About the Test
The Preparent™ Carrier Screen – Global+ Panel is a test that looks at your genes to determine whether you are a carrier of certain genetic disorders. A list of disorders included in this test appears on page 2. A positive result tells you with greater than 99% certainty that you are a carrier of a genetic disorder, and you could be at risk of having an affected child. If a risk is identified, you may wish to consider genetic carrier screening for your partner, consult with your healthcare provider, or pursue genetic counseling. If you are pregnant, prenatal testing can be performed to find out whether your baby has inherited the genetic disorder. You could also learn that you may be affected by a genetic disorder, although this is extremely rare. In addition, results may reveal clinical health implications for you which may require follow-up with your health care provider.

Limitations
Like all tests, this test has limitations. It is a screening test and is not intended to diagnose genetic conditions. If a risk is identified in your pregnancy, a prenatal diagnostic test such as chorionic villus sampling or amniocentesis is recommended. False positive and false negative results are rare but possible.

This test is designed to look for specific genetic changes. It cannot detect all genetic changes that could cause health problems. Normal results do not guarantee a healthy pregnancy or baby.

In the course of performing the test, information regarding other chromosomal alterations may become evident (called Incidental Findings). The laboratory does not report or comment on any Incidental Findings that may be noted in the course of analyzing the test data.

Risks
This test is performed on a blood draw. Side effects of having blood drawn are uncommon, but may include dizziness, fainting, soreness, bleeding, bruising, and, rarely, infection.

Required Information and Confidentiality
We keep test results confidential. Your test results will be sent only to the healthcare provider who ordered the test, or his/her agent, unless otherwise authorized by you or required by law. You may also contact us if you would like a copy of your test results. Your healthcare provider is responsible for interpreting your test results, explaining them to you, and determining the best next steps for your care. No other test will be performed and reported on your sample unless authorized by your healthcare provider. Leftover specimens from New York State will be destroyed within 60 days.

For the most accurate interpretation of test results, the laboratory needs to collect information about your health history. This may include information about your pregnancy (gestational age, number of babies), your health (height and weight, diabetes status, transplant status), and your family history (ethnic background, any known family history of genetic disease). This information is kept confidential.

Collecting information about your pregnancy after testing is part of a laboratory’s standard practice for quality purposes, and is required in several states. The laboratory may contact your healthcare provider to obtain this information.

Genetic Discrimination
The U.S. government has enacted laws to protect Americans against discrimination based on their genetic information for health insurance and employment. The laws may not protect against genetic discrimination in other circumstances, such as when applying for life insurance or long-term disability insurance. Talk to your healthcare provider or genetic counselor if you have concerns about genetic discrimination prior to testing.

Testing is Voluntary
The decision to accept or decline testing is completely yours. You may wish to consult with a certified genetic counselor before consenting to this test. Ask your healthcare provider for information about genetic counseling resources that are available to you. You can also find a genetic counselor through the National Society of Genetic Counselors at www.nsgc.org.

Before signing this form, I have had the opportunity to discuss this testing with my healthcare provider or someone he/she has designated, and genetic counseling has been recommended before and after testing. My questions have been answered and I have all the information I need to make a decision at this time. I have decided that:

☐ Yes, I want to receive the Preparent Carrier Screen – Global+ Panel.
☐ No, I do not want testing at this time.

__________________________  __________________________  ________________________
Patient Name (please print)  Patient Signature  Date
Disorders Tested – Global+ Panel

17-alpha-hydroxylase deficiency
17-beta-hydroxysteroid dehydrogenase deficiency, type III
3-beta-hydroxysteroid dehydrogenase deficiency, type II
3-hydroxy-3-methylglutaryl CoA lyase deficiency
3-methylcrotonyl-CoA carboxylase deficiency, MCCC1-related
3-methylcrotonyl-CoA carboxylase deficiency, MCCC2-related
3-methylglutaconic aciduria, type III
3-phosphoglycerate dehydrogenase deficiency
Abetalipoproteinemia
Achalasia-Addisonism-alacrima syndrome
Achromatopsia, CNGA3-related
Achromatopsia, CNGB3-related
Acrodermatitis enteropathica
Adenosine deaminase deficiency
Adrenoleukodystrophy (X-linked)
Acidic-Goutieres syndrome, RNAE2KC-related
Acidic-Goutieres syndrome, SAMHD1-related
Acidic-Goutieres syndrome, TREX1-related
Alkaptonuria
Alpha-1 antitrypsin deficiency
Alpha-mannosidosis
Alpha-thalassemia
Alport syndrome, autosomal recessive
Alport syndrome, X-linked
Amish infantile epilepsy syndrome
Andermann syndrome
Argininosuccinic aciduria
Aromatase deficiency
Arthrogryposis, mental retardation and seizures
Ataxia (X-linked)
Aspartylglycosaminuria
Ataxia neuropathy spectrum
Ataxia with vitamin E deficiency
Ataxia-telangiectasia
Ataxia-telangiectasia-like disorder
Autoimmune polyglandular syndrome, type I
Autosomal recessive poly cyclic kidney disease
Autosomal recessive woolly hair/hypotrichosis
Bardet-Biedl syndrome, BB51-related
Bardet-Biedl syndrome, BB52-related
Bardet-Biedl syndrome, BB53-related
Barker syndrome, type IV
Beta-ketothiolase deficiency
Beta-thalassemia
Bilateral frontoparietal polymicrogyria
Biotinidase deficiency
Bloom syndrome
Canavan disease
Carnitine palmitoyltransferase I deficiency
Carnitine palmitoyltransferase II deficiency
Carpenter syndrome
Cartilage-hair hypoplasia
Charcot-Marie-Tooth disease, GJB1-related (X-linked)
Charcot-Marie-Tooth disease, MPZ-related (X-linked)
Chediak-Higashi syndrome
Cholesterol ester storage disease
Choroideremia (X-linked)
Citrinemia, type I
Citronemia, type I
Congenital adrenal hyperplasia
Congenital amegakaryocytic thrombocytopenia
Congenital disorder of glycosylation, type IA
Congenital disorder of glycosylation, type IB
Congenital glycogen storage disease Ia
Congenital lipoid hyperplasia
Congenital neutropenia, autosomal recessive
Coneal dystrophy and perceptive deafness syndrome
Corticosteroid methyl oxidase deficiency
Creatine transporter defect, SLC6A9-related (X-linked)
Crigler-Najjar syndrome
Cystic fibrosis (600 mutations)
Cystinosis
D-bifunctional protein deficiency
Desbuquois dysplasia
Dihydropyrimidinase dehydrogenase deficiency
Du Pan syndrome
Dyskeratosis congenita, autosomal recessive
Dyskeratosis congenita, X-linked
Dystrophic epidermolysis bullosa, autosomal recessive
Early onset myopathy with fatal cardiomyopathy
Ehlers Danlos syndrome, type VIIC
Energy-Defruss muscular dystrophy (X-linked)
Enhanced S-cone syndrome
Ethylmalonic encephalopathy
Fabry disease (X-linked)
Familial dysautonomia
Familial hyperinsulinism, ABCC8-related
Familial hyperinsulinism, KCNJ11-related
Familial Mediterranean fever
Fanconi anemia, type A
Fanconi anemia, type C
Fetal akinesia deformation sequence, DOK7-related
Fragile X syndrome (X-linked)
Fumarase deficiency
Galactokinase deficiency
Galactosemia
Gardner syndrome
Geroferdinostriodysplasia
Gitelman syndrome
Glucose-6-phosphate dehydrogenase deficiency (X-linked)
Glutaric acidemia, type I
Glyceine encephalopathy, AMT-related
Glyceine encephalopathy, GLDC-related
Glycogen storage disease, type I A
Glycogen storage disease, type IB
Glycogen storage disease, type II
Glycogen storage disease, type III
Glycogen storage disease, type IV
Glycogen storage disease, type V
Glycogen storage disease, type VI
Glycogen storage disease, type VII
GM1-gangliosidosis
GRACILE syndrome
Guanosine triphosphate methyltransferase deficiency
Hemoglobinopathy, type evaluation
Hemoglobinopathy, Hb C
Hemoglobinopathy, Hb D
Hemoglobinopathy, Hb E
Hemoglobinopathy, Hb O
Hemoglobinopathy, Sickle cell anemia (HbS)
Hemophilia A (X-linked)
Hemophilia B (X-linked)
Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related
Hereditary fructose intolerance
Hereditary hemochromatosis, HFE-related
Hereditary hemochromatosis, HFE2-related
Hereditary hemochromatosis, HPRT2-related
Herlitz junctional epidermolysis bullosa, LAMB3-related
Hermansky-Pudlak syndrome, type III
Holocarboxylase synthetase deficiency
Homocystinuria, CBS-related
Hyperphosphatemic familial tumoral calcinosis
Hypophosphatotic ectodermal dysplasia
Hypophosphatasia, autosomal recessive
Inclusion body myopathy, type II
Isovaleric acidemia
Ioubert syndrome 2
Juvenile retinoschisis, X-linked
Krabbe disease
Lamellar ichthyosis, type I
Leber congenital amaurosis, LCA5-related
Leber congenital amaurosis, RDH12-related
Leigh syndrome, French Canadian
Limb-girdle muscular dystrophy, type 2A
Limb-girdle muscular dystrophy, type 2B
Limb-girdle muscular dystrophy, type 2D
Limb-girdle muscular dystrophy, type 2E
Limb-girdle muscular dystrophy, type 2f
Lipidosis lipase deficiency
Long-chain 3-hydroxacyl-CoA dehydrogenase deficiency
Luteinizing hormone resistance
Maple syrup urine disease, type IA
Maple syrup urine disease, type IB
Medium-chain acyl-CoA dehydrogenase deficiency
MEN1 syndrome
Metachromatic leukodystrophy
Methylaminocitric aciduria, cblA type
Methylaminocitric aciduria, cblB type
Methylaminocitric aciduria, cblC type
Methylaminocitric aciduria, MUL-related
Mitochondrial complex IV deficiency
Mitochondrial myopathy and sideroblastic anemia I
Mucolipidosis, type IIIB
Mucolipidosis, type IV
Mucopoly saccharidosis, type I (Hurler syndrome)
Mucopoly saccharidosis, type II (Hunter syndrome)
Mucopoly saccharidosis, type IIIC
Mucopoly saccharidosis, type VI
Mutlybrevianism
Multiple sulfatase deficiency
Muscle-eye-brain disease
Myotubular myopathy, MTM1-related (X-linked)
Nemaline myopathy 2
Nephrotic syndrome, type I
Neuronal ceroid lipofuscinosis, CLN5-related
Neuronal ceroid lipofuscinosis, CLN6-related
Neuronal ceroid lipofuscinosis, CLN8-related
Neuronal ceroid lipofuscinosis, PNSF5-related
Neuronal ceroid lipofuscinosis, PTP11-related
Neuronal ceroid lipofuscinosis, TPP1-related
Niemann-Pick disease, type A and B
Niemann-Pick disease, type CI
Niemann-Pick disease, type C1
Nijmegen breakage syndrome
Nonsyndromic hearing loss, GJB2-related
Onemman syndrome
Oxidative transcarbamylase deficiency (X-linked)
Oxidative translocase deficiency
Pendred syndrome
Phenylalanine hydroxylase deficiency
Pontocerebellar hypoplasia, type IA
Primary congenital glaucoma
Primary hyperoxaluria, type I
Primary hyperoxaluria, type II
Progressive familial intrahepatic cholestasis, type II
Prolidase deficiency
Propionic acidemia, PCCA-related
Propionic acidemia, PCCB-related
Pseudocholesterase deficiency
Pseudoxanthoma elasticum
Pychnodysostosis
Pyruvate dehydrogenase deficiency, autosomal recessive
Pyruvate dehydrogenase deficiency, X-Linked
Retinitis pigmentosa 59
Rhizomelic chondrodysplasia punctata, type I
Salla disease
Sandhoff disease
Severe combined immunodeficiency, RAG1-related
Severe combined immunodeficiency, X-linked
Short-chain acyl-CoA dehydrogenase deficiency
Shwachman-Diamond syndrome
Sjögren-Larsson syndrome
Smith-Lemli-Opitz syndrome
Spastic ataxia of Charlevoix-Saguenay, autosomal recessive
Spastic paraplegia, ZFYVE26-related
Spinal muscular atrophy
Steroid-resistant nephrotic syndrome
Stuve-Wiedemann syndrome
Sulfate transporter-related osteochondrodysplasia
Tay-Sachs disease
Tumoral calcinosis, normophosphatemic
Tyrosinemia, type I
Tyrosinemia, type II
Usher syndrome, type IB
Usher syndrome, type IC
Usher syndrome, type II
Usher syndrome, type IF
Usher syndrome, type III
Very-long chain acyl-CoA dehydrogenase deficiency
Vitamin D-dependent rickets, type I
Walker-Warburg syndrome
Wilson disease
Xeroderma pigmentosum
Zellweger syndrome spectrum, PEX-related