MyScreen

NGS based carrier screening

MyScreen - בדיקת סקר גנטי מורחב - כי מגיע לך לדעת יותר!

מהי בדיקת MyScreen?

MyScreen הינה בדיקת סקר גנטי מורחב המקיפה והמותאמת ביותר לאוכלוסייה בישראל. הבדיקה מבוצעת במכונים הגנטיים בארץ בסטנדרטים מחמירים. מטרתה של הבדיקה להפחית את הסיכון להולדת ילד חולה. **הבדיקה כוללת בירור למעל 1800 מוטציות שכיחות ביותר מ-500 גנים למחלות תורשתיות מוכרות בקרב האוכלוסיה הישראלית מכלל העדות והמוצאים (יהודים ולא יהודים).**

בבדיקה מכוסות כל המוטציות למחלות בעדות ספציפיות אשר נכללו בסל הבריאות וכן אלו שאושרו ע"י האיגוד הגנטי וטרם הוכנסו לסל הבריאות (למעט בדיקות ה-Fragile X, DMD ו-SMA הכלולות בסל הבריאות ויבוצעו בשיטה אחרת). בנוסף מכילה הבדיקה בירור למוטציות רבות לפי המלצות המכונים הגנטיים אשר לא נכללות בסל הבריאות כיום או בבדיקות סקר המבוצעות בחו"ל. ניתן לקבל הסבר נוסף במכון הגנטי.

מהי בדיקת סקר גנטי?

בדיקת סקר גנטי מזהה האם הינך נשא/ית לאחת מהתסמונות הגנטיות הנכללות בבדיקה. בבדיקה כלולות בדר"כ מחלות רבות ובכל מחלה מגוון המוטציות השכיחות בישראל. המונח "נשאות" מתייחס למצב שבו אדם בריא לחלוטין נושא במטען הגנטי שלו שינוי/ליקוי גנטי ("מוטציה") באחד מהעותקים של גן מסויים בעוד העותק השני תקין. הסיכוי להיות נשא למחלה גנטית תורשתית כלשהי בקרב האוכלוסיה הישראלית מוערך ב-20%-40. רק כאשר שני בני הזוג נמצאו נשאים לאותה מחלה קיים סיכוי של 25% לילדית ילד חולה במחלה.

בישראל קיימת תוכנית סקר גנטי מצוינת ובדיקות רבות נכללות בסל הבריאות (כגון Tay Sachs, CF ועוד). עם זאת, תוכנית זו מכסה רק את המחלות השכיחות ביותר בהתאם לקריטריונים של שכיחות המחלה וחומרתה. דוגמא לכך היא מחלת ה-CF שבה נבדקות כיום רק 19 מוטציות בסל הבריאות מתוך מאות מוטציות אפשריות. בדיקת ה-MyScreen מכסה 236 מוטציות ידועות בגן ה-CF ובכך הסיכוי לאתר נשאים למחלה זו עולה. כמו כן מכסה הבדיקה מאות מוטציות למחלות נוספות שכלל לא כלולות כיום בסל הבריאות.

Gene	Disease	
TNNT1	Nemaline myopathy 5, Amish type	467
TPP1	Ceroid lipofuscinosis, neuronal, 2	468
TRAK1	Encephalopathy, fatal	469
TRAPPC9	Mental retardation, Autosomal Recessive 13	470
TRIM32	Bardet-Biedl syndrome 11	471
TRIOBP	Deafness, Autosomal Recessive 28	472
TRMT10A	Microcephaly, short stature	473
TRMU	LIFT, Liver failure infantile transient	474
TRPM1	Night blindness, 1C	475
TRPM6	Hypomagnesemia 1, intestinal	476
TSHR	Hypothyroidism, congenital, nongoitrous, 1	477
TSPAN12	Vitroretinal vascular malformations, congenital	478
TTN	Cardiomyopathy, dilated - Lethal Congenital Arthrogryposis	479
TUBGCP4	Microcephaly, primary, Autosomal Recessive	480
TULP1	Retinitis pigmentosa 14	481
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type)	482
TYR	Albinism, oculocutaneous, type IA (OCA1A)	483
UNC13D	Hemophagocytic lymphohistiocytosis, familial, 3	484
UNC80	Hypotonia infantile (HPFR2)	485
UPB1	Beta-ureidopropionase deficiency	486
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4	487
USH1C	Usher syndrome, type 1C	488
USH1G	Usher syndrome, type 1G	489
USH2A	Usher syndrome, type 2A	490
USMG5	Leigh syndrome realted to USMG5	491
VDR	Rickets, vitamin D-resistant, type IIA	492
VIPAS39	ARC syndrome 2	493
VPS11	Hypomyelination and developmental delay	494
VPS13A	Choreoacanthocytosis	495
VPS13B	Cohen syndrome	496
VPS33B	ARC syndrome	497
VPS37A	Spastic paraplegia 53, Autosomal Recessive	498
VPS45	Neutropenia, severe congenital, 5, Autosomal Recessive	499
VPS53	Pontocerebellar hypoplasia, type 2E (PCCA2)	500
VRK1	Pontocerebellar hypoplasia type 1A	501
WFS1	Wolfram-like syndrome, Autosomal Dominant	502
WISP3	Arthropathy, progressive pseudorheumatoid, of childhood	503
XPC	Xeroderma pigmentosum, group C	504
XRCC2	Fanconi Anemia	505
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome-2	506
ZMPSTE24	Mandibuloacral dysplasia with type B lipodystrophy	507
ZNF469	Brittle cornea syndrome 1	508

Gene	Disease	
SLC39A4	Acrodermatitis enteropathica	425
SLC45A2	Albinism, oculocutaneous, type IV	426
SLC46A1	Folate malabsorption, hereditary	427
SLC4A4	Renal tubular acidosis (RTA)	428
SLCO2A1	Hypertrophic osteoarthropathy	429
SMARCAL1	Schimke immunoosseous dysplasia	430
SMN1	Spinal muscular atrophy-1	431
SMPD1	Niemann-Pick disease type B, SMPD1-related	432
SNAP29	CEDNIK Syndrome	433
SNX10	Osteopetrosis, Autosomal Recessive 8	434
SPG11	Spastic paraplegia 11, Autosomal Recessive	435
SPINK5	Netherton syndrome	436
ST3GAL3	Early infantile epileptic encephalopathy 15	437
STRA6	Microphthalmia	438
STRC	Deafness, Autosomal Recessive 16	439
SUCLA2	Mitochondrial DNA depletion syndrome 5	440
SUMF1	Multiple sulfatase deficiency	441
SURF1	Leigh syndrome, due to COX deficiency	442
SYNE4	Deafness, Autosomal Recessive 76	443
SZT2	Epileptic encephalopathy, early infantile, 18	444
SepSecS	Pontocerebellar hypoplasia type 2D	445
TAF2	Mental retardation, Autosomal Recessive 40	446
TBCD	Infantile neurodegenerative disorder (PEBAT)	447
TBCE	Hypoparathyroidism retardation dysmorphism syndrome	448
TBX19	Adrenocorticotropic hormone deficiency	449
TCIRG1	Osteopetrosis, Autosomal Recessive 1	450
TCTN2	Meckel syndrome 8	451
TECPR2	Spastic paraplegia 49, Autosomal Recessive	452
TGM1	Ichthyosis, congenital, Autosomal Recessive 1	453
THG1L	Cerebellar ataxia and developmental delay	454
TIMM50	3-methylglutaconic aciduria, type IX	455
TK2	Mitochondrial DNA depletion syndrome 2 (myopathic type)	456
TKT	TRANSKETOLASE DEFICIENCY	457
TMC1	Deafness, Autosomal Recessive 7	458
TMEM165	Congenital disorder of glycosylation	459
TMEM216	Joubert syndrome 2 (MKS2)	460
TMEM231	Meckel syndrome 11	461
TMEM260	Neurodevelopmental, Cardiac, and Renal Syndrome	462
TMEM38B	Osteogenesis imperfecta, type XIV	463
TMEM67	Joubert syndrome type 6 (MSK3)	464
TMEM70	ATPase deficiency, nuclear encoded	465
TMPRSS3	Deafness, Autosomal Recessive 8/10	466

Gene	Disease	
PSMB8	Autoinflammation, lipodystrophy, and dermatosis syndrome	372
PTPN23	Developmental delay	373
PUS1	Mitochondrial myopathy and sideroblastic anemia 1	374
RAB27A	Griscelli syndrome, type 2	375
RAB28	Cone-rod dystrophy 18	376
RAG1	Severe combined immudeficiency, RAG1-related	377
RAG2	Severe combined immudeficiency, RAG2-related	378
RAPSN	Myasthenic syndrome (CMS4C)	379
RAPSN	Severe combined immudeficiency, RAG2-related	380
RARS2	Pontocerebellar hypoplasia, type 6	381
RDH12	Leber congenital amaurosis 13	382
RECQL2	Werner syndrome	383
RFX5	Bare lymphocyte syndrome, type II (SCID)	384
RIN2	Macrocephaly, alopecia, cutis laxa, and scoliosis	385
RNASEH2B	Aicardi-Goutieres syndrome 2	386
ROGDI	Kohlschutter-Tonz syndrome	387
RP1	Retinitis pigmentosa 1	388
RPE65	Leber congenital amaurosis 2	389
RPGRIP1	Cone-rod dystrophy 13	390
RPGRIP1L	Meckel syndrome 5	391
RRM2B	Mitochondrial DNA depletion syndrome 8	392
RSPH9	Ciliary dyskinesia, primary, 12	393
RTEL1	Dyskeratosis congenita	394
RYR1	Minicore myopathy with external ophthalmoplegia	395
SAMD9	Tumoral calcinosis, familial, normophosphatemic	396
SAMHD1	Aicardi Goutieres syndrome	397
SARS2	Hyperuricemia	398
SCAPER	Retinitis pigmentosa with intellectual disability	399
SCN9A	Insensitivity to pain, congenital, with anhidrosis (CIPA)	400
SCNN1A	Pseudohypoaldosteronism type I - SCNN1A gene	401
SCNN1B	Pseudohypoaldosteronism type I - SCNN1B gene	402
SDHA	Cardiomyopathy, dilated , 1GG neonatal isolated	403
SEC23B	Dyserythropoietic anemia, congenital, type II	404
SERAC1	3-methylglutaconic aciduria (MEGDEL)	405
SGCG	Muscular dystrophy, limb-girdle, type 2C	406
SGSH	Mucopolysaccharidisis type IIIA (Sanfilippo A)	407
SLC12A3	Bartter Syndrome, Gitelman Variant	408
SLC17A5	Sialic acid storage disorder, infantile (ISSD)	409
SLC18A3	Myasthenia gravis, congenital	410
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome	411
SLC1A4	Spastic tetraplegia	412
SLC22A5	Carnitine deficiency, systemic primary	413
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	414
SLC25A1	Combined D-2- and L-2-hydroxyglutaric aciduria	415
SLC25A20	Carnitine-acylcarnitine translocase deficiency - CACT	416
SLC26A3	Congenital chloride diarhhea (CLD)	417
SLC26A4	Pendred syndrome	418
SLC29A3	Histiocytosis-lymphadenopathy plus syndrome	419
SLC2A2	Fanconi-Bickel syndrome	420
SLC30A9	Birk-Landau-Perez cerebro-renal syndrome	421
SLC35A3	Arthrogryposis, mental retardation, and seizures	422
SLC35C1	Congenital disorder of glycosylation, type IIc	423
SLC37A4	Glycogen storage disease Ib	424

Gene	Disease	
NGLY1	Congenital disorder of deglycosylation	315
NNT	Glucocorticoid deficiency 4	316
NPC1	Niemann-Pick disease type C1	317
NPHP1	Joubert syndrome	318
NPHS1	Nephrotic syndrome type 1	319
NPHS2	Nephrotic syndrome	320
NRL	Retinitis pigmentosa 27	321
NTRK1	Insensitivity to pain, congenital, with anhidrosis (CIPA)	322
NUP62	Striatonigral degeneration (IBSN)	323
OAT	Gyrate atrophy	324
OCA2	Albinism, oculocutaneous, type II	325
OPA3	3-methylglutaconic aciduria, type III - Costeff	326
OTC	Ornithine transcarbamylase deficiency	327
OTOA	Deafness, Autosomal Recessive 22	328
OTOF	Deafness, Autosomal Recessive 9	329
P3H2	Myopia	330
PAH	Phenylketonuria	331
PARK2	Parkinson disease, early onset	332
PAX7	Myopathy, congenital, progressive, with scoliosis	333
PCCA	Propionic acidemia, PCCA-related	334
PCCB	Propionic acidemia, PCCB-related	335
PCDH12	Microcephaly, (MISSBC)	336
PCDH15	Usher syndrome, type 1F	337
PCK1	Phosphoenolpyruvate carboxykinase-1, cytosolic, deficiency	338
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II	339
PDE6A	Retinitis pigmentosa 43	340
PDE6B	Retinitis pigmentosa-40	341
PDE6G	Retinitis pigmentosa 57	342
PEPD	Prolidase deficiency	343
PEX1	Peroxisome biogenesis disorder 1A (Zellweger)	344
PEX2	Peroxisome biogenesis disorder 5A (Zellweger)	345
PEX6	Peroxisome biogenesis disorder 4B (Zellweger syndrome)	346
PEX7	Rhizomelic chondrodysplasia punctata type 1	347
PGAP3	Hyperphosphatasia with mental retardation syndrome 4	348
PGM1	Congenital disorder of glycosylation, type It	349
PHGDH	Phosphoglycerate dehydrogenase deficiency	350
PHKG2	Glycogen storage disease IXc	351
PHYH	Refsum disease	352
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1	353
PIGT	Multiple congenital anomalies-hypotonia-seizures syndrome 3	354
PIP5K1C	Lethal congenital contractural syndrome 3	355
PJVK	Deafness, Autosomal Recessive 59	356
PKHD1	Polycystic kidney & hepatic disease, PKHD1-related	357
PLA2G6	Infantile neuroaxonal dystrophy 1 (INAD)	358
PLAA	Neurodevelopmental disorder	359
PLEKHG2	Leukodystrophy and acquired microcephaly with or without dystonia	360
PMM2	Congenital disorder of glycosylation Ia	361
POC1A	Short stature, onychodysplasia	362
POMGNT2	Muscular dystrophy-dystroglycanopathy., type A, 8	363
POMT1	Walker-Warburg Syndrome, type A, 1	364
POMT2	Walker-Warburg Syndrome, type A, 2	365
POR	Antley-Bixler syndrome (ABS1)	366
PPIB	Osteogenesis imperfecta, type IX	367
PPP1R13L	Cardio-Cutaneous Syndrome DCM	368
PPT1	Ceroid lipofuscinosis, neuronal, 1	369
PRCD	Retinitis pigmentosa 36	370
PRICKLE1	Epilepsy, progressive myoclonic 1B	371

לפרטים נוספים והזמנת הבדיקה

יש לפנות למזכירות המכון הגנטי בבתי החולים הבאים:

בילינסון 03-9377659/8 | מאיר 09-7471059/962 | כרמל 04-9932777

בני ציון 04-8359851 | מעבדות זר 03-5247261

מידע נוסף על הבדיקה ניתן למצוא ב:

www.MyScreen.co.il

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Member of the gamida diagnostics division

עלות בדיקת MyScreen משתלמת יותר

עלות הבדיקה נמוכה בהשוואה לבדיקות שמציעות מעבדות פרטיות בארץ ובחו"ל. לבירור פרטים יש לפנות למכונים הגנטיים המבצעים את הבדיקה.

למי מומלץ לבצע את הבדיקה?

- זוגות המתכננים הריון ו/או הקמת משפחה בעתיד, כולל זוגות שכבר נבדקו בעבר לבדיקות גנטיות אחרות
- זוגות המצפים לתינוק
- זוגות בהם אחד מבני הזוג או שניהם נמצאו כנשאים של תסמונת גנטית
- בעלי היסטוריה משפחתית של תסמונת גנטית המעוניינים להעריך את גורמי הסיכון שלהם לפני תכנון משפחה.

מה משמעות תוצאות הבדיקה?

אם נמצא שאחד מבני הזוג הוא נשא של מוטציה למחלה ספציפית מאלו שנבדקות, והשני שנבדק במקביל לא נשא למוטציות שבפאנל, הסיכון הסטטיסטי להשנות אותה תסמונת ייחשב כנמוך. אם שני בני הזוג נמצאו נשאים לאותה תסמונת תופנו ליעוץ במכון הגנטי בו יינתן הסבר מקיף על האפשרויות העומדות בפניכם לצורך אבחון מדויק ואמין של המחלה בעובר (בבדיקת סיסי שיליה, דיקור מי שפיר או אבחון טרום השרשתי).

רשימת המחלות הנכללות בבדיקה:

Gene	Disease	
AN05	Limb-girdle muscular dystrophy	32
AP4B1	Spastic paraplegia 47, Autosomal Recessive	33
APTX	Ataxia (EAOH)	34
AQP2	Diabetes insipidus, nephrogenic	35
ARFGEF2	Periventricular heterotopia with microcephaly	36
ARHGDIA	Nephrotic syndrome, type 8	37
ARL6	Bardet-Biedl syndrome 3	38
ARSA	Metachromatic leukodystrophy - MLD	39
ARSG	Usher syndrome, type IV	40
ASL	Argininosuccinic aciduria	41
ASNS	Asparagine synthetase deficiency	42
ASPA	Canavan Disease	43
ASS1	Citrullinemia,classic	44
ATM	Ataxia-tyelangiectasia	45
ATP6V0A2	Cutis laxa, Autosomal Recessive, type IIA	46
ATP7B	Wilson disease	47
ATP8B1	Cholestasis, progressive familial intrahepatic 1	48
AVP	Familial neurohypophyseal diabetes insipidus	49
B3GALNT2	Muscular dystrophy-dystroglycanopathy , type A, 11	50
B4GALT1	Congenital disorder of glycosylation, type IId	51
BBS10	Bardet-Biedl syndrome 10	52
BBS1	Bardet-Biedl syndrome 1	53
BBS2	Bardet-Biedl syndrome 2	54
BBS4	Bardet-Biedl syndrome 4	55
BBS7	Bardet-Biedl syndrome 7	56
BBS9	Bardet-Biedl syndrome 9	57
BCKDHA	Maple syrup urine disease, type Ia	58
BCKDHB	Maple syrup urine disease, type Ib	59
BLM	Bloom syndrome	60
BMPER	Diaphanospondylodysostosis	61
BMPR1B	Brachydactyly type A2	62
BSND	Bartter syndrome, type 4a	63
BTD	Biotinidase deficiency	64
C12ORF65	Spastic paraplegia 55, Autosomal Recessive	65

Gene	Disease	
2p21	Hypotonia-cystinuria syndrome	1
ABCA12	Ichthyosis, congenital, Autosomal Recessive 4A	2
ABCA3	Congenital surfactant deficiency (SMDP3)	3
ABCA4	Cone-rod dystrophy 3	4
ABCB11	Cholestasis, progressive familial intrahepatic type 2	5
ABCC8	Hyperinsulinemic hypoglycemia, familial, 1 (PHHI/HHF1)	6
ABCD1	X-linked adrenoleukodystrophy	7
ABHD5	Chanarin-Dorfman syndrome	8
ACADM	Medium-chain Acyl-CoA dehydrogenase deficiency	9
ACADVL	Acyl-CoA dehydrogenase, very long-chain, VLCAD deficiency	10
ACO2	Infantile cerebellar-retinal degeneration	11
ACP5	Spondyloenchondrodysplasia with immune dysregulation	12
ACSF3	Combined malonic and methylmalonic aciduria	13
ADA	Severe combined immunodeficiency due to ADA deficiency	14
ADAM9	Cone-rod dystrophy 9	15
ADAMTS2	Ehlers Danlos syndrome, type VIIC	16
ADGRG1	Bilateral Frontoparietal Polymicrogyria (BFPP)	17
ADGRV1	Usher syndrome, type 2C	18
AGA	Aspartylglucosaminuria	19
AGL	Glycogen storage disease III	20
AGXT	Hyperoxaluria, primary, type I	21
AHI1	Joubert syndrome-3	22
AIMP1	Leukodystrophy, hypomyelinating, 3	23
AIPL1	Leber congenital amaurosis 4	24
AIRE	APS-1	25
ALDH1A3	Microphthalmia, isolated 8	26
ALDH7A1	Epilepsy, pyridoxine-dependent	27
ALDOB	Fructose intolerance	28
ALMS1	Alstrom syndrome	29
ALPL	Hypophosphatasia, infantile	30
AMT	Glycine encephalopathy, AMT-related	31

Gene	Disease	
JAK3	SCID, autosomal recessive, T-negative/B-positive type	256
KCNJ10	SESAME syndrome	257
KIAA1279	Goldberg-Shprintzen megacolon syndrome	258
KIF1C	Spastic ataxia 2, Autosomal Recessive	259
KIZ	Retinitis pigmentosa 69	260
KLHL40	Nemaline myopathy 8, Autosomal Recessive	261
KREMEN1	Ectodermal dysplasia	262
KRT14	Epidermolysis bullosa simplex	263
KY	Myopathy, myofibrillar, 7	264
LAMA2	Muscular dystrophy, LAMA2 deficiency	265
LAMA3	Laryngoonychocutaneous Syndrome	266
LAMB3	Epidermolysis bullosa, junctional, non-Herlitz type	267
LAMC2	Epidermolysis bullosa, junctional, Herlitz type	268
LCA5	Leber congenital amaurosis 5	269
LIFR	Stuve-Wiedemann syndrome	270
LIPA	Wolman disease	271
LOXHD1	Deafness, Autosomal Recessive 77	272
LRBA	Immunodeficiency, common variable, 8, with autoimmunity	273
MAK	Retinitis pigmentosa 62	274
MAN1B1	Mental retardation, Autosomal Recessive 15	275
MATN3	Spondyloepimetaphyseal dysplasia	276
MCIDAS	Mucociliary clearance disorder	277
MCOLN1	Mucolipidosis type IV - ML4	278
MECR	Dystonia (DYTOABG)	279
MED17	Microcephaly (ICCA)	280
MED25	Basel-Vanagaite-Smirin-Yosef syndrome	281
MEGF10	Myopathy	282
MERTK	Retinitis pigmentosa 38	283
MFSD8	Ceroid lipofuscinosis, neuronal, 7	284
MKS1	Meckel syndrome 1	285
MLC1	Megalencephalic leukoencephalopathy	286
MLPH	Griscelli syndrome, type 3	287
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	288
MOCS1	Molybdenum cofactor deficiency A	289
MOCS2	Molybdenum cofactor deficiency Type B	290
MPDU1	Congenital disorder of glycosylation, type If	291
MPL	Thrombocytopenia, congenital amegakaryocytic	292
MPV17	Mitochondrial DNA depletion syndrome 6	293
MRE11A	Ataxia Telangiectasia like disorder	294
MTHFR	Homocystinuria due to MTHFR deficiency	295
MTTP	Abetalipoproteinemia ABL	296
MUT	Methylmalonic acidemia, mut(0) type	297
MVK	Hyper-IgD syndrome	298
MYBPC1	Lethal congenital contracture syndrome 4	299
MYH2	Proximal myopathy and ophthalmoplegia	300
MYO15A	Deafness, Autosomal Recessive 3	301
MYO7A	Usher syndrome, type 1B	302
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B)	303
NARS2	Combined oxidative phosphorylation deficiency 24	304
NBEAL2	Gray platelet syndrome	305
NCF1	Chronic granulomatous disease due to deficiency of NCF-1	306
NCF2	Chronic granulomatous disease due to deficiency of NCF-2	307
NDUFA11	Mitochondrial complex I deficiency - NDUFA11 gene	308
NDUFAF5	Mitochondrial complex I deficiency - NDUFAF5 gene	309
NDUFS2	Mitochondrial complex I deficiency-NDUFS2 gene	310
NDUFS4	Leigh syndrome	311
NDUFS6	Mitochondrial complex I deficiency - NDUFS6 gene	312
NEB	Nemaline myopathy 2	313
NECTIN1	Zlotogora-Ogur syndrome	314

Gene	Disease	
FKRP	Muscular dystrophy-dystroglycanopathy, type C, 5	193
FKTN	Walker Warburg syndrome	194
FLT4	Autosomal Recessive Hereditary Lymphedema	195
FOXRED1	Mitochondrial encephalomyopathy complex I deficiency	196
FRMD4A	Microcephaly intellectual disability and dysmorphism	197
FTO	Growth retardation	198
G6PC3	Neutropenia, severe congenital 4, Autosomal Recessive	199
G6PC	Glycogen storage disease Ia - GDS1a	200
GAA	Pompe (Glycogen storage disease type II)	201
GALC	Krabbe disease	202
GALNT3	Tumoral calcinosis, hyperphosphatemic, familial	203
GALT	Galactosemia	204
GAN	Giant axonal neuropathy 1	205
GATC	Hypertophic Cardiomyopathy	206
GATM	Cerebral creatine deficiency syndrome 3	207
GBA	Gaucher disease, , type I	208
GCDH	Glutaricaciduria type I	209
GH1	Growth hormone deficiency, isolated, type IA	210
GHR	Laron dwarfism	211
GHRHR	Growth hormone deficiency, isolated, type IB	212
GIPC3	Deafness, autosomal recessive 15	213
GJB2	Deafness, autosomal recessive 1A	214
GJB6	Deafness, Autosomal Recessive 1B	215
GLB1	GM1-gangliosidosis, type I	216
GLDC	Glycine encephalopathy	217
GLRA1	Hyperekplexia	218
GMPPA	Alacrima, achalasia, and mental retardation syndrome	219
GMPPB	Muscular dystrophy-dystroglycanopathy	220
GNE	Hereditary inclusion body myopathy (HIBM)	221
GNPTAB	Mucolipidosis III alpha/beta	222
GNPTG	Mucolipidosis III gamma	223
GPC6	Omodysplasia 1	224
GPSM2	Chudley-McCullough syndrome	225
GRHPR	Hyperoxaluria, primary, type II	226
GUCY2D	Leber congenital amaurosis 1 , Cone-rod dystrophy 6	227
HACD1	Congenital myopathy	228
HADHA	Long-Chain hydroxylacyl-CoA dehydrogenase deficiency	229
HAX1	Kostmann disease	230
HBB	Hemoglobinopathies	231
HEXA	Tay-Sachs disease	232
HEXB	Sandhoff disease, infantile, juvenile, and adult forms	233
HGD	Alkaptonuria	234
HGSNAT	Retinitis pigmentosa 73	235
HIKESHI	Leukodystrophy, optic atrophy	236
HMGCL	HMG-CoA lyase deficiency	237
HOGA1	Hyperoxaluria, primary, type III	238
HPD	Thyrosinemia type III	239
HPS1	Hermansky-Pudlak syndrome 1	240
HPS3	Hermansky-Pudlak syndrome 3	241
HPS6	Hermansky-Pudlak syndrome 6	242
HSPD1	Leukodystrophy, hypomyelinating, 4, HLD4 (HSP60)	243
IBA57	Spastic paraplegia 74, Autosomal Recessive	244
IDUA	Mucopolysaccharidosis Type IH - Hurler syndrome	245
IGHMBP2	Neuronopathy, distal hereditary motor, type VI	246
IL10RA	Inflammatory bowel disease 28	247
INSR	Leprechaunism, Donohue syndrome	248
INVS	Nephronophthisis 2, infantile	249
ISPD	Muscular dystrophy-dystroglycanopathy (MDDGA7)	250
ITGA2B	Glanzmann thrombasthenia, ITGA2B-related	251
ITGB3	Glanzmann thrombasthenia, ITGB3-related	252
ITGB4	Carmi syndrome	253
ITK	Lymphoproliferative syndrome	254
IVD	Isovaleric academia	255

Gene	Disease	
CYP11A1	Adrenal insufficiency (P450scc)	130
CYP11B2	Hypoaldosteronism, congenital, due to CMO II deficiency	131
CYP1B1	Glaucoma 3A	132
CYP27A1	Cerebrotendinous xanthomatosis	133
CYP4F22	Congenital recessive ichthyoses (CRI)	134
CYP4V2	Bietti crystalline corneoretinal dystrophy	135
CYP7B1	Spastic paraplegia 5A, Autosomal Recessive	136
DAG1	Muscular dystrophy-dystroglycanopathy , type A, 9	137
DARS2	Leukoencephalopathy, LBSL	138
DBT	Maple syrup urine disease, type II	139
DCAF17	Woodhouse-Sakati syndome	140
DCLRE1C	Severe combined immunodeficiency, Athabascan type	141
DDR2	Spondylometaepiphyseal dysplasia, short limb-hand type	142
DDRGK1	Spondyloepimetaphyseal dysplasia (Shohat-type)	143
DDX11	Warsaw breakage syndrome	144
DGAT1	Diarrhea 7, congenital	145
DGUOK	Mitochondrial DNA depletion syndrome	146
DHCR24	Desmosterolosis	147
DHCR7	Smith Lemli Opitz syndrome	148
DHDDS	Retinitis pigmentosa 59	149
DLD	Dihydrolipoamide Dehydrogenase Deficiency	150
DLL3	Spondylocostal dysostosis 1, Autosomal Recessive	151
DNAH11	Ciliary dyskinesia, primary, 7	152
DNAH5	Ciliary dyskinesia (CILD3/PCD)	153
DNAI1	Ciliary dyskinesia, primary, 1	154
DNAI2	Ciliary dyskinesia, primary, 9	155
DNAL1	Ciliary dyskinesia, primary, 16	156
DOCK8	Hyper-IgE recurrent infection syndrome	157
DOLK	Congenital disorder of glycosylation, type Im	158
DSG1	Erythroderma (EPKHE)	159
DST	Epidermolysis bullosa simplex, Autosomal Recessive 2	160
DSTYK	Spastic paraplegia, complicated	161
DYSF	Muscular dystrophy, limb-girdle, type 2B	162
ECHS1	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	163
ECM1	Urbach-Wiethe disease	164
EDAR	Ectodermal dysplasia 10B	165
ELP1	Dysautonomia, familial	166
EOGT	Adams-Oliver syndrome 4	167
EPG5	Vici syndrome	168
EPM2A	Epilepsy, progressive myoclonic 2A (Lafora)	169
ERBB3	Lethal congenital contractural syndrome 2	170
ERCC2	Xeroderma pigmentosum, group D	171
ERCC5	Xeroderma pigmentosum/Cockayne	172
ERCC6	Cockayne syndrome, type B	173
ERCC8	Cockayne syndrome, type A	174
ESCO2	Roberts-SC phocomelia syndrome	175
ETFDH	Glutaric acidemia IIC	176
EXOSC3	Pontocerebellar hypoplasia, type 1B	177
EXOSC8	Pontocerebellar hypoplasia, type 1C	178
EYS	Retinitis pigmentosa 25	179
F7	Factor VII deficiency	180
FA2H	Spastic paraplegia 35, Autosomal Recessive	181
FAH	Tyrosinemia, type I	182
FAM161A	Retinitis pigmentosa 28	183
FAM20A	Amelogenesis imperfecta, type IG (enamel-renal syndrome)	184
FANCA	Fanconi anemia, complementation group A	185
FANCC	Fanconi anemia, complementation group C	186
FANCG	Fanconi Anemia - complementation group G	187
FDX1L	Mitochondrial muscle myopathy	188
FERMT1	Kindler syndrome	189
FGB	Afibrinogenemia congenital	190
FH	Fumarase deficiency, leiomyomatosis and renal cell cancer	191
FKBP10	Osteogenesis imperfecta, type XI	192

Gene	Disease	
C21orf59	Ciliary dyskinesia, primary, 26	66
C2ORF71	Retinitis pigmentosa 54	67
C8orf37	Retinitis pigmentosa 64	68
CAPN3	Muscular dystrophy, limb-girdle, type 2A	69
CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, 2	70
CBS	Homocystinuria, thrombosis, hyperhomocysteinemic	71
CC2D1A	Mental retardation, Autosomal Recessive 3 - MRT3	72
CC2D2A	Mental retardation, Autosomal Recessive 3 - MRT3	73
CCDC114	Ciliary dyskinesia, primary, 20	74
CCDC174	Birk Volodarsky PMR Synderome	75
CCDC65	Ciliary dyskinesia, primary, 27	76
CCDC88C	Hydrocephalus, nonsyndromic, Autosomal Recessive	77
CCNO	Ciliary dyskinesia, primary, 29	78
CD59	Hemolytic anemia	79
CDAN1	Dyserythropoietic anemia, congenital, type Ia	80
CDH23	Usher Syndrome Type ID	81
CDK10	Al Kaissi syndrome	82
CDK5	Lissencephaly 7 with cerebellar hypoplasia	83
CEACAM16	Deafness, autosomal recessive	84
CECR1	Adenosine deaminase 2 deficency	85
CECR1	Polyarteritis nodosa, childhood-onset	86
CENPJ	Microcephaly, primary, Autosomal Recessive	87
CEP104	Joubert syndrome (JBTS)	88
CEP152	Microcephaly 9, primary, Autosomal Recessive	89
CEP290	Meckel syndrome 4	90
CERKL	Retinitis pigmentosa 26	91
CFH	Hemolytic uremic syndrome, complement factor H deficiency	92
CFTR	Cystic fibrosis	93
CHRNE	Myasthenic syndrome, congenital, 4B, fast-channel	94
CLCN1	Myotonia congenita, Autosomal Recessive	95
CLCN1	Myotonia congenita,Autosomal Recessive	96
CLCN5	Proteinuria	97
CLCNKB	Bartter syndrome, type 3 and Gitelman syndrome	98
CLN5	Ceroid lipofuscinosis, neuronal, 5	99
CLN6	Ceroid lipofuscinosis, neuronal, 6	100
CLN8	Neuronal ceroid lipofuscinosis type 8	101
CLRN1	Usher syndrome, type 3A	102
CNGA1	Retinitis pigmentosa 49	103
CNGA3	Achromatopsia-2 - total color blindness	104
CNGB1	Retinitis pigmentosa 45	105
CNGB3	Achromatopsia-3,macular degeneration, juvenile	106
CNNM4	Jalili syndrome	107
CNTNAP1	Lethal congenital contracture syndrome 7	108
COL11A2	Otospondylomegaepiphyseal dysplasia (ZW)	109
COL17A1	Epidermolysis bullosa, junctional,	110
COL4A3	Alport Syndrome, COL4A3-Related	111
COL4A4	Alport syndrome, COL4A4-Related	112
COL4A5	Alport syndrome, COL4A5-Related	113
COL7A1	Dystrophic epidermolysis bullosa	114
COLEC11	3MC syndrome 2	115
COLQ	Myasthenic syndrome, congenital, 5	116
COQ4	Coenzyme Q10 deficiency, primary, 7	117
CPS1	Carbamoylphosphate synthetase I deficiency	118
CPT1A	Carnitine palmitoyltransferase 1 deficiency	119
CPT2	CPT deficiency, hepatic, type II	120
CRB1	Leber congenital amaurosis 8	121
CRB2	Ventriculomegaly with cystic kidney disease	122
CRTAP	Osteogenesis imperfecta, type VII	123
CSTA	Exfoliative ichthyosis (PSS4)	124
CTNS	Cystinosis,CTNS-related	125
CTSC	Haim-Munk syndrome	126
CTSK	Pycnodysostosis	127
CYBA	Chronic granulomatous disease	128
CYBB	Chronic granulomatous disease, X-linked	129