FamilyFirst
סקר גנטי טרום הריון

רשימת המחלות הנסקרות בבדיקה

FamilyFirst - בדיקת סקר גנטי טרום הריוני מורחב, הסוקרת כ-1,700 מוטציות העלולות לחולל כ- 360 מחלות תורשתיות. הבדיקה כוללת את כל המחלות המומלצות ע"י משרד הבריאות ואיגוד הגנטיקאים הישראלי. בדיקת סקר לתלסמיה מבוצעת באמצעות ספירת דם - יש לברר מול הרופא המטפל.

Disease Name
Alkaptonuria
Carnitine deficiency, systemic primary
Gitelman syndrome
Hemophilia A
Hemophilia B/Thrombophilia, X-linked, due to factor IX defect
Methylmalonic aciduria and homocystinuria, cblC type
Methylmalonic aciduria, mut(0) type
Pseudohermaphroditism, male, with gynecomastia
Segawa syndrome, recessive
Bare lymphocyte syndrome, type II, complementation group A
17,20-lyase deficiency, isolated/17-alpha-hydroxylase/17,20-lyase deficiency
2-methylbutyrylglycinuria
3-Methylcrotonyl-CoA carboxylase 1 deficiency
3-Methylcrotonyl-CoA carboxylase 2 deficiency
3-methylglutaconic aciduria, type III
3-phosphoglycerate dehydrogenase deficiency
4 optional diseases
5-fluorouracil toxicity/Dihydropyrimidine dehydrogenase deficiency
Abetalipoproteinemia
Achalasia-addisonianism-alacrimia syndrome
Achondrogenesis Ib/Atelosteogenesis, type II/De la Chapelle dysplasia
Achromatopsia 3/Macular degeneration, juvenile

Disease Name
Achromatopsia, type 2
Acromesomelic dysplasia, Demirhan type
Acyl-CoA dehydrogenase, short-chain, deficiency of
Adams–Oliver syndrome
Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency
Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency/Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency
Adrenocorticotropic hormone deficiency
Adrenoleukodystrophy
Aicardi-Goutieres syndrome, type 5
Albinism, oculocutaneous, type IA
Albinism, oculocutaneous, type II/Albinism, brown oculocutaneous/
Albinism, oculocutaneous, type IV
Alport syndrome
Alport syndrome 2, autosomal recessive
Alstrom syndrome
Anauxetic dysplasia 1/Cartilage-hair hypoplasia/Metaphyseal dysplasia without hypotrichosis
Argininosuccinic aciduria
Arterial calcification, generalized, of infancy, 2/Pseudoxanthoma elasticum
Arthrogryposis, autism spectrum disorder, and epilepsy
Arthropathy, progressive pseudorheumatoid, of childhood
Asparagine synthetase deficiency
Aspartylglucosaminuria

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Disease Name
Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia
Ataxia-telangiectasia
Ataxia-telangiectasia-like disorder
Autoimmune polyendocrinopathy syndrome, type I, with or without reversible metaphyseal dysplasia
Bardet-Biedl syndrome 1
Bardet-Biedl syndrome 10
Bardet-Biedl syndrome 11
Bardet-Biedl syndrome 12
Bardet-Biedl syndrome 13/Joubert syndrome 28/Meckel syndrome 1
Bardet-Biedl syndrome 2
Bardet-Biedl syndrome 3
Bardet-Biedl syndrome 4
Bartter syndrome, type 3
Bartter syndrome, type 4a
Basel-Vanagait-Smirin-Yosef syndrome
Bernard-Soulier syndrome, type B/Giant platelet disorder, isolated
Bernard-Soulier syndrome, type C
Biotinidase Deficiency
Bjornstad syndrome/Leigh syndrome/Mitochondrial complex III deficiency, nuclear type 1
Blood group, ABO system
Bloom syndrome
Bothnia retinal dystrophy/Fundus albipunctatus/Retinitis punctata albescens
Brittle cornea syndrome 1
Canavan disease
Carbamoylphosphate synthetase I deficiency
Cardiomyopathy, dilated, 1GG
Carnitine palmitoyltransferase deficiency, hepatic, type IA

Disease Name
Carnitine palmitoyltransferase deficiency, hepatic, type II, infantile,lethal neonathal
Carnitine-acylcarnitine translocase deficiency
Cerebral creatine deficiency syndrome 1
Cerebral creatine deficiency syndrome 2
Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome
Cerebrotendinous xanthomatosis
Ceroid lipofuscinosis, neuronal, 2/Spinocerebellar ataxia, autosomal recessive 7
Ceroid lipofuscinosis, neuronal, 3
Ceroid lipofuscinosis, neuronal, 5
Cholestasis; progressive familial intrahepatic 2
Choreoacanthocytosis
Chronic granulomatous disease (cytochrome b-negative)
Chronic granulomatous disease (cytochrome b-positive, type 1)
Chronic granulomatous disease, X-linked
Ciliary dyskinesia, primary, 1, with or without situs inversus
Ciliary dyskinesia, primary, 16
Citrullinemia
Citrullinemia, adult-onset type II/Citrullinemia, type II, neonatal-onset
Clopidogrel, impaired responsiveness to/Mephenytoin poor metabolizer/Omeprazole poor metabolizer/Proguanil poor metabolizer
Cockayne syndrome
Cockayne, type A
Coenzyme Q10 deficiency, primary, 7
Combined malonic and methylmalonic aciduria
Complement factor H deficiency
Complex hereditary spastic paraparesis

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Disease Name
Congenital adrenal insufficiency with 46,XY sex reversal
Congenital amegakaryocytic thrombocyto-penia
Congenital arthrogryposis with anterior horn cell disease/Lethal congenital contracture syndrome 1
Congenital disorder of deglycosylation
Congenital disorder of glycosylation type Ia
Congenital disorder of glycosylation, type Ib
Congenital disorder of glycosylation, type Im
Cornelia de Lange like (Birk Flusser) syn-drome
Crigler-Najjar syndrome, type I/Cri-gler-Najjar syndrome, type II/Hyperbiliru-binemia, familial transient neonatal/Gilbert syndrome
Cystic fibrosis
Cystinosis, nephropathic
Cystinuria
D-bifunctional protein deficiency/Perrault syndrome 1
Deafness, autosomal recessive 1
Deafness, autosomal recessive 12
Deafness, autosomal recessive 16
Deafness, autosomal recessive 22
Deafness, autosomal recessive 3
Deafness, autosomal recessive 59
Deafness, autosomal recessive 7
Deafness, autosomal recessive 76
Deafness, autosomal recessive 77
Deafness, autosomal recessive 8/10
Desmosterolosis
Diabetes insipidus, nephrogenic
Diabetes insipidus, neurohypophyseal

Disease Name
Diaphanospondylodysostosis
Diarrhea 7, protein-losing enteropathy type
Dihydrolipoamide dehydrogenase deficiency
Disordered steroidogenesis due to cyto-chrome P450 oxidoreductase
Duchenne muscular dystrophy
Dysautonomia, familial
Dyserythropoietic anemia, congenital, type Ia
Dyserythropoietic anemia, congenital, type II
Dyskeratosis congenita, autosomal recessive 4 (TERT-related)
Dyskeratosis congenita, autosomal recessive 5 (RTEL1-related)
Dysprothrombinemia/Hypoprothrombinemia
Ectodermal dysplasia 1, hypohidrotic, X-linked
Ehlers-Danlos syndrome, type VII-C
Encephalopathy, neonatal severe/Mental retardation, X-linked syndromic, Lubs type/ Mental retardation, X-linked, syndromic 13
Enhanced S-cone syndrome
Epidermolysis bullosa, junctional, Herlitz type (LAMA3-related)
Epidermolysis bullosa, junctional, Herlitz type (LAMB3-related)
Epidermolysis bullosa, junctional, Herlitz type (LAMC2-related)
Epidermolysis bullosa; junctional; with pyloric atresia
Epilepsy, progressive myoclonic 1B
Epilepsy, progressive myoclonic 2A (Lafora)
Epilepsy, pyridoxine-dependent
Fabry disease
Factor VII deficiency
Fanconi anemia, complementation group A
Fanconi anemia, complementation group C

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Disease Name
Fanconi anemia, complementation group G
Fanconi-Bickel syndrome
Fatty liver, acute, of pregnancy/HELLP syndrome, maternal, of pregnancy
Fructose intolerance, hereditary
Fumarase deficiency
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 acidemia IIA
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/IIIb (Cori or Forbes disease)
Glycogen storage disease IV
Glycogen storage disease VII (Tarui disease)
GM1-gangliosidosis, type I/typeII/typeIII/Mucopolysaccharidosis type IVB (Morquio)
Gray platelet syndrome
Growth hormone deficiency, isolated, type IA (GH1-related)
Growth hormone deficiency, isolated, type IB (GHRHR - related)

Disease Name
Growth retardation, developmental delay, facial dysmorphism (GDFD)
Haim-Munk syndrome
Hemochromatosis, type 2A
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
Homocystinuria, B6-responsive and nonresponsive types
Homocystinuria, cblD type, variant 1/Methylmalonic aciduria and homocystinuria, cblD type
Hyperinsulinemic hypoglycemia, familial, 1
Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome
Hyperoxaluria, primary, type 1
Hyperphenylalaninemia, BH4-deficient, A
Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis
Hypoaldosteronism, congenital, due to CMO I deficiency/Hypoaldosteronism, congenital, due to CMO II deficiency
Hypomagnesemia 1, 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

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Disease Name
Inflammatory bowel disease 28; early onset; autosomal recessive
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 13
Leber congenital amaurosis 2
Leber congenital amaurosis 4
Leber congenital amaurosis 5
Leigh syndrome, due to COX deficiency
Leprechaunism
Lethal congenital contractural syndrome 2
Lethal congenital contractural syndrome 3
Lethal congenital contracture syndrome 4
Leukodystrophy and acquired microcephaly with or without dystonia
Leukodystrophy, hypomyelinating, 3
Leukodystrophy, hypomyelinating, 4
Lipoprotein lipase deficiency
Liver failure, transient infantile
Lysinuric protein intolerance
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

Disease Name
Medium-chain acyl-CoA dehydrogenase deficiency
Megalencephalic leukoencephalopathy with subcortical cysts
Megaloblastic anemia-1, Norwegian type (Imerslund-Gräsbeck syndrome)
Mental retardation; autosomal recessive 15
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 (NDUFAF5-related)
Mitochondrial complex I deficiency (NDUFS4-related)
Mitochondrial complex I deficiency (NDUFS6-related)
Mitochondrial complex I deficiency, nuclear type 20
Mitochondrial complex III deficiency, nuclear type 4
Mitochondrial DNA depletion syndrome 1 (MNGIE type)
Mitochondrial DNA depletion syndrome 2 (myopathic type)
Mitochondrial DNA depletion syndrome 3 (hepatocerebral type)
Mitochondrial DNA depletion syndrome 4A (Alpers type)/Mitochondrial DNA depletion syndrome 4B (MNGIE type)
Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)

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Disease Name
Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)
Mitochondrial myopathy and sideroblastic anemia 1
Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy
Molybdenum cofactor deficiency A
Mucolipidosis II alpha/beta/Mucolipidosis III alpha/beta
Mucolipidosis III gamma
Mucolipidosis IV
Mucopolysaccharidisis type IIIA (Sanfilippo A)
Mucopolysaccharidosis Ih
Mucopolysaccharidosis type IIIB (Sanfilippo B)
Mucopolysaccharidosis type IIIC (Sanfilippo C)/Retinitis pigmentosa 73
Mucopolysaccharidosis type IIID
Multiple congenital anomalies-hypotonia-seizures syndrome 1
Multiple sulfatase deficiency
Muscular dystrophy, limb-girdle, autosomal recessive 1
Muscular dystrophy, limb-girdle, autosomal recessive 3
Muscular dystrophy, limb-girdle, type 2B
Muscular dystrophy, limb-girdle, type 2C
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5
Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency
Myasthenic syndrome, congenital, 23, presynaptic/Combined D-2- and L-2-hydroxyglutaric aciduria
Myotonia congenita; recessive
Nemaline myopathy 2, autosomal recessive

Disease Name
Nephronophthisis 2, infantile
Nephrotic syndrome, type 1
Nephrotic syndrome, type 2
Neuronopathy, distal hereditary motor, type VI
Neutropenia, severe congenital 4, autosomal recessive
Niemann-Pick disease, type C1
Niemann-pick disease, type C2
Niemann-Pick disease, types A/B
Nijmegen breakage syndrome
Odontoonychodermal dysplasia/Schopf-Schulz-Passarge syndrome
Omenn syndrome / T- B- severe combined immunodeficiency
Omenn syndrome/Severe combined immunodeficiency, Athabascan type
Ornithine transcarbamylase deficiency
Osteogenesis imperfecta, type xiv
Osteopetrosis, autosomal recessive 1
Osteopetrosis, autosomal recessive 8
Otospondylomegaepiphyseal dysplasia
Pendred syndrome
Peroxisome biogenesis disorder 1A (Zellweger)
Peroxisome biogenesis disorder 4A (Zellweger)
Peroxisome biogenesis disorder 5A (Zellweger)
Peroxisome biogenesis disorder 9B/Rhizomelic chondrodysplasia punctata, type 1
Phenylalanine hydroxylase deficiency (including phenylketonuria)
Polycystic kidney disease, autosomal recessive
Polymicrogyria, bilateral frontoparietal
Pontocerebellar hypoplasia, type 1A

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Disease Name
Pontocerebellar hypoplasia, type 2D (Progressive cerebello-cerebral atrophy, type 2D)
Pontocerebellar hypoplasia, type 2E
Primary ciliary dyskinesia-12 (RSPH9-related)
Primary ciliary dyskinesia-9 (DNAI2-related)
Prolidase deficiency
Propionicacidemia
Proximal myopathy and ophthalmoplegia
Pycnodysostosis
Pyridoxamine 5›-phosphate oxidase deficiency
Pyruvate carboxylase deficiency
Renal tubular acidosis; proximal; with ocular abnormalities
Retinitis pigmentosa 12
Retinitis pigmentosa 14
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
Roberts syndrome/SC phocomelia syndrome
Sandhoff disease, infantile, juvenile, and adult forms
Schimke immunoosseous dysplasia
Severe combined immunodeficiency due to ADA deficiency
Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis
Sialic acid storage disorder, infantile

Disease Name
Sjogren-Larsson syndrome
Smith-Lemli-Opitz syndrome
Spastic paraparesis 49, autosomal recessive
Spastic paraplegia 53, autosomal recessive
Spinal Muscular Atrophy
Spondylometaepiphyseal dysplasia, short limb-hand type
Stargardt disease 1 including Cone-rod dystrophy 3
Striatonigral degeneration, infantile
Stuve-Wiedemann syndrome/ Schwartz-Jampel type 2 syndrome
Surfactant metabolism dysfunction, pulmonary, 3
Tay-Sachs disease
Thiamine-responsive megaloblastic anemia syndrome
Thyroid dyshormonogenesis 5
Trichohepatoenteric syndrome 1
Tumoral calcinosis, familial, hyperphosphatemic
Tumoral calcinosis, familial, normophosphatemic
Tyrosinemia, type I
Tyrosinemia, type III
Usher syndrome, type 1B
Usher syndrome, type 1C
Usher syndrome, type 1F
Usher syndrome, type 2A
Usher syndrome, type 3A
Ventricular tachycardia, catecholaminergic polymorphic, 2
Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
Vitamin D-dependent rickets, type I
Werner syndrome

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Disease Name
Wilson disease
Wiskott-Aldrich syndrome
Wolman disease
Woodhouse-Sakati syndrome
Xeroderma pigmentosum, group C
Xeroderma pigmentosum, group G/Cock-ayne syndrome
Xeroderma pigmentosum, variant type

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