GENEPLANET

NIFTY PREMIUM
Seznam testiranih bolezni

- Downov sindrom (trisomija 21), Edwardsov sindrom (trisomija 18) in Patauov sindrom (trisomija 13)
- Trisomije 9, 16, in 22
- Anevploidije spolnih kromosomov (XO, XXX, XXY, XYY)
- 92 delecijsko/duplikacijskih sindromov (popoln seznam si lahko ogledate spodaj)
- Vse ostale avtosomalne anevploidije in delecije/duplikacije*
- 218 monogenskih bolezni (popoln seznam si lahko ogledate spodaj)
- Informacija o spolu

*V primeru, da pacientka izbere naključne najdbe (delecije in duplikacije, večje od 5 M baznih parov).

92 delecijsko/duplikacijskih sindromov (PRO)

Alagillov sindrom 1, Duplikacija 10p, Sindrom delecije 10p12-p11, Sindrom delecije 11q23, Duplikacija 12p, Sindrom mikrodelecije 12p12.1, Duplikacija 14q, Sindrom mikrodelecije 15q24, Sindrom prekomerne rasti 15q26, Sindrom duplikacije 16p12.2-p11.2, Sindrom duplikacije 16p13.3, Duplikacija 17p, Duplikacija 20p, Delecija 21q22, Duplikacija 2q, Sindrom mikrodelecije 2q31.1, Sindrom delecije 2q37, Duplikacija 3q, Duplikacija 4p, Duplikacija 5p, Sindrom duplikacije 5p13, Delecija 6p, Sindrom delecije 6q15-q23, Sindrom delecije 6q25-qter, Sindrom delecije 6q26-q27, Delecija 7q21-q32, Delecija 7q31-q32, Duplikacija 8p, Duplikacija 8q, Duplikacija 9p, Sindrom delecije Xq22.3, Distalna delecija 13q, Distalna delecija 15q, Distalna duplikacija 3p, Distalna delecija 4q, Distalna duplikacija 4q, Proksimalna delecija 14q, Proksimalna duplikacija 16q, Sindrom distalne delecije 18q, Sindrom delecije 11p11.2 (Potocki-Shafferjev sindrom), Sindrom delecije 11p13 (Sindrom WAGR), Sindrom delecije 14q22 (Friasov sindrom), Sindrom delecije 4p16.3 (Sindrom Wolf- Hirschhorn), Angelmanov sindrom, Prader-Willijev sindrom, Sindrom delecije 10q22.3-q23.2, Sindrom delecije 10q26, Sindrom mikrodelecije 12q14, Sindrom delecije 13q14, Sindrom delecije 14q11-q22, Sindrom delecije 15q14, Sindrom delecije 15q26-qter, Sindrom delecije 16p12.2-p11.2, Sindrom delecije 16p13.3, Sindrom delecije 17p13.3, Sindrom duplikacije 17p13.3, Sindrom delecije 18p, Sindrom delecije 1p32-p31, Sindrom delecije 1p36, Sindrom delecije 1q41-q42, Sindrom delecije 22q11.2 (DiGeorgejev sindrom), Sindrom delecije 2p16.1-p15, Sindrom duplikacije 2q31.1, Sindrom delecije 2q33.1 ("Glass syndrome"), Sindrom delecije 3pter-p25, Sindrom delecije 3q13.31, Sindrom delecije 4q21, Sindrom delecije 5q12, Sindrom delecije 5q14.3, Sindrom delecije 6pter-p24, Sindrom delecije 6q11-q14, Sindrom delecije 6q24-q25, Delecija 7q, Sindrom delecije 7q11.23, Sindrom delecije 8p23.1, Sindrom duplikacije 8p23.1, Sindrom delecije 8q22.1, Sindrom duplikacije 8q22.1, Sindrom delecije 9p, Sindrom duplikacije Xp11.23-p11.22, Sindrom delecije Xp21, Sindrom delecije Xq21, Sindrom duplikacije Xq27.3-q28, Cri du Chat sindrom, Dandy-Walkerjev sindrom (DWS), DiGeorgejev sindrom tipa 2 (DGS2), Sindrom Jacobsenove, Langer-Giedion sindrom (LGS), Sindrom Levy-Shanske, Potocki-Lupski sindrom (sindrom duplikacije 17p11.2), Sindrom Smith-Magenis, Sindrom Yuan-Harel-Lupski (YUHAL)

218 monogenskih bolezni (MONO)

GEN	MONOGENSKA BOLEZEN	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	Progresivni okosteneli miozitis (fibrodisplazija ali FOP-Fibrodysplasia Ossificans Progressiva)	Fibrodysplasia Ossificans Progressiva
ADNP	Avtosomno 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	Avtosomno 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 v otroštvu 1	Alternating Hemiplegia of Childhood 1
ATP1A2	Razvojna in epileptična encefalopatija 98	Developmental and epileptic encephalopathy 98
ATP1A3	Razvojna in epileptična encefalopatija 99	Developmental and epileptic encephalopathy 99
BCL11A	Dias-Logan sindrom	Dias-Logan syndrome
BICD2	Spinalna mišična atrofija, prevladujoča za spodnje okončine, 2B, avtosomno 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 in epileptična encefalopatija 42	Developmental and epileptic encephalopathy 42
CAMTA1	Disfunkcija možganov s spremenljivimi kognitivnimi in vedenjskimi motnjami	Cerebellar dysfunction with variable cognitive and behavioral abnormalities
CASK	Intelektualna prizadetost, povezana z mikrocefalijo s pontinsko in cerebralno hipoplazijo (MICPCH)	Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia
CBL	Noonanovemu sindromu podobna motnja z ali brez juvenilne mielomonocitne levkemije	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia
CDKL5	Razvojna in epileptična encefalopatija 2	Developmental and epileptic encephalopathy 2
CHD2	Razvojna in epileptična encefalopatija 94	Developmental and epileptic encephalopathy 94
CHD7	CHARGE sindrom	CHARGE syndrome
CHD8	Intelektualna razvojna motnja z avtizmom in makrocefalijo	Intellectual developmental disorder with autism and macrocephaly
COL11A1	Sticklerjev sindrom tipa II	Stickler syndrome, type II
COL1A1	Osteogenesis Imperfecta (OI) tipa I	Osteogenesis Imperfecta type I

COL1A1/COL1A2	Osteogenesis Imperfecta (OI) tipa II	Osteogenesis Imperfecta type II
COL1A1/COL1A2	Osteogenesis Imperfecta (OI) tipa III	Osteogenesis Imperfecta type III
COL1A1/COL1A2	Osteogenesis Imperfecta (OI) tipa IV	Osteogenesis Imperfecta type IV
COL2A1	Sticklerjev sindrom tipa I	Stickler syndrome, type I
COL2A1	Platispondilna letalna skeletna displazija tipa Torrance	Platyspondylic Lethal Skeletal dysplasia, Torrance type
COL2A1	Ahondrogeneza tipa II ali hipohondrogeneza	Achondrogenesis, type II or hypochondrogenesis
COL2A1	SED congenita	SED congenita
COL4A1	Porencefalija 1	Porencephaly 1
COL9A2	Epifizna displazija, multiplazija, 2	Epiphyseal dysplasia, multiple, 2
COL9A3	Epifizna displazija, multiplazija, 3, z ali brez miopatije	Epiphyseal dysplasia, multiple, 3, with or without myopathy
COMP	Psevdoahondroplazija	Pseudoachondroplasia
COMP	Epifizna displazija, multiplazija, 1	Epiphyseal dysplasia, multiple, 1
CREBBP	Rubinstein-Taybi sindrom 1	Rubinstein-Taybi Syndrome 1
CREBBP	Menke-Hennekam sindrom 1	Menke-Hennekam syndrome 1
CTCF	Intelektualna razvojna motnja, avtosomno dominantna 21	Intellectual developmental disorder, autosomal dominant 21
CTNNB1	Nevrološko razvojna motnja s spastično diplegijo in okvarami vida (NEDSDV)	Neurodevelopmental disorder with spastic diplegia and visual defects
DNM1	Razvojna in epileptična encefalopatija 31	Developmental and epileptic encephalopathy 31
DYNC1H1	Intelektualna razvojna motnja, avtosomno dominantna 13	Intellectual developmental disorder, autosomal dominant 13
DYRK1A	Intelektualna razvojna motnja, avtosomno dominantna 7	Intellectual developmental disorder, autosomal dominant 7
EBP	Hondrodisplazija punktata, vezana na X, dominantna	Chondrodysplasia punctata, X-linked dominant
EFNB1	Kraniofrontonazalna displazija	Craniofrontonasal dysplasia
EFTUD2	Mandibulofacialna disostoza tipa 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-Weissov sindrom	Jackson-Weiss Syndrome
FGFR1	Trigonocefalija 1	Trigonocephaly 1
FGFR1/FGFR2	Pfeifferjev sindrom	Pfeiffer syndrome
FGFR2	Saethre-Chotzen sindrom	Saethre-Chotzen Syndrome
FGFR2	Sindrom displazije upognjenih kosti	Bent Bone Dysplasia Syndrome
FGFR2	Beare-Stevenson Cutis Gyrata sindrom	Beare-Stevenson Cutis Gyrata syndrome
FGFR2	Antley-Bixler sindrom brez genitalnih anomalij ali motene steroidogeneze	Antley-Bixler Syndrome Without Genital Anomalies Or Disordered Steroidogenesis
FGFR2	Crouzonov sindrom	Crouzon syndrome
FGFR2	Apertov sindrom	Apert syndrome
FGFR3	Tanatoforična displazija tipa II	Thanatophoric Dysplasia, type II
FGFR3	SADDAN (huda ahondroplazija z zaostankom v razvoju in acanthosis nigricans (temni poroženevajoči predeli kože v predelih kožnih pregibov in gub))	SADDAN (severe achondroplasia with developmental delay and acanthosis nigricans)
FGFR3	Muenke sindrom	Muenke Syndrome
FGFR3	Crouzonov sindrom z acanthosis nigricans (temni poroženevajoči predeli kože v predelih kožnih pregibov in gub)	Crouzon syndrome with acanthosis nigricans
FGFR3	Tanatoforična displazija tipa I	Thanatophoric Dysplasia, type I
FGFR3	Hipohondroplazija	Hypochondroplasia
FGFR3	Ahondroplazija	Achondroplasia

FLNA	Otopalatodigitalni sindrom tipa II	Otopalatodigital syndrome, type II
FLNB	Atelosteogeneza tipa I	Atelosteogenesis, type I
FLNB	Larsenov sindrom	Larsen Syndrome
FLNB	Atelosteogeneza tipa III	Atelosteogenesis, type III
FLNB	Bumerangova displazija	Boomerang dysplasia
FOXG1	Rettov sindrom, prirojena različica	Rett syndrome, congenital variant
FOXP1	Intelektualna razvojna motnja z motnjami v govoru in z ali brez avtističnih značilnosti	Intellectual developmental disorder with language impairment with or without autistic features
FREM1	Trigonocefalija 2	Trigonocephaly 2
GABRA1	Razvojna in epileptična encefalopatija 19	Developmental and epileptic encephalopathy 19
GABRB2	Razvojna in epileptična encefalopatija 92	Developmental and epileptic encephalopathy 92
GATAD2B	Sindrom GAND (nevrološko razvojna motnja, povezana z GATAD2B)	GAND syndrome
GFAP	Alexandrova bolezen	Alexander Disease
GNAO1	Razvojna in epileptična encefalopatija 17	Developmental and epileptic encephalopathy 17
GNAO1	Nevrološko razvojna motnja z nehotenimi gibi	Neurodevelopmental disorder with involuntary movements
GRIN1	Nevrološko razvojna motnja z ali brez hiperkinetičnimi gibi in napadi, avtosomno dominantna	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant
GRIN2B	Razvojna in epileptična encefalopatija 27	Developmental and epileptic encephalopathy 27
GRIN2B	Intelektualna razvojna motnja, avtosomno dominantna 6, z napadi ali brez 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 in epileptična encefalopatija 54	Developmental and epileptic encephalopathy 54
HRAS	Costello sindrom	Costello Syndrome
IFITM5	Osteogenesis imperfecta tipa 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	Genitopatelarni sindrom	Genitopatellar syndrome
KCNB1	Razvojna in epileptična encefalopatija 26	Developmental and epileptic encephalopathy 26
KCNJ2	Andersenov sindrom	Andersen syndrome
KCNQ2	Razvojna in epileptična encefalopatija 7	Developmental and epileptic encephalopathy 7
KCNT1	Razvojna in epileptična encefalopatija 14	Developmental and epileptic encephalopathy 14
KIF1A	NESCAV sindrom	NESCAV syndrome
KMT2A	Wiedemann-Steinerjev 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	Prirojena mišična 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 in značilne obrazne poteze z ali brez okvar srca	Impaired intellectual development and distinctive facial features with or without cardiac defects
MEF2C	Nevrološko razvojna motnja s hipotonijo, stereotipnimi gibi rok in motnjami govora	Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language
MSX2	Kraniosinostoza 2	Craniosynostosis 2
MSX2	Parietalna foramina s kleidokranialno displazijo (PFMCCD)	Parietal Foramina With Cleidocranial Dysplasia

NALCN	Prirojene kontrakture okončin in obraza, hipotonija in zaostanek v razvoju (CLIFAHDD)	Congenital Contractures Of The Limbs And Face, Hypotonia, And Developmental delay
NF1	Nevrofibromatoza 1	Neurofibromatosis 1
NF2	Nevrofibromatoza 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 optične 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	Intelektualna razvojna motnja, avtosomno dominantna 36	Intellectual developmental disorder, autosomal dominant 36
PPP2R5D	Intelektualna razvojna motnja, avtosomno dominantna 35	Intellectual developmental disorder, autosomal dominant 35
PRKAR1A	Akrodisostoza 1, z ali brez hormonske rezistence	Acrodysostosis 1, with or without Hormone Resistance
PTPN11	Noonanov sindrom 1	Noonan Syndrome 1
PURA	Nevrološko razvojna motnja z dihalno stisko novorojenčka, hipotonijo in težavami pri hranjenju	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	Nevrološko razvojna motnja z ali brez anomalij možganov, oči ali 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	Metafizična displazija z maksilarno hipoplazijo in z ali brez brahidaktilije	Metaphyseal Dysplasia with Maxillary Hypoplasia with or without Brachydactyly
RUNX2	Kleidokranialna displazija	Cleidocranial dysplasia
SATB2	Glass sindrom	Glass Syndrome
SCN1A	Zgodnja dojenčkova epileptična ecefalopatija 6	Early Infantile Epileptic Encephalopathy 6
SCN1A	Razvojna in epileptična encefalopatija 6B, ne Dravetova	Developmental and epileptic encephalopathy 6B, non-Dravet
SCN2A	Razvojna in epileptična encefalopatija 11	Developmental and epileptic encephalopathy 11
SCN2A	Epizodična ataksija tipa 9	Episodic ataxia, type 9
SCN8A	Razvojna in epileptična encefalopatija 13	Developmental and epileptic encephalopathy 13
SCN8A	Kognitivna motnja z ali brez cerebelarne ataksije	Cognitive Impairment With Or Without Cerebellar Ataxia
SETBP1	Schinzel-Giedionov sindrom	Schinzel-Giedion syndrome
SETBP1	Intelektualna razvojna motnja, avtosomno dominantna 29	Intellectual developmental disorder, autosomal dominant 29
SETD2	Luscan-Lumish sindrom	Luscan-Lumish Syndrome
SETD5	Intelektualna razvojna motnja, avtosomno dominantna 23	Intellectual developmental disorder, autosomal dominant 23
SHANK3	Phelan-McDermid sindrom	Phelan-McDermid Syndrome
SHOC2	Noonanu podoben sindrom z izpadajočimi anagenimi lasmi	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	Blefarofimozni sindrom - sindrom oslabljenega intelektualnega razvoja	Blepharophimosis-impaired intellectual development syndrome
SMARCA4	Avtosomno dominantna duševna zaostalost 16	Autosomal Dominant Mental Retardation 16

SMARCB1	Avtosomno 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 in epileptična encefalopatija 5	Developmental and epileptic encephalopathy 5
SRCAP	Sindrom plavajočega pristanišča	Floating-Harbor Syndrome
SRCAP	Zaostanek v razvoju, hipotonija, mišično-skeletne okvare in vedenjske motnje	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities
STAT3	Hiper IgE sindrom	Hyper-IgE recurrent infection syndrome
STXBP1	Razvojna in epileptična encefalopatija 4	Developmental and epileptic encephalopathy 4
SYNGAP1	Intelektualna razvojna motnja, avtosomno dominantna 5	Intellectual developmental disorder, autosomal dominant 5
TBL1XR1	Pierpont sindrom	Pierpont Syndrome
TBL1XR1	Intelektualna razvojna motnja, avtosomno dominantna 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-Dietz sindrom 1	Loeys-Dietz syndrome 1
TGFBR2	Loeys-Dietz sindrom 2	Loeys-Dietz syndrome 2
TRAF7	Srčne, obrazne in digitalne anomalije z zaostankom v 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	Prirojene simetrične obodne kožne gube 1	Symmetric circumferential skin creases, congenital, 1
TUBB	Kompleksna kortikalna displazija z drugimi možganskimi malformacijami tipa 6	Cortical Dysplasia, Complex, with Other Brain Malformations 6
TUBB2A	Kompleksna kortikalna displazija z drugimi možganskimi malformacijami tipa 5	Cortical Dysplasia, Complex, with Other Brain Malformations 5
TUBB4A	Hipomielinizacijska levkodistrofija tipa 6	Leukodystrophy, hypomyelinating, 6
TWIST1	Saethre-Chotzenov sindrom z ali brez anomalij vek	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 kopičenjem železa v možganih tipa 5	Neurodegeneration with brain iron accumulation 5
ZBTB20	Primrose sindrom	Primrose syndrome
ZC4H2	Wieacker-Wolffov sindrom (samo pri ženskah)	Wieacker-Wolff syndrome, female-restricted
ZEB2	Mowat-Wilsonov sindrom	Mowat-Wilson syndrome
ACTB	Sindrom distonija-gluhost 1	Dystonia- Deafness Syndrome 1
ATP1A3	Alternirajoča hemiplegija otroštva 2	Alternating hemiplegia of childhood 2
ATP1A3	Sindrom CAPOS	CAPOS syndrome
COL1A1	Kombinirana osteogeneza imperfecta in Ehlers-Danlosov sindrom 1	Combined osteogenesis imperfecta and ehlers-danlos syndrome 1
COL2A1	Sticklerjev sindrom tip I, nesindromski očesni	Atypical stickler syndrome, type I
COL2A1	Kniestova displazija	Kniest dysplasia
FLNA	Periventrikularna nodularna heterotopija 1	Periventricular nodular heterotopia 1

KCNT1	Nočna epilepsija frontalnega režnja 5	Epilepsy, nocturnal frontal lobe, 5
KIF1A	Spastična paraplegija 30, avtosomno dominantna	Spastic paraplegia 30, autosomal dominant
NF1	Neurofibromatoza-Noonanov sindrom	Neurofibromatosis-noonan syndrome
NF1	Watsonov sindrom	Watson syndrome
NFIX	Malanov sindrom	Malan syndrome
PIK3CA	Sindrom prekomernega rasti in/ali cerebralne malformacije	Overgrowth syndrome and/or cerebral malformations
PTPN11	Sindrom LEOPARD 1	Syndrome LEOPARD 1
SETD2	Intelektualna razvojna motnja, avtosomno dominantna 70	Intellectual developmental disorder, autosomal dominant 70
SMC1A	Razvojna in epileptična encefalopatija 85 z ali brez sredinskih možganskih okvar	Developmental and epileptic encephalopathy 85 with or without midline brain defects
SPTAN1	Razvojni zaostanek z ali brez epilepsije	Developmental delay with or without epilepsy

NIFTY PREMIUM STATISTIKA

SINDROM		OBČUTLJIVOST	SPECIFIČNOST	PPV
Trisomija 21		99.17%	99.95%	92.19%
Trisomija 18		98.24%	99.95%	76.61%
Trisomija 13		>99.9%	99.96%	32.84%
CNV	**≥10 Mb**	>99.9%	99.97%	NA
	<10 Mb	>99.9%	99.86%	NA
Monogenske bolezni		>99%	>99%	NA
Določitev spola		99,53%	99.20%	NA
ANEVPLOIDIJE SPOLNIH KROMOSOMOV		**OBČUTLJIVOST**	**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%

Opomba: Podatki v tabeli temeljijo na obstoječi literaturi in internih podatkih ter odražajo le preteklo zaznavanje, ne pa dejanskega stanja testiranega vzorca ali podane vrednosti.

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