Az iGen Nifty-mono teszt által szűrt betegségek listája. A lista csak tájékoztató jellegű, kérjük, minden esetben egyeztessen klinikai genetikussal a szűrések megbízhatósága, a betegség észlelésének valószínűsége (detektációs ráta) és a magas, illetve alacsony kockázatú eredmények értelmezése kapcsán!

No.	Gene	Disease_EN	OMIM
1	ACTB	Baraitser-Winter syndrome 1	243310
2	ACTB	Dystonia- Deafness Syndrome 1	607371
3	ACTG1	Baraitser-Winter syndrome 2	614583
4	ACTG2	Visceral myopathy 1	155310
5	ACVR1	Fibrodysplasia Ossificans Progressiva	135100
6	ADNP	Autosomal Dominant Mental Retardation 28	615873
7	AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2	615937
8	ANKRD11	KBG Syndrome	148050
9	ARID1A	Autosomal Dominant Mental Retardation 14	614607
10	ARID1B	Coffin-Siris syndrome 1	135900
11	ASXL1	Bohring-Opitz Syndrome	605039
12	ASXL3	Bainbridge-Ropers Syndrome	615485
13	ATP1A2	Alternating Hemiplegia of Childhood 1	104290
14	ATP1A2	Developmental and epileptic encephalopathy 98	619605
15	ATP1A3	Developmental and epileptic encephalopathy 99	619606
16	ATP1A3	Alternating hemiplegia of childhood 2	614820
17	ATP1A3	CAPOS syndrome	601338
18	BCL11A	Dias-Logan syndrome	617101
19	BICD2	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant	618291
20	BRAF	Noonan Syndrome 7	613706
21	BRAF	Cardiofaciocutaneous Syndrome	115150
22	BRAF	LEOPARD syndrome 3	613707
23	CACNA1A	Developmental and epileptic encephalopathy 42	617106
24	CAMTA1	Cerebellar dysfunction with variable cognitive and behavioral abnormalities	614756
25	CASK	Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia	300749
26	CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia	613563
27	CDKL5	Developmental and epileptic encephalopathy 2	300672
28	CHD2	Developmental and epileptic encephalopathy 94	615369
29	CHD7	CHARGE syndrome	214800
30	CHD8	Intellectual developmental disorder with autism and macrocephaly	615032
31	COL11A1	Stickler syndrome, type II	604841
32	COL1A1	Osteogenesis Imperfecta type I	166200
33	COL1A1	Combined osteogenesis imperfecta and ehlers-danlos syndrome 1	619115
34	COL1A1/COL1A2	Osteogenesis Imperfecta type II	166210
35	COL1A1/COL1A2	Osteogenesis Imperfecta type III	259420
36	COL1A1/COL1A2	Osteogenesis Imperfecta type IV	166220
37	COL2A1	Stickler syndrome, type I	108300
38	COL2A1	Platyspondylic Lethal Skeletal dysplasia, Torrance type	151210
39	COL2A1	Achondrogenesis, type II or hypochondrogenesis	200610
40	COL2A1	SED congenita	183900
41	COL2A1	Atypical stickler syndrome, type I	609508
42	COL2A1	Kniest dysplasia	156550
43	COL4A1	Porencephaly 1	175780
44	COL9A2	Epiphyseal dysplasia, multiple, 2	600204
45	COL9A3	Epiphyseal dysplasia, multiple, 3, with or without myopathy	600969
46	COMP	Pseudoachondroplasia	177170

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No.	Gene	Disease_EN	OMIM
47	COMP	Epiphyseal dysplasia, multiple, 1	132400
48	CREBBP	Rubinstein-Taybi Syndrome 1	180849
49	CREBBP	Menke-Hennekam syndrome 1	618332
50	CTCF	Intellectual developmental disorder, autosomal dominant 21	615502
51	CTNNB1	Neurodevelopmental disorder with spastic diplegia and visual defects	615075
52	DNM1	Developmental and epileptic encephalopathy 31	616346
53	DYNC1H1	Intellectual developmental disorder, autosomal dominant 13	614563
54	DYRK1A	Intellectual developmental disorder, autosomal dominant 7	614104
55	EBP	Chondrodysplasia punctata, X-linked dominant	302960
56	EFNB1	Craniofrontonasal dysplasia	304110
57	EFTUD2	Mandibulofacial dysostosis, Guion-Almeida type	610536
58	EHMT1	Kleefstra Syndrome 1	610253
59	EP300	Rubinstein-Taybi Syndrome 2	613684
60	EP300	Menke-Hennekam syndrome 2	618333
61	ERF	Craniosynostosis 4	600775
62	ERF	Chitayat syndrome	617180
63	FBN1	Marfan Syndrome	154700
64	FGFR1	Trigonocephaly 1	190440
65	FGFR1/FGFR2	Jackson-Weiss Syndrome	123150
66	FGFR1/FGFR2	Pfeiffer syndrome	101600
67	FGFR2	Saethre-Chotzen Syndrome	101400
68	FGFR2	Bent Bone Dysplasia Syndrome	614592
69	FGFR2	Beare-Stevenson Cutis Gyrata syndrome	123790
70	FGFR2	Antley-Bixler Syndrome Without Genital Anomalies Or Disordered Steroidogenesis	207410
71	FGFR2	Crouzon syndrome	123500
72	FGFR2	Apert syndrome	101200
73	FGFR3	Thanatophoric Dysplasia, type II	187601
74	FGFR3	SADDAN	616482
75	FGFR3	Muenke Syndrome	602849
76	FGFR3	Crouzon syndrome with acanthosis nigricans	612247
77	FGFR3	Thanatophoric Dysplasia, type I	187600
78	FGFR3	Hypochondroplasia	146000
79	FGFR3	Achondroplasia	100800
80	FLNA	Otopalatodigital syndrome, type II	304120
81	FLNA	Periventricular nodular heterotopia 1	300049
82	FLNB	Atelosteogenesis, type I	108720
83	FLNB	Larsen Syndrome	150250
84	FLNB	Atelosteogenesis, type III	108721
85	FLNB	Boomerang dysplasia	112310
86	FOXG1	Rett syndrome, congenital variant	613454
87	FOXP1	Intellectual developmental disorder with language impairment with or without autistic features	613670
88	FREM1	Trigonocephaly 2	614485
89	GABRA1	Developmental and epileptic encephalopathy 19	615744
90	GABRB2	Developmental and epileptic encephalopathy 92	617829
91	GATAD2B	GAND syndrome	615074
92	GFAP	Alexander Disease	203450
93	GNAO1	Developmental and epileptic encephalopathy 17	615473
94	GNAO1	Neurodevelopmental disorder with involuntary movements	617493

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No.	Gene	Disease_EN	OMIM
95	GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant	614254
96	GRIN2B	Developmental and epileptic encephalopathy 27	616139
97	GRIN2B	Intellectual developmental disorder, autosomal dominant 6, with or without seizures	613970
98	HDAC8	Cornelia de Lange syndrome 5	300882
99	HNRNPK	Au-Kline Syndrome	616580
100	HNRNPU	Developmental and epileptic encephalopathy 54	617391
101	HRAS	Costello Syndrome	218040
102	IFITM5	Osteogenesis imperfecta, type V	610967
103	JAG1	Alagille Syndrome 1	118450
104	KANSL1	Koolen-De Vries Syndrome	610443
105	KAT6B	SBBYSS syndrome	603736
106	KAT6B	Genitopatellar syndrome	606170
107	KCNB1	Developmental and epileptic encephalopathy 26	616056
108	KCNJ2	Andersen syndrome	170390
109	KCNQ2	Developmental and epileptic encephalopathy 7	613720
110	KCNT1	Developmental and epileptic encephalopathy 14	614959
111	KCNT1	Epilepsy, nocturnal frontal lobe, 5	615005
112	KIF1A	NESCAV syndrome	614255
113	KIF1A	Spastic paraplegia 30a, autosomal dominant	610357
114	KMT2A	Wiedemann-Steiner syndrome	605130
115	KMT2D	Kabuki Syndrome 1	147920
116	KRAS	Cardiofaciocutaneous Syndrome 2	615278
117	KRAS	Noonan Syndrome 3	609942
118	LMNA	Muscular dystrophy, congenital	613205
119	LMNA	Hutchinson-Gilford Progeria Syndrome	176670
120	LZTR1	Noonan Syndrome 10	616564
121	MAP2K1	Cardiofaciocutaneous Syndrome 3	615279
122	MAP2K2	Cardiofaciocutaneous Syndrome 4	615280
123	MECP2	Rett syndrome	312750
124	MED13L	Impaired intellectual development and distinctive facial features with or without cardiac defects	616789
125	MEF2C	Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language	613443
126	MSX2	Craniosynostosis 2	604757
127	MSX2	Parietal Foramina With Cleidocranial Dysplasia	168550
128	NALCN	Congenital Contractures Of The Limbs And Face, Hypotonia, And Developmental delay	616266
129	NF1	Neurofibromatosis 1	162200
130	NF1	Neurofibromatosis-noonan syndrome	601321
131	NF1	Watson syndrome	193520
132	NF2	Neurofibromatosis 2	101000
133	NFIX	Marshall-Smith syndrome	602535
134	NFIX	Malan syndrome	614753
135	NIPBL	Cornelia de Lange syndrome 1	122470
136	NOTCH2	Hajdu-Cheney Syndrome	102500
137	NOTCH2	Alagille Syndrome 2	610205
138	NR2F1	Bosch-Boonstra-Schaaf Optic Atrophy Syndrome	615722
139	NRAS	Noonan syndrome 6	613224
140	NSD1	Sotos Syndrome 1	117550
141	NSDHL	CHILD syndrome	308050

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No.	Gene	Disease_EN	OMIM
142	PACS1	Schuurs-Hoeijmakers syndrome	615009
143	PIK3CA	Overgrowth syndrome and/or cerebral malformations	MONDO:0100283
144	PIK3R2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1	603387
145	PPP2R1A	Intellectual developmental disorder, autosomal dominant 36	616362
146	PPP2R5D	Intellectual developmental disorder, autosomal dominant 35	616355
147	PRKAR1A	Acrodysostosis 1, with or without Hormone Resistance	101800
148	PTPN11	Noonan Syndrome 1	163950
149	PTPN11	Leopard syndrome 1	151100
150	PURA	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties	616158
151	RAD21	Cornelia de Lange syndrome 4	614701
152	RAF1	Noonan Syndrome 5	611553
153	RERE	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart	616975
154	RIT1	Noonan Syndrome 8	615355
155	RPS6KA3	Coffin-Lowry Syndrome	303600
156	RUNX2	Metaphyseal Dysplasia with Maxillary Hypoplasia with or without Brachydactyly	156510
157	RUNX2	Cleidocranial dysplasia	119600
158	SATB2	Glass Syndrome	612313
159	SCN1A	Early Infantile Epileptic Encephalopathy 6	607208
160	SCN1A	Developmental and epileptic encephalopathy 6B, non-Dravet	619317
161	SCN2A	Developmental and epileptic encephalopathy 11	613721
162	SCN2A	Episodic ataxia, type 9	618924
163	SCN8A	Developmental and epileptic encephalopathy 13	614558
164	SCN8A	Cognitive Impairment With Or Without Cerebellar Ataxia	614306
165	SETBP1	Schinzel-Giedion syndrome	269150
166	SETBP1	Intellectual developmental disorder, autosomal dominant 29	616078
167	SETD2	Luscan-Lumish Syndrome	616831
168	SETD2	Intellectual developmental disorder, autosomal dominant 70	620157
169	SETD5	Intellectual developmental disorder, autosomal dominant 23	615761
170	SHANK3	Phelan-McDermid Syndrome	606232
171	SHOC2	Noonan-Like Syndrome with Loose Anagen Hair	607721
172	SKI	Shprintzen-Goldberg Syndrome	182212
173	SLC25A24	Fontaine Progeroid Syndrome	612289
174	SMAD3	Loeys-Dietz syndrome 3	613795
175	SMAD4	Myhre Syndrome	139210
176	SMARCA2	Nicolaides-Baraitser Syndrome	601358
177	SMARCA2	Blepharophimosis-impaired intellectual development syndrome	619293
178	SMARCA4	Autosomal Dominant Mental Retardation 16	614609
179	SMARCB1	Autosomal Dominant Mental Retardation 15	614608
180	SMARCE1	Coffin-Siris Syndrome 5	616938
181	SMC1A	Cornelia de Lange syndrome 2	300590
182	SMC1A	Developmental and epileptic encephalopathy 85 with or without midline brain defects	301044
183	SMC3	Cornelia de Lange syndrome 3	610759
184	SOS1	Noonan Syndrome 4	610733
185	SOS2	Noonan Syndrome 9	616559
186	SOX9	Campomelic Dysplasia	114290
187	SPECC1L	Opitz GBBB Syndrome, Type II	145420
188	SPTAN1	Developmental and epileptic encephalopathy 5	613477

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No.	Gene	Disease_EN	OMIM
189	SPTAN1	Developmental delay with or without epilepsy	620540
190	SRCAP	Floating-Harbor Syndrome	136140
191	SRCAP	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities	619595
192	STAT3	Hyper-IgE recurrent infection syndrome	147060
193	STXBP1	Developmental and epileptic encephalopathy 4	612164
194	SYNGAP1	Intellectual developmental disorder, autosomal dominant 5	612621
195	TBL1XR1	Pierpont Syndrome	602342
196	TBL1XR1	Intellectual developmental disorder, autosomal dominant 41	616944
197	TBX5	Holt-Oram Syndrome	142900
198	TCF4	Pitt-Hopkins syndrome	610954
199	TGFB2	Loeys-Dietz syndrome 4	614816
200	TGFBR1	Loeys-Dietz syndrome 1	609192
201	TGFBR2	Loeys-Dietz syndrome 2	610168
202	TRAF7	Cardiac, Facial, and Digital Anomalies with Developmental Delay	618164
203	TRPS1	Trichorhinophalangeal syndrome, type I	190350
204	TSC1	Tuberous Sclerosis-1	191100
205	TSC2	Tuberous sclerosis-2	613254
206	TUBA1A	Lissencephaly 3	611603
207	TUBB	Symmetric circumferential skin creases, congenital, 1	156610
208	TUBB	Cortical Dysplasia, Complex, with Other Brain Malformations 6	615771
209	TUBB2A	Cortical Dysplasia, Complex, with Other Brain Malformations 5	615763
210	TUBB4A	Leukodystrophy, hypomyelinating, 6	612438
211	TWIST1	Saethre-Chotzen syndrome with or without eyelid anomalies	101400
212	TWIST1	Craniosynostosis 1	123100
213	TWIST1	Sweeney-Cox syndrome	617746
214	TWIST1	Robinow-Sorauf Syndrome	180750
215	WDR45	Neurodegeneration with brain iron accumulation 5	300894
216	ZBTB20	Primrose syndrome	259050
217	ZC4H2	Wieacker-Wolff syndrome, female-restricted	301041
218	ZEB2	Mowat-Wilson syndrome	235730