(2020-11-26) Ver 2 - Bedouin רשימת מחלות ומוטציות בפאנל
Hybrid Capture-Based Next Generation Sequencing

Mutation #	Gene: Disease Name {(Transcsript) "Mutation name"}
1	2p21:Hypotonia-cystinuria syndrome {"2p21"}
2 - 2	ABCA4:Cone-rod dystrophy 3 {(NM_000350.3) "c.834delT"}
3 - 3	ABCC8:Hyperinsulinemic hypoglycemia, familial, 1 (PHHI/HHF1) {(NM_000352.4) "c.2506C>T"}
4 - 4	ACADVL:Acyl-CoA dehydrogenase, very long-chain, VLCAD deficiency {(NM_000018.4) "c.65C>A"}
5 - 5	AIMP1:Leukodystrophy, hypomyelinating, 3 {(NM_004757.3) "c.292_293delCA"}
6 - 6	AQP2:Diabetes insipidus, nephrogenic {(NM_000486.5) "c.83T>C"}
7 - 7	ARL6:Bardet-Biedl syndrome 3 {(NM_032146.5) "c.364C>T"}
8 - 8	ATM:Ataxia-tyelangiectasia {(NM_000051.3) "c.7241_7244delAAGC"}
9 - 9	B4GALT1:Congenital disorder of glycosylation, type IId {(NM_001497) "c.61C>T"}
10 - 10	BBS2:Bardet-Biedl syndrome 2 {(NM_031885.4) "c.224T>G"}
11 - 12	BBS4:Bardet-Biedl syndrome 4 {(NM_033028.5) "c.77-1422_221-753del ", "c.884G>C"}
13 - 13	BCKDHA:Maple syrup urine disease, type Ia {(NM_000709.4) "c.859C>T"}
14 - 14	BSND:Bartter syndrome, type 4a infantile variant with sensorineuronal deafness {(NM_057176.3) "c.28G>A"}
15 - 15	CCDC174:Birk Volodarsky PMR Synderome Hypotonia and psychomotor developmental delay {(NM_016474.5) "c.1404A>G"}
16 - 16	CFH:Hemolytic uremic syndrome, complement factor H deficiency {(NM_000186.3) "c.3677_*4del"}
17 - 35	CFTR:Cystic fibrosis {(NM_000492.3) "c.1521_1523delCTT", "c.1585-1G>A", "c.1624G>T", "c.1647T>G", "c.2051_2052delAAinsG", "c.254G>A", "c.273+1G>A", "c.2988+1173_c.3468+2111del8898", "c.2989-1G>A", "c.3266G>A", "c.3276C>G", "c.3700A>G", "c.3718-2477C>T", "c.3846G>A", "c.3883_3886delATTT", "c.3909C>G", "c.54-5940_273+10250del21Kb", "c.761delA", "c.[1075C>A;1079C>A]"}
36 - 37	CLCN1:Myotonia congenita, Autosomal Recessive {(NM_000083) "c.1444G>A", "c.1586C>T"}
38 - 38	COL11A2:Otospondylomegaepiphyseal dysplasia (ZW) {(NM_080680.2) "c.3991C>T"}
39 - 39	CPS1:Carbamoylphosphate synthetase I deficiency {(NM_001875.5) "c.3374C>T"}
40 - 40	DHCR24:Desmosterolosis {(NM_014762.4) "c.307C>T"}
41 - 41	DLD:Dihydrolipoamide Dehydrogenase Deficiency {(NM_000108.5) "c.1436A>T"}
42 - 42	DOCK8:Hyper-IgE recurrent infection syndrome, autosomal recessive {(NM_203447) "c.5132C>A"}
43 - 43	EOGT:Adams-Oliver syndrome 4 {(NM_001278689.2) "c.1074delA"}
44 - 44	ERBB3:Lethal congenital contractural syndrome 2 {(NM_001982.3) "c.1184-9A>G"}

45 - 45	FRMD4A:Microcephaly intellectual disability and dysmorphism {(NM_018027) "c.2134_2146dup13"}
46 - 46	GCDH:Glutaricaciduria type I {(NM_000159.4) "c.505+1G>A"}
47 - 47	GH1:Growth hormone deficiency, isolated, type IA {(NM_000515.5) "c.456+5G>C"}
48 - 48	GJB2:Deafness, autosomal recessive 1A {(NM_004004.6) "c.35delG"}
49 - 49	GLDC:Glycine encephalopathy and non-ketoic hyperglycinemia, GLDC-related {(NM_000170.2) "c.2607C>A"}
50 - 50	GUCY2D:Leber congenital amaurosis 1 , Cone-rod dystrophy 6 {(NM_000180.3) "c.2129C>T"}
51 - 51	HEXA:Tay-Sachs disease {(NM_000520.5) "c.459+2dupT"}
52 - 52	IL10RA:Inflammatory bowel disease 28, early onset, autosomal recessive {(NM_001558) "c.537G>A"}
53 - 53	INVS:Nephronophthisis 2, infantile {(NM_014425.5) "c.2719C>T"}
54 - 54	ITGB4:Epidermolysis bullosa, junctional, with pyloric atresia - Carmi syndrome {(NM_000213.5) "c.3224_3793+120del"}
55 - 55	KRT14:Epidermolysis bullosa simplex {(NM_000526) "c.915G>A"}
56 - 57	LAMA2:Muscular dystrophy, congenital, due to partial LAMA2 deficiency {(NM_000426.3) "c.8665G>A", "c.8689C>T"}
58 - 58	MRE11A:Ataxia Telangiectasia like disorder {(NM_005591.3) "c.290A>G"}
59 - 59	MYBPC1:Lethal congenital contracture syndrome 4 {(NM_002465.4) "c.952C>T"}
60 - 60	NDUFA11:Mitochondrial complex I deficiency - NDUFA11 gene {(NM_001193375.1) "c.97+5G>A"}
61 - 61	NPC1:Niemann-Pick disease type C1 {(NM_000271.5) "c.1211G>A"}
62 - 62	NTRK1:Insensitivity to pain, congenital, with anhidrosis (CIPA) {(NM_002529.3) "c.1860_1861insT"}
63 - 63	NUP62:Striatonigral degeneration, Infantile Bilateral Striatal Necrosis (IBSN) {(NM_016553.4) "c.1172A>C"}
64 - 64	PEX6:Peroxisome biogenesis disorder 4B (Zellweger syndrome) {(NM_000287.4) "c.1947delG"}
65 - 65	PIP5K1C:Lethal congenital contractural syndrome 3 {(NM_012398.2) "c.757G>A"}
66 - 67	PKHD1:Polycystic kidney & hepatic disease, PKHD1-related {(NM_138694.4) "c.2279G>A", "c.6122-12G>A"}
68 - 68	PLA2G6:Infantile neuroaxonal dystrophy 1 (INAD) {(NM_003560.4) "c.2070_2072delTGT"}
69 - 69	POR:Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis {(NM_000941.3) "c.1615G>A"}
70 - 70	RFX5:Bare lymphocyte syndrome, type II (SCID) {(NM_000449) "c.715C>T"}
71 - 71	SCAPER:Retinitis pigmentosa with intellectual disability {(NM_020843) "c.2806delC"}
72 - 72	SCN9A:Insensitivity to pain, congenital, with anhidrosis (CIPA) {(NM_002977.3) "c.2687G>A"}
73 - 73	SDHA:Cardiomyopathy, dilated , 1GG neonatal isolated {(NM_004168.4) "c.1664G>A"}
74 - 74	SLC17A5:Sialic acid storage disorder, infantile (ISSD) {(NM_012434.5)

	"c.983G>A"}
75 - 75	SLC25A20:Carnitine-acylcarnitine translocase deficiency - CACT {(NM_000387.6) "c.713A>G"}
76 - 76	SLC30A9:Birk-Landau-Perez cerebro-renal syndrome {(NM_016474.5) "c.1047_1049delCAG"}
77 - 78	SLC37A4:Glycogen storage disease Ib {(NM_001164277.1) "c.1042_1043delCT", "c.83G>A"}
79 - 79	SLC4A4:Renal tubular acidosis (RTA), proximal, with ocular abnormalities and mental retardation {(NM_003759.3) "c.2321G>A"}
80 - 80	SMN1:Spinal muscular atrophy-1 {(NM_000344) "c.835_*3del"}
81 - 81	SUMF1:Multiple sulfatase deficiency {(NM_182760.3) "c.1043C>T"}
82 - 83	TBCE:Hypoparathyroidism retardation dysmorphism syndrome {(NM_003193.5) "c.155_166delGCCACGAAGGGA", "c.355_356del"}
84 - 84	TCIRG1:Osteopetrosis, Autosomal Recessive 1 {(NM_006019.4) "c.674delG"}
85 - 85	TMC1:Deafness, Autosomal Recessive 7 {(NM_138691.2) "c.100C>T"}
86 - 86	TMEM38B:Osteogenesis imperfecta, type XIV {(NM_018112) "c.455_542del"}
87 - 87	TRPM6:Hypomagnesemia 1, intestinal {(NM_017662.5) "c.2009+1G>A"}
88 - 88	UNC80:Hypotonia, infantile, with psychomotor retardation and characteristic facies 2 (HPFR2) {(NM_032504.1) "c.151C>T"}
89 - 89	UQCRQ:Mitochondrial complex III deficiency, nuclear type 4 {(NM_014402.5) "c.134C>T"}

*** לפי החלטת איגוד הגנטיקאים הישראלי, המוטציה מדווחת חיובית רק ליהודים ממוצא קווקזי De boer M, Gavrieli R, Van leeuwen K, et al. A false-carrier state for the c.579G>A mutation in the NCF1 gene in Ashkenazi Jews. J Med Genet. 2018;55(3):166-172.**

**** לפי החלטת איגוד הגנטיקאים הישראלי, המוטציה מדווחת חיובית רק לאנשים ממוצא דרוזי Belostotsky R, Seboun E, Idelson GH, et al. Mutations in DHDPSL are responsible for primary hyperoxaluria type III. Am J Hum Genet. 2010;87(3):392-399. doi:10.1016/j.ajhg.2010.07.023.**