(2020-11-21) 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.**

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