to receiving the results of the ordered genetic test?

- [ ] Reproductive testing options
- [ ] New/Change in recurrence risk
- [ ] Identification/Testing of at-risk family members
- [ ] Other (specify):

- [ ] None

Have you discussed or do you plan to discuss any of the following with the deceased patient's **family members** prior to receiving the results of the ordered genetic test?

- [ ] Referral to support organizations
- [ ] Changes in financial planning
- [ ] Discrimination in employment and/or in obtaining insurance
- [ ] Detection of misattributed paternity or adoption
  - [ ] Legal implications (specify):

- [ ] Other (specify):

- [ ] None

Are there any additional details you would like to provide regarding the patient's **family members'** treatment or management (optional)?
Raffle Block

Thank you for completing the survey. Would you like to enter a raffle for the chance to win a $50 Amazon gift card?

- Yes
- No

Powered by Qualtrics
You are currently completing this survey for ${m://ExternalDataReference}.

Survey Cover Page

Dear Healthcare Provider:

You are invited to complete your participation in a research study exploring how the clinical management of patients may change based on the results of genetic testing.

This study is being conducted by Jill Dolinsky, MS, CGC at Ambry Genetics. Healthcare providers (e.g. physicians, genetic counselors, PhDs and nurses) who practice in the USA and are involved in genetic testing are welcome to participate. This survey relates to your patient:

Patient Name: ${m://ExternalDataReference}

- This is the second survey in a series of two surveys about this patient. You completed the first survey prior to the reporting of genetic testing results. This survey should be completed after the receipt of genetic testing results.
- This survey should take approximately 5-10 minutes to complete.
- Please respond based on the treatment/management options discussed with your patient rather than just the options your patient elected to pursue.
- Participation is voluntary and you may decide to leave the survey at any time.
- De-identified clinical and test result information about your patient may be included in the analysis of your responses. In no way will your responses or your patient’s information be identifiable to the investigators.
- After completing the survey you will have the option of entering a drawing for a $100 Amazon gift card. Drawings will be held four times a year on a quarterly basis. If you wish to enter the drawing you will be prompted to enter your email address, which will be stored separately from your survey responses.

Should you have any questions regarding this research study you may contact the study team at ambrystudies@ambrygen.com.
By continuing with the survey, you acknowledge that you have read the information above and consent to participate in this survey. Thank you in advance for your time and consideration.

Sincerely,

Jill Dolinsky, RN, MS, CGC
Director, Clinical Affairs
jdolinsky@ambrygen.com

Starting Block

Genetic testing category for this patient:

- Oncology
- Cardiology
- Neurology/General Genetics

Is your patient living?

- Yes
- No

Alive Oncology Block

Based on your patient's genetic test results, do you recommend increases in frequency/earlier age at onset/additions to screening and/or interventions for any of the following cancers? Select all that apply:

- Brain
- Breast
- Colorectal
- Leukemia/Lymphoma
- Melanoma
Based on your patient’s genetic test results, do you recommend decreases in frequency/delay in age at onset/discontinuation of screening and/or interventions for any of the following cancers? Select all that apply:

- Ovarian
- Pancreatic
- Prostate
- Uterine
- GI polyps

Other type (specify):

- No recommendations for increased screening/intervention

Based on your patient’s genetic test results, do you recommend recommended increase in frequency/earlier age of onset/additions to brain cancer screening/interventions that were discussed with the patient (select all that apply):

- Biopsy
- Imaging
- Surgery
- Medications
- Radiation
Based on your patient's genetic test results, recommended decrease in frequency/delay in age of onset/discontinuation of brain cancer screening/interventions that were discussed with the patient (select all that apply):

- Biopsy
- Imaging
- Surgery
- Medications
- Radiation
- Systemic therapy/chemotherapy
- Clinical Trial
- Referral to Specialist
- Recommendations for changes in lifestyle/health habits
- Patient education in symptom awareness
- Other (specify):

Based on your patient's genetic test results, recommended increase in frequency/earlier age of onset/additions to breast cancer screening/interventions that were discussed with the patient (select all that apply):

- Self-Breast Exam
- Biopsy
- Breast MRI
- Digital/Analog Mammogram (MMG)
- Tomosynthesis/3D Mammogram
- Ultrasound
- Risk-Reducing Mastectomy (RRM)
Based on your patient's genetic test results, recommended **decrease in frequency/delay in age of onset/dischotoination** of **breast cancer** screening/interventions that were discussed with the patient (select all that apply):

- Risk-Reducing Salpingo-Oophorectomy (RRSO)
- Chemopreventive Medications
- Radiation
- Systemic therapy/chemotherapy
- Clinical Trial
- Referral to Specialist
- Recommendations for changes in lifestyle/health habits
- Patient education in symptom awareness
- Other (specify):
Based on your patient's genetic test results, recommended **increase in frequency/earlier age at onset/additions** to colorectal cancer screening/interventions that were discussed with the patient (select all that apply):

- [ ] Biopsy
- [ ] Colectomy
- [ ] Colonoscopy
- [ ] Sigmoidoscopy
- [ ] Upper Endoscopy
- [ ] Video Capsule Endoscopy
- [ ] Chemopreventive Medications
- [ ] Radiation
- [ ] Systemic therapy/chemotherapy
- [ ] Clinical Trial
- [ ] Referral to Specialist
- [ ] Recommendations for changes to lifestyle/health habits
- [ ] Patient education in symptom awareness
- [ ] Other (specify):

Based on your patient's genetic test results, recommended **decrease in frequency/delay in age at onset/discontinuation** of colorectal cancer screening/interventions that were discussed with the patient (select all that apply):

- [ ] Biopsy
- [ ] Colectomy
- [ ] Colonoscopy
- [ ] Sigmoidoscopy
- [ ] Upper Endoscopy
- [ ] Video Capsule Endoscopy
- [ ] Chemopreventive Medications
- [ ] Radiation
- [ ] Systemic therapy/chemotherapy
- [ ] Clinical Trial
- [ ] Referral to Specialist
Based on your patient's genetic test results, recommended increase in frequency/earlier age at onset/additions to melanoma screening/interventions that were discussed with the patient (select all that apply):

- Biopsy
- Skin Cancer Screening
- Eye Exam/Ocular Melanoma Screening
- Sun Screen Agents
- Medications
- Radiation
- Systemic therapy/chemotherapy
- Clinical Trial
- Referral to Specialist
- Recommendations for changes in lifestyle/health habits
- Patient education in symptom awareness

Other (specify):

Based on your patient's genetic test results, recommended decrease in frequency/delay in age at onset/discontinuation of melanoma screening/interventions that were discussed with the patient (select all that apply):

- Biopsy
- Skin Cancer Screening
- Eye Exam/Ocular Melanoma Screening
- Sun Screen Agents
- Medications
- Radiation
- Systemic therapy/chemotherapy
- Clinical Trial
- Referral to Specialist
- Recommendations for changes in lifestyle/health habits
- Patient education in symptom awareness

Other (specify):
Based on your patient's genetic test results, recommended **increase in frequency/earlier age at onset/additions** to **ovarian cancer** screening/interventions that were discussed with the patient (select all that apply):

- [ ] Biopsy
- [ ] CA-125
- [ ] Transvaginal Ultrasound
- [ ] Salpingo-Oophorectomy
- [ ] Bilateral Salpingo-Oophorectomy (BSO)
- [ ] Total Abdominal or Laparoscopic Hysterectomy and Bilateral Salpingo-Oophorectomy (TAH-BSO)
- [ ] Chemopreventive Medications
- [ ] Oral Contraceptives (OCP)
- [ ] Radiation
- [ ] Systemic therapy/chemotherapy
- [ ] Clinical Trial
- [ ] Referral to Specialist
- [ ] Recommendations for changes in lifestyle/health habits
- [ ] Patient education in symptom awareness
- [ ] Other (specify):

Based on your patient's genetic test results, recommended **decrease in frequency/delay in age at onset/discontinuation** of **ovarian cancer** screening/interventions that were discussed with the patient (select all that apply):

- [ ] Biopsy
- [ ] CA-125
- [ ] Transvaginal Ultrasound
- [ ] Salpingo-Oophorectomy
- [ ] Bilateral Salpingo-Oophorectomy (BSO)
Based on your patient's genetic test results, recommended increase in frequency/earlier age at onset/additions to pancreatic cancer screening/interventions that were discussed with the patient (select all that apply):

- Total Abdominal or Laparoscopic Hysterectomy and Bilateral Salpingo-Oophorectomy (TAH-BSO)
- Chemopreventive Medications
- Oral Contraceptives (OCP)
- Radiation
- Systemic therapy/chemotherapy
- Clinical Trial
- Referral to Specialist
- Recommendations for changes in lifestyle/health habits
- Patient education in symptom awareness
- Other (specify):

Abdominal Ultrasound
- Angiography
- Biopsy
- CA 19-9
- Carcinoembryonic Antigen (CEA)
- CT Scan
- Endoscopic Ultrasound
- Endoscopic Retrograde Cholangiopancreatography (ERCP)
- Liver Function Tests
- MR Angiography (MRA)
- MR Cholangiopancreatography (MRCP)
- Percutaneous Transhepatic Cholangiography (PTC)
- Positron Emission Tomography (PET)
- Somatostatin Receptor Scintigraphy (SCS)
- Medications
- Radiation
- Systemic therapy/chemotherapy
- Clinical Trial
- Referral to Specialist
Based on your patient's genetic test results, recommended **decrease in frequency/delay in age at onset/discontinuation** of **pancreatic cancer** screening/interventions that were discussed with the patient (select all that apply):

- [ ] Abdominal Ultrasound
- [ ] Angiography
- [ ] Biopsy
- [ ] CA 19-9
- [ ] Carcinoembryonic Antigen (CEA)
- [ ] CT Scan
- [ ] Endoscopic Ultrasound
- [ ] Endoscopic Retrograde Cholangiopancreatography (ERCP)
- [ ] Liver Function Tests
- [ ] MR Angiography (MRA)
- [ ] MR Cholangiopancreatography (MRCP)
- [ ] Percutaneous Transhepatic Cholangiography (PTC)
- [ ] Positron Emission Tomography (PET)
- [ ] Somatostatin Receptor Scintigraphy (SCS)
- [ ] Medications
- [ ] Radiation
- [ ] Systemic therapy/chemotherapy
- [ ] Clinical Trial
- [ ] Referral to Specialist
- [ ] Recommendations for changes in lifestyle/health habits
- [ ] Patient education in symptom awareness

Other Blood Tests (specify):

Other (specify):
Based on your patient's genetic test results, recommended **increase in frequency/earlier age at onset/additions** to **prostate cancer** screening/interventions that were discussed with the patient (select all that apply):

- [ ] Biopsy
- [ ] Prostate Specific Antigen (PSA)
- [ ] Prostate MRI
- [ ] Hormone Therapy
- [ ] Radiotherapy
- [ ] Prostatectomy
- [ ] Medications
- [ ] Radiation
- [ ] Systemic therapy/chemotherapy
- [ ] Clinical Trial
- [ ] Referral to Specialist
- [ ] Recommendations for changes in lifestyle/health habits
- [ ] Patient education in symptom awareness
- [ ] Other (specify):

Based on your patient's genetic test results, recommended **decrease in frequency/delay in age at onset/discontinuation** of **prostate cancer** screening/interventions that were discussed with the patient (select all that apply):

- [ ] Biopsy
- [ ] Prostate Specific Antigen (PSA)
- [ ] Prostate MRI
- [ ] Hormone Therapy
- [ ] Radiotherapy
- [ ] Prostatectomy
- [ ] Medications
- [ ] Radiation
- [ ] Systemic therapy/chemotherapy
- [ ] Clinical Trial
- [ ] Referral to Specialist
Based on your patient's genetic test results, recommended **increase in frequency/earlier age at onset/additions** to uterine cancer screening/interventions that were discussed with the patient (select all that apply):

- Biopsy
- CA-125
- CT Scan
- MRI
- Cystoscopy and Proctoscopy
- Hysteroscopy
- Ultrasound
- Dilation and Curettage (D&C)
- Medications
- Radiation
- Systemic therapy/chemotherapy
- Clinical Trial
- Referral to Specialist
- Recommendations for changes in lifestyle/health habits
- Patient education in symptom awareness
- Other Blood Tests (specify):
- Other (specify):

Based on your patient's genetic test results, recommended **decrease in frequency/delay in age at onset/discontinuation** of uterine cancer screening/interventions that were discussed with the patient (select all that apply):

- Biopsy
- CA-125
- CT Scan
- MRI
Based on your patient's genetic test results, recommended **increase in frequency/earlier age at onset/additions** to **leukemia/lymphoma** screening/interventions that were discussed with the patient (select all that apply):

- [ ] Biopsy
- [ ] Bone Marrow Aspiration and Biopsy
- [ ] Lumbar Puncture
- [ ] Complete Blood Count (CBC)
- [ ] Chest X-Ray or Chest CT Scan
- [ ] Blood Transfusion
- [ ] Stem Cell Transplant
- [ ] Radiotherapy
- [ ] Medications
- [ ] Radiation
- [ ] Systemic therapy/chemotherapy
- [ ] Clinical Trial
- [ ] Referral to Specialist
- [ ] Recommendations for changes in lifestyle/health habits
- [ ] Patient education on symptom awareness
- [ ] Other Blood Tests (specify):
- [ ] Other (specify):
Based on your patient's genetic test results, recommended decrease in frequency/delay in age at onset/discontinuation of leukemia/lymphoma screening/interventions that were discussed with the patient (select all that apply):

- Biopsy
- Bone Marrow Aspiration and Biopsy
- Lumbar Puncture
- Complete Blood Count (CBC)
- Chest X-Ray or Chest CT Scan
- Blood Transfusion
- Stem Cell Transplant
- Radiotherapy
- Medications
- Radiation
- Systemic therapy/chemotherapy
- Clinical Trial
- Referral to Specialist
- Recommendations for changes in lifestyle/health habits
- Patient education on symptom awareness
- Other Blood Tests (specify):
- Other (specify):

Based on your patient's genetic test results, recommended increase in frequency/earlier age at onset/additions to Other Cancer screening/interventions that were discussed with the patient (select all that apply):

- Biopsy
- Blood Testing
- CT Scan
- MRI
- Ultrasound
- Referral to Specialist
Based on your patient's genetic test results, recommended **decrease in frequency/delay in age at onset/discontinuation** of **Other Cancer** screening/interventions that were discussed with the patient (select all that apply):

- [ ] Biopsy
- [ ] Blood Testing
- [ ] CT Scan
- [ ] MRI
- [ ] Ultrasound
- [ ] Referral to Specialist
- [ ] Medications
- [ ] Radiation
- [ ] Systemic therapy/chemotherapy
- [ ] Clinical Trial
- [ ] Recommendations for changes in lifestyle/health habits
- [ ] Patient education in symptom awareness
- [ ] Other Imaging (specify):
- [ ] Other (specify):

Based on your patient's genetic test results, recommended **increase in frequency/earlier age at onset/additions** to **GI polyps** screening/interventions that were discussed with the patient (select all that apply):

- [ ] Biopsy
- [ ] Blood Testing
Based on your patient's genetic test results, recommended decrease in frequency/delay in age at onset/discontinuation of GI polyps screening/interventions that were discussed with the patient (select all that apply):

- Biopsy
- Blood Testing
- CT Scan
- MRI
- Ultrasound
- Clinical Trial
- Referral to Specialist
- Medications
- Chemopreventive medications
- Recommendations for changes in lifestyle/health habits
- Patient education in symptom awareness

Other Imaging (specify):

Other (specify):

Based on your patient's genetic test results, have you discussed any of the following genetic counseling implications with your patient? Select all that apply:
Based on your patient’s genetic test results, have you discussed any of the following with your patient? Select all that apply:

- Reproductive testing options
- New/change in recurrence risk
- Testing/identification of at risk family members
- Other (specify):
- None

Do any increases/decreases/omissions in the screening/interventions options described previously differ from published management recommendations?

- Yes
- No
- I’m not sure

From which organization do your recommendations differ (select all that apply)?

- National Comprehensive Cancer Network (NCCN)
- American College of Gastroenterology (ACG)
- International Gastric Cancer Linkage Consortium (IGCLC)
- International Cancer of the Pancreas Screening (CAPS) Consortium
- Other (please specify):
Please describe the reason(s) for recommendations that differ from published recommendations (select all that apply):

- [ ] Patient wishes
- [ ] Financial/Insurance reasons
- [ ] Medical condition/health status that prevents intervention
- [ ] Limited evidence for published recommendation
- [ ] No access/Service unavailable
- [ ] Referred to specialist for management discussion
- [ ] Other (please specify):

Are there any additional details you would like to provide regarding the impact of your patient's genetic testing results on their treatment or medical management (optional)?

Deceased Oncology Block

Based on your patient's genetic testing results, have you discussed any of the following genetic counseling implications with his/her family members? Select all that apply:

- [ ] Reproductive testing options
- [ ] New/change in recurrence risk
- [ ] Identification/Testing of at-risk family members
- [ ] Other (specify):
- [ ] None

Based on your patient's genetic testing results, have you discussed any of the following with his/her family members? Select all that apply:

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Are there any additional details you would like to provide regarding the impact of your patient's genetic testing results on his/her family members (optional)?

Alive Cardiology Block

Based on your patient’s genetic testing results, do you recommend increases in/additions of any of the following in their current screening/monitoring plan? Select all that apply:

- Blood pressure monitoring
- Cardiac MRI
- Chest X-Ray/Angiography
- CT Scan
- Echocardiogram
- Electrophysiology study (EPS)
- Exercise stress test
- 24-hour ECG monitoring (Holter)
- Pulse monitoring

Other (specify):
Based on your patient's genetic testing results, do you recommend **decreases** in/discontinuation of any of the following in their current screening/monitoring plan? Select all that apply:

- [ ] Blood pressure monitoring
- [ ] Cardiac MRI
- [ ] Chest X-Ray/Angiography
- [ ] CT Scan
- [ ] Echocardiogram
- [ ] Electrophysiology study (EPS)
- [ ] Exercise stress test
- [ ] 24-hour ECG monitoring (Holter)
- [ ] Pulse monitoring
- [ ] Other (specify):
- [ ] No recommendations for decreased screening/monitoring

Based on your patient's genetic testing results, do you recommend **increases** in/addition of any of the following interventions in their current management plan? Select all that apply:

- [ ] Ablation
- [ ] Cardiac catheterization/angiogram
- [ ] Implantable Cardioverter Defibrillator (ICD)
- [ ] Pacemaker
- [ ] Lipoprotein apheresis
- [ ] Lifestyle modification
- [ ] Stimulant avoidance
- [ ] Other (specify):
- [ ] No recommendations for increased interventions
Based on your patient's genetic testing results, do you recommend **decreases in/discontinuation of** any of the following interventions in their current management plan? Select all that apply:

- Ablation
- Cardiac catheterization/angiogram
- Implantable Cardioverter Defibrillator (ICD)
- Pacemaker
- Lipoprotein apheresis
- Lifestyle modification
- Stimulant avoidance
- Other (specify):
- No recommendations for decreased interventions

Based on your patient's genetic testing results, do you recommend modifications to their current medication regimen? Select all that apply:

- **Increase** in medication dose
- **Decrease** in medication dose
- **Additional** medication
- **Discontinuation** of medication
- No recommended modifications

Please specify recommended **increase** in dose of any of the following medications (select all that apply):

- Anticoagulants
- Antiplatelet agents and Dual Antiplatelet Therapy (DAPT)
- Angiotensin-converting enzyme (ACE) Inhibitors
- Angiotensin II receptor blockers (ARBs)
- Alpha-adrenoreceptor antagonists
- Beta-adrenergic blocking agents
- Combined alpha- and beta-blocking agents
- Cholesterol lowering medications
Please specify recommended decrease in dose of any of the following medications (select all that apply):

- [ ] Anticoagulants
- [ ] Antiplatelet agents and Dual Antiplatelet Therapy (DAPT)
- [ ] Angiotensin-converting enzyme (ACE) Inhibitors
- [ ] Angiotensin II receptor blockers (ARBs)
- [ ] Alpha-adrenoreceptor antagonists
- [ ] Beta-adrenergic blocking agents
- [ ] Combined alpha- and beta-blocking agents
- [ ] Cholesterol lowering medications
- [ ] Calcium channel antagonists
- [ ] Digitalis preparations
- [ ] Diuretics
- [ ] Pain Medications
- [ ] Sodium channel antagonists
- [ ] Supplements
- [ ] Vasodilators
- [ ] Other (specify): ___

Please specify recommended addition of any of the following to your patient's current medication regimen (select all that apply):

- [ ] Anticoagulants
- [ ] Antiplatelet agents and Dual Antiplatelet Therapy (DAPT)
Please specify recommended **discontinuation** of any of the following in your patient's current medication regimen (select all that apply):

- Anticoagulants
- Antiplatelet agents and Dual Antiplatelet Therapy (DAPT)
- Angiotensin-converting enzyme (ACE) Inhibitors
- Angiotensin II receptor blockers (ARBs)
- Alpha-adrenoreceptor antagonists
- Beta-adrenergic blocking agents
- Combined alpha- and beta-blocking agents
- Cholesterol lowering medications
- Calcium channel antagonists
- Digitalis preparations
- Diuretics
- Pain Medications
- Sodium channel antagonists
- Supplements
- Vasodilators

Other (specify):
Based on your patient's genetic test results, have you discussed any of the following genetic counseling implications with your patient? Select all that apply:

- Reproductive testing options
- New/change in recurrence risk
- Identification/Testing of at-risk family members
- Other (specify):
- None

Based on your patient's genetic test results, have you discussed any of the following with your patient? Select all that apply:

- Referral to support organizations
- School interventions (IEP, etc.)
- Changes in financial planning
- Discrimination in employment and/or in obtaining insurance
- Detection of misattributed paternity or adoption
- Other (specify):

Are there any additional details you would like to provide regarding the impact of your patient's genetic testing results on their treatment or medical management (optional)?

Deceased Cardiology Block
Based on your patient’s genetic testing results, have you discussed any of the following **genetic counseling implications** with their **family members**? Select all that apply:

- [ ] Reproductive testing options
- [ ] New/Change in recurrence risk
- [ ] Identification/Testing of at-risk family members
- [ ] Other (specify):
- [ ] None

Based on your patient’s genetic testing results, have you discussed any of the following with his/her **family members**? Select all that apply:

- [ ] Referral to support organizations
- [ ] Changes in financial planning
- [ ] Discrimination in employment and/or in obtaining insurance
- [ ] Detection of misattributed paternity or adoption
- [ ] Other (specify):
- [ ] Legal implications (specify):
- [ ] Other (specify):
- [ ] None

Are there any additional details you would like to provide regarding the impact of your patient’s genetic testing results on his/her **family members** (optional)?

Alive General Block

Based on your patient’s genetic testing results, do you recommend **increases** in/additions of any of the following in their current screening/monitoring plan? Select all
Based on your patient's genetic testing results, do you recommend decreases in/discontinuation of any of the following in their current screening/monitoring plan? Select all that apply:

- Biochemical testing
- Bone Age
- Echocardiogram
- EEG
- EMG
- X-Ray/Skeletal Survey
- CT Scan (specify location)
- MRI (specify location)
- Ultrasound (specify location)
- Other imaging (specify type and location):
- Other (specify):
- No recommendations for increased screening/monitoring

Based on your patient's genetic testing results, do you recommend decreases in/discontinuation of any of the following in their current screening/monitoring plan? Select all that apply:

- Biochemical testing
- Bone Age
- Echocardiogram
- EEG
- EMG
- X-Ray/Skeletal Survey
- CT Scan (specify location)
- MRI (specify location)
- Ultrasound (specify location)
- Other imaging (specify type and location):
- Other (specify):
- No recommendations for decreased screening/monitoring
Based on your patient's genetic testing results, do you recommend **modification** *(i.e., increase/decrease)* of any of the following interventions in their current management plan? Select all that apply:

- [ ] Modification of dietary intervention
- [ ] Increase/decrease of current dose in **medication** regimen
- [ ] Increase/decrease of current dose in **supplement** regimen
- [ ] Other (specify):
- [ ] No recommendations for modification of current interventions

Please specify modifications to your patient's current dietary intervention:



Please specify modifications to current doses in your patient's **medication** regimen (include medication and if increased/decreased):



Please specify modifications to current doses in your patient's **supplement** regimen (include supplement and if increased/decreased):



Based on your patient's genetic testing results, do you recommend **addition of** any of the following interventions in their current management plan? Select all that apply:
Please specify recommended **addition of** dietary intervention:



Please specify recommended **medication addition** to your patient's current regimen:



Please specify recommended **supplement addition** to your patient's current regimen:



Based on your patient's genetic testing results, do you recommend **discontinuation of** any of the following interventions in their current management plan? Select all that apply:

- Discontinuation of dietary intervention
- Discontinuation of **medication** in current regimen
- Discontinuation of **supplement** in current regimen
Please specify recommended **discontinuation of** dietary intervention:

Please specify recommended **discontinuation of medication** in your patient's current regimen:

Please specify recommended **discontinuation of supplement** in your patient's current regimen:

Based on your patient's genetic testing results, do you recommend any **additional** referrals to specialists for this patient? Select all that apply:

- [ ] Cardiology
- [ ] Developmental/Behavioral Pediatrics
- [ ] Endocrinology
- [ ] Gastroenterology
- [ ] Genetics
Based on your patient's genetic testing results, do you recommend the patient **discontinue** seeing any specialists previously treating this patient? Select all that apply:

- [ ] Cardiology
- [ ] Developmental/Behavioral Pediatrics
- [ ] Endocrinology
- [ ] Gastroenterology
- [ ] Genetics
- [ ] Hematology/Oncology
- [ ] Nephrology
- [ ] Neurology
- [ ] Pulmonology

□ Other (specify):

- [ ] No recommendation for additional specialists

Based on your patient's genetic testing results, have you discussed any of the following **genetic counseling implications** with your patient?

- [ ] Referral to support organizations
- [ ] School interventions (e.g. IEP, etc.)
- [ ] Changes in financial planning
- [ ] Discrimination in employment and/or in obtaining insurance
- [ ] Detection of misattributed paternity or adoption

□ Other (specify):

- [ ] None
Based on your patient's genetic testing results, have you discussed any of the following with your patient?

- [ ] Reproductive testing options
- [ ] New/change in recurrence risk
- [ ] Identification/Testing of at-risk family members
- [ ] Other (specify):
- [ ] None

Are there any additional details you would like to provide regarding the impact of your patient's genetic testing results on their treatment or management (optional)?

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Deceased General Block

Based on your patient's genetic testing results, have you discussed any of the following genetic counseling implications with their family members? Select all that apply:

- [ ] Reproductive testing options
- [ ] New/change in recurrence risk
- [ ] Identification/Testing of at-risk family members
- [ ] Other (specify):
- [ ] None

Based on your patient's genetic testing results, have you discussed any of the following with his/her family members? Select all that apply:
Referral to support organizations

Discrimination in employment and/or in obtaining insurance

Detection of misattributed paternity or adoption

Legal implications (specify):

Other (specify):

None

Are there any additional details you would like to provide regarding the impact of your patient's genetic testing results on his/her family members (optional)?

Raffle Block

Thank you for completing the survey. Would you like to enter a raffle for the chance to win a $100 gift card?

Yes

No

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