NutriPATH

Conversation Starter: NutriPATH Carrier Screening

Carrier screening provides you and your patient with information regarding their reproductive risks and how their children may be impacted. By knowing each parent's genetic risks, families can understand the chance that a child may or may not be affected by certain hereditary conditions that run in the family.

Fast and Affordable Carrier Screening using Next Generation Sequencing

What information does this test provide?

• A **negative** test can reassure your patient that there is a reduced risk of having a child with a genetic disorder.

• A **positive** test allows your patient the chance to pursue reproductive options that can reduce the chance of having an affected child.

What genes/conditions are tested?

We perform ***full exonic (versus hotspot only)*** sequencing of 106 genes linked to 171 conditions. A complete list is available on the back page of this brochure.

What can we do with this information?

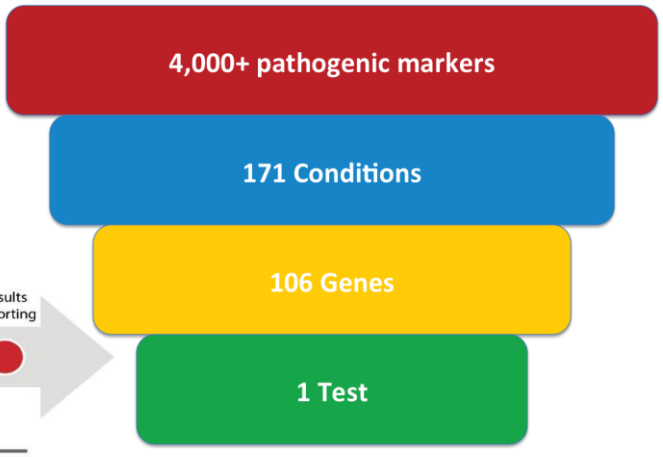
With positive results you can (1) find a specialist, (2) pursue Pre-implementation Genetic Diagnosis/Screening (PGD/PGS), (3) go through In-Vitro Fertilization (IVF) or (4) make other plans (adoption, etc.).

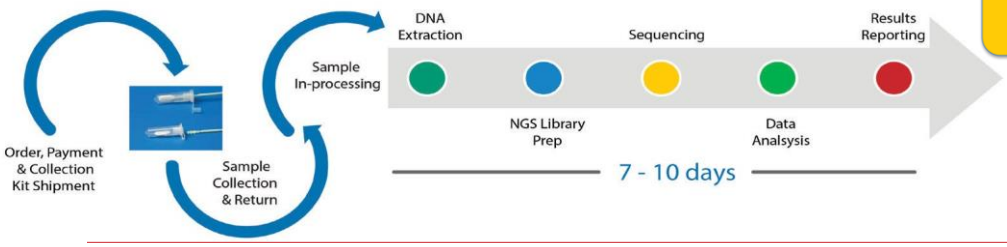
Most carriers of a recessive disorder are unaware of their status due to the lack of symptoms or family history!

How do we order the test?

To order a test and obtain a collection kit, start by taking this brochure to your doctor and discussing your options. A physician-ordered test requisition form and payment are all you need. NutriPATH Customer Reps are available to answer any questions via:

e-mail - **info@NutriPATH.com.au** or
call - **1300-688-522.**





DON'T RISK IT...SCREEN IT!!!

NutriPATH Customer Care
1300-688-522 | info@NutriPATH.com.au

SCAD Deficiency 17-beta-hydroxysteroid dehydrogenase X deficiency 2-methylbutyryl-CoA Dehydrogenase Deficiency 3-hydroxyacyl-CoA dehydrogenase deficiency 3-Methylcrotonyl-CoA carboxylase 1 deficiency (MCC1D) 3-Methylcrotonyl-CoA carboxylase 2 deficiency (MCC2D) 3-methylglutaconic aciduria type I (MCGA1) 3-methylglutaconic aciduria, type III 3-methylglutaconic aciduria, type V Adrenoleukodystrophy	Hb Barts Hb C disease (Hb CC) Hb C/ Beta^{0} thalassemia Hb C/Beta^{+} thalassemia Hb D disease (Hb DD) Hb D/ Beta^{0} thalassemia Hb D/Beta^{+} thalassemia Hb E/ Beta^{0} thalassemia Hb E/Beta^{+} thalassemia Hb EE
Alpha-methylacetoacetic aciduria (3-ketothialase deficiency) Argininemia (Arginase Deficiency) Arginosuccinic Aciduria Ault-onset citrullinemia Type II Autosomal dominant deafness Type 3A Autosomal dominant deafness Type IIB Autosomal dominant deafness Type IIIB Autosomal dominant persistent hypermethioninemia due to methionine adenosyltransferase I/III deficiency Autosomal recessive deafness Autosomal recessive deafness Type 1A	Hb H (3 gene deletion) Hb H/Constant Spring disease Hb S/ Beta^{0} thalassemia Hb S/Beta $^{+}$ thalassemia Hb Variant/ Beta^{0} thalassemia Hb Variant/Beta^{+} thalassemia Hb variants Hemolytic anemia due to G6PD deficiency Hepatic carnitine palmitoyl transferase deficiency Type I Hepatic carnitine palmitoyl transferase deficiency Type II
Autosomal recessive deafness Type IB Autosomal recessive deafness type IV Autosomal recessive Methionine adenosyltransferase deficiency Barth Syndrome Bart-Pumphrey Syndrome Beta thalassemia major BH4-deficient Hyperphenylalaninemia A BH4-deficient Hyperphenylalaninemia B BH4-deficient Hyperphenylalaninemia C BH4-deficient Hyperphenylalaninemia D	Hereditary persistence of fetal hemoglobin Hex A pseudodeficiency HMG-CoA Lyase Deficiency Holocarboxylase synthetase deficiency Homocystinuria due to MTHFR deficiency Homocystinuria, B6-responsive and nonresponsive types Hyperhomocysteinemic thrombosis Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase Hystrix-like ichthyosis with deafness Isobutyryl-CoA dehydrogenase deficiency
Biotinidase deficiency Bloom Syndrome Canavan disease Carnitine-acylcarnitine translocase (CACT) deficiency cbI E complementation type homocystinuria-megaloblastic anemia cbI G complementation type homocystinuria-megaloblastic anemia cblB complement type Vitamin B-12 responsive methylmalonic aciduria (due to defect in synthesis of adenosylcobalamin) cblD complement type homocystinuria (Variant 1) cblD complement type homocystinuria (Variant 2) cblD complement type Methylmalonic aciduria and homocystinuria	Isovaleric acidemia Keratitis ichthyosis deafness syndrome Krabbe disease LCHAD deficiency Lethal neonatal CPT2 deficiency Malonyl-CoA decarboxylase deficiency Maple syrup urine disease type II Maple syrup urine disease, type Ia Maple syrup urine disease, type Ib MCAD Deficiency
cblJ Type Methylmalonic aciduria and homocystinuria Citrullinemia Clouston type ectodermal dysplasia Type II Cogenital bilateral absence of the vas deferens (CVAD) Combined malonic and methylmalonic aciduria Congenital Adrenal Hyperplasia due to 21-hydroxylase deficiency Congenital hypothyroidism due to thyroid dysgenesis or hypoplasia Congenital nongoitrous hypothyroidism 1 Congenital nongoitrous hypothyroidism 4 Congenital nongoitrous hypothyroidism 6	Mental retardation X-linked syndromic 10 (MRXS10) Methylmalonic aciduria and homocystinuria, cblC type Methylmalonic aciduria due to Methylmalonyl-CoA Mutase deficiency Methylmalonic aciduria due to transcobalamin receptor defect Methylmalonyl-CoA epimerase deficiency Mucolipidosis IV Mucopolysaccharidosis Ih Mucopolysaccharidosis Ih/s Mucopolysaccharidosis Is Neonatal hypertrypsinemia
CPT2 deficiency associated myopathy Cystic Fibrosis Digenic deafness GJB2/GJB3 Digenic GJB2/GJB6 deafness Dihydrolipoamide dehydrogenase deficiency DOPA-responsive dystonia (with or without hyperphenylalaninemia) Erythrokeratodermia variabilis et progressiva Fabry disease Familial dilated cardiomyopathy Familial dysautonomia	Neonatal onset citrullinemia Type II Niemann-Pick disease, type A Niemann-Pick disease, type B Niemann-Pick disease, type C1 Niemann-pick disease, type C2 Niemann-Pick disease, type D Nonautoimmune hyperthyroidism Non-classic hyperandrogenism due to 21-hydroxylase deficiency Non-PKU hyperphenylalanemia Optic atrophy 3 with cataract
Familial gestational hyperthyroidism Familial hyperinsulinemic hypoglycemia type 4 Favism Galactokinase deficiency with cataracts Galactose epimerase deficiency Galactosemia Gaucher disease Type I Gaucher disease Type II Gaucher disease Type III Gaucher disease Type IIIC	Ornithine transcarbamylase deficiency Palmoplantar keratoderma with deafness Partial adenosine deaminase deficiency Pendred syndrome Perinatal lethal Gaucher disease Phenylketonuria Propionicacidemia Severe combined immunodeficiency (SCID) due to adenosine deaminase deficiency (ADAD) Sickle cell anemia (S/S) Sickle cell disease variants
Glutaric acidemia IIA Glutaric acidemia IIB Glutaric acidemia IIC Glutaric aciduria Type I Glycine N-methyltransferase deficiency Glycogen storage disease Ia Glycogen storage disease II GM2- gangliosidosis	Sickle hemoglobin C disease Sickle hemoglobin D disease Sickle hemoglobin E disease Susceptibility to acute-infection induced encephalopathy Suscptibility to autoimmune thyroid disease Type III Systemic primary carnitine deficiency Tay-Sachs disease Thryoid dyshormonogenesis 6 Thyroid dyshormonogenesis 1
	Thyroid dyshormonogenesis 2A Thyroid dyshormonogenesis 3 Thyroid hormone resistance Transcobalamin II deficiency Trifunctional protein deficiency Tyrosinemia, type I Tyrosinemia, type II Tyrosinemia, type III Vitamin B-12 responsive methylmalonic aciduria VLCAD deficiency Vohwinkel syndrome X-linked mental retardation with methylmalonic acidemia and homocysteinemia X-linked severe combined immunodeficiency (SCID)