PATHWAY GENOMICS®

CARRIER STATUS DNA INSIGHT®

Protected Health Information

PERSONAL DETAILS	
PATIENT ID	SAMPLE PATIENT
DOB	Jan 1, 19XX
GENDER	M
ETHNICITY	Asian

ORDERING HEALTHCARE PROFESSIONAL

Glenn Braunstein M.D.
4755 Nexus Center Drive
San Diego, CA 92121 US

LABORATORY INFO	
ACCESSION NUMBER	XXXXXXXX
ACTIVATION CODE	XXXXXXXX
SPECIMEN TYPE	BUCCAL SWAB
COLLECTED DATE	Feb 2, 2017
RECEIVED DATE	Feb 13, 2017
REPORT DATE	Feb 27, 2017

Test Results Reviewed & Approved by:
Laboratory Director,
Nilesh Dharajiya, M.D.

Summary of Results

Propionic Acidemia

Risk to Child: Any child of this patient has a 50% chance of inheriting the patient's mutation associated with this disease and being a carrier. If the patient's partner also carries a mutation for this disease, there is a 25% chance that each child of the patient will inherit both parents' mutations and may develop the disease.

Risk to Patient: This patient is a carrier of a genetic mutation for this disease but is not likely to be affected. Since there are many rare mutations, it is possible to carry an untested mutation in addition to the one found in the patient's DNA.

Recommendation: Genetic counseling is recommended for the patient and his or her partner to discuss the potential clinical and/or reproductive implications of this result and to discuss genetic testing of the patient's partner and close relatives.

Result:
Carrier, Heterozygote

Mutations:
PCCB [c.1228C>T (p.R410W)]

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PATIENT IS NOT A CARRIER FOR THE FOLLOWING:			
21-Hydroxylase-deficient congenital adrenal hyperplasia	Costeff optic atrophy syndrome	Hereditary fructose intolerance	Pendred syndrome
3-Methylcrotonyl-CoA carboxylase deficiency	Crigler-Najjar syndrome	Herlitz junctional epidermolysis bullosa, LAMA3-related	Phenylketonuria
Achromatopsia	Cystic fibrosis	Herlitz junctional epidermolysis bullosa, LAMB3-related	Polycystic kidney disease
Acrodermatitis enteropathica	Cystinosis	Herlitz junctional epidermolysis bullosa, LAMC2-related	Pompe disease
Alkaptonuria	Diabetes, permanent neonatal	HMG-CoA lyase deficiency	Prekallikrein deficiency
Alpha-1 antitrypsin deficiency	Dihydropyrimidine dehydrogenase deficiency	Homocystinuria, cblE type	Primary hyperoxaluria, type 1
Alpha-mannosidosis	Dubin-Johnson syndrome	Homocystinuria, classic	Primary hyperoxaluria, type 2
Amyotrophic lateral sclerosis	Ehlers-Danlos syndrome, dermatosparaxis	Hurler syndrome	Primary hyperoxaluria, type 3
Andermann syndrome	Ehlers-Danlos syndrome, hypermobility	Hypophosphatasia, autosomal recessive	Prothrombin deficiency
Argininosuccinate lyase deficiency	Ehlers-Danlos syndrome, kyphoscoliotic	Inclusion body myopathy 2	Rh-null syndrome
ARSACS	Factor V Leiden thrombophilia	Juvenile retinoschisis, X-linked	Rhizomelic chondrodysplasia punctate type 1
Aspartylglucosaminuria	Factor XI deficiency	Krabbe disease	Rickets, pseudovitamin D-deficiency
Ataxia with vitamin E deficiency	Familial dysautonomia	Lipoamide dehydrogenase deficiency	Salla disease
Ataxia-telangiectasia	Familial Mediterranean fever	Lipoprotein lipase deficiency, familial	Sandhoff disease
Autoimmune polyglandular syndrome, type I	Fanconi anemia	Maple syrup urine disease	Short-chain acyl-CoA dehydrogenase deficiency
Bardet-Biedl syndrome, BBS1-related	Galactokinase deficiency	Medium-chain acyl-CoA dehydrogenase deficiency	Sick sinus syndrome
Bartter syndrome, type 4a	Galactosemia	Megalencephalic leukoencephalopathy with subcortical cysts	Sickle cell disease
Beta-ketothiolase deficiency	Gaucher disease	Metachromatic leukodystrophy	Smith-Lemli-Opitz syndrome
Beta-thalassemia	Glutaric acidemia, type 1	Methylmalonic acidemia	Spherocytosis, hereditary
Biotinidase deficiency	Glycogen storage disease, type 1a	Mucolipidosis II	Tay-Sachs disease
Bloom syndrome	Glycogen storage disease, type Ib	Mucolipidosis III	Tay-Sachs pseudodeficiency
Canavan disease	Glycogen storage disease, type III	Mucolipidosis IV	Thrombocytopenia, congenital amegakaryocytic

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TAY-SACHS DISEASE

(p.Y427IfsX5, 1278insTATC), c.574G>C (p.V192L), c.346+1G>C (IVS2+1G>C)]

TAY-SACHS PSEUDODEFICIENCY

Not a Carrier of: HEXA [c.739C>T (p.R247W), c.745C>T (p.R249W)]

THROMBOCYTOPENIA, CONGENITAL AMEGAKARYOCYTIC

Not a Carrier of: MPL [c.305G>C (p.R102P), c.127C>T (p.R43X)]

TYROSINE HYDROXYLASE DEFICIENCY

Not a Carrier of: TH [c.698G>A (p.R233H), c.707T>C (p.L236P)]

TYROSINEMIA

Not a Carrier of: FAH [c.192G>T (p.Q64H), c.554-1G>T, c.607-6T>G, c.782C>T (p.P261L), c.786G>A (p.W262X), c.1009G>A (p.G337S), c.1062+5G>A]

USHER SYNDROME, TYPE 1F

Not a Carrier of: PCDH15 [c.733C>T (p.R245X)]

VERY LONG-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY

Not a Carrier of: ACADVL [c.848T>C (p.V283A)]

VON WILLEBRAND DISEASE, TYPE 2 NORMANDY

Not a Carrier of: VWF [c.2311A>G (p.M771V), c.2561G>A (p.R854Q), c.2451T>A (p.H817Q), c.2287A>G (p.R763G), c.2344C>T (p.R782W), c.2354G>A (p.G785E), c.2359G>A (p.E787K), c.2362T>C (p.C788R), c.2363G>A (p.C788Y), c.2372C>T (p.T791M), c.2384A>G (p.Y795C), c.2635G>A (p.D879N), c.3159G>T (p.Q1053H), c.3178T>C (p.C1060R),

VON WILLEBRAND DISEASE, TYPE 2 NORMANDY

c.2411G>T (p.C804F), c.2435C>T (p.P812L), c.2447G>A (p.R816Q), c.2446C>T (p.R816W), c.3232G>A (p.E1078K), c.3673T>G (p.C1225G)]

VON WILLEBRAND DISEASE, TYPE 3

Not a Carrier of: VWF [c.3940delG (p.V1314SfsX34), c.1384delG (p.A462QfsX15), c.3258_3259insT (p.D1087X), c.3736_3737dupCC (p.P1247LfsX7), c.4324_4331dupAGTGTGGA (p.D1444EfsX84), c.7172_7173insT (p.E2391DfsX3), c.1693C>T (p.Q565X), c.3800T>A (p.L1267X), c.2016_2019del (p.S673TfsX67), c.2269_2270del (p.L757VfsX22), c.3943C>T (p.R1315C), c.4036C>T (p.Q1346X), c.4092_4093del (p.L1365VfsX11), c.4368C>A (p.Y1456X), c.5053+1G>A (IVS28+1G>A), c.5170+10C>T (IVS29+10C>T), c.5557C>T (p.R1853X), c.6182delT (p.F2061SfsX38), c.6520T>G (p.C2174G), c.6977-1G>C (IVS40-1G>C), c.7085G>T (p.C2362F), c.7603C>T (p.R2535X), c.7630C>T (p.Q2544X), c.7683delT (p.Q2562SfsX2), c.7729+7C>T (IVS45+7C>T), c.8012G>A (p.C2671Y), c.8155+3G>T (IVS50+3G>T), c.8216G>A (p.C2739Y), c.8262T>G (p.C2754W), c.139G>C (p.D47H), c.276delT (p.F92LfsX11), c.817C>T (p.R273W), c.970C>T (p.R324X), c.1071C>A (p.Y357X), c.1093C>T (p.R365X), c.1110-1G>A (IVS9-1G>A), c.1830C>A (p.Y610X), c.1858G>T (p.E620X), c.191delG (p.G64AfsX19), c.212C>A (p.S71X), c.652C>T (p.Q218X), c.666G>A (p.W222X), c.1117C>T (p.R373X), c.1131G>T (p.W377C), c.2157delA (p.D720TfsX21), c.7300C>T (p.R2434X), c.374_387del (p.G125VfsX3), c.874+1G>A (IVS7+1G>A), c.893dupG (p.M299YfsX4), c.1657dupT (p.W553LfsX97), c.3212G>T (p.C1071F), c.4626C>G (p.Y1542X), c.7139dupT (p.L2380FfsX11), c.7674dupC (p.S2559LfsX8), c.8411G>A (p.C2804Y)]

WILSON DISEASE

Not a Carrier of: ATP7B [c.2333G>T (p.R778L), c.3207C>A (p.H1069Q)]

ZELLWEGER SYNDROME SPECTRUM, PEX1-RELATED

Not a Carrier of: PEX1 [c.2097dupT (p.I700YfsX42), c.2528G>A (p.G843D)]

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RESIDUAL RISK AFTER NEGATIVE TEST RESULTS

In the case of a negative test result (not a carrier), there is a residual risk that the patient may have a mutation that is not part of the test panel. Included in the table below are the residual risk estimates for the carrier conditions in the Pathway Genomics carrier status test. Population carrier rate, carrier detection rate and residual risk are shown for conditions and specific populations for which the data is known. For other conditions listed below and populations that are not shown, the prevalence is rare, the mutation detection rate is unknown and residual risk is not calculable.

For individuals with a “NOT A CARRIER” result for a condition for which there is suggestive personal and/or family history, additional genetic testing may be indicated.

For questions regarding the interpretation of residual risk information, please contact Pathway Genomics' genetic counseling department at (877) 505-7374 or counselors@pathway.com.

21-HYDROXYLASE-DEFICIENT CONGENITAL ADRENAL HYPERPLASIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Yupik Eskimos	1:9	100.0%	Negligible
General	1:60	69.0%	1:191

3-METHYLCROTONYL-COA CARBOXYLASE DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
German and Turkish	1:146	4.0%	1:151

ACHROMATOPSIA

CNGB3

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Pingelapese	1:3	100.0%	Negligible
European	1:91	91.0%	1:1001

CNGA3

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
European	1:181	42.0%	1:311

ACRODERMATITIS ENTEROPATHICA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Tunisian	1:500	78.0%	1:2269

ALKAPTONURIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Czech, Slovak	1:90	50.0%	1:179
European (non-Slovak or Czech)	1:250	11.0%	1:281

ALPHA-1 ANTITRYPSIN DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Southern European	1:7	95.0%	1:121
North American	1:12	95.0%	1:221
African	1:14	95.0%	1:261
Northern European	1:15	95.0%	1:281
Middle East and North African	1:16	95.0%	1:301
Southeast Asian	1:84	95.0%	1:1661
Far East Asian	1:570	95.0%	1:11381

ALPHA-MANNOSIDOSIS

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Pan-ethnic	1:354	35.0%	1:544

AMYOTROPHIC LATERAL SCLEROSIS

DATA NOT AVAILABLE

There is insufficient published data to determine the carrier rate, detection rate, and residual risk for this condition.

ANDERMANN SYNDROME

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
French-Canadian	1:23	100.0%	Negligible

ARGININOSUCCINATE LYASE DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Pan-ethnic	1:194	50.0%	1:387

ARSACS

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
French-Canadian	1:21	96.0%	1:501

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TEST METHODOLOGY

Genotyping by PCR-based enrichment and next-generation sequencing.

DISCLAIMER

This test was developed and its performance characteristics determined by Pathway Genomics Corporation. It has not been cleared or approved by the FDA. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research.

If you have any questions about this report or wish to speak with one of Pathway Genomics' genetic counselors, please call (877) 505.7374.

RISKS AND LIMITATIONS

Risk of Laboratory Technical Problems or Laboratory Error

The certified testing laboratory has standard and effective procedures in place to protect against technical and operational problems. However, such problems may still occur. The testing laboratory receives samples collected by patients and physicians. Problems in shipping to the laboratory or sample handling can occur, including but not limited to damage to the specimen or related paperwork, mislabeling, and loss or delay of receipt of the specimen. Laboratory problems can occur that might lead to inability to obtain results. Examples include, but are not limited to, sample mislabeling, DNA contamination, un-interpretable results, and human and/or testing system errors. In such cases, the testing laboratory may need to request a new sample. However, upon re-testing, results may still not be obtainable.

As with all medical laboratory testing, there is a small chance that the laboratory could report inaccurate information. For example, the laboratory could report that a given genotype is present when in fact it is not. Any kind of laboratory error may lead to incorrect decisions regarding medical treatment and/or diet and fitness recommendations. If a laboratory error has occurred or is suspected, a health care professional may wish to pursue further evaluation and/or other testing. Further testing may be pursued to verify any results for any reason.

Limitations

The purpose of this test is to provide information about how a tested individual's genes may affect carrier status for some inherited diseases, responses to some drugs, risk for specific common health conditions, and/or selected diet, nutrition and/or exercise responses, as well as to learn more about the tested individual's ancient ancestry, depending upon the specific genetic testing that is ordered by the health care professional. Tested individuals should not make any changes to any medical care (including but not limited to changes to dosage or frequency of medications, diet and exercise regimens, or pregnancy planning) based on genetic testing results without consulting a health care professional.

The science behind the significance or interpretation of certain testing results continues to evolve. Although great strides have been made to advance the potential usefulness of genetic testing, there is still much to be discovered. Genetic testing is based upon information, developments and testing techniques that are known today. Future research may reveal changes in the interpretation of previously obtained genetic testing results. For example, any genetic test is limited by the variants being tested. The interpretation of the significance of some variants may change as more research is done about them. Some variants that are associated with disease, drug response, or diet, nutrition and exercise response may not be tested; possibly these variants have not yet been identified in genetic studies.

Many of the conditions and drug responses that are tested are dependent on genetic factors as well as nongenetic factors such as age, personal health and family health history, diet, and ethnicity. As such, an individual may not exhibit the specific drug response, disease, or diet, nutrition and exercise response consistent with the genetic test results.

Another limitation for some conditions, particularly in the areas of diet and exercise, is that genetic associations have been studied and observed in Caucasian populations only, and in some cases only in one gender. In this case, the interpretations and recommendations are made in the context of Caucasian studies, but the results may or may not be relevant to tested individuals who are of non-Caucasian or mixed ethnicities or the non-studied gender. If patient ethnicity is not disclosed in the test requisition form the ethnicity field in the report will read as "Ethnicity: Not Reported". Such reports will be defaulted to phenotype list displayed for Caucasian ethnicity.

Based on test results and other medical knowledge of the tested individual, health care professionals might consider additional independent testing, or consult another health care professional or genetic counselor.

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RESULT STATUS DEFINITIONS

Amended 	Test results and/or patient information that have been revised in a way that does not impact the clinical significance of the result(s) and/or patient diagnosis, treatment or management.
Corrected 	Test results and/or patient information that have been revised in a way that may impact the clinical significance of the result(s) and/or patient diagnosis, treatment or management.
Final 	Test results that are available at the time of report issue or have been revised from pending status to final status.
Pending 	Test results that are not available at the time of report issue. All pending results will be specified in the report.

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