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App: Healthcare Pro

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Healthcare Professional

Genetic Report

Confidential Identification Number

7941848

Confidential Information

Report Sections

Section 1
STAT Alerts

Please note that the following information is based solely on an analysis of the pt's genes and does not take into account non-genetic factors that may also significantly affect the pt's risk of a disease or response to a medication, such as the pt's age, BMI, lifestyle, habits, and medical conditions as well as any supplements and medications the pt may be taking. These non-genetic factors may be important to take into consideration in-addition to the genetic analysis presented below.

Please also note that the information in this report is based upon current research pertaining to the genetic basis of disease and pharmacogenomics and that this may change overtime as additional genetic research is conducted. Reanalysis of the pt's genetic data throughout the pt's life may therefore be beneficial in making sure the pt's genetic analysis is always up-to-date.

- <<**ALERT**>>Thrombophilia: **Factor V Leiden Detected**. Patient has one Factor V Leiden mutation, which increases risk of thrombophilia.

    - On its own, a single Factor V Leiden mutation only increases risk of thrombosis a small amount. Thrombosis is more likely to manifest and cause DVT, PE, MI, or CVA if combined with one or more the following non-genetic factors:
        - smoking cigarettes
        - being overweight or obese
        - medications and illicit drugs that increase the risk of thrombosis, such as OCPs, HRT, and cocaine
        - prolonged states of limited mobility such as may occur when traveling, post-op, or for certain professions (i.e. working inside a tank in the military)
    - Whether there is an increased risk of DVTs due to long plane or car rides is still not conclusively known. Regardless, consider educating pt that while on flights or car rides longer than a couple hours, getting up and walking around for 5-10 mins every hour may help decrease risk of DVTs.
    - Consider prophylaxis against thrombosis if clinically appropriate.
        - Non-pharmacologic intervention may include anti-embolism compression stockings
        - Depending on clinical assessment, the pt may not need pharmacologic prophylaxis. If meds are warranted, they may include one or more of the following:
            - Fish Oil supplements
            - Aspirin
                - Based upon a further analysis of this pt's genes, aspirin is likely to be effective in decreasing risk of MIs and CVAs.
            - Clopidogrel (Plavix®)
                - Based upon a further analysis of this pt's genes, clopidogrel is likely to be effective as an antiplatelet agent at routine dosing.
            - Warfarin
                - Please see the medication section for specific information on the likely optimal warfarin dose based upon this pt's genes.
            - Heparin

    - Consider educating the pt on warning signs of a DVT and that if a DVT develops, it may be a medical emergency that can cause death if not treated.

- Pt should understand the importance of seeking medical care immediately if a DVT is suspected and that most ERs can not only assess whether a blood clot exists but can also start the pt on appropriate interventions, such as fast acting blood thinning medications, if necessary.

- Counsel pt on importance of alerting other doctors, including surgeons, anesthesiologists, internists, and any doctor that may prescribe medication that the pt has the Factor V Leiden mutation and may be predisposed to blood clots.

Section 2
Increased Risk Detected

Please note that the following information is based solely on an analysis of the pt's genes and does not take into account non-genetic factors that may also significantly affect the pt's risk of a disease, such as the pt's age, BMI, lifestyle, habits, and medical conditions as well as any supplements and medications the pt may be taking. These non-genetic factors may be important to take into consideration in-addition to the genetic analysis presented below.

Please also note that the information in this report is based upon current research pertaining to the genetic basis of disease and that this may change overtime as additional genetic research is conducted. Reanalysis of the pt's genetic data throughout the pt's life may therefore be beneficial in making sure the pt's genetic analysis is always up-to-date.

- CAD & MI: Increased Risk Detected**.** This patient has a moderate risk of heart attack (68% lifetime risk compared to the general population risk of 42% for men). Solely based upon the pt's genes, the pt is at risk of early-onset CAD & MI. Because of this, the pt is at risk both before and after the age of 50 – consider instituting CAD screening and preventions starting at age 21 or within the next few months if patient is older than 21 and hasn't received a cardiovascular workup by a physician within the last year. Preventive measures include:

    - Annual cholesterol level screening as per routine screening guidelines and instituting interventions if high cholesterol levels are detected.
        - Statins
            - Based upon a further analysis of this pt's genes, the pt is at low risk of adverse reactions with statins.
        - Aspirin
            - Based upon a further analysis of this pt's genes, aspirin is likely to be effective in decreasing risk of MIs and CVAs.
        - Clopidogrel (Plavix®)
            - Based upon a further analysis of this pt's genes, clopidogrel is likely to be effective as an antiplatelet agent at routine dosing.
        - Warfarin
            - Please see the medication section for specific information on the likely optimal warfarin dose based upon this pt's genes.

    - Consider monitoring CRP and homocysteine levels. Even if cholesterol levels are WNL, consider starting this pt on a statin if CRP levels are elevated.
        - This pt is at low risk of experiencing myopathy with statins.
    - Regular cardiovascular exercise.

    - Counsel patient on benefits of a low fat/low cholesterol diet.

    - While a low fat/low cholesterol diet is a general target for all individuals, a study by Do (2011) found that the risk of MI due to the specific genetic variants detected in this pt may be significantly reduced if the pt eats at least one serving of raw vegetables (especially green leafy vegetables) and/or one serving of fruit during meals (lunch, dinner, and snacks).
        - The pt has a specific change in a taste bud gene indicates that, unlike some people, the pt should be able to tolerate the taste of cruciferous vegetables even if they are eaten on their own.

- For complementary prevention, consider:
  - Fish Oil (Omega-3) supplementation such as 1,200mg/day
  - 1-2 glasses of red wine or grape juice per day

- The pt's genetic risk of CAD and MI is also linked to a risk of periodontitis.
  - Preliminary research indicates that good oral hygiene and yearly dental visits may reduce the risk of both periodontitis and heart disease.

- Consider discussing with the pt that this genetic risk assessment is solely based upon their genes and does not take into account lifestyle factors that may cause the pt's *total* risk (genetic + nongenetic factors) of CAD and MI to be higher or lower than the pt's genetic risk. For example, if the pt has led a sedentary lifestyle for many years and is overweight, then the pt's total risk of CAD and MI is likely higher than 68%.

- Salt-sensitive Hypertension: Increased Risk Detected. Due to mutations in the AGT gene, this pt may be predisposed to salt-sensitive HTN.
  - Monitor pt's blood pressure each year.
  - If elevated HTN or borderline HTN is detected, counsel patient on decreasing sodium intake and consider low-sodium diets such as the lower sodium DASH diet (max of 1,500mg of sodium/day).

- Atrial Fibrillation: Increased Risk Detected. Based on this patient's genes, the lifetime risk of a-fib is increased from ~26% to ~40-50%. While it is not yet possible to predict the age of onset of a-fib via a genetic test, many times the age of diagnosis even in those genetically predisposed to this condition is in the pt's 60's, 70's or 80's.
  - Prevention includes abstaining from drinking alcohol as alcohol may trigger the onset of a-fib.
  - Surveillance measures may include taking a baseline EKG, conducting a work-up for a-fib if the patient ever has any symptoms, and educating the patient on symptoms associated with a-fib so that if they manifest, the patient will know to contact the pt's healthcare provider.
  - Even if the pt is asymptomatic, consider performing an EKG to have a baseline EKG in the pt's chart.
  - Consider counseling pt on the symptoms associated with a-fib.

- Osteoarthritis: Increased Risk Detected.
  - Knee OA = Slightly Increased (41% lifetime risk compared to the population lifetime risk of 40%)
  - Hip OA = Slightly Increased (27% compared to 25%)
  - Wrist OA = Slightly Increased (33% compared to 30%)
    - The patient has an increased risk of knee, hip, wrist OA. Consider counseling pt on decreasing impact and repetitive stress upon those specific joints through the pt's life, such as during physical exercise and sports.
      - Pt should still continue to perform cardiovascular exercise and resistance training as well as playing sports but consider focusing on low-impact exercises for those joints that have a higher risk of OA.
      - For cardiovascular exercise, consider biking, using the elliptical machine, or swimming as opposed to running or jogging in-order to decrease repetitive stress to the knees and hips.
      - While skiing a couple times a year should be fine, consider counseling pt to avoid skiing on a frequently basis as this sport entails tremendous stress upon the knees and hips.

- Counsel pt on using correct posture during ambulation. Also consider an orthotic referral for feet orthotics if optimizing pt's foot alignment is warranted, such as if the pt stands during a significant portion of the day or if pt is experiencing pain or discomfort in the feet, knees, or hips.
- Avoid or significantly limit boxing, heavy overhead lifting, and other activities that put significant strain upon the wrist and hand joints.
- Counsel patient on using correct posture and ergonomics when typing and using the mouse in-order to limit repetitive strain on the wrists and hands.

- While unneeded exposure to medical radiation should always be avoided, based on a further assessment of the pt's genes, this pt is not genetically predisposed to radiation-induced CA.
  - Because of this, x-ray imaging may be an appropriate method to screen for and monitor arthritis.
- Consider counseling pt on the importance of weight control in helping to mitigate risk of OA.

- Colorectal Cancer: Increased Risk Detected. This patient contains a set of genetic markers that slightly increases lifetime risk of CRC from ~7% to ~10%. Preventive measures to consider include:
  - Colonoscopy and other CRC screening exams according to routine CRC screening guidelines.
  - Avoidance of smoking, consuming processed meats, and significantly decrease consumption of red meat to no more than one time every other week (max two times per month).
  - Checking vitamin D levels at least once a year to make sure they are WNL as vitamin D deficiency may further increase risk of colorectal cancer.
    - Consider vitamin D_3 supplementation (such as 1,000mg/day) if vitamin D levels are low or if deemed worthwhile as a CRC risk-reduction supplement.
  - Regular cardiovascular exercise (three or more times/week for 30mins or longer) has been shown to significantly decrease the risk of CRC.

- Melanoma: Risk Detected. This patient has a moderate risk of melanoma (lifetime risk between 4-10%).
  - Due to the pt's high risk of melanoma, consider referral to a dermatologist in-order to perform full body skin exams as indicated for high risk individuals, such as every 9-12 months.
  - Counsel pt on the importance of avoiding the sun and conducting self-skin exams.
    - Consider educating the pt with images of melanoma, especially at early stages, so that the pt is familiar with how melanoma may present. Pictures of melanoma can be found online, such as at this site: www.mayoclinic.com/health/melanoma/DS00575
    - Consider reinforcing the need to strictly avoid tanning beds throughout life.
  - As a child, the patient should be protected from sunburns through the use of sunscreen as well as sun-protective hats and clothing. Children with increased risk of melanoma may also benefit from wearing sunglasses while outside on a sunny day in-order to protect their eyes from ocular melanoma.
  - Consider genetic testing for all first degree and possibly second degree relatives in-order to identify any other family members that may also have a very high risk of melanoma and benefit from preventive measures.

- Brain Aneurysm: Increased Risk Detected. The patient's lifetime risk of a brain aneurysm is increased from ~5% to ~7%. This primarily results from mutations in genes that cause a slight weakness of blood vessel walls. While the pt's blood vessels function normally, the slight structural weakness predisposes them to aneurysm formation if the vessel is exposed to long periods of increased pressure.
  - The primary preventive measure is to monitor for hypertension and be aggressive in treating/controlling

both HTN and borderline HTN.

- Noise-induced Hearing Loss: Increased Risk Detected. This patient is at increased risk of permanent hearing loss following repetitive exposure to loud noise, such as very loud music or construction equipment.
  - Preventive measures include avoiding exposure to loud noise, such as wearing hearing protection if going to a concert or in a situation with repetitive loud noise (such as a helicopter).
  - iPods, when used with headphones, have been shown to be able to pass the decibel threshold necessary to cause trauma to the auditory system.
    - Consider counseling pt on avoidance of listening to music at excessively loud volumes, especially when wearing headphones. The pt can still listen to an iPod with headphones without a risk of pathology as long as moderate volumes are maintained.

- Asthma: Increased Risk Detected. This patient is at increased risk of bronchial hyperreactivity and asthma, especially if the pt is exposed to cigarette smoke during childhood.
  - If pt is a newborn or child, consider counseling patient's family on the importance of aggressively avoiding exposing the pt to any secondhand cigarette smoke, including smoke particles that may cling to a person's clothing (for example, as a child this pt should not be held by, or come into contact with, an adult that was smoking that day even if the person is not smoking at that exact moment).

- Disseminated infection with Mycobacterium avium and Salmonella enteritidis: Increased Risk Detected. Patient is at significantly increased risk of disseminated infection with Mycobacterium avium and Salmonella enteritidis.
  - Consider counseling pt on avoiding travel to endemic areas.
  - Also consider counseling pt on symptoms associated with infection with these organisms so that pt knows to seek medical treatment if symptoms manifest.

- Hirschsprung Disease: Increased Risk Detected. Due to a mutation in the RET gene(referred to in genetic terms as IVS 1, ds, +9277 and also as +9349 relative to MET) this patient has a 5.7 fold increased risk of Hirschsprung Disease. This disease results in absence of nerves at the end of the large intestine, which can cause abnormalities with bowel function.
  - This does *not* mean the patient has Hirschsprung Disease, only that the pt is at increased risk compared to the general population due to a mutation in one of the pt's RET genes.
    - Symptoms of Hirschsprung Disease include failure to pass meconium, constipation, watery diarrhea, poor feeding, poor weight gain and slow growth, and abdominal distention.
    - While this disease is usually identified when the patient is a newborn, some individuals may not be diagnosed with this condition until later in childhood or, sometimes, not even until adulthood.
    - If symptoms suggest possible Hirschsprung Disease, consider abdominal x-ray and referral to gastroenterologist.
      - Based upon the pt's genes, this pt is <u>not</u> at increased risk of radiation-induced breast CA.

- MTHFR Deficiency: Carrier: This patient contains a single mutation known as Ala222Val in the MTHFR gene. The pt is not likely affected by this condition.
  - While being a carrier of MTHFR deficiency is usually not associated with increased homocysteine levels, a single mutation in the MTHFR gene can decrease the activity of the gene by as much as 35-40%.
    - Consider checking homocysteine levels at least periodically in-order to confirm no effect from this mutation.
  - If homocysteinemia is detected, it may be successfully treated with L-Methylfolate.

- Consider daily L-Methylfolate supplementation.
  - L-Methylfolate supplementation may be beneficial even if homocysteine levels aren't elevated because some preliminary studies have also found that MTHFR deficiency may also increase the risk of other diseases, such as depression.

- If the pt plans to have children in the future, counsel pt that prenatal vitamins specifically containing L-Methylfolate may be important for the mother of the pt's child to take even before the pt is aware she is pregnant. This is because the pt's child will have at a minimum a 50% chance of inheriting at least one MTHFR mutation and if the mother of the child also has one or more MTHFR mutations then the risk of the child inheriting one or more MTHFR mutations will be even higher. MTHFR mutations in the developing fetus may predispose the fetus to spina bifida. If the mother is taking L-Methylfolate supplementation during the first few weeks of pregnancy, however, this may counteract the potentially harmful effects of the MTHFR mutations and may significantly protect the fetus against spina bifida.

Section 3
Medications (Pharmacogenomics)

Please note that the following information is based solely on an analysis of the pt's genes and does not take into account non-genetic factors that may also significantly affect medication pharmacodynamics, pharmacokinetics, effectiveness, ADR risk, and dosing such as the pt's age, BMI, lifestyle, habits, and medical conditions as well as any supplements and medications the pt may be taking. These non-genetic factors may be important to take into consideration in-addition to the genetic analysis presented below.

Please also note that the information in this report is based upon current research pertaining to pharmacogenomics and that this may change overtime as additional genetic research is conducted. Reanalysis of the pt's genetic data throughout the pt's life may therefore be beneficial in making sure the genetic analysis is always up-to-date.

Cardiovascular medications:

- Clopidogrel (Plavix®) Effectiveness: Significantly Impaired Clopidogrel Effectiveness Detected.
  - The patient is a poor metabolizer (poor activator) of clopidogrel and is likely to not effectively convert clopidogrel to its active metabolite.
  - Consider starting clopidogrel at 600mg (loading dose) and continue with 150mg daily (maintenance dose).
  - Consider using platelet aggregation assay (or other platelet function assessment) to monitor the effect of the clopidogrel.
  - Avoid using clopidogrel with omeprazole, a CYP2C19 inhibitor.
  - Due to clopidogrel's potential ineffectiveness, it may be appropriate to consider starting the patient on a different drug.

- Clopidogrel (Plavix®) Effectiveness: Impaired Clopidogrel Effectiveness Detected.
  - The patient is an intermediate metabolizer (intermediate activator) and may not effectively convert clopidogrel to its active metabolite.
  - Consider starting at a higher dose of clopidogrel (loading dose).
  - Consider using platelet aggregation assay (or other platelet function assessment) to monitor the effect of the clopidogrel.
  - Avoid using clopidogrel with omeprazole, a CYP2C19 inhibitor.

Other medications:

- Mercaptopurine and Azathioprine ADR: Possible Increased Risk Detected due to a mutation in the ITPA gene. Patient may be at increased risk of adverse reactions to Mercaptopurine and Azathioprine medications due to a single mutation in the ITPA gene that is known to cause partial ITPase deficiency.
  - Partial ITPase deficiency is not always associated with Mercaptopurine and Azathioprine ADR. If the ITPase deficiency is significant enough then the pt may be at increased risk of this ADR.
    - If these medications are required, consider starting patient on a lower dose and carefully monitoring patient for signs of toxicity.

- 5-fluorouracil Toxicity: Increased Risk Detected. This patient has an increased risk of toxicity with 5-FU due to a mutation in both of the pt's DPYD genes.
  - Increased risk of leukopenia, thrombocytopenia, medium to severe nausea and vomiting, diarrhea, stomatitis, hair loss, and neurological sequelae including cerebellar ataxia and possible coma.

- If this medication is needed, consider an alternative chemotherapeutic agent or starting with a lower dose.

- Phenytoin: Slightly Decreased Metabolism Detected. Due to mutation in the CYP2C9 gene referred to as I359L, if phenytoin is ever needed then the pt may need a slightly lower dose (approx. mean daily dose of 309mg).

- Nitrous Oxide ADR: Increased risk not detected but patient is a Carrier. Due to a single mutation in the patient's MTHFR gene known as Ala222Val, the patient may have elevated homocysteine levels and be at increased risk of adverse reactions to nitrous oxide.
  - While being a carrier of is usually not associated with increased risk of nitrous oxide ADR, a single mutation in the MTHFR gene can decrease the activity of the gene by as much as 35-40%.
    - Consider checking homocysteine levels at least periodically in-order to confirm no effect from this mutation. If elevated, then that may increase the risk of ADR with NO.
      - Nitrous oxide usually leads to increases in homocysteine levels. If hyperhomocysteinemia already exists when NO is administered, this further increase in homocysteine levels may significantly increase risk of thromboembolic events.
      - If homocysteine levels are WNL then this pt may not have an increased ADR risk w/ NO.
  - This condition is *recessive*: If both parents are carriers of a mutation in this same gene then each child will have a 25% chance of being affected by this condition but if only the patient is a carrier then each child will have close to a 0% (zero percent) chance of being affected by this condition and ~50% chance of being a carrier like the pt.
    - Newborns and young children affected by this condition may be at highest risk of severe adverse reactions to nitrous oxide. Therefore, if the person this pt is going to have a child with is also a carrier of a MTHFR mutation, consider genetic testing for all of their children as newborns in-order to assess whether each child is affected by this condition. If so, consider avoidance of NO for that child.

Section 4
Screen for Rare Diseases, Conditions, and Traits

For additional information about this screen, including the full list of over 1,200 rare diseases, conditions, and traits that were screened for, please visit: https://sequencing.com/carrier-screening-rare-diseases-syndromes-and-traits.

Important note: the pt may still be a carrier of or even affected by a disease, condition, or trait even if it is not detected in this screen if the pt has a mutation that was not tested by the genetic test, not analyzed as part of this screen or if the pt has a yet-to-be documented mutation

Based on Sequencing.com's rare disease screen for more than 1,200 rare diseases, conditions, and traits the following was detected:

- Cystic Fibrosis: Carrier. This patient is a carrier of, but is not affected by, this disease due to a single mutation in the CFTR gene known as Deletion at coordinate 117149181. CF is a serious condition that affects the lungs, pancreas, liver, and intestines and usually causes early mortality, although improved treatments are now allowing a person with CF to live much longer than before.
    - This disease is *recessive*: If both parents are carriers of a mutation in this same gene then each child may have a 25% chance of being affected by this disease but if only the patient is a carrier then each child will have close to a 0% (zero percent) chance of being affected by this disease and ~50% chance of being a carrier like the pt.

- Adenosine Monophosphate Deaminase Deficiency: Carrier. This patient is a carrier of, but is not affected by, this disorder due to a single mutation in the AMPD1 gene known as K287I. This condition can cause fatigue, muscle pain and muscle cramping.
    - This disorder is *recessive*: If both parents are carriers of a mutation in this same gene then each child will have a 25% chance of being affected by this condition but if only the pt is a carrier then each child will have close to a 0% (zero percent) chance of being affected by this disorder and ~50% chance of being a carrier like the pt.

Section 5
No Increased Risk Detected

Please note that the following information is based solely on an analysis of the pt's genes and does not take into account non-genetic factors that may also significantly affect the pt's risk of a disease, such as the pt's age, BMI, lifestyle, habits, and medical conditions as well as any supplements and medications the pt may be taking. These non-genetic factors may be important to take into consideration in-addition to the genetic analysis presented below.

Please also note that the information in this report is based upon current research pertaining to the genetic basis of disease and that this may change overtime as additional genetic research is conducted. Reanalysis of the pt's genetic data throughout the pt's life may therefore be beneficial in making sure the pt's genetic analysis is always up-to-date.

Based on this comprehensive genetic screen, no increased risk has been detected for the following diseases, conditions, and traits:

- ✓ Sudden Cardiac Death (SCD): Increased risk not detected. Patient not likely to be affected by SCD conditions, including Hypertrophic Cardiomyopathy (HCM), Dilated Cardiomyopathy (DCM), Restrictive Cardiomyopathy (RCM), Long QT Syndrome (LQTS), Arrhythmogenic Right Ventricular Dysplasia (ARVD), Brugada Syndrome, and Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT).
- ✓ Malignant Hyperthermia: Increased risk not detected.
- ✓ Breast Cancer: Increased risk not detected.
- ✓ Breast Cancer Risk with Radiation Exposure: Increased risk not detected.
- ✓ Adrenocortical Carcinoma: Increased risk not detected.
- ✓ Multiple Sclerosis Risk due to Low Vitamin D Levels: Increased risk not detected.
- ✓ Lactose Intolerance: Increased risk not detected.
- ✓ Clopidogrel (Plavix®) Effectiveness: Ineffectiveness not detected. Plavix likely to be effective as an antiplatelet agent.
- ✓ Aspirin Effectiveness: Ineffectiveness not detected. Aspirin is likely to be effective in helping to prevent heart attacks and strokes.
- ✓ Statin ADR: Increased adverse reaction risk not detected. Patient is at low (3%) risk of statin-induced myopathy.
    - Genetic studies have found that simvastatin (Zocor®) is the primary statin associated with this specific ADR risk assessment. This genetic test may not be predictive of ADR risk with other statins, including atorvastatin and pravastatin.
- ✓ Statin Dosing: Routine dosing likely optimal. Unlike some patients, this patient is not likely to benefit from high dose atorvastatin compared to a regular statin dose.
- ✓ Postanesthetic Apnea: Increased risk not detected.
- ✓ Sulfonylureas ADR: Increased risk not detected.
- ✓ Abacavir ADR: Increased risk not detected.

Section 6
Analysis Alerts

This section contains important information about to the analysis of the pt's genetic data. The alerts below are provided so that you are aware of any limitations that were identified during the analysis of the data. Additional information about alerts: https://sequencing.com/knowledge-center/app-alerts

Ambiguity in genetic data

✓ **No ambiguity exists in the genetic data.**

Incomplete genetic data

While the genetic data file contained a lot of useful data, it did not contain enough data for a complete analysis of some diseases, conditions or traits.

As a generic example, this app may attempt to analyze 20 different genetic variants in-order to provide a risk assessment for condition X. While many genetic data files do contain data for all 20 variants, some genetic data files only contain data for 18 out of the 20. Some files may even only provide data on just two or three variants out of the 20.

The analysis conducted by this app will use whatever data is available and will also provide this alert if data isn't available for 100% of the variants that the app is configured to analyze.

The results may be different if more data was available. The genetic data was incomplete for the diseases, conditions and traits listed below. If not listed below then the data for that disease, condition or trait was complete.
Note: The Rare Disease assessment (Section 4) is not included in the table below. This data can be obtained from the report generated by the Rare Disease Screen app accessible at Sequencing.com.

Disease, Condition, Trait or Medication *Analysis used incomplete data*	**Available Data** *used for analysis*	**Pt's Analysis** *available variants*	**Required for Full Analysis** *total known variants*
Increased Risk Of Breast And/or Ovarian Cancer	99.9%	6199	6203
Increased Risk Of Breast Cancer And/or Ovarian Cancer With Radiation Exposure	99.9%	6073	6077
Variant Of Unknown Significance (possibly Associated With Breast And/or Ovarian Cancer)	99.9%	5167	5171
Likely Harmless Variant (previously Associated With Variant Of Unknown Significance)	99.9%	778	779
Rare Conditions	99%	5753	5783
Likely Harmless Variant (previously Associated With Breast And/or Ovarian Cancer)	99%	2895	2926
Hypertrophic Cardiomyopathy	99%	88	89
Warfarin (Coumadin®) Dosing	98%	52	53
Niemann-Pick Disease, Type C1	97%	170	175

Disease, Condition, Trait or Medication *Analysis used incomplete data*	Available Data *used for analysis*	Pt's Analysis *available variants*	Required for Full Analysis *total known variants*
Best Macular Dystrophy	97%	84	87
Central Core Disease	94%	16	17
Tay-Sachs Disease	90%	9	10
Gaze Palsy, Horizontal, With Progressive Scoliosis	90%	9	10
Prostate Cancer	88%	7	8
Coronary Heart Disease	83%	5	6
Leber Congenital Amaurosis, Type VI	80%	4	5
Dyskeratosis Congenita	80%	24	30
Noise-induced Hearing Loss	75%	3	4
Hartnup Disorder	50%	1	2
Complement C3 Deficiency	50%	1	2
Vitelliform Macular Dystrophy	33%	1	3

Lack of genetic data (analysis not possible)

There was no genetic data available, and no genetic analysis could occur, for the following diseases, conditions, traits or medications. Because of this, genetic analysis was possible only for the diseases, conditions and traits that appear in Sections 1-5 of this report.

This table differs from the 'Incomplete genetic data' table above because analysis was still performed for everything listed above. This was possible because for those diseases, conditions, traits or medications there was at least *some* genetic data available.

The table below, however, identifies diseases, conditions, traits or medications that were *not* analyzed because there was no genetic data available. While the diseases, conditions and traits listed below are normally analyzed as part of this app, they could not be analyzed for this pt because the pt's genetic data file did not contain *any* genetic data for these diseases, conditions or traits.

In order to obtain analysis of the diseases, conditions, traits and medications listed below, the pt may need to have additional genetic testing that provides a greater amount of genetic data, such as Sequencing.com's Ultimate Genome Sequencing service, which includes 30x whole genome sequencing.

Disease, Condition, Trait or Medication *Analysis not possible*	Available Data	Pt's Analysis *available variants*	Required for Full Analysis *total known variants*
Lethal Congenital Contracture Syndrome	0%	0	1
Zinsser-Cole-Engman Syndrome	0%	0	1
Peters Anomaly	0%	0	1
Porphyria, Acute Intermittent, Nonerythroid Variant	0%	0	2

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Disease, Condition, Trait or Medication ***Analysis not possible***	**Available Data**	**Pt's Analysis** ***available variants***	**Required for Full Analysis** ***total known variants***
Colorblindness, Deutan	0%	0	1
Unna-Thost Disease	0%	0	1
Defective Spermatogenesis	0%	0	1