FamilyFirst
סקר גנטי טרום הריון
By igentify

רשימת המחלות הנסקרות בבדיקה

FamilyFirst - בדיקת סקר גנטי טרום הריוני מורחב, הסוקרת כ-850 מוטציות העלולות לחולל 255 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
3-methylglutaconic aciduria, type III Also known as: Costeff syndrome
3-phosphoglycerate dehydrogenase deficiency
Abetalipoproteinemia
Achromatopsia, type 2
Acyl-CoA dehydrogenase, short-chain, deficiency of
Adams–Oliver syndrome
Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency
Adrenocorticotropic hormone deficiency
Adrenoleukodystrophy
Aicardi-Goutieres syndrome, type 5
Albinism, oculocutaneous, type IA
Alport syndrome
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 1
Bardet-Biedl syndrome 10
Bardet-Biedl syndrome 11
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
Mitochondrial respiratory chain complex II deficiency
Carnitine palmitoyltransferase deficiency, hepatic, type IA
Carnitine palmitoyltransferase deficiency, hepatic, type II, infantile,lethal neonathal
Carnitine-acylcarnitine translocase deficiency
Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome
Cerebrotendinous xanthomatosis Also known as: CTX
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

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Disease Name
Deafness, autosomal recessive 12
Deafness, autosomal recessive 16
Deafness, autosomal recessive 1A Also known as: Connexin 26
Deafness, autosomal recessive 1B Also known as: Connexin 30
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
Diaphanospondylodysostosis
Dihydrolipoamide dehydrogenase deficiency Also known as: Lipoamide dehydrogenase deficiency LDD
Duchenne muscular dystrophy
Dysautonomia, familial
Dyserythropoietic anemia, congenital, type Ia
Dyserythropoietic anemia, congenital, type II
Dyskeratosis congenita, autosomal recessive 4 (TERT-relate)
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, Herlitz type (LAMC2-related)
Epidermolysis bullosa, junctional, with pyloric atresia
Epilepsy, progressive myoclonic 1B

Disease Name
Factor VII deficiency
Familial Mediterranean fever, AR
Fanconi anemia, complementation group A
Fanconi anemia, complementation group C
Fanconi-Bickel syndrome
Fragile X
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 (Tarui disease)
Gray platelet syndrome
Growth hormone deficiency, isolated, type IA (GH1-related)
Growth hormone deficiency, isolated, type IB (GHRHR - related)
Growth retardation, developmental delay, facial dysmorphism (GDFD)
Haim-Munk syndrome
Hemolytic anemia, with or without immune-mediated polyneuropathy
Hermansky-Pudlak syndrome 3
Hermansky-Pudlak syndrome 6

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Disease Name
HMG-CoA lyase deficiency
Homocystinuria due to MTHFR deficiency
Hyperinsulinemic hypoglycemia, familial, 1
Hyperoxaluria, primary, type 1
Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis
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
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
Leber congenital amaurosis 5
Leigh syndrome, due to COX deficiency
Lethal congenital contractural syndrome 3
Lethal congenital contracture syndrome 2
Lethal congenital contracture syndrome 4
Leukodystrophy, hypomyelinating, 3
Leukodystrophy, hypomyelinating, 4
Liver failure, transient infantile
Mandibuloacral dysplasia

Disease Name
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 (NDUFAF5-related)
Mitochondrial complex I deficiency (NDUFS4-related)
Mitochondrial complex I deficiency (NDUFS6-related)
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 5 (encephalomyopathic with or without methylmalonic aciduria)
Mitochondrial myopathy and sideroblastic anemia 1
Molybdenum cofactor deficiency A
Mucolipidosis III gamma

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Disease Name
Mucolipidosis IV Also known as ML-4
Mucopolysaccharidisis type IIIA (Sanfilippo A)
Mucopolysaccharidosis Ih
Multiple congenital anomalies-hypotonia-seizures syndrome 1
Multiple sulfatase deficiency
Muscular dystrophy, limb-girdle, type 2B Also known as: lgmd2b
Muscular dystrophy, limb-girdle, type 2C
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4 Also known as: Walker Warburg
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5
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
Neuronopathy, distal hereditary motor, type VI
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 1
Osteopetrosis, autosomal recessive 8
Otospondylomegaepiphyseal dysplasia
Pendred syndrome
Peroxisome biogenesis disorder 1A (Zellweger)
Peroxisome biogenesis disorder 4A (Zellweger)

Disease Name
Peroxisome biogenesis disorder 5A (Zellweger)
Phenylalanine hydroxylase deficiency (including phenylketonuria)
Polycystic kidney disease, autosomal recessive
Polymicrogyria, bilateral frontoparietal
Pontocerebellar hypoplasia, type 1A
Pontocerebellar hypoplasia, type 2D (Progressive cerebello-cerebral atrophy, type 2D)
Pontocerebellar hypoplasia, type 2E Also known as: PCCA2
Primary ciliary dyskinesia-12 (RSPH9-related)
Primary ciliary dyskinesia-16 (DNAL1-related)
Primary ciliary dyskinesia-9 (DNAI2-related)
Prolidase deficiency
Propionicacidemia
Proximal myopathy and ophthalmoplegia
Pycnodysostosis
Pyridoxamine 5›-phosphate oxidase deficiency
Leber congenital amaurosis 8
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
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
Sickle cell anemia

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Disease Name
SMA
Smith-Lemli-Opitz syndrome Also knomn as: SLO syndrome
Spastic paraparesis 49, autosomal recessive
Spastic paraplegia 53, autosomal recessive
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
Thalassemia, alpha
Thiamine-responsive megaloblastic anemia syndrome
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
Wilson disease
Wiskott-Aldrich syndrome
Wolman disease
Woodhouse-Sakati syndrome
Xeroderma pigmentosum, group C
Xeroderma pigmentosum, group G/Cockayne syndrome
Xeroderma pigmentosum, variant type

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