PATHWAY GENOMICS®

CARRIER STATUS DNA INSIGHT®

Protected Health Information

PATIENT ID	SAMPLE PATIENT
GENDER	M
ACCESSION #	99999999
REPORT DATE	Feb 27, 2017

PENDRED SYNDROME

Not a Carrier of: SLC26A4 [c.1246A>C (p.T416P), c.707T>C (p.L236P), c.1001+1G>A, c.1151A>G (p.E384G), c.716T>A (p.V239D), c.919-2A>G, c.2168A>G (p.H723R), c.1540C>A (p.Q514K)]

PHENYLKETONURIA

Not a Carrier of: PAH [c.143T>C (p.L48S), c.473G>A (p.R158Q), c.727C>T (p.R243X), c.782G>A (p.R261Q), c.842C>T (p.P281L), c.1066-11G>A (IVS10-11G>A), c.1208C>T (p.A403V), c.1222C>T (p.R408W), c.1223G>A (p.R408Q), c.1241A>G (p.Y414C), c.728G>A (p.R243Q), c.838G>A (p.E280K), c.442-1G>A (IVS4-1G>A), c.611A>G (IVS6-96A>G), c.1068C>A (p.Y356X)/c.1068C>G (p.Y356X), c.117C>G (p.F39L), c.194T>C (p.I65T), c.734T>C (p.V245A), c.331C>T (p.R111X), c.721C>T (p.R241C), c.1238G>C (p.R413P)]

POLYCYSTIC KIDNEY DISEASE

Not a Carrier of: PKHD1 [c.8829dupC (p.I2944HfsX6), c.107C>T (p.T36M), c.1486C>T (p.R496X), c.10412T>G (p.V3471G), c.10444C>T (p.R3482C), c.2414C>T (p.P805L), c.9530T>C (p.I3177T), c.10174C>T (p.Q3392X), c.664A>G (p.I222V), c.9689delA (p.D3230VfsX34), c.8870T>C (p.I2957T)]

POMPE DISEASE

Not a Carrier of: GAA [c.2741delinsCAG (p.Q914PfsX30), c.1935C>A (p.D645E), c.2560C>T (p.R854X), c.525delT (p.E176RfsX45), c.925G>A (p.G309R)]

PREKALLIKREIN DEFICIENCY

Not a Carrier of: KLKB1 [c.1205G>A (p.W402X), c.1643G>A (p.C548Y)]

PRIMARY HYPEROXALURIA, TYPE 1

Not a Carrier of: AGXT [c.508G>A (p.G170R), c.33dupC (p.K12QfsX156)]

PRIMARY HYPEROXALURIA, TYPE 2

Not a Carrier of: GRHPR [c.103delG (p.D35TfsX11), c.403_404+2del]

PRIMARY HYPEROXALURIA, TYPE 3

Not a Carrier of: HOGA1 [c.700+5G>T, c.944_946del (p.E315del)]

PROTHROMBIN DEFICIENCY

Not a Carrier of: F2 [c.481G>T (p.D161Y, D118Y), c.787C>T (p.R263C, R220C), c.124C>T (p.R42W, R-2W), c.542G>A (p.C181Y, C138Y), c.940C>T (p.R314C, R271C), c.1054G>A (p.E352K, E309K), c.1499G>A (p.R500Q, R457Q), c.1741C>T (p.R581C, R538C)]

RH-NULL SYNDROME

Not a Carrier of: RHAG [c.808G>A (p.V270I)]

RHIZOMELIC CHONDRODYSPLASIA PUNCTATE TYPE 1

Not a Carrier of: PEX7 [c.875T>A (p.L292X), c.653C>T (p.A218V), c.649G>A (p.G217R)]

RICKETS, PSEUDOVITAMIN D-DEFICIENCY

Not a Carrier of: CYP27B1 [c.1166G>A (p.R389H), c.262delG (p.V88WfsX71, 958delG), c.589+1G>A (IVS3+1G>A), c.1319_1325dupCCCACCC (p.F443PfsX24, 3398dupCCCACCC)]

SALLA DISEASE

Not a Carrier of: SLC17A5 [c.115C>T (p.R39C)]

SANDHOFF DISEASE

Not a Carrier of: HEXB [c.76delA (p.M26CfsX5), c.445+1G>A (IVS2+1G>A)]

SHORT-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY

Not a Carrier of: ACADS [c.1170C>G (p.I390M), c.1138C>T (p.R380W), c.1058C>T (p.S353L), c.529T>C (p.W177R), c.319C>T (p.R107C), c.136C>T (p.R46W), c.417G>C (p.W139C), c.1095G>T (p.Q365H), c.1108A>G (p.M370V), c.596C>T (p.A199V), c.505A>C (p.T169P)]

SICK SINUS SYNDROME

Not a Carrier of: SCN5A [c.3893C>T (p.P1298L), c.659C>T (p.T220I), c.4222G>A (p.G1408R), c.4895G>A (p.R1632H)]

SICKLE CELL DISEASE

Not a Carrier of: HBB [c.20A>T (p.E7V, Hemoglobin S)]

SMITH-LEMLI-OPITZ SYNDROME

Not a Carrier of: DHCR7 [c.452G>A (p.W151X), c.1210C>T (p.R404C), c.278C>T (p.T93M), c.506C>T (p.S169L), c.724C>T (p.R242C), c.725G>A (p.R242H), c.906C>G (p.F302L), c.976G>T (p.V326L), c.1054C>T (p.R352W), c.1228G>A (p.G410S), c.1342G>A (p.E448K), c.832-1G>C (IVS8-1G>C)]

SPHEROCYTOSIS, HEREDITARY

Not a Carrier of: ANK1 [c.444+16C>T (5703+16C>T), c.1387G>A (p.V463I)]; **EPB42** [c.357G>A (p.W119X), c.424G>A (p.A142T), c.929G>A (p.R310Q), c.523G>T (p.D175Y), c.922+1G>A (IVS6+1G>A), c.949C>T (p.R317C)]

TAY-SACHS DISEASE

Not a Carrier of: HEXA [c.613delC (p.L205WfsX2), c.986G>A (p.W329X), c.1003A>T (p.I335F), c.1373G>A (p.C458Y), c.1074-1G>T (IVS9-1G>T), c.533G>A(p.R178H)/c.533G>T(p.R178L), c.1510C>T (p.R504C), c.1073+1G>A (IVS9+1G>A), c.805G>A (p.G269S), c.1496G>A (p.R499H), c.509G>A (p.R170Q), c.915_917del (p.F305del, deltaTTC910-912), c.629C>T (p.S210F), c.508C>T (p.R170W), c.1421+1G>C (IVS12+1G>C), c.571-1G>T (IVS5-1G>T), c.1274_1277dupTATC

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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)]

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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ASPARTYLGLUCOSAMINURIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Finnish	1:68	98.0%	1:3351

ATAXIA WITH VITAMIN E DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Mediterranean, North African	1:274	80.0%	1:1366

ATAXIA-TELANGIECTASIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Amish	1:100	100.0%	Negligible
Costa Rican	1:100	86.0%	1:708
North African Jewish	1:100	100.0%	Negligible
Norwegian	1:100	55.0%	1:221
Polish	1:100	39.0%	1:163
Sardinian	1:100	95.0%	1:1981
Turkish	1:100	33.0%	1:149

AUTOIMMUNE POLYGLANDULAR SYNDROME, TYPE I

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Iranian Jewish	1:48	100.0%	Negligible
Finnish	1:80	71.0%	1:273
Slovenian	1:104	67.0%	1:313
Norwegian	1:150	48.0%	1:288
Polish	1:250	71.0%	1:860

BARDET-BIEDL SYNDROME, BBS1-RELATED

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
North American, European	1:387	79.0%	1:1839

BARTTER SYNDROME, TYPE 4A

DATA NOT AVAILABLE

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

BETA-KETOTHIOLASE DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Vietnamese	1:500	88.0%	1:4159

BETA-THALASSEMIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Mediterranean	1:7	91.0%	1:68
Thai	1:11	91.0%	1:112
West African	1:11	75.0%	1:41
Middle Eastern	1:49	91.0%	1:534
Chinese	1:100	91.0%	1:1101

BIOTINIDASE DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Pan-ethnic	1:120	89.0%	1:1083

BLOOM SYNDROME

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:107	99.0%	1:10601

CANAVAN DISEASE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:41	97.0%	1:1540

CARNITINE DEFICIENCY, PRIMARY SYSTEMIC

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Taiwanese	1:1000	35.0%	1:153

CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY

DATA NOT AVAILABLE

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

CARTILAGE-HAIR HYPOPLASIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Old Order Amish	1:10	100.0%	Negligible
Finnish	1:76	92.0%	1:939

CEREBROTENDINOUS XANTHOMATOSIS

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
North African Jewish	1:5	79.0%	1:20
Dutch	1:111	100.0%	Negligible

CHOROIDEREMIA

DATA NOT AVAILABLE

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

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CITRULLINEMIA, TYPE I

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Pan-ethnic	1:119	46.0%	1:220

COHEN SYNDROME

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Old Order Amish	1:11	99.0%	1:1001

COMBINED PITUITARY HORMONE DEFICIENCY, PROP1-RELATED

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Worldwide	1:63	55.0%	1:139

CONGENITAL DISORDER OF GLYCOSYLATION TYPE IA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Danish	1:60	88.0%	1:493

COSTEFF OPTIC ATROPHY SYNDROME

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Iraqi Jewish	1:10	100.0%	Negligible

CRIGLER-NAJJAR SYNDROME

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Caucasian	1:500	75.0%	1:1997

CYSTIC FIBROSIS

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:24	94.0%	1:384
Non-Hispanic Caucasian	1:25	88.0%	1:206
Hispanic Caucasian	1:58	72.0%	1:205
African American	1:61	64.0%	1:171
Asian American	1:94	49.0%	1:183

CYSTINOSIS

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
US general, Northern European	1:158	50.0%	1:315

DIABETES, PERMANENT NEONATAL

DATA NOT AVAILABLE

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

DIHYDROPYRIMIDINE DEHYDROGENASE DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Caucasian	1:56	52.0%	1:116

DUBIN-JOHNSON SYNDROME

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Iranian Jewish	1:18	100.0%	Negligible
Moroccan Jewish	1:18	100.0%	Negligible

EHLERS-DANLOS SYNDROME, DERMATOSPARAXIS

DATA NOT AVAILABLE

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

EHLERS-DANLOS SYNDROME, HYPERMOBILITY

DATA NOT AVAILABLE

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

EHLERS-DANLOS SYNDROME, KYPHOSCOLIOTIC

DATA NOT AVAILABLE

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

FACTOR V LEIDEN THROMBOPHILIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
European American	1:18	100.0%	0
Hispanic American	1:45	100.0%	0
Native American	1:80	100.0%	0
African American	1:83	100.0%	0
Asian American	1:222	100.0%	0

FACTOR XI DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:11	98.0%	1:501
U.K. Pan-ethnic	1:500	39.0%	1:819

FAMILIAL DYSAUTONOMIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:31	99.0%	1:3001

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FAMILIAL MEDITERRANEAN FEVER

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Armenian	1:3	79.0%	1:11
Ashkenazi Jewish	1:4	54.0%	1:8
Non-Ashkenazi Jewish	1:4	69.0%	1:11
Turkish	1:6	76.0%	1:22
Arab	1:7	53.0%	1:14

FANCONI ANEMIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:89	99.0%	1:8801

GALACTOKINASE DEFICIENCY

DATA NOT AVAILABLE

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

GALACTOSEMIA

Duarte Variant

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Northern European	1:9	100.0%	0
Western European	1:11	100.0%	0
Eastern European	1:12	100.0%	0
Southern European	1:18	100.0%	0
Asian	1:56	100.0%	0

Classic

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Northern European	1:111	80.0%	1:551
Southern European	1:234	80.0%	1:1166
Western European	1:270	80.0%	1:1346
African American	1:1010	80.0%	1:5046
Eastern European	1:1016	80.0%	1:5076

GAUCHER DISEASE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:18	90.0%	1:171
Pan-ethnic	1:50	64.0%	1:137

GLUTARIC ACIDEMIA, TYPE 1

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Caucasian	1:159	38.0%	1:256

GLYCOGEN STORAGE DISEASE, TYPE 1A

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:71	93.0%	1:1001
Non-Jewish	1:158	62.0%	1:414

GLYCOGEN STORAGE DISEASE, TYPE IB

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
European	1:354	47.0%	1:667
Japanese	1:354	50.0%	1:707

GLYCOGEN STORAGE DISEASE, TYPE III

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
General	1:158	20.0%	1:197

GLYCOGEN STORAGE DISEASE, TYPE V

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
US general	1:158	41.0%	1:267
Spanish	1:206	41.0%	1:348

GM1-GANGLIOSIDOSIS

DATA NOT AVAILABLE

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

HEARING LOSS, DFNB1 AND DFNB9 NONSYNDROMIC

DATA NOT AVAILABLE

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

HEARING LOSS, DFNB59 NONSYNDROMIC

DATA NOT AVAILABLE

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

HEMOCHROMATOSIS

HFE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Northern European	1:3	63.0%	1:6

HFE2

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
French-Canadian	1:500	100.0%	Negligible
Greek	1:500	70.0%	1:1664
Italian	1:500	4.0%	1:521

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HEMOGLOBIN C

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
African American	1:52	100.0%	0
Native American	1:489	100.0%	0
Hispanic American	1:1517	100.0%	0
Caucasian	1:2754	100.0%	0
Asian Indian	1:4768	100.0%	0
Filipino	1:4775	100.0%	0
Middle Eastern	1:5476	100.0%	0
Asian	1:6607	100.0%	0
Southeast Asian	1:14200	100.0%	0

HEMOGLOBIN D

HbD-Punjab

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
South Asian	1:232	100.0%	0

HEMOGLOBIN E

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Bangladeshi	1:24	100.0%	0
Chinese	1:221	100.0%	0
Pakistani	1:529	100.0%	0
Asian Indian	1:578	100.0%	0
White Irish	1:1961	100.0%	0
White British	1:9091	100.0%	0
African	1:10000	100.0%	0

HEMOGLOBIN O

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
South Asian	1:233	100.0%	0
General	1:1428	100.0%	0
African American	1:30000	100.0%	0

HEREDITARY FRUCTOSE INTOLERANCE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Middle Eastern	1:97	50.0%	1:193
US general	1:122	50.0%	1:243
African American	1:226	50.0%	1:451

HERLITZ JUNCTIONAL EPIDERMOLYSIS BULLOSA, LAMA3-RELATED

LAMA3, LAMB3, LAMC2

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
US general	1:781	45.0%	1:1419

HERLITZ JUNCTIONAL EPIDERMOLYSIS BULLOSA, LAMB3-RELATED

LAMA3, LAMB3, LAMC2

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
US general	1:781	45.0%	1:1419

HERLITZ JUNCTIONAL EPIDERMOLYSIS BULLOSA, LAMC2-RELATED

LAMA3, LAMB3, LAMC2

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
US general	1:781	45.0%	1:1419

HMG-COA LYASE DEFICIENCY

DATA NOT AVAILABLE

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

HOMOCYSTINURIA, CBLE TYPE

DATA NOT AVAILABLE

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

HOMOCYSTINURIA, CLASSIC

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
UK	1:500	50.0%	1:999
US general	1:500	26.0%	1:675

HURLER SYNDROME

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Caucasian	1:159	79.0%	1:753

HYPOPHOSPHATASIA, AUTOSOMAL RECESSIVE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Japanese	1:194	41.0%	1:328

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INCLUSION BODY MYOPATHY 2

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Middle Eastern Jewish	1:15	100.0%	Negligible
Japanese	Unknown	100.0%	Negligible

JUVENILE RETINOSCHISIS, X-LINKED

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Finnish	1:65	95.0%	1:1281

KRABBE DISEASE

DATA NOT AVAILABLE

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

LIPOAMIDE DEHYDROGENASE DEFICIENCY

DATA NOT AVAILABLE

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

LIPOPROTEIN LIPASE DEFICIENCY, FAMILIAL

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Caucasian	1:500	46.0%	1:925

MAPLE SYRUP URINE DISEASE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:97	99.0%	1:1921

MEDIUM-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Caucasian	1:40	70.0%	1:131

MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS

DATA NOT AVAILABLE

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

METACHROMATIC LEUKODYSTROPHY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Australian	1:100	46.0%	1:184
Polish	1:100	54.0%	1:216

METHYLMALONIC ACIDEMIA

MUT

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Japanese	1:187	22.0%	1:239
Black	1:237	35.0%	1:364
Caucasian	1:237	19.0%	1:292
Hispanic	1:237	41.0%	1:401

MMAA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Japanese	1:448	64.0%	1:1243
Caucasian	1:568	43.0%	1:996

MUCOLIPIDOSIS II

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Japanese	1:500	60.0%	1:1249
Predominantly white	1:500	56.0%	1:1135

MUCOLIPIDOSIS III

DATA NOT AVAILABLE

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

MUCOLIPIDOSIS IV

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:127	95.0%	1:2521

MULTIPLE CARBOXYLASE DEFICIENCY

DATA NOT AVAILABLE

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

NEPHROTIC SYNDROME, STEROID-RESISTANT

DATA NOT AVAILABLE

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

NEURONAL CEROID LIPOFUSCINOSIS, CLN3-RELATED

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Finnish	1:70	85.0%	1:461
West German	1:188	85.0%	1:1248

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NEURONAL CEROID LIPOFUSCINOSIS, CLN5-RELATED

DATA NOT AVAILABLE

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

NEURONAL CEROID LIPOFUSCINOSIS, CLN8-RELATED

DATA NOT AVAILABLE

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

NEURONAL CEROID LIPOFUSCINOSIS, PPT1-RELATED

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Finnish	1:70	98.0%	1:3451

NEURONAL CEROID LIPOFUSCINOSIS, TPP1-RELATED

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Newfoundlander	1:53	69.0%	1:169

NIEMANN-PICK DISEASE

Type A

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:90	97.0%	1:2968

NIJMEGEN BREAKAGE SYNDROME

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Eastern European Slavic	1:155	100.0%	Negligible
North American	1:158	70.0%	1:524

PENDRED SYNDROME

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Chinese	1:50	84.0%	1:307
Japanese	1:50	53.0%	1:105
Northern European Caucasian	1:60	50.0%	1:119

PHENYLKETONURIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Irish	1:34	72.0%	1:119
Turkish	1:34	65.0%	1:95
French-Canadian	1:45	56.0%	1:101
Polish	1:45	78.0%	1:201
Spanish	1:51	41.0%	1:86
Chinese	1:53	54.0%	1:114
Danish	1:55	43.0%	1:96
US Caucasian	1:62	51.0%	1:125
Korean	1:102	62.0%	1:267
Japanese	1:174	70.0%	1:578

POLYCYSTIC KIDNEY DISEASE

DATA NOT AVAILABLE

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

POMPE DISEASE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
African American	1:59	60.0%	1:146
Dutch	1:185	40.0%	1:308
Taiwanese, Chinese	1:185	80.0%	1:921

PREKALLIKREIN DEFICIENCY

DATA NOT AVAILABLE

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

PRIMARY HYPEROXALURIA, TYPE 1

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
European	1:173	44.0%	1:308
Worldwide	1:289	44.0%	1:515

PRIMARY HYPEROXALURIA, TYPE 2

DATA NOT AVAILABLE

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

PRIMARY HYPEROXALURIA, TYPE 3

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Worldwide	1:913	75.0%	1:3649

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PROPIONIC ACIDEMIA

PCCA, PCCB

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Japanese	1:160	35.0%	1:246

PCCB

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Northern European	1:160	30.0%	1:228
Spanish	1:160	50.0%	1:320

PROTHROMBIN DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Iranian, Italian	1:707	54.0%	1:1536

RH-NULL SYNDROME

DATA NOT AVAILABLE

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

RHIZOMELIC CHONDRODYSPLASIA PUNCTATE TYPE 1

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Pan-ethnic	1:158	51.0%	1:321

RICKETS, PSEUDOVITAMIN D-DEFICIENCY

DATA NOT AVAILABLE

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

SALLA DISEASE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Northeastern Finnish	1:100	95.0%	1:1981

SANDHOFF DISEASE

DATA NOT AVAILABLE

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

SHORT-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:15	65.0%	1:41

SICK SINUS SYNDROME

DATA NOT AVAILABLE

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

SICKLE CELL DISEASE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
African American	1:15	100.0%	0
Native American	1:150	100.0%	0
Hispanic American	1:203	100.0%	0
Middle Eastern	1:478	100.0%	0
Caucasian	1:642	100.0%	0
Asian Indian	1:652	100.0%	0
Filipino	1:879	100.0%	0
Asian	1:1315	100.0%	0
Southeast Asian	1:2365	100.0%	0

SMITH-LEMLI-OPITZ SYNDROME

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Northwestern European	1:50	69.2%	1:150
General	1:68	69.2%	1:219
Southern European	1:83	69.2%	1:267
Middle Eastern	1:129	69.2%	1:417
Hispanic	1:135	69.2%	1:436
African American	1:339	69.2%	1:1098

SPHEROCYTOSIS, HEREDITARY

DATA NOT AVAILABLE

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

TAY-SACHS DISEASE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Ashkenazi Jewish	1:31	99.0%	1:3001
Non-Jewish	1:250	46.0%	1:462

TAY-SACHS PSEUDODEFICIENCY

DATA NOT AVAILABLE

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

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THROMBOCYTOPENIA, CONGENITAL AMEGAKARYOCYTIC

DATA NOT AVAILABLE

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

TYROSINE HYDROXYLASE DEFICIENCY

DATA NOT AVAILABLE

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

TYROSINEMIA

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
French-Canadian	1:66	87.0%	1:501
Ashkenazi Jewish	1:100	99.0%	1:9901
US general	1:150	60.0%	1:374

USHER SYNDROME, TYPE 1F

DATA NOT AVAILABLE

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

VERY LONG-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY

DATA NOT AVAILABLE

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

VON WILLEBRAND DISEASE, TYPE 2 NORMANDY

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Pan-ethnic	1:500	75.0%	1:1997

VON WILLEBRAND DISEASE, TYPE 3

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Swedish, Finnish	1:500	10.0%	1:555

WILSON DISEASE

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Asian	1:90	57.0%	1:208
European	1:90	35.0%	1:138

ZELLWEGER SYNDROME SPECTRUM, PEX1-RELATED

POPULATION	CARRIER RATE	DETECTION RATE	RESIDUAL RISK
Pan-ethnic	1:147	80.0%	1:731

See Disclaimer(s) on page 22 of this Report

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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