

FamilyFirst - בדיקת סקר גנטי טרום הריוני מורחב, הסוקרת מעל 850 מוטציות העלולות לחולל 261 מחלות תורשתיות. הבדיקה כוללת את כל המחלות המומלצות ע"י משרד הבריאות ואיגוד הגנטיקאים הישראלי

Disease Name
3-methylglutaconic aciduria, type III
3-phosphoglycerate dehydrogenase deficiency
Abetalipoproteinemia
Achromatopsia, type 2
Acyl-CoA dehydrogenase, short-chain, deficiency
Adams-Oliver syndrome
Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency
Adrenocorticotrophic hormone deficiency
Adrenoleukodystrophy
Aicardi-Goutieres syndrome, type 5
Albinism, oculocutaneous, type IA
Alport syndrome
Apnea, postanesthetic
Argininosuccinic aciduria
Arthrogryposis, autism spectrum disorder, and epilepsy
Arthropathy, progressive pseudorheumatoid, of childhood
Asparagine synthetase deficiency
Aspartylglucosaminuria
Ataxia-telangiectasia
Autoimmune polyendocrinopathy syndrome, type I, with or without reversible metaphyseal dysplasia
Bardet-Biedl syndrome 10
Bardet-Biedl syndrome 11
Bardet-Biedl syndrome 1
Bardet-Biedl syndrome 2
Bardet-Biedl syndrome 3
Bardet-Biedl syndrome 4
Bartter syndrome, type 3
Bartter syndrome, type 4a

Disease Name
Biotinidase Deficiency
Bloom syndrome
Brittle cornea syndrome 1
Canavan disease
Carbamoylphosphate synthetase I deficiency
Cardiomyopathy, dilated, IGG
Carnitine acylcarnitine translocase deficiency
Carnitine palmitoyltransferase deficiency, hepatic, type II, infantile,lethal neonatal
Carnitine palmitoyltransferase deficiency, hepatic, type IA
Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome
Cerebrotendinous xanthomatosis
Choreoacanthocytosis
Chronic granulomatous disease (cytochrome b-negative)
Chronic granulomatous disease (cytochrome b-positive, type 1)
Chronic granulomatous disease, X-linked
Cockayne, type A
Complement factor H deficiency
Complex hereditary spastic paraparesis
Congenital adrenal insufficiency with 46,XY sex reversal
Congenital amegakaryocytic thrombocytopenia
Congenital disorder of deglycosylation
Congenital disorder of glycosylation type Ia
Congenital disorder of glycosylation, type Im
Cornelia de Lange like (Birk Flusser) syndrome
Cystic fibrosis
Cystinosis, nephropathic
Deafness, autosomal recessive 12

Disease Name
Deafness, autosomal recessive 16
Deafness, autosomal recessive 1A
Deafness, autosomal recessive 1B
Deafness, autosomal recessive 22
Deafness, autosomal recessive 3
Deafness, autosomal recessive 59
Deafness, autosomal recessive 7
Deafness, autosomal recessive 76
Deafness, autosomal recessive 8/10
Deafness, autosomal recessive 77
Desmosterolosis
Diabetes insipidus, nephrogenic
Diaphanospondylodysostosis
Dihydrolipoamide dehydrogenase deficiency
Duchenne muscular dystrophy
Dysautonomia, familial
Dyserythropoietic anemia, congenital, type 1a
Dyserythropoietic anemia, congenital, type 2
Dyskeratosis congenita, autosomal recessive 4 (TERT-related)
Dyskeratosis congenita, autosomal recessive 5 (RTEL1-related)
Ehlers-Danlos syndrome, type VII-C
Enhanced S-cone syndrome
Epidermolysis bullosa, junctional, Herlitz type (LAMA3-related)
Epidermolysis bullosa, junctional, Herlitz type (LAMB3-related)
Epidermolysis bullosa, junctional, with pyloric atresia
Epidermolysis bullosa, junctional, Herlitz type (LAMC2-related)
Epilepsy, progressive myoclonic 1B
Factor V and factor VIII combined deficiency

Disease Name
Factor VII deficiency
Factor XI deficiency
Familial hypercholesterolemia (LDLR-related)
Familial Mediterranean fever
Fanconi anemia, complementation group C
Fanconi anemia, complementation group A
Fanconi-Bickel syndrome
Fragile X syndrome
Fundus albipunctatus
Galactosemia
Gaucher disease, type I
Glanzmann thrombasthenia (ITGA2B-related)
Glanzmann thrombasthenia (ITGB3-related)
Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset
Glutaric aciduria type IIC
Glutaricaciduria, type I
Glycine encephalopathy (AMT-related)
Glycine encephalopathy (GLDC-related)
Glycogen storage disease Ia (von Gierke disease)
Glycogen storage disease Ib
Glycogen storage disease II (Pompe disease)
Glycogen storage disease IIIa/Glycogen storage disease IIIb (Cori or Forbes disease)
Glycogen storage disease IV
Glycogen storage disease VII
Gray platelet syndrome
Growth hormone deficiency, isolated, type IB (GHRHR - related)
Growth hormone deficiency, isolated, type IA (GHI-related)
Growth retardation, developmental delay, facial dysmorphism (GDFD)

Disease Name
Haim-Munk syndrome
Hemolytic anemia, with or without immune-mediated polyneuropathy
Hermansky-Pudlak syndrome 3
Hermansky-Pudlak syndrome 6
HMG-CoA lyase deficiency
Homocystinuria due to MTHFR deficiency
Hyperinsulinemic hypoglycemia, familial, 1
Hyperoxaluria, primary, type I
Hyperoxaluria, primary, type III
Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis
Hypoaldosteronism, congenital, due to CMO I deficiency/Hypoaldosteronism, congenital, due to CMO II deficiency
Hypomagnesemia I, intestinal
Hypoparathyroidism-retardation-dysmorphism syndrome
Hypophosphatasia, infantile
Hypophosphatemic rickets with hypercalciuria
Hypothyroidism, congenital, nongoitrous, 1
Hypotonia-cystinuria syndrome
Inclusion body myopathy, autosomal recessive
Infantile neuroaxonal dystrophy 1
Insensitivity to pain, congenital
Insensitivity to pain, congenital, with anhidrosis
Isovaleric acidemia
Joubert syndrome 2
Kohlschutter-Tonz syndrome
Krabbe disease
Laron dwarfism
Leber congenital amaurosis 1
Leber congenital amaurosis 2
Leber congenital amaurosis 4

Disease Name
Leber congenital amaurosis 5
Leigh syndrome, due to COX deficiency
Lethal congenital contractural syndrome 2
Lethal congenital contractural syndrome 3
Lethal congenital contracture syndrome 4
Leukodystrophy, hypomyelinating, 3
Leukodystrophy, hypomyelinating, 4
Liver failure, transient infantile
Mandibuloacral dysplasia
Maple syrup urine disease, type Ia
Maple syrup urine disease, type Ib
Maple syrup urine disease, type II
McArdle disease
Meconium ileus, familial
Medium-chain acyl-CoA dehydrogenase deficiency
Megalencephalic leukoencephalopathy with subcortical cysts
Megaloblastic anemia-1, Norwegian type (Imerslund-Gräsbeck syndrome)
Mental retardation, autosomal recessive 3
Metachromatic leukodystrophy
Microcephaly 9, primary, autosomal recessive
Microcephaly, postnatal progressive, with seizures and brain atrophy
Minicore myopathy with external ophthalmoplegia
Mitochondrial complex I deficiency (NDUFA11-related)
Mitochondrial complex I deficiency (NDUFS4-related)
Mitochondrial complex I deficiency (NDUFAF5-related)
Mitochondrial complex I deficiency (NDUFS6-related)

Disease Name
Mitochondrial complex III deficiency, nuclear type 4
Mitochondrial DNA depletion syndrome 2 (myopathic type)
Mitochondrial DNA depletion syndrome 1 (MNGIE type)
Mitochondrial DNA depletion syndrome 3 (hepatocerebral type)
Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)
Mitochondrial myopathy and sideroblastic anemia 1
Molybdenum cofactor deficiency A
Mucopolipidosis III gamma
Mucopolipidosis IV
Mucopolysaccharidosis type IIIA (Sanfilippo A)
Mucopolysaccharidosis I <sub>H</sub>
Multiple congenital anomalies-hypotonia-seizures syndrome 1
Multiple sulfatase deficiency
Muscular dystrophy, limb-girdle, type 2B
Muscular dystrophy, limb-girdle, type 2C
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4
Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency
Nemaline myopathy 2, autosomal recessive
Nephronophthisis 2, infantile
Nephrotic syndrome, type 1
Nephrotic syndrome, type 2
Neuropathy, distal hereditary motor, type VI

Disease Name
Neutropenia, severe congenital 4, autosomal recessive
Niemann-Pick disease, type A
Niemann-Pick disease, type B
Niemann-Pick disease, type C1
Omenn syndrome / T- B- severe combined immunodeficiency
Omenn syndrome / T- B- severe combined immunodeficiency
Osteopetrosis, autosomal recessive 8
Osteopetrosis, autosomal recessive 1
Otospondylomegaepiphyseal dysplasia
Pendred syndrome
Peroxisome biogenesis disorder 5A (Zellweger)
Peroxisome biogenesis disorder 1A (Zellweger)
Peroxisome biogenesis disorder 4A (Zellweger)
Phenylalanine hydroxylase deficiency (including phenylketonuria)
Polycystic kidney disease, autosomal recessive
Polymicrogyria, bilateral frontoparietal
Pontocerebellar hypoplasia, type 2D (Progressive cerebello-cerebral atrophy, type 2D)
Pontocerebellar hypoplasia, type 2E
Pontocerebellar hypoplasia, type 1A
Primary ciliary dyskinesia-12 (RSPH9-related)
Primary ciliary dyskinesia-16 (DNALI1-related)
Primary ciliary dyskinesia-9 (DNAI2-related)
Prolidase deficiency
Propionicacidemia
Proximal myopathy and ophthalmoplegia
Pycnodysostosis
Pyridoxamine 5'-phosphate oxidase deficiency
Retinitis pigmentosa 12
Retinitis pigmentosa 14

Disease Name
Retinitis pigmentosa 25
Retinitis pigmentosa 26
Retinitis pigmentosa 28
Retinitis pigmentosa 36
Retinitis pigmentosa 57
Retinitis pigmentosa 59
Retinitis pigmentosa 64
Rickets, vitamin D-resistant, type IIA
Sandhoff disease, infantile, juvenile, and adult forms
Severe combined immunodeficiency due to ADA deficiency
Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis
Sialic acid storage disorder, infantile
Smith-Lemli-Opitz syndrome
Spastic paraplegia 49, autosomal recessive
Spastic paraplegia 53, autosomal recessive
Spinal muscular atrophy (SMA)
Spondylometaepiphyseal dysplasia, short limb-hand type
Stargardt disease I including Cone-rod dystrophy 3
Striatonigral degeneration, infantile
Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome
Surfactant metabolism dysfunction, pulmonary, 3
Tay-Sachs disease
Thalassemia, alpha
Thalassemias, beta
Thiamine-responsive megaloblastic anemia syndrome
Tumoral calcinosis, familial, normophosphatemic
Tumoral calcinosis, familial, hyperphosphatemic
Tyrosinemia, type III

Disease Name
Tyrosinemia, type I
Usher syndrome, type IB
Usher syndrome, type IC
Usher syndrome, type IF
Usher syndrome, type 2A
Usher syndrome, type 3A
Ventricular tachycardia, catecholaminergic polymorphic, 2
Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
Wilson disease
Wiskott-Aldrich syndrome
Wolman disease
Woodhouse-Sakati syndrome
Xeroderma pigmentosum, variant type
Xeroderma pigmentosum, group C
Xeroderma pigmentosum, group G/Cockayne syndrome