Fore Genomics

FORESITE 360 Disease List

vQ42023

Cardiovascular

- Dilated Cardiomyopathy 3b
- Alagille Syndrome 1 / Tetralogy Of Fallot
- Familial Hypercholesterolemia
- Long-Chain 3-Hydroxyacyl-Coa Dehydrogenase Deficiency
- Primary Carnitine Deficiency
- Very Long Chain Acyl-Coa Dehydrogenase Deficiency
- Aortic Aneurysm, Familial Thoracic 4
- Aortic Aneurysm, Familial Thoracic 6
- Arrhythmogenic Right Ventricular Cardiomyopathy, Type 5
- Arrhythmogenic Right Ventricular Cardiomyopathy, Type 8
- Arrhythmogenic Right Ventricular Cardiomyopathy, Type 9
- Arrhythmogenic Right Ventricular Cardiomyopathy, Type 10
- Arrhythmogenic Right Ventricular Cardiomyopathy, Type 11
- Dilated Cardiomyopathy 1e
- Catecholaminergic Polymorphic Ventricular Tachycardia
- Dilated Cardiomyopathy
- Dilated Cardiomyopathy
- Dilated Cardiomyopathy 1a
- Familial Hypercholesterolemia
- Familial Hypercholesterolemia
- Familial Hypertrophic Cardiomyopathy 1
- Familial Hypertrophic Cardiomyopathy 3
- Familial Hypertrophic Cardiomyopathy 4
- Familial Hypertrophic Cardiomyopathy 6
- Familial Hypertrophic Cardiomyopathy 7
- Familial Hypertrophic Cardiomyopathy 8
- Familial Hypertrophic Cardiomyopathy 10
- Familial Hypertrophic Cardiomyopathy 11
- Osler-Weber-Rendu Disease
- Telangiectasia, Hereditary Hemorrhagic, Type 2
- Hypercholesterolemia
- Left Ventricular Noncompaction 6
- Loeys-Dietz Syndrome Type 1a
- Loeys-Dietz Syndrome Type 1b
- Loeys-Dietz Syndrome Type 2b
- Loeys-Dietz Syndrome Type 3
- Long Qt Syndrome 1
- Long Qt Syndrome 2
- Long Qt Syndrome 3
- Pompe Disease
- Barth Syndrome

Congenital Abnormalities

- Smith-Lemli-Opitz Syndrome
- Gm1 Gangliosidosis
- Mucopolysaccharidosis
- Mucopolysaccharidosis Type 7
- Heterotaxy Syndrome

Dermatologic

- Ehlers-Danlos
- Osteogenesis Imperfecta Type I, Ehlers-Danlos Syndrome
- Xeroderma Pigmentosum
- Xeroderma Pigmentosum
- Xeroderma Pigmentosum Group A
- Xeroderma Pigmentosum
- Ehlers-Danlos Syndrome, Type 4
- Pilomatrixoma
- Pten Hamartoma Tumor Syndrome

Dermatologic, Pediatric Oncology

- Neurofibromatosis, Type 1

Dermatologic, Vision Loss

- Hermansky-Pudlak Syndrome 1
- Hermansky-Pudlak Syndrome

Dermatological, Immunodeficiency

- Acrodermatitis Enteropathica, Transient Neonatal Zinc Deficiency

Endocrine

- Cold-Induced Sweating Syndrome 1
- Congenital Adrenal Hyperplasi
- Maturity-Onset Diabetes Of The Young Type 1, Type Ii Diabetes Mellitus, Fanconi Renotubular Syndrome 4
- Glucocorticoid Deficiency 1
- Congenital Adrenal Hypoplasia
- Congenital Adrenal Hyperplasia
- 3-Beta-Hydroxysteroid Dehydrogenase Type Ii Deficiency
- Central Hypothyroidism And Testicular Enlargement
- Combined Pituitary Hormone Deficiency 1
- Combined Pituitary Hormone Deficiency 2
- Combined Pituitary Hormone Deficiency 3
- Congenital Adrenal Hyperplasia Due To 11-Beta-Hydroxylase Deficiency
- Congenital Hypothyroidism Due To Thyroid Dysgenesis Or Hypoplasia
- Congenital Nongoitrous Hypothryoidism 1 / Nonautoimmune Hyperthyroidism
- Congenital Nongoitrous Hypothryoidism 4
- Congenital Nongoitrous Hypothryoidism 6
- Corticosterone Methyloxidase Deficiency

- Familial Hyperinsulinemic Hypoglycemia 4 / 3-Hydroxyacyl-Coa Dehydrogenase Deficiency
- Familial Hyperinsulinism
- Familial Hyperinsulinism
- Generalized Thyrotropin-Releasing Hormone Resistance
- Hyperinsulinism-Hyperammonemia Syndrome
- Lipoid Adrenal Hyperplasia
- Neonatal Hypoparathyroidism / Autosomal Dominant Hypocalcemia
- Permanent Neonatal Diabetes Mellitus
- Thyroid Dyshormonogenesis 1
- Thyroid Dyshormonogenesis 2a
- Thyroid Dyshormonogenesis 3
- Thyroid Dyshormonogenesis 4
- Thyroid Dyshormonogenesis 5
- Thyroid Dyshormonogenesis 6
- Maturity-Onset Of Diabetes Of The Young
- Multiple Endocrine Neoplasia, Type 1
- Multiple Endocrine Neoplasia, Type 2a
- Multiple Endocrine Neoplasia, Type 2b

Endocrine, Gastrointestinal

- Wolman Disease / Cholesteryl Ester Storage Disease

Gastrointestinal

- Insulin-Dependent Diabetes Mellitus Secretory Diarrhea Syndrome
- Lipoprotein Lipase Deficiency
- Lysinuric Protein Intolerance
- Adenomatous Polyposis Coli, Gardner Syndrome
- Juvenile Polyposis Syndrome
- Peutz-Jeghers Syndrome

Hearing Loss

- Autosomal Recessive Nonsyndromic Hearing Loss 1a, Autosomal Dominant Nonsyndromic Hearing Loss 3a, Ichthyosis
- Autosomal Recessive Nonsyndromic Hearing Loss
- Pendred Syndrome
- Hearing Impairment
- Autosomal Recessive Nonsyndromic Hearing Loss 6
- Autosomal Recessive Nonsyndromic Hearing Loss 8
- Autosomal Recessive Nonsyndromic Hearing Loss 79
- Autosomal Recessive Nonsyndromic Hearing Loss 28
- Neurofibromatosis, Type 2

Hearing Loss, Cardiovascular

- Congenital Long Qt Syndrome
- Jervell And Lange-Nielsen Syndrome 2
- Long Qt Syndrome 5

Hearing Loss, Dermatologic

- Waardenburg Syndrome
- Craniofacial-Deafness-Hand Syndrome

Hearing Loss, Vision Loss

- Autosomal Recessive Nonsyndromic Hearing Loss 12
- Usher Syndrome Type 1d
- Usher Syndrome Type 1c
- Usher Syndrome Type 1g
- Usher Syndrome Type 2a
- Usher Syndrome Type 2d
- Autosomal Recessive Nonsyndromic Hearing Loss 31

Hematologic

- Upshaw-Schulman Syndrome
- Pyruvate Kinase Deficiency Of Red Cells
- Elliptocytosis, Spherocytosis,
- Alpha-Thalassemia
- Beta-Globin-Related Hemoglobinopathies
- Congenital Amegakaryocytic Thrombocytopenia
- Factor Ix Deficiency
- Hemolytic Anemia
- Homocystinuria-Megaloblastic Anemia
- Cobalamin G Type
- Spherocytosis, Type 1
- Spherocytosis, Type 5

Hepatic

- Steatohepatitis
- Acute Infantile Liver Failure
- Glycogen Storage Disease, Type 0
- Glycogen Storage Disease, Type Ia
- Glycogen Storage Disease, Type Ii
- Glycogen Storage Disease, Type Iii
- Glycogen Storage Disease, Type Ixb
- Glycogen Storage Disease, Type Vi
- Hmg-Coa Synthase 2 Deficiency
- Holocarboxylase Synthetase Deficiency
- Hereditary Hemochromatosis
- Wilson Disease

Hepatic, Gastrointestinal

- Congenital Bile Acid Synthesis Defect

Hepatic, Neurological

- Crigler-Najjar Syndrome, Types 1 And 2 / Gilbert Syndrome

Immunodeficiency

- X-Linked Agammaglobulinemia With Growth Hormone Deficiency
- Hyper-Igm Syndrome Type 1

- Mhc Class Ii Deficiency
- Combined Immunodeficiency Due To Dock8 Deficiency
- Familial Mediterranean Fever
- Granulomatous Disease,
- Familial Hemophagocytic Lymphohistiocytosis 2
- Mhc Class Ii Deficiency
- Mhc Class Ii Deficiency
- Mhc Class Ii Deficiency
- Zap70-Related Severe Combined Immunodeficiency
- Adenosine Deaminase Deficiency
- Chronic Granulomatous Disease
- Chronic Granulomatous Disease
- Congenital Neutropenia
- Immunodeficiency 18
- Immunodeficiency 19
- Omenn Syndrome
- Omenn Syndrome / Severe Combined Immunodeficiency, Athabaskan-Type
- Omenn Syndrome And Other Rag1-Related Disorders
- Severe Combined Immunodeficiency
- Severe Combined Immunodeficiency
- Severe Combined Immunodeficiency
- X-Linked Severe Combined Immunodeficiency

Immunodeficiency, Dermatologic

- Vitamin D-Dependent Rickets Type Ii With Alopecia

Inborn Errors Of Metabolism

- Deficiency Of Isobutyryl-Coa Dehydrogenase
- Menkes Kinky-Hair Syndrome
- X-Linked Distal Spinal Muscular Atrophy Type 3
- Cutis Laxa, X-Linked
- Vitamin D-Dependent Rickets, Type 1
- Niemann-Pick Disease
- Glycogen Storage Disease, Type I
- Transcobalamin Ii Deficiency
- 3-Methylcrotonyl-Coa Carboxylase Deficiency
- 3-Methylcrotonyl-Coa Carboxylase Deficiency
- 3-Phosphoglycerate Dehydrogenase Deficiency
- 6-Pyruvoyl-Tetrahydropterin Synthase Deficiency
- Abetalipoproteinemia
- Argininemia
- Argininosuccinic Aciduria
- Bh4-Deficient Hyperphenylalaninemia C
- Bh4-Deficient Hyperphenylalaninemia D
- Biotinidase Deficiency
- Carbamoylphosphate Synthetase I Deficiency
- Cerebral Creatine Deficiency Syndrome 2
- Citrin Deficiency
- Citrullinemia, Type 1
- Congenital Disorder Of Glycosylation, Type Ib
- Cystinosis
- Dopa-Responsive Dystonia / Bh4-Deficient Hyperphenylalaninemia B
- Fabry Disease
- Fructose-1,6-Bisphosphatase Deficiency
- Galactokinase Deficiency
- Galactose Epimerase Deficiency
- Galactosemia
- Hereditary Fructose Intolerance
- Maple Syrup Urine Disease, Type 1a
- Maple Syrup Urine Disease, Type 1b
- Maple Syrup Urine Disease, Type 2
- Methylmalonyl-Coa Epimerase Deficiency
- Niemann-Pick Disease
- Ornithine Transcarbomylase Deficiency
- Propionic Acidemia
- Propionic Acidemia
- Tyrosinemia, Type I
- Tyrosinemia, Type Ii
- Tyrosinemia, Type Iii
- Biotinidase Deficiency
- Fabry's Disease
- Malignant Hyperthermia
- Ornithine Carbamoyltransferase Deficiency
- Beta-Ketothiolase Deficiency

Neurological

- Carnitine Acylcarnitine Translocase Deficiency
- Carnitine Palmitoyltransferase Ia Deficiency
- Carnitine Palmitoyltransferase Ii Deficiency
- Cerebral Folate Transport Deficiency
- Galactosylceramide Beta-Galactosidase Deficiency
- Mucopolysaccharidosis
- Migraine, Familial Hemiplegic, 3, Epilepsy, Generalized Epilepsy With Febrile Seizures
- Leigh Syndrome
- Adrenoleukodystrophy, X-Linked
- Ataxia With Isolated Vitamin E Deficiency
- Cerebrotendinous Xanthomatosis
- Early Infantile Epileptic Encephalopathy 11
- Benign Familial Infantile Seizures 3
- Early Infantile Epileptic Encephalopathy 13
- Benign Familial Infantile Seizures 5
- Early Infantile Epileptic Encephalopathy 7
- Benign Neonatal Seizures
- Ethylmalonic Encephalopathy
- Familial Infantile Convulsions With Paroxysmal Choreoathetosis
- Glucose Transporter 1 Deficiency Syndrome And Other Slc2a1-Related Disorders
- Glutaric Acidemia, Type I

- Glutaric Acidemia, Type Iia
- Glutaric Acidemia, Type Iib
- Glutaric Acidemia, Type Iic
- Glutathione Synthetase Deficiency
- Hmg-Coa Lyase Deficiency
- Homocystinuria
- Homocystinuria, Cobalamin E Type
- Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome
- Isovaleric Acidemia
- Lipoamide Dehydrogenase Deficiency
- Malonyl-Coa Decarboxylase Deficiency
- Medium Chain Acyl-Coa Dehydrogenase Deficiency
- Metachromatic Leukodystrophy
- Methionine Adenosyltransferase I/Iii Deficiency
- Methylmalonic Acidemia
- Methylmalonic Acidemia
- Methylmalonic Acidemia
- Methylmalonic Aciduria And Homocystinuria, Cobalamin C Type
- Methylmalonic Aciduria And Homocystinuria, Cobalamin D Type
- Methylmalonic Aciduria And Homocystinuria, Cobalamin F Type
- Mitochondrial Trifunctional Protein Deficiency
- Mucopolysaccharidosis Type I
- Mucopolysaccharidosis Type Ii
- Mucopolysaccharidosis Type Iva
- Mucopolysaccharidosis Type Vi
- N-Acetylglutamate Synthase Deficiency
- Neurodegeneration Due To Cerebral Folate Transport Deficiency
- Phenylalanine Hydroxylase Deficiency
- Pyridoxamine 5’-Phosphate Oxidase Deficiency
- Pyridoxine-Dependent Epilepsy
- Segawa Syndrome
- Sepiapterin Reductase Deficiency
- Spinal Muscular Atrophy

Neurological, Muscular

- Cerebral Creatine Deficiency Syndrome 3

Pediatric Oncology

- Gorlin Syndrome
- Familial Medullary Thyroid Carcinoma
- Hereditary Paraganglioma-Pheochromocytoma Syndrome
- Transmembrane Protein 127
- Lynch Syndrome
- Myh-Associated Polyposis
- Paragangliomas 1
- Paragangliomas 2
- Paragangliomas 3
- Paragangliomas 4
- Retinoblastoma
- Tuberous Sclerosis 1
- Tuberous Sclerosis 2
- Von Hippel-Lindau Syndrome
- Wilms' Tumor
- Familial Medullary Thyroid Carcinoma
- Retinoblastoma
- Breast-Ovarian Cancer, Familial 1
- Breast-Ovarian Cancer, Familial 2
- Li-Fraumeni Syndrome 1

Pulmonary

- Bronchiectasis
- Cystic Fibrosis

Renal

- Polycystic Kidney Disease 2
- Autosomal Recessive Polycystic Kidney Disease
- Alport Syndrome
- Alport Syndrome
- Alport Syndrome
- Nephrogenic Diabetes Insipidus, Type Ii
- Nephrogenic Syndrome Of Inappropriate Antidiuresis / Nephrogenic Diabetes Insipidus
- Primary Hyperoxaluria, Type 1
- Primary Hyperoxaluria, Type 2
- Primary Hyperoxaluria, Type 3
- Wilms Tumor, Type 1 And Other Wt1-Related Disorders
- Distal Renal Tubular Acidosis And Other Slc4a1-Related Disorders

Skeletal

- Craniometaphyseal Dysplasia
- Neutropenia
- Hidrotic Ectodermal Dysplasia Syndrome
- Hypophosphatemic Bone Disease
- Hypophosphatasia
- Marfan Syndrome And Other Fbn1-Related Disorders
- Osteopetrosis 1
- Marfan's Syndrome

Vision Loss

- Ocular Albinism
- Autosomal Dominant Optic Atrophy
- Rpe65-Related Retinopathy
- Oculocutaneous Albinism Type 4
- Ornithine Aminotransferase Deficiency