GENEPLANET

NIFTY PREMIUM
Popis bolesti koje se otkrivaju testom

- Downov (trisomija 21), Edwardsov (trisomija 18) i Patauov (trisomija 13) sindrom
- Trisomije 9, 16 i 22
- Aneuploidije spolnih kromosoma (XO, XXX, XXY, XYY)
- 92 sindroma kromosomskih delecija/duplikacija (cijeli popis možete pronaći u nastavku)
- Sve ostale autosomne aneuploidije i delecije/duplikacije*
- 218 monogenskih bolesti (cijeli popis možete pronaći u nastavku)
- Podaci o spolu

*Ako pacijentica želi da se kao slučajni nalazi uključe i sve delecije ili duplikacije veće od 5 milijuna parova baza.

92 sindroma kromosomskih delecija/duplikacija (PRO)

Alagilov sindrom 1, Angelmanov sindrom, Duplikacija kromosoma 10p, Sindrom delecije kromosoma 10p12-p11, Sindrom delecije kromosoma 10q22.3-q23.2, Sindrom delecije kromosoma 10q26, Sindrom delecije kromosoma 11p11.2 (Potocki-Shafferov sindrom), Sindrom delecije kromosoma 11p13 (WAGR sindrom), Sindrom delecije kromosoma 11q23, Duplikacija kromosoma 12p, Sindrom mikrodelecije kromosoma 12p12.1, Sindrom mikrodelecije kromosoma 12q14, Sindrom delecije kromosoma 13q14, Duplikacija kromosoma 14q, Sindrom delecije kromosoma 14q11-q22, Sindrom delecije kromosoma 14q22 (Friasov sindrom), Sindrom delecije kromosoma 15q14, Sindrom mikrodelecije kromosoma 15q24, Sindrom prekomjernog rasta kromosoma 15q26, Sindrom delecije kromosoma 15q26-qter, Sindrom delecije kromosoma 16p12.2-p11.2, Sindrom duplikacije kromosoma 16p12.2-p11.2, Sindrom delecije kromosoma 16p13.3, Sindrom duplikacije kromosoma 16p13.3, Duplikacija kromosoma 17p, Sindrom delecije kromosoma 17p13.3, Sindrom duplikacije kromosoma 17p13.3, Sindrom delecije kromosoma 18p, Sindrom delecije kromosoma 1p32-p31, Sindrom delecije kromosoma 1p36, Sindrom delecije kromosoma 1q41-q42, Duplikacija kromosoma 20p, Delecija kromosoma 21q22, Sindrom delecije kromosoma 22q11.2 (DiGeorgeov sindrom), Sindrom delecije kromosoma 2p16.1-p15, Duplikacija kromosoma 2q, Sindrom duplikacije kromosoma 2q31.1, Sindrom mikrodelecije kromosoma 2q31.1, Sindrom delecije kromosoma 2q33.1 ("Glassov sindrom"), Sindrom delecije kromosoma 2q37, Sindrom delecije kromosoma 3pter-p25, Duplikacija kromosoma 3q, Sindrom delecije kromosoma 3q13.31, Duplikacija kromosoma 4p, Sindrom delecije kromosoma 4p16.3 (Wolf-Hirschhornov sindrom), Sindrom delecije kromosoma 4q21, Duplikacija kromosoma 5p, Sindrom duplikacije kromosoma 5p13, Sindrom delecije kromosoma 5q12, Sindrom delecije kromosoma 5q14.3, Delecija 6p kromosoma, Sindrom delecije kromosoma 6pter-p24, Sindrom delecije kromosoma 6q11-q14, Sindrom delecije kromosoma 6q15-q23, Sindrom delecije kromosoma 6q24-q25, Sindrom delecije kromosoma 6q25-qter, Sindrom delecije kromosoma 6q26-q27, Delecija kromosoma 7q, Sindrom delecije kromosoma 7q11.23, Delecija kromosoma 7q21-q32, Delecija kromosoma 7q31-q32, Duplikacija kromosoma 8p, Sindrom delecije kromosoma 8p23.1, Sindrom duplikacije kromosoma 8p23.1, Duplikacija kromosoma 8q, Sindrom delecije kromosoma 8q22.1, Sindrom duplikacije kromosoma 8q22.1, Sindrom delecije kromosoma 9p, Duplikacija kromosoma 9p, Sindrom duplikacije kromosoma Xp11.23-p11.22, Sindrom delecije kromosoma Xp21, Sindrom duplikacije kromosoma Xq21, Sindrom delecije kromosoma Xq22.3, Sindrom duplikacije kromosoma Xq27.3-q28, Sindrom Cri du Chat, Dandy-Walkerov sindrom, DiGeorgeov sindrom 2, Distalna delecija kromosoma 13q, Distalna delecija kromosoma 15q, Sindrom delecije distalnog kromosoma 18q, Duplikacija distalnog kromosoma 3p, Distalna delecija 4q kromosoma, Duplikacija distalnog kromosoma 4q, Jacobsenov sindrom, Langer-Giedionov sindrom, Levy-Shanske sindrom, Sindrom Potocki-Lupski, Prader-Willijev sindrom, Proksimalna delecija kromosoma 14q, Proksimalna duplikacija kromosoma 16q, Smith-Magenisov sindrom, Yuan-Harel-Lupski sindrom

218 monogenskih bolesti (MONO)

GEN	MONOGENSKA BOLEST	MONOGENIC CONDITION
ACTB	Baraitser-Winterov sindrom 1	Baraitser-Winter syndrome 1
ACTG1	Baraitser-Winterov sindrom 2	Baraitser-Winter syndrome 2
ACTG2	Visceralna miopatija 1	Visceral myopathy 1
ACVR1	Progresivna osificirajuća fibrodisplazija (FOP – Fibrodysplasia Ossificans Progressiva)	Fibrodysplasia Ossificans Progressiva
ADNP	Autosomno dominantna duševna zaostalost 28	Autosomal Dominant Mental Retardation 28
AKT3	Sindrom megalencefalije-polimikrogirije-polidaktilije-hidrocefalusa 2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2
ANKRD11	KBG sindrom	KBG Syndrome
ARID1A	Autosomno dominantna duševna zaostalost 14	Autosomal Dominant Mental Retardation 14
ARID1B	Coffin-Sirisov sindrom 1	Coffin-Siris syndrome 1
ASXL1	Bohring-Opitzov sindrom	Bohring-Opitz Syndrome
ASXL3	Bainbridge-Ropersov sindrom	Bainbridge-Ropers Syndrome
ATP1A2	Alternirajuća hemiplegija u dječjoj dobi 1	Alternating Hemiplegia of Childhood 1
ATP1A2	Razvojna i epileptična encefalopatija 98	Developmental and epileptic encephalopathy 98
ATP1A3	Razvojna i epileptična encefalopatija 99	Developmental and epileptic encephalopathy 99
BCL11A	Dias-Loganov sindrom	Dias-Logan syndrome
BICD2	"Spinalna mišićna atrofija, pretežito donjih ekstremiteta, 2B, autosomno dominantna"	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant
BRAF	Noonanov sindrom 7	Noonan Syndrome 7
BRAF	Kardiofaciokutani sindrom	Cardiofaciocutaneous Syndrome
BRAF	LEOPARD sindrom 3	LEOPARD syndrome 3
CACNA1A	Razvojna i epileptična encefalopatija 42	Developmental and epileptic encephalopathy 42
CAMTA1	Cerebelarna disfunkcija s promjenjivim kognitivnim poremećajima i poremećajima u ponašanju	Cerebellar dysfunction with variable cognitive and behavioral abnormalities
CASK	"Poremećaj intelektualnog razvoja i mikrocefalija s pontinskom i cerebelarnom hipoplazijom"	Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia
CBL	Poremećaj sličan Noonanovom sindromu, s juvenilnom mijelomonocitnom leukemijom ili bez nje	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia
CDKL5	Razvojna i epileptična encefalopatija 2	Developmental and epileptic encephalopathy 2
CHD2	Razvojna i epileptična encefalopatija 94	Developmental and epileptic encephalopathy 94
CHD7	Sindrom CHARGE	CHARGE syndrome
CHD8	Poremećaj intelektualnog razvoja s autizmom i makrocefalijom	Intellectual developmental disorder with autism and macrocephaly

COL11A1	Sticklerov sindrom, tip II.	Stickler syndrome, type II
COL1A1	Osteogenesis imperfecta, tip I.	Osteogenesis Imperfecta type I
COL1A1/COL1A2	Osteogenesis imperfecta, tip II.	Osteogenesis Imperfecta type II
COL1A1/COL1A2	Osteogenesis imperfecta, tip III.	Osteogenesis Imperfecta type III
COL1A1/COL1A2	Osteogenesis imperfecta, tip IV.	Osteogenesis Imperfecta type IV
COL2A1	Sticklerov sindrom, tip I.	Stickler syndrome, type I
COL2A1	Platispondilna letalna skeletna displazija, Torranceov tip	Platyspondylic Lethal Skeletal dysplasia, Torrance type
COL2A1	Ahondrogeneza (tip II.) ili hipohondrogeneza	Achondrogenesis, type II or hypochondrogenesis
COL2A1	Spondiloepimetafizna dysplasia congenita	SED congenita
COL4A1	Porencefalija 1	Porencephaly 1
COL9A2	Multipla epifizna displazija, 2	Epiphyseal dysplasia, multiple, 2
COL9A3	Multipla epifizna displazija, 3, s miopatijom ili bez nje	Epiphyseal dysplasia, multiple, 3, with or without myopathy
COMP	Pseudoahondroplazija	Pseudoachondroplasia
COMP	Multipla epifizna displazija, 1	Epiphyseal dysplasia, multiple, 1
CREBBP	Rubinstein-Taybi sindrom 1	Rubinstein-Taybi Syndrome 1
CREBBP	Menke-Hennekam sindrom 1	Menke-Hennekam syndrome 1
CTCF	Poremećaj intelektualnog razvoja, autosomno dominantan 21	Intellectual developmental disorder, autosomal dominant 21
CTNNB1	Neurorazvojni poremećaj sa spastičkom diplegijom i oštećenjima vida	Neurodevelopmental disorder with spastic diplegia and visual defects
DNM1	Razvojna i epileptična encefalopatija 31	Developmental and epileptic encephalopathy 31
DYNC1H1	Poremećaj intelektualnog razvoja, autosomno dominantan 13	Intellectual developmental disorder, autosomal dominant 13
DYRK1A	Poremećaj intelektualnog razvoja, autosomno dominantan 7	Intellectual developmental disorder, autosomal dominant 7
EBP	Chondrodysplasia punctata, X-vezana, dominantna	Chondrodysplasia punctata, X-linked dominant
EFNB1	Kraniofrontonazalna displazija	Craniofrontonasal dysplasia
EFTUD2	Mandibulofacijalna disostoza, tip Guion-Almeida	Mandibulofacial dysostosis, Guion-Almeida type
EHMT1	Kleefstra sindrom 1	Kleefstra Syndrome 1
EP300	Rubinstein-Taybi sindrom 2	Rubinstein-Taybi Syndrome 2
EP300	Menke-Hennekam sindrom 2	Menke-Hennekam syndrome 2
ERF	Kraniosinostoza 4	Craniosynostosis 4
ERF	Chitayatov sindrom	Chitayat syndrome
FBN1	Marfanov sindrom	Marfan Syndrome
FGFR1/FGFR2	Jackson-Weissov sindrom	Jackson-Weiss Syndrome
FGFR1	Trigonocefalija 1	Trigonocephaly 1
FGFR1/FGFR2	Pfeifferov sindrom	Pfeiffer syndrome
FGFR2	Sindrom Saethre-Chotzen	Saethre-Chotzen Syndrome
FGFR2	Sindrom displazije savijenih kostiju	Bent Bone Dysplasia Syndrome
FGFR2	Sindrom Beare-Stevenson Cutis Gyrata	Beare-Stevenson Cutis Gyrata syndrome
FGFR2	"Sindrom Antley-Bixler bez genitalnih anomalija ili poremećene steroidogeneze"	Antley-Bixler Syndrome Without Genital Anomalies Or Disordered Steroidogenesis
FGFR2	Crouzonov sindrom	Crouzon syndrome
FGFR2	Apertov sindrom	Apert syndrome
FGFR3	Tanatoforna displazija, tip II.	Thanatophoric Dysplasia, type II
FGFR3	SADDAN (teška ahondroplazija sa zastojem u razvoju i crnom akantozom / acanthosis nigricans)	SADDAN (severe achondroplasia with developmental delay and acanthosis nigricans)
FGFR3	Muenkeov sindrom	Muenke Syndrome
FGFR3	Crouzonov sindrom s crnom akantozom (acanthosis nigricans)	Crouzon syndrome with acanthosis nigricans
FGFR3	Tanatoforna displazija, tip I.	Thanatophoric Dysplasia, type I

FGFR3	Hipohondroplazija	Hypochondroplasia
FGFR3	Ahondroplazija	Achondroplasia
FLNA	Orofaciodigitalni sindrom, tip II.	Otopalatodigital syndrome, type II
FLNB	Atelosteogeneza, tip I.	Atelosteogenesis, type I
FLNB	Larsenov sindrom	Larsen Syndrome
FLNB	Atelosteogeneza, tip III.	Atelosteogenesis, type III
FLNB	Boomerang displazija	Boomerang dysplasia
FOXG1	Rettov sindrom, kongenitalna varijanta	Rett syndrome, congenital variant
FOXP1	Poremećaj intelektualnog razvoja s poremećajima u govoru i autističnim obilježjima ili bez njih	Intellectual developmental disorder with language impairment with or without autistic features
FREM1	Trigonocefalija 2	Trigonocephaly 2
GABRA1	Razvojna i epileptična encefalopatija 19	Developmental and epileptic encephalopathy 19
GABRB2	Razvojna i epileptična encefalopatija 92	Developmental and epileptic encephalopathy 92
GATAD2B	Sindrom GAND	GAND syndrome
GFAP	Alexanderova bolest	Alexander Disease
GNAO1	Razvojna i epileptična encefalopatija 17	Developmental and epileptic encephalopathy 17
GNAO1	Neurorazvojni poremećaj s nevoljnim pokretima	Neurodevelopmental disorder with involuntary movements
GRIN1	Neurorazvojni poremećaj s hiperkinezom i napadajima ili bez njih, autosomno dominantan	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant
GRIN2B	Razvojna i epileptična encefalopatija 27	Developmental and epileptic encephalopathy 27
GRIN2B	Poremećaj intelektualnog razvoja, autosomno dominantan 6, s napadajima ili bez njih	Intellectual developmental disorder, autosomal dominant 6, with or without seizures
HDAC8	Sindrom Cornelia de Lange 5	Cornelia de Lange syndrome 5
HNRNPK	Sindrom Au-Kline	Au-Kline Syndrome
HNRNPU	Razvojna i epileptična encefalopatija 54	Developmental and epileptic encephalopathy 54
HRAS	Costellov sindrom	Costello Syndrome
IFITM5	Osteogenesis imperfecta, tip V.	Osteogenesis imperfecta, type V
JAG1	Alagilleov sindrom 1	Alagille Syndrome 1
KANSL1	Koolen–De Vriesov sindrom	Koolen-De Vries Syndrome
KAT6B	Sindrom SBBYS	SBBYS syndrome
KAT6B	Genitopatelarni sindrom	Genitopatellar syndrome
KCNB1	Razvojna i epileptična encefalopatija 26	Developmental and epileptic encephalopathy 26
KCNJ2	Andersenov sindrom	Andersen syndrome
KCNQ2	Razvojna i epileptična encefalopatija 7	Developmental and epileptic encephalopathy 7
KCNT1	Razvojna i epileptična encefalopatija 14	Developmental and epileptic encephalopathy 14
KIF1A	Sindrom NESCAV	NESCAV syndrome
KMT2A	Wiedemann-Steinerov sindrom	Wiedemann-Steiner syndrome
KMT2D	Kabuki sindrom 1	Kabuki Syndrome 1
KRAS	Kardiofaciokutani sindrom 2	Cardiofaciocutaneous Syndrome 2
KRAS	Noonanov sindrom 3	Noonan Syndrome 3
LMNA	Kongenitalna mišićna distrofija	Muscular dystrophy, congenital
LMNA	Hutchinson-Gilfordov sindrom progerije	Hutchinson-Gilford Progeria Syndrome
LZTR1	Noonanov sindrom 10	Noonan Syndrome 10
MAP2K1	Kardiofaciokutani sindrom 3	Cardiofaciocutaneous Syndrome 3
MAP2K2	Kardiofaciokutani sindrom 4	Cardiofaciocutaneous Syndrome 4
MECP2	Rettov sindrom	Rett syndrome
MED13L	Kašnjenje u intelektualnom razvoju i karakteristične crte lica sa srčanim greškama ili bez njih	Impaired intellectual development and distinctive facial features with or without cardiac defects
MEF2C	Neurorazvojni poremećaj s hipotonijom, stereotipnim pokretima ruku i poremećajima u govoru	Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language

MSX2	Kraniosinostoza 2	Craniosynostosis 2
MSX2	Parijetalna foramina s kleidokranijalnom displazijom	Parietal Foramina With Cleidocranial Dysplasia
NALCN	Kongenitalne kontrakture u udovima i licu, hipotonija i zastoj u razvoju	Congenital Contractures Of The Limbs And Face, Hypotonia, And Developmental delay
NF1	Neurofibromatoza 1	Neurofibromatosis 1
NF2	Neurofibromatoza 2	Neurofibromatosis 2
NFIX	Marshall-Smithov sindrom	Marshall-Smith Syndrome
NIPBL	Sindrom Cornelia de Lange 1	Cornelia de Lange syndrome 1
NOTCH2	Sindrom Hajdu-Cheney	Hajdu-Cheney Syndrome
NOTCH2	Alagilleov sindrom 2	Alagille Syndrome 2
NR2F1	Sindrom optičke atrofije Bosch-Boonstra-Schaaf	Bosch-Boonstra-Schaaf Optic Atrophy Syndrome
NRAS	Noonanov sindrom 6	Noonan syndrome 6
NSD1	Sotosov sindrom 1	Sotos Syndrome 1
NSDHL	Sindrom CHILD	CHILD syndrome
PACS1	Schuurs-Hoeijmakersov sindrom	Schuurs-Hoeijmakers syndrome
PIK3R2	Sindrom megalencefalije-polimikrogirije-polidaktilije-hidrocefalusa 1	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1
PPP2R1A	Poremećaj intelektualnog razvoja, autosomno dominantan 36	Intellectual developmental disorder, autosomal dominant 36
PPP2R5D	Poremećaj intelektualnog razvoja, autosomno dominantan 35	Intellectual developmental disorder, autosomal dominant 35
PRKAR1A	Akrodizostoza 1, s hormonskom rezistencijom ili bez nje	Acrodysostosis 1, with or without Hormone Resistance
PTPN11	Noonanov sindrom 1	Noonan Syndrome 1
PURA	Neurorazvojni poremećaj s respiratornom insuficijencijom, hipotonijom i problemima s hranjenjem kod novorođenčadi	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties
RAD21	Sindrom Cornelia de Lange 4	Cornelia de Lange syndrome 4
RAF1	Noonanov sindrom 5	Noonan Syndrome 5
RERE	Neurorazvojni poremećaj s anomalijama mozga, oka ili srca ili bez njih	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart
RIT1	Noonanov sindrom 8	Noonan Syndrome 8
RPS6KA3	Coffin-Lowryjev sindrom	Coffin-Lowry Syndrome
RUNX2	Metafizealna displazija s maksilarnom hipoplazijom i brahidaktilijom ili bez nje	Metaphyseal Dysplasia with Maxillary Hypoplasia with or without Brachydactyly
RUNX2	Kleidokranijalna displazija	Cleidocranial dysplasia
SATB2	Glassov sindrom	Glass Syndrome
SCN1A	Rana infantilna epileptična encefalopatija 6	Early Infantile Epileptic Encephalopathy 6
SCN1A	Razvojna i epileptična encefalopatija 6B (nije Dravet sindrom)	Developmental and epileptic encephalopathy 6B, non-Dravet
SCN2A	Razvojna i epileptična encefalopatija 11	Developmental and epileptic encephalopathy 11
SCN2A	Epizodična ataksija, tip 9	Episodic ataxia, type 9
SCN8A	Razvojna i epileptična encefalopatija 13	Developmental and epileptic encephalopathy 13
SCN8A	Oštećenja kognitivnih funkcija s cerebelarnom ataksijom ili bez nje	Cognitive Impairment With Or Without Cerebellar Ataxia
SETBP1	Schinzel-Giedionov sindrom	Schinzel-Giedion syndrome
SETBP1	Poremećaj intelektualnog razvoja, autosomno dominantan 29	Intellectual developmental disorder, autosomal dominant 29
SETD2	Luscan-Lumishov sindrom	Luscan-Lumish Syndrome
SETD5	Poremećaj intelektualnog razvoja, autosomno dominantan 23	Intellectual developmental disorder, autosomal dominant 23
SHANK3	Phelan-McDermidov sindrom	Phelan-McDermid Syndrome
SHOC2	Poremećaj sličan Noonanovom sindromu s gubitkom anagene gustoće kose	Noonan-Like Syndrome with Loose Anagen Hair
SKI	Shprintzen-Goldbergov sindrom	Shprintzen-Goldberg Syndrome
SLC25A24	Fontaineov progeroidni sindrom	Fontaine Progeroid Syndrome
SMAD3	Loeys–Dietzov sindrom 3	Loeys-Dietz syndrome 3
SMAD4	Myhreov sindrom	Myhre Syndrome
SMARCA2	Nicolaides-Baraitserov sindrom	Nicolaides-Baraitser Syndrome

SMARCA2	Blefarofimoza sindrom – sindrom kašnjenja u intelektualnom razvoju	Blepharophimosis-impaired intellectual development syndrome
SMARCA4	Autosomno dominantna duševna zaostalost 16	Autosomal Dominant Mental Retardation 16
SMARCB1	Autosomno dominantna duševna zaostalost 15	Autosomal Dominant Mental Retardation 15
SMARCE1	Coffin-Sirisov sindrom 5	Coffin-Siris Syndrome 5
SMC1A	Sindrom Cornelia de Lange 2	Cornelia de Lange syndrome 2
SMC3	Sindrom Cornelia de Lange 3	Cornelia de Lange syndrome 3
SOS1	Noonanov sindrom 4	Noonan Syndrome 4
SOS2	Noonanov sindrom 9	Noonan Syndrome 9
SOX9	Kampomelna displazija	Campomelic Dysplasia
SPECC1L	Opitz GBBB sindrom, tip II.	Opitz GBBB Syndrome, Type II
SPTAN1	Razvojna i epileptična encefalopatija 5	Developmental and epileptic encephalopathy 5
SRCAP	Floating-Harbor sindrom	Floating-Harbor Syndrome
SRCAP	"Zastoj u razvoju, hipotonija, mišićno-skeletne anomalije i poremećaji u ponašanju"	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities
STAT3	Hiper-IgE sindrom	Hyper-IgE recurrent infection syndrome
STXBP1	Razvojna i epileptična encefalopatija 4	Developmental and epileptic encephalopathy 4
SYNGAP1	Poremećaj intelektualnog razvoja, autosomno dominantan 5	Intellectual developmental disorder, autosomal dominant 5
TBL1XR1	Pierpont sindrom	Pierpont Syndrome
TBL1XR1	Poremećaj intelektualnog razvoja, autosomno dominantan 41	Intellectual developmental disorder, autosomal dominant 41
TBX5	Holt-Oramov sindrom	Holt-Oram Syndrome
TCF4	Pitt-Hopkinsov sindrom	Pitt-Hopkins syndrome
TGFB2	Loeys–Dietzov sindrom 4	Loeys-Dietz syndrome 4
TGFBR1	Loeys–Dietzov sindrom 1	Loeys-Dietz syndrome 1
TGFBR2	Loeys–Dietzov sindrom 2	Loeys-Dietz syndrome 2
TRAF7	Anomalije srca, lica i prstiju sa zastojem u razvoju	Cardiac, Facial, and Digital Anomalies with Developmental Delay
TRPS1	Trihorinofalangealni sindrom, tip I.	Trichorhinophalangeal syndrome, type I
TSC1	Tuberozna skleroza 1	Tuberous Sclerosis-1
TSC2	Tuberozna skleroza 2	Tuberous Sclerosis-2
TUBA1A	Lisencefalija 3	Lissencephaly 3
TUBB	Simetrični obodni nabori kože, urođeni, 1	Symmetric circumferential skin creases, congenital, 1
TUBB	Složena kortikalna displazija s drugim malformacijama mozga, tip 6	Cortical Dysplasia, Complex, with Other Brain Malformations 6
TUBB2A	Složena kortikalna displazija s drugim malformacijama mozga, tip 5	Cortical Dysplasia, Complex, with Other Brain Malformations 5
TUBB4A	Hipomijelinizirajuća leukodistrofija, tip 6	Leukodystrophy, hypomyelinating, 6
TWIST1	Sindrom Saethre-Chotzen s anomalijama kapaka ili bez njih	Saethre-Chotzen syndrome with or without eyelid anomalies
TWIST1	Kraniosinostoza 1	Craniosynostosis 1
TWIST1	Sindrom Sweeney-Cox	Sweeney-Cox syndrome
TWIST1	Sindrom Robinow-Sorauf	Robinow-Sorauf Syndrome
WDR45	Neurodegeneracija s nakupljanjem željeza u mozgu, tip 5	Neurodegeneration with brain iron accumulation 5
ZBTB20	Primroseov sindrom	Primrose syndrome
ZC4H2	Wieacker-Wolffov sindrom (samo kod žena)	Wieacker-Wolff syndrome, female-restricted
ZEB2	Mowat-Wilsonov sindrom	Mowat-Wilson syndrome
ACTB	Sindrom distonije i gluhoće 1	Dystonia- Deafness Syndrome 1
ATP1A3	Alternirajuća hemiplegija djetinjstva 2	Alternating hemiplegia of childhood 2
ATP1A3	CAPOS sindrom	CAPOS syndrome
COL1A1	Kombinirana osteogenesis imperfecta i Ehlers-Danlos sindrom 1	Combined osteogenesis imperfecta and ehlers-danlos syndrome 1
COL2A1	Atipični sindrom ljepljive kosti, tip I	Atypical stickler syndrome, type I

COL2A1	Kniestova displazija	Kniest dysplasia
FLNA	Periventrikularna nodularna heterotopija 1	Periventricular nodular heterotopia 1
KCNT1	Epilepsija, noćni frontalni režanj, 5	Epilepsy, nocturnal frontal lobe, 5
KIF1A	Spastična paraplegija 30, autosomno dominantna	Spastic paraplegia 30, autosomal dominant
NF1	Neurofibromatoza-Noonan sindrom	Neurofibromatosis-noonan syndrome
NF1	Watsonov sindrom	Watson syndrome
NFIX	Malanov sindrom	Malan syndrome
PIK3CA	Sindrom prekomjernog rasta i/ili cerebralne malformacije	Overgrowth syndrome and/or cerebral malformations
PTPN11	Sindrom LEOPARD 1	Syndrome LEOPARD 1
SETD2	Poremećaj intelektualnog razvoja, autosomno dominantno 70	Intellectual developmental disorder, autosomal dominant 70
SMC1A	Razvojna i epileptička encefalopatija 85 sa ili bez defekata u središnjoj liniji mozga	Developmental and epileptic encephalopathy 85 with or without midline brain defects
SPTAN1	Zastoj u razvoju sa ili bez epilepsije	Developmental delay with or without epilepsy

NIFTY PREMIUM – STATISTIKA

SINDROM		OSJETLJIVOST	SPECIFIČNOST	PPV
Trisomija 21		99.17%	99.95%	92.19%
Trisomija 18		98.24%	99.95%	76.61%
Trisomija 13			99.96%	32.84%
CNV	**≥10 Mb**	>99.9%	99.97%	NA
	<10 Mb	>99.9%	99.86%	NA
Monogenske bolesti		>99%	>99%	NA
Spol fetusa		99,53%	99.20%	NA
ANEUPLOIDIJE SPOLNIH KROMOSOMA		**OSJETLJIVOST**	**PPV**	**NPV**
XYY		>99.9%	50.00%	>99.9%
XXY		>99.9%	42.86%	>99.9%
XXX		>99.9%	70.00%	>99.9%
XO		>99.9%	40.00%	>99.9%

Napomena: Podaci u tablici temelje se na povijesnoj literaturi i internim podacima te odražavaju samo prošle slučajeve otkrivanja, a ne stvarno stanje testiranog uzorka niti obećanu vrijednost.

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