GENEPLANET

NIFTY PREMIUM
Lista testiranih bolesti

- Daunov sindrom (trizomija 21), Edvardsov sindrom (trizomija 18), Patauov sindrom (trizomija 13)
- Trizomije 9, 16 i 22
- Aneuploidije polnih hromozoma (XO, XXX, XXY, XYY)
- 92 sindroma delecija/duplikacija (kompletnu listu možete pogledati ispod)
- Sve ostale autosomalne aneuploidije i delecije/duplikacije*
- 218 monogenskih bolesti (kompletnu listu možete pogledati ispod)
- Informacija o polu

*U slučaju da pacijentkinja odabere slučajne nalaze (delecije i duplikacije, veće od 5 M baznih parova).

92 sindroma delecija/duplikacija (PRO)

Alagilleov sindrom, Duplikacija 10p hromozoma, Sindrom delecije 10p12-p11 hromozoma, Sindrom delecije 11q23 hromozoma, Duplikacija 12p hromozoma, Sindrom mikrodelecije 12p12.1 hromozoma, Duplikacija 14q hromozoma, Sindrom mikrodelecije 15q24 hromozoma, Sindrom prekomerne rasti 15q26 hromozoma, Sindrom duplikacije 16p12.2-p11.2 hromozoma, Sindrom duplikacije 16p13.3 hromozoma, Duplikacija 17p hromozoma, Duplikacija 20p hromozoma, Delecija 21q22 hromozoma, Duplikacija 2q hromozoma, Sindrom mikrodelecije 2q31.1 hromozoma, Sindrom delecije 2q37 hromozoma, Duplikacija 3q hromozoma, Duplikacija 4p hromozoma, Duplikacija 5p hromozoma, Sindrom duplikacije 5p13 hromozoma , Delecija 6p hromozoma, Sindrom delecije 6q15-q23 hromozoma, Sindrom delecije 6q25-qter hromozoma, Sindrom delecije 6q26-q27 hromozoma, Delecija 7q21-q32 hromozoma, Delecija 7q31-q32 hromozoma, Duplikacija 8p hromozoma, Duplikacija 8q hromozoma, Duplikacija 9p hromozoma, Sindrom delecije Xq22.3 hromozom , Distalna delecija 13q hromozoma, Distalna delecija 15q hromozoma, Distalna duplikacija 3p hromozoma, Distalna delecija 4q hromozoma, Distalna duplikacija 4q hromozoma, Proksimalna delecija 14q hromozoma, Proksimalna duplikacija 16q hromozoma, Sindrom distalne delecije 18q hromozoma, Sindrom delecije 11p11.2 hromozoma (Potocki-Shafferin sindrom), Sindrom delecije 11p13 hromozoma (WAGR sindrom), Sindrom delecije 14q22 hromozoma (Friasov sindrom), Sindrom delecije 4p16.3 hromozoma (Wolf-Hirschhrnov sindrom), Angelman sindrom, Prader-Willijev sindrom, Sindrom delecije 10q22.3-q23.2 hromozoma, Sindrom delecije 10q26 hromozoma, Sindrom mikrodelecije 12q14 hromozoma, Sindrom delecije 13q14 hromozoma, Sindrom delecije 14q11-q22 hromozoma, Sindrom delecije 15q14 hromozoma, Sindrom delecije 15q26-qter hromozoma, Sindrom delecije 16p12.2-p11.2 hromozoma, Sindrom delecije 16p13.3 hromozoma, Sindrom delecije 17p13.3 hromozoma, Sindrom duplikacije 17p13.3 hromozoma, Sindrom delecije 18p hromozoma, Sindrom delecije 1p32-p31 hromozoma, Sindrom delecije 1p36 hromozoma, Sindrom delecije 1q41-q42 hromozoma, Sindrom delecije 22q11.2 hromozoma (DiGeorgeov sindrom), Sindrom delecije 2p16.1-p15 hromozoma, Sindrom duplikacije 2q31.1 hromozoma, Sindrom delecije 2q33.1 hromozoma, Sindrom delecije 3pter-p25 hromozoma, Sindrom delecije 3q13.31 hromozoma, Sindrom delecije 4q21 hromozoma, Sindrom delecije 5q12 hromozoma, Sindrom delecije 5q14.3 hromozoma, Sindrom delecije 6pter-p24 hromozoma, Sindrom delecije 6q11-q14 hromozoma, Sindrom delecije 6q24-q25 hromozoma, Delecija 7q hromozoma, Sindrom delecije 7q11.23 hromozoma, Sindrom delecije 8p23.1 hromozoma, Sindrom duplikacije 8p23.1 hromozoma, Sindrom delecije 8p22.1 hromozoma, Sindrom duplikacije 8q22.1 hromozoma, Sindrom delecije 9p hromozoma, Sindrom duplikacije Xp11.23-p11.22 hromozoma, Sindrom delecije Xp21 hromozoma, Sindrom delecije Xq21 hromozoma, Sindrom duplikacije Xq27.3-q28 hromozoma, Sindrom mačijeg plača (Sindrom Cri du Chat), Dandy-Walkerov sindrom, DiGeorgeov sindrom 2, Jacobsenov sindrom, Langer-Giedionov sindrom, Levy-Shanskeov sindrom, Potocki-Lupskijev sindrom, Smith-Magenisov sindrom, Yuan-Harel-Lupskijev sindrom.

218 monogenskih bolesti (MONO)

Rezultati MONO analize biće na engleskom jeziku, tako da ovde možete pogledati imena monogenskih bolesti kako na srpskom, tako i na engleskom jeziku.

GEN	MONOGENSKA BOLEST	MONOGENIC CONDITION
ACTB	Baraitser-Winterjev sindrom 1	Baraitser-Winter syndrome 1
ACTG1	Baraitser-Winterjev sindrom 2	Baraitser-Winter syndrome 2
ACTG2	Visceralna miopatija 1	Visceral myopathy 1
ACVR1	Progresivna osifikantna 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-Siris sindrom 1	Coffin-Siris syndrome 1
ASXL1	Bohring-Opitz sindrom	Bohring-Opitz Syndrome
ASXL3	Bainbridge-Ropers sindrom	Bainbridge-Ropers Syndrome
ATP1A2	Alternativna hemiplegija u detinjstvu 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-Logan sindrom	Dias-Logan syndrome
BICD2	Spinalna mišićna atrofija, dominantna za donje ekstremitete, 2B, autosomno dominantna	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant
BRAF	Noonanov sindrom 7	Noonan Syndrome 7
BRAF	Kardiofaciokutani sindrom (CFCS)	Cardiofaciocutaneous Syndrome
BRAF	LEOPARD sindrom 3	LEOPARD syndrome 3
CACNA1A	Razvojna i epileptična encefalopatija 42	Developmental and epileptic encephalopathy 42
CAMTA1	Disfunkcija mozga sa promenljivim kognitivnim i ponašajnim poremećajima	Cerebellar dysfunction with variable cognitive and behavioral abnormalities
CASK	Intelektualna ometanost povezana sa mikrocefalijom i pontinskom i cerebralnom hipoplazijom (MICPCH)	Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia
CBL	Poremećaj sličan Noonanovom sindromu sa ili bez juvenilne mijelomonocitne leukemije	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	CHARGE sindrom	CHARGE syndrome
CHD8	Intelektualna ometanost s autizmom i makrocefalijom	Intellectual developmental disorder with autism and macrocephaly
COL11A1	Sticklerov sindrom tip II	Stickler syndrome, type II
COL1A1	Osteogenesis Imperfecta (OI) tip I	Osteogenesis Imperfecta type I
COL1A1/COL1A2	Osteogenesis Imperfecta (OI) tip II	Osteogenesis Imperfecta type II
COL1A1/COL1A2	Osteogenesis Imperfecta (OI) tip III	Osteogenesis Imperfecta type III
COL1A1/COL1A2	Osteogenesis Imperfecta (OI) tip IV	Osteogenesis Imperfecta type IV
COL2A1	Sticklerov sindrom tip I	Stickler syndrome, type I
COL2A1	Platispondilna letalna skeletna displazija tip Torrance	Platyspondylic Lethal Skeletal dysplasia, Torrance type
COL2A1	Ahondrogeneza tip II ili hipohondrogeneza	Achondrogenesis, type II or hypochondrogenesis
COL2A1	SED congenita	SED congenita
COL4A1	Porencefalija 1	Porencephaly 1
COL9A2	Epifizna displazija, multipla, 2	Epiphyseal dysplasia, multiple, 2
COL9A3	Epifizna displazija, multipla, 3, sa ili bez miopatije	Epiphyseal dysplasia, multiple, 3, with or without myopathy
COMP	Pseudoahondroplazija	Pseudoachondroplasia
COMP	Epifizna displazija, multipla, 1	Epiphyseal dysplasia, multiple, 1
CREBBP	Rubinstein-Taybi sindrom 1	Rubinstein-Taybi Syndrome 1
CREBBP	Menke-Hennekam sindrom 1	Menke-Hennekam syndrome 1
CTCF	Duševna zaostalost, autosomno dominantna 21	Intellectual developmental disorder, autosomal dominant 21
CTNNB1	Neurološko razvojni poremećaj sa spastičkom diplegijom i oštećenjima vida (NEDSDV)	Neurodevelopmental disorder with spastic diplegia and visual defects
DNM1	Razvojna i epileptična encefalopatija 31	Developmental and epileptic encephalopathy 31
DYNC1H1	Duševna zaostalost, autosomno dominantna 13	Intellectual developmental disorder, autosomal dominant 13
DYRK1A	Duševna zaostalost, autosomno dominantna 7	Intellectual developmental disorder, autosomal dominant 7
EBP	Hondrodisplazija punktata, vezana za X, dominantna	Chondrodysplasia punctata, X-linked dominant
EFNB1	Kraniofrontonazalna displazija	Craniofrontonasal dysplasia
EFTUD2	Mandibulofacijalna disostoza tip Guion-Almeida (MFDGA)	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	Chitayat sindrom	Chitayat syndrome
FBN1	Marfanov sindrom	Marfan Syndrome
FGFR1/FGFR2	Jackson-Weiss sindrom	Jackson-Weiss Syndrome
FGFR1	Trigonocefalija 1	Trigonocephaly 1
FGFR1/FGFR2	Pfeifferov sindrom	Pfeiffer syndrome
FGFR2	Saethre-Chotzen sindrom	Saethre-Chotzen Syndrome
FGFR2	Sindrom displazije savijenih kostiju	Bent Bone Dysplasia Syndrome
FGFR2	Beare-Stevenson Cutis Gyrata sindrom	Beare-Stevenson Cutis Gyrata syndrome
FGFR2	Antley-Bixler sindrom 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	Tanatoforična displazija tip II	Thanatophoric Dysplasia, type II
FGFR3	SADDAN (teška ahondroplazija sa zastojem u razvoju i acanthosis nigricans (tamni, zadebljali delovi kože u kožnim naborima i pregibima))	SADDAN (severe achondroplasia with developmental delay and acanthosis nigricans)
FGFR3	Muenke sindrom	Muenke Syndrome

FGFR3	Crouzonov sindrom sa acanthosis nigricans (tamni, zadebljali delovi kože u kožnim naborima i pregibima)	Crouzon syndrome with acanthosis nigricans
FGFR3	Tanatoforična displazija tip I	Thanatophoric Dysplasia, type I
FGFR3	Hipohondroplazija	Hypochondroplasia
FGFR3	Ahondroplazija	Achondroplasia
FLNA	Otopalatodigitalni 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	Bumerangova displazija	Boomerang dysplasia
FOXG1	Rettov sindrom, kongenitalna varijanta	Rett syndrome, congenital variant
FOXP1	Intelektualna ometanost sa poremećajima govora i sa ili bez autističnih karakteristika	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 (neurološko razvojni poremećaj povezan sa GATAD2B)	GAND syndrome
GFAP	Alexanderova bolest	Alexander Disease
GNAO1	Razvojna i epileptična encefalopatija 17	Developmental and epileptic encephalopathy 17
GNAO1	Neurološko razvojni poremećaj sa nevoljnim pokretima	Neurodevelopmental disorder with involuntary movements
GRIN1	Neurološko razvojni poremećaj sa ili bez hiperkinezom i napadima, autosomno dominantna	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant
GRIN2B	Razvojna i epileptična encefalopatija 27	Developmental and epileptic encephalopathy 27
GRIN2B	Mentalna zaostalost, autosomno dominantna 6, sa napadima ili bez njih	Intellectual developmental disorder, autosomal dominant 6, with or without seizures
HDAC8	Cornelia de Lange sindrom 5	Cornelia de Lange syndrome 5
HNRNPK	Au-Kline sindrom	Au-Kline Syndrome
HNRNPU	Razvojna i epileptična encefalopatija 54	Developmental and epileptic encephalopathy 54
HRAS	Costello sindrom	Costello Syndrome
IFITM5	Osteogenesis imperfecta tip V	Osteogenesis imperfecta, type V
JAG1	Alagille sindrom 1	Alagille Syndrome 1
KANSL1	Koolen-De Vries sindrom	Koolen-De Vries Syndrome
KAT6B	SBBYS sindrom (Say-Barber-Biesecker-Young-Simpson sindrom)	SBBYS syndrome
KAT6B	Genitopatilarni 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	NESCAV sindrom	NESCAV syndrome
KMT2A	Wiedemann-Steiner sindrom	Wiedemann-Steiner syndrome
KMT2D	Kabuki sindrom 1	Kabuki Syndrome 1
KRAS	Kardiofaciokutani sindrom 2 (CFC2)	Cardiofaciocutaneous Syndrome 2
KRAS	Noonanov sindrom 3	Noonan Syndrome 3
LMNA	Kongenitalna mišićna distrofija	Muscular dystrophy, congenital
LMNA	Hutchinson-Gilfordov sindrom progerije (HGPS)	Hutchinson-Gilford Progeria Syndrome
LZTR1	Noonanov sindrom 10	Noonan Syndrome 10
MAP2K1	Kardiofaciokutani sindrom 3 (CFC3)	Cardiofaciocutaneous Syndrome 3
MAP2K2	Kardiofaciokutani sindrom 4 (CFC4)	Cardiofaciocutaneous Syndrome 4
MECP2	Rettov sindrom	Rett syndrome

MED13L	Oslabljen intelektualni razvoj i karakteristične crte lica sa ili bez srčanih defekata	Impaired intellectual development and distinctive facial features with or without cardiac defects
MEF2C	Neurološko razvojni poremećaj sa hipotoničnim pokretima ruku i poremećajem govora	Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language
MSX2	Kraniosinostoza 2	Craniosynostosis 2
MSX2	Parijetalna foramina sa kleidokranijalnom displazijom (PFMCCD)	Parietal Foramina With Cleidocranial Dysplasia
NALCN	Kongenitalne kontrakture udova i lica, hipotonia i zastoj u razvoju (CLIFAHDD)	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	Cornelia de Lange sindrom 1	Cornelia de Lange syndrome 1
NOTCH2	Hajdu-Cheney sindrom	Hajdu-Cheney Syndrome
NOTCH2	Alagille 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	CHILD sindrom	CHILD syndrome
PACS1	Schuurs-Hoeijmakers sindrom	Schuurs-Hoeijmakers syndrome
PIK3R2	Sindrom megalencefalije-polimikrogirije-polidaktilije-hidrocefalusa 1	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1
PPP2R1A	intelektualni razvojni poremećaj, autosomno dominantna 36	Intellectual developmental disorder, autosomal dominant 36
PPP2R5D	intelektualni razvojni poremećaj, autosomno dominantna 35	Intellectual developmental disorder, autosomal dominant 35
PRKAR1A	Akrodisostoza 1, sa ili bez hormonske rezistencije	Acrodysostosis 1, with or without Hormone Resistance
PTPN11	Noonanov sindrom 1	Noonan Syndrome 1
PURA	Neurološko razvojni poremećaj sa respiratornim distresom novorođenčeta, hipotonia i problemi sa hranjenjem	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties
RAD21	Cornelia de Lange sindrom 4	Cornelia de Lange syndrome 4
RAF1	Noonanov sindrom 5	Noonan Syndrome 5
RERE	Neurološko razvojni poremećaj sa ili bez anomalija mozga, očiju ili srca	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart
RIT1	Noonanov sindrom 8	Noonan Syndrome 8
RPS6KA3	Coffin-Lowry sindrom	Coffin-Lowry Syndrome
RUNX2	Metafizička displazija sa maksilarna hipoplazija i sa ili bez brahidaktilije	Metaphyseal Dysplasia with Maxillary Hypoplasia with or without Brachydactyly
RUNX2	Kleidokranijalna displazija	Cleidocranial dysplasia
SATB2	Glass sindrom	Glass Syndrome
SCN1A	Rana infantilna epileptična encefalopatija 6	Early Infantile Epileptic Encephalopathy 6
SCN1A	Razvojna i epileptična encefalopatija 6B, ne Dravetov sindrom	Developmental and epileptic encephalopathy 6B, non-Dravet
SCN2A	Razvojna i epileptična encefalopatija 11	Developmental and epileptic encephalopathy 11
SCN2A	Epizodična ataksija tipa 9	Episodic ataxia, type 9
SCN8A	Razvojna i epileptična encefalopatija 13	Developmental and epileptic encephalopathy 13
SCN8A	Kognitivna motnja s ili bez cerebelarne ataksije	Cognitive Impairment With Or Without Cerebellar Ataxia
SETBP1	Schinzel-Giedionov sindrom	Schinzel-Giedion syndrome
SETBP1	Intelektualna razvojna motnja, autosomno dominantna 29	Intellectual developmental disorder, autosomal dominant 29
SETD2	Luscan-Lumish sindrom	Luscan-Lumish Syndrome
SETD5	Intelektualna razvojna motnja, autosomno dominantna 23	Intellectual developmental disorder, autosomal dominant 23
SHANK3	Phelan-McDermid sindrom	Phelan-McDermid Syndrome
SHOC2	Noonanov sindrom s gubitkom anagene kose	Noonan-Like Syndrome with Loose Anagen Hair
SKI	Shprintzen-Goldbergov sindrom	Shprintzen-Goldberg Syndrome
SLC25A24	Fontainov progeroidni sindrom	Fontaine Progeroid Syndrome
SMAD3	Loeys-Dietz sindrom 3	Loeys-Dietz syndrome 3

SMAD4	Myhre sindrom	Myhre Syndrome
SMARCA2	Nicolaides-Baraitser sindrom	Nicolaides-Baraitser Syndrome
SMARCA2	Blefarofimoza sindrom - sindrom intelektualnog zaostajanja	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-Siris sindrom 5	Coffin-Siris Syndrome 5
SMC1A	Cornelia de Lange sindrom 2	Cornelia de Lange syndrome 2
SMC3	Cornelia de Lange sindrom 3	Cornelia de Lange syndrome 3
SOS1	Noonanov sindrom 4	Noonan Syndrome 4
SOS2	Noonanov sindrom 9	Noonan Syndrome 9
SOX9	Kampomelična displazija	Campomelic Dysplasia
SPECC1L	Opitz GBBB sindrom tipa II	Opitz GBBB Syndrome, Type II
SPTAN1	Razvojna i epileptična encefalopatija 5	Developmental and epileptic encephalopathy 5
SRCAP	Sindrom plavajućeg pristaništa	Floating-Harbor Syndrome
SRCAP	Zaostatak u razvoju, hipotonija, mišićno-skeletne anomalije i poremećaji ponašanja	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	Intelektualna razvojna motnja, autosomno dominantna 5	Intellectual developmental disorder, autosomal dominant 5
TBL1XR1	Pierpont sindrom	Pierpont Syndrome
TBL1XR1	Intelektualna razvojna motnja, autosomno dominantna 41	Intellectual developmental disorder, autosomal dominant 41
TBX5	Holt-Oramov sindrom	Holt-Oram Syndrome
TCF4	Pitt-Hopkinsov sindrom	Pitt-Hopkins syndrome
TGFB2	Loeys-Dietz sindrom 4	Loeys-Dietz syndrome 4
TGFBR1	Loeys-Dietz sindrom 1	Loeys-Dietz syndrome 1
TGFBR2	Loeys-Dietz sindrom 2	Loeys-Dietz syndrome 2
TRAF7	Anomalije srca, prstiju i lica s zaostatkom u razvoju	Cardiac, Facial, and Digital Anomalies with Developmental Delay
TRPS1	Trihorinofalangealni sindrom tipa 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	Prirođene simetrične obodne kožne gube 1	Symmetric circumferential skin creases, congenital, 1
TUBB	Kompleksna kortikalna displazija s drugim moždanim malformacijama tipa 6	Cortical Dysplasia, Complex, with Other Brain Malformations 6
TUBB2A	Kompleksna kortikalna displazija s drugim moždanim malformacijama tipa 5	Cortical Dysplasia, Complex, with Other Brain Malformations 5
TUBB4A	Hipomijelinizacijska leukodistrofija tipa 6	Leukodystrophy, hypomyelinating, 6
TWIST1	Saethre-Chotzenov sindrom s ili bez anomalija kapaka	Saethre-Chotzen syndrome with or without eyelid anomalies
TWIST1	Kraniosinostoza 1	Craniosynostosis 1
TWIST1	Sweeney-Cox sindrom	Sweeney-Cox syndrome
TWIST1	Robinow-Sorauf sindrom	Robinow-Sorauf Syndrome
WDR45	Nevrodegeneracija s nakupljanjem željeza u mozgu tipa 5	Neurodegeneration with brain iron accumulation 5
ZBTB20	Primrose sindrom	Primrose syndrome
ZC4H2	Wieacker-Wolffov sindrom (samo kod žena)	Wieacker-Wolff syndrome, female-restricted
ZEB2	Mowat-Wilsonov sindrom	Mowat-Wilson syndrome
ACTB	Distonija sindrom gluhoće tip 1	Dystonia- Deafness Syndrome 1
ATP1A3	Alternirajuća hemiplegija dečjeg doba tip 2	Alternating hemiplegia of childhood 2
ATP1A3	CAPOS sindrom	CAPOS syndrome

COL1A1	Kombinovani sindrom osteogeneze imperfekte i Ehlers–Danlosovog sindroma tip 1 (OI/EDS tip 1)	Combined osteogenesis imperfecta and ehlers-danlos syndrome 1
COL2A1	Atipični Stiklerov sindrom, tip I	Atypical stickler syndrome, type I
COL2A1	Knistova displazija	Kniest dysplasia
FLNA	Periventrikularna nodularna heterotopija 1	Periventricular nodular heterotopia 1
KCNT1	Epilepsija noćnog frontalnog režnja tip 5	Epilepsy, nocturnal frontal lobe, 5
KIF1A	Autozomno dominantna spastična paraplegija tip 30	Spastic paraplegia 30, autosomal dominant
NF1	Nunanov sindrom (varijanta neurofibromatoze tip 1)	Neurofibromatosis-noonan syndrome
NF1	Vatsonov sindrom	Watson syndrome
NFIX	Malanov sindrom	Malan syndrome
PIK3CA	Sindrom prekomernog rasta i/ili cerebralne malformacije	Overgrowth syndrome and/or cerebral malformations
PTPN11	LEOPARD sindrom 1	Syndrome LEOPARD 1
SETD2	Poremećaj intelektualnog razvoja, autozomno dominantni tip 70	Intellectual developmental disorder, autosomal dominant 70
SMC1A	Razvojna i epileptična encefalopatija tip 85 sa ili bez defekta srednje linije mozga	Developmental and epileptic encephalopathy 85 with or without midline brain defects
SPTAN1	Razvojno kašnjenje sa ili bez epilepsije	Developmental delay with or without epilepsy

NIFTY PREMIUM STATISTIKA

SINDROM		OSJETLJIVOST	SPECIFIČNOST	PPV
Trizomija 21		99.17%	99.95%	92.19%
Trizomija 18		98.24%	99.95%	76.61%
Trizomija 13		>99.9%	99.96%	32.84%
CNV	**≥10 Mb**	>99.9%	99.97%	NA
	<10 Mb	>99.9%	99.86%	NA
Monogenske bolesti		>99%	>99%	NA
Određivanje pola		99,53%	99.20%	NA
ANEUPLOIDIJE POLNIH HROMOZOMA		**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 tabeli baziraju se na postojećoj literaturi i internim podacima te odražavaju samo prethodno zapažanje, a ne stvarno stanje uzorka koji je testiran ili navedene vrednosti.

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